

Visual Analytics in Personalized Health: A Study of the Expert Analyst – Health Consumer Relationship in a Direct-to-Consumer Service

by

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Abstract

In the era of “big data analytics” for healthcare, the personalized medicine promise offers a shift to the provision of care enabled by our technical ability to quantify and assess large volumes of biomedical data. This message however, often seems to strengthen a notion of healthcare from a “biomedical positivism framework”, that is, that diagnosis of disease, medical image analysis, integration of devices, and ultimately, the selection of the appropriate therapy is empowered by volumes of data and algorithmic accuracy, thus improving the patient’s illness. In this research program, we approached expert biomolecular analysts, recorded their sensemaking process, and analyzed the role of data visualization technologies while they performed analysis of multi-omic data for a direct-to-consumer service of personalized health.

We uncovered the nature of the analysts turning to their human-interaction skillset to address the health reality of each consumer they worked for. Assertions about the scientific validity and the amount of data, often emphasize the claims of this personalized health approach, but in practice, the analysts turned to attend goals, preferences, to find actionable evidence in the data, and to frame a relatable health summary story for the clients. The role of technology design in scenarios like this one will be fundamental in properly translating and bridging the effort from these emergent providers (the analysts) in communication with the end consumers. Our findings suggest that both parties benefit from analytic capacities to explore and understand the strength of each piece of evidence in the case, including the evidence that is provided by the clients themselves beyond their biological samples.

We believe that this work, along with the research methodologies deployed in work-settings, are a contribution to the Visual Analytics community to support the tasks of bio scientists in personalized medicine, as much as an HCI initiative in support of evidence-based models of preventive healthcare with large amounts of data.

Keywords: personalized medicine; visual analytics in healthcare; human computer interaction; omics analysis; pair analytics; qualitative research

A Roberto y Águeda,
El contenido es hacerlo,
El camino es la meta.

To my family across the continent,
with Love and gratitude.

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Table of Contents

Approval.....	ii
Ethics Statement.....	iii
Abstract.....	iv
Dedication.....	v
Acknowledgements.....	vi
Table of Contents.....	vii
List of Tables.....	x
List of Figures.....	xi
List of Acronyms.....	xii
Chapter 1. Introduction.....	1
Context: The Personalized Medicine Revolution.....	5
Definitions & Settings of the Study.....	6
Understanding the user experience: Studies of multiple stakeholders in personalized medicine.....	9
Research Questions.....	10
Thesis Overview.....	12
Chapter 2. Related Works and Theoretical Frameworks.....	14
2.1. Related works on Visual Analytics and Studies of Sensemaking.....	14
2.2. Theory: The Sensemaking Loop of Intelligence Analysis.....	16
2.3. Theory: Evidence-Based Medicine.....	19
2.4. Theory: The Doctor-Patient Relationship.....	23
2.5. Theory: The Role of Expertise and Task Characteristics.....	27
Studies of Expertise.....	27
Tasks Characteristics and the Expert's Environment.....	30
2.6. Joint Action Theory in Visual Analytics Research.....	33
Chapter 3. Methodological Approach: Pair Analytics and In-Vivo Studies of Sensemaking.....	38
3.1. Design Research and Studies of Cognition in Responsive Environments.....	38
3.2. In-Vivo Studies of Cognition: The Pair-Instruction-Analysis Method and Protocol Analysis.....	40
The original pair analytics method.....	40
Variation in context: the instructional interaction.....	43
The researcher's role in an embedded capacity.....	44
Pair Analytics and other fieldwork approaches to studying sensemaking in work environments.....	45
3.3. Bounding the Study.....	48
Setting.....	48
Actors.....	48
The role of expertise: Studying medical professionals compared to molecular biology scientists.....	50

Data collection	51
Sample size sufficiency and data adequacy	52
Ethical considerations	53
3.4. Data Analysis: Thinking with Pair Analysis Data	54
Transcripts	54
Coding	55
Analysis Results: Emergence from an Inductive Approach	58
The sense-making loop	59
Actionable capacity, trust, and confidence in health evidence	61
“The fourth person in the room”	63
Reflective expertise (or the metacognitive ability)	65
Team collaboration in a distributed process	67
3.5. Summary	67
Chapter 4. Interpreting Multi-Omics Data in a Personalized Medicine	
Organization	69
4.1. Overview of the Socio-Technical System: People, Project Planning, and Data Reporting Tools	70
Managing the Analysis Process	71
The Data Report	72
The OMICS analytics and reporting information system	74
4.2. What Constitutes Evidence?	81
A Central Repository of Scientific Knowledge	82
Types of Evidence	83
Visualizing Confidence of Evidence	89
Interpreting Evidence: Schemas or Frames of Reference	91
4.3. Cycles of Analysis Working with Biomolecular and Self-reported Health Data	95
Creating the Health Data Summary	97
Creating Diet and Supplement Recommendations	103
Physical Activity Assessment and Recommendations	108
4.4. Summary	115
Chapter 5. Joint Action Theory and the Client-Analyst Relationship in Direct-to-Consumer Personalized Health	118
5.1. Joint Action Theory and the Creation of Meaning in an Asynchronous Distributed Process	120
The Expert Analyst– Client Relationship as a Joint Activity	121
Advancing Joint Activities	124
Coordination Devices	128
Signaling	130
5.2. A Comparative Examination with the Doctor – Patient Relationship	134
Chapter 6. Discussion of Results and Implications	138
6.1. Overview of Results	139
6.2. Implications	143

Role Participation and Role Awareness: The Tacit Health Provider	143
Roles' Dynamics: Mutual Participation	145
Transparency and Completeness of Evidence	145
Defining Quality in Self-Reported Health Information.....	148
The Power of Narratives: The Machine Learning and Big Data Assisted Medicine Rhetoric	149
6.3. Study Limitations.....	150
Chapter 7. Conclusions and Future Work	152
7.1. Thesis Summary	152
7.2. Contributions	154
7.3. Future Directions.....	156
References.....	158
Appendix A. Pair Analysis Session Instruction Sheet for Participants	169
Appendix B. Consent Form	170
Informed Consent Form	170
Appendix C. Transcripts	173

List of Tables

Table 2.1	Task characteristics associated with good and poor performance in experts. From (Shanteau, 1992).....	31
Table 2.2	Features of face-to-face conversation. In (Clark & Brennan, 1991)	36
Table 3.1	Study participants and data collection summary.....	49
Table 3.2	Codes in The Sense-making Loop theme	60
Table 3.3	Example quotes of the Actionability, trust, and familiarity of evidence	62
Table 3.4	Codes in the Actionable capacity theme	62
Table 3.5	Example quotes about communicating risk, and creating a narrative for the client.....	63
Table 3.6	Codes in “The fourth person in the room” theme	65
Table 3.7	Codes in the Reflective expertise theme.....	67
Table 4.1	Actors with anonymized identities	70
Table 4.2	Categories of scientific evidence from (Kumar, 2007)	84
Table 5.1	Features of face-to-face conversation. In (Clark & Brennan, 1991)	122
Table 5.2	Features of the setting of mediated DTC services viewed as a joint activity.	123
Table 5.3	Expert – Client signaling instances in the recorded data.....	131

List of Figures

Figure 1.1	From blood samples to a consumer-tailored health data report: an overview of the process.....	9
Figure 2.1	The sensemaking loop for intelligence analysis (Pirulli & Card, 2005)....	18
Figure 2.2	A model of evidence-based clinical decisions with evidence, circumstances, and preferences in the decision-making process (Haynes, 2002).....	21
Figure 3.1	Data Collection for the Pair Analysis Protocols.....	51
Figure 3.2	Atlas.ti view of the Pair Analysis data.....	57
Figure 3.3	Frequency of codes in the dataset. Groundness.	61
Figure 4.1	Card and board design of the platform used to manage the process.	72
Figure 4.2	The landing page of the Provider view (top) and the details of information provided as evidence of a risk assessment (bottom).....	76
Figure 4.3	Screencapture of the note editing for prediabetes	77
Figure 4.4	Landing page of the Client view (top) shows the health data summary as a text summary. (Bottom) Details of Pre-Diabetes obtained when clicking on the corresponding title from the summary or navigating through the menu.	79
Figure 4.5	Nutritional data hierarchy.....	81
Figure 4.6	Results reported for four measures. Visualization of the reference range.	85
Figure 4.7	A network of molecular measures that are outside of the reference ranges and for which there is evidence of association with organ systems and diseases.....	90
Figure 4.8	Confidence score from 1 to 4 in the Provider view, listed with each biomarker in the context of a disease.....	91
Figure 4.9	Overview of nutrition recommendations in an action plan. Clients get a list of actions (left) and can obtain details about how does it connect to their health data results (right).....	96
Figure 4.10	Visual depiction of a list of recommendations in a single action plan.....	97
Figure 4.11	List of biomarkers and diseases bookmarked with flags for the action plan team.	112
Figure 5.1	Experts and Client coordinate on content in the interface. Each participant contributes pieces at different moments in time.....	124
Figure 5.2	Timeline of events with call for action in the Client Interface	127
Figure 5.3	Top: Action plan example. Information displayed to the client with arguments in text. Bottom: Health risk assesment example.....	130
Figure 6.1	Roles and type of relationship in the DTC personalized health provision model.....	144
Figure 6.2	The evidence displayed in the <i>Client Interface</i> in a flat layout, without hierarchies.....	147

List of Acronyms

EBM	Evidence-Based Medicine
VA	Visual Analytics
JAT	Joint Action Theory
DCOG	Distributed Cognition

Chapter 1.

Introduction

Fred is doing his best to interpret the health report he is reading on his electronic device. Fred is a 55 years old man who has been motivated to invest in lifestyle changes that can help him maintain his health and wellness into his elderly age. Fred has made the choice to introduce more short walks in his day and he is conscious about getting on his bike on a regular basis for the sake of his fitness and the pleasure of Vancouver's sightseeing. Recently, Fred walked into the pharmacy and learned about a new type of advanced test based on his body's molecular markup that can help him tailor his diet and physical activity routine to achieve his best. The providers of the test claim they can help him to tailor lifestyle actions to target the goal of maintaining his health. Fred completes an intake form which includes his health history and current activity routine. He submits the form along with a blood test and a couple of weeks later Fred is checking the results of a digital report that contains a vast amount of data. The report starts with a health brief and a tailored action plan created by a group of experts. The brief talks about early signals of risks for diabetes and cardiovascular disease. It highlights that his body is currently showing signs of inflammation and weak immune function, two very important factors to consider in the prevention of illness. The report includes a full dataset of blood markers measured, risks assessed, and the scientific evidence backing up the interpretation of the results. The amount of data is beyond what Fred is able to process to fully understand his own health. Fred is able to focus on the brief containing the highlights and in the tailored health plan with diet, supplements, and physical activity recommendations. Fred may not be able to understand much of what the report is saying, but according to the healthcare industry, Fred is now a health consumer of personalized medicine. This is a shift in the delivery of medicine that focuses on prediction, personalization, prevention, and participation (or democratization) of healthcare, empowered by our technological ability to measure and analyze biomolecular data at a scale not conceived of before.

The analytics of large amounts of biomolecular data is a topic of interest in multiple computing areas: AI and mixed-initiative research groups focus on developing

accurate and efficient assisting technologies to put in clinical practice, while HCI, Visual Analytics (VA) and even Healthcare informatics groups are also interested in the systemic approach to the practice of medicine that involves providers, clinicians, and most specially, health consumers. In this dissertation my goal is to bring forward the role of expertise, the role of environment in which the expert analysts operate, and the role of the task characteristics at hand, in an effort to reframe the way in which technologists in the visual analytics community have adopted cognitive psychology research about heuristics, biases and the communication of results within the decision making process

Fred's story weaves the narrative of the research interest in this dissertation, to describe the process of decision making, validation, and data visualization technologies a group of experts use in the process of interpreting biomolecular data profiles for healthcare consumers. Adding to the complexity of capturing digital health information is the increasing complexity of medical tests and their interpretation. Declines in price of biomolecular testing and Big Data advances in analysis methods have made commercial precision medicine tests more affordable, increasing the complexity of medical records. These more complex documents create additional challenges for health analysts in understanding the patient experience in order to better communicate analyses in ways that are compatible with the mental models of patients and caregivers.

This will be especially important to consider in more recent models of healthcare where many traditional providers are removed from their role as information gatekeepers and so, the role of the health data analyst now requires to pay attention to the interpretation and specially, the communication of health results directly to an individual. It is a model that continues to align and evolve with the promotion of collaborative, patient-centered and patient-driven practices of decision-making in the healthcare industry.

“Human health is too precious to turn it to automated procedures only” –argues Eric Topol in a *Nature Medicine* think piece (Topol, 2019). Our technological ability to create machines for ingesting and drawing meaningful inferences from massive sets of data quickly, accurately, and inexpensively, will ultimately lay the foundation for high-performance medicine. The proposal is that high performance medicine is truly data-driven, decompressing our reliance on human resources only; It will eventually take us well beyond the sum of the parts of human and machine intelligence, but as technology

designers, we are obliged to face this process with a detailed characterization of what we consider autonomous technologies *today*. In Topol's review (2019), the opportunities and pitfalls of AI in medical practice, highlight the progress made on the application of neural networks –a type of AI algorithm, highlighting the common pitfall of believing that just validation of the performance of the algorithm in terms of its accuracy is equivalent to demonstrating clinical efficacy (Topol, 2019). Drawing an analogy between self-driving cars and medicine, Topol describes the vision of the AI assisted medicine future as a middle-point synergy between the functions of the clinician (or provider) and the machines, a change from the human-only approach in diagnosing, but distant from the idea of full automation with no potential for clinicians backup (p.51). What I take as important from this vision is that there is an important part and place for emerging roles of decision making within this synergic process.

Visual Analytics (VA) – “*the science of analytical reasoning facilitated by visual interactive interfaces*” (Thomas & Cook, 2005) is an interdisciplinary science that synthesizes methods and techniques from multiple disciplines including information visualization, HCI, data mining, data management, decision science, mathematical and computational science, statistics, cognitive psychology, and perceptual science . VA was born separately from the algorithm-devoted research communities making a strong argument about crediting the creative ingenuity of expert analysts instead of replacing the decision-making process with automated algorithms.

With that in mind, in this dissertation I document the process that a group of interdisciplinary experts follows to provide a human-interpretation layer in a biomolecular test to facilitate the understanding of evidence-based risk assessments for an individual. The interpretation layer –or highlights of findings– is the starting point for a consumer into their health assessment created from hundreds or molecular markers measured from their own plasma samples and genetic analysis. The product of the interpretation by experts plays a key component in the communication with a client challenged with understanding health outcomes derived from black-boxed analysis, using large amounts of data. I present the account of events in this analysis process documented via a collaboration and involvement with a small for-profit organization that provides biomolecular testing directly to consumers (DTC). The service of this organization is described as a non-diagnostic, empowering individuals with preventive lifestyle interventions based on the latest scientific evidence about their molecular results. Direct-

to-consumer is a model that has gained strength in emerging trends in the healthcare industry. It implies that health products or services are marketed directly to consumers and tests can be bought online or in stores. The most relevant feature is that consumers receive their results directly via interactive apps, websites, or written reports, without the mediation of a provider that has requested the tests for them, or that can lead the interpretation of results.

In this work I turn to the use of a prominent theory from psycholinguistics and the cognitive sciences, *Joint Action Theory (JAT)* to investigate the features of the expert-consumer relationship in the DTC healthcare model. In Visual Analytics, the community has formerly incorporated JAT for the analysis of expert cognition, reasoning, and tacit knowledge from conversational data. Herbert Clark presents *Joint Action Theory* as a framework for looking at interaction (activities) in which two or more individuals participate, by studying how do they construct meaning in their use of communicative acts. According to Clark, language use and joint activities are inseparable, we cannot understand one without the other, and so the theory provides a set of constructs to look at roles, activities, markers of entry and exit participation, and grounding in communication (Clark, 1996). And so, following Clark's proposal of language use, in this dissertation I take a lens on how the design of visual analytics tools for analysts in personalized medicine can be better informed not only from the individual requirements for analyst and client separately, but by looking at the performance of their situation as a joint activity.

With a JAT lenses, researchers have a novel conceptual model to unpack the features of communication between experts and consumers, mediated by interactive information reporting tools. The goal is to better understand the emerging roles in the provision of preventive care that, when not a clinician, the analysts seem to cover to support a consumer in their interpretation of risk, evidence, and overall uncertainty. In this way, the underlying goal of this research is to broaden our understanding of the application of *Joint Action Theory* in shifting the way in which Visual Analytics researchers can study reasoning, expertise, and the process of creating meaning within an evidence-driven, decision-making processes for personalized medicine. Ultimately, with the goal to translate this understanding to the design of digital health technologies.

This will be relevant to the goal of advancing the personalized medicine industry because while the promise to power the innovation cycle of systems medicine via clouds of data from patients and consumers currently holds true, the ultimate users of the data, whether a health consumer or a provider, are still challenged by the complexity and difficulty to bridge data into a direct health action or clinical practice. This niche is a valuable research arena for visual analytics experts and technology designers interested in supporting the process of deriving health insight from an expert and a consumer synergy.

Context: The Personalized Medicine Revolution

*Humans are too complex to be treated as average. — Leroy Hood.
Institute for systems biology*

Today's healthcare is largely based on advances that benefit the population as a whole rather than a particular individual, but this one-size-fits-all approach does not take into account the fact that one may differ significantly from the "average" person and that individuals have different goals for their own health. Being "*healthy*" and "*in better health*" are moving targets from person to person and throughout an individual's life. However, major advances in bioinformatics and clinical research of molecular measures in the past decades have made possible to understand our health more precisely than ever before.

Personalized Medicine –also known as Precision Health or Personalized Health, a rapidly advancing field of health care that is informed by each person's unique clinical, genetic, genomic, and environmental information is based in the use of information from genomes and their derivatives (RNA, proteins, and metabolites) to guide medical decision making ((Ginsburg & Willard, 2009)). Personalized medicine bloomed over the past decade with more accessible genetic sequencing and continues to grow into clinical practice and direct-to-consumer markets now with the ability to measure concentrations of hundreds of other molecular blood markers like proteins and metabolites in the body. These blood markers, known as **biomarkers** in the molecular biology circles, take further the genetic blueprint and our ability to assess the risk of "*what could happen*" to a present picture of "*what is happening*", a present-day molecular picture of an individual's health (Cullis, 2015). With personalized health, we enter the era of providing the right treatment to fit an individual's biological profile.

The power to measure all these indicators together enables us to create a picture of susceptibility to diseases, systemic performance of the body, as well as tapping into the potential to see the physical predisposition to diets and fitness activity for each body. The promise with Personalized health is to shift our understanding of health from “*the absence of illness*” towards “*a wellness and optimization of lifestyle*” tailored to each individual with biomolecular precision for the treatment of maladies, as much as for prevention and performance of individual bodies.

The relevance of creating information aids for consumers from their own health data, responds to the different challenges faced by current models of healthcare where consumers and providers have agency and active roles on the “health jobs”¹, or the tasks that are being done as part of the provision of care for a person, whether those tasks are related to diagnosis, treatment, or prevention. As practitioners in Visual Analytics and the design of interactive visualization decision-support systems, we see the opportunities and challenges, because realizing this vision will require not only innovative computational solutions and data science methods, but also innovative ways to deliver decision support to both clinicians and consumers, specially before the later become active patients. Research already suggests that not only poor interface design, but also poor fit with clinical work practices, and the disconnect between the system design and existing knowledge on clinical reasoning and decision-making as the chief reasons for low impact of decision-support systems on clinical practice(Sittig et al., 2008).

Definitions & Settings of the Study

The FDA defines a **biomarker** as a “*characteristic that is measured as an indicator of normal biological processes, pathogenic processes, or responses to an exposure or intervention, including therapeutic interventions*” (FDA-NIH Biomarker Working Group, 2016) and specifies biomarkers in types such as: Molecular (e.g. blood glucose), radiographic (e.g. tumor size), and physiologic (e.g. blood pressure).

¹ A Health Job is a term introduced by Dr. Brendan Byrne, a BC physician advocator of the long due digital health and personalized health transformations in healthcare. <https://www.linkedin.com/pulse/health-jobs-basic-unit-work-digital-brendan-byrne/>

In the domain of personalized health, we use the term *biomarker* to refer mostly to the measurable indicators at the molecular level that can be used for the assessment of some biological state or condition. The subsets of these type of markers are used in the field in terms of their biological composition or role, for example (1) proteins measured mostly from plasma samples such as C-Reactive Protein; (2) metabolites, or products of the metabolism, measured both in plasma and urine samples, for example glucose or serotonin; (3) SNP variants, variations in the DNA sequence at particular locations that generate biological variation between people by causing differences in the recipes for proteins that are written in genes. Each of these types is studied under an “omic” science, *proteomics*, *metabolomics*, and *genomics*, respectively, and for that reason the biomarkers are also known as “omics” markers.

For the specific situation in this dissertation, I looked at the process to transform raw omics data into a health data report directed to a consumer. In this study, **a health data report**—or just data report—is the collection of quantified biomarkers and the collection of evidence that links those analytes to health dimensions for an individual. Dimensions such as: disease risks, organs health, functions of the body assessment, genetic predisposition to medications, or nutritional and physical activity highlights.

Overall, due to policies on data control, ownership, and the ethics on reportable findings that binds the service providers to report back on known risks given the data, the resulting report contains hundreds of data points and a myriad of connections to research evidence. For this reason, the **health data summary** (or health data highlights) is the interpretation process and summary findings that experts create from the data analyzed and reported. The goal of the expert-interpretation process is to create a personal health narrative for an individual based on the multiple dimensions of data collected and assessed. Through the analysis, the scientists highlight what are the more relevant results given multiple factors. I will look to understand what these factors are when experts evaluate biomolecular data results. For example, a well-known determinant in the assessment is the clinical and scientific confidence of some markers (e.g. finding that the glucose concentration in a fasting sample of plasma is higher than 6000 μ M, has the highest confidence in the current medical literature to indicate a risk, or even a diagnosis, of diabetes and related comorbidities); but not all biomarkers have the same level of confidence or have been so widely studied in relationship to a single disease thus, experts go into an assessment process to identify what can be most

relevant. This is a case of decision making under uncertainty and incomplete data that we can observe. In the context of this study, the analysts looking at the data are qualified experts, but the data can still be scarce for each possible diagnostic case, the process is highly distributed, the expertise is interdisciplinary, and the goal is to keep the interpretation efficient (in volume of reports to look at and time spent) and reliable for clinical practice. Finally, the **action plan**, is a set of lifestyle recommendations that the experts create from the biomolecular results and the client health history and preferences. In this dissertation we will follow the process that a nutritionist and a physical activity expert follow to create actions for their clients.

Figure 1.1 depicts a high-level overview of the process from taking blood samples to having a report returned to a consumer². After measuring the levels of hundreds of proteins, metabolites, and the presence of thousands of genetic variants (snp variants), the data goes through automated quality control validations to then algorithmically create a health data report. This report includes the assessment of risk for close to a hundred diseases and other early warning signs. Behind the scenes, this analysis is drawing from a vast array of scientific literature and clinical research to bring the personal insights based on the latest scientific knowledge. However, the results are long and complex. Understanding the confidence in the different assessments of risk still requires the interpretation of expert analyst. The teal section of the process shows experts integrating the health records and lifestyle preferences reported by clients to add an interpretation layer to the results. Experts highlight what is most relevant from the results and create tailored health recommendations from those findings. The end result is a dashboard delivered to the consumer that includes all the data and the interpretation reports from the experts.

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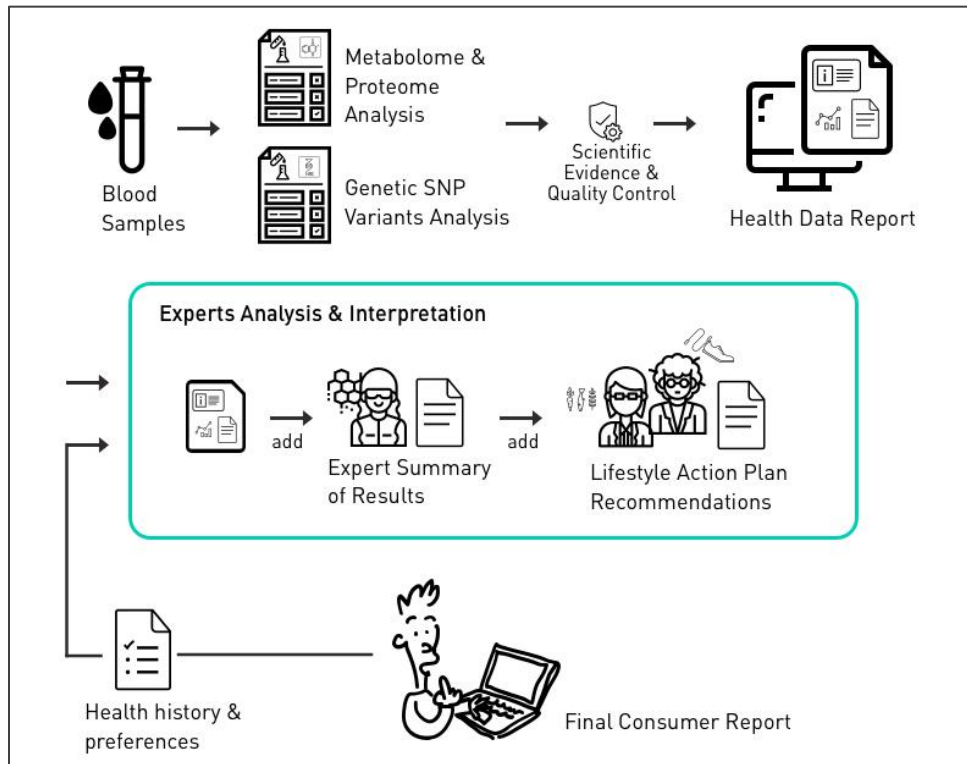


Figure 1.1 From blood samples to a consumer-tailored health data report: an overview of the process.

Understanding the user experience: Studies of multiple stakeholders in personalized medicine

In any healthcare research endeavor, understanding the journey of patients or health consumers intrinsically involves understanding a journey of provision of care with multiple stakeholders. In this dissertation the focus is on the providers, particularly the emergent roles completing that help a patient understand data, take informed decisions, and be empowered.

The case documented in this dissertation as an interpretation process, is an effort to respond to the issue of communicating personalized health data that has been challenged with low trust and difficulty interpreting results, as it has happened to many initiatives in the industry (Bloss et al., 2010; Carere et al., 2016; Lachance et al., 2010; Sillence et al., 2006).

In a service model where physicians no longer act as gatekeepers and a confident source of information, personalized medicine initiatives are under the constant pressure

for reinventing how to communicate big data results, preserve uncertainty, and remain valid in its effort to become a preventive, predictive and personalized approach to healthcare. In different services, the addition of layers of interpretation such as the *Health Data Summary* could be seen as an effort to mitigate the challenges faced by end users interpreting and understanding “*the signal from the noise*” after receiving data results as an assessment that provided a wealth of health information.

Research Questions

This study explores the process of experts in action whom systematically deal with interpreting biomolecular profiles of data to create health summaries directed to health consumers and their care teams. The relevance of the human in the loop in this process, the human-technical setting, and the characteristics of the task at hand using data makes it an interesting case for Visual Analytics research. Focusing on the reasoning process, the validity of expertise, the pitfalls of intuitive judgement in evidence-based reasoning, and the attributes of the communication of findings, the study in this dissertation seeks to understand:

How do analysts of multi-omics (biomolecular) data complete a consumer-tailored review of the data for personalized health? What factors, related to sensemaking and data-visualization technologies, influence their decisions for selecting the most relevant health evidence?

Motivated by this overarching question, I investigate the following research questions:

RQ1: What is the process that experts follow to derive interpretations and health recommendations from biomolecular data?

The goal is to explore and describe the interpretation process that analysts perform using multi-omic biomolecular data profiles: Proteome, Metabolome, and Genome data. The actors are experts in different professional backgrounds in biosciences and healthcare. The opportunity to understand reasoning towards evidence-based wellness and disease prevention, is a unique setting emerging with the advances of personalized medicine. Presenting the power that these data poses and the potential to bring the promise of predictive and preventive healthcare has been extensively documented as the computational power and the personalized medicine field advances (Chen et al., 2016; Hirsch & Abernethy, 2012; Price et al., 2017). However, studies in the health informatics field have also documented that a present need for health consumers

when using this information has to do with the difficulty interpret results from information that is complex, lengthy, and hardly actionable (Bloss et al., 2010; Carere et al., 2016; Lachance et al., 2010; Parthasarathy, 2010).

The issue of how to translate the value into clinical practice with actionable validity also remains object of investigation (Berg et al., 2011; Julian-Reynier et al., 2008; Laurence, 2009; Manolio et al., 2013). For this reason, gaining an understanding of how practitioners are dealing with the issue of creating interpretations of the data is a unique opportunity for the different disciplines working towards advancing personalized health. Moreover, the study of in-depth decision making in the VA literature is mostly found in the space of intelligence analysis, my case settings are a unique opportunity for the understanding of expert reasoning with visual interfaces.

RQ2: What mechanisms (if any) do biomolecular experts use to validate their argumentation and courses of action working with incomplete data, seeking evidence for health assessments? Which of these mechanisms can be attributed to skilled expertise?

In the process of understanding what is the role of the human in the creation of actionable recommendations from readings of data, we also need to understand how do experts use their expertise to make judgement calls and what mechanisms (if any) do they use to validate their own hypotheses testing. For example, Shanteau (1992) found that decision makers in professions such as nurses, physicians and auditors, exhibit good expert performance, and he determined expertise factors such as predictability of outcomes, amount of experience, and availability of good feedback. But to what extent is the expert knowledge and the characteristics of expertise the only drivers in the process? With this question I am interested in investigating the attributes of the task, the attributes of the communication, or other interactions that can be observed to be argumentation warrants in the decision-making process of the experts in this case.

From a methodological standpoint this research program assesses and evaluates the application of the *Pair Analytics Methodology* (R. Arias-Hernandez et al., 2011) to capture the nuances of the problem-solving process and richness of a distributed process in personalized medicine.

Thesis Overview

This monograph dissertation is organized in 6 chapters. Chapter 1 introduced the motivation, the guiding research objective, and set the basic context for the research as a *Visual Analytics* endeavour in the *Personalized Health* domain. In Chapter 2 I discuss related works to ground the state of the art in the study of expert decision making in *Visual Analytics*, *Evidence-Based Medicine (EBM)*, and *Patient-Centered medicine*. I also present the background for the *Joint Action Theory* framework.

Chapter 3 addresses the methodological commitments. The methodological approach is an in-situ qualitative research study where the empirical data are based on three years of work at the organization including observations, semi-structured interviews, field notes, and the deployment of *Pair Analysis* protocols. This research builds on the *Pair Analytics Methodology* for the study of cognition in a technology-mediated problem. I introduce the *Pair-instructional-analysis* method as an adaptation of the method to fit the characteristics of the situation in which I collected data. I conclude the chapter presenting the open-coding, inductive approach to the analysis of the data before turning to an in-depth analysis in chapters 04 and 05.

In Chapter 4 I present the descriptive account of the process documented in which a team of interdisciplinary experts works collaboratively to create a layer of interpretation and actionable insights for a client. This chapter turns to answer the first research question addressing the observed process, the actors, and tools in the socio-technical system. In this chapter I also discuss the attributes of evidence and confidence of the biomolecular data that are at the core of the analysis process in the observed situation.

Chapter 5 is devoted to the discussion with JAT as the analytic lens to unpack some of the attributes of the process described in chapter 04, specifically, that of communicating results to clients. This chapter is a walk through the constructs of JAT in the settings of the case recorded in the dissertation along with a comparative examination with the traditional doctor-patient relationship.

In chapter 06, I conclude the research work with a discussion of the findings and the implications from the analysis performed through chapters 04 and 05. Finally, in Chapter 7 I summarize the contributions to our understanding of the provision of care

tasks of new roles of expert performance in the domain of personalized health and present the future work.

Chapter 2. Related Works and Theoretical Frameworks

2.1. Related works on Visual Analytics and Studies of Sensemaking

The focus of my research is situated in the broader context of Visual Analytics research. Since its conception, “*the science of analytical reasoning facilitated by interactive visual interfaces*” has made the effort to drive research in both, engineering and cognitive sciences directions. The field is known to be represented by computer scientist and HCI researchers in the design of visual interfaces area, and translational researchers looking to provide actionable models of cognition in action that can inform and justify decisions on systems design. The goal is building systems that can be used to synthesize information and derive knowledge from massive, often ambiguous, dynamic and even conflicting sources of data –to “*detect the expected and discover the unexpected*” (Thomas & Cook, 2005, p. 4).

From its origins at the time of *Illuminating the path* (Thomas and Cook 2005), visual analytics has been grounded on the basis of exploiting a technology-mediated dialogue between an analyst and her information to produce a judgment about an issue. Human analytical reasoning has been considered fundamental to tackle the complex, exploratory, insight discovery and knowledge building problems that the field aims to address. A main stream of research in decision making with visual interfaces has covered the bulk of experimental psychology studies that have established the bottlenecks in the human abilities to perceive, attend, and process information (Cleveland & McGill, 1984; Fekete et al., 2008; B. Shneiderman, 1994; Ware, 2008). The goal is to understand and make use of the human visual system broad bandwidth to inform the design of data representations and interaction techniques that allow deep questions and reflection in the process to derive insight. However, data representations cannot be taken for granted, analytical reasoning is central to the task of applying human judgments to reach conclusions from a combination of evidence and assumptions. For this reason, a second stream of research efforts are concerned with advancing our understanding of cognition in decision making, particularly in the context of expert

analytics thinking under uncertainty (Richard Arias-Hernandez et al., 2012; Endert et al., 2014; Fisher et al., 2011; Ribarsky et al., 2009).

VA tasks have been regularly defined by practitioners as exploratory or ill-defined in nature. The problems that interest the community, often pose questions that are unknown a priori and for which training data is not available: “*tasks are strategic in nature and must be translated into operational questions during the course of the analysis*” (Endert et al., 2014). For this reason, VA research has found to be a fit to investigate the design of interactive data interfaces to support decision-making and data reasoning, in a diversity of fields. In the literature, VA research has documented the work with experts as diverse as intelligence analysts (Youn-ah Kang & Stasko, n.d.; Kretz, 2018); healthcare stakeholders and provision of care specialists (Chetta et al., 2015; B. Shneiderman et al., 2013); experts in genetics research (Nielsen et al., 2009; Parsons et al., 2015); or scientists in molecular biology (Gehlenborg et al., 2010; Livengood et al., 2011; Price et al., 2017), amongst other domains. I

In healthcare informatics, a domain characterized by a diversity of systems landscape beyond the clinical settings, the analytic interest is typically tilted towards the scientific analysis of data, image processing, and the improvement of longitudinal electronic health records (EHR). There is little research on the specific niche of supporting expert analytics for personalized health results with biomolecular data. That is, the visual analytics and scientific visualization research with *omics* data is focused on expert interpretation for cohorts of data (scientific visualization), but little to none research has been done in the use interactive visualization tools to support the emergent expert analyst-to-patient communication for the understanding of consumer-oriented results. A close effort made in this direction is the work by Parson’s et al, about the design of a tool for the interpretation of molecular cytogenomic findings (Parsons et al., 2015). This work aligns with this dissertation in the study of tasks that experts perform for the interpretation of large amounts of molecular data (in their case geneticists and cytogeneticists, in both research and clinical settings). The authors describe the analytic goal of their user group, as “*interpreting the findings from patient analysis to determine their clinical significance*” (p.1), which is the same overarching activity of the participants in this study. Their focus, however, is on the design of a visualization tool for mining the scientific literature to help the researchers with the interpretation process. In chapter 04 we will review that this task is also part of the process documented in this dissertation

but differs from the main goal, which is to create a consumer-facing summary of results and a set of lifestyle action recommendations. As suggested earlier, this is a close research effort from a Visual analytics perspective in the domain of personalized health, but the specific goal, settings of the user group, and data diverge.

This thesis is motivated to adopt Visual Analytics methodologies and techniques not only to deal with information overload but also to help analysts to address wicked and ill-structured health problems using biomolecular data. Ill-defined health problems cannot be solved by humans alone or by computers independently. Especially in the provision of care, a domain where there is traditionally a strong human component related to the communication of health outcomes. Therefore, the goal is to integrate Visual Analytics tool and techniques to bridge the gap and synthesizes the strengths of both humans and computers to efficiently and effectively integrate domain knowledge to address the interpretation of biomolecular profiles of data and derive personalized lifestyle interventions in a timely fashion. With this in mind, in this research I look at the factors that influence the decision-making process of biomolecular experts and that extend into the communication of outcomes for a patient.

Furthermore, another pressing priority for the VA community the issue of problem solving that can be audited and traceable: *“even though we implement more powerful tools for automated data analysis, we still face the problem of understanding and analysing our analyses”* (Keim et al., 2010). Something that was earlier described as the *analytical provenance* in the original Thomas & Cook’s VA agenda. Given the relevance of warrants and provenance in the creation of health interventions and the communication of health evidence, in this research program we are also interested in documenting mechanisms that analysts of the data use to trace and backup their analysis process.

2.2. Theory: The Sensemaking Loop of Intelligence Analysis

Nearly every type of analysis is a goal-oriented activity requiring the processing and manipulation of information. Analysis often involves collecting a number of data points, sorting through them, forming hypotheses, performing calculations and drawing conclusions. “Sensemaking” is described as the process of searching for a

representation and encoding data in that representation to answer task-specific questions (Pirolli & Card, 2005; Wade, 2011)

One of the reference models of sensemaking for the Visual Analytics community is the sensemaking loop for intelligent analysis by Pirolli and Card (Pirolli & Card, 2005). In this model (See figure 2.1), two major loops characterize the main tasks that intelligence analysts perform: (1) information retrieval (marked as the foraging loop), refers to gathering and sorting information, and is often most prevalent during early stages of analysis. (2) interpreting information, or sensemaking, represents the higher order cognitive functions such as hypothesis generation and matching information to a schema.

The general overview of tasks the model consists of information gathering, re-representation of the information in a schema that aids analysis, the development of insight through the manipulation of this representation, and the creation of some knowledge product or direct action based on the insights.



The sensemaking loop takes the development of schemas iteratively testing new hypotheses to make best fit with the evidence. The work suggests that analysts invoke an opportunistic mix of top-down processes (from theory to data) and bottom-up processes (from data to theory) to approach the foraging and sensemaking tasks.

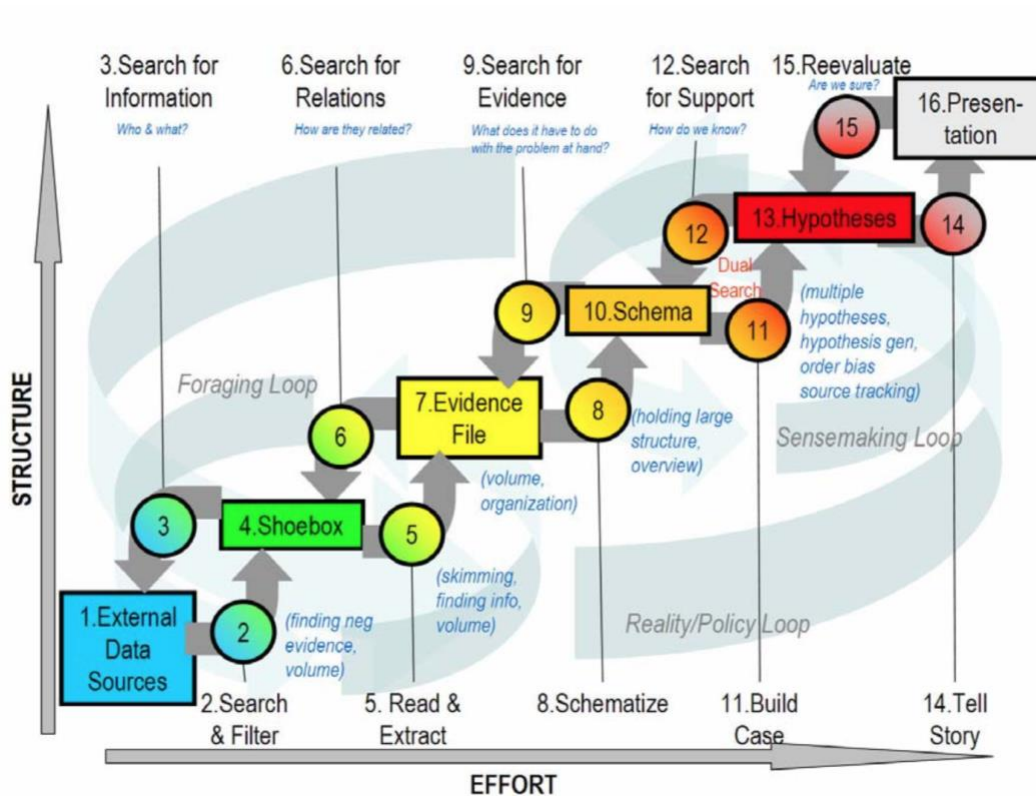


Figure 2.1 The sensemaking loop for intelligence analysis (Pirolli & Card, 2005)

We are interested in this model as because of the constructs to observe evidence-based decisions by experts. Even if experts in different domains vary in the specific goal and data sources they use in their processes, the sensemaking loop has proven to be useful in the VA community to characterize higher-order cognitive functions that take place for experts looking to approach evidence-based decision making in a structured fashion.

The model builds up on work that emphasizes the role of schematic knowledge structures built from expertise and experience (Klein, 2008). One of the best-established phenomenon in expertise from a psychological perspective is that *expertise involves large and more integrated cognitive units*, and that *expertise involves functional, abstracted representations of presented information* (Anders Ericsson et al., 2006). The research for these claims builds on the perceptual chunking experiments and find out that with increased experience and practice, people organize the perceptually available information into larger units. Research also shows that experts acquire skills to develop complex representations that allow them immediate and integrated access to information and knowledge relevant to the demands of action in current situations and tasks. In

consequence, the expectation is that experts will have built up from extensive experience a set of patterns around the important elements of their tasks. These sets of patterns are part of what Pirolli and Card name “*schemas*”.

The sensemaking loop for intelligence analysis not only is a seminal work for the Visual Analytics community, but as we will see, it can be viewed in concert with the research models on Evidence-Based clinical decision making. Together create a battery of constructs that can help us describe the process of experts dealing with biomolecular and other health data. In the next sections of this chapter, I review the literature on the use of evidence in clinical sensemaking and dig into the literature of expertise from a psychological perspective.

2.3. Theory: Evidence-Based Medicine

Foraging information and searching evidence are primary tasks to analytic endeavors. In this section I survey the literature on what constitutes evidence in clinical decision making.

The concept of evidence is central to epistemology and the philosophy of medical sciences. The standard account for knowledge in analytic philosophy is that *knowledge is justified true belief* – and is common to distinguish among different kinds of knowledge such as *the knowledge-that* or the *knowledge-how*, that is, propositional knowledge such as knowing a fact and having good reason to believe that is correct, or the know-how, the knowledge from experience an intuition that cannot be reduced to proposition (Fantl, 2017). This distinction has been present in the philosophy of medical sciences for decades and is especially important in the discussion of *evidence-based medicine* (EBM), the movement that seeks to increase the use of high-quality evidence, or best evidence, in making decisions about the care of individual patients. The revised and improved definition of evidence-based medicine is “*a systematic approach to clinical problem solving which allows the integration of the best available research evidence with clinical expertise and patient values*” (Ashcroft, 2005; Masic et al., 2008).

As John Worrall introduces, a typical reaction from anyone who is not a scholar of epistemology in medicine sciences would ask themselves, if not based on the best evidence, what was medicine based on before? (Worrall, 2002). For Worrall, there are

two main areas in which EBM debated the view of evidence that it recommends: the first concerns the role and evidential power of randomization; the second concerns the role and evidential power of clinical judgment and expertise (the know-how of clinicians). While it is perhaps more difficult to evaluate knowledge claims such as “this patient has this illness, and this treatment will be the most beneficial under these circumstances”, clinical judgment is within the scope of EBM’s techniques experimentally and analytically (Ashcroft, 2005). On the other hand, despite the common misconception, EBM is not restricted to randomized trials, the literature reveals that what accounts for evidence in EBM involves tracking down the best external evidence to answer clinical questions and distinguishes whether is in the interest of studies of causation or for therapeutic evidence. Randomize experimentation is best suited to study therapeutic effects since non-experimental approaches more often lead to false-positive effects about efficacy; but even some questions about therapy cannot be tested with randomized trials or cannot wait for the trials to be conducted (for example interventions for fatal conditions). *“If no randomized trial has been carried out for our patient’s predicament, we must follow the trail to the next best external evidence and work from there”* – cites Worrall (2002).

Moreover, according to Masic et al., one of the greatest achievements of evidence-based medicine has been the development of systematic reviews and meta-analyses, methods by which researchers identify multiple studies on a topic, separate the best ones and then critically analyze them to come up with a summary of the best available evidence (Masic et al., 2008). For questions about prognosis, researchers follow up studies of patients from early points in the clinical course of the disease. But as the basic science technologies move forward to provide more timely evidence, genetics, immunology, and the assessment of molecular components in the body become valid measures in the study of a disease. (D. Kumar, 2007; Worrall, 2002) This implies the incorporation of personalized medicine methods as a category of evidence in EBM.

While EBM concepts and models evolve with the adoption of bioinformatics tools and biomolecular technologies, the models and practice of medicine have also evolved towards the more inclusive, patient-based process that accounts for their preferences and actions. From that perspective, evidence from research can be no more than one component of any decision. Other key components are the circumstances of the patient (assessed through self-reports and the expertise of the clinician), and the very prevalent, preferences of the patient (Haynes, 2002). The circumstances set is meant to include all

additional considerations of the clinical situation that may prevent the direct application of evidence to particular patients. How to combine the different pieces of evidence to derive the optimal decision has not been clearly stated, but it is widely accepted that “*clinical knowledge and expertise*” are essential to success as a complement to the research evidence (Haynes, 2002). A similar position for visual analytics researchers in the healthcare domain states that “*evidence is fundamentally information that takes various forms—i.e., artifacts, mental structures, or communication processes*” and that the issue of decision making becomes a matter of understanding the information flow and creation of knowledge in a distributed cognitive system. That is, the role of expertise goes beyond the mere access to ready-resources (Sedig et al., 2015).

Figure 2.2 reproduced from Haynes’ work is a guide for thinking about how decisions are made with patient’s preferences in mind. In this model, clinical expertise is the central force that glues together activities such as diagnosis, prognosis, effective patient communication, the correct performance of a treatment or test, and the application of population-based evidence to particular individual patients, a goal that often involves the sorting of trade-offs. (Wieten, 2018).

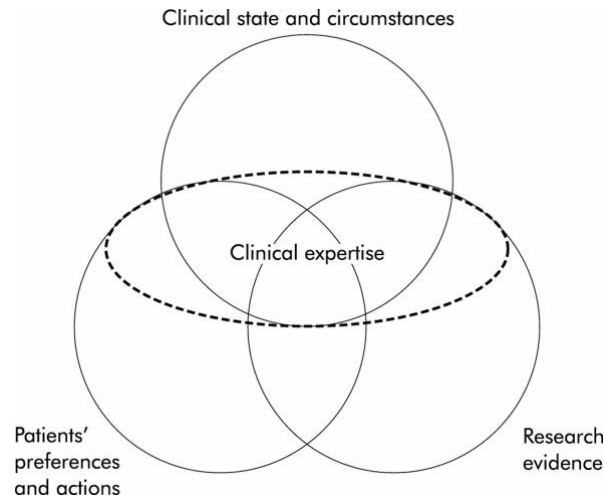


Figure 2.2 A model of evidence-based clinical decisions with evidence, circumstances, and preferences in the decision-making process (Haynes, 2002).

Haynes’s model is the third approach on the evolution of evidence within the development of EBM. It is important for the account of expert judgement as evidence in the creation of their practices. In the early days of the EBM movement, expertise was

taken to be exactly what it was rebelling against —the authoritarian pronouncements about “best” interventions learned in medical schools. But since then, the community has reworked the idea and role of expertise in the equation. The expert knowledge in EBM can be equated to “experience, expert opinion, clinical skills, expert judgement” and they all refer to the knowledge gained by subjects in the course of clinical interactions, in contrast with knowledge gained from sources such as journal articles reporting on the findings of RCTs, or meta-analyses and systematic reviews (Wieten, 2018).

A central discussion in EBM about “*expert knowledge*” is whether it can be considered a type of evidence or otherwise something external to the evidence but relevant in the process of evaluating it, like bringing together the patient’s preferences and the research evidence for a diagnosis, or even the effective communication with patients. The discussion is however alive because the original models presented the expert opinion as a type of evidence at the bottom of a bottom-up pyramid-model, that developed evidence from low warrant-quality “expert opinions” to the warranted sources of evidence such as amalgamations of RCTs from meta-analysis and systematic reviews (see more in Wieten, 2018). But however confusing is classifying expert opinion as a type of evidence, clinical expertise does constitute a kind of evidence in some ways to the community, although for the most part expertise is the judgment necessary for the interpretation of all evidence, whether that evidence is high or low quality.

Under optimal conditions, decision makers become translators, with a cognitive apparatus that works like a choice-selecting mechanism. But aligned with the sensemaking model of intelligence analysts, cognition has a more significant role when decisions are made with limited information, under the pressure of time, when uncertainties are incalculable, and there is no universally correct answer. In such cases, medical decision making is more about problem-solving than deliberation and choice. Expert decision makers frequently are called upon to engage in original thinking; at these times, they are generating options as well as selecting from them (Falzer, 2004).

In consequence, one of the major hurdles is faced by clinicians on daily basis is selecting the best available evidence. It has been widely recognized that the clinical staff cannot be expected to undertake this evaluation themselves prior to undertaking clinical decisions across a busy practice (D. Kumar, 2007). Hence, pressing matters in the assessment of evidence include the accurate and efficient communication with

practitioners as much with as patients(Haynes, 2002). And this issues of translation between the research and clinical practice communities go beyond the challenges for an individual organization, they represent central debates in the international Personalized Medicine gatherings such as *Personalized Medicine Initiative Summit* held in Vancouver in 2017. During our participation in the summit, I was able to better understand that the translation from the laboratories to clinical practice is also an issue about the communication of *two tribes*³, scientists and clinicians, because they approach their practice with different world views.

2.4. Theory: The Doctor-Patient Relationship

Let us start recalling that personalized health DTC services, like the one studied in this dissertation, do not address *patients* because the service is described as *not diagnostic* and can be provided to health consumers that are not convalescent of any disease. In this dissertation I study the voice and analytic work of expert analysts creating a data interpretation and a list of therapeutic lifestyle actions that clients can take to have an impact on any foreseeable health risk from the results of the biomolecular data. As a result of the analysis I will present in chapters 04 and 05, I found that the process and product of the analysts in this situation, is highly shaped not only by the biomolecular information, but by the goals and interests of the client they address. From that perspective it was worth reviewing the situation as that of a participatory activity where both analysts and health consumers contribute to a common goal. I summarize the literature on the doctor-patient relationship as the foundation on how to understand the provider-consumer relationship in clinical settings, to find commonalities and important differences when taken out of the clinical realm and into the preventive, wellness sphere of healthcare.

The doctor – patient relationship study describes an evolving association that remained a paternalistic model between a patient seeking help and a doctor whose decisions were silently complied with by the patient, for centuries (Kaba & Sooriakumaran, 2007). Three basic models describe the evolution of the relationship as

³ The Two tribes speaking different languages, scientist and clinicians, was a metaphor used during the summit by Sir Malcom Grant, former Chairman of NHS England, as he described the national initiatives and challenges in personalized medicine in England.

seen by Szasz and Hollender (1956) in their seminal contribution to the philosophy of medicine, and that demarked a common lingo to describe this relation: (1) the active-passive model, (2) the guidance-co-operation model and (3) the mutual participation model. *The active-passive model* is the oldest and perhaps the longest sustained through history. It is based on the physician acting on the patient, who is treated as an inanimate object. Despite this may not be a current model of general values, it is valid and appropriate for specific situations, for example emergency rooms (Chipidza et al., 2015). In *the guidance-cooperation model*, a doctor is seen as the one having medical knowledge that the patient lacks. The doctor is expected to decide what is in the patient's best interest and to make recommendations accordingly from a higher power position. It is a model that also remains appropriate for specific situations. However, over the last several decades there has been increasing support for the mutual participation model when medically feasible. This is how Kaba et al., describe *the mutual participation model*:

“The model of mutual participation is based on the belief that equality amongst human beings is mutually advantageous. In this model the doctor does not confess to know exactly what is best for the patient [...] For the concept of mutual participation between the doctor and patient to exist, it is important that the interaction between them is based on having equal power, mutual independence, and equal satisfaction. This ultimately allows the patients to take care of themselves. The management of chronic disease provides a good example. This model therefore provides the patient with a greater degree of responsibility and is characterized by a high degree of empathy and has elements often associated with friendship and partnership, as well as the imparting of expert medical advice” (Kaba & Sooriakumaran, 2007).

Let us highlight that in this model the role of the physician is to *help patients to help themselves* and that the patient participates in partnership with a physician who brings expertise to discuss evidence and collaborate with the patient in the understanding of evidence about disease and therapies. Kaba et al., describe it as a more mature exemplar of relationship. One that emulates an adult-to-adult relation with no drastic power imbalance. In clinical applications, the model can be seen in action with most chronic illness management or psychotherapy. Each model influence on the content and style of the relationship between doctor and patient, where relationship is defined as “*neither structure nor function but rather an abstraction embodying the activities of two interacting systems (persons)*” (Szasz & Hollender, 1956)”

This model conveys the values of preventive and participatory care. Something also aligned with the rise of the patient-centered model to medical practice that, as it becomes the predominant model, it changes structural normative models in the provision of care. Changes from what constitutes evidence as we discussed in the previous section, and changes to role dynamics and participation.

The patient-centered approach has been described as one where “*the physician tries to enter the patient's world, to see the illness through the patient's eyes*” (McWhinney, 1989). And it is for this reason of seeing illness through someone else's experience and current circumstances, that Kleinman et al., justify clinical science to be thought of as both a biomedical and a social science. For the authors, the biological malfunctioning of a specific process denotes what we understand as disease, but it deserves a distinction with the experience of illness, “*the personal, interpersonal, and cultural reactions to disease or discomfort*” (Kleinman et al., 1978).

According with Mead and Bower (2000), the development of patient-centred medicine is the result of the perceived limitations in the conventional way of doing medicine often labelled as the *biomedical model*. In the biomedical model patient's illness is reduced to a set of signs and symptoms which are investigated and interpreted within a positivist biomedical framework (Mead & Bower, 2000). To these authors, the most comprehensive model on patient-centeredness was described by (Stewart et al., 2013) whose model of the patient-centred clinical method identifies six overlapping components: (1) exploring both the disease and the illness experience; (2) understanding the whole person; (3) finding common ground regarding management; (4) incorporating prevention and health promotion; (5) enhancing the doctor–patient relationship; (6) “*being realistic*” about personal limitations and issues such as the availability of time and resources (2000).

Mead and Bower extend the doctor-patient relationship component stating that that patient-centred medicine differs from the exclusive biomedical model in terms of five key dimensions, each representing a particular aspect of the relationship between doctor and patient: (1) *the biopsychosocial perspective*, which we have described before as broadening the explanatory perspective on illness to include social and psychological factors, and that extends the scope of medicine beyond the seemingly realm of ‘healthy’ bodies. (2) the “*patient as a person*” regards the patient as an experiencing individual

rather than the object of some disease or entity. In consequence, a full understanding of the patient's experience of illness depends on her particular "*biography*", a recollection of the patient's history and circumstances. (3) "*Sharing power and responsibility*" as a dimension suggests the shift in the doctor-patient relationship from the "guidance-cooperation" model to "mutual participation". In clinical consultation, this translates into behaviours such as encouraging the patient to voice ideas, listening, reflecting, and offering collaboration. According to Mead and Bower, related notions like "user involvement", "negotiation", "concordance" and "patient empowerment" have been implemented in health policy since the 80s. Shared power and responsibility studies have reported positive associations with health outcomes (Kaplan et al., 1989), and suggest that information enables patients to take greater responsibility for their health (Grol et al., 1990).

Regarding the fourth dimension in their proposal, (4) *the therapeutic alliance* proposes that patient-centred medicine ensures a far greater priority to the personal relationship between the doctor and patient, if based on therapist attitudes of empathy, congruence and unconditional positive care. "*A common understanding of the goals and requirements of treatment is crucial to any therapy, whether physical or psychological*" (Mead & Bower, 2000). Something that will also impact the likelihood of adherence to treatment.

The final dimension for Mead and Bower concerns the influence of the personal qualities of the doctor: (5) the "*doctor as a person*". For Balint et al. (1993), the biomedical model is a "*one-person medicine*" in that a satisfactory clinical description does not require consideration of the doctor. That is, there is no theoretical reason why well-trained doctors should not be essentially interchangeable since doctor subjectivity does not impact on diagnosis and treatment. By contrast, patient-centred medicine is "*two-person medicine*", in which the doctor is an integral aspect of any description: "*the doctor and patient are influencing each other all the time and cannot be considered separately*". So, this dimension highlights the affective relationship as it develops between the parties.

2.5. Theory: The Role of Expertise and Task Characteristics

Experts certainly know more, but they also know differently. – Feltovich, Priteula & Ericsson (Anders Ericsson et al., 2006)

As we have seen, understanding the warrants of expert judgements is a topic of interest for any different analytic endeavors, as in clinical decisions or intelligence analysis. Whenever the subject of an expert intuition is discussed, there is always the question of how we can trust on the validity of an individual's judgement or how to identify if such are the results of overconfident claims. In this section I survey the psychology literature of expertise in an effort to better understand how researchers approach the issue of understanding what constitutes true expertise skill as opposed to overconfident subjective experience?

Studies of Expertise

An evident requirement on the study of expert performance is to define who is an expert. Following Shanteau's suggestion to let those in a domain define an expert, in this research I will use his definition: "*experts are operationally defined as those who have been recognized within their profession as having the necessary skills and abilities to perform at the highest level*" (Shanteau, 1992). In this dissertation we will be following the work of 5 different experts in their own unique niches.

Studying the reasoning process of experts is a subject that interests research in psychology, different decision sciences, and as I introduced before, a topic that remains central to the visual analytics research agenda. The characteristics of true expertise can be often described as heuristic reasoning, a strategy to judge a situation or problem with incomplete information available. Despite this is an efficient and common procedure, it can also be the source of systematic errors, or biases. "*Experts are not immune to the cognitive illusions that affect other people*" (Kahneman, 2011).

We are interested in identifying expert intuitions in an analytics process, and to characterize what we can better support with the use of Visual Analytics tools, including the role of data visualization in the process and when and how expert intuitions are likely to introduce biased judgements in this process. To this end, multiple streams of research

can agree that Expert intuitions, as defined by Herbert Simon and later introduced by Nobel laureate, psychologist Daniel Kahneman, are based on recognition:

“The situation has provided a cue; this cue has given the expert access to information stored in memory, and the information provides the answer. Intuition is nothing more and nothing less than recognition.” (H. Simon in Kahneman 2011, page 237).

There is an extensive body of research on what constitutes expertise. From a psychological perspective, the theoretical interest in expertise and expert performance is based on the assumptions that there are shared psychological constraints on the structure and acquisition of expert performance across different domains, and the evidence suggests the necessary integration of automated basic strokes by experts (Anders Ericsson et al., 2006). These findings suggest that the character of cognitive operations changes with adaptation and practice of experts in their domains. In other words, automaticity of fundamental encoding and decoding of relevant input as well as the ability to access knowledge in relevant situations, are fundamental skills of experts but they also are what can potentially be modeled as reasoning pitfalls and cognitive biases.

In an effort to characterize mechanisms and aspects of expertise that generalize across domains Feltovich & Ericsson (2006, p. Chapter 04) survey the psychology research to report on 8 general characteristics of what constitutes expertise:

1. Expertise is limited in its scope and elite performance not transferable.
2. Knowledge and content matter are important to expertise.
3. Expertise involves large and more integrated cognitive units, and
4. Expertise involves functional, abstracted representations of presented information.
5. Expertise involves automated basic strokes.
6. Expertise involves selective access to relevant information.
7. Expertise involves reflection.
8. Expertise is adaptation.

Expertise is limited in its scope implies that there is little transfer from high-level proficiency in one domain to another, even when the domains seem intuitively similar.

According to Shanteau (Shanteau 1992) some professionals exhibit genuine expertise in some of their activities but not in others. Kahneman and Klein named this as “*fractionated expertise*” and they believe this is the rule, not the exception (Kahneman & Klein, 2009). Regarding *knowledge and content*, the authors are rejecting a classical view on human cognition in which abilities such as learning, reasoning, problem solving, and concept formation were understood as capacities that can be studied independently of the content domains. They describe it as the growing recognition that knowledge-based features of perception and cognition such as perceptual chunking, knowledge organization, knowledge differentiation, and effective perceptual-knowledge coupling, play an important role in expertise.

One of the best-established phenomena in expertise from a psychological perspective is that expertise *involves large and more integrated cognitive units*, and that *expertise involves functional, abstracted representations of presented information*. Feltovich et al., present the extensive research built on the perceptual chunking experiments to conclude that with increased experience and practice, people organize the perceptually available information into larger units. Experts recognize about the same number of units, but the chunk size is larger. In these areas, they say, research also shows that experts acquire skills to develop complex representations that allow them immediate and integrated access to information and knowledge relevant to the demands of action in current situations and tasks. Chase and Simon (Chase & Simon, 1973) performed more of their well-known chess players experiments and found out that when given the task to reconstruct a chessboard, professional players perform with more precision and higher recall, by being able to work with chunks of patterns associated to moves. Instead, novices deal with the reconstruction of a board in piece-by-piece matter.

The issues of *automated basic strokes and selective access to relevant information* represent rather controversial characteristics in the study of expertise because they are perhaps processes that easily translate as overconfident judgement calls from a heuristics & biases perspective. Feltovich et al., include the recognition of operations that become fast, less deliberate, and that can run in parallel with other processes as one of the generalizable characteristics of expertise. According to the authors, research shows that a major limitation in novices is their ability to access knowledge in relevant situations and that a key constraint for skilled encoding is that

experts are able to anticipate potential future contexts where the encountered information might become relevant.

Finally, the last two generalizations *expertise involves reflection*, and *expertise is adaptation* describe the metacognitive ability to test an individual's own understanding and partial solutions to a problem. There is a common belief that “*true experts know when they don't know*” which more than a cultural expectation reflects on the manifestation of metacognitive skills. Experts who claim an intuition are aware of the limits of their knowledge, and the ability to recognize that a situation is anomalous and poses a challenge is one of the manifestations of authentic expertise according to the Kahneman and Klein (2009). From these works, the advancing the argument is that “*experts restructure, reorganize, and refine their representation of knowledge and procedures for effective application to their work-a-day environments. Experts certainly know more, but they also know differently.*” (Anders Ericsson et al. 2006, page 57).

Expertise is appropriately viewed not as simple matter of fact acquisition but as a complex construct of adaptations that include self-monitoring and control mechanisms to make use of the environment in service of the tasks and goals.

Tasks Characteristics and the Expert's Environment

Findings in which the performance of human judges is inferior to that of simple algorithms are often cited as evidence of cognitive ineptitude, but this conclusion is unwarranted. The correct conclusion is that people perform significantly more poorly than algorithms in low-validity environments. – Kahneman and Klein 2009

Taking a shift towards the role of task characteristics and the conditions of the environment in expert performance, James Shanteau argued that the two contrasting views: (1) the judgement and decision research showing that experts make flawed decisions due to biasing effects of using heuristics, and (2) the cognitive science research that views experts as competent and different from novices, are both correct but incomplete (Shanteau, 1992). Shanteau's research interest in the competence of experts led him to systematically look at what factors drive experts to do well and what factors lead them to do poorly. He describes five key factors that play a role in the emerging of expert-level skills and abilities: *domain knowledge, psychological traits, cognitive skills, decision strategies, and task characteristics*. While those that require to

look at the psychology and cognitive strategies of the decision maker overlap with characteristics described in the previous chapter, in this section I will focus on the often-overlooked role of the characteristics of the task at hand. Looking at task characteristics can shed light on whether it is possible for experts to behave competently or not even when they have developed genuine expertise-intuitive skills.

According to Shanteau’s work, research shows that experts in fields such as weather forecasters, livestock judges, astronomers, test pilots, chess masters, physicists or mathematicians, have consistently being reported as having exceled competent performance; and that experts in domains such as clinical psychologists, psychiatrists, court judges, behavioral researchers, counselors, intelligence analysts or stockbrokers more often exhibit poor performance in research studies of their tasks (Shanteau, 1992).

In some domains, experts such as nurses, physicians, and auditors, have been described to perform both, competently and poorly. The differences in those cases could be attributed to the different tasks selected for the different studies. The tasks characteristics that can be associated with exceling competence by experts in these different domains are highlighted in table 2.1

Table 2.1 Task characteristics associated with good and poor performance in experts. From (Shanteau, 1992).

Good Performance	Poor Performance
Static stimuli Decisions about things Experts agree on stimuli More predictable problems Some errors expected Repetitive tasks Feedback available Objective analysis available Problem decomposable Decision aids common	Dynamic (changeable) stimuli Decisions about behaviour Experts disagree on stimuli Less predictable problems Few errors expected Unique tasks Feedback unavailable Subjective analysis only Problem not decomposable Decision aids rare

Shanteau’s take on these findings of the literature was, that we observe competent performance by experts executing tasks that involve static objects or things, where the experts are asked to make decisions or evaluate about stimuli relatively constant. In contrast, poor performance was observed in cases where the stimuli are

dynamic and often involve human behavior. “*Experts in this case are asked to decide about what is in effect a moving target*”. (Shanteau 1992). Other insights captured in the analysis included the role of predictability and having an objective analysis available: human behavior is inherently less predictable than physical stimuli, says the author. More differences are attributed to how much similar conditions appear from time to time and make the task repetitive while also having the opportunity to receive and respond to feedback, in other words, the chances to learn from past decisions.

Another insightful observation is that of historical dynamism of the different domains, most domains listed as “good performance” started as “poor performance” domains. Take for example weather forecasters who moved from relying on intuitions and guesswork to using detailed climatic information as scientific understanding of meteorology developed and decision-aids were placed. Moreover, studying the competent performance without looking at decision-making aids is a faulty method. “*An unaided expert may be an oxymoron since competent experts will adopt whatever aids are needed to assist their decision making.*”(Edwards and von Winterfeldt 1986).

In a similar fashion Kahneman and Klein found that the characteristics of the environment are what results common in their often seen as contradictory, approaches to intuitive judgement(Kahneman & Klein, 2009). In an effort to reconcile what the two traditions can accept to be conditions that separate true intuitive skill from overconfident and biased impressions, Kahneman and Klein conclude that subjective experience is not a reliable indicator of accuracy and they reconcile their approaches under a recognition model proposed by cognitive scientist H. Simon: “*The situation has provided a cue; this cue has given the expert access to information stored in memory, and the information provides the answer. Intuition is nothing more and nothing less than recognition.*” (Kahneman 2011, page 237).

Kahneman and Klein suggest that the recognition model implies two conditions that need to be satisfied to make an intuitive judgement the product of genuine expert skill: (1) *the environment must provide adequately valid cues to the nature of the situation*, which implies that skilled intuitions will only develop in an environment of sufficient regularity providing such cues to the situation. (2) *People must have an opportunity to learn the relevant cues*. The conclusion of the Kahneman & Klein debate is that developing intuitive expertise depends on the quality and speed of feedback, and

sufficient opportunity to practice (Kahneman & Klein, 2009). We notice how on this perspective, there is also a shift from the qualities of the decision-maker to the characteristics of the environment under which such expert operates. These pointers are important to the study of intuitive judgments in expert decision making because they allow us to evaluate validity by focusing on the provenance rather than by looking at the individual judgement instances.

2.6. Joint Action Theory in Visual Analytics Research

Language use and joint activities are inseparable, we cannot understand one without the other, Clark would argue in his seminal work of *Using Language* (Clark, 1996). This idea can be taken further as Levinson's proposal of a "*human interaction engine*" stating that humans did not evolve language to then get involved in a special kind of social life, but just the opposite, language must have evolved for something for which there was already a need: for communication in interaction. "*It is interactional intelligence that made language possible as a means of communication*" (Enfield & Levinson, 2006, p. 44). Levinson and Clark's works give us a framework for looking at interaction (activities) in which two or more individuals participate, by studying how do they construct meaning in their use of communicative acts.

Contributing to the pragmatic tradition of the study of language use, the main proposal in JAT is that language use is an instance of a broader category of human actions: joint actions. A joint action is one that is carried out by an ensemble of participants in coordination with each other (Clark, 1996). In Clark's theory, the coordination of actions happens at the level of content –what participants intend to do– or at the level of process –the physical and mental systems used in the execution of the intentions. Take for example the actions performed between a customer and a pharmacist at a drugstore, or two people competing in a chess match without exchanging spoken words. Both are examples of joint activities that require individual actions to be coordinated in order to move together the intentionality and commitment taken by entering the activity. I would like to highlight that not all joint activities have a conventional formal intention (take for example joining a gossip session in the corner of the room), nor they require verbal exchanges, like in the chess game. Clark's proposition is that language use arises in all these type joint activities.

In this sense, language use is more than the sum of a speaker speaking and a listener listening. It is the action that emerges in coordination of the participants, having each of them performing their individual actions. This occurs in different settings of language use including discourse in a spoken or written media. As a joint action, language must be seen within the complex context of intentions, beliefs, and assumptions in which is used.

For the Visual Analytics community and the studies of technology, Arias-Hernandez et al., extended the use of the framework to include the study of interacting with technology, specifically the role of visualization technologies in joint actions: “*since collaborative visual analysis is an instance of a joint action, the starting theoretical point to apply joint action theory to visual analytics was that: humans will use language and will work together to solve coordination problems in collaborative, visual analytics*” (Richard Arias-Hernandez et al., 2011).

In this dissertation, we are interested in the framework from two perspectives. In the first place, JAT has been the seminal theory used in the *Pair Analytics Methodology* to study the social and technological distribution of cognitive processes (reasoning, attention, awareness, learning) in collaborative analytical tasks involving visual interfaces. The methodology is based on the idea that information about some cognitive processes can be inferred by studying two (or more) people engaged in a joint analytical task. Using JAT, researchers can characterize a “collaborative analytic task” as the relevant phenomena of study. They do so analysing *pair analysis protocols*, a method designed for capturing reasoning processes using video and conversational data between two expert collaborators. Originally, pair analytics data was analyzed using conversational markers and tagging speech acts, following a *Clarkian* approach (Richard Arias-Hernandez et al., 2011, 2011; Kaastra & Fisher, 2014). From this perspective, JAT offers a theoretical framework to empirically investigate the social and cognitive aspects of collaborative analytics using visualization interfaces. Note that in studying reasoning in Visual Analytics settings, our research goal is not about understanding language as a joint action, but we are concerned with understanding interaction in the setting of a pair of analysts, a dataset, and a data analysis tool; we are set to study language used to coordinate problem solving in visual analysis activities.

The second approach is contributing to *design knowledge*, by looking at different instances of technology mediated discourse as joint actions, using the constructs of the theory. This means that the theory provides relevant constructs to characterize joint activities and dissect how the coordination devices used by members of the activity, particularly in mediated settings, are contributing to the progress of the public goal. For the research interest of this dissertation, we will take as example the joint activity between the expert analysts and their client readers via the written *health data summary* and the interactive report. I will address this approach in Chapter 05.

The starting point to apply JAT towards design knowledge in this situation is that according to Clark, it is possible for two individuals to participate in a joint activity asynchronously, with little to none knowledge of each other, and without being in the same room. Although the spoken setting is the richer, and most obvious setting of language use, the range of instances goes beyond conversation, and includes “reading a novel”, “putting on a play”, or “talking to oneself”. The settings of language use can be used to understand the creative variety of situations that are settings for joint activities, and language use. (1996 p.8). With this mind, in Chapter 05 I will use JAT to propose a characterization of the asynchronous joint activity between our experts and their clients.

Considering this, for the remaining of this section I summarize some relevant constructs of JAT including *Settings for language use*, *Joint Commitment*, *Coordination Devices* and the accumulation of *Common Ground*.

Settings of Language Use: Clark classifies the ranging different instances of language use according to the *scene* –where language use takes place, and the *medium* – the nature of the message whether is spoken, written, printed or mixed. The *setting* refers to both medium and scene of an instance. Face-to-Face discourse is the principal basic setting because (1) is universal to all human societies and (2) does not require special skills, you gain the skills by virtue of being born and raised as a human part of a society. This implies (3) face-to-face discourse is the basic setting for children's acquisition of language. Looking at the attributes of the conversational setting, we can find characteristics of other non-basic settings. Table 2.2 shows a list of the features of conversation listed in (Clark & Brennan, 1991). Feature 1 to 4 reflect the *immediacy* of face-to-face conversation. Features 5 to 7 characterize the face to face spoken *medium* and features 8 to 10 describe the *control* – who controls what gets done in the activity.

Other settings lack the immediacy, medium, or control of face-to-face conversation, so they require special techniques or practices.

Table 2.2 Features of face-to-face conversation. In (Clark & Brennan, 1991)

1	Copresence	Participants share the same physical environment
2	Visibility	Participants can see each other
3	Audibility	Participants can hear each other
4	Instantaneity	The participants perceive each other's actions at no perceptible delay
5	Evanescence	The medium fades quickly
6	Recordlessness	The participant's actions leave no record or artifact
7	Simultaneity	Participants can produce and receive at once and simultaneously
8	Extemporaneity	Participants formulate and execute their actions, without preparation, in real time. (Notice that this does not refer to the content but to the sequence of actions in the conversation)
9	Self-determination	Participants determine for themselves what actions to take when
10	Self-expression	Participants take actions as themselves (different to an actor speaking for the intentions of a character, or a politician presenting the voice of an office).

Common Ground (CG): In Clark's terms, the problem for the participants in a joint activity is to infer what individual actions they can expect from each other so that they can pursue the public goal of their joint activity. To solve the coordination problems, participants rely on what Clark called after Stalnaker the "*common ground*": the knowledge, beliefs and suppositions participants share about the activity, and that accumulate over the course of actions.

It is important to note that CG is not just what people know at the time of entering the interaction, is rather a set of basic mechanisms for coordinating in interaction. Instead of understanding CG as "a space" for participants to "share" the same knowledge, data and goals, we can refer to the CG as *a process of building and maintaining a mutual understanding* ((Monk, 2003)). This means that as joint activities advance, what they accumulate is *common ground* about the activity for all participants. Clark argues that for most activities, common ground at any moment divides into three parts:

- *Initial common ground*: the set of background facts, assumptions, and beliefs the participants assumed when they entered the activity. In a store transaction, for example, participants enter the activity with assumptions about the standard procedure in a culture for the exchange of money for goods, or knowledge about a specific type of store (a drugstore transaction may be

different to that of a grocery store). Participants also presuppose they both spoke English, that the purchaser has enough money, etc.

- *Current state of the activity*: External representations of the change of states convey the current state of a scene, for example taking the desired items to the counter for them to be scanned / charged. In a chess game, the chess board and its pieces are a precise representation of the current state of the activity, as it is the time elapsed. External representations are especially valuable as a medium for the actions, but signaling mechanisms and awareness of the other's understanding can also be taken into the current state of common ground.
- *Public events so far*: Clark argues that people keep a track of the actions that have made possible the activity, a record of actions taken from the beginning.

Coordination devices: The mechanisms for advancing and solving problems as the activity move forward. Coordination devices are a rationale for mutual expectations that make partners believe that they will converge on the same joint action. Participating individuals use the Coordination Devices as "*something to tell them which actions are expected*" (Clark, 1996, p. 91). The use of Coordination Devices constitutes an essential component in maintaining coordination and collaboration among individuals. Individuals need to be on "the same page" so as to ensure effective communication and coordination of actions (Sebanz et al., 2006). There are different types of devices:

- Explicit Agreements
- Conventions (including explicit agreements that could evolve into conventions if they are established as the agreed solution to a recurrent problem)
- Perceptual Saliency: manifest elements from the environment. It refers to situations in which the environment (or the available information) makes the next move apparent within the many moves that could conceivably be chosen
- Non-Conventional coordination devices like *Precedents*, norms and expectations developed within the on-going experience of the joint activity.

Chapter 3. Methodological Approach: Pair Analytics and In-Vivo Studies of Sensemaking

In this dissertation I set to explore the systematic process of experts that deal with interpreting biomolecular profiles of clouds of data to create health assessments and recommendations that go directly back to consumers. This new trend of health products put the consumers as the primary users of health data, even before they share it with their care teams. Moreover, the research goal was set to understand the human sensemaking process in the creation of actionable lifestyle recommendations from big amounts of data: to understand how experts make use of their acquired competence to make judgement calls and what mechanisms (if any) do they use to validate their own hypotheses and courses of action when evaluating data, working with incomplete data, and seeking evidence.

Whereas a study of genuine applied expertise or the identification of mechanisms that professionals use to counteract on pitfalls that they recognize about their thinking process, the case and methodological considerations proposed in this dissertation are relevant to advance research informed design to support an analytic discourse –the technology-mediated dialogue between an analyst and her information to produce a judgment about an issue, as it is defined by the VA community. (Thomas and Cook 2005, page 38).

3.1. Design Research and Studies of Cognition in Responsive Environments

The methodological commitments in this study consider the intersectional, interdisciplinary, and transdisciplinary practices that create knowledge in HCI, Design, and Visual analytics. We considered appropriate to review notions of *responsive (applied) design research* given our commitment to the study of cognition and tacit knowledge as distributed, situated processes. In general terms, we aim at developing knowledge about a community of practice to then apply this knowledge to episodes of individual activities, in an effort to reveal how cognitive activities are accomplished in real-world settings. In that regard, our process of inquiry borrows from the social sciences a commitment to the empirical approach along with the conviction that the

accounts of action, communication, and tacit expertise knowledge in real work settings, cannot be simulated in the lab. There are however, opportunities to bring the lab to environments of deep engagement in the application of design thinking, where the affiliation to cycles of innovation in technology allow for the introduction of artificial case-situations that are designed to capture the collaborative intricacies of the human-technology systems of thinking and problem solving; from this, a fundamental assumption in our methodological approach is that, in real world problems, information about some cognitive processes can be inferred by studying two (or more) people engaged in a joint analytical task.

This is made possible through the engagement of researchers in a partnering environment, and through observational methods that relay on digital media support like the video capture of work snippets. While this is not uncommon in the studies of technology from the HCI, or CSCW, it has been rare to develop these engagements with the industry-sector without it being about the design cycle of a specific end product. For the Visual Analytics community, this is evident in the creation and wide adoption of the *Design Studies Methodology* (Munzner, 2009; Sedlmair et al., 2012) where the engagement and the cycle of responsive research are created with the specific goal of documenting research in the application process of creating a custom visualization product.

As Perri 6 and Bellamy (2011) state, in observational research designs in the social sciences, “*the methodological purpose served by method is control*”, even if control in observational research is short compared to the experimental approach, we still try to exercise control via a carefully designed procedure with three main elements: (1) the categories of interest that delimit units of interest, (2) the means by which data is collected and made capable of analysis, and (3) the analytical procedures to perform comparisons, identification of patterns, or partitioning of interest.

With this in mind, the emergent and evolving, *Pair Analytics Research Methodology* is based on the idea that information about some cognitive processes can be inferred by studying the collaboration of people engaged in a joint analytical task along with an information interface. In its original conception, a typical pair analysis project will study how a visual analytic expert works with a subject matter expert to pose and solve analytical problems that engage interactive data visualizations. Most of the

studies are situated and involve a preliminary immersion within an organization to identify the analytical problems, the appropriate analysts, and their ecosystem of tools in place. The pair analytics methodology takes seriously the idea that two people collaborating at a single workstation are more effective than two people working separately; teaming them together provides a more natural way of making explicit and capturing their processes of reasoning. This provides a basis for going beyond tool evaluation to a deeper understanding of the underlying mechanisms of joint analytical work (Kaastra, 2015; Kaastra et al., 2012; Kaastra & Fisher, 2014)

The inference-warrant claims under the pair analytics methodology is supported by the use of a conceptual framework based in the work of Herbert H. Clark (1996), namely Joint Action Theory (JAT). JAT allows us to unpack and label multi-modal communication (e.g. word, inflection, gesture, gaze, and posture), multi-level communication (e.g. a word or gesture having more than one layer of meaning), joint awareness (i.e. of aspects of the task or environment), the state of the task, embedded activities, interpersonal roles, and personal, shared, or public goals.

Using the *Pair Analytics Research Methodology* as a departing framework for this research, the following is a synthesis of the methodological premises in the setup of the current study, that is, the adoptions, and the adaptations that we feel, allow us to draw meaningful inferences from the situation and the data.

3.2. In-Vivo Studies of Cognition: The Pair-Instruction-Analysis Method and Protocol Analysis

The original pair analytics method

Pair analytics was originally developed as a research method for capturing reasoning processes in visual analytics. As we have already discussed, the presumption is that in the study of how humans interact with abstract, visual representations of data, provides knowledge about how cognition works in visual analytics, and thus, poses implications for design and evaluation of visualization systems. Pair analytics builds on the tradition of “in-vivo” studies of cognition, or studies that collect ethnographic data of cognition in action. As a research method, its authors claim that the pair analysis method offers two advantages to other methods for capturing visual, analytical reasoning: (1) it is

a more natural way of making explicit and capturing reasoning processes, by pairing participants with different expertise and from different work environments, it reduces the chances of tacit knowledge not being verbalized, one of the limitations of in-vivo studies of cognition in social settings. The second benefit claimed is that (2) pair analytics makes evident certain cognitive processes used to structure and coordinate collaborative analysis. These kinds of processes cannot be captured with other individually-focused methods, since these are interactive processes and can only be captured in social, interactive settings (R. Arias-Hernandez et al., 2011).

In its original form, pair analytics requires a dyad of participants: one Subject Matter Expert (SME) and one Visual Analytics Expert (VAE). The pair are given one analytical task, a data set, and one computer with one or several visual analytics (VA) tool(s). The VAE has technical expertise in the operation of a suite of VA tools, but may lack the contextual knowledge that would be required to conduct meaningful analysis of the data set they are working on. The SME, on the other hand, has expertise in a specific analytic domain, but VA tools and their features may be unfamiliar to her. The pairing of SME and VAE is designed to generate a human-to-human dialog that will make explicit mental models and cognitive processes of SME and VAE during their visual analysis (R. Arias-Hernandez et al., 2011).

The Highly Integrative Basic and Responsive (HIBAR) Research it's a call to academia to improve research and the benefits to society through deepening the engagements with external partners. This research is characterized by (1) embracing the creation of knowledge coupled with the solution to of real world important problems, (2) combining the academic research methods with practical design thinking aimed at application, (3) partnering in efforts by academics and real-world experts, (4) develop cycles of research and production in a more direct-application timeframes (*The HIBAR Research Alliance*, 2020).

Successful pair analytics situations have been rooted in the deep engagement with private partners. The initial pilot studies and the development of the original pair protocol was conducted in the context of a partnership with Aeroinfo and Boeing Company, looking at aviation safety. It was the immersion in these organizations that allowed researchers, Wade and team, to explore the deployment of ethnographic methods, interviewing analysts in their workspaces, and collecting observations about

the data and the tasks that would then become suitable for a paired session (Wade, 2011). The immersion would grant researchers access to data repositories and a deep understanding of what the best task and data available would become the research case. As Wade describes in his manuscript, the internship role he took at Boeing allowed him to constantly refine and revisit the topics and specific questions for analysis, the selection of a task and scenario (from topics like pilot probe icing, runway excursions, volcanic ash, lightning, turbulence, kinetic energy of bird strikes, narrow body vs. wide body aircraft strikes and pilot response to bird strikes), and to engage in a dialog with the “subject matter experts” to establish a common understanding of the scope and the possibilities of taking upon the collaborative analysis with him acting as a navigator of the visual analytics tools.

The following deployment of the method, and a new iteration in what the methodology grants us in terms of inferences, was developed by Al-Hajj et al., in the context of the public health domain. The authors worked in a collaborative setting, bringing together multiple stakeholders looking at national injury prevention data. Similar to the Wade case, the immersion in the domain, required the researchers to delve into the data, and to take the opportunity to understand the tasks, identify stakeholders and appropriate scenarios (Al-Hajj et al., 2013)(Al-Hajj et al., 2017).

The pair analytics methodology serves as the framework to navigate the creation of knowledge in either the formation of new fundamental hypothesis worth testing in the lab or to broadening the understanding of the situatedness of cognition and tacit knowledge elicitation in applied domains. The creation of a visualization product and the reflection on design guidelines are not explicitly a piece of the methodology but of the overarching engagement with the reflective environment. In this dissertation, pair analytics is not concerned with the design of an artifact as a product but with delving deeper into the fundamental notion of human-reasoning in an analytics loop: Visual Analytics was born and developed on the foundation of crediting the creative ingenuity of experts, and with this methodology we are able to address research questions about what constitutes the expertise, eliciting the knowledge of experts, discerning the role of the automation algorithms, and as discussed in this dissertation, avoiding the pitfall of generalizing and reducing the human judgement to a black-box list of biases and cognitive pitfalls, to instead reframing the approach by looking at the characteristics of the task, the environment, and the situatedness of the problem.

Variation in context: the instructional interaction

While the pair analysis is a method that produces a rich amount of interactional data of language in use, the assumption of the use of a toolset and the characteristics of the datasets in different domains, required us to consider an adaptation for this dissertation. We adapted the protocol considering two characteristics of the task and environment of application: In the first place, the nature of the data and the toolset that analysts use in their work, did not fit the requirement for a VAE participant with knowledge of visual analytics tools. The situation required us to observe analysts working with their custom-built tools. On the second hand, the task of interest, although confirmatory and correlational in principle, had a very different driver: the outcomes are ought to be created for a specific non-expert audience sitting at the conclusion of the analysis; communication of outcomes, although prevalent and acknowledge as a core component in any analytics pipeline, hasn't taken the central stage in a pair analytics scenario so far.

With those characteristics in mind, we would expect to see different dynamics exhibited between the roles in the dyad. In the adapted instructional setting, the visual analytics expert will perform as the driver of the interfaces that the domain experts regularly use for their task, and we have used the term "training" rather than "collaborating" to introduce the protocol to the participants. From that perspective, we expected that the issue of interrupting a participant while thinking or verbalizing would feel more accepted and less intrusive. We invited the intern to enquire on the fly as needed, to ask questions to the expert like: "what are you thinking" or "why is something important" or "what are you looking at for that decision" as he saw fit during their session.

Instruction in the pairing format has been documented as a successful practice for on-boarding engineers using pair programming and related variants of training (Coman et al., 2008; Hulkko & Abrahamsson, 2005). Despite the difference in the nature of the collaboration, we expected to capture the rich communication data relying on the expected "social contract" when a domain expert solving a real problem and wearing the hat of instruction, engages to on-board a new analyst.

The researcher's role in an embedded capacity

Following the tradition of ethnographic approaches, the researcher is a key instrument in the methodology. Data creation, collection, and interpretation is on hands of the pair analytics researcher. In a traditional qualitative ethnographic approach, we are required to make explicit the identification of personal values, assumptions, and biases at the outset of the studies(Creswell, 2009). But more important, in the adoption of the qualitative inquiry set of values for HCI and Design research, the question of how designers gain knowledge of the nature of collaboration in work settings while often being also design practitioners, becomes a relevant issue. A wave of practices borrowed from the ethnographic tradition, especially after Lucy Suchman's work, on the idea of work practice as resource in the design of systems. This set the stage for the participation of sociologists (or HCI researchers in a sociologist hat) interested in the naturalistic study of interaction involved with computer technology (Button, 2000).

Thus, in the interest of making explicit the biases and assumptions at outset, we consider pertinent to clarify that the researcher and author of this manuscript, myself, participated as researcher, observer, design practitioner, and co-worker of the expert participants. This was a product of the in-depth engagements that are carried out via internships in-situ. I was involved with the organization for close to 3 years in different roles, starting with a MITACS accelerate internship as UX researcher and Data Visualization developer. Given the engagement from an early stage of the product design, I was part of a team of designers and developers that strategically designed the service and the data reporting platform. From a UX standpoint I designed and executed initial user testing with a cohort of initial consumers and with partner physicians. From a Visual Analytics perspective, during the initial stages of my involvement I developed the client-facing data visualizations but later switched to support an analytic capacity for scientists and researchers as richer cohorts of data became available for analysis. My rich understanding of the process the experts follow as well as understanding the bottlenecks come from my involvement with the organization. Similarly, identifying interesting research questions and potential gaps in the analytic process was a product of my involvement with the everyday works in the organization. I became interested in the issue of communicating health outcomes as I would often hear the scientists expressing their task of "Interpreting the data to create a story for the client's" with its different variations.

Additionally, similar to the Wade's situation with Boeing, my involvement as researcher at the partner organization had implications in the observer-participation during the data collection protocols. In Wade's case, his participation took the central role of "analyst" working side by side with the experts, turning him into the data wrangler and visualization tool navigator. In my case, my role as that of a designer and UX researcher put me in a position with the scientists as the one often eliciting requirements and understanding user's needs including their own in relation to analytic tools. This had a direct consequence at the time participating of the pair analysis protocols, I designed the situations to put myself as a passive participant observing the instructional situation occurring between the experts and a VA junior intern. However, the passivity of my role was rapidly broken as the experts treated each situation as a collaborative session in which they would direct their body posture, gaze and acknowledge my presence in the room similarly to other circumstances when I had interacted with them in different requirements elicitation process. This is transparently reflected on the transcripts of the data when often there is a "researcher" voice as a ratified participant of the situation.

Ericsson and Simon (Ericsson & Simon, 1993) discussed the negative influence of interrupting participants when they are in the verbalization process of their thinking. However, if we look at the situation of multiple people in a room, JAT provides good lenses to understand that in the negotiation of meaning, and within the instructional setting created for training our intern, the conversation and the expectation of feedback is natural to the situation. As a researcher I also took an active participation in the conversations with the experts. Depending on the individual personalities, some of them felt more inclined to truly engage and instruct both of us, intern and researcher, in their craft and knowledge, and similarly responded without conflict to the questions and interventions to either intern or researcher in the room.

Pair Analytics and other fieldwork approaches to studying sensemaking in work environments

The paired analytics protocol was developed through the work of several researchers who were interested in studying cognition in technologically enabled cognitive tasks. The most important distinction is that researchers carrying out these projects aimed to do more than evaluate the effectiveness of the tool. A primary defining characteristic for projects in Pair Analytics was to study the social and technological

distribution of cognitive processes like reasoning, attention, awareness, and learning in these tasks. This required methods for capturing not only the behaviours of participants, but aspects of their reasoning, attention, awareness, and learning (Kaastra, 2015).

However, other suitable research methodologies and observational field methods that researchers use to the study of socio-technical systems include *contextual Inquiry*, a method (Holtzblatt & Beyer, 2017), *distributed cognition (cognitive ethnography)* (Hollan et al., 2000; Hutchins, 1995), and even traditional *case studies* (Harrison et al., 2017).

Contextual inquiry is the fundamental interviewing method to implement contextual design. Contextual interviews are conducted with people within their working context. Four interaction principles are proposed to guide the interviews: partnership, focus, interpretation, and context. The principle of “partnership” states that the interviewer should try to establish a master–apprenticeship relationship with the interviewee, making it clear that they have inferior knowledge about the domain but a high interest in understanding it (Holtzblatt & Beyer, 2017). In that regard it compares to the pair-instruction-analysis dynamic I described in this chapter. Contextual inquiry interviews require a clear focus on the relevant units of analysis for the research question at hand, and although is a form of interview, it is designed to be performed while participants work on the completion of a task. Implementation of contextual inquiries has proven successful for the study of technology in workplace and the understanding of what encompasses “work” beyond a task, in the ecosystem of healthcare it is a method used to understand patients, providers, and technology design (Coble et al., 1995; Graan et al., 2016; S. Kumar et al., 2015). However designing relevant questions to inquiry about a participant task and domain can be difficult and requires research training (Berndt et al., 2015). Moreover, contextual inquiry is constrained by the limitations of an engineering project. Field interviews are restricted to a few hours, not days or weeks, and the interaction between interviewer and user is defined as a focused conversation.

Considering the similarities in the approaches, the main difference with implementing pair analysis protocols sits in the epistemological commitments of their traditions of origin. Contextual Design was designed for practical application with commercial design teams. In that regard contextual design has been tailored to designers and their teams (Holtzblatt & Beyer, 2020).

Case studies are another research strategy and empirical inquiry that investigates a phenomenon within its real-life context. The methodology relies on observational and multiple sources of data to gather an in-depth understanding of an individual, group or event (Pamela Baxter and Susan Jack, 2008). The selection of a case study for my case and the implementation of ethnomethods for data collection and analysis would reflect the constraints in the sample size and the work setting.

Robert Stake (1995) described instrumental case studies as those in which there is an interest in providing insight into a particular issue, redrawing generalizations, or building theory. The case can be instrumental in *understanding something else, the issue is dominant over the case* (ibid). In Stake's terms, the instrumental case study is used to accomplish something other than understanding a particular situation. The difference between an intrinsic (exploratory in nature) and instrumental case study is not the case but rather the purpose of the study(Grandy, 2010). On the other hand, intrinsic case studies (descriptive in nature) are set with propositions. Propositions are part of descriptive and explanatory case studies for Robert Ying. Ying's approach to case studies is primarily positivistic in perspective, hence his take on propositions is that theory should inform propositions, and propositions inform data collection and analysis. To apply case studies in this situation, we would follow Robert Stake's approach to case study as an instrumental case. Robert Stake (1995) takes an approach to case study research in a constructivist paradigm. Based on this viewpoint, the case develops in close collaboration between the researcher and the participant, enabling participants to tell their stories and describe their views of reality. This enables the researcher to better understand the participants' actions (Pamela Baxter and Susan Jack 2008).

However, in order to gather problem solving data with verbal protocols, we needed to introduce the pair analytics methods, which in turn, implied the creation of a synthetic situation out of everyday life for the analysts: the training of the intern while being recorded.

Finally, distributed cognition, like any other cognitive theory, seeks to understand the organization of cognitive systems. However, it extends the reach of what is considered cognitive beyond the individual to encompass interactions between people and with resources and materials in the environment (Hutchins, 1995). Distributed cognition has a special role to play in understanding interactions between people and

technologies, for its focus has always been on whole environments: what we really do in them and how we coordinate our activity in them (Hollan et al., 2000). However, the mappings of distributed cognition to the analysis of ethnographic data for design can be limited. Researchers have made efforts to create conceptual frameworks to map into the analysis of a situation, but the potential criticism of this work is the lack of method that takes them from analysis to design (Fields et al., 2000).

In this regard, rather than a method to contrast, DCOG is a theoretical framework to understand and analyze the situation. It can be a relevant perspective to analyse data from multiple methodological approaches like pair analysis and contextual inquiry interviews.

3.3. Bounding the Study

Setting

The setting for this study is a small bio-sciences organization that provides personalized health services directly to consumers. The business of this organization is to provide direct-to-consumer personalized health analysis by testing on multiple-omics profiles of data namely, genomics, proteomics, and metabolomics. The promise is to infer early signals of disease and wellness patterns to provide an individual with a personalized health plan. The purpose of the case in this research is to capture and describe the process of decision making, validation, and data visualization technologies that analysts with different professional backgrounds need in the process of (1) interpreting the biomolecular data profiles, and (2) creating personalized health recommendations based on those profiles. The case is narrowed to the interpretation process that analysts of the organization perform with the data. Currently, the process is heavily human-mediated and dependent on specific expertise.

Actors

Four different roles are involved in completing the core process of interest for this dissertation. They constitute the set of multidisciplinary expert participants. The different roles are: 1) Health Data Review Analysts, 2) Nutrition Analysts, 3) Physical Activity Analyst, and 4) Adjunct clinicians. These roles are selected given their involvement in

the process of analyzing the biomolecular data and they are known to me because of my in-situ engagement with the organization under the MITACS Accelerate research program.

In partnership with the organization, there is a team of physicians that work closely to evaluate and provide feedback on the clinical validity of the product. Physicians are not involved in the process of interpreting the original results, but the motivation to have a customer-tailored review is to bridge the clinical validity and provide guidance for a consumer and their care team, thus, physicians feedback and experience loop back into informing how to improve the product. I collected semi-structured interviews with one of the physicians, with a focus on how they interpret a report in consultation with a client, once they receive a health data review and action plan completed. Table 3.1 summarizes the pair analysis data collection summary with each different role involved in the case.

Table 3.1 Study participants and data collection summary

Role	ID	Participant Background	Task	Num. of sessions	Avg. time per session
Health Data Review Analyst	E02	Biosciences, PhD	Create data review summaries	3	1h
Health Data Review Analyst	E01	Geneticist, PhD	Create data review summaries	3	1h
Health Plan – Nutrition Analyst	E03	Nutrition and Food Scientist, PhD	Create nutritional health recommendations	3	45 mins
Health Plan – Exercise Analyst	E04	Kinesiology and Biomolecular Sciences PhD Candidate	Create physical activity recommendations	2	45 mins
			Estimated Total	11	10hs

Considering the nature of the protocol to collect the data, an intern will be the one common actor in conversation with each of our experts. Our intern is a Visual Analytics master student with a background in medical technologies. He’s recently joined the organization as part of the same MITACS research program. During each of the sessions, we asked our experts to talk to the intern as if they are training them to perform the same task they go through in the interpretation and creation of an action plan for an individual.

The role of expertise: Studying medical professionals compared to molecular biology scientists

Since the launch of genetic testing and the rapid decline on testing, research in the adoption of personalized medicine methods and philosophy has shown the barriers in the adoption of the practice by clinicians. *“Adoption of this approach has progressed slowly and unevenly because the trial-and-error treatment model still governs how the health care system develops, regulates, pays for, and delivers therapies”* say Aspinall, the president of Genzyme Genetics, and Hamermesh, chair of a Harvard Business School (Aspinall & Hamermesh, 2007).

At the forefront, medical professionals' lack of understanding in genetics and in the detailed *omics* data poses a barrier for the adoption of personalized medicine approaches in clinical practice. *“The bulk of the 700,000 practicing U.S physicians lack an understanding of issues in personalized medicine”* (ibid), since most of them received their medical education before the genomics revolution and the subsequent multi-omic profiling. This issue has been reported widely, as researchers, policy decision makers, and medical schools move slowly towards the inclusion of proactive, and preventive standards of care, such as the one promoted with personalized medicine models. (Gurwitz et al., 2003; Julian-Reynier et al., 2008; Najafzadeh et al., 2012; Weldon et al., 2012).

Needless to say, medical professionals are not excluded from the design and implementation of personalized medicine services and products. In my own case, I conducted pilot studies on the interpretation of data and the attitudes of clinical professionals in the use of multi-omic reporting with our service. These early experiences lead me to identify who were the more knowledgeable and had the most training experience on creating health assessment and lifestyle inferences from the biomarkers data. More research in the adoption of the technologies and methods for early detection of disease need to be conducted with physicians and medical experts.

However, for an in-depth understanding of how an *expert* works with the data, the most suitable candidates were the scientist building the knowledge databases and working from the trenches on a systematic interpretation of reports. Moreover, as we will see in Chapter 04, expertise in this case goes beyond the understanding and selection of scientific evidence, our interdisciplinary trained scientists deploy their expertise on

human kinetics, nutrition sciences and provision of health experience to create a compelling case.

Data collection

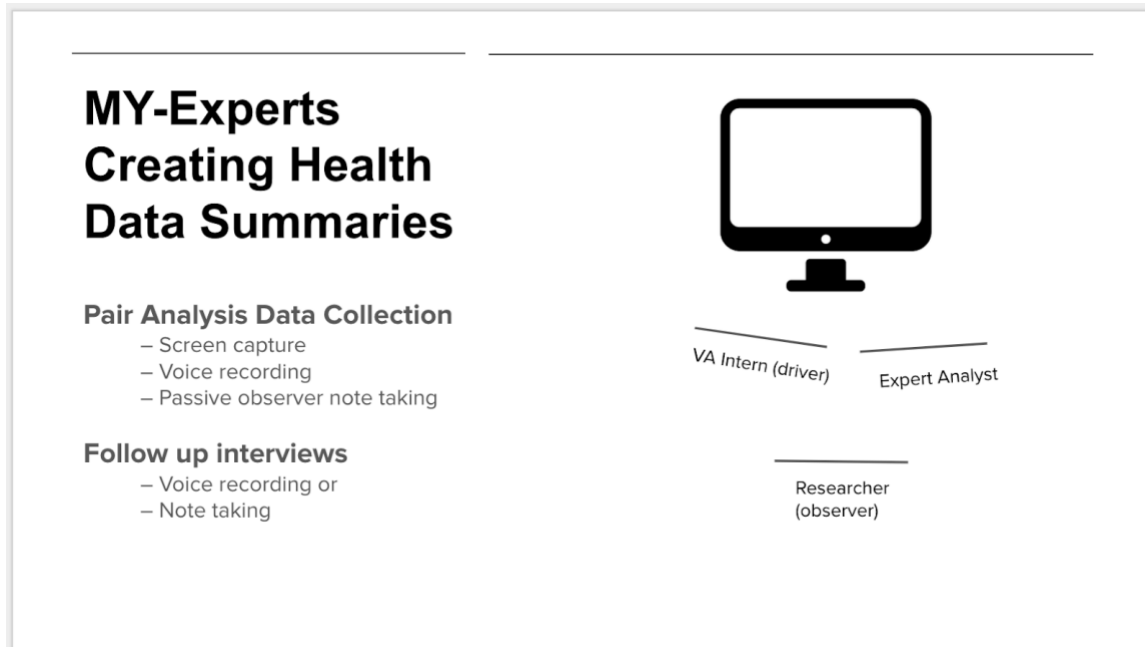


Figure 3.1 Data Collection for the Pair Analysis Protocols

I implemented a systematic data collection procedure in longitudinal sessions of pair-instructional-analysis with each of the stakeholders and a visual analytics intern. The setting in the room and the data collection protocol summary is depicted in the figure. I collected the following type of data:

- Screen capture: Using CAMTASIA screen capture software, I recorded the screen that the pair was sharing during the analysis. The data collection station used a 27' monitor so we could ensure both analysts had a good perspective on the screen.
- Voice recording: CAMTASIA is enabled to capture sound on top of the video recording. I enabled the sound recording feature, so all videos have the conversation recorded on top.
- Note taking I sat in the back of each session and took notes about the information and events of interest.

- Voice recording for interviews: I recorded the semi-structured interview using a voice recorder.

Considering the need to capture the decision-making process with cases that are new to them (not rehearsed or known in advance), I collected data along with the schedule for releasing reports at the organization. A single batch of reports is released every 3 weeks approximately. Each analyst goes through more than one report per batch, but I performed data collection just once per batch with each participant in order to account for (1) performance effects, (2) aiming to be the least intrusive in the process, and (3) to be considerate of time constraints they face with each batch of data.

At the end of the last session with each of my participants I asked them to stay for an extra 20 minutes. I held a brief semi-structured interview with each of them to capture their professional background, training, their opinions about the task they perform, and the set of skills they consider necessary to complete it (see appendix for the guiding questions).

Sample size sufficiency and data adequacy

Choosing a suitable sample size in qualitative research is an area of conceptual debate and practical uncertainty. Sample adequacy in qualitative inquiry refers to the appropriateness of the sample *composition* and *size* (Vasileiou et al., 2018). Table 3.1 presented an estimate of 11 sessions with 4 different experts for about 10hs of data collection. Within the VA community, there is a tradition of taking *deployed expertise* as a core argument to justify sample size. Convenience and limitations of access to this type of participants has been well accepted for a community working on a tradition of naturalistic sampling for studies of expert reasoning with interactive interfaces (see (Y. Kang & Stasko, 2012; Sedlmair et al., 2012; Ben Shneiderman & Plaisant, 2006)).

Following the field's tradition, the data collection for this study was design around the constraints of the expert participants. We have established the study as in-situ naturalistic research complemented with the design of pair-analysis sessions. From this we justify the selection of the sample size to be 4 experts that lead their analytic tasks in a small organization. However, regarding the decision of collecting 3 sessions of data with each of the participants, the justification comes with 2 arguments: data saturation and availability of non-rehearsed cases.

Qualitative inquiry researchers describe qualitative samples as purposive. That is, selected by virtue of their capacity to provide rich information, relevant to the phenomenon under investigation (M. N. Marshall, 1996; Vasileiou et al., 2018). In this dissertation, the selection of cases with each of the experts took into consideration that for us to capture an *instruction* session rich in verbalizations of the reasoning process of an expert, we needed to record non-rehearsed cases. Hence, each of the sessions recorded is captured at the moment experts go for the first time to work on a case of their backlog. For this reason, and in consideration to the amount of reports they go through every time there is data available, cases were available only every 3 to 5 weeks. We considered necessary to allow the relationship between the intern and the expert to develop and allow data collection post feedback with the intern regarding his level of interaction. For this reason, we captured at least 2, optimally 3 sessions between the experts and the intern. Regarding data saturation, we asked the experts to screen the cases they had available each time to work with us, if possible, so that they would choose cases with different data leads. That is, we aimed to capture different health cases, from low risk, mostly optimization, to high levels of disease risk management. Or cases with complete vs. incomplete self-reported health data included. This perspective on the diversity of cases to determine the saturation of data was based on the inside-knowledge and my own understanding of the data. As I have mentioned earlier, I was involved with the organization in multiple data initiatives from the beginning of the service. In the end, the resultant dataset covers a variety of analysis cases for which our experts have to expose their rationale and strategies to deal with each situation.

Ethical considerations

In this study we interact with a group of experts in their workplace. The team of experts corresponds with at least 4 members of the scientific team at a local small personalized medicine organization. The principal investigator has been involved with the organization through ENGAGE and MITACS internship grants since 2017. The participants are not required to provide any comment about the specific organization. Furthermore, the study data and results have no repercussion in the evaluation or assessment of any participant. The observations in the protocol designed, and the information gathered via interviews focus solely on analytical work, health information needs, the use of information technologies, and not on any other aspect of the

organization or agency that participants are working for. Participants are free to refuse to participate or withdrawal after agreeing to participate with no adverse effects. This study was evaluated by the Research Ethics Board at the University of British Columbia in a harmonized fashion with Simon Fraser University.

3.4. Data Analysis: Thinking with Pair Analysis Data

Transcripts

Pair analysis, protocol analysis and conversation data are often incomplete until we create textual transcripts. Although one common issue with which qualitative research methodologists disagree is the amount of the data corpus – the total body of data – that should be coded, the creation of the transcripts allows us, the researchers, to ensure a full overview of the data before the creation of the data that would be coded using salient portions of the corpus related to the research questions.

Transcriptions are labour intensive tasks, for this dissertation, a professional medical transcriptionist and me (the principal investigator) transcribed the full body of data from the sessions that were originally captured via screen capture with voice recording on top. I transcribed the first session captured with each of the participants. For the first iteration of creation of transcripts, I timestamped the full sessions but later noticed that the timestamps would not be key piece in the analysis I would perform considering the cadence and rhythm of the conversations unfolding. My take on the transcription process allowed me to determine the level of details I would use in the transcriptions, and to create templates and standards to provide to the transcriptionist. I used the multimedia files (screen capture video with voice recorded on top) for this first part of the transcription process. For each recording I provided notes of context to the transcriptionist and instructions on how to label the actors for the case using “expert”, “intern”, and “researcher”. The transcriptionist used sound files only (without access to the screens) for the task.

There are many valuable resources on how to create detailed transcripts for conversation e.g. (Clark & Brennan, 1991), (Jefferson, G. 2004, McNeill, 2006). For the purpose of this dissertation we did not include the transcription of gestures, except on few instances where it provides context that goes missing if not looking at the video.

The Atlas.ti qualitative research software proved to be better suited for the analysis of video data with transcriptions and for the first iteration on transcripts. At the time of my dissertation Atlas.ti allowed the horizontal alignment of the panels containing the video and the transcription.

Coding

In this dissertation, I started the coding process in a inductive approach to coding. Kathy Charmaz (Charmaz, 2014) describes coding as “the pivotal link between collecting data and developing an emergent theory to explain the data” (p. 46). Following the ethnographic tradition, this study uses open and selective coding to organize the properties of each the process distributed among the actors (Corbin & Strauss, 2008). During the first phase of analysis, I highlighted quotations as “key moments” or “dialogic sound bites” that rise to surface as relevant. I used research memos (available in Atlas.ti) to document initial ideas and themes that became repetitive and sounding for my research as I went through the videos first. From that, I created moments, extracts from the whole data that became the main data source for analysis. I adopted multiple coding techniques including open and in-vivo coding first. Because of the nature of the data collection, and truthful with an ethnographic approach, the coding and analysis process were intertwined and iterative. I transcribed and conducted initial open coding of the sessions as they were available, but by the last session recorded and transcribed I was able to use focused-coding with the codes and categories from earlier sessions. I remained open and allowed more codes to emerge until the last session.

Traditional protocol analysis is built on the basis that the thinking aloud protocol and retrospective reports that human subjects produce, can reveal in remarkable details what information they are attending while performing their tasks, and by revealing such information, they can provide a detailed picture of the way in which the tasks are being performed: the strategies employed, the inferences drawn from information, and the accessing of memory by recognition. Although more emphasis is given to the ability to capture this information in the form of task analysis, research has also shown how to study problem solving tasks by looking at the general processes, which is what the instance of my case study requires (Ericsson & Simon, 1993).

Regarding the development of codes from verbal data protocols, Ericsson and Simon propose that there are two main approaches to encoding and characterizing verbal data, either as an analysis of information revealed in a protocol following a task analysis process (i.e. the analysis of how a task is accomplished typically having one or multiple metrics of performance by knowing what is success in the execution of the task), or as an encoding and analysis in terms of task-independent general processes (Ericsson & Simon, 1993). In this research, and in the study of decision making with incomplete and novel data, we cannot define objectively what constitutes success, hence we will deal with the analysis of task-independent processes.

For this direct assessment of general processes, examples of coding schemes tend to categorize activities in terms of type and the status of information used by them. Whereas for the Task Analysis and heeded information approach the coding looks at the input/output of the cognitive process (information retrieved from memory), in this assessment we would be looking at the more high-level entities. According to Simon, “most theories that fit this description characterize cognitive processes by the types of processes involved, rather than the information heeded”.(Ericsson and Simon 1993, page 204).

A concrete example of how I started to instantiate a coding scheme in this fashion, is similar to Goor (1974), a system of 7 categories that can be seen as sets of activities in terms of type and status of information used by them.

1. Surveying given information
2. Generating new information or hypotheses
3. Developing or working on a hypothesis
4. Unsuccessful solution
5. Changing the conditions of the problem
6. Self-reference or Self-criticism
7. Silence

This particular approximation provided an initial set of categories to look at the sensemaking process, but the highly iterative coding-analysis process yielded to a broader codification.

Atlas.ti allows to annotate or highlight quotations in each source file independently, and so, I used the annotations in the video to highlight stages of analysis (i.e chunks of the video that I can identify as whole activities and projects) while working side by side on the transcript file for all the content quotations and analysis codes. Figure 3.2 shows a view of the working space, note that Atlas.ti devotes margins on the right of each file to label codes and quotations. The package provided a good overview of process and content in a single window.

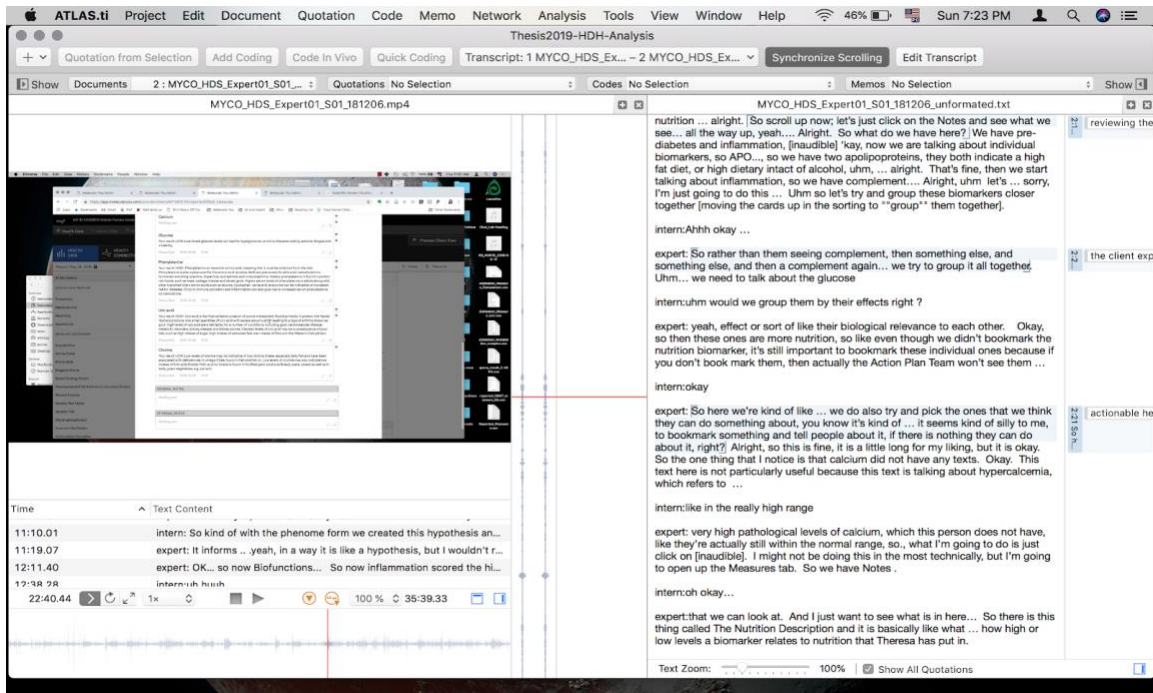


Figure 3.2 Atlas.ti view of the Pair Analysis data

The coding process was conducted by a single coder – me, the principal investigator and author of this manuscript. Reaching consensus with an intercoder reliability score is argued to be a standard measure of research quality, and a low level of intercoder reliability may suggest weakness in coding methods (Lavrakas, 2019). However, for a project of this magnitude and with the introduction of Joint Action Theory later in a deductive analysis, training coders to define categories without the fundamental knowledge in visual analytics and the domain of molecular biology was out of the scope. This is a considerable limitation in this study but also an opportunity to identify narrower goals and concepts in the data that can lead further investigation in the domain.

Analysis Results: Emergence from an Inductive Approach

Informed by an inductive approach to the analysis of my data, I set to understand the situation looking at the emergent categories in the data first. Because I participated as an observant of the sessions as they were happening, I took note of the differences in the analysis process between the action plan participants and the health data summary participants, mainly in the analytic tasks they were performing.

Besides the created pair-instruction-analysis situations for data collection, through my involvement with this organization I documented in research memos, opinions, observations, reflections and issues that I considered relevant for the analysis process. For the most part, I took note on challenges with the interpretation of the data, or due to the evident changes introduced in the DTC model of personalized medicine that positions the patients/clients as the direct consumers of data results. At the time of coding it became evident that there were enough instances that represented some of the same topics. By the end of the last iteration, five major themes had emerged from my analysis. This is a summary description of the major themes:

- **The sense-making loop:** In this theme, the data shows clear instances of an iterative process of retrieving, linking, summarizing information, posing questions to oneself, making explicit potential courses of investigation, testing them as hypotheses, and working within the constraints of incomplete data.
- **Actionable capacity, trust, and confidence in health evidence:** While working on a task, the participants would verbalize their rationale for attending a piece of data as important evidence, or seeking for more details about it because it would fall in a category of confidence or relevance given (a) its scientific validity, (b) the ability to target and intervene it via lifestyle actions, or (c) how familiar can it be for a lay person. The codes in this theme emerged annotating the verbalizations of those reflective references.
- **“The fourth person in the room”:** This is one of the most interesting themes in my data. It describes the actions, reflections, and reasoning arguments that the expert participants made about the data by virtue of emulating a conversation with the client or reflecting on what they consider the client’s expectations to be.
- **Team collaboration in a distributed process:** This captures the instances where each participant is referring to or taking actions that involve another team member in the process.

- **Reflective expertise (or the metacognitive ability):** For this theme I captured verbalizations that indicated any metacognitive reflection about the participants' own process.

Other themes that I didn't pursue in depth:

- **Expert suggest other features or automation:** the full automation of the process was a recurrent debate and topic in my research memos. However, in the pair analysis data, I found only few, sporadic comments from experts about expecting to have specific algorithmic features process. Under the "sense-making loop" I kept note of automation references that were used as explanations of their decision making.
- Bugs and issues with the tool
- UI styling, preferences

The sense-making loop

In this category, the data shows clear instances of an iterative process of retrieving, linking, summarizing information, posing questions to oneself, making explicit potential courses of investigation, testing those hypotheses, and working within the constraints of incomplete data. This is the largest and most present theme in my data. The codes in this theme represent explicit actions undertaken by the participants while working on one of the two general goals of the activity: (1) to identify what are the more relevant results and create a summary of the data for the client; or (2) to create a plan of lifestyle actions recommended to the client.

Interpreting molecular data to determine risk factors and significance in health is the higher-level sensemaking task for the first case. To interpret the significance of a given set of findings, our experts get involved in sub-activities such as evaluating confidence of evidence, posing and testing hypothesis or courses of action, annotating warrants and reasons, and determining importance of specific risk assessments taking into consideration the client's goals and expectations. In chapter 04 we will describe a process that is filled with recognition of patterns in biomolecular signatures, a task aided with algorithmic assessment but that requires further human evaluation. For the case of creating an action plan, experts engage in the overarching activity of creating a set of lifestyle actions including diet, exercise, and supplementation, tailored to a subset of relevant health risks and molecular measures. To do this, we observe that these experts engaged in sub-activities such as evaluating the confidence of scientific evidence,

annotating tailored justifications and warrants for an action choice and comparing/evaluating alternatives. In both cases, and as it has been documented elsewhere, complex activities of this nature do not take place in a linear fashion (Pirolli & Card, 2005; Sedig & Parsons, 2013), we documented experts performing loops of analysis and interacting with their tools in a way that allows them to consume the same piece of evidence from different dimensions of health impact. Chapter 04 is devoted to present the details of the sense-making loop. Table 3.2 lists the codes used in the dataset that make up this theme.

Table 3.2 Codes in The Sense-making Loop theme

The sense-making loop	
Codes: <ul style="list-style-type: none"> - Working on a hypothesis; - Generating new information or hypotheses; - Dealing with incomplete data; - Expert's intuitions and knowledge; - Linking or summarizing information; - Recalling known data; - Framing a narrative story; - Reviewing the output. 	In this theme, the data shows clear instances of an iterative process of retrieving, linking, summarizing information, posing questions to oneself, making explicit potential courses of investigation, testing those hypotheses, and working within the constraints of incomplete data.

The sense-making loop is the highest frequency category in my data. It should be noted that all the other categories emerge within activities in the loop, meaning they are embedded as part of the sensemaking process. See image 3.3 for the frequency of the codes in the pair-instruction-analysis dataset.

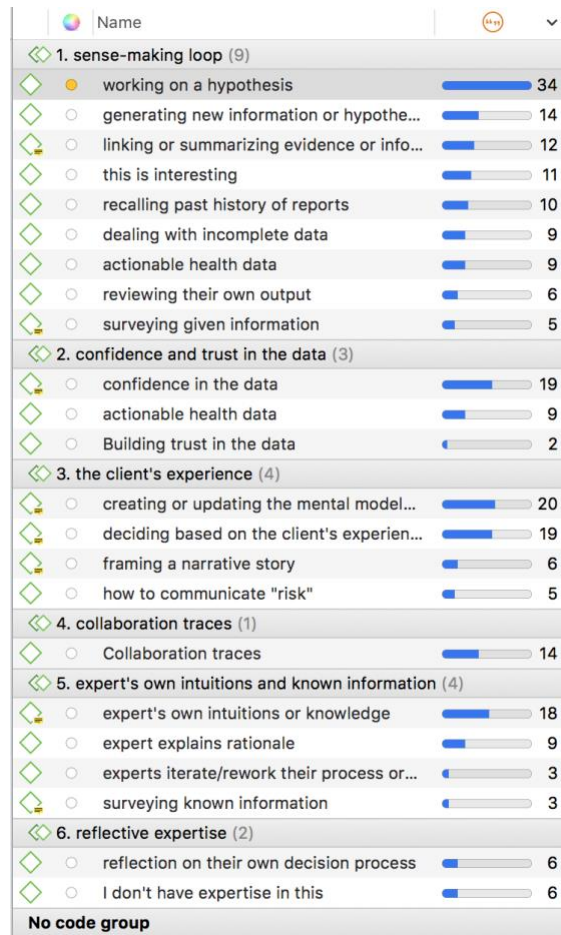


Figure 3.3 Frequency of codes in the dataset. Groundness.

Actionable capacity, trust, and confidence in health evidence

The codes in this theme emerged by capturing the verbalizations of reflective references the experts made about their rationale for attending a piece of data as important evidence or seeking for more details about it. Three main reasons explained the relevance of evidence (a) its scientific validity, (b) the ability to target and intervene it via lifestyle actions, or (c) how familiar can it be for a non-expert person. Alternatively, some instances of the same codes in this category capture when the participants are questioning the confidence or reflecting about what they trust or not in the information provided in the interface.

Examples of verbalizations ascribed in this category were: *“this protein is important for the inflammation pathway and is well understood”*, *“this measure is important because is something that can be changed via lifestyle interventions, not all of them are actionable”*, *“I will highlight this measure because is likely that people is familiar*

with it'. Dealing with risk and actionable capacity, was a recurrent topic for the experts to teach to (or comment to) Alan. See examples in table 3.3:

Table 3.3 Example quotes of the Actionability, trust, and familiarity of evidence

Code	Quote
Actionable health data	Gail: Okay, so then these ones are more nutrition, so like even though we didn't bookmark the nutrition function, it's still important to bookmark these individual [biomarkers]. [...]We try and pick the ones that we think they can do something about, you know it's kind of ... it seems kind of silly to me, to bookmark something and tell people about it, if there is nothing they can do about it, right?
Actionable health data	Alan: We have bookmarked vitamin C... this kind of derived from bookmarking this one or is it more from like a prior analysis? Gail: No, it's just because it is nutritional actionable.
Confidence of evidence	Antonio: For me, I personally, sometimes I feel like going through a report is kind of a ... it's like an art, right? And we get to make decisions and depending upon what your background is as well, you kind of have a feeling for certain things. For me I probably will not make a comment on this, because the clinical significance of this level is not well understood, it is low confidence and I look at the number ... is low, so I'm like there is no point in kind of telling this person to focus on the biomarker, because it's probably not relevant. So, I'll skip over that.
Familiarity for non-experts	Antonio: I'm just going to highlight haptoglobin, because that is something people are familiar with ... or sort of are familiar with... they might have heard of it, is what I mean by that.

Additionally, from the research memos, I documented early opinions from collaborator physicians about their concerns with the clinical interpretation of the large amounts of bimolecular data and the varying degrees of confidence in this data. This coding theme seems to uncover an effort to tackle the communication of reportable outcomes using *confidence*, *trust*, *familiarity* and, the *actionable capacity* as attributes of the evidence to convey and warrant meaning. In chapter 04 we will dig deeper into the details of attributes of evidence. Table 3.4 presents the list of codes that make up the theme.

Table 3.4 Codes in the Actionable capacity theme

Actionable capacity, trust, and confidence in health evidence	

<p>Codes:</p> <ul style="list-style-type: none"> - Actionable health data; - Confidence in the data; - Dealing with incomplete data; - Familiarity of the data; - Reflecting on trust on the data. 	<p>The codes in this theme emerged mostly via verbalizations of reflective references in the middle of the sensemaking process. That is, while working on a task, the participants would verbalize their rationale for attending a piece of data as important evidence given (a) its scientific validity, (b) the ability to target and intervene it via lifestyle actions, or (c) how familiar can it be for a lay person.</p>
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“The fourth person in the room”

Codes in this team describe the actions, reflections, and reasoning arguments that the participants made about the data by virtue of emulating a conversation with the client or by reflecting on what they are taken to be the expectations of the client given the data provided such as “goals”. As we will see in chapter 04, the schema of the particular client as well as the generic “health consumer”, the end recipient subject of the summary and action plan that they are creating, plays a key role in the decision-making process.

The creation of “the client” schema begins with the reading of the phenome data (for 3 out of the 4 participants), the data provided by each client about their health history and lifestyle habits. The sessions with John, the exercise expert, also showed us how the model can be created from a subset of the molecular data first. The completion of the intake health history data (*the phenome form*) is not mandatory. In the analysis of chapter 04 we would look at how experts work without a complete picture of the client’s data. The codes in this theme were used for instances in the data where we could see a participant doing maintenance of the mental model of the client, and for any reference about a decision action that was based on satisfying the expectations of the end reader, not only on the data results. See example quotes in table 3.5.

Table 3.5 Example quotes about communicating risk, and creating a narrative for the client.

Code	Quote
Communication of Risk	<p>Gail: I’ll do a quick check to make sure if there is a significant increased risk [genetic] variant that it’s [an outlier]. This is data I trust...</p> <p>[...] uhm, and genetics in general just freaks people out. So, we want to be sure that *that* data is reliable. If there is no significant increased risk, that’s fine.</p>

<p>Creating or updating a model of the client</p>	<p><u>Antonio</u>: So in my head it might be likely the person is old and it's caused through joint use and part of aging that the person has arthritis rather than something else, but I will also look for markers of inflammation in case I see those kinds of things, so yes, the same process. Old female, BMI is a little high.</p> <p>She must do something, I don't think people usually say "I want to sustain my wellness, if they aren't doing something like eating ..."</p> <p><u>Alan</u>: So, you assume that they are already doing something?</p> <p><u>Antonio</u>: mm hmmm, or at least they are in that mind set of doing that. People who I see are like obese and they're not taking care of their health, they will more likely say something like "I am dealing with a chronic issue" or you know, something else.</p>
<p>Deciding based on "the client's experience"</p>	<p><u>John</u>: we take a look at what their goals are... we really are going to frame what we write for this individual around these goals... [...] his goal is... disease prevention... increase muscle mass.... and fitness and flexibility ...</p> <p>We can see that this person wants more stamina, more strength... And they're not worried about injuries, and they're not worried about their weight, so we don't have to worry or give any sort of recommendations based around that. And so our Action Plan is going to be mostly centered around this, because we know we don't have any limitations in terms of injuries, we don't have any limitations in terms of health risks, we don't have any limitations in terms of biomarkers, and so it's all going to rely uhm on addressing this persons goals.</p>
<p>Framing a narrative story</p>	<p><u>John</u>: why a personal trainer is effective is because you pay someone, and now you are stuck to go there whether you want to or not, and even though you might hate it for the first couple of minutes, you are always satisfied and happy at the end, because you went through it, and someone watched you, guided you, and pushed you. And so... we keep that all in mind, because that is how we're going to write it.</p>
<p>Framing a narrative story</p>	<p><u>Gail</u>: So, working off that limited amount of information... I am going to write something that says like, "you have indicated that you have a chronic ... or ... you are looking to manage a chronic condition" I mean that is what they said in their goals, so we can address that. To tell him, "you know, in general you may benefit from reducing your levels of inflammatory markers, of which you have many that are in the high range. And inflammation is the key component of chronic conditions". "Proper diet and exercise as outlined in your Action Plan may help with control of your inflammation".</p>

I added to this category the actionable health data code as well because the importance of actionable capacity was often linked to satisfying the needs or expectations of the client. Talking about "risk", was observed as helping a client navigate and frame what a risk for disease means in this molecular evidence model.

In my memos, I often recorded discussions from the data team describing their efforts (individually and as a collaborative group) updating their processes to tackle the need to create a consistent argumentation narrative for the clients. In the coding of the data, there are enough instances about argumentation and warrants that are highlighted by the expert, even considered urgent to communicate, but not clearly materialized to the end user. Table 3.6 lists the codes that make up the theme.

Table 3.6 Codes in “The fourth person in the room” theme

“The fourth person in the room”	
<p>Codes:</p> <ul style="list-style-type: none"> - Creating or updating a model of “the client”; - Deciding based on “the client’s experience”. - Commenting on the “communication of risk”; - Framing a narrative story; - Actionable health data. 	<p>Actions, reflections, and reasoning arguments that the participants made about the data by virtue of emulating a conversation with or reflecting on what they think are the expectations of the client.</p>

Reflective expertise (or the metacognitive ability)

In this theme I captured verbalizations that indicated any metacognitive reflection on the participants’ own decision-making process. This category emerged in an early session when our intern asked one of the experts if what they are doing is testing hypotheses. The expert goes about explaining why they cannot call it as such in the formal sense because they are not looking to disprove the leads that they find but they are looking to check if the data supports what the client is reporting. See the excerpt from Gail:

Gail: in a way it is like testing a hypothesis, but I wouldn’t really call it a hypothesis, because just like when you are looking at a hypothesis, you are trying to disprove it, whereas here we are looking for “does it support what they say?”, “does the phenome form and the data line up and make sense together?” or “are they saying different things?”. Uhm, and I do think that introduces a little bit of a bias, but at the same time, this is very similar to what doctors do: you have to see what the person is like, and then you look at the data to see whether or not it supports what the person reports.

I then started capturing verbalizations that indicated any metacognitive reflection on their process. I found instances of the participants commenting on purposefully

avoiding extra information in the phenome form “*because it is not relevant to my process, and I don’t want to cause it to influence me*”, or acknowledging that they are bringing their own expertise-domain lenses to the process and how this influences the way they each would look at the same data. The following examples in this category capture John’s understanding of his own judgements in the closure of an analysis loop to move on to the next task (creating an action based on his analysis), and record Antonio reflecting on how each expert brings their own lenses:

Example 01

John: that’s it ... I can now leave this. So, I have an idea that this person is –by my bias– is more tending towards disease and extremely sedentary. And so then, I would go click my other tab that we’ve opened.

Example 02

Antonio: For me, I personally, sometimes I feel like going through a report is kind of a ... it’s like an art, right? And we get to make decisions and depending upon what your background is as well, you kind of have a feeling for certain things.

I also captured instances where they would transparently talk about how they don’t feel like experts in the details of some pieces of the data (for example Antonio talking about digging into the genetic variant details “*if it is not an obvious case*”), or instances where they confidently rely on the others’ expertise to complete the evaluation of what they are doing, for example:

Antonio: I don’t want [the client] exercising without you know, this joint issue and all these kinds of things... and I don’t want them eating something without proper consultation of people who are experts among that. So, John and Helena who deal with that... they will look at this same report and they will realize that this is pretty, pretty unique...

Finally, I added to this category a few instances where my participants commented on how they have iterated on guidelines and agreements that the entire team can use. The improvements are based on the reflections of how each of them works and what are the issues they each find when going through the data. This turned for me into evidence of a maturing and constant learning process. The codes for this category of data, are backed by the literature in expertise and expert performance, as I described in the literature review in chapter 02. Table 3.7 lists the codes in the data that make up this theme.

Table 3.7 Codes in the Reflective expertise theme

Reflective expertise (or the metacognitive ability)	
Codes: <ul style="list-style-type: none"> - Reflecting on their own decision process; - "I don't have expertise in this"; - Iterating over team guidelines. 	In this theme I captured any metacognitive reflection on the participants' own decision process.

Team collaboration in a distributed process

This self-explanatory category captures the instances where each participant is referring to or taking actions that involve another team member in the process. Examples in this category include, highlighting measures in the *health data highlights* stage for the *action plan* team to work on them; checking notes associated to measures that may have been written by a different team member; creating a request (with a note) to a team member with a different expertise to review a piece of the data; working out a partial solution of the text and relying on the complementary part of the team to complete it with their expertise. Additionally, instances where participants verbalize what they have learned from each other. I used a single code to annotate collaboration evidence in the pair analysis data: "Collaboration traces". Through the collaboration traces we capture the evidence of a process that require close coordination between numerous people, artifacts, and other elements of the environment.

3.5. Summary

In this chapter I described the methods I used to collect and analyze data from a synthetic situation designed to be deployed in a real world setting by virtue of the collaboration with a personalized health organization. The synthetic component refers to setting up a "training" scenario to capture the analytic courses of action that a group of experts take to complete the interpretation of each report they need to deliver back to clients. This is a routine task for them. For the data collection proposed, each expert worked in a paired setup with an intern and we captured data while they worked together in real cases. The dataset collected is a rich multimedia source of screen recordings with audio conversations. For the analysis, I started with a grounded theory approach to identifying relevant categories in the data related to the sensemaking process. Through my analysis, I identified emergent themes descriptive of the experts' analysis process

such as: the use of evidence, the strategies and pitfalls for building a communication artifact with a client, the evidence of exhibited *expertise* by these individuals, and the features of a collaborative, distributed process.

For the next two chapters I will elaborate on the analysis and discussion of the complex socio-technical system at play in the effort of creating and communicating health risks, evidence, and recommendations to a client in a direct-to-consumer personalized health service.

Chapter 4. Interpreting Multi-Omics Data in a Personalized Medicine Organization

I started this research program with a question of elements that influence the decision-making process of a group of experts working with large amounts of molecular data. Their task is to interpret the data to highlight health implications for an individual. In the case recorded in this dissertation, the experts work on a case-by-case fashion to for the analysis of the data. They conclude with a summary of findings and a list of lifestyle interventions to target any health or wellness issues found. The full analytic process involves an automated part that takes the results of the biomarkers' analysis and creates an assessment of health risks. However, early in the creation of this service, the team identified the challenges related to communicating directly to consumers the results of these assessments of risk along with the vast amount of data. This challenge is further emphasized in a model that delivers data results without the traditional health information keepers –the physicians. For this reason, the service extended the analysis process to add the human-interpretation component that deals with identifying and creating a summary of findings and a plan of lifestyle interventions to tackle risks.

In this chapter I present the account of the events that constitute the process recorded with experts in the interpretation and creation of a data summary product for a client. The narrative in this chapter is intended to convey the temporality and complexity of the socio-technical system where both, automation of data analytics and human judgement take place. I pay especial emphasis to the succession of events as well as the questions of what information is represented, where and how it is used, what patterns of information flow are formed in the collaborative analysis, and what patterns of information are taking place to account for the communication of results. Before a procedural description of the process that my participants (the experts) follow, I first introduce the details of the data, knowledge databases, and what constitutes evidence for the participants in their task. The inferences and highlights I make for these categories are based on the analysis of the data collected but also from the observation process from my extended involvement with the organization. I will discuss in further analysis, that the use of evidence complies with an *evidence-based medicine* approach to working with health data.

Because of the nature of the data collection and analysis, the coding and analysis process were intertwined and iterative, I coded categories of data that required me to go back to the literature iteratively. I conclude the chapter with a brief discussion on the findings that relate to research on decision-making in healthcare and the less evident uncovered nuances of the role of information interfaces in the communication of health assessments without provider mediators. This chapter is a detailed analysis of the case and the relationship with the evidence-based medicine literature. In chapter 05 I will investigate the use of JAT as a characterization of the situation between experts and their clients, to conclude with a discussion of the need to establish clear roles and technologies for emerging tasks that analysts take upon in the provision of care to health consumers.

4.1. Overview of the Socio-Technical System: People, Project Planning, and Data Reporting Tools

The *data team*, a group of individuals in charge of reviewing the results and create the lifestyle recommendations for each client, receives a notification with each batch of data, there will be 18 reports ready for reviewing in the current batch. The team is a group of about 10 people, but in this dissertation, I worked with a subgroup that represent the experts for each specialty in the team⁴. They were selected for their expertise and because they are the main participants in the process of interpreting the data for a client. They also have an important role in creating and managing the underlining knowledge databases, and in training new scientists and analysts at the organization. Through the dissertation I use fictitious names to anonymize their identity and as a way to easily refer back to each of them in the narrative. Thus, the actors in the process I recorded are described in table 4.1.

Table 4.1 Actors with anonymized identities

Participant	Specialization	Relevant Tasks for the Dissertation
Antonio, PhD – <i>The data team lead</i>	He is a specialist in molecular biology and biochemistry with expertise in experimental medicine.	<ul style="list-style-type: none"> – Research and development of the knowledge databases. – Team management. – Analyses reports to create the Health Data Highlights.

⁴ See description of the participants in the previous chapter (table 3.1)

Gail, PhD – <i>Genetics expert</i>	She is an expert in cellular developmental biology. Specialized in genetics of oxidative stress responses.	<ul style="list-style-type: none"> – Research and development of the knowledge databases. – Analyses reports to create the Health Data Highlights.
Helena, PhD – <i>Nutritionist and Food Science expert</i>	She is a Nutrition researcher specialized in food chemistry.	<ul style="list-style-type: none"> – Research and development of the knowledge databases. – Creates the Nutritional Action Plan recommendations.
John, PhD Candidate – <i>Physical activity expert</i>	Canadian-level former athlete with vast experience in coaching high-performance teams. He specializes in kinesiology and molecular biology for athletic performance.	<ul style="list-style-type: none"> – Research and development of the knowledge databases. – Creates the Exercise Action Plan recommendations.
Alan – <i>Visual Analytics intern</i>	Biotechnology engineer.	<ul style="list-style-type: none"> – VA intern involved in the design and product team. – He was involved with the organization in the part-time intern role for 1 year. – He is acting as the training participant in the paired-analysis protocol.

Managing the Analysis Process

Antonio opens up the project management tool and sets up the list of cards in the project board. The boards emulate a cork-board where sticky notes or index cards are used to visualize the flow of work through a process. Each card is the representation of the algorithmically-generated health report for one client. The card is the management representation used to move a report through the last stage of the analysis process: the human-expert interpretation and communication part.

There are five columns in the board, one for each step of the process that the reports will need to go through: (1) The “report ready” column, lists cards for all 18 reports with an identifier and a priority assigned that ranks the order in which the team should tackle the reports. (2) The “health data highlights” column shows a card with each

report assigned to either Antonio or Gail. They are in charge of interpreting the data and creating the *health data summary*. The next column (3) begins empty but it will show the list of cards for reports that are ready for the “diet and supplements” experts to proceed to create the nutritional component of the *action plan*. Once the card is in this column, it shows who has claimed responsibility for the task, in this dissertation we will read Helena’s process working on this step. The fourth column (4) also begins empty, is the list of cards that will be ready for the “physical activity” expert, John, after they are done with the nutritional component. John will work on the reports as they become available for him completing the *action plan* for each individual with the tailored exercise activities. (5) The last column shows the reports that are ready for a “language review and release”. Figure 4.1 is a schematic of the cards and the salient information used to manage the process.

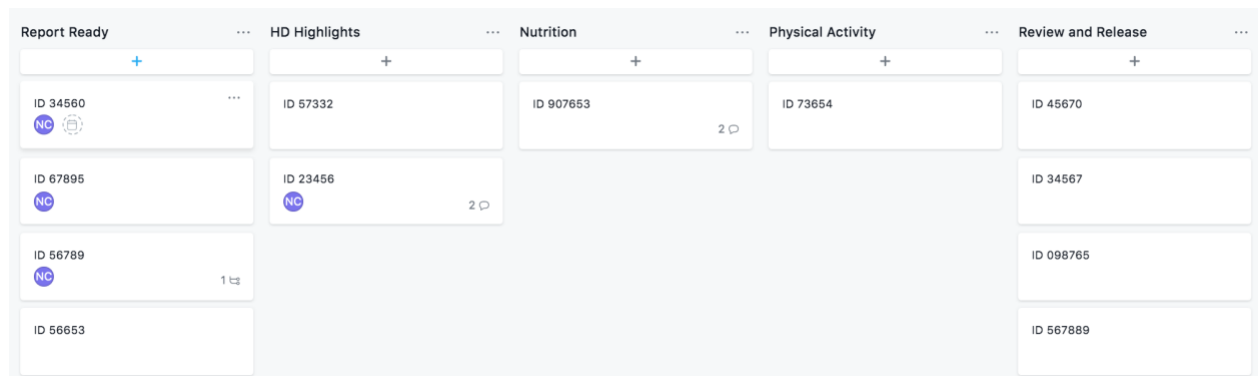


Figure 4.1 Card and board design of the platform used to manage the process.

The cards can be dragged and dropped between columns and they preserve the comments and the history of the conversation between each responsible for that report through the process. The team uses a feature for note taking in the cards to ask questions and communicate relevant pieces of information of the individual cases to the rest of the team. For example, in one of the report cards, Antonio gets a comment from the data quality control team that reads:

Client indicated they have arthritis, yet there was no signal of risk in the report results. –Just needs to provide clarification.

The Data Report

As an extremely simplified example of what constitutes a multi-omics report, consider what a simple blood test result is: a list of blood markers tested from plasma

samples with an evaluation of whether the quantified level of each measure is within the clinical reference for what is considered “normal” in a population. This means that for each marker, we obtain the quantified concentration in the body sample taken and whether this is within expected levels, too low, or too high. Typically, the results also include additional notes and highlights for the attending clinician.

In analogy, a report with the results of multi-omic testing could be thought in first instance, as the extensive list of blood markers of different types and the quantified assessment of whether they are found to be within the reference ranges for a population. In the case of the genetic variants (or snps) the results do not concern concentration levels in the body, instead, these can be thought as the list of thousands of variants tested with the corresponding genotype for the individual. For each variant annotated in the scientific literature, the report can tell if the individual carries a genotype that may be important for a disease risk, for medication metabolism effects, or if it corresponds to some interesting trait. Yet, the higher value of the *omic* data is in the evidence that links subsets of measures to the assessment of risk of disease or assessment of the health status for systems of the body, especially to capture early signals of disease. Continuing with the analogy of the simple blood test, the data report would include not only the results for the blood markers and the highlights for anything that seems out of norm, it would also include an evaluation of all the potential health implications from having some of the blood markers out of the reference levels.

The data report is then, the collection of quantified levels for hundreds of proteins and metabolites in the body, the assessment of the genotype for thousands of snp variants, and the complex network of health implications that can be inferred from this information in terms of signals of disease for hundreds of diseases, and the functioning of important systems in the body that can act as pathways towards disease (for example the immune system, the liver metabolism function, or the inflammation pathways in the body). Considering the nature of the data and the structure of interconnections, the report is not a lineal written document, but an interactive digital platform to explore the network of information.

However, after studying the response that consumers have with the data results, the reaction from clinicians as a second user group of all this information, and the challenges reported by other initiatives in the industry, the report now includes a human-

interpretation layer, what they have called *the health data summary*, and *the lifestyle action plan*. The goal of the health data summary is to help the users navigate the information by means of notes from experts on the items that deserve more attention. These annotations aim to support the user with the interpretation of risk assessments, and to better tailor what is relevant for each individual depending on their own goals (for example to discriminate if the goal is to manage a chronic condition or to improve wellness and performance). On the other hand, the goal of the action plan is to provide the clients with a set of lifestyle recommendations targeting the risk areas and the underlying measures that are outside of reference levels. The action plan aims at empowering the individuals to manage their health goals.

However, the biomolecular data is not the only source of information relevant in the interpretation process. Besides the plasma samples from blood, the clients provide self-reported health information, medical history, family health history, dietary information, and physical activity routines by means of a digital intake form-like questionnaire. The data team refers to this form as “*the Phenome*” information. The use of health history is not uncommon in the domain of public health for models of disease risk estimation, and in this case, we will see that the self-reported information is key in how the analysts frame schemas and decide courses of action.

The OMICS analytics and reporting information system

The interpretation process starts with our experts selecting a report to work on. They choose the identifier from the list of cards on the management board and look for the report in their custom-built *OMICS analytics and reporting system* internally known as the “*health intelligence system*”. This system is a single-entry point to all the information for an individual including the health analysis from the molecular results, and the self-reported health information in the “*phenome*” form. Additionally, from the OMICS reporting system, the analysts can launch the action-plan builder module, and they have access to a preview of the reporting interface that the client receives.

In summary, there are four core modules or views of the data that are the most relevant for the process described in this dissertation: (1) the *phenome* self-reported form information, (2) the *provider view*, an interactive visualization dashboard of the data report for each individual. This view presents the report (all the health assessment and

molecular results) with features for annotating, bookmarking, and reviewing the scientific evidence. This is the module that Antonio and Gail use for the core part of their task of creating a health data summary. (3) The *action plan tool*, a module for creating and managing the lifestyle plans created for each individual. An action plan is always linked to the biomolecular results, thus, the bookmarking feature in the *provider view* of results is a tool to select and communicate what are the most relevant findings for the action plan team members to work on. Through the process we will hear our experts talking about how they built the tools to link the data results and the lifestyle recommendations. Finally, (4) the *client view* is a preview of the interface that the consumer sees and uses to access their results. Some experts use this interface in their process to preview what the client would receive, while others use it actively as information source because it presents the health data summary that captures the conclusions from the former team member.

The interpretation process starts with a quick glimpse of the data results and the careful reviewing of self-reported medical data. Figure 4.2 presents an overview of the “*provider view*” that analysts use to access one report at a time and to launch all other modules. The landing page of the provider view shows an assessment of disease risks evaluated by the algorithm (left column). For each dimension of risk analysis (diseases, organ systems, functions of the body), the tool presents a data visualization summary of the molecular biomarkers used in the assessment (center bar chart in the image), and the full list of biomarkers with details such as the concentration values, the reference ranges, and the body of scientific evidence that is behind the association between the biomarker and the risk estimation (see bottom image in Figure 4.2).

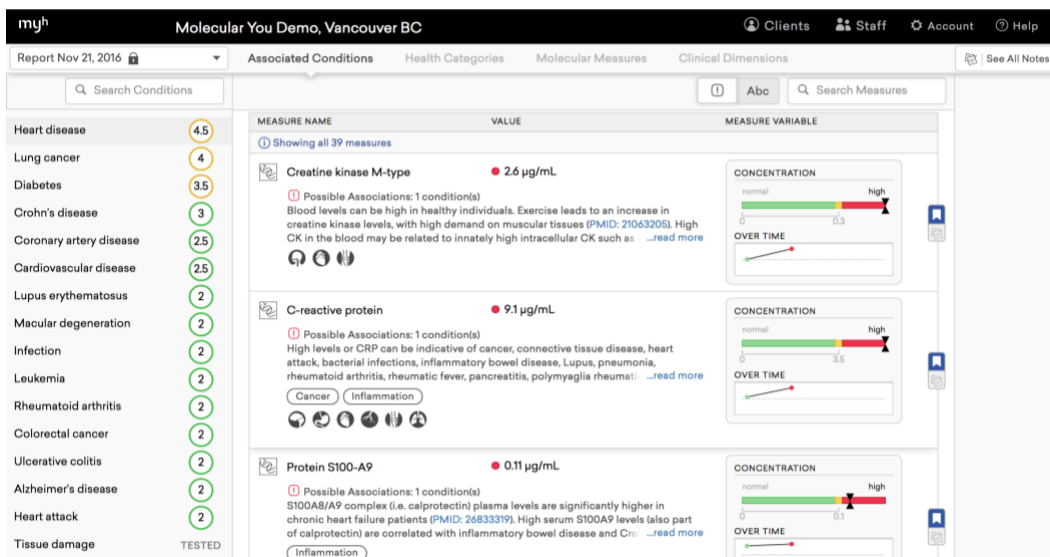
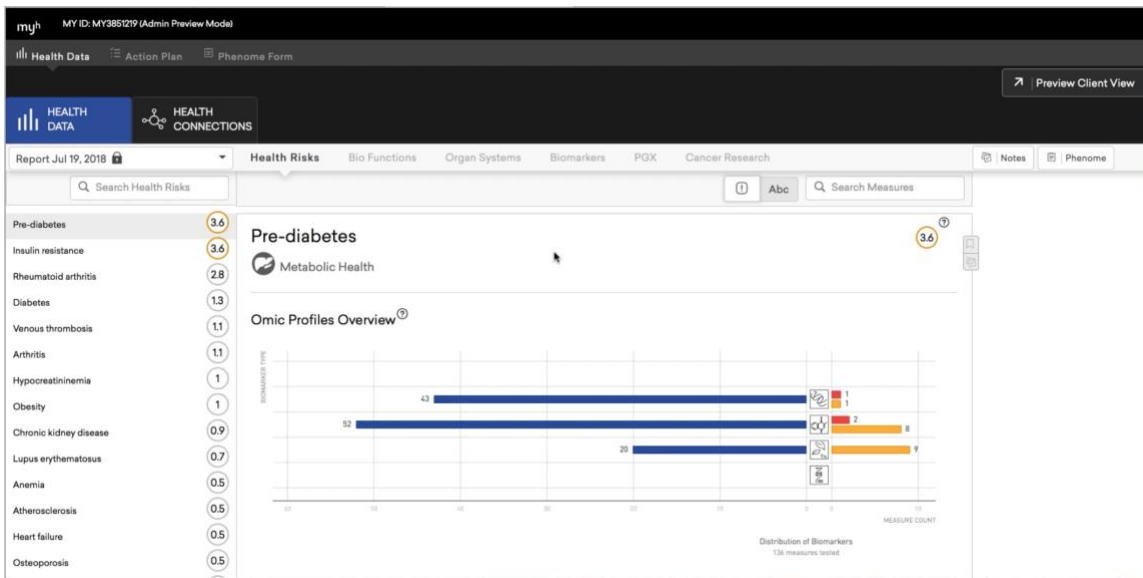


Figure 4.2 The landing page of the Provider view (top) and the details of information provided as evidence of a risk assessment (bottom).

Note. Each piece of information in the provider view has associated buttons in the right-margin for bookmarking and creating notes. The most important evidence is typically a list of biomarkers used for estimating risk of a health dimension as shown in the bottom picture.

Along with the display of data, the tool has features characteristic of a reviewing process such as creating and editing general notes, highlighting pieces of information (via bookmarking), and making comments for a specific data-result (similar to the comments you can create for a sentence or paragraph in a word document). Figure 4.3 shows the note editing feature. Bookmarking and comments can be applied to different dimensions of the data results such as: individual disease risks, specific functions of the body, or the individual biomarkers from the set of hundreds. In the example in Figure

4.3, there is an annotation for a disease risk (Pre-Diabetes), and both the disease and the first biomarker (c-reactive protein) have been bookmarked. The annotations are later reformatted and displayed to the client as a single *health data summary*. The bookmarks serve two functions: (1) a bookmarked measure that is part of a bookmarked category of health will be displayed to the client as an important piece of evidence (a highlight to pay attention to something amongst the bulk of measures); and (2) bookmarks are a key collaborative feature in the way the team has designed the workflow to move from health data assessments to creating an action plan. During the first stage of analysis one expert bookmarks the measures that they consider important because of their implications in the health results and because they can potentially be tackled with lifestyle interventions. During the second stage, two other different experts begin their analytic process using the bookmarked information. I will show the details of this flow of information in section 4.3

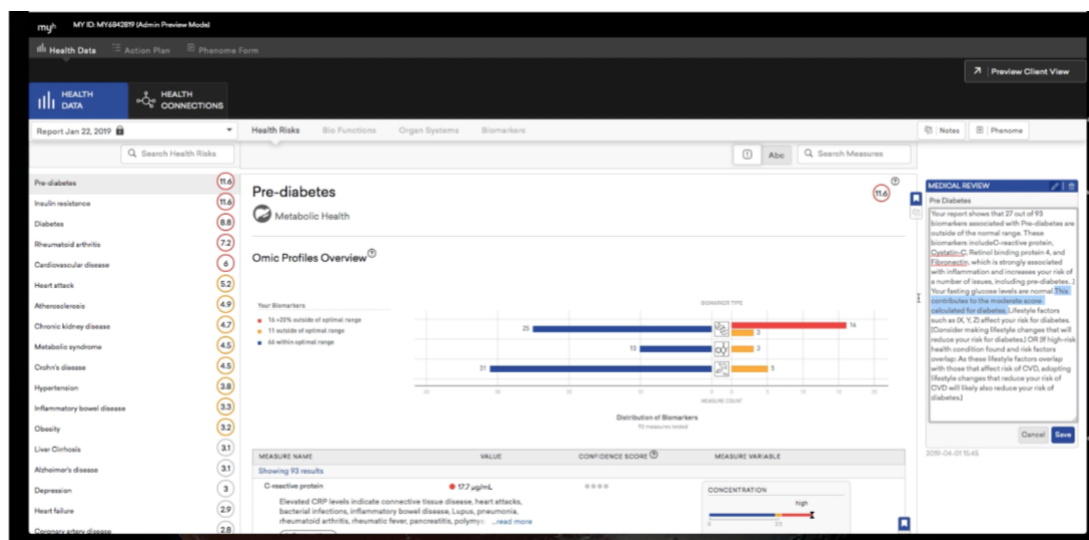


Figure 4.3 Screenshot of the note editing for prediabetes

The *provider view* is closer to a Visual Analytics toolkit in that it allows our analysts to explore the data set of results in an interactive fashion supported with a data visualization component. The features to capture the comments and highlights are also descriptive of an information system designed to support and document the reasoning process with multiple sources of evidence. The notes enable the communication of final outcomes from the analysis process. As a member of the product team involved in the design of the information systems, I was in direct charge of the design and implementation of the data visualization components, and I was also an active

participant of the design process for the addition of the reviewing features (bookmarking and notes) in the platform.

Our analysts mainly work on the *provider view*, because this is the interface in which they have access to the bookmarking and notes features, but they also access the data via the *client view* to preview the results of their work (the annotations). Sometimes they access the *client view*, to see some aggregates of the data that they find better represented in the client view, although this is more of a personal preference than a trend. In both views, whether an analyst working on a case or a client navigating the results, the report presents the full list of biomarkers assessed, and the same categories for the health assessments with the corresponding associated evidence: analysis of disease risks, organs and functions of the body assessment, genetic medication effects, etc.

The “*client view*” (figure 4.4) is the interactive reporting interface for the clients. The current version of the client view lands in a text-based page that compiles the notes that experts made during the interpretation process. The layout organizes the annotated results showing the summary for any risk of diseases first, followed by functions of the body assessments, individual biomarker notes, and finally other general notes and a disclaimer. Each section in the text is hyperlinked to the page in the platform where the user can obtain more details and the complete set of evidence backing up the results. The bulk of information, that is, everything that was measured and assessed but was not highlighted by experts, is also accessible via categories of information in the navigation menu. As a member of the product team, I was also involved in the design, development, and evaluation of the system. Particularly of the data visualization strategies used to communicate the network of scientific evidence connections (Figure 4.9 presented in the next section of this chapter).

myh MY ID: MY7579154 | Fred Home Health Data Action Plan Order Learn About Me Help

Report Date: Oct 25, 2018
Package Type: Premium Plus

Health Data Highlights

Welcome to Health Data Highlights.

Our team of scientific experts has reviewed your entire report. They have drawn attention to the most important findings.

Health Risks:

Pre-diabetes
Your previous report also showed a high risk of diabetes. Your current report indicated diabetes is now in the low risk zone. This is great news! Along with improved markers for Diabetes Risk, your risk for pre-diabetes is currently in the MODERATE risk zone. This risk has fallen from the high risk zone from your last report. Great job! Your key biomarkers associated with diabetes risk, such as fasting glucose, branched chain amino acids, and lipopolysaccharide binding protein are within optimal ranges. Below are the 6 markers that contribute to your moderate risk.

Relevant results

- Phosphatidylinositol-glycan-specific phospholipase D (high)
- Copper (low)
- Betaine (low)
- Glycine (low)
- Complement C4-B (low)
- Creatinine (low)

Body Functions:

Nutrition
You have 10 results related to diet that are outside of the optimal range. These biomarkers can be influenced by following your personalized action plan.

① Your individual biomarkers have been compared to levels associated with various health risks. Our database is regularly updated by our scientific team to represent the latest research on health status and risks. References to peer-reviewed scientific and clinical evidence are available throughout your report.

You can explore your health data through your interactive report. The health data visuals show how your biomarkers relate to specific health risks and imbalances. We also graph each biomarker in respect to your personal optimal range.

MYhi offers you a directional tool. It is not a diagnostic report.

myh MY ID: MY7579154 | Fred Home Health Data Action Plan Order Learn About Me Help

Pre-diabetes

Pre-diabetes means your long-term blood sugar levels are heading toward a diabetic profile. It usually means that your body is becoming less responsive to insulin. Insulin's main role is to signal the sugar in your blood - that came from your diet - to enter your cells. This lowers your blood sugar and allows you to use sugar for energy. When your body is having trouble responding to insulin; insulin rises in your blood, but your blood sugar doesn't lower. Over longer periods of time, climbing blood sugar levels can lead toward diabetes. These rising blood sugar levels can occur because of diet, lifestyle, genetics and environmental factors. Pre-diabetes is a warning sign to make some lifestyle changes. Usually diabetes risk is assessed with blood sugar measurements. Our assessment looks at blood sugar, along with early signals of pre-diabetes related to insulin's role. It also includes biomarkers known to be altered with pre-diabetes. A diet high in fibre and low in sugar can benefit and reverse pre-diabetes. Regular exercise specifically improve your body's response to

Dynamic Biomarkers

Your Biomarkers

- 3 >20% outside of optimal range
- 18 outside of optimal range
- 115 within optimal range

Distribution of Biomarkers
136 measures tested

Dynamic Biomarkers connected to Pre-diabetes

BIOMARKER	RESULT	UNIT SCALE	RANGE	CONFIDENCE SCORE
Showing 136 results				
Creatinine	48.1 µM	concentration	low	○○○○

Figure 4.4 Landing page of the Client view (top) shows the health data summary as a text summary. (Bottom) Details of Pre-Diabetes obtained when clicking on the corresponding title from the summary or navigating through the menu.

Finally, the action plan builder tool is a specialized filtering system that helps the action plan experts (John and Helena in this dissertation) to find recommendations in the lifestyle recommendations knowledge database. The information system allows smart filtering to find recommendations starting by selecting all the targets (the bookmarked measures and disease risks) that can be addressed with nutrition or exercise respectively. In this tool, the first list of data that experts find is the list of bookmarked

biomarkers that were highlighted by a team member creating the health data summary. In the case of Nutrition, the system filters diets associated as interventions for those targets, and drills down to select detailed food categories and foods. For example, the system shows a mark in 7 targets out of a list of about 15 items that were bookmarked in the previous stage for which there are nutritional recommendations in the data base. Each of the recommendations are individual diets that have been studied to have an impact. Helena is in charge of identifying common patterns for all the targets she is interested in, and making a call when there are conflicting dietary suggestions. The system also shows food categories ranked by a proprietary algorithm she developed and that takes the confidence levels from the diets recommended for all targets and then ranks the food categories like “legumes”, “leafy greens”, or “soy products” amongst many others. In other words, the system allows Helena to find what are ALL the diets known to have an impact on each of the selected targets and what is the level of confidence in the scientific evidence backing up the connection.

Due to intellectual property restrictions, I do not present here an image of the system but as an example, figure 4.5 is a representation of the hierarchy of the data that Helena uses⁵. The process is replicated for the counterpart of exercise actions in charge of John. The action plan builder allows an iterative filtering and selection of actions that are complemented with details of frequency, amount, alternatives, and notes. Both Helena and John use the notes to justify the rationale that connects each action to the highlights made in the former step.

⁵ All icons from the Noun Project licensed under Creative Commons free. The required credits are: Food collection by Made from the Noun Project. <http://thenounproject.com>

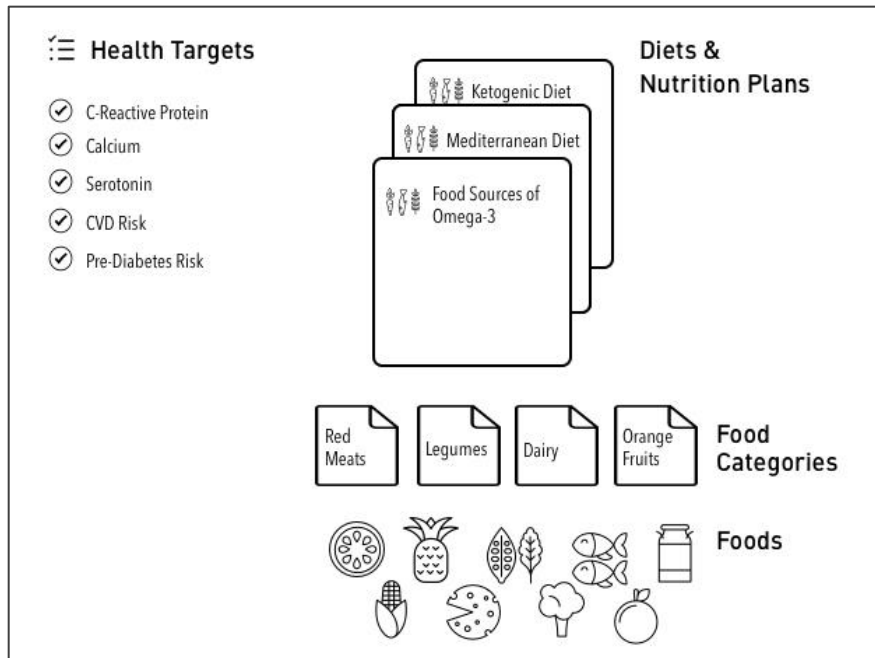


Figure 4.5 Nutritional data hierarchy

Note. Health targets are linked to evidence in diets and nutrition plans. Each diet includes a set of food categories. Each of these categories are commonly understood nutritional categories that includes a set of foods.

In addition to their own custom-built tools, the team uses “*Nuclino*”, a cloud-based team collaboration system that allows users to create and share information they want to re-use through multiple cases. Nuclino allows them to keep templates for repetitive notes and comments they make in the reports, different versions of the type of notes, the styling of the language, and even work-in-progress scientific annotations that they want to have for individual biomarkers. Most of the biomarker text descriptions are already in the OMICS reporting system but because the scientific content is in constant reviewing, the team creates scientific annotations ad-hoc as the experts see need for them. For this reason, some of the initial drafts live in Nuclino before they get approved to be imported to the OMICS reporting system. (*Nuclino*, 2019)

4.2. What Constitutes Evidence?

The report is a collection of information that starts with the quantification of hundreds of proteins, metabolites, and thousands of snp variants. The analysis and quantification of all these biomarkers becomes health evidence only when each of the measure results has been linked to one or multiple health risks and systemic

disfunctions of the body. For example, low levels of Serotonin in the blood, a biochemical messenger and regulator, have been linked to clinical depression with a high degree of confidence. Low serotonin has also been linked to chronic fatigue, sleep disorders, loss of appetite, hot flushes and headaches but with different, lower degrees of confidence (De-Miguel & Trueta, 2005). In this section we talk about how the data team creates evidence by collecting and assessing scientific information. But more important, we discuss other types of evidence that play a significant role in the analysis process and are aligned with the implementation of evidence-based medicine practices. Before moving to the description of the process that different experts undertake to interpret and create lifestyle recommendations for each individual, we frame their analytic endeavor in the use of evidence to create schemas or frames of reference about the case they have at hand. My goal is that with both, evidence and schemas, we can better unpack the analytic process delivered by different specialists.

A Central Repository of Scientific Knowledge

Following Evidence-Based Medicine (EBM) methodological commitments, the data team approaches the collection of evidence via systematic reviews and meta-analysis of published evidence. The data team has created and curated knowledge databases of scientific evidence with links for all individual biomarkers to multiple health risks. They have developed a system to manage the lifecycle of the evidence that they use through the decision-making process and that they include in the delivery of results back to their clientele.

The knowledge databases store the network of associations and the confidence level for each of these associations as dynamic entities whose lifecycle is updated and managed by a data mining system. In-house developed algorithms assess the strength of the evidence compiled. However, despite the efforts to automate and manage the vast amounts of information algorithmically, the quality assurance of this process includes a human-curated process for approving new evidence before it is added to the data reports with high degree of confidence. When I interviewed the experts, I would ask what part of their tasks they see more suited for automation as more data becomes available for knowledge discovery, to this issue, Helena was poignant to respond that she considered *critical thinking*, the ability to read and understand scientific evidence, and to be able to judge the robustness of the methodology used in each paper, as a task that required

skilled research expertise. Accordingly, she highly values the role of her PhD interns in the maintenance of the knowledge databases and the development of the supporting mining algorithms. On the other hand, she could foresee the suggestion and selection of lifestyle actions as an automated algorithmic task considering that the suggestions are available given the effort the team takes in curating evidence.

The knowledge base integrates new discoveries as they become available in the scientific literature repositories. This means that the confidence to link and use a biomarker in the estimation of risk for a specific disease or health dimension, is for the most part not static (with the exception of a handful of clinically established associations well studied and known in clinical practice). As more studies (evidence) become available, new links are drawn and the strength of the association is constantly re-evaluated over time. The information in these curated knowledge databases is the basis for the automatic creation of the health data reports. Similarly, the *action plan* experts have compiled scientific evidence about lifestyle interventions known to have an impact in the body at the molecular level. The evidence about the impact of lifestyle interventions for disease prevention and treatment is more widely understood (for example that an active lifestyle is a preventive intervention for the risk of Diabetes) but in the case under discussion, the knowledge databases also include evidence links between interventions and their effect on individual measures. This means that the team has a measure of confidence to evaluate that a nutritional or physical activity intervention will have a direct effect on specific individual biomarkers, and with that basis, it should be expected that a pre-post intervention testing on individuals would demonstrate the changes of targeted biomarker levels.

Types of Evidence

Based on the process observed with our experts, we will be interested in three categories of evidence: (1) scientific evidence, (2) the health history and self-reported goals and preferences of an individual and (3) expert intuitions and judgements. While the boom of personalized medicine technologies brings forward a myriad of scientific research from the genetics and molecular biomarkers, the data I collected in my sessions made evident the key role of self-reported goals of individual patients in the decision-making process of experts.

Scientific Evidence

Clearly, there is no intention to dispute that scientific evidence represents the “gold standard” when it comes to that which justifies the practitioner’s belief on a judgement or claim. However, as we discussed in the related works, what is arguable is the claim that only RCTs account for real scientific evidence. For this reason, one of the important components of evidence-based practice includes the use and dissemination of standards, practices, and procedures essential to achieve the EBM goal. For example, table 4.2 show categories of evidence taken from the literature that the team uses as reference for classifying scientific information.

Categories of evidence	
Ia	Evidence from meta-analysis of randomized controlled trials
Ib	Evidence from at least one randomized controlled trial
IIa	Evidence from at least one controlled study without randomization
IIb	Evidence from at least one other quasi-experimental study
III	Evidence from descriptive studies, such as comparative studies, correlation studies and case-control studies
IV	Evidence from expert committee reports or opinions or clinical experience of respected authorities or both

Table 4.2 Categories of scientific evidence from (D. Kumar, 2007)

The scientific evidence that links biomarkers to diseases, and that links therapies to effects at the molecular level, is found in different categories and at different volume of studies. The mining algorithms download information in bulk into the databases that is later reviewed and assessed with a confidence score. This means that the team has stored evidence from more than the categories above. The mining system also downloads scientific publications for indirect studies (observing the association as a collateral result of a study with a different goal), or studies that have not been performed in human subjects yet. The data team of experts has developed their own assessment of confidence to determine if a piece of evidence stored will be used in the assessment of risk for the data reports or not. The goal for storing information that does not make it to the report is to monitor and remain informed from data trends and open experimentation topics that despite not being marked as strong evidence in the present, may in time make it over the evaluation threshold as more studies become available.

Moreover, the networks of connections between molecular measures and health risks are not the only pieces of evidence that the team are interested in. A second piece of relevant information is the consensus on the expected concentration levels, or the

amount of a specific measure in the body in different samples like plasma or urine. *The reference ranges* are sets of values used by a health professional to interpret a set of medical test results from such samples. The expectation is that the reference ranges become a consensus for clinical practice, discriminating between different population groups. Take for example the expected levels of insulin in the blood for females vs. males or children vs. adults. The consensus on the reference ranges may be very well established for a set of clinical biomarkers, but for most of the vast array of *omics* biomarkers, the ranges vary in the scientific literature, and often in practice between different testing labs. For that reason, the team also collects the reported reference ranges in the scientific literature studying each of these biomarkers.

Putting it all together, the scientific evidence will create a link between a measure and risk only in the expected level. For example, while decrease levels in haptoglobin can support a diagnosis of hemolytic anemia, increased levels of haptoglobin may be a signal of an inflammatory process (infection, burns, major crush injury, allergy, etc.). This means that a measure outside of the expected range can only be interpreted as “evidence” in the context of a specific risk, and that not all measures found outside of their reference ranges are risks for itself. Figure 4.6 shows a visualization of the reference range used to report back to both scientists and clients of the product. In the figure, *Fibrinogen gamma chain* and *Haptoglobin* results are high based on the expected ranges, and *Apolipoprotein A-IV* is low. The data report will evaluate the risk of disease for which **high-levels** of *Haptoglobin* and *Fibrinogen gamma chain* are relevant and so on.



Figure 4.6 Results reported for four measures. Visualization of the reference range.

Note. The second column reports the quantified concentration. The fourth column shows the scale with the reference range and where does the current result falls within that scale. Notice that a measure level can be within the range (labeled as “normal”), below, or above the range.

Self-reported health information and patient's preferences

The self-reported information –known for the organization in this dissertation as the *phenome* data– is the medical history information that clients provide by means of a comprehensive digital intake form-like questionnaire. *Phenome* data plays an important role as health evidence for experts looking to connect patterns and identify trends. In the absence of an in-person consultation or the knowledge developed in relationship with a family physician, the digital questionnaire captures information about the health goals, concerns, health history, family history, and lifestyle routines from the individual. The team has evolved in the valuation and usage of the self-reported information, from being considered mostly, information to tackle the personalization of actions, it has now grown to be part of risk models and a key component of the analysis that experts follow. Through the description of the experts' process we will see that self-reported goals are determinant in the schematization of a case and that while serving as a valuable knowledge source for health care workers and analysts, the self-reported health records replace dialogue with the patient by a 1-sided extraction of information, which in turn raises the challenge to identify *the right* information to obtain from patients. In this organization the self-reported data questionnaires began emulating a clinical intake form and have evolved as our analysts see need for different or less information.

We found two main challenges in the use of self-reported health information: (1) the ability to capture accurate diagnoses and disease information from patients is not standardized by any practice and relies on the usability-design of the form, and (2) comprehensive health information is often scarce and incomplete. The capacity to understand and represent the human body is variable in the patient care spectrum. Moreover, the nuances of how individuals perceive and communicate their health also have implications in the mediated-recollection of information. In the design of form questions, the data team and the usability team often opt for capturing extra-information even if they are aware of flaws and general biases of the average user when asked about their lifestyle habits. For example, John uses a question that asks the user to fill-in their physical activity routine to get a sense of the type of activities that an individual already practices, but he discards the details about the regime they report. In his words:

“Unfortunately, when people report their current exercise, this is the least accurate way of getting the information. It's extremely inaccurate. More often than not people will think back to the last year, if not years,

when have they done anything, if ever anything... and then they put it down.”

But it is relevant to let the user describe what they perceive as their routine. To get a better picture, John combines the answer to the routine question with other questions that asks the client to rate themselves in aerobic fitness and muscular fitness, and a question on the client’s fitness goals. Together, those answers provide the information he needs for creating his frame of reference about the physical fitness of the person. For the case documented in this dissertation, we observed that the data team can work with the minimum (required) information available: age, weight, height, and gender. However, a more complete phenome form will lead to more targeted summary and lifestyle recommendations. The goals and preferences of an individual are of particular importance since it is through that knowledge that experts frame the way they communicate results back, as we will see through the detailed description of their process in section 4.3.

Expert’s Knowledge and Opinions

As we have said, our experts rely extensively on scientific evidence, and on self-reported health information that includes the goals and preferences of the client. They also make use of their individual *tacit* knowledge or *know-how* within their individual specialties. Moreover, we will see that an instrumental piece of tacit knowledge in the decision-making process is the behavioural and physical expression knowledge about how patients communicate their goals and put in action any behaviour change linked to lifestyle.

Following the work of our experts we will encounter that what determines the utility of evidence will be attributes of confidence that stand not only for the validity in experimentation but also for how much actionable information it provides back to practitioners and consumers. Not surprisingly, the action-plan team members, John and Helena, have a strong opinion about the type of expertise that is better suited to continue the task they perform. Both of them highlight the knowledge on translational practice, that is, the knowledge on human behaviour, habit formation, and physiology of sedentary vs. active individuals. For Helena, *dieticians* (in North America) have been trained to give nutrition *advice in practice*. Dieticians have a better translational practice, while nutritionists like herself, are strong in the interpretation of scientific evidence, especially

with graduate-level training. Similarly, according to John having expertise in *physical training* determines the success of the type of analysis and the task at hand. See the following excerpt in answer to the skillset he would look for an analyst performing his task:

John: I would say it would be more useful to have a background in training, than it would be in microbiology. Because it is easier to automate the microbiology component than it is the training component. We can provide a robust database to support a new person with the biology component.

But do you have any idea of how does a 30-year-old female exercise versus a 70-year-old or 60-year-old? Or a 50-year-old that weighs 210 lbs on a 5-foot 2 frame? If you had that understanding and insight, then you have a better leeway into giving the actions... because the actions are physical things. There're physical limitations to some exercises if your body shape or body composition is of a certain sort. Uhm, so that is invaluable. That's life experience that translates well into the Action Plan.

As a summary, we discussed different types of evidence that are present in the decision-making process documented in this dissertation, but that we can also find in the literature related to evidence-based decision medicine:

- **Scientific evidence:** All the experimental evidence compiled from scientific publications within the different categories of methodology used. In the case of this dissertation, the data team compiles all references that study the relationship between a biomarker and a health dimension and creates a measure of confidence for that piece. As we discussed earlier, there are different types of studies accepted as evidence in this category. The scientific evidence also includes the reference ranges used to estimate whether a biomarker is within or outside the expected levels for an individual in a specific population group.
- **Self-reported health information, goals and preferences:** Known internally as the phenome data, the self-reported information plays an important role as health evidence for experts looking to connect patterns and identify trends. In the absence of an in-person consultation or the knowledge developed in relationship with a family physician, the digital questionnaire captures information about the health goals, concerns, health history, family history, and lifestyle routines from the individual.
- **Expert knowledge or Know-how:** Following the Evidence-Based Medicine shift and the adoption of its practices and models, “expertise” has been considered in the clinical practice both a *type of evidence* category and an external component that brings together the components for judgement in clinical settings. In this dissertation, the participant-experts are not applying their knowledge in clinical settings, yet they are dealing with the creation of a product directed to health consumers and finding their role filling-in the gaps in the communication of findings and lifestyle health recommendations. In that

sense, we will be uncovering the extent to which their role becomes an evidence-based translational practice.

Visualizing Confidence of Evidence

The estimation of a confidence metric for the scientific evidence is intended to capture the quality of the evidence for each biomarker and its health associations. Different to attributing statistical confidence from experimental or observational data, the assessment of validity within the organization is a score assigned depending on the type of study, the population of study, and the direct or indirect assessment of the relationship of interest in the literature. The goal of the confidence score is to have a metric to determine the inclusion or not of a piece of evidence as an estimative of risk for each possible health dimension associated to it. Some clinically established biomarkers for diagnosis of disease have the highest confidence value, while links between markers and indirect studies of a disease (observing the association as a collateral result of a study with a different goal) have a lower one.

Additionally, in some cases the confidence is created by looking at a set of biomarkers rather than at individual values. For example, the set of 11 proteins in the complement pathway are evidence of the activation of the mechanism most responsible for immune-mediated injury. The activation of the complement cascade has a variety of biologic consequences which can result in injury to the host (James, 1982). Finding evidence of the complement factors activation in the blood can be indicative of serious inflammation in the body (as a response to immunity) and it has been associated with diabetes, systemic lupus erythematosus, kidney disease, and rheumatoid arthritis. In cases like these, our experts look the cascade of measures that are expected to be caught out of their reference levels.

A key component for the use of evidence is the transparency about its confidence value. This is true for both, analysts using the information through the sensemaking process, but also to the consumers of their results, the clients and their care teams. Figure 4.7 shows a network visualization of the relationships between a biomarker associated to disease risks and to organ systems with different levels of confidence. In this interactive visualization, users (expert and naïve) explore the strength of confidence for a link that has been established as evidence. The external layer (black icons) represents organ systems and health dimensions such as “Lung health” or

“Immune function”. The middle layer (colored rings) shows disease risks associated to the status of each of the higher order dimensions. For example, Diabetes and Insulin resistance are Metabolic conditions. Hypertension, Heart attack, and Atherosclerosis are under the Cardiovascular hierarchy. The middle layer shows all the biomarkers that were found out of their reference ranges for the individual. So, while there are hundreds of measures assessed (without considering the genetic component yet), in the average case, some dozens are found to be out of their reference ranges. The red and yellow dots represent individual measures that are either out of their normal range by 20% or more (red) or within 20% off from their reference ranges (yellow). Details can be obtained on demand for any element in the network. More importantly, on selection of a measure, the thickness of the link, maps the value of the confidence score for the association between a measure and a disease (see the different weights assigned to the links of Creatine in the image).

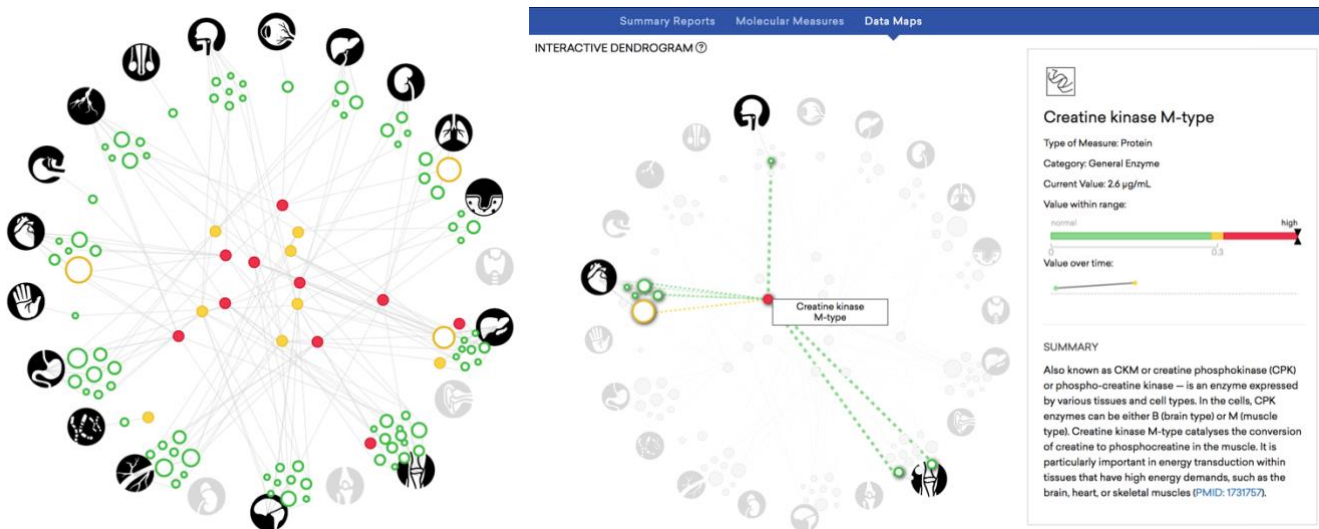


Figure 4.7 A network of molecular measures that are outside of the reference ranges and for which there is evidence of association with organ systems and diseases.

Note. By selecting data elements inside the visualization, the user can see how the selection connects to the rest of the information. Supporting text cards with details appear on the side of the diagram on demand. The thickness of the link represents the level of confidence in the association.

The evidence network was designed as a visual aid to understand that an individual biomarker is linked with varying degrees of confidence to multiple risks. After evaluating the use of such visualization strategies for clients and analysts, we found that

analysts are better suited with a straightforward table view with the confidence score listed in context. The network was appreciated by clinicians approaching for the first time the complexity of the bimolecular information. Figure 4.8, shows the confidence score reported in the data tables of the *Provider view*.

When we were evaluating this type of visual narratives, we received feedback from physicians stating that the evidence network helped them to create an introductory narrative to discuss the data with the patients that brought their biomolecular test results. One of the physicians commented that they used the interactive visualization to establish the ground with their patients: *“I tell them the data we are reviewing together should be seen as a network of interconnections from which to pick some signals for prevention or disease treatment”*. Although we have discussed that in the direct-to-consumer product the data is returned to clients directly, understanding physicians’ perspectives and uses of the data gave us insight into the issue of communicating back to individuals the concepts of risk, confidence and uncertainty in results.

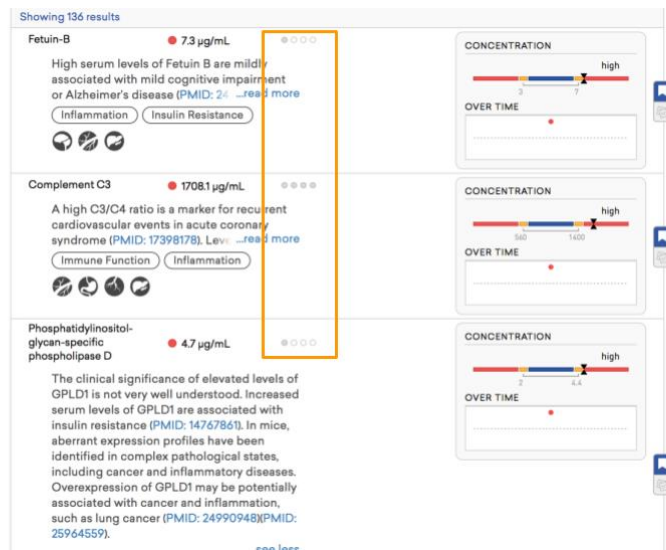


Figure 4.8 Confidence score from 1 to 4 in the Provider view, listed with each biomarker in the context of a disease.

Interpreting Evidence: Schemas or Frames of Reference

Decision-making schemas in problem solving are typically described as mental templates against which a decision maker or problem solver maps a specific situation (S. P. Marshall & Seel, 2012). We covered in the literature review the works by Pirolli and Card on intelligence analysis and by Klein in the NDM model about recognition-primed

decision making that emphasizes the role of schematic knowledge structures built from expertise and experience (Klein, 2008; Pirolli & Card, 2005). The expectation is that experts will have built up from extensive experience a set of patterns around the important elements of their tasks. And those sets of patterns are part of what Pirolli and Card denote “*schemas*”.

Through the process recorded in this dissertation, the data exposes an evident case on the use of schemas in the decision-making process for the experts in this domain. While the biomolecular data results are organized in a risk-assessment schema, finding a personalized health narrative that suits the goals and preferences of the client seems to be a strong requirement driving decisions.

The *risk assessment schema* is framed with the data and the health dimensions that are automatically added to a health report. It includes categories such as, risks of disease (risk of diabetes, risk of atherosclerosis), health status of organ systems (liver health), triggers of functional systems (inflammation pathway, immune health), etc. With the categories of information and the data evidence presented, the analysts can frame the case as one of either *disease management*, or *prevention and optimization*. That is, the assessment of disease risk and molecular measures allow the experts to put the case in one of these categories. As they go on adding evidence from the different sources they use, the observational data recorded shows our experts creating and updating the schema on the type of health category of report they are dealing with.

But it is only through the inclusion of the phenome data that the information is re-organized centered on a *schema of the client*. At this point, and in alignment with what Pirolli and Card report, the raw evidence is organized into small-scale stories about typical topics or in answer to typical questions (e.g., who, what, when, where, why, how). The schema of the client may include a previous history of disease –the *what* and *when* are recorded– the *why* and *how* become leads to investigate in the *biomolecular schema*. But most importantly, it is the *who* that is being framed as an individual with specific goals, and a disposition towards their own health what drives most of the hypotheses leads.

A main task the experts take at this point is to create a client-model as a working schema. Experts may work from the biomolecular evidence to create a reference (as John

does given that he works with a much narrower dataset) or from the phenome self-reported information to help the experts frame the boundaries of the analysis. In the end, the goal is to bring together both schemas and a story about the client as the final product. Take for example the following quotes from Antonio about the role of the phenome data in his schematization of the case in comparison with a quote from John about developing his frame of reference from the biomolecular information:

Example 01: Antonio

Antonio: The way the phenome form for me is to kind of get a feeling of what I can expect or not expect to see. Uhm, this portion is really important, right here: *the personalized health goals and motivations*, uhm, this section I feel is really, like the person is talking to me.

They're just like okay... "you know here's why I did the test" or "here's why I have an issue" –this really guides me into how I'm going to write the story of the report.

Example 02: John

John: Before I even look at what they've said that they do, I'll go to the biomarkers tab I opened. And I can quickly just punch in a few... [types in a few measure names] and if you scroll down, we're going to look for apolipoproteins A1 and we see for this individual it is super low; we know that apolipoprotein A1 is extremely relevant for people that are currently exercising.

Apolipoprotein A1 can move with numerous things; if you're exercising and in somewhat of a healthy state, even if there's lots of issues, it should be on a higher end... and so by this biomarker, either this person rarely ever exercises, or they could be exercising to such a point that their body is completely breaking down. I would say that it's usually not the case.

We can use the Phenome Form afterwards to confirm. I'm going to go with the approach of not knowing anything about the individual, just look at the biomarkers, and see where do they lie? Do they lie with athletes, or do they lie with sedentary, or do they lie in disease? And if they are, then we can make recommendations accordingly.

Schemas also play a role ranking and prioritizing the tasks in the interpretation process. From the many signals picked up in the biomolecular data, our experts decide what is more relevant either based on warning diagnostic flags from the biomarkers or based on what the client is telling them about their goals, concerns, or history of disease. Tackling disease risk seems to be the leading priority for the team. And it has mostly to do with the need to help the client to understand the uncertainty in an assessment of

risk. Their goal is to clarify for a client to what degree the probability of disease, as measured by the algorithms, is a priority area to focus on. For this purpose, Antonio, Gail and Helena have as part of their general schema of a “*client*” not only the details of the specific individual, but also a general reference of the sensitiveness for individuals when it comes to talk about “health risk”. In some ways design thinking and user-centered practices fall within a reasonable analogy with the work that our experts perform. See examples of the topic discussed by Gail and Helena (in different cases):

Example 01: Gail

Gail: [The notes] are general recommendations. You want to empower [the client] right? Like you want them to feel like, this is not a death sentence, is a moderate risk of CVD. It’s more like “*this is what you can do to reduce your risk of CVD*”, right?

Before this [update to the OMICS reporting tool] we didn’t have the feature where you would highlight a health risk and it would give you all the biomarkers associated with it, so you were trying, for **each** biomarker of those highlighted, to have paragraphs to talk about them and the feedback we got back was that it was really overwhelming for people.

People tend to focus on... “*oh my God I have this *one* biomarker that is associated with depression. Should I see my doctor for depression?*” and [what we try to tell is] like “*no, no, big picture... you have other things to worry about*” Uhm so that’s why we came up with these guidelines for bookmarking biomarkers and communicating their risks.

Example 02: Helena

Helena: So, we have got the feedback that when people see that they’re like “*oh my god I’m at risk for such and such disease, what am I going to do about it?*” and so it’s good if we have it in the action plan right? because then they’re like “*oh okay, *that’s* what I can do about it*”. Yah, and so it seems like the diseases and the [systemic] functions seem to worry people the most. But that is why the Action Plan is important.

Finally, as what is expected with schemas from a psychological perspective, schemas are formed and accessed from experience. By this point in their operation task of creating reports, the team of experts has identified recurrent patterns in the data they work with, and “types” of reports that would be candidates for pre-screenings and more automation tasks. See the following excerpts from the participants:

Example 01 Helena

Helena: This was actually quite a quick and easy action plan, because there was not a lot of information, it's like inflammation and diabetes, I've seen it a hundred times.

Example 02 John

Alan: and so, for each of these reports did you write a comprehensive summary like this one?

John: so luckily for the 'Comprehensive Summaries' a lot of them are already written out, and what I do is copy/paste from the ones that I have, to make it consistent. And so ... whether it's addressing joint stiffness, or injury rehabilitation, or specifically weight management... whether it's uh increasing muscle mass or muscle tone, there is already things I've written with enough iterations that I think I'm now finally at a point where I've seen so many times that I know there are differences[...] There are still, even to this very day, examples of the lactic acid / glucose profile, that it's so unique that it needs its own approach to it. Which is why I haven't made any clear-cut automation. I should have possibly about 8-12 clear distinctions, that can give me massive prompts, and then of course, uhmm... that would be maybe our first attempt at the automation... but as you can see with the exercise, it is incorporating so many different avenues that it is not just as simple as "here's a biomarker – here's the exercise".

4.3. Cycles of Analysis Working with Biomolecular and Self-reported Health Data

After an overview of the components of a system of people, data, information tools and evidence, in this section I follow the temporal events of the process that the expert participants complete for the analysis of each report. Let us recall that a data report with the assessment of health risks is algorithmically created from the results of the biomarker set measured from plasma samples. Following this, experts go through each report to create notes, highlights, and calls for action.

The health data interpretation stage is the analysis devoted to identifying what are the most relevant results for an individual's health, including risks of disease and related individual biomarkers that are outside of the optimal ranges. The action plan stage is the process of creating lifestyle recommendations that are known to have an effect on the identified disease risks, biomarkers, or both. The final health data interpretation product is the *health data summary* that compiles a written narrative from notes and highlights created by the scientists. The final action plan is a set of *lifestyle actions* created by experts in nutrition and physical activity.

Figure 4.4 showed in previous sections presents the result of the health data highlights and the details of the data in the report as seen by a client. The screenshots on Figures 4.9 and 4.10 shows the details of actions delivered to a customer and the network of targeted goals. Each intervention is linked to risks and biomarkers that can be tackle with lifestyle changes.

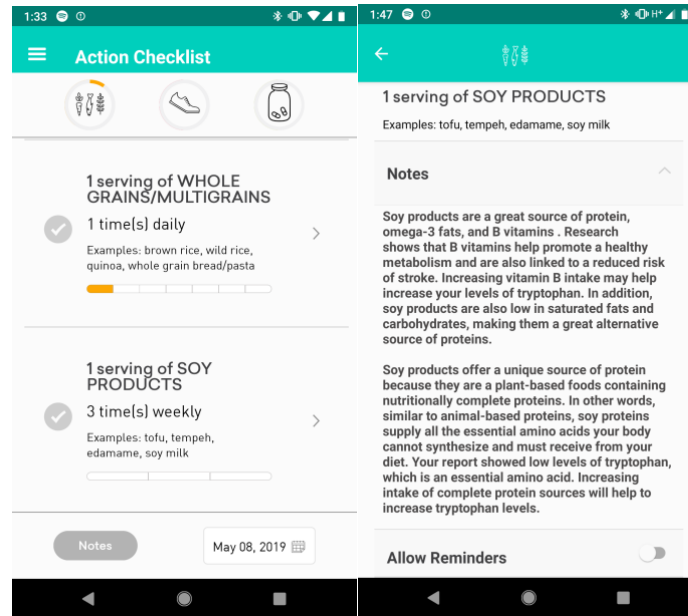


Figure 4.9 Overview of nutrition recommendations in an action plan. Clients get a list of actions (left) and can obtain details about how does it connect to their health data results (right).

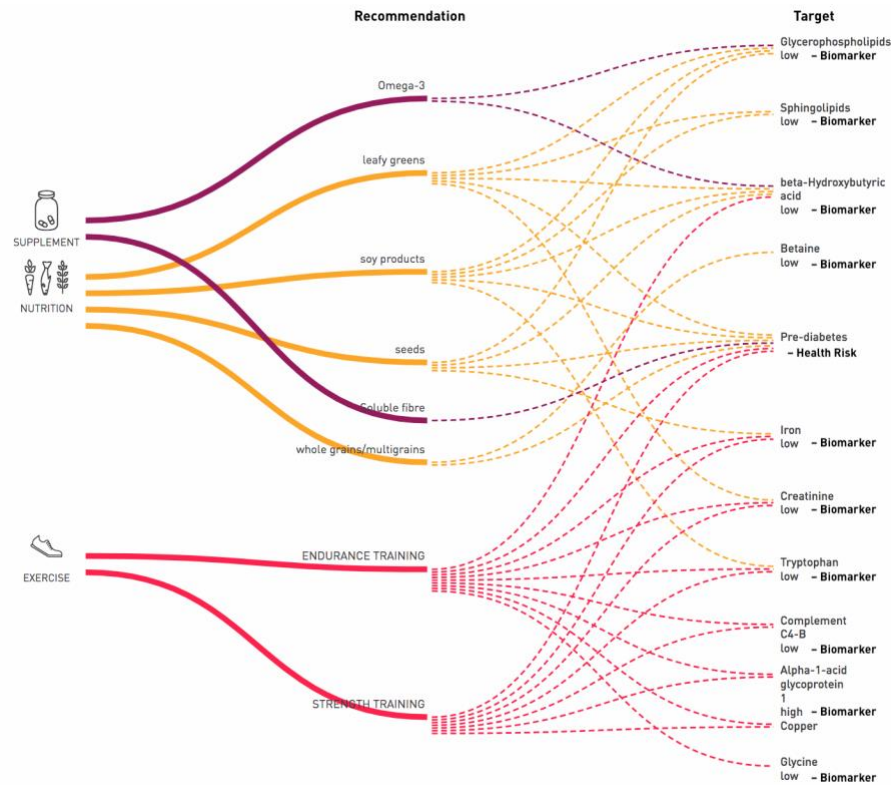


Figure 4.10 Visual depiction of a list of recommendations in a single action plan.

Note. Each action (middle column) belongs to a category of lifestyle interventions (left column) nutritional, supplementation, or exercise recommendation. The evidence links each recommendation to a target, biomarkers and health risks in this case (right column).

Creating the Health Data Summary

Antonio asks Alan to open 3 different tabs in the screen, the “*Provider view*” of an individual report, the “*Client view*” of the same report, and “*Nuclino*”. The process starts at the landing page of the *provider view* that shows the list of health risks (see figure 4.1 at the beginning of the chapter). Risks in the report come with the assessment score color-coded as gray (green in an older version) to convey low risk, yellow maps moderate risk and red represents high risk areas. Before they start exploring the data results, both Antonio, and Gail do the same: they go straight to the self-reported health information in the phenome questionnaire. This is an overview of what the initial piece of the phenome data reads for one of Antonio’s cases:

Antonio: First thing, let’s look a phenotype form. Okay, this one has stuff, so yeah, old female, BMI is a little high. Normal BMI is around 24-25, so this is probably in the overweight range [...] So her hope is to

achieve her best and sustain wellness... Yeah, I guess she has got... she exercises, or does something...

Alan: 70... okay old.

Antonio: yeah, so she must do something. I don't think people usually say "*I want to sustain my wellness*", if they aren't doing something like eating well.

Alan: So, you assume that they are already doing something

Antonio: mm hmmm, or at least they are in the mind set of doing that. People who I see are like obese and they're not taking care of their health, they will more likely say something [in their goals] like "*I am dealing with a chronic issue*" or you know, something else. Most of the time.

The highlights in the first paragraph show that the phenome data is not always completed by the client. As we discussed earlier, the goals and preferences reported by a client play an important role as evidence in the decision-making process, but experts have also learned to work with general cases. After taking some time with the phenome data our experts go into analysis loops similar to what has been described in the literature of intelligence analysis (Pirolli & Card, 2005). During this stage, experts use the information from the phenome form to create a schema of reference and set the boundaries for the case. Both Gail and Antonio use the self-reported goal to get a sense of what the client's expectations are and what is the type of conversation they could have with them. In the absence of the self-reported information, they are driven only by the results in the data report and their understanding of the different patterns of risk. From the phenome data, they may or may not have information about the history of diabetes for the client's family, or the background, or the diagnosis of specific conditions, but they do have a recollection of different signatures of diabetes they have seen in the reports, and in some very well understood cases, such as diabetes, they also have the strong confidence in the clinical biomarkers. A risk of diabetes may be driven by inflammatory markers, by the lipid metabolism measures, or diagnosed with measures of glucose levels. They will tailor how in-depth can they get for each client, and how specific with regards to the goals if those were provided in the phenome form.

The following excerpt from our first session with Antonio captures the moment when he is verbalizing a summary of what he's decided is relevant from the phenome data before they move into the analysis of the data results.

Antonio: so right away for me some indicators from the Phenome form have been, metabolic health, have been liver, kidney, and some kind of joint stuff. Some joint, or some kind of like arthritis stuff. And from Family History, there was cancer in the family history and dementia. So those are two things I may look at, but these are my focus areas that I want to focus on right away.

So, I kind of have this like backstory of what they're telling me. Oh! and I should also mention that they said they have kind of an active lifestyle-ish or their concern was wanting to '*maintain my wellness*' so, maintain wellness. They've complained, like I said, of some kind of joint issue, so joints.... I'll make those notes, and now what I'll do is I'll look at this area.

The assessment of the data through cycles of analysis (or loops of analysis) works following pattern recognition tasks. Once there is a schema of the client, Antonio may have in mind that is important to talk about the risk of arthritis for the client, but he is also seeing the recurrent pattern of inflammatory markers that drive a risk for other conditions so, he chooses to make a note about those as well.

As a window into how they process the data, the following excerpt follows a trail of explanations that Gail gives to Alan as they navigate the data. They have already visited the *phenome data*, the case is for a 32 years old male individual that reported as goal "*to better manage a chronic condition*" but without further specification. In this example she is visiting multiple risks highlighted in yellow or red and drawing conclusions given the patterns of information:

Example 01 Gail

So, I am seeing a lot of inflammation markers, so that is like fibronectin, complement serum amyloid, some I think thrombospondin is inflammatory. Uric acid is high, which is interesting, and then 1APO C protein, which is a fat metabolism marker. Okay... scroll back up.

You know, a lot of these [markers] are related to inflammation, and that may be a more general thing. but it just so happens that most of them are associated with diabetes. Uhm... given that this person has low glucose, I would say that pre-diabetes is probably not exactly what they have to worry about... It just so happens that a lot of these other markers are associated with diabetes, so that is why pre-diabetes is scoring high.

Okay, now if you go to Coagulation and healing, this one is a tricky one, because often times you might get sort of contradictory patterns or you might not really know now to interpret this.

uhm, and also, it's not really actionable ...

But it is the second highest one on this list [they are looking at the ranking of risk-assessment]. So, let's have a look at what's in there ... so confluent factor 18 ... prothrombin, fibronectin ... okay, can you click... read more on that... okay, nothing okay, great uhm, okay, go back up. Uhm I want to think about that one for a while, so let's go to Immune Function. They are almost the same.

[...] alright, finally if you go to Biomarkers. There are some individual biomarkers that I have seen so far that I think might be worth highlighting for this person. Let's bookmark calcium, just because it is a little bit high, okay. Complement C5, okay... and maybe we should bookmark glucose because the glucose is low and we did point that out in diabetes, but ... uhm, actually, bookmark phenylalanine as well.

Depending on the complexity of the data reported (many disease risks identified, or very little risk identified), the task is to iterate over the results that need clarification for the client, making sure to evaluate the evidence for disease risks and pathways of disease first. They click on the categories of information in the report in the order of priorities they established when creating their schema. This is evident in the screen capture videos as we see that they do not click on the diseases in the same order they are ranked and listed in the interface. For all the cases we recorded, the experts started with the disease priorities that they determined from the phenome form and did not cover the full list because as they read through the biomolecular evidence, the patterns of information become saturated and repetitive while moving between categories.

If the case represents a generally healthy individual for whom there aren't any foreseeable disease risks in the data, our experts will work with the systemic functions, organ systems, and other categories of information that can be addressed for prevention. The system will generate an automatic note for the client stating that "*they have not found any alarming risk of disease*" and display the other pieces of information highlighted. For example, inflammation, immune health, lipid metabolism, nutritional markers, are some of the functional dimensions that are part of this assessment, and that can be used to frame the review story from prevention and optimization. Whether there are or not many signals for disease, there is often a case in the data to talk about some of the functions such as nutritional biomarkers that are showing to be out of the optimal ranges established in the literature, or immune system signals that may be indicative of a deficiency that can later translate into disease. The experts will talk about the functions in the context of anything the client has reported back to them: "*he*

mentioned he has pain in his joints, so I'll address that on my note about inflammation" for example.

In this stage, the experts use the bookmarking feature of the tool for two main purposes: (1) they bookmark information they will later want to make notes about, or (2) they are highlighting information for the team members in charge of creating the action plan recommendations. The notes they write for each of the bookmarked pieces will turn into the health data summary, for the client.

The team uses templated language to keep consistency and uniformity in the product, but we can see how the development of a narrative and a story, along with the careful selection of the language they use for talking about risk, is an iterative process that runs between decision-making and storytelling. The following is an excerpt from an unusually long and difficult report that Antonio worked through during our last session with him. He has gone through the bookmarking process and he is ready to create notes starting with the disease risks he's found important. I have added annotations in squared brackets to highlight details and add context. Figure 4.3 (earlier in this chapter) shows a screenshot of the notes feature taken from the video as he is writing the following note.

Antonio: Right uh, so... let's go here, let's make this bigger...[adjusting the note input area]. Alright, so now here's where the complicated part comes in for this one: *"Your report shows that you have ... shows - that - 27 out of 93 biomarkers associated with"*... oh yeah, and then I will say *"prediabetes are outside of normal range. "These biomarkers include..."* because it seems like there is a strong trend of something... so what I will do is I'll say: *"C-reactive protein, cystatin C..."* and what was the other one? yah, we'll use RBP4 here... You know what? I'll use both of them: *"retinol binding protein 4"* I probably spelled that wrong, but anyways... *"ret-in-ol... protein 4 and..."* What was the last one? Oh, fibronectin, right? Yah... thank you! *"fib-ro-nec-tin, which is strongly associated with the inflammation"*.

I'll add that because it's really important here. Then I will say: *"Inflammation increases your risk of a number"* because they're everywhere right? *"a number of issues including prediabetes"*. Okay, uh.... *"your risk of prediabetes is primarily being driven by inflammation"*. I will say that the fast glucose blood levels are normal. Lifestyle factors...[reading], lifestyle factors such as... I will actually not comment on this.... uhhh no, I will not comment on *these*... I'll let the Action Plan [team] deal with these.

That's it. Then what I'll do is I will copy this thing here [selecting part of the note he already wrote for prediabetes], and then we'll go into *"rheumatoid arthritis"*.

Although their goal is to develop a personalized story about the client's results, some pieces of information like the notes that show up for a highlighted biomarker are automatic text, the same for every case, in contrast to the notes on a disease risk or a function of the body. The latter have more room for the personalization components because the comments at the disease and functional level reference back to the self-reported data or to previous reports if they exist.

The evidence is assessed both, top-down –from risk categories to biomarker evidence– and bottom-up –from individual biomarkers to health risks. But the process of creating notes and highlights is always a top-down approach: they highlight and create notes for the categories of disease, functions, and systems before they highlight individual biomarkers to display to the user. For our experts the disease risks will lead to investigating the underlying biomarkers as evidence and it seems they would expect the client to follow the same trail. Because of that, the last individual biomarkers annotated are those that are important because of their clinical confidence or because they just were not captured as obvious highlights within a specific risk. It is unclear that these reasoning is captured in the interface, or that the client will read the evidence in the same way.

At the end of a cycle of analysis, they go through a review of the outcome they have produced. Sometimes a first iteration in the review takes them back to highlight more information or just to re-organize the content according to what they consider would be “*a better narrative*” for the user, so keeping related pieces of information together. The following excerpt captures Gail reviewing and reflecting about the final output of the report she worked through the second session:

Gail: Let's click on the “Notes” and see what we see... Alright. So, what do we have here? We have pre-diabetes and inflammation... okay. Now we are talking about individual biomarkers, so APO... we have two apolipoproteins, they both indicate a high fat diet, or high dietary intake of alcohol, uhm, ... alright. That's fine. Then we start talking about inflammation, so we have complements... Alright, uhm so let's try and group these biomarkers closer together [changing the sorting of the notes' cards].

So rather than them seeing complement, then something else, and something else, and then a complement again... we try to group it all together. Uhm... we need to talk about the glucose... [there isn't a note for it yet]

Alan: uhm would we group them by their effects, right?

Gail: yeah, effect or sort of like their biological relevance to each other [...] This is what shows up when they first open the page. Uhm, and sometimes I just... I will bookmark as many as I think I need to, and then if I go in and see that, like "okay, yeah I've been talking about complement six times", then I am not going to keep talking about complement, I will remove some of it.

The *health data highlights* stage finishes when the analyst assigns the case to the next responsible in the workflow, an expert in nutritional sciences in charge of creating the diet and supplement recommendations as part of a tailored action plan.

As a summary observation after observing their process and the result of the analysis, we could say that the overarching goal of the health data highlights is *to reduce the information space that a client is most likely to interact with*. It adds a layer in the hierarchy of health information, and the general assumption is that at the very least, a client should pay attention to the information presented in the summary, if nothing else.

Creating Diet and Supplement Recommendations

Let's recall that in the scientific knowledge databases the data team has collected information about the connection between disease risks with each of the individual biomarkers. They have also compiled scientific evidence about lifestyle interventions known to have an impact at the molecular level (see figure 4.5 earlier in this chapter). The evidence about the impact of lifestyle interventions for disease prevention and treatment is more widely understood but entering the personalized medicine revolution promises a level of detail from detection to action, and so their knowledge databases also include evidence links between interventions and their effect on individual measures. The ultimate goal is to go beyond the prescription of therapeutic interventions to be able to predict what effect will the interventions have in the levels of targeted molecular measures.

For Helena, two main sources of data serve as evidence –the phenome data and the result from the work on the health data summary. In her own words, “[*the phenome data*] gives me a general idea of who we're dealing with”. The two sources make up the initial setup in creating a schema of the case she's working on. Instead of reviewing the history of disease in the phenome form again, Helena uses the *health data highlight*

notes created by the team to frame the case of disease or optimization that she'll be dealing with. However, through the recordings we capture that Helena uses more than the data she verbally reports as her sources of evidence. Overall, she uses (1) the phenome demographic characteristics of the individual, the diet diary, and food preferences information, the (2) the health data selected by the team in the previous step. She also looks at (3) a subset of the biomolecular measure results, and make use of (4) her knowledge about patterns for disease and nutritional actions using some standard reference, for example the Canadian Food Guidelines, and (5) her own intuition about what an individual of a certain gender, within a weight, height, and age range looks like. The following is an excerpt from Helena commenting on the data, phenome and health highlights that she is using to frame the case:

Helena: Alright, first uhm, let's check the Phenome form, it just gives me a general idea of who we're dealing with. uhmmm it looks like someone who didn't fill in too much, you can see there's not a lot. Okay.... Uhm, so it's just a 61-year-old male I suppose.

Okay... so let's go back to the "Health Data" tab, and then click on the "Notes" and this basically shows you what the Health Data highlights are... and it tells you what they [team member reviewing data] thought was important. We are trying to align the Action plan with the Health Data highlights.

So, obviously cardiovascular disease seemed important and rheumatoid arthritis... those were the diseases. And now for functions it is inflammation and immune function. Okay let's just scroll to the end and then see that there's... okay and then obviously there were some things that were pointed out, so complements ... okay... this is... like C-reactive protein is high and its' kind of important to know because it's a good indicator of inflammation. So just in my head, I'm like just remembering this, I used to take notes for that, and to just remember to make sure there is a mention of cardiovascular disease, rheumatoid arthritis, inflammation and immune function maybe, when we have the action plan made.

Helena's next step in the process is to check the results of a specific group of biomarkers that she is knowledgeable about and that are relevant for nutritional assessment. She tells us she may find the markers she is interested in under the already bookmarked list, but in two out of the three sessions we recorded with her, she still checked results on the list of all measure results, and under the "nutrition" function in the data report in the *provider view* interface, before proceeding to creating recommendations. When exploring data results, similar to the process that the previous data team member follows, Helena is looking for anything that "stands out".

Helena:[Scrolling through a list of biomarker results] for example what I'm looking for is if there is any of the vitamins that are out of range or any of the other ... like big indicators of nutrition that are out of range, and the only one I see here is choline, although Cesium being high is a weird one, I haven't seen that, but there's nothing we can do about it in the action plan. Yah, okay, that's... that's fine, so let's ... and then... let's go to "biofunctions". And so here if you ... basically what I want to know is how high is nutrition ranked.

Helena: so that will give us good idea as to how many biomarkers that are out of range can be addressed through the action plan and how important it is for this person to change their diet, and it's not ranked very high. Uhm, so it gives me an idea that it's going to be rather difficult to build an action plan because they're actually... they don't have a lot of markers that we are going to be able to address, and then... oh, here... so something that I would note is that choline is low and also TMAO and I know they're in the same pathway. TMAO is a metabolite of choline... I'm finding some patterns. Alright, cool, let's open "action plan" in the new tab.

For the remaining of the process, Helena's task consists of selecting a set of nutritional recommendations based on the highlighted (bookmarked and annotated) data. The outcome of her task will be a set of dietary actions, linked to the relevant bookmarked targets, and completed with details of the rationale to how a specific group of foods would target some of the highlighted results and so why are they relevant for this client.

As we discussed in section 4.2, she uses an information system that allows smart filtering to find recommendations starting by selecting all the targets (the bookmarked measures and disease risks) that can be addressed with nutrition. The system filters diets associated as interventions for those targets, to finally select detailed food categories and foods.

For example, the system shows a mark in 7 targets out of a list of about 15 items that were bookmarked in the previous stage by a different team member. The mark indicates that there are nutritional recommendations in the data base. Each of the recommendations are individual diets that have been studied to have an impact on the desired target. Helena is in charge of identifying common patterns for all the targets she is interested in and making a call when there are conflicting dietary suggestions. The system also shows food categories ranked by a proprietary algorithm she developed and that takes the confidence levels from the diets recommended for all targets to rank and

sort suggestions for food categories like “legumes”, “leafy greens”, or “soy products” amongst many others.

In other words, the system allows Helena to find what are ALL the diets known to have an impact on each of the selected targets and what is the level of confidence of the scientific evidence backing up the connection. Helena is in charge of identifying the common pattern (for example seeing that foods in the *Mediterranean diet* are associated to 6 out the 7 selected targets) and dealing with any conflicting cases when they appear. In the following excerpt, Helena is explaining the process to Alan:

Helena: Okay, so then to build actions, what we have to do is check off these diseases. So, I want to build an action based on prediabetes ... yah, so... anything you see here that has an “N” on it, means that there is a nutrition action to it.

So, complement C3 would be the one, haptoglobin too high would be one, and then uric acid too high, serum amyloid A4, and that’s it. And then if you scroll down ... so here I look at this... [pointing to the diets filtered by the system] so basically it gives me these diets down here ...

Basically it tells me how each of these biomarkers that we highlighted are linked to a dietary intervention and so for example complement C3, if it’s too high, you can lower it by avoiding red meat, and the “1” that’s next to it means the evidence is a “1”, which is the poorest evidence.

Helena: and if there is a “4”, which we don’t have right now, that would be the highest evidence, and then yeah.. you know “3” and “2” are in between! uuhmm so I’m just looking if there are any patterns, so obviously avoiding red meat would be a good one, which also targets inflammation, yeah, it’s a great idea, it’s a 30-year-old male, chances are they’re eating quite a bit of red meat. Uhm... then we have some food sources of omega 3 fats for amyloid A4, for choline. And then uric acid is the Mediterranean diet and then for prediabetes it is just sort of your classic fibre and low glycemic index foods.

She relies in this custom-built database filtering system and the richness of the knowledge database. The underline sentence above is an example of the assumptions that she does when looking for courses of action based on incomplete data. Whenever the client has completed the diet diary and their food preferences, Helena devotes her time in the process to write down all the reasons she used for choosing an action within a specific food category for them. But in the absence of detailed information she uses guidelines she has created for different “types” of reports, for example:

Helena: so... like this one [report] gets a generic list of recommendations, for example... the legumes, they just get the generic explanation, and sometimes you can see they would fit better. uhm I can give some tips for people who say more... if I see someone who in the morning eats a granola bar and you give them oats, then I would give them "try to have oatmeal in the morning" or like I would try to give them some little tips that I can think of... uhm... if I don't know if he already eats all of that.

This was actually quite a quick and easy action plan, because there was not a lot of information, it's like inflammation and diabetes, I've seen it a hundred times.

Finally, Helena reviews the list of nutritional actions she has proposed and arranges based on a "better narrative" for the client. Similar to what happens with the *Health Data Summary*, it is unclear if all the argumentation and her reasoning for organizing the list of actions it's captured and communicated to the client.

Helena: And that's it. And then we go to "review". Uhm, and so here I just try to have them organized right? So, what I do here is... right now we have like legumes, whole grains, white fish... If I've changed my mind I re organize. I think it's fine, but I could change the order of them and sometimes... I try to have the "avoid" action as the last one, just because of the way it displays, and it looks the nicest, but sometimes I feel that it's the first one [chuckles]. And it's also important that the first action actually explains everything, so this one explains why we put fiber and omega 3 fats, and then because none of the other ones explains that, right?

Regarding the underlying challenge of creating a narrative to prompt the client to take action with the lifestyle recommendations, Helena talked to us about her role as a Nutrition scientist compared to that of a Dietician. In her words, a dietician in North America has been trained to give nutrition advice in practice. They receive individuals with a problem that can be tackled through diets; dieticians are enabled to create meal plans for individuals under specific conditions, for example patients of a disease with dietary restrictions. Dietetic practice is also more regulated, closer related to clinical practice in that sense. Nutritionists, like herself, remain mostly in research and are not allowed to practice directly with private clients. "*When it comes to giving advice, dieticians are better equipped to understand the psychology of behaviour change, but a nutritionist like me*" – she says, "*understands the science, especially because you go beyond the undergraduate training.*"

In Helena's opinion, creating and maintaining the knowledge databases requires a biochemistry, nutrition, or molecular biology skillset, but most important, a critical thinking graduate-level skillset to read and interpret scientific evidence. However, on the issue of creating actual recommendations once the evidence is in place, she thinks that a dietician is better trained for the "human link". For the same reasons, she also thinks that once a knowledge database has been curated and has a process for managing its lifecycle, the subset of recommendations for an individual could be created automatically by an algorithm to a large extent, and the focus of the human intervention could be on curating the knowledge and managing the non-information components of behavior change intervention design. In the next chapter I will return to the role of design for interactive visualization technologies in the mediation of personalized medicine communication between providers and clients, particularly in making evident the hidden or emergent roles that scientists and the clients fulfill or fail to fulfill in the service.

Physical Activity Assessment and Recommendations

The last task in the analysis process that produces an interpretation of the health data results and a list of lifestyle recommendations for a client, is to create the physical activity recommendations. Let us recall that John, the expert in charge of this task, has an academic background in biology, regenerative medicine, and is currently completing a PhD program in kinesiology. But John's skills for this task go beyond the biomolecular academic background, he is also a former water polo Canadian athlete, and has extensive coaching history with international competing youth athletes, and with private clients as a personal trainer. John is keen to share his opinions with us about the significance of the "human interaction" skillset first and foremost in this personalized medicine approach because, in his opinion, the molecular biology part of his task can more easily be learned and understood given the effort the team has already put into building knowledge databases. In his own words:

"the molecular markers or the fact that we can quantify them, *that* can be accelerated. And maybe even an easier way to simplify it... the molecular biology is static... it is a snap shot. Versus the recommendations and the exercise regime, that requires more complexity".

Similar to Helena's process in the counterpart of the Action Plan, the information available and relevant for John is: 1) a subset of results of the molecular assessment, 2)

a specific section from the phenome data related to fitness, 3) the results of the health data highlights summary that the team created in the former steps and, 4) his knowledge and intuition about the physiology of the human body for training. In the quote below he explains to Alan the rationale behind the schema he is initially creating for the case starting with a review to the *health data highlights* created by another team member:

John: we're going to open up three other windows. We're just going to open up... one window for "Action Plan"; one window for "Phenome Form"; and one window for "Preview Client". Okay. So, we'll click that last window we opened for "Preview Client" and we're just going to see what our team has passed off onto us.

Because this is uhm, this is the end of the line... so to speak for the report ... the report gets put for review and then gets sent out to the client and so we want to be able to tie it in with what they have addressed as health risks. And so here the only health risk the [data team] really addressed for this person because they're healthy and none of their health risks have jumped up into the moderate zone, is nutrition and immune function, coagulation and healing, but none of these seemed to have jumped so high that they cause a health risk, so we really don't have to worry in terms of any restrictions for the exercise, but we can use some of this information to guide what we would suggest.

John runs his analysis with a significantly reduced subset of biomarkers. From the list of about 300 proteins and metabolites, John begins his task instructing Alan to check the results on less than 10 that he is interested in. John's process is also different to all others in that he creates the schema for the case starting with the biomolecular data results. In his own words, he "*use(s) a set of biomarkers to create the profile of the person for fitness. That is, to assess whether they are currently exercising or not and where do they lie within terms of fitness: do they lie with athletes, sedentary or disease profiles*". Here is a brief from the beginning of the first session recorded with John:

John: so, you know you're dealing a little bit backwards here with the exercise recommendations. You're seeing a blood profile and suggesting what they do versus the normal approach is you see them, and you can give them a physical assessment and then give the recommendations, but uhm... you know, there are a lot of advantages that we have to this insight, is that it's really not biased in the sense of okay we're going to get you to do stuff that has worked for everybody else... uh, yes we do stuff if this has worked for other people in literature, this has worked for other people we've worked with, but it is tailored to your unique profile...

John: so, you don't have to do a fasted exercise unless we actually detect any rationale for you to do a fasted exercise. Of course, there are benefits of doing it, in terms of growth hormone, in terms of

testosterone, in terms of hormone balance for anybody, uhm... but here is you can actually get the specifics: "are you going to benefit more than the other person?"

John's process is also different in his use of the reference ranges for assessing the importance of the measures. John uses the standard reference ranges for an initial assessment of disease status, and then he applies a different range to make an assessment of fitness. These extra set of ranges are extracted from the literature that studies the molecular biomarkers with control groups such as professional athletes vs. non athletes, or different levels of athletic performance.

According to John, this means that if the biomarker is outside the reference ranges, this puts the biomarker into the "*disease*" evidence category, and it cannot be used to understand the fitness profile. But if the measure of interest falls within the reference ranges, then John applies a second "fitness range" to evaluate what he needs from the measure. For example, in the following excerpt, what he means when he says that Cystatin C "*should be low*" is better read as "*should be on the lower end of the reference range*". Which is, he looks that the concentration level is within the reference range, and then on which end of that range.

John: we're going to go to 'serum P', so, serum P is in the disease range. We can't really have an idea of whether this is associated with fitness or not, but with fitness it should be on the lower end of the normal range. Because it is in disease, we don't take it into account.

John: Now, Uhhh... if we look at cystatin C... cystatin C should be low in people who have good muscular strength. Good muscular strength obviously can come from both an endurance-based or simply a resistance-based regime, and we see this person has through the roof... amount of cystatin C, and this one is associated with inflammation, so even though in some athletes it might be great and they should be *down here* [pointing in the screen], because they have inflammatory or systemic inflammation... it can go higher, but it usually doesn't exceed such a high range. So, this person again, can't assess this for fitness, because it is so heavily in the disease range.

From this assessment, John creates a frame of reference for the case. After he has checked the biomarkers of interest, John now reads into the Phenome form. He has designed and refined a set of questions to learn details from an individual's fitness. He combines his knowledge on human behaviour (as a coach) to interpret the answers and correlate the patterns of how people talk about their physical activity. For example, in the following excerpt he is explaining to Alan how he uses the answer to three different

questions: (1) the first question asks clients to list their physical activity routines. We mentioned earlier that instead of trusting the reported frequency and history, he looks at the types of activities people report (see part 01 below). (2) Then he combines that answer with a second question that asks the client to rate themselves in aerobic fitness and muscular fitness and (3) finally he looks at a question about the fitness goals. The last 2 are captured in part 02 below.

Part 01

John: And so, we go to “Exercise and Energy Levels” next. Uhm, and then we kind of take a look at what does [the client] do... Unfortunately, when people report their current exercise, this is the least accurate way of getting the information. ... it’s extremely inaccurate. More often than not people will think back to the last year, if not years, when have they done anything, if ever anything... and then they put it down, right? I mean, it is just the way it is when a human is asked...

There are very few people that can be uhm... uhm.... honest, care about it, and understand what we’re trying to capture with it... and so this person says they do elliptical... that’s all we really take, is okay, they’re already on something that they’re familiar with. We’re going to go with elliptical and recommend, in terms of types of exercise but we’re going to kind of ignore how often they say they do it and for how long...

Part 02

John: And so, the next thing we do to actually get an idea of how fit and how the person feels about themselves, we just quickly scroll down to the self-reported fitness score [scrolling]. So, they say they are... aerobic fitness is a 1 and muscular fitness is a 2. [The scale is] “poor, fair, good, very good, excellent”.

So, before, he gave himself how much exercise? but this is... for some reason a little bit more informative of their current state, because more often than not people are going to report if they’re not in a good shape or if they’re in good shape. If they’re in very good shape, they do one of two things... They either report they’re ‘excellent’ or they’re so hard on themselves they say ‘no, I’m only good or very good’. But which either case... it still gives you an idea of where this person is in terms of comfort.

The next thing you do is you go back up... we look at this statement “*I am physically active once in a while, but not regularly; I intend to start doing more regular within the next six months*”. So, you can see that what they reported here, doesn’t coincide with...

Alan: oh right????!!! So, the elliptical, and the gardening, and the walking and... how interesting.

After surveying the information from the three sources, John is ready to create a list of physical activity recommendations. Using the same tool that Helena uses for creating nutritional actions, John starts by selecting all bookmarked conditions and biomarkers that can be tackled with exercise activities. He has also created an underlying knowledge database that links physical activity to the change in levels of individual biomarkers and diseases. During this part of the process he goes almost automatically asking Alan to select everything that exercise can bring back to its expected levels. The list of bookmarked information and the flags for exercise and nutrition looks like figure 4.11. He takes most of the session with us giving us an explanation of how exercise tackles each of the biomarkers that Alan is clicking on. It is a rich intense discussion on the molecular biology component.

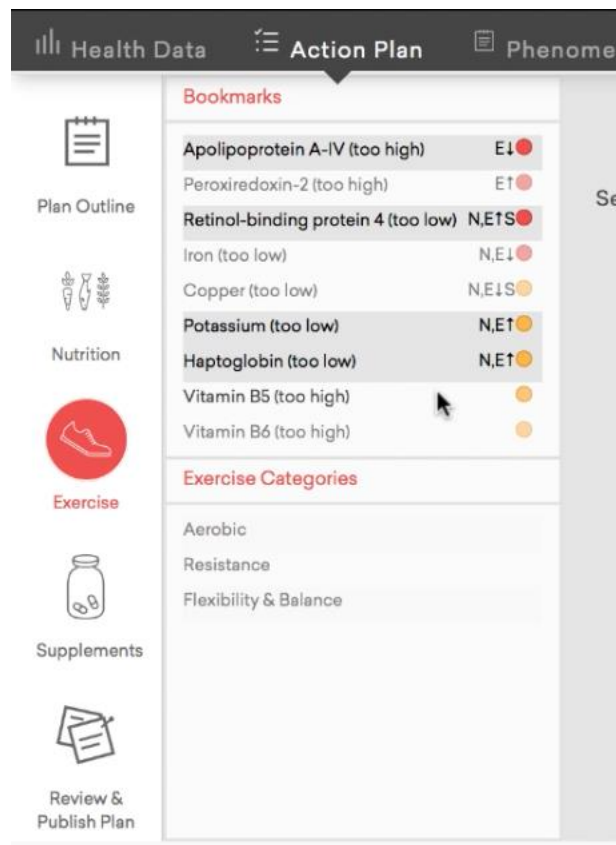


Figure 4.11 List of biomarkers and diseases bookmarked with flags for the action plan team.

Note. The bookmarks' list shows the biomarkers and diseases selected by the team in the former step. The flags on the right are the signals for the Action Plan team to recognize which of those can be targeted with Exercise (E) and with Nutrition (N) according to the curated knowledge databases.

John creates between 3 to 5 actions and additional notes that he uses to explain to the client the rationale behind every decision in terms of activity suggested, intensity, duration, and even the type of training to perform. John brings to his recommendations what you would expect from a trainer, he goes beyond evidence linked to biomolecular impact and takes into account, adherence to exercise, motivations and the goals listed by the client. He also aims to provide details to the extent that he would when seeing a person at the gym. Through this pair-instruction analysis data collection we could capture the extent to which John goes about in his rationale for selecting a fitness recommendation, however, we can also see that most of the richness of the explanation does not make it to the client using only what they have at hand in the tool: written notes.

In this example, John goes at length into explaining “muscle antagonism” in the context of muscle stiffness, and the rationale to recommend yoga:

John: the muscle antagonism is a little bit of ... you have to have an education in fitness to have it make sense. But all your muscles exist and work in terms of an antagonism. And so ... in the arm your arm can flex and extend based off your biceps and your triceps. And so, your muscles only do one thing, they contract, and so for you to have movement, you need another muscle to contract...

So, the other muscle will stop contracting... and so what happens to us, is that we have a tendency to grow certain muscles stronger than others, and they will then overpower those weaker muscles when we do these big movements.

Yoga does a great job of putting us in ranges of motion where we have to engage those weaker muscles, and it helps us identify which areas you can actually target with strength training.

John: I'd explain this in the gym... so I have no idea, because I don't see these individuals, this is the best way to get them on track to be able to help them help themselves.

Alan: like a self-assessment. That's great.

John: and so, I tell them look for power yoga or flow basic to increase movement and less static holds, I would say the opposite to this individual. Uhm... the Vinyasa I was just talking about, a more of a strength-based yoga.

The sessions with John bring forward more clearly the topic of behaviour change and the roles involved in the provision of care for an individual in the personalized health model. The scope of the case documented in this dissertation ends with the creation of lifestyle recommendations and the health data interpretation summary. The data team in charge of this process goes as far as creating the recommendations and we could say that is in hands of the client (the health consumer) to manage their own behaviour change and lifestyle adaptations. There is a myriad of research into digital health technologies for behaviour change interventions, but this is out of scope for the service at the time of documenting the process for this dissertation. Equally important, we can see in the pair analysis data that for the scientists involved in the creation of the summary and the lifestyle recommendations, the knowledge in human behaviour and the interest in addressing a client's needs to promote change, goes along with the analytic task they perform with biomolecular data. The understanding of human behaviour is as relevant for some of them in performing the task as it is their biomolecular background. This was more evident in the sessions with John and Helena but also present in the work of the health data summary experts when they take into account their knowledge about how people talk about health and the issues regarding understanding "risk" for the general public.

The following two examples present the rich rationale that John is providing to a client and why he considers it so necessary to go into the details. Comparing the verbal explanation that we receive to what he captures in the notes, it is unclear if the rich argumentation will be clearly communicated to clients.

Example 01

John: [they have selected all the measures that will be targeted with exercise] After we have clicked all that, we can now click on "Exercise Recommendation". We're going to go to "Aerobic", and now we're going to take everything we've seen from the profile and sum it up into a recommendation for this individual.

So, we're going to write for example, instead of all of these things [the examples in the template] we're going to write first "elliptical". And the next thing we're going to say is "brisk walking". Why I choose brisk walking over the other ones first? uhm... is because.... uhm ... "easy does it".

Right, When the person sets really high goals and are hard on themselves, they're not going to continue, and people think that "*if I'm hard on myself, it will be better for me in the future, because then I'll stick more to it*", but it actually has the opposite effect on our behavior. It is when you are compassionate to yourself that you are more likely to stick with something, and you are more likely to continue that regime. Brisk walking – because he already said that he does walking... brisk is for the help prompt, yeah, it's... you're not just walking, you can do something else within your walking regime.

Example 02

John: so, why do we say what we said? What we've said is everything we've said just to get this person motivated into doing this on a regular basis. The hardest part for exercise with people is commitment, is doing it. And so, there are lots of ways, there are lots of tricks to make yourself do it, and there hasn't been a single person that has done exercise and has said "*I regretted doing it*".

right? And that's my time as a personal trainer and that's my time with, you know, everything I've done.

4.4. Summary

In this chapter I went through an in-depth description of the process that experts follow to complete the interpretation of biomolecular data for each individual report. My goal was to address the richness of the socio-technical system at play. With this chapter I have addressed the descriptive component of the research questions I proposed.

We looked at the details of categories of evidence and the forming of confidence to assess evidence. In the process observed we found expected attributes of confidence that turn a piece of information into health evidence: scientific validity. However, in the recorded process, evidence from scientific research was no more than one component of any decision. We captured the heavy influence of less common attributes beyond the scientific standards, that is, the clients' goals and preferences, and the actionable capacity in health information in relation to answer to client's goals and to empower behaviour change.

Connecting the findings with what is described in the Evidence-Based Medicine (EBM) literature, we uncovered a process where an individual's goals, preferences and self-reported health information play a key role as evidence in the decision-making

process. EBM is described as “a systematic approach to clinical problem solving which allows the integration of the best available research evidence with clinical expertise and patient values”, and aligned with the models in the literature, we recorded a process that weights evidence from scientific literature, as well as patients values, and expert judgement.

In the EBM literature of clinical decision-making, expert knowledge is described as the “*experience, expert opinion, clinical skills, or expert judgement*” that is gained by practitioners in the course of clinical interactions, in contrast with knowledge gained from sources such as journal articles reporting on the findings of RCTs, or meta-analyses and systematic reviews (Wieten, 2018). As expected, in the process we documented in this chapter, the scope of expert judgements goes beyond the understanding of scientific evidence. With varying degrees of involvement, each of the participants in the workflow used their knowledge and expert intuitions about human behaviour to complete the sensemaking tasks. We reviewed instances such as (1) applying knowledge/intuitions about the communication of health information, (2) understanding the emotional / psychological component of a person’s attitudes towards *risk and uncertainty* in health results, and (3) expert knowledge of the psychology of behaviour change at play.

The most important takeaway is the fact that we are looking at a team of *analysts* putting in action their human-interaction skillset, as a health provider would do, but in a setting distant from the traditional clinical scenario. The novelty in our case is the seamless translation in the process. We observed attributes of the EBM model emerging in a technology-mediated, preventive approach to healthcare, outside of clinical settings.

Furthermore, the *actionable capacity of evidence* was a new emerging key attribute. Similar to what we may qualify in the design-lingo as a human-centered decision-making, the actionable capacity in the evidence, turned this process towards a human-centered approach to preventive health with biomolecular data. Confidence of evidence was assessed based on standard industry metrics for scientific validity (applying a threshold to judge the quality of evidence based on the underlying study methodologies), but more importantly, identifying evidence that can translate to action, agency and participation by a client seems to be the signature of the preventive approach documented in this dissertation.

Similarly, from our earlier discussion on evidence types in the evidence-based model for clinical decision making, and the observations on the works of the group of experts in this case, we can see that there are different schemas that guide the user-centered analysis work of experts using biomolecular results. While the data results are organized into a risk-assessment schema, finding a personalized health narrative that suits the goals and preferences of the client seem to be a strong model to drive decisions.

It seems appropriate to recall what Cognitive Scientists, Philosopher and precursor of the Design Sciences, Herbert Simon denoted as the professional *designer*:

Everyone designs who devises courses of action aimed at changing existing situations into preferred ones. The intellectual activity that produces material artifacts is no different fundamentally from one that prescribes remedies for a sick patient or the one that devises a new sales plan for a company or a social welfare policy a state. Design, so construed, is the core of all professional training: it is the principal mark that distinguishes the professions from the sciences (Simon, 1988).

As a Visual Analytics problem, I set to study factors related to sensemaking, hypothesis testing, information foraging, and data visualization technologies, that are associated with the courses of action that experts take in their observable decision-making process. We have discussed some important attributes of evidence and the role of expertise in human behaviour, beyond molecular biology, as factors driving the decision-making process.

In chapter 05 I will continue with the analysis of the communication between experts and their clients and conclude with the implications that all the discussed attributes have in the design of direct-to-consumer health information technologies.

Chapter 5. Joint Action Theory and the Client-Analyst Relationship in Direct-to-Consumer Personalized Health

From the emergent categories in the grounded approach to my data, I became interested in three main ones in relation to the driving research questions: (1) the sense-making loop theme, covered at length in chapter 04, captures the nature of the decision-making process, the tools, evidence sources, and warrants in framing a case. (2) I found the attributes of evidence: *confidence*, *trust*, *familiarity* and, *actionable capacity* to be highly exploited communication and decision tools in the process. This topic turned into a relevant category to discuss the factors that influence evidence-based decision making for experts in the domain of personalized health. This was also discussed in chapter 04. Finally, (3) from a human-computer interaction perspective, I became interested in the observable gaps in the communication of results, particularly, of the gaps in the communication of the argumentative warrants that are part of the results of an analysis process.

As cognitive psychologist and father of the Distributed Cognition framework, Ed Hutchins states, “the need for a framework that helps us understand cognitive systems larger than an individual become even more apparent when we consider how cultural factors may affect performance characteristics” (Hutchins, 2006). In this chapter, I demonstrate the power of taking the pragmatics lenses to the particular account of human interaction between the analysts and their clients, a complex situation where computations (or cognitions) occur both internally and externally as the interaction occurs. This is the basis of the Distributed Cognition approach. The appearance of “the third person in the room” category in my analysis invited for a dissection to look at how is meaning achieved between these two actors in their particular mediated setting. As language use researchers state, I became interested in looking to explain how the gap between sentence meaning and speaker’s meaning is bridged.

Distributed cognition is a powerful theory to set the units of analysis in complex situations beyond the individuals. In this sense, as a researcher I treat the activity as a socio-technical distributed system where patterns of information flow in cognitive artifacts as representations. However, as a theoretical framework, Joint Action Theory provides a

robust set of constructs to look at the attributes of the setting and the interaction of individuals participating in an activity. Ed Hutchings incorporates some of the general concepts of pragmatics and language in use in his own analysis. For example, in his work that examines the instruments and procedures found on commercial airline flight decks, Hutchings uses common ground, indicators as signaling mechanisms, and redundant processing, as constructs to describe the actions in the cabin (Hutchins, 1999).

So, while a socially distributed theoretical approach, sets the stage of the units of analysis to look at the complex system, deepening into the attributes of communication, meaning creation and negotiation, researchers have a more robust decomposition method with Joint Action Theory.

In my pair-analysis data there are sounding observations about the extent of effort that experts make to capture and communicate the rationale of their decisions, something that seems as a priority to provide to their clients and their care teams. There is, however, a discernible disparity in how such efforts materialize in the resultant product. With this in mind, in this chapter I will propose that in the direct-to-consumer model documented in this dissertation, the analysis conducted by experts can be seen as a *joint activity* occurring between the analysts and their clients. As such, we can use the constructs of “common ground”, “advancing a joint activity” and the “methods of signaling” to gather an insight into how the activity is advancing and how to characterize the gaps in the mediated communication. This in turn has implications in the design of evidence-based visualization interfaces for personalized medicine products in a direct-to-consumer model.

In collecting pair analysis data, I intended to capture the richness of interaction between the analysts and the interactive tools they use to navigate the complexity of their task and dataset. As a member of the Visual Analytics research lab that gave origin to the Pair Analysis methodology, we use H. Clark’s framework of Joint Action Theory to look at instances of collaborative analysis. That is, multiple analysts, often from with different expertise areas, developing meaning from data while interacting with visualization technologies. While not originally intended, the emergence of “*the fourth person in the room*” became a relevant occurrence of the workings of language use as a different potential *joint activity*. Instead of looking at the activity between analyst-intern,

the analyst-client activity is one of particular interest because of its prospective implications for the design of services and solutions in the developing personalized medicine industry, and the booming of its direct-to-consumer products.

5.1. Joint Action Theory and the Creation of Meaning in an Asynchronous Distributed Process.

Herbert Clark's Joint Action Theory main proposal is that language use is an instance of a broader category of human actions: joint actions. As we discussed in Chapter 02, a joint action is one that is carried out by an ensemble of participants in coordination with each other (Clark, 1996). In Clark's theory, the coordination of actions happens at the level of content –what participants intend to do– or at the level of process –the physical and mental systems used in the execution of the intentions. Arias-Hernandez et al., extended the use of the framework to include the study of interacting with technology, specifically the role of visualization technologies in joint actions: “*since collaborative visual analysis is an instance of a joint action, the starting theoretical point to apply joint action theory to visual analytics was that: humans will use language and will work together to solve coordination problems in collaborative, visual analytics*” (Richard Arias-Hernandez et al., 2011). With JAT, in this chapter we are interested in taking a perspective on how a group of experts collaboratively build a “document” as the summary of the data interpretation and a set of recommendations for the client.

In chapter 2 I introduced the sensemaking loop for intelligent analysts, we described the sensemaking loop consisting of higher order cognitive functions involving information manipulation to answer goal-specific tasks. The model is safely described as highly iterative. During the sensemaking loop (after the foraging loop), experts go through the schematization tasks, hypothesis testing, and the development of the case to tell a story, reiteratively. The case documented in this dissertation can be easily seen as an instance of that proposition. During the coding process there were evident instances of the goal to “*frame a story*” as a task that takes place from the beginning of the analysis process. The story in this case, is built from the bookmarking, annotations, and selection of lifestyle actions by the analysts. The “*presentation*” artifact with the end-results from the analysis, is a mash-up of the former rather than a newly developed deliverable. What I propose in this chapter is to look at the client as a more active agent in the task. Addressing client's goals, in a particular way that resembles face-to-face

conversation, or “talking back to them”, remains a core effort for the analysts while they work through the analytic endeavor of interpreting health data for an individual.

For that reason, following Clark’s proposal of Language Use, what I describe in this section is an attempt to understand how the design of visual analytics and health reporting tool can be better informed not only from the individual requirements from analyst and client separately, but by looking at the performance of their situation as a *joint activity*.

The Expert Analyst– Client Relationship as a Joint Activity

*Being in the same transaction is like **believing** we are in the same room at the same time. — H. Clark (1996, p. 36)*

I propose that an instantiation of a Joint Activity emerged in my data as the mediated interaction between the team of experts interpreting data and their end user, the health consumer. Let us begin with the basic question of how it is possible for two individuals to participate in a joint activity asynchronously, with little to none knowledge of each other, and without being in the same room.

We discussed that with Clark’s framework we can characterize the settings of the activity taking place. Although the spoken setting is the richer, and most obvious setting of language use, the range of instances goes beyond conversation, and includes “reading a novel”, “putting on a play”, or “talking to oneself”. The classification of the written summary as a joint activity in this case can be seen as an activity occurring in a setting that is mixed in nature: partly written, partly diagrammatic, partly interactive. It involves the communication of both personal intentions (from the client to the provider), and institutional intentions (the experts do not respond about their personal intentions but represent the institution and the service).

Clark specifies that in written settings messages are directed not at individuals known to the writer, but at a *type* of individual, such as “clients that are dealing with a chronic condition” or “clients that love data and are looking to optimize their health”. The activity does not have to occur in synchronous time in order to be a joint activity. In asynchronous Joint Actions (1996, p. 90) particularly in writing and reading, participants coordinate mostly in content. In conversation, speakers and addresses synch the

phases of their actions. In asynchronous settings, speakers try to make processing optimal for their addresses.

Let us revisit the features of face-to-face conversation as presented in chapter 02 (see table 5.1). Using these features as reference, I will describe the attributes of the settings in our case.

Table 5.1 Features of face-to-face conversation. In (Clark & Brennan, 1991)

1	Copresence	Participants share the same physical environment
2	Visibility	Participants can see each other
3	Audibility	Participants can hear each other
4	Instantaneity	The participants perceive each other's actions at no perceptible delay
5	Evanescence	The medium fades quickly
6	Recordlessness	The participant's actions leave no record or artifact
7	Simultaneity	Participants can produce and receive at once and simultaneously
8	Extemporaneity	Participants formulate and execute their actions, without preparation, in real time. (Notice that this does not refer to the content but to the sequence of actions in the conversation)
9	Self-determination	Participants determine for themselves what actions to take when
10	Self-expression	Participants take actions as themselves (different to an actor speaking for the intentions of a character, or a politician presenting the voice of an office).

One immediate difference is that the attributes of *immediacy* (features 1 to 4 in the original table) are entirely gone given our setting is *remote and asynchronous* in nature. The *medium* (features 5 to 7) offer differences that can be taken into consideration of collaborative technology design. The most important characteristic to denote is that in this setting, actions, and signals *only exist to all parties when recorded*. Finally, the attributes of *control*, Clark estates in his book that “*the less control participants have over the formulation, timing, and meaning of their actions, the more specialized techniques they require*” (p. 11). Our situation is one where most individual actions are scripted, participants operate on a timeline that prompts what needs to be done when, instead of jointly determining who says what, when, as what happens in face-to-face conversation. Moreover, as I will discuss later in the chapter, the current *control features* are ambiguous because there is no consensus on the type of relationship we would be interested in modelling: is the expert-client activity one that naturally embodies a power imbalance with a provider prescriptive behaviour; or does it promote higher mutual participation? Table 5.1 summarizes the features of the mediated

expert-client settings of communication, compared to the standard face-to-face conversation.

Table 5.2 Features of the setting of mediated DTC services viewed as a joint activity.

Mediated Personalized Medicine Service	Face to Face Conversation
<p><u>Remoteness</u></p> <p>Participants in this activity don't share the same physical environment. They don't see nor hear each other at any time.</p>	<p><u>Immediacy</u></p> <p>In face-to face conversation the immediacy of the setting is described with: Copresence, Visibility, Audibility, and Instantaneity.</p>
<p><u>Asynchronous, Recorded Medium</u></p> <p>The participants perceive each other's actions with <i>delay</i> and require <i>awareness aids</i>. More importantly, the actions only EXIST for all participants when <i>recorded</i>.</p>	<p><u>Simultaneous, Evanescent Medium</u></p> <p>In face-to face conversation, the medium was described as: Simultaneous, Evanescent, and without Records.</p>
<p><u>Control Features: Institutional Expression</u></p> <p>In our mediated settings, the actions of the activity are mostly <i>deliberated</i>, <i>scripted</i> in the order they occur.</p> <p>Participants express in conformity with a business transaction on behalf of an institution and are limited by the institutional rules: Experts are providers that do not speak for themselves, but that act on the role of an information provider. Clients do express as themselves but under the etiquette of a provision of health model.</p>	<p><u>Control Features: Self Expression</u></p> <p>In face-to-face conversation actions emerge extemporaneously and represent participants as themselves. The features were described as: Extemporaneity, Self-determination, Self-expression</p>

Figure 5.1 depicts a snapshot of a temporal representation of participants in the activity. A couple of experts and a client enter the activity at different times and contribute content to the scene. The middle area represents the interface they share as a medium for communication. Arrows point to the scripted actions that each participant is expected to contribute for the duration of their engagement. An important question at this point is to what extent are the consumers aware of the *analyst-provider role* and their interaction capacity with them? In a traditional doctor-patient consultation, the settings of the scene and participant roles are clearly exposed. But as we discussed in chapter 02, even within its traditional notion, the practice of medicine has evolved in relation to information keepers, power dynamics, and the modality of participation (passive patients vs. active)

etc. In our personalized-medicine provision model, the roles are not obvious, and the mediated scene may further hinder this issue.

A major proposition in this representation is then, that the medium needs not only to provide the means for each participant to complete their individual actions at hand at any given time, but it also needs to accumulate what is necessary for all members to advance in the activity, a representation of the states, initial, current state, and public events so far. Particularly, we discussed the initial agreement, the *handshake* to enter the activity, to make evident what is the public state of common ground entering the activity.

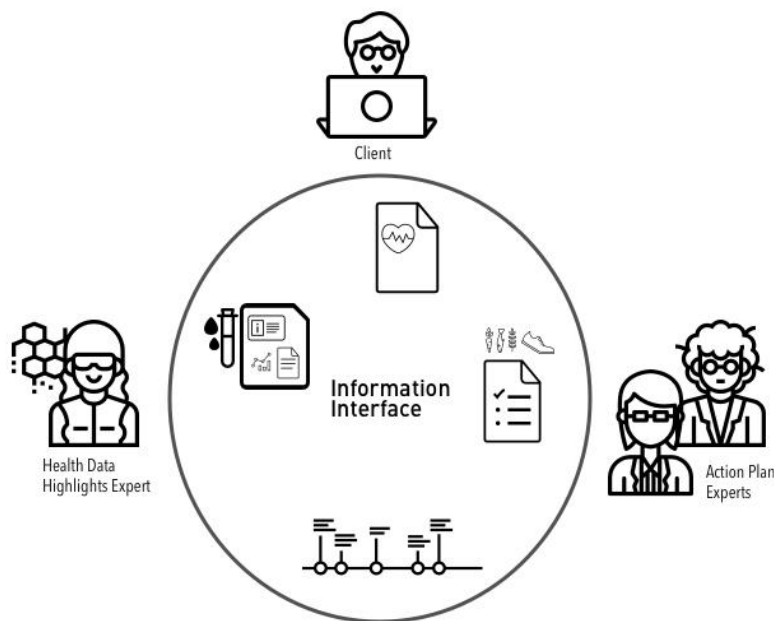


Figure 5.1 Experts and Client coordinate on content in the interface. Each participant contributes pieces at different moments in time.

Advancing Joint Activities

In entering the joint activity our experts assume that they are performing and optimizing the processing for the clients. “*Being in the same transaction is like believing we are in the same room at the same time.*” says Clark. Let us take a look at some of the verbalizations that reflect the experts’ clear motivation to attend to an individual’s needs. In the first case, Antonio reads the phenome form in a simulation of what the client is telling him, and a simulation of what he would answer to that. That is, as if he

portrays the client in the room. In the second example, John explains why he goes so at length with the exercise written summary. He is compensating for the obvious shortcoming of not being able to see his client, moreover, he is not able to *follow up* with his client in the way he would do with a trainee.

Example 01:

Antonio: They're just like okay... "you know here's why I did the test" or "here's why I have an issue" or whatever and that really – this really guides me into how I'm going to write the story of the report, right? How am I going to interpret it?

Because I really want to answer their question – like, if this individual, if they're just like "I just want to achieve my best and sustain my wellness" so this tells me that you know what? They more or less think they're healthy... so let me see if I can back that up for them, or if they have some concerns, I will address their concerns in that sense. Right?

Example 02:

[Expert has gone at length about specifics of the exercise type and his rationale for each decision. Researcher and Expert are reviewing the written end result].

Researcher: This is one of the most detailed instructions that I've seen you give, and I wonder is it because he is healthy, and you can get more into the specifics of the training?

John: even for people who are not necessarily that comfortable with it, we can give something like this, but there is just less chance I will. [Because] there's usually other things I want to address, and so I can't write you know, an essay on how to undergo from one to seven, I have to go one to two ...

The hard part is you don't get to see them, you don't know if they're doing it right, you have no idea. You are hoping that they will take this as a.... uhm... like a trigger, for them to actually go and find out how to do all this stuff

I have no idea, because I don't see these individuals, and this is the best way to get them on track to be able to help them help themselves.

If joint activities are cumulative, what do they accumulate? Clark's proposal is that they accumulate *common ground*: joint actions accumulate the common ground of the participants in the activity - the knowledge, beliefs, and suppositions they believe they share about the activity (1996, p. 38). Let us take a look at the different stages of common ground in this activity:

Initial common ground: One strong assumption is that of the *roles* of participants. As a health service, consumers approach the service with the expectation of that of a service provided by health professionals, but as a DTC lifestyle intervention service, they are also aware of their participation and responsibility. Both experts and clients are aware of a shared responsibility, although depending on the onboarding process, the clear scope and tasks may still be unclear. Brendan Byrne, a BC based primary care physician and digital health entrepreneur, describes this assumptions as the “health jobs” in digital care, or “the specific tasks that are being done as part of the provision of care for a person” (Byrne, 2016). Byrne specifies that these health jobs may or may not be specific to a disease or diagnosis; they may be done by health professionals for patients; or they may be done by consumers themselves. A clear understanding of the tasks that each of the roles are responsible for, is part of the initial common ground. As we have described in detail, our experts are aware of their role of *providers* and that part of their tasks interpreting the data is to reduce the information space and provide confidence handles on the results. Besides the roles, the DTC model, the expectation of data and consent provided by clients and returned to clients is part of the common ground.

Current state of the activity: In chapter 02 we discussed that external representations are especially valuable as a medium for the joint activity. The information interface in this case is a recording environment where the changes of status take place and notifications get shared. On the client’s end, the interface provides a timeline that displays the status of the analysis process and prompts the client to action when required. See figure 5.2. On the expert’s end, however, we arrive to the data analysis without a clear understanding of whether the client has completed their phenome information, or how engaged they were in the process of doing it. Besides the content provided, gathering information about what the client considers more important to share, or what pieces of self-reported information are confusing, are relevant attributes to develop common ground. We observed Gail, Antonio, and Helena working on cases where they had made an inference about whether the client had: partially completed, not started, or fully completed the phenome information.

Public events so far: Besides the aforementioned, there is an interesting question on investigating if more context-awareness features that keep record of the events so far, and more updates about the process would result in higher trust from a client perspective.

Please complete your order

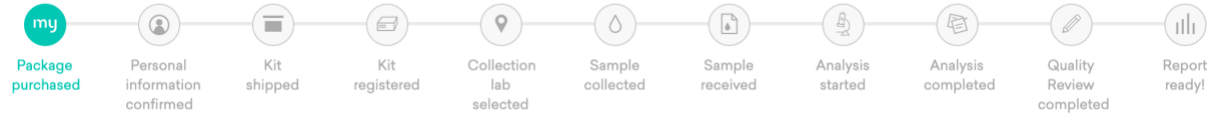


Figure 5.2 Timeline of events with call for action in the Client Interface

Moreover, the situation in this dissertation is an interaction that accumulates common ground, not at all times with an individual, but with *types* of individuals in the role of the health consumer. In creating a story for a client, Antonio has set his mind to address pivotal pieces given to him in the phenome form, he advances through the assumptions of the goals and intentions of his audience. On the other hand, the team has received feedback about the expectations and the knowledge of the clients, and they accumulate that for the activity as well. For example:

Example 01:

Gail: Heart attack is a scary one to try and bookmark because that's like a very scary outcome to try and tell people that they're at risk of. Uhm... and unless they have something like C-Reactive protein, which is kind of a clinical marker for a high risk of heart attack or cardiovascular disease, we usually try and highlight things that are more chronic [not heart attack].

Example 02:

Helena: so, we've got the feedback that when people see that they're like "oh my God I'm at risk for cardiovascular disease, what am I going to do about it?" and so it's good if we have it in the action plan right? because then they're like "oh okay, that's what I can do about it".

Yeah, and so it seems like the diseases and the body functions [systemic functions] seem to worry people the most.

Example 03:

Gail: [she is describing to Alan a former practice about providing information to the client about all highlighted biomarkers]

For **each** one of those highlighted biomarkers we have paragraphs to talk about them and it was ... the feedback we got back was that it was really overwhelming for people. And people tend to focus in on ..." oh my God I have this one biomarker that is associated with depression. Should I see my doctor for depression?" and it's like ... no big picture...

you have other things to worry about... uhm so that's why we came up with these guidelines for the biomarkers.

And so, if we are to assume that our participants will coordinate in content, what are the coordination devices that can be better supported via the design of interactive interfaces for biomolecular data and health reportable outcomes? One direction is to look at attributes in the text content but equally important will be to look at the non-linguistic methods for signaling that convey meaning via *interactivity*.

Coordination Devices


Considering the mediated settings, from the coordination devices described in chapter 02 such as *conventions, joint salience, precedent etc.* required to advance common ground according to Clark, the most obvious feature in our case are those of *perceptual or joint salience*. An example of an existent coordination device is a timeline in the interface that displays to the client the status of the analysis process. Figure 5.2 shows an example of the full journey for a client, the timepoints demark calls for action or notifications of status. As we mentioned earlier, there isn't an equivalent mechanism that communicates to the experts the status of clients.

On the other hand, despite the client is prompt during different times before the analysis of results to provide their health information, the consumer participation involves a continuous *learning* engagements⁶. Currently, there are no features for experts, or other providers to be aware of the learning process by the client. As part of our initial findings with consumers (*Visualization in Practice: Personalized Health as a Human-Centered Design Process at Molecular You*, 2017), we gained the understanding that, an experience for a consumer is enhanced with the sense of *learning something about themselves*. This goes hand in hand with the experience of health as *a moving target*. For some "healthy" participants that received results indicating little concerns about their general health, there was a sense of powerlessness and surprise from not having a biomolecular signal to represent the experience of their own health status. They expressed a sense of disconnection between the results in the data and their daily

⁶ Not originally intended and remained out of the scope of the thesis proposal, we did not include user studies conducted with clients of the organization. However, as I have mentioned earlier, as part of the product team at the organization, I designed and participated of Focus Groups and other user studies conducted with end consumers.

experience of themselves. For these participants, reading the results felt like they needed to keep doing what they were already doing to stay “healthy”, but this had no new learning about themselves.

These considerations are quite relevant when it comes to *signaling* the argumentation of results for an individual, and for the design of better coordination devices between experts and the client. The final deliverable is a composition of rich written justifications to back up each area identified as important. In the action plan, each of the actions in the list of lifestyle recommendations, also presents rich text content. What is relevant to our case is that experts review their process asserting that the different states of evidence and the warrants of the decisions, are *explicit* in the written text. However, as presented in Figure 5.3, the literal pieces of information miss many of the decision attributes. In most cases, the text may contain references that show that experts have paid attention to the client’s goals, to his/her history of illness, or family history reported. But there are gaps in presenting the argumentation that they were so eager to describe to us. In the top image, “brisk walking” is part of a list of example activities, but there is not a direct reference about our expert arguments: “brisk walking is part of what they reported doing already. Easy does it.” Similarly, there is a list of biomarkers presented both in the action plan and the health data highlights summary, as “relevant”. But there are no indications of what are the attributes in the evidence that makes this particular set, more important than the rest. We have discussed at length that some pieces of information stand out due to the scientific confidence, while other pieces are selected because of how actionable or familiar they can be. Finally, there are no mentions to the self-reported information used as evidence in the assessment of risk, something that would leverage context-awareness of the decision process for the client.



ENDURANCE TRAINING
[4 time(s) weekly]

Examples: cycling, swimming, circuit training, aerobics or brisk walking

Max heart rate: 40-80%

Low Beta-Hydroxybutyric Acid levels inform the body is not burning FATs adequately. Longer bouts >30min will stimulate FAT metabolism but avoid foods that will raise your blood sugar, as it will prevent muscles from burning FAT during and post exercise and preferentially use sugar as a fuel source. Additionally, morning exercise bouts prior to first meal can greatly impact FAT metabolism. Avoid sustained high-intensities or long durations while exercising in a FASTED state. Temporarily increasing heart rate above 80% to recruit fast twitch muscles is great but avoid sprint style training while in a FASTED state. Stay consistent, even on low energy days a brisk walk is enough to stimulate the body to burn fat as a fuel source.

Relevant insights:
Creatinine (low), Pre-diabetes, Complement C4-B (low), Alpha-1-acid glycoprotein 1 (high), Iron (low), Copper, Tryptophan (low), beta-Hydroxybutyric acid (low), and Glycine (low)

Health Risks:

Pre-diabetes

Your previous report also showed a high risk of diabetes. Your current report indicated diabetes is now in the low risk zone. This is great news! Along with improved markers for Diabetes Risk, your risk for pre-diabetes is currently in the MODERATE risk zone. This risk has fallen from the high risk zone from your last report. Great job! Your key biomarkers associated with diabetes risk, such as fasting glucose, branched chain amino acids, and lipopolysaccharide binding protein are within optimal ranges. Below are the 6 markers that contribute to your moderate risk.

Relevant results

- Phosphatidylinositol-glycan-specific phospholipase D (high)
- Betaine (low)
- Glycine (low)
- Copper (low)
- Complement C4-B (low)
- Creatinine (low)

Figure 5.3 Top: Action plan example. Information displayed to the client with arguments in text. Bottom: Health risk assesment example.

Signaling

In JAT, signals are linguistic and non-linguistic signs by which one person means something to another. Signals are deliberate and any deliberate action can be a signal in the right circumstances. More specifically, it is the *method of signaling* what can be used in 3 scenarios:

- Icons: a method used in demonstrating a thing
- Indexes: a method used for indicating something (e.g. pointing)

- Symbols: describing something as a type of a thing (mostly linguistic)

Table 5.3 provides some examples of signals found in the pair-analysis data. While the specific paragraphs of notes for each the *health data summary*, and the *action plan*, are the obvious linguistic signals, there is many other embedded interactions with the information that the experts are using as signals:

Table 5.3 Expert – Client signaling instances in the recorded data

Signal	Meaning
Analyst rearranges order of cards with notes.	<p>Non-linguistic, Indexing</p> <p>In both tasks, health data highlights and action plan, the analysts use the feature that allows them to sort cards with the intention to communicate a ranking of importance, but also to construct the written discourse with style (as in not repeating an argument twice if it has already been presented on a previous card)</p>
The specific paragraphs or the attention paid to word selection by analysts.	<p>Symbolic</p> <p>In both tasks, health data highlights and action plan, the analysts pay special attention to the words they choose to communicate risk, confidence, and what a signal means for an individual's health while aiming at preserving the uncertainty they have with the data.</p>
Client providing information in the phenome form.	<p>Both linguistic (content) and perhaps index of intentionality.</p> <p>The proposal here is that besides the obvious intention of sharing information for the case, actually completing the phenome form, and the level of detail to which is attended by the client, carries commitment, and a signal of the state of mind (anxiety, worrisome, curiosity, or just plain duty) of the client entering the activity.</p> <p>From the pair-analysis data we see that analysts read both content, and attribute intention based on the detail of information provided.</p>
Client not completing the phenome form.	<p>An important clarification is that, if completing the phenome carries more than content, not completing the form cannot be used as a signaling mechanism. Our experts cannot make any interpretation from an empty form, neither in content nor intention.</p>
Timeline of events	<p>Symbolic</p> <p>The timeline of events informs the client of the state and stages of analysis that the data is going through. It is also a call for action when they are expected to participate, for example, completing the phenome data.</p>

Signals can convey intentions that do not carry any content. *Gricean intentions* are those intentions that drive behaviors whose sole function is to have an effect by virtue of having their intentions recognized. For example, when our experts sort the cards, we can read the action with two different possible intentions: on the one hand sorting cards in the screen is a physical action of the reviewing and checkmark process our experts perform to conclude the decision process. In this case, similar to what Kirsh and Maglio (Kirsh, 2006) found out about Tetris players performing actions not for pragmatic advantage but rather for epistemic reasons, rotating figures in the screen is faster than doing the mental rotation. Sorting the cards can be seen as an epistemic action with the system in their reasoning process, the goal is to mark the task as done, and moving cards around is a physical instantiation of the review process.

In the second circumstance, sorting the cards for the experts could actually be carrying a communicative intention. We hear our experts talk about their ability to sort the cards as carrying the meaning of *order, ranking, and grouping*. The choice of the *first* card in some cases is important because there are more literal explanations in it. The *grouping* of biological related terms means coherence for the expert, and they deliberately do it *for the user*. See the two verbalizations below as examples:

Example 01:

Gail: When [clients] log in this is basically the first thing they will see. So, they will see that... hmhhh, okay I might want to rearrange this. Go back to Notes, alright, let's move Apo C2 down to under glucose [sorting cards in the notes again]. And let's make C4 under C2. [mouse clicking]. Yeah, that should be fine, close it.

So now the inflammatory markers are up here, and down here are mostly nutrition or nutrition-related markers, and the additional notes are showing up.

Example 02:

Helena: [reviewing the final action plan cards she has created] I think it's fine, but I could change the order of them and sometimes... yah, sometimes I try to have the "avoid" action as the last one, just because of the way it displays, and it looks the nicest, but sometimes I feel that it's the first one, and then I would move it down. And it's also important that the first action actually explains everything, so this one explains why we put fibre and omega 3 fats, and then because none of the other ones explains that, right?

But in this case, can the recipient decode the signal with an interface in which there isn't a mechanism for moving cards around? Nor a signal that decodes that order and ranking of the cards are intentional? Note that for the joint activity occurring between the expert and the intern, the signaling of sorting the cards advances common ground, so from a design implication it is worth asking if the recipient design is. In this case, signaling and the breaking of signaling mechanisms are a powerful construct in the study of developing meaning and communicating with data in non-co-present settings.

In summary, instead of a complex version of a user-centred design approach to interfaces for the display of information in healthcare, what I am proposing is the validity of joint action theory's constructs as a design tool to capture the requirements about the analysis and communication of large amounts of health outcomes that is so needed in the emerging industry of personalized health.

Designers typically approach the understanding of these mediated interactions with the implementation of UX research techniques. What I have proposed here is to bring JAT as a set of constructs to unpack and analyze the same situation. Taking JAT lenses, we can see the mediated interaction of expert analysts creating a summary for a client –that has provided their self-reported health information and goals– as a joint activity. As such, it displays the characteristics of joint commitment, advances in an asynchronous setting, displays the need for coordination devices, and displays the evidence of accumulation of common ground.

The proposal is then that we can gain a new perspective for the design of interactive data visualization technologies by tracing the progress of speaker's meaning and recipient understanding, in a specific implementation of discourse. In the particular case recorded in this dissertation this became clearer to me as I analyzed the use of information exchange, in the final *written format* that the experts used to communicate meaning (as described in chapter 04). However, the pair analysis data showed, that their *meaning* includes much more than what was captured in the written interface.

Standard UX methodologies and frameworks can be (and have been) applied to the design of interactive interfaces for the reporting of health assessments based on biomolecular testing (Andress, 2016). But the case in this dissertation, is highlighting an often-overlooked direction about transformation design: emphasizing on aspects of the

asynchronous activity in which analysts and clients engage with their new roles. They operate in a collaborative evidence-building and trust-developing process.

5.2. A Comparative Examination with the Doctor – Patient Relationship

To the best of my knowledge, the manifested expert analyst-health consumer relationship has not been a focus of attention in any medicine or HCI research agenda outside of clinical settings. It emerged in this dissertation as we observed the tasks of the analysts taking upon some of the *health provider* characteristics, that is, interested in finding relevant evidence to explain risk, and to answer a consumer needs and goals. Based on the instances of the data where this was highlighted, my proposition was that, similar to that of a doctor – patient relationship, in this mediated scenario we could see the activity with the *joint activity* lens. With that in mind there was an evident need to further investigate what are the characteristics of the doctor – patient relationship in traditional clinical settings? What is the relationship to the roles and tasks performed in this new wave of data-centric approach to preventive healthcare? Since I arrived to these questions is a result of the analysis of my data, I did not go in depth pursuing this research avenue, although it does highlight important and relevant future work.

In chapter 02, I summarized the literature on the doctor-patient relationship, particularly as depicted from a patient-centered medicine perspective. Let us recall that personalized health DTC services, as the one documented in this dissertation, do not address *patients* because the service is *not diagnostic* and can be provided to health consumers that are not convalescent of any disease yet. However, after a thorough analysis of the tasks and types of evidence that experts go through to support a client in understanding their biomolecular results and implications in their health, my research demanded to attend to the attributes of the relation unfolding between the analysts and their clients. In this section I draw a comparison of the literature on the doctor-patient relationship with our own specific settings and discuss the implications.

The doctor – patient relationship study describes an evolving association that remained a paternalistic model between a patient seeking help and a doctor whose decisions were silently complied with by the patient, for centuries (Kaba & Sooriakumaran, 2007). According with the literature, the model has evolved to

characterize the relationship as one of *mutual participation*, where the role of the physician is to *help patients to help themselves*. And in which the patient participates in partnership with a physician who brings expertise to discuss evidence and collaborate with the patient in the understanding of evidence about disease and therapies. Researchers in this area use the concept of “relationship” as “*neither structure nor function but rather an abstraction embodying the activities of two interacting systems (persons)*” (Szasz & Hollender, 1956)”

There is for us an immediate comparison in the depiction of the expert-client relationship as a *joint activity*, one that is carried out by an ensemble of participants and that requires individual actions to be coordinated in order to move together the intentionality and mutual commitment taken. The concept of *relationship* in Szasz and Hollender’s model, suggests the necessity to capturing more than content and process alone in the interaction between patients and their clinicians, highlighting the evolution that has taken from a single-individual passive-patient model to a participatory, interactive activity. The attributes of *joint activities* that I presented in this chapter provide a rich schema to describe precisely those type of activities where participants need to infer what individual actions, they can expect from each other so they can pursue their public goal. Through Clark’s framework, the patient and doctor entering in a *relationship* (set of activities) with a public goal that considers shared responsibility, can be inspected via the coordination of their individual actions, and the accumulation of common ground.

What seems rather obvious is that one of the initial shared beliefs should be that of their active roles. As we discussed in chapter 02, *sharing power and responsibility* implies the shift in the doctor-patient relationship from the “guidance-co-operation” model to “mutual participation”. Which translates into behaviours such as encouraging the patient to voice ideas, listening, reflecting, and offering collaboration. Each of these actions can be seen within the framework of *joint actions* as devices for the accumulation of knowledge and beliefs (about the activity) for both participants. In analogy, within our case of experts dealing with the molecular profile of individuals, we observed at length the expert’s fundamental principle to attend to these essential, although non biomolecular, aspects of the evidence when available. However, we also described earlier, that the control features of this setting describe a highly scripted interaction, and it would seem that the most important current gap is that there is little awareness of the participants on each other’s role. That is, we have evidence of experts

working in relation to an individual or a model of *client*, but on the other hand, little to no knowledge on the client's end about the medium they are in with their providers.

Moreover, in research literature the patient-centered approach has been described as one where "*the physician tries to enter the patient's world, to see the illness through the patient's eyes*" (McWhinney, 1989). Through chapter 04 I went at length presenting the multiple instances of the process of experts where their decision-making process is driven by what the client is telling them in terms of their goals, expectations, and lifestyle. The most immediate difference is that in the preventive and optimization model, not all clients are subject to an illness. The reader may agree with me in that this characterization, however, does not exclude the application of the patient-centered (consumer-centered) approach. I could argue that the understanding of what constitutes "health" for an individual, in the absence of disease, can be complex, and thus demands the same commitment from the providers to enter their client's world.

One interesting dimension that has entered the realm of the patient-centered approach to medicine is that of the "*two-person medicine*", in which the doctor is an integral aspect of any description. Biomedical expertise is considered but one of the components that the physician brings to the collaboration, there is a higher emphasis on the personal-level relationship that builds on trust. The relationship is described as one that develops through time between the parties. The most evident difference to highlight with our specific case, is that in the literature of patient-centered medicine, the rhetoric of the relationship between patients and their physicians, denotes an involvement and development through time shared in participation of their roles. In the DTC model to preventive care, most products provide services that do not go beyond a one-time point interaction. The most obvious case are DTC genetic services. However, as I depicted earlier, there is space to consider how to augment participatory actions through the course of the journey for a client from samples to results, in which they can feel they add their voice to a conversation more actively.

Moreover, in this aspect seems relevant the distinction between the analysts devoted to data interpretation for health risks and those devoted to the recommendation of lifestyle interventions based on relevant data that the former experts identified. We observed the efforts made by the nutritionist and the physical activity expert to warrant their choices and communicate to the client as if they *could see them in person*. Mostly

based on their knowledge of behaviour change. Coming to an interpretation of the data is a first challenge, but adherence to treatment, even within the scope of lifestyle recommendations, seems to depend in part on how far the expert can “*talk back*” to an individual and his needs.

Similar to what happened with the emergence of *genetic counsellors*, defined as “*health care professionals who provide individuals and families with information on the nature, inheritance and implications of genetic disorders to help them make informed medical and personal decisions*” (Genetic Counsellors | CIHI, 2019); the roles and information technologies that can accompany consumers through their journey, post-data delivery, would play a key factor on the adoption and adherence to personalized, preventive models.

Chapter 6. Discussion of Results and Implications

In this dissertation I approached the analysis work of biomolecular experts working on the interpretation of risk assessments with large amounts of biomolecular data and various types of evidence. I proposed to frame the problem as a visual analytics endeavor. One for which I recorded the decision-making process, the technological ecosystem, the expert actors, and their strategies to interpret the data results. This research set out with the general goal of *understanding the process that analysts of multi-omics (biomolecular) data follow to complete a consumer-tailored assessment for personalized health. And to understand the characteristics of sensemaking and data-visualization technologies that influence the expert's decisions for selecting the most relevant health evidence.* With that in mind, the two specific goals were: (1) to have an in-depth understanding of the process that expert analysts follow to derive interpretations and health recommendations from biomolecular data; and (2) to investigate the mechanisms that biomolecular experts use (if any) to validate their argumentation and courses of action working with incomplete data.

Considering this, I performed an inductive analysis of the pair-analysis data collected that gave room to five main themes:

- (1) **The sense-making loop of expert analysis:** The evidence of an iterative process of retrieving, linking, summarizing information, posing questions to oneself, making explicit potential courses of investigation, testing them as hypotheses, and working within the constraints of incomplete data.
- (2) **The actionable capacity, trust, and confidence in health evidence:** Observations that expose the experts' rationale for attending a piece of data as important evidence, or seeking for more details about it because of its relevance given (a) its scientific validity (confidence value), (b) the ability to target and intervene it via lifestyle actions, or (c) how familiar they think it is for a lay person.
- (3) **“The fourth person in the room”:** Within this theme I observed the evidence of a health consumer-centered biomolecular analysis. The research data showed arguments that the expert participants made about the data by virtue of emulating a conversation with the client or reflecting on what they inferred as the client's own goals and expectations.
- (4) **Reflective expertise:** Verbalizations that indicated any metacognitive reflection about the experts' own process.

- (5) **Team collaboration features:** In this theme I captured instances of participants referring to or taking actions that involve another team member in the process.

By working iteratively through the inductive analysis, I gained an understanding of the process, the types of evidence the experts use, and the characteristics of the relationship between the analysts and their clients. An in-depth examination of these themes was covered in chapter 04. In chapter 05 I expanded the analysis of the relationship between analysts and their clients introducing ideas from Joint Action Theory and revising medical epistemology postures on the traditional doctor-patient relationship.

6.1. Overview of Results

From the analysis conducted to investigate the socio–technical system in this personalized health organization, these are the main takeaways:

1. We observed a process that exhibits the characteristics of sensemaking analysis loops for each individual expert as described by Pirolli and Card's *sensemaking of intelligence analysis* (2005). The experts go through loops of information foraging and schematization and developing a personalized health story of the results. Schematization of the case for these experts refers to creating a sense of who is their client, including a biomedical and psychosocial perspective. To do so, they look at self-reported (*phenomic*) health information to understand past history of disease and the health goals of an individual. In most cases they create analysis leads from the phenome information to then investigate the signals picked in the biomolecular data. They can also approach the biomolecular evidence first to create a profile of the individual to then confirm if it aligns with the self-reported data.
2. We uncovered the different types of evidence that bio scientists are using in the analysis process: That is, the biomolecular data linked to scientific research, the self-reported health data, and their expert know-how. We found out that the use of the consumer preferences and the inclusion of context of lifestyle and circumstances go in line with research in epistemology of medicine, the study of what constitutes evidence in clinical practice. The Evidence-based medicine

(EBM) movement stresses the examination of evidence from clinical research and deemphasizes intuition and unsystematic clinical experience rationale as sufficient grounds for clinical decision making. For this reason, EBM requires new skills of the physician, including efficient literature searching and the application of formal rules of evidence evaluating the clinical literature (Upshur & Colak, 2003). More recent research in EBM models, reflects that evidence from scientific research is no more than one component of any decision. With respect to each of the types of evidence and the methodologies used to assess it, we can summarize:

- In this dissertation, the scientific evidence that links biomarkers to diseases, and that links therapies to effects at the molecular level, is found in different categories of confidence and with different number of studies performed (reproducibility) by the scientific community. This also goes in line with an EBM approach. Our experts explore the literature to identify multiple studies on a topic, not restrictive to RCTs. With the aid of data mining algorithms, they store scientific publications into knowledge databases, they separate the best publications and then critically analyze them to classify the best available evidence.
- The phenome information is the source of information for the psychosocial factors, goals, and other lifestyle data that is pivotal for the analysts schematizing a case. We could say that our experts work in line with the believe of "*health as a moving target*", that is, they are looking to capture insights regarding health areas, but also looking for what is the right frame or angle to take for each individual based on inferences of such individual's goals and current circumstances. Based on this, a signal of risk for cardiovascular disease or pre-diabetes and what to do about it, can be framed differently for individuals using the individual biomarker results that contribute to the evidence, and using the client's reported goals about their own health.
- Regarding expertise, the know-how of analysts required in this case is more than biomolecular and biomedical knowledge. We observed a set of experts deploying their knowledge and expert intuitions about human behaviour to complete the sensemaking tasks. We reviewed instances such as (1) applying knowledge/intuitions about the communication of health information, (2) understanding the emotional / psychological component of a person's attitudes towards risk and uncertainty in health results, and (3) expert knowledge of the psychology of behaviour change at play.

The main takeaway from looking at the use of evidence is that besides the evident biomolecular data, the phenotypical –or self-reported health data– is a key component in the investigative process. Our analysts use the two different sources of data to complete their schemas and compare results. It is important to

highlight that we are looking at a team of *analysts* implementing the EBM practices in a setting distant from the traditional clinical scenario. The novelty in our case is the seamless translation of the practice. We observed attributes of the EBM model emerging in a technology-mediated, preventive approach to healthcare, outside of clinical settings.

3. With respect to expert performance, an important takeaway is the evidence on the unstated tasks and the shifting role that analysts perform as health providers. Implementing efficient mining techniques for research literature and use of algorithmic rules for assessing the evidence, is part of the established practice for bio scientists in an *analyst role*. But we also observed them putting in action their human-interaction skillset, as a *health provider* would do using their expertise in this area. Like (Haynes, 2002) presents, “*clinical knowledge and expertise*” are essential to success as a complement to the research evidence. In recent versions of the EBM model, clinical expertise is the central force that glues together activities such as diagnosis, prognosis, effective patient communication, the correct performance of a treatment or test, and the application of population-based evidence to particular individual patients, a goal that often involves the sorting of trade-offs (Wieten, 2018). In this dissertation we recorded our analysts turning to a type of “clinical hat” to cover the communication of risk, the details of lifestyle interventions, and putting it all together selecting the best evidence for the individual case at hand.

In this dissertation we learned that although personalized medicine DTC services remain for the most part non-diagnostic, and promote individual’s empowering via clouds of data, the role of health providers has remained active and has shifted to the tasks that bio scientist perform for an individual. This assertion goes hand in hand with our discussion about relevant attributes of evidence (number 4 below) and the role of expertise and know-how in preventive care. Psychosocial factors in the relationship between a provider and a health consumer are key constituents of any delivery of care model. Compared with the evolution of the traditional doctor-patient relationship, I concluded that a model of *mutual participation* is a relevant framework to look at the emerging analyst-client relationship. The idea of mutual participation is based on “*the belief that the doctor does not confess to know exactly what is best for the patient*” hence,

requiring to actively listen and take into account the consumer experience of their health. Their interaction is characterized “*based on having equal power, mutual independence, and equal satisfaction*” (Kaba & Sooriakumaran, 2007). It ultimately results in empowering patients to take care of themselves.

4. We also identified different attributes that experts use for the assessment of biomolecular evidence. We found as expected, the use of *confidence* as a measure to evaluate the quality of evidence from scientific literature. Confidence of evidence was assessed based on standard industry metrics for scientific validity (applying a threshold to judge the quality of evidence based on the underlying study methodologies). But more importantly, we recorded that *actionability* and *familiarity* of the data are determinant attributes for bio scientists creating an assessment of the results for an individual. Similar to what we may qualify in the design-lingo as a “user-centered” decision-making process, the actionable capacity in the evidence turns the process towards an approach to preventive healthcare with biomolecular data that positions the client as an active user, rather than a passive agent. That is, identifying evidence that can translate to action, agency, and participation by a client seems to be the signature of the preventive approach documented in this dissertation.
5. Finally, for an innovative take upon the mediated expert analyst – health consumer relationship, in chapter 05 I explored the constructs of Joint Action Theory (JAT) and proposed to portray the analytic task within a larger type of joint activity occurring between the analysts and a health consumer. The main takeaway from this analysis is that JAT provides a valuable lens to understand people coordinating meaning in complex socio-technical situations. Ultimately, I propose to take this lens as a toolkit for designers looking to understand the nuances of the creation of meaning via the study of communication. As a toolkit, it can be integrated into the design cycles of technology in healthcare. The specific concepts of *common ground*, *role awareness*, *coordination devices*, and *signaling methods* are robust constructs to study the pitfalls of communication in mediated DTC personalized health services.

6.2. Implications

Based on the insight I gained into the socio-technical system as a service in personalized health, in this section I present the implications that can have an impact on the design of technology in this domain of digital healthcare. It is important to clarify that the scope of these reflections belong to the in-depth understanding of the needs and challenges of the complex emerging economy of DTC personalized health. I conducted a qualitative study to understand sensemaking practices on how experts infer and communicate meaning with biomolecular data. However, the results provided a broader overview of a system that includes experts, consumers, evidence, information as health stories, and technologies that mediate the participation.

With this in mind, I present the following implications:

Role Participation and Role Awareness: The Tacit Health Provider

The results showed that in the interpretation of data and communication of health risks assessments, our expert analysts also perform tasks of a health provider. A shifting responsibility of the provision of care role suggests that designers of communication technology in this domain should emphasize to support mutual awareness.

The tasks that experts perform go beyond data analysis and include a relationship with the psychosocial factors of an individual in order to create a health narrative of highlights and lifestyle actions for them. Moreover, as we presented in early findings with consumers (*Visualization in Practice: Personalized Health as a Human-Centered Design Process at Molecular You*, 2017), clients often referred to a sense of isolation with the data, “missing the voice of the physician”, and felt challenged to assume health decision making with this complex data. From these results, figure 6.1 depicts the proposal to understand the relationship in a 3-directional way. For the case we recorded in this dissertation, experts embodied tasks for two of the roles.

This does not imply a call to fictionalize a *health provider*, but rather to account the voice of an expert in the design of the communication tools. The expert has taken into account the strength of evidence in the case, including the evidence that is provided by the clients themselves beyond their biological samples. This also implies, that designers can stress the client’s own agency and create communication channels or

strategies to accentuate the high value of the self-reported information as it feeds into the analysis. Finally, the figure features a relationship between the expert in its two roles and opens the consideration of how to design toolkits and analysis tools to support the ability of the analysts to move into the role of the provider.

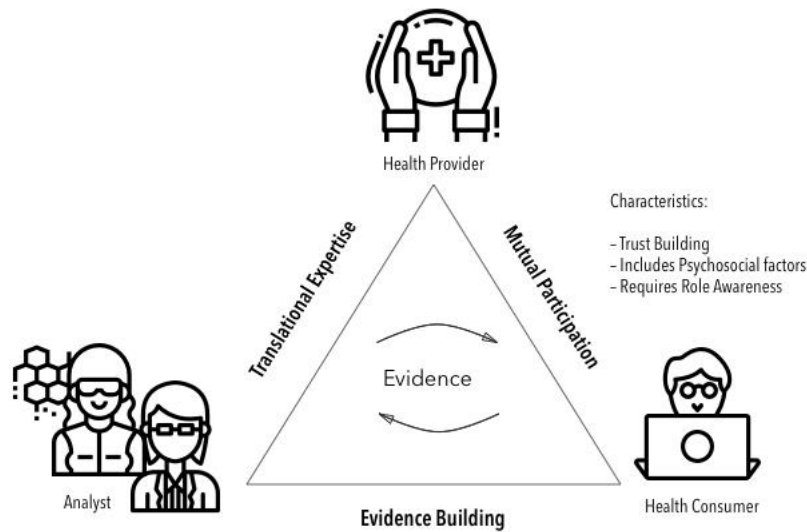


Figure 6.1 Roles and type of relationship in the DTC personalized health provision model.

More studies with medical practitioners would deepen the understanding of the attributes of the relationship between a provider and a health consumer. The literature in this field is rich for a traditional model, but out of the scope in this dissertation. As I discussed in the methodological decisions in chapter 03, understanding the attitudes and adoption of medical practitioners for the Personalized medicine methods should consider barriers from knowledge (training) to the systemic rewarding system in place with current healthcare models. Studying the attitudes of different type of providers like genetic counsellors and specialists that have integrated genetic testing into their practice will certainly better shed light in the design of reporting and mediation tools. However, many current preventive services in personalized health are designed without the mediation of a health provider, better understanding of the barriers for consumers and to support active roles by other experts continues to be relevant in the personalized medicine ecosystem.

Roles' Dynamics: Mutual Participation

In figure 6.1 above, I also depict what we observed in this dissertation as the information flows between the different roles. The results showed that the *mutual participation* model fits the relationship between the emerging analyst-providers and their clients in personalized medicine.

Through this dissertation we followed a decision-making process of experts whose end product closely embodies a *guidance-cooperation* model between providers and consumers. Our experts are expected to have the biomedical knowledge that the client lacks; different experts are expected to prescribe lifestyle therapies in the client's best interest; the health consumer sits at the end of a workflow where she only provided an experience of her health through information forms. She consumes her data results and advice.

In the practical details of the process however, there is less evidence of a dramatic power disbalance, and more of a participation gap. Through the process, we observed that a key driver for the analysts was to “answer” to their client's goals and preferences. We also saw the significant role of self-reported health information, that is, the recorded “biography” of a client. And in the end, some of our experts verbalized their goal as that of “helping their clients help themselves”, or “empowering them with biomedical information” in the absence of a direct interaction with them.

So, while the media rhetoric for personalized medicine services seems to overemphasize a biomedical positivist framework, our analysts in the trenches deliver healthcare analytics aiming for a framework of mutual participation between them and their clients. Their information interfaces, however, convey a guidance-cooperation at best. To that end, in this dissertation I proposed that the concepts of common ground, signaling mechanisms, and coordination devices from JAT can be a toolkit for unpacking communication, coordination, and participation features for the design of balanced participation in evidence-based DTC health tools.

Transparency and Completeness of Evidence

As we saw in number (2) and (4) of the overview results, *evidence* in this process is not a single item but a data structure of different types of information with multiple

attributes. It is both a challenge and a responsibility for designers of communication technology in this domain to aim to preserve the completeness of the data that justifies the decisions and warrants the results.

Figure 6.2 below highlights the pieces of evidence that are displayed in the client interface. A client can read a list of all biomolecular data as evidence that contributes to the results (see highlight 2), but each item is presented at the same level of importance. Despite some of the attributes are displayed to the client (like the confidence level highlighted in 3), there is not a clear signal that other attributes made a piece of information relevant for the expert. The images present two different screens that a client visits to obtain information about her assessment of risk for pre-diabetes. As we saw before, the *health data summary*, includes a personal narrative that an expert created for the individual. There is also a set of biomarkers highlighted as “relevant” in the summary (highlight 1). The client can obtain more details navigating to the “Pre-Diabetes” page. Yet, in this page again, any signal for how to read the relevance of each piece of evidence listed, is missing. Except for the (scientific) confidence score. More importantly, there are not references to the use of self-reported information as source of evidence.

Following the analysis that our experts made, we can highlight that although all biomolecular data is presented, and the results of the interpretation highlight some information as “relevant” pieces of evidence, there is a gap in displaying the hierarchy of the evidence. Presenting transparently that each piece of information stands out for an expert given different attributes, makes part of the development of trust in the data and the provider relationship with the consumer.

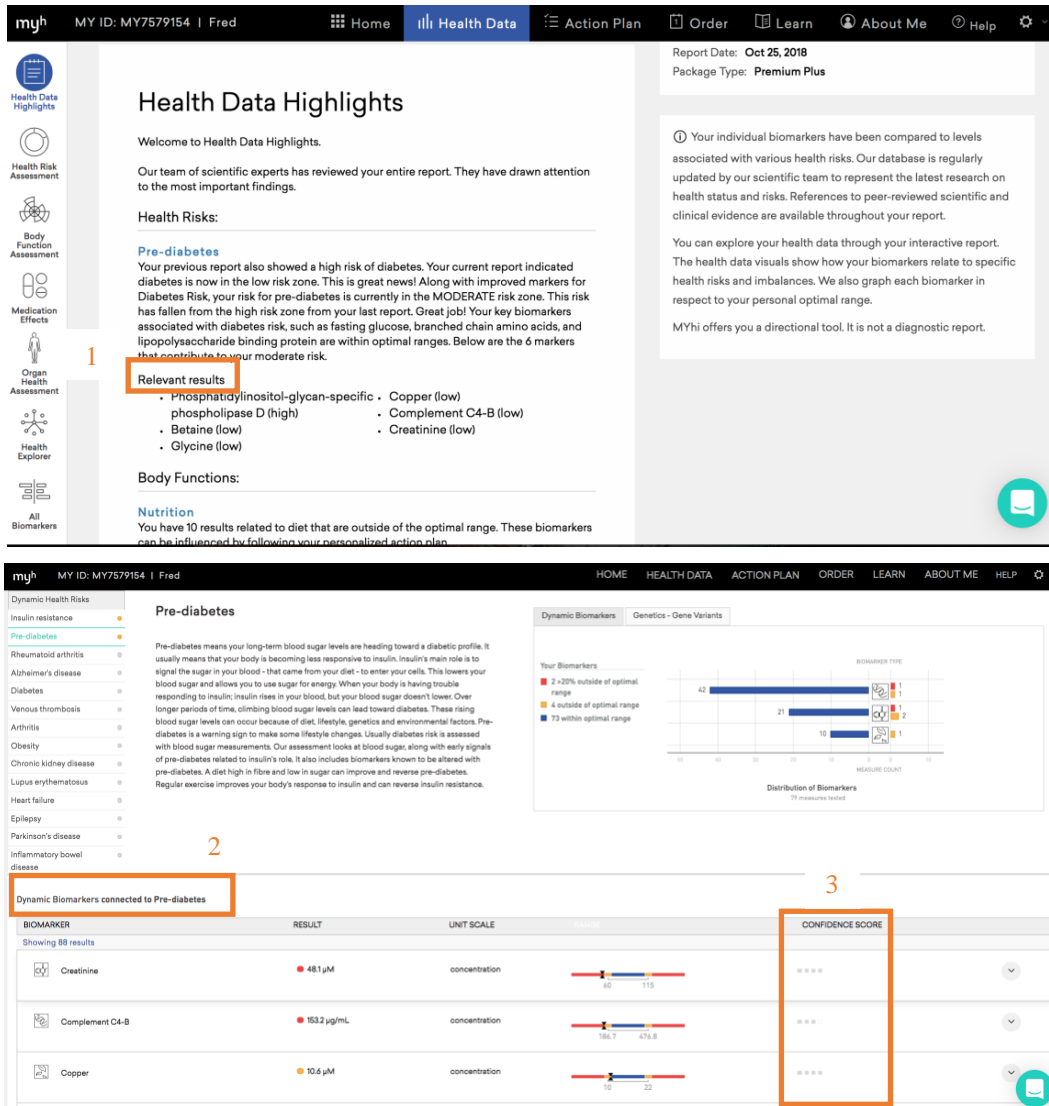


Figure 6.2 The evidence displayed in the *Client Interface* in a flat layout, without hierarchies.

When we followed the verbalizations from our experts about how they developed the summary and described the details that go into the actions, it was made clear that their own appreciation of what constitutes *reportable evidence* is limited to the biomolecular scientific evidence, and to the confidence value about it. Despite their case is built around all the other types and attributes aforementioned, there seems to be a tendency to talk about evidence from the scientific-standard alone. Designers can have a great impact on understanding the information flows and working towards representing the completeness of the case that ultimately strengthens the value of information and the trust-building relationship between all parties.

It is out of the scope of this dissertation to run a comparative analysis of different DTC personalized medicine services to analyze how does each present evidence and warrants of argumentation. But we can say there is a trend in big-data initiatives in health care to focus the attention in the volume of data that we are now able to quantify, overlooking at what turns a piece of data into evidence for both experts and consumers, particularly within the preventive approach that differs from a clinical diagnostic one. Current data visualization design guidelines can help bring perceptual cues to the display of hierarchies of evidence, this discussion is about acknowledging the attributes of evidence often overlooked by the predisposition to present evidence as methodological *scientific evidence* alone.

Defining Quality in Self-Reported Health Information

We discussed at length the weight of the self-reported information in the decision-making process. Including all pieces of relevant evidence, besides the biomolecular results, also make part of trust-development and a clear analytic provenance trail. For the case we studied in this dissertation, the phenotypical information plays an important role in the analysis process of experts, but it is downplayed in the communication of end results.

How to better capture information from health consumers is a research area in its own, particularly if it takes into account the social perspectives of illness and disease as we discussed in chapter 05. Initial efforts in this direction, and as part of the work done with this organization have already been made part of the HCI – Healthcare discussions see (Barrera-Machuca et al., 2019). In this work, Barrera-Machuca and colleagues discuss how electronic health records (EHR), although designed to provide instant access to patient information across networks of health providers, carry on the western tradition of the “depersonalization of health care”. While serving as a valuable knowledge source for health providers, the EHR replaces dialogue with the patient by a one-sided extraction of information. This raises the need for tools that can provide a two-sided dialogue in which the interface provides the patient information as well as medical understanding. They evaluate if describing the experience of illness in a somatic graphic representation would give patients a more accurate and descriptive understanding of their illness, while continuing to provide health providers accurate information for their analyses. More efforts in this direction are highlighted as relevant research gaps and

elucidate that interdisciplinary approaches can address the current limitations in understanding the complexity of technology for healthcare.

Besides the evident continuation of that work, in this dissertation I also discuss the integration of phenotypical information as a content-coordination device that requires to be recorded and present for the exchange occurring between experts and the clients during the analytic tasks. This goes along with Barrera-Machuca call for a “holistic approach” in understanding information flows for patients, caregivers and analysts.

The Power of Narratives: The Machine Learning and Big Data Assisted Medicine Rhetoric

Machine Learning and Big Data assisted medicine rhetoric tend to overlook at the cultural constructions of health realities, but analysts from the trenches do not. This is a final reflection about the reality in which our expert analysts operate in face of the promise of big data analytics in biomedical practice. In the era of “big data analytics” for healthcare, the rhetoric of a shift to the provision of care enabled by our technical ability to quantify and assess large volumes of biomedical data, seems to strengthen a notion of healthcare from a “biomedical positivism framework”. That is, that diagnosis of disease, medical image analysis, integration of devices, and ultimately, the selection of the appropriate therapy is empowered by volumes of data and algorithmic accuracy, thus improving the patient’s illness.

I addressed this research program within the framework of visual analytics endeavors. This due to the amount and complexity of the data, and the ill-defined task of identifying preventive signals of disease, made the case into a fitting VA challenge. As I reported in the introduction to this work, big-data analytics is a trend often documented as the radical force in the transformation of personalized medicine (see (Cirillo & Valencia, 2019; Topol, 2019). Multi-omics, medical image analysis, devices data, and electronic health records represent some of the main big data types in biomedical research from a post-positivist view. Researchers and promoters rightly identify significant scientific and technical developments required for the creation of powerful systems for the effective use of biomedical big data in Personalized Medicine, including infrastructure, engineering, project and financial management(Cirillo & Valencia, 2019).

However, the medical reality for individuals and its study from a socio-technical perspective justify the distinction between the experience of disease, illness, prevention, wellness, and aging well. Disease understood as “*the malfunctioning or maladaptation of biologic and psychophysiological processes in the individual*” (Kleinman et al., 1978) may very well benefit from all the biomedical research progress, but the experience of illness as, “*the personal, interpersonal, and cultural reactions to disease or discomfort*” (ibid), requires the intervention of the social sciences to deal with these essential, but nonbiomedical, aspects of clinical practice. This is something even at higher risk of being overlooked outside of the clinical practice, in a model of prevention, where individuals are not patients yet.

What we documented in this dissertation was the nature of the analysts turning to their human-interaction skillset to address the health reality of each individual they worked with. Although assertions about the scientific validity and the amount of data, overemphasize the claims of this personalized health approach, in practice, the analysts turned to attend goals, preferences, to find actionable evidence in the data, and to frame a relatable health summary story for their clients.

The role of technology design in scenarios like this one will be fundamental in properly translating and bridging the effort from these emergent providers (the analysts) in communication with their clients. Both parties can benefit from analytic capacities to explore and understand the strength of each piece of evidence in the case, including the evidence that is provided by the clients themselves beyond their biological samples. In the end, this may be as relevant for the Visual Analytics community to support the tasks of bio scientists in personalized medicine, as much as an HCI initiative in support of data-centric models of healthcare.

6.3. Study Limitations

Firstly, I would like to acknowledge that there is a gap in the lack of the *client* perspective in the current study. I entered this research program with questions about *expert analytics*, the factors that influence their decision-making process with biomolecular data. As the analysis of my research data evolved, I noticed the evident presence of the *client* schema in the process, and the weight it took, the assumptions experts made, and the feedback they integrated to update their process. When I

proposed to investigate the activity as a *joint activity*, I funded my observations in the recorded data, but extended the depiction of the situation also based on knowledge from my involvement in early user studies with consumers (*Visualization in Practice: Personalized Health as a Human-Centered Design Process at Molecular You*, 2017). That is, through my deep involvement as UX researcher with the organization, and other studies not covered under the ethics and scope of this dissertation project. As I will discuss later, getting a current comparative *client* perspective about their participation of the activity is a good next step for future research.

Regarding the scope of generalizability, this study was framed as descriptive of a real-world application of visual analytics with biomedical data. The engagement with the organization and the delimitation to study the experts at such organization presents an obvious limitation in scope. However, through this program, we have uncovered new testable hypothesis and research avenues that can be extended for the communication of risk in healthcare evidence-based endeavors. The implications discussed in the previous section were presented within the broader emerging industry of DTC personalized health services, and for the evolving dynamics between the roles of providers and consumers in this model. As a translational science, and research conducted in centers of application, Visual Analytics endeavors like the one in this dissertation have been depicted before as translational in strength – “*a science that bridges centers of production of knowledge and applications where knowledge is put to use*” (Fisher et al., 2011).

Furthermore, I acknowledge the potential delimitations of a single coder of the research data. I discussed that the pair analysis data with real biomolecular client cases required unique skills and knowledge in various fields to generate reliable and efficient coding schemes. I possessed the contextual knowledge to examine and code the phenomena in the collaborative visual analytics scenarios that we proposed. In this dissertation I have done an effort to document the process such that more researchers can potentially take advantage of the rich dataset that I created with anonymized transcriptions and use the data for further investigation. With the case described at length in this dissertation, I hope that the categories resultant from my analysis, or new perspectives on the data can be approached by researchers without the restriction of being embedded in the organization.

Chapter 7. Conclusions and Future Work

7.1. Thesis Summary

The research program I presented in this dissertation began with my interest in understanding in-depth the decision-making process that bio scientists take to interpret large amounts of data in a DTC model of provision of care. I became interested in this problem as an in-situ participant of the operations of an organization that serves health consumers with a comprehensive health assessment that uses scientific evidence to interpret multi-omics biomolecular results. The biomolecular data integrates genetics, metabolomics and proteomics, along with the self-reported health history information of a client to create a snapshot of the current state of the body at the molecular level. The promise of this approach to *personalized health*, is that the data analysis capacity allows us to identify early signals of disease and to tailor lifestyle therapies to each individual's molecular mark-up. However, early in the process I witnessed challenges related to the scalability of the service if dependant on human experts to go through the data, and the actual level of *personalization* that was feasible to achieve without the goals and preferences of individuals. More importantly, there were challenges associated with communicating complex evidence and health risk assessments to health consumers without the participation of physicians. As a Visual Analytics scenario, I approached the problem to investigate expert analysts, their sensemaking process, and the role of data visualization technologies.

In chapter 02 I presented the literature to introduce the field of Visual Analytics and works in healthcare analytics relevant to my research. As I discussed in the chapter, there is not up to date a body of works in the specific domain of analytics in direct-to-consumers healthcare services, except for those that are devoted to the scientific visualization of each of the *omics* domains I mentioned before. In the chapter I also introduced the literature in trends and evolution of medical practice, particularly the psychosocial components of what constitutes evidence in medical research, and how has the doctor – patient relationship evolved over time. These works became relevant in my research as I was progressing in an inductive analysis of the data. Went back and forth between my data and the literature looking for research done about themes that started to appear in my data. The Evidence-Based Medicine model and the literature in

the Doctor-Patient relationship were theories I identified and used in the interpretation of my observational data. Moreover, as I started the research program concerned with *expertise* and the attributes that differentiate skilled expertise from intuitive judgement, I summarized the literature on expertise from a psychological perspective. Finally, because of the relevance in my interpretation of the expert – consumer relationship and the methodological commitments, I introduced *Joint Action Theory*.

In chapter 03 I presented the methodological approach. This research program deployed and assessed the application of the *Pair Analytics Methodology* (R. Arias-Hernandez et al., 2011) to capture the nuances of the problem-solving process and richness of a distributed process in the specific situation under study with experts in a personalized medicine organization. As documented elsewhere, deploying pair analysis methods is a successful strategy to gather verbal protocols of people solving a problem together. I experienced the same challenges reported, associated with time-consuming transcriptions of audio and screen recording data. Moreover, my adaptation to the protocols produced data that was less rich on collaboration and discussion, but richer on the level of detail of explanations that experts gave to a trainee intern. To truly implement Join Action Theory with pair analysis data however, my conclusion is that the situation requires a more balanced participation by both participants, as in Wade's case (Wade, 2011). We could say that there is a higher inference power with JAT for situations of expert collaboration and equal participation.

Through chapters 04 and 05 I presented the in-depth analysis of the data. In Chapter 04 I provide a sociotechnical account of the detailed collaborative and asynchronous process that a team of experts goes through with the results for each client. The intervention of human experts responds to the need to clarify data results for health consumers. Experts create a *health data summary* with the data that they consider are most relevant and for which different attributes of evidence are stronger for the individual's case. I described the attributes of evidence exhibited in the process and a major observation was the role of the self-reported goals and health history data from an individual, guiding the analysis process. In the same theme, the key value of evidence that represented actionable capacity also was a noticeable component of the process. In the same chapter I documented data visualization approaches I had taken to support the understanding and communication of confidence of evidence.

Finally, I dedicated chapter 05 to investigate the mediated expert analyst – health consumer relationship. I proposed to use the constructs of Joint Action Theory and to portray the analytic task as a larger type of *joint activity* occurring between the analysts and a health consumer. In entering that part of the analysis, the motion was that, as in traditional models of medicine, there are more dimensions beyond the scientific evidence against the treatment of disease. Psychosocial factors in the relationship between provider and health consumer are key constituents of the delivery of care and need to become part of technology design considerations.

In chapter 06 I conclude the analysis with an overview of the main takeaways after conducting the qualitative analysis, and a discussion of the implications from these findings.

7.2. Contributions

With the emergence of direct-to-consumer personalized health services, traditional health professionals and arising roles of health providers, constantly undergo the challenging task of analysing and interpreting heterogeneous biomolecular data in order to provide tailored health insight to individuals. Understanding the gaps and challenges in this new trend of care delivery is an important contribution. Given the focus of my study in the tasks and processes of analysts, my analysis can provide valuable contributions to understanding the tacit tasks that bio scientists perform in a consumer-centered analysis process.

In my analysis, I demonstrate that non-explicit attributes of evidence play a key role in the sensemaking process for bio scientists. This is in part due that the analysts perform tasks tuned to provision of care beyond analysis of scientific evidence. The latest research in Evidence-Based Medicine proposes a model that includes patient preferences, psychosocial circumstances, and scientific evidence, as evidence types in clinical decision making. In that model, physician's expertise glue together the diverse sources in clinical practice. The case in this dissertation contributes to the understanding of the same phenomena outside of clinical settings, and as such, emphasizes the relevance of understanding emerging *health provider roles* and their information practices. Along the same line, a specific contribution was to unmask that apart from the confidence in scientific validity, the potential *actionable capacity* –or the data for which

there is direct evidence about lifestyle interventions that has a therapeutic effect; and the *familiarity* of all the data measured –based on their known exposure on public dissemination of science; are both important attributes that experts use in their decision making process. Based on these findings, discuss implications that informs visual analytics practitioners and technology designers working in the DTC personalized medicine domain. I highlight that designers have an important role ensuring the communication of everything that is included as *evidence* and *attributes of evidence* for a consumer.

In an effort to better understand information practices, tasks, and needs in the workflow from analysis to communication of personalized health results, I contributed an analysis of the expert analyst-consumer as a *joint activity* and compared it with the more evident doctor-patient relationship. From this analysis I proposed a future line of work to investigate the characteristics of the relationship between health consumers and health providers in the personalized medicine paradigm. From an HCI perspective such work requires to take a clear posture about what is to empower a health consumer, and in what tasks the active role of a consumer has place during the service. The analysis itself and the use of Joint Action theory is a contribution to the toolkits of UX researchers looking to understand the nuances of how to create meaning in the communication of evidence in personalized health.

The work in dissertation presents a contribution to the social studies of digital technology for personalized healthcare and to the Visual Analytics community from a methodological standpoint. By taking a VA approach to the challenges, I was able to unpack the pressing issue of attending analytical reasoning in personalized healthcare as a collaborative, social construct that requires to be understood from a participatory provision of care model rather than exclusively from silos that look at the needs of analysts and clients apart. I consider it a novel approach to understanding the issues in this domain. This study provides methodological strategies for examining sensemaking, collaborative decision-making, and cognition in workplace environments. I followed a clear and repeatable protocol that can be beneficial to HCI, and VA research conducted in centers of application. This is a contribution in benefit of translational sciences and a different adoption of applied research in Visual Analytics. As a contribution to further research in this direction, I have also contributed all the text transcripts for the 11 hours of recordings that I gathered in pair analysis sessions.

Moreover, for the pressing issue of understanding the social constructs of digital technologies in the provision of evidence-based and data oriented personalized health, this dissertation contributes the methods, theories, and lessons for investigating the evidence based provision of care in the asynchronous fashion. The implementation of JAT adds a substantial new tool to the toolkits of HCI designers, because it goes into goes in the detail about the negotiation mechanisms to create meaning between two participants of an activity in the provision of care in the personalized medicine model.

7.3. Future Directions

There are several directions for extending this research. First, it would be worthwhile to investigate the constructs of the *joint activity* proposed, that is, the accumulation of common ground, the issues with signaling mechanisms, and the expectations of entering a joint activity, with the client's perspective.

Secondly, future work can take propositions from this dissertation to investigate more DTC initiatives in personalized health. Instead of replicating the same methods and analysis, a future study can be designed to investigate hierarchies of evidence, attributes of evidence, and incorporation of consumers' goals and preferences in personalized health products and services. Extending the scope to other products can help to ground a more robust conversation in the field.

Third, future research will continue to investigate the digitalization of the experience of health, and its implications in the provision of care in the personalized health paradigm. This dissertation showed at length that phenotypes, or expressed health information in the form of symptoms, history of disease, lifestyle habits, or what is consider *the biography* of an individual, play a key role in the analysis of biomolecular data. More research in how to better capture a psychosocial picture of illness and health goals, will have an impact moving forward with more DTC services.

The personalized medicine community has often manifested the challenge of bridging the scientific and the clinical communities. It has been described as a challenge to bridge tribes that look at problems from a different perspective. Continuing the investigation of the relationship between the physician and its patients is a promising

avenue to identify the similarities and differences with the emerging analyst – health consumer relationship needs. Moreover, how can data visualization interfaces better support the mediation of the service.

References

- FDA-NIH Biomarker Working Group. BEST (Biomarkers, EndpointS, and other Tools) Resource [Internet]. Silver Spring (MD): Food and Drug Administration (US); 2016-. Available from: [https://www.ncbi.nlm.nih.gov/books/NBK326791/Co-published by National Institutes of Health \(US\), Bethesda \(MD\)](https://www.ncbi.nlm.nih.gov/books/NBK326791/Co-published%20by%20National%20Institutes%20of%20Health%20(US),%20Bethesda%20(MD).).
- Barrera-Machuca, Aldo A; Calderon, Nadya; Anwar, Ash; Fraser, Robert; Fisher, Brian. "Graphical Mapping of a Data Ontology For Capturing Descriptive Patient Data", 1-3, ACM CHI, Glasgow, UK, 2019
- 6, P., & Bellamy, C. (2011). *Principles of Methodology: Research Design in Social Science*. Sage Publications.
- Al-Hajj, S., Fisher, B., Smith, J., & Pike, I. (2017). Collaborative Visual Analytics: A Health Analytics Approach to Injury Prevention. *International Journal of Environmental Research and Public Health; Basel, 14(9)*, 1056. <http://dx.doi.org.proxy.lib.sfu.ca/10.3390/ijerph14091056>
- Al-Hajj, S., Pike, I., Riecke, B., & Fisher, B. (2013). Visual Analytics for Public Health: Supporting Knowledge Construction and Decision-Making. *2013 46th Hawaii International Conference on System Sciences (HICSS)*, 2416–2423. <https://doi.org/10.1109/HICSS.2013.599>
- Anders Ericsson, K., Charness, N., Feltovich, P. J., & Hoffman, R. R. (2006). *The Cambridge Handbook of Expertise and Expert Performance*. Cambridge University Press.
- Andress, S. (2016, June 22). *Designing the New 23andMe Homepage*. <https://blog.23andme.com/23andme-and-you/designing-the-new-23andme-homepage/>
- Arias-Hernandez, R., Kaastra, L. T., Green, T. M., & Fisher, B. (2011). Pair Analytics: Capturing Reasoning Processes in Collaborative Visual Analytics. *2011 44th Hawaii International Conference on System Sciences (HICSS)*, 1–10. <https://doi.org/10.1109/HICSS.2011.339>
- Arias-Hernandez, Richard, Green, T. M., & Fisher, B. (2012). From Cognitive Amplifiers to Cognitive Prostheses: Understandings of the Material Basis of Cognition in Visual Analytics. *Interdisciplinary Science Reviews, 37(1)*, 4–18. <https://doi.org/10.1179/0308018812Z.0000000001>
- Arias-Hernandez, Richard, Kaastra, L. T., & Fisher, B. (2011). Joint action theory and pair analytics: In-vivo studies of cognition and social interaction in collaborative visual analytics. *Proceedings of the 33rd Annual Conference of the Cognitive Science Society*, 3244–3249.

- Ashcroft, R. E. (2005). Current epistemological problems in evidence-based medicine. In *Evidence-based Practice in Medicine and Health Care* (pp. 77–85). Springer.
- Aspinall, M. G., & Hamermesh, R. G. (2007). Realizing the promise of personalized medicine. *Harvard Business Review*, 85(10), 108–117, 165.
- Balint, E. (1993). *The doctor, the patient and the group: Balint revisited*. Taylor & Francis.
- Berg, J. S., Khoury, M. J., & Evans, J. P. (2011). Deploying whole genome sequencing in clinical practice and public health: Meeting the challenge one bin at a time. *Genetics in Medicine*, 13(6), 499–504.
<https://doi.org/10.1097/GIM.0b013e318220aaba>
- Berndt, E., Furniss, D., & Blandford, A. (2015). Learning Contextual Inquiry and Distributed Cognition: A case study on technology use in anaesthesia. *Cognition, Technology & Work*, 17(3), 431–449. <https://doi.org/10.1007/s10111-014-0314-y>
- Bloss, C. S., Ornowski, L., Silver, E., Cargill, M., Vanier, V., Schork, N. J., & Topol, E. J. (2010). Consumer perceptions of direct-to-consumer personalized genomic risk assessments. *Genetics in Medicine*, 12(9), 556–566.
<https://doi.org/10.1097/GIM.0b013e3181eb51c6>
- Button, G. (2000). The ethnographic tradition and design. *Design Studies*, 21(4), 319–332. [https://doi.org/10.1016/S0142-694X\(00\)00005-3](https://doi.org/10.1016/S0142-694X(00)00005-3)
- Byrne, B. (2016). *Health Jobs are the Basic Unit of Work in Digital Health*. Daedalus Innovation. <https://www.daedalusinnovation.com/blog/health-jobs-are-the-basic-unit-of-work-for-digital-health-platforms>
- Carere, D. A., Kraft, P., Kaphingst, K. A., Roberts, J. S., Green, R. C., & Group, for the Pg. S. (2016). Consumers report lower confidence in their genetics knowledge following direct-to-consumer personal genomic testing. *Genetics in Medicine*, 18(1), 65–72. <https://doi.org/10.1038/gim.2015.34>
- Charmaz, K. (2014). *Constructing Grounded Theory*. SAGE.
- Chase, W. G., & Simon, H. A. (1973). The mind's eye in chess. In *Visual information processing*. (pp. xiv, 555–xiv, 555). Academic.
- Chen, R. C., Gabriel, P. E., Kavanagh, B. D., & McNutt, T. R. (2016). How Will Big Data Impact Clinical Decision Making and Precision Medicine in Radiation Therapy? *International Journal of Radiation Oncology • Biology • Physics*, 95(3), 880–884.
<https://doi.org/10.1016/j.ijrobp.2015.10.052>

- Chetta, A., Carrington, J. M., & Forbes, A. G. (2015). Augmenting EHR Interfaces for Enhanced Nurse Communication and Decision Making. *Proceedings of the 2015 Workshop on Visual Analytics in Healthcare*, 4:1–4:6. <https://doi.org/10.1145/2836034.2836038>
- Chipidza, F. E., Wallwork, R. S., & Stern, T. A. (2015). Impact of the Doctor-Patient Relationship. *The Primary Care Companion for CNS Disorders*, 17(5). <https://doi.org/10.4088/PCC.15f01840>
- Cirillo, D., & Valencia, A. (2019). Big data analytics for personalized medicine. *Current Opinion in Biotechnology*, 58, 161–167. <https://doi.org/10.1016/j.copbio.2019.03.004>
- Clark, H. H. (1996). *Using language*. Cambridge University Press.
- Clark, H. H., & Brennan, S. E. (1991). Grounding in communication. *Perspectives on Socially Shared Cognition*, 13(1991), 127–149.
- Cleveland, W. S., & McGill, R. (1984). Graphical perception: Theory, experimentation, and application to the development of graphical methods. *Journal of the American Statistical Association*, 79(387), 531–554.
- Coble, J. M., Maffitt, J. S., Orland, M. J., & Kahn, M. G. (1995). Contextual inquiry: Discovering physicians' true needs. *Proceedings of the Annual Symposium on Computer Application in Medical Care*, 469–473.
- Coman, I. D., Sillitti, A., & Succi, G. (2008). Investigating the Usefulness of Pair-Programming in a Mature Agile Team. In P. Abrahamsson, R. Baskerville, K. Conboy, B. Fitzgerald, L. Morgan, & X. Wang (Eds.), *Agile Processes in Software Engineering and Extreme Programming* (pp. 127–136). Springer Berlin Heidelberg.
- Corbin, J., & Strauss, A. (2008). *Basics of Qualitative Research (3rd ed.): Techniques and Procedures for Developing Grounded Theory*. SAGE Publications, Inc.
- Creswell, J. W. (2009). *Research design: Qualitative, quantitative, and mixed methods approaches* (3rd ed). Sage Publications.
- Cullis, P. (2015). *The personalized medicine revolution: How diagnosing and treating disease are about to change forever*. Greystone Books.
- De-Miguel, F. F., & Trueta, C. (2005). Synaptic and extrasynaptic secretion of serotonin. *Cellular and Molecular Neurobiology*, 25(2), 297–312.
- Endert, A., Hossain, M. S., Ramakrishnan, N., North, C., Fiaux, P., & Andrews, C. (2014). The human is the loop: New directions for visual analytics. *Journal of Intelligent Information Systems*, 1–25. <https://doi.org/10.1007/s10844-014-0304-9>

- Enfield, N. J., & Levinson, S. C. (2006). *Roots of human sociality: Culture, cognition and interaction*. Berg.
- Ericsson, K. A., & Simon, H. A. (1993). *Protocol Analysis: Verbal Reports as Data*. Bradford Books.
- Falzer, P. R. (2004). Cognitive schema and naturalistic decision making in evidence-based practices. *Journal of Biomedical Informatics*, 37(2), 86–98. <https://doi.org/10.1016/j.jbi.2004.02.002>
- Fantl, J. (2017). Knowledge How. In E. N. Zalta (Ed.), *The Stanford Encyclopedia of Philosophy* (Fall 2017). Metaphysics Research Lab, Stanford University. <https://plato.stanford.edu/archives/fall2017/entries/knowledge-how/>
- Fekete, J.-D., Wijk, J. J., Stasko, J. T., & North, C. (2008). *Information Visualization* (A. Kerren, J. T. Stasko, J.-D. Fekete, & C. North, Eds.; pp. 1–18). Springer-Verlag. http://dx.doi.org/10.1007/978-3-540-70956-5_1
- Fields, R., Marti, P., Termini, V., & Palmonari, M. (2000). *Air Traffic Control as a Distributed Cognitive System: A study of external representations*.
- Fisher, B., Green, T. M., & Arias-Hernández, R. (2011). Visual Analytics as a Translational Cognitive Science. *Topics in Cognitive Science*, 3(3), 609–625. <https://doi.org/10.1111/j.1756-8765.2011.01148.x>
- Gehlenborg, N., O'Donoghue, S. I., Baliga, N. S., Goesmann, A., Hibbs, M. A., Kitano, H., Kohlbacher, O., Neuweger, H., Schneider, R., Tenenbaum, D., & Gavin, A.-C. (2010). Visualization of omics data for systems biology. *Nature Methods*, 7(3), S56–S68. <https://doi.org/10.1038/nmeth.1436>
- Genetic Counsellors | CIHI*. (2019, January 17). Canadian Institute for Health Information. <https://www.cihi.ca/en/genetic-counsellors>
- Ginsburg, G. S., & Willard, H. F. (2009). Genomic and personalized medicine: Foundations and applications. *Translational Research*, 154(6), 277–287. <https://doi.org/10.1016/j.trsl.2009.09.005>
- Graan, A. C. van, Williams, M. J. S., & Koen, M. P. (2016). Professional nurses' understanding of clinical judgement: A contextual inquiry. *Health SA Gesondheid*, 21, 280–293.
- Grandy, G. (2010). Instrumental case study. *Encyclopedia of Case Study Research*, 1, 473–475.
- Grol, R., De Maeseneer, J., Whitfield, M., & Mokkink, H. (1990). Disease-centred versus patient-centred attitudes: Comparison of general practitioners in Belgium, Britain and The Netherlands. *Family Practice*, 7(2), 100–103.

- Gurwitz, D., Weizman, A., & Rehavi, M. (2003). Education: Teaching pharmacogenomics to prepare future physicians and researchers for personalized medicine. *Trends in Pharmacological Sciences*, 24(3), 122–125. [https://doi.org/10.1016/S0165-6147\(03\)00024-5](https://doi.org/10.1016/S0165-6147(03)00024-5)
- Harrison, H., Birks, M., Franklin, R., & Mills, J. (2017). Case Study Research: Foundations and Methodological Orientations. *Forum Qualitative Sozialforschung / Forum: Qualitative Social Research*, 18(1). <http://www.qualitative-research.net/index.php/fqs/article/view/2655>
- Haynes, R. B. (2002). What kind of evidence is it that Evidence-Based Medicine advocates want health care providers and consumers to pay attention to? *BMC Health Services Research*, 2(1), 3. <https://doi.org/10.1186/1472-6963-2-3>
- Hirsch, B. R., & Abernethy, A. P. (2012). Leveraging informatics, mobile health technologies and biobanks to treat each patient right. *Personalized Medicine*, 9(8), 849–857. <https://doi.org/10.2217/pme.12.102>
- Hollan, J., Hutchins, E., & Kirsh, D. (2000). Distributed cognition: Toward a new foundation for human-computer interaction research. *ACM Trans. Comput.-Hum. Interact.*, 7(2), 174–196. <https://doi.org/10.1145/353485.353487>
- Holtzblatt, K., & Beyer, H. (2017). 3—Principles of Contextual Inquiry. In K. Holtzblatt & H. Beyer (Eds.), *Contextual Design (Second Edition)* (pp. 43–80). Morgan Kaufmann. <https://doi.org/10.1016/B978-0-12-800894-2.00003-X>
- Holtzblatt, K., & Beyer, H. (2020). Contextual Design. In *The Encyclopedia of Human-Computer Interaction. 2nd Edition*. <https://www.interaction-design.org/literature/book/the-encyclopedia-of-human-computer-interaction-2nd-ed/contextual-design>
- Hulkko, H., & Abrahamsson, P. (2005). A Multiple Case Study on the Impact of Pair Programming on Product Quality. *Proceedings of the 27th International Conference on Software Engineering*, 495–504. <https://doi.org/10.1145/1062455.1062545>
- Hutchins, E. (1995). *Cognition in the wild*. MIT Press.
- Hutchins, E. (1999). *The Cognitive Consequences of Patterns of Information Flow*. <https://ntrs.nasa.gov/search.jsp?R=20000004537>
- Hutchins, E. (2006). The distributed cognition perspective on human interaction. *Roots of Human Sociality: Culture, Cognition and Interaction*, 375–398.
- James, K. (1982). Complement: Activation, consequences, and control. *Am. J. Med. Technol.*, 48(9), 735–742.

- Julian-Reynier, C., Nippert, I., Calefato, J.-M., Harris, H., Kristoffersson, U., Schmidtke, J., Ten Kate, L., Anionwu, E., Benjamin, C., Challen, K., Plass, A.-M., & Harris, R. (2008). Genetics in clinical practice: General practitioners' educational priorities in European countries. *Genetics in Medicine*, *10*(2), 107–113. <https://doi.org/10.1097/GIM.0b013e3181616693>
- Kaastra, L. T. (2015). *Pair Analytics Research Manual* [Lab research manual].
- Kaastra, L. T., Arias-Hernandez, R., & Fisher, B. (2012). Evaluating Analytic Performance. *Proceedings of the 2012 BELIV Workshop: Beyond Time and Errors - Novel Evaluation Methods for Visualization*, *14*:1–14:3. <https://doi.org/10.1145/2442576.2442590>
- Kaastra, L. T., & Fisher, B. (2014). Field Experiment Methodology for Pair Analytics. *Proceedings of the Fifth Workshop on Beyond Time and Errors: Novel Evaluation Methods for Visualization*, 152–159. <https://doi.org/10.1145/2669557.2669572>
- Kaba, R., & Sooriakumaran, P. (2007). The evolution of the doctor-patient relationship. *International Journal of Surgery*, *5*(1), 57–65. <https://doi.org/10.1016/j.ijisu.2006.01.005>
- Kahneman, D. (2011). *Thinking, Fast and Slow*. Doubleday Canada.
- Kahneman, D., & Klein, G. (2009). Conditions for Intuitive Expertise: A Failure to Disagree. *American Psychologist*, 515–526.
- Kang, Y., & Stasko, J. (2012). Examining the Use of a Visual Analytics System for Sensemaking Tasks: Case Studies with Domain Experts. *IEEE Transactions on Visualization and Computer Graphics*, *18*(12), 2869–2878. <https://doi.org/10.1109/TVCG.2012.224>
- Kang, Youn-ah, & Stasko, J. (n.d.). Characterizing the intelligence analysis process: Informing visual analytics design through a longitudinal field study. *Proc. VAST*, 21–30.
- Kaplan, S. H., Greenfield, S., & Ware Jr, J. E. (1989). Assessing the effects of physician-patient interactions on the outcomes of chronic disease. *Medical Care*, S110–S127.
- Keim, D. A., Mansmann, F., & Thomas, J. (2010). Visual Analytics: How Much Visualization and How Much Analytics? *SIGKDD Explor. Newsl.*, *11*(2), 5–8. <https://doi.org/10.1145/1809400.1809403>
- Kirsh, D. (2006). Distributed cognition: A methodological note. *Pragmatics & Cognition*, *14*(2), 249–262. <https://doi.org/10.1075/pc.14.2.06kir>
- Klein, G. (2008). Naturalistic Decision Making. *Human Factors*, *50*(3), 456–460. <https://doi.org/10.1518/001872008X288385>

- Kleinman, A., Eisenberg, L., & Good, B. (1978). Culture, Illness, and Care: Clinical Lessons From Anthropologic and Cross-Cultural Research. *Annals of Internal Medicine*, 88, 251–258. <https://doi.org/10.1176/foc.4.1.140>
- Kretz, D. R. (2018). Experimentally Evaluating Bias-Reducing Visual Analytics Techniques in Intelligence Analysis. In G. Ellis (Ed.), *Cognitive Biases in Visualizations* (pp. 111–135). Springer International Publishing. https://doi.org/10.1007/978-3-319-95831-6_9
- Kumar, D. (2007). From evidence-based medicine to genomic medicine. *Genomic Medicine*, 1(3–4), 95–104. <https://doi.org/10.1007/s11568-007-9013-6>
- Kumar, S., Hanss, T., Johnson, L., Friedman, C., Donaldson, K., Tyus, J., Omollo, K., & Rubin, J. (2015). Leveraging Contextual Inquiry Methods to Empower Patients in a Learning Health System. *2015 48th Hawaii International Conference on System Sciences*, 3141–3147. <https://doi.org/10.1109/HICSS.2015.379>
- Lachance, C. R., Erby, L. A. H., Ford, B. M., Allen, V. C., & Kaphingst, K. A. (2010). Informational content, literacy demands, and usability of websites offering health-related genetic tests directly to consumers. *Genetics in Medicine*, 12(5), 304–312. <https://doi.org/10.1097/GIM.0b013e3181dbd8b2>
- Laurence, J. (2009). Getting personal: The promises and pitfalls of personalized medicine. *Translational Research*, 154(6), 269–271. <https://doi.org/10.1016/j.trsl.2009.09.010>
- Lavrakas, P. (2019). *Encyclopedia of Survey Research Methods*. <https://doi.org/10.4135/9781412963947>
- Livengood, P., Maciejewski, R., Chen, W., & Ebert, D. S. (2011). A visual analysis system for metabolomics data. *2011 IEEE Symposium on Biological Data Visualization (BioVis)*, 71–78. <https://doi.org/10.1109/BioVis.2011.6094050>
- Manolio, T. A., Chisholm, R. L., Ozenberger, B., Roden, D. M., Williams, M. S., Wilson, R., Bick, D., Bottinger, E. P., Brilliant, M. H., Eng, C., Frazer, K. A., Korf, B., Ledbetter, D. H., Lupski, J. R., Marsh, C., Mrazek, D., Murray, M. F., O'Donnell, P. H., Rader, D. J., ... Ginsburg, G. S. (2013). Implementing genomic medicine in the clinic: The future is here. *Genetics in Medicine*, 15(4), 258–267. <https://doi.org/10.1038/gim.2012.157>
- Marshall, M. N. (1996). Sampling for qualitative research. *Family Practice*, 13(6), 522–526. <https://doi.org/10.1093/fampra/13.6.522>
- Marshall, S. P., & Seel, N. M. (2012). Schemas and Decision Making. In N. M. Seel (Ed.), *Encyclopedia of the Sciences of Learning* (pp. 2956–2959). Springer US. https://doi.org/10.1007/978-1-4419-1428-6_264

- Masic, I., Miokovic, M., & Muhamedagic, B. (2008). Evidence Based Medicine – New Approaches and Challenges. *Acta Informatica Medica*, 16(4), 219–225. <https://doi.org/10.5455/aim.2008.16.219-225>
- McWhinney, I. (1989). The need for a transformed clinical method. *Communicating with Medical Patients*, 9, 25–40.
- Mead, N., & Bower, P. (2000). Patient-centredness: A conceptual framework and review of the empirical literature. *Social Science & Medicine*, 51(7), 1087–1110. [https://doi.org/10.1016/S0277-9536\(00\)00098-8](https://doi.org/10.1016/S0277-9536(00)00098-8)
- Monk, A. (2003). Common ground in electronically mediated communication: Clark's theory of language use. In Carroll (Ed.), *HCI Models, Theoris and Frameworks* (pp. 265–290). Morgan Kaufmann. http://www.ee.upatras.gr/hci/courses/colaborative_technology/Monk_CommonGround.pdf
- Munzner, T. (2009). A Nested Model for Visualization Design and Validation. *IEEE Transactions on Visualization and Computer Graphics*, 15(6), 921–928. <https://doi.org/10.1109/TVCG.2009.111>
- Najafzadeh, M., Lynd, L. D., Davis, J. C., Bryan, S., Anis, A., Marra, M., & Marra, C. A. (2012). Barriers to integrating personalized medicine into clinical practice: A best–worst scaling choice experiment. *Genetics in Medicine*, 14(5), 520–526. <https://doi.org/10.1038/gim.2011.26>
- Nielsen, C. B., Cantor, M., Dubchak, I., Gordon, D., & Wang, T. (2009). Visualizing genomes: Techniques and challenges. *Nature Methods*, 7, S5–S15. <https://doi.org/10.1038/nmeth.1422>
- Nuclino. (2019, May 29). Nuclino: The Easy Knowledge Base for Teams. <https://www.nuclino.com/>
- Pamela Baxter and Susan Jack. (2008). Qualitative case study methodology: Study design and implementation for novice researchers. *The Qualitative Report Article*, 13(4), 544–559.
- Parsons, P., Sedig, K., Mercer, R. E., Khordad, M., Knoll, J., & Rogan, P. (2015). Visual Analytics for Supporting Evidence-based Interpretation of Molecular Cytogenomic Findings. *Proceedings of the 2015 Workshop on Visual Analytics in Healthcare*, 2:1–2:8. <https://doi.org/10.1145/2836034.2836036>
- Parthasarathy, S. (2010). Assessing the social impact of direct-to-consumer genetic testing: Understanding sociotechnical architectures. *Genetics in Medicine*, 12(9), 544–547. <https://doi.org/10.1097/GIM.0b013e3181e71c70>

- Pirolli, P., & Card, S. (2005). The Sensemaking Process and Leverage Points for Analyst Technology as Identified Through Cognitive Task Analysis. *Proceedings of the 2005 International Conference on Intelligence Analysis*. 2005 International Conference on Intelligence Analysis.
- Price, N. D., Magis, A. T., Earls, J. C., Glusman, G., Levy, R., Lausted, C., McDonald, D. T., Kusebauch, U., Moss, C. L., Zhou, Y., Qin, S., Moritz, R. L., Brogaard, K., Omenn, G. S., Lovejoy, J. C., & Hood, L. (2017). A wellness study of 108 individuals using personal, dense, dynamic data clouds. *Nature Biotechnology*. <https://doi.org/10.1038/nbt.3870>
- Ribarsky, W., Fisher, B., & Pottenger, W. M. (2009). Science of Analytical Reasoning. *Information Visualization*, 8(4), 254–262. <https://doi.org/10.1057/ivs.2009.28>
- Sebanz, N., Bekkering, H., & Knoblich, G. (2006). Joint action: Bodies and minds moving together. *Trends in Cognitive Sciences*, 10(2), 70–76. <https://doi.org/10.1016/j.tics.2005.12.009>
- Sedig, K., & Parsons, P. (2013). Interaction Design for Complex Cognitive Activities with Visual Representations: A Pattern-Based Approach. *AIS Transactions on Human-Computer Interaction*, 5(2), 84–133.
- Sedig, K., Parsons, P., Naimi, A., & Willoughby, K. (2015). Reconsidering healthcare evidence as dynamic and distributed: The role of information and cognition. *International Journal of Evidence-Based Healthcare*, 13(2), 43–51. <https://doi.org/10.1097/XEB.0000000000000030>
- Sedlmair, M., Meyer, M., & Munzner, T. (2012). Design Study Methodology: Reflections from the Trenches and the Stacks. *IEEE Transactions on Visualization and Computer Graphics*, 18(12), 2431–2440. <https://doi.org/10.1109/TVCG.2012.213>
- Shanteau, J. (1992). Competence in experts: The role of task characteristics. *Organ. Behav. Hum. Decis. Process.*, 53, 252–252.
- Shneiderman, B. (1994). Dynamic queries for visual information seeking. *Software, IEEE*, 11(6), 70–77. <https://doi.org/10.1109/52.329404>
- Shneiderman, B., Plaisant, C., & Hesse, B. W. (2013). Improving Healthcare with Interactive Visualization. *Computer*, 46(5), 58–66. <https://doi.org/10.1109/MC.2013.38>
- Shneiderman, Ben, & Plaisant, C. (2006). Strategies for Evaluating Information Visualization Tools: Multi-dimensional In-depth Long-term Case Studies. *Proceedings of the 2006 AVI Workshop on BEyond Time and Errors: Novel Evaluation Methods for Information Visualization*, 1–7. <https://doi.org/10.1145/1168149.1168158>

- Sillence, E., Briggs, P., Harris, P., & Fishwick, L. (2006). A framework for understanding trust factors in web-based health advice. *International Journal of Human-Computer Studies*, 64(8), 697–713. <https://doi.org/10.1016/j.ijhcs.2006.02.007>
- Simon, H. A. (1988). The Science of Design: Creating the Artificial. *Design Issues*, 4(1/2), 67–82. JSTOR. <https://doi.org/10.2307/1511391>
- Sittig, D. F., Wright, A., Osheroff, J. A., Middleton, B., Teich, J. M., Ash, J. S., Campbell, E., & Bates, D. W. (2008). Grand challenges in clinical decision support. *Journal of Biomedical Informatics*, 41(2), 387–392. <https://doi.org/10.1016/j.jbi.2007.09.003>
- Stake, R. E. (1995). *The Art of Case Study Research*. SAGE.
- Stewart, M., Brown, J. B., Weston, W., McWhinney, I. R., McWilliam, C. L., & Freeman, T. (2013). *Patient-centered medicine: Transforming the clinical method*. CRC press.
- Szasz, T. S., & Hollender, M. H. (1956). A Contribution to the Philosophy of medicine: The Basic Models of the Doctor-Patient Relationship. *A.M.A. Archives of Internal Medicine*, 97(5), 585–592. <https://doi.org/10.1001/archinte.1956.00250230079008>
- The HIBAR Research Alliance*. (2020). <https://hibar-research.org/>
- Thomas, J. J., & Cook, K. A. (Eds.). (2005). *Illuminating the Path: The Research and Development Agenda for Visual Analytics*. National Visualization and Analytics Ctr.
- Topol, E. J. (2019). High-performance medicine: The convergence of human and artificial intelligence. *Nat. Med.*, 25(1), 44–56.
- Upshur, R. E. G., & Colak, E. (2003). Argumentation and Evidence. *Theoretical Medicine and Bioethics*, 24(4), 283–299. <https://doi.org/10.1023/A:1026006801902>
- Vasileiou, K., Barnett, J., Thorpe, S., & Young, T. (2018). Characterising and justifying sample size sufficiency in interview-based studies: Systematic analysis of qualitative health research over a 15-year period. *BMC Medical Research Methodology*, 18(1), 148. <https://doi.org/10.1186/s12874-018-0594-7>
- Visualization in Practice: Personalized Health as a Human-Centered Design Process at Molecular You*. (2017). [Poster Abstracts]. Personalized Medicine Summit, Vancouver, BC.
- Wade, A. (2011). *Visual analytics for aviation safety: A collaborative approach to sensemaking* [MSc. Thesis, Simon Fraser University]. <http://proxy.lib.sfu.ca/login?url=https://search-proquest-com.proxy.lib.sfu.ca/docview/1624924737?accountid=13800>

- Ware, C. (2008). *Visual Thinking: For Design* (1st ed.). Morgan Kaufmann.
- Weldon, C. B., Trosman, J. R., Gradishar, W. J., Benson, A. B., & Schink, J. C. (2012). Barriers to the Use of Personalized Medicine in Breast Cancer. *Journal of Oncology Practice*, 8(4), e24–e31. <https://doi.org/10.1200/JOP.2011.000448>
- Wieten, S. (2018). Expertise in evidence-based medicine: A tale of three models. *Philosophy, Ethics, and Humanities in Medicine : PEHM*, 13. <https://doi.org/10.1186/s13010-018-0055-2>
- Worrall, J. (2002). What Evidence in Evidence-Based Medicine? *Philosophy of Science*, 69(S3), S316–S330. <https://doi.org/10.1086/341855>

Appendix A. Pair Analysis Session Instruction Sheet for Participants

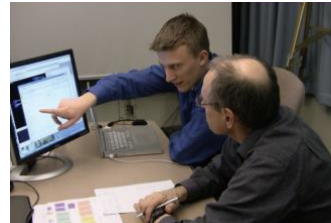
MY-Experts: Creating Health Data Summaries, Action Plans, and Performing QA/QC

Summary

During this session you will work in collaboration with a visual analytics intern on one of the reports from the current batch. The goal is to create a health data summary (or an action plan) by instructing the intern step by step where to begin and how to proceed. We want to learn about different pieces of the data that are relevant, the use of the tools available, and your reasoning process.

The goals of the session are:

- To instruct and create a health data summary or action plan
- To capture information hierarchies
- To capture the support and the gaps from tools



The session will develop as:

- Part 01: [5 mins] Setup and protocols
- Part 02: [35-40 mins] Collaborative analysis session.
- Part 03: [10 mins] Analyst feedback wrap-up

Data Collection

During the session, and with your consent, we will be recording research data using screen capture, audio recording, and notes.



Appendix B. Consent Form



Informed Consent Form

Research Project Title

Translational Research in Visual Analytics for Personalized Health

Principal Investigator

Dr. Brian Fisher, Ph.D. Simon Fraser University and University of British Columbia

E-mail: removed

Phone: removed

Researchers

Nadya Calderón, PhD student Simon Fraser University

Aldo Barrera, MSc student, Simon Fraser University

Primary Contact

Nadya Calderón

E-mail: removed

Phone: removed

This informed consent form describes the purpose of the study and what your participation will entail. If you have questions or would like more detail, you should feel free to ask the investigator(s). Please take the time to read this carefully and be certain that you understand the information.

Your signature on this form will signify that you have been informed about the goals of the study, the procedures to be used, and any potential risks and/or benefits of your participation. Your signature signifies that you understand the goals, risks, and procedures, and that you voluntarily agree to participate in this study.

Purpose

Personalized health is based on the discovery that individuals have a unique molecular makeup, which, in part, defines or reflects their susceptibility to certain diseases. Scientists and other healthcare professionals require advanced data visualization strategies to interpret personalized health data and turn it into actionable recommendations. We have previously designed interfaces allowing for the display of personalized health data. In the current study, we propose to use a research approach that happens in the context of your daily work life, to explore the cognitive flow of scientists and healthcare professionals in the use of our interfaces. This will allow us to evaluate and improve the design of our interfaces.

Sponsor:

This research study is sponsored by Molecular You inc. and MITACS as part of the accelerate grant program.

Why Have I Been Invited?

You have been invited to participate of these user sessions because you are considered a member of the scientific team at Molecular You or a healthcare professional working with Molecular You.

What Will I Be Asked To Do?

If you agree to participate, we will schedule a time that is convenient to you outside your work hours during which you will be invited to come to a single session (~1 hour) in an office at the MAGIC lab (Forestry Building, UBC Campus).

You will be presented with a selected case for analysis. The case scenarios are a new dataset on the Molecular You environment you are trained and used to work. You will be asked to verbalize aloud your thinking process while performing the analysis of the case scenarios. Describe *what* and *why* are you taking each step on the process. Examples of the verbalization process includes narrating why are you clicking on a specific piece of information (e.g. *I will select the item Diabetes" because is the first in the ranked list*), or what are you thinking during periods of no interactivity with the software (e.g. *I am thinking that the client didn't report having history for this condition but that is not congruent with what I am seeing*).

We will record the screen using screen capture software. This procedure doesn't take pictures of you. Additionally, during this time, Nadya or Aldo will sit, observe, and take notes but never intervene or interrupt your analysis process.

At the end of the interpretation part, Nadya or Aldo will conduct a semi-structured interview to clarify any details that they could have, expand on your experience, and reflect on specific moments of your analysis.

My Participation

Participation is voluntary. If you agree to participate, you are free to withdraw at any time for any reason. All you study data will be destroyed if you decide to withdraw.

What Type of Personal Information Will Be Collected?

Activities that require asking for your opinion or experience will be recorded with note taking and voice recorders. We will take screen captures of the workflow and activities during the session.

Your confidentiality will be strictly maintained. Any data collected will be kept confidential to only the researchers involved. For publication of research outcomes, we will use only descriptions of the tasks and results.

Molecular You will not know that you have agreed to participate in the study. They will not be able to link your responses to you. Audio recordings will only be accessible to Brian Fisher, Nadya Calderon and Aldo Barrera (the researchers), and will not be shared with any personnel at Molecular You.

Are There Risks or Benefits if I Participate?

A potential risk may be that participants might be identified from quotes that are being recorded during the study, given the small group of eligible participants. We are taking this risk into consideration and all notes will be de-identified as well as quotes will be edited, e.g. removing identifiable information such as specific job descriptions or tasks, before being accessible to anyone outside the study team, i.e. Nadya Calderon, Aldo Barrera, and Brian Fisher.

During this research participants will not be expected to do anything that may be harmful to themselves or others. When observed or interviewed, you, as a participant, will not be required to provide any comment about your organization. The observations and interviews will focus solely on analytical work, and use of information and communication technologies, and not on any other aspect of your organization or agency.

What Happens to the Information I Provide?

No one other than the involved researchers will be permitted to see or hear any recordings or interviews. Data is kept in a secure location at UBC and will only be accessible by the involved researchers at

Molecular You Inc. and their collaborators in Dr. Fisher's laboratory at Simon Fraser University. All data collected as part of the study will be kept on a secure server computer in Canada.

We expect to publish reports and presentations describing this research. Public presentations of the results will primarily present the results in an aggregate form that combines the actions and opinions of more than one participant. Where individual participant data is disclosed, such as exemplar interview comments or quotes, we will ensure that the selected data does not suggest participant identities.

Acceptance of this Form

Your signature on this form indicates that you 1) understand to your satisfaction the information provided to you about your participation in this research project, and 2) agree to participate as a research subject. In no way does this waive your legal rights nor release the investigators, sponsors, or involved institutions from their legal and professional responsibilities. You are free to withdraw from this research project at any time. You should feel free to ask for clarification or additional information from the investigators at any time during your participation.

Participant's Name (please print) _____

Participant's Signature _____

Date _____

Questions/Concerns

If you have any concerns or complaints about your rights as a research participant and/or your experiences while participating in this study, contact the Research Participant Complaint Line in the UBC Office of Research Ethics at 604-822-8598 or if long distance e-mail RSIL@ors.ubc.ca or call toll free 1-877-822-8598.

Appendix C. Transcripts

The full dataset of screen recordings with voice over and transcripts belong to the Science-Lab under direction of professor Brian Fisher. Materials can be asked to be accessed for research purposes with the author or professor Fisher.