Interpreting Direct-to-Consumer Genetic Tests in the Public Health System: Exploring the Trade-offs

by

Carla Beak

B.Sc. (Hons., Cell & Molecular Biology), Simon Fraser University, 2002

RESEARCH PROJECT SUBMITTED IN PARTIAL FULFILLMENT
OF THE REQUIREMENTS FOR THE DEGREE OF
MASTER OF PUBLIC POLICY

in the
School of Public Policy
Faculty of Arts and Social Sciences

© Carla Beak 2012

SIMON FRASER UNIVERSITY

Spring 2012

All rights reserved.
However, in accordance with the Copyright Act of Canada, this work may be reproduced, without authorization, under the conditions for “Fair Dealing.” Therefore, limited reproduction of this work for the purposes of private study, research, criticism, review and news reporting is likely to be in accordance with the law, particularly if cited appropriately.
Approval

Name: Carla Beak
Degree: Master of Public Policy

Examining Committee:

Chair: Firstname Surname, Position

Firstname Surname
Senior Supervisor
Assistant/Associate/Professor

Firstname Surname
Supervisor
Assistant/Associate/Professor

Firstname Surname
Assistant/Associate/Professor
Supervisor

Firstname Surname
Supervisor

Firstname Surname
Internal Examiner
Assistant/Associate/Professor
School/Department or Faculty

Firstname Surname
External Examiner
Assistant/Associate/Professor, Department
University

Date Defended: March 23, 2012
Partial Copyright Licence

The author, whose copyright is declared on the title page of this work, has granted to Simon Fraser University the right to lend this thesis, project or extended essay to users of the Simon Fraser University Library, and to make partial or single copies only for such users or in response to a request from the library of any other university, or other educational institution, on its own behalf or for one of its users.

The author has further granted permission to Simon Fraser University to keep or make a digital copy for use in its circulating collection (currently available to the public at the "Institutional Repository" link of the SFU Library website (www.lib.sfu.ca) at http://summit/sfu.ca and, without changing the content, to translate the thesis/project or extended essays, if technically possible, to any medium or format for the purpose of preservation of the digital work.

The author has further agreed that permission for multiple copying of this work for scholarly purposes may be granted by either the author or the Dean of Graduate Studies.

It is understood that copying or publication of this work for financial gain shall not be allowed without the author's written permission.

Permission for public performance, or limited permission for private scholarly use, of any multimedia materials forming part of this work, may have been granted by the author. This information may be found on the separately catalogued multimedia material and in the signed Partial Copyright Licence.

While licensing SFU to permit the above uses, the author retains copyright in the thesis, project or extended essays, including the right to change the work for subsequent purposes, including editing and publishing the work in whole or in part, and licensing other parties, as the author may desire.

The original Partial Copyright Licence attesting to these terms, and signed by this author, may be found in the original bound copy of this work, retained in the Simon Fraser University Archive.

Simon Fraser University Library
Burnaby, British Columbia, Canada

revised Fall 2011
Ethics Statement

The author, whose name appears on the title page of this work, has obtained, for the research described in this work, either:

a. human research ethics approval from the Simon Fraser University Office of Research Ethics,

or

b. advance approval of the animal care protocol from the University Animal Care Committee of Simon Fraser University;

or has conducted the research

  c. as a co-investigator, collaborator or research assistant in a research project approved in advance,

or

  d. as a member of a course approved in advance for minimal risk human research, by the Office of Research Ethics.

A copy of the approval letter has been filed at the Theses Office of the University Library at the time of submission of this thesis or project.

The original application for approval and letter of approval are filed with the relevant offices. Inquiries may be directed to those authorities.

Simon Fraser University Library
Burnaby, British Columbia, Canada

update Spring 2010
Abstract

Direct-to-consumer (DTC) genetic testing poses a growing problem in the field of health policy. Consumers who purchase the tests often do not have the knowledge required to interpret the tests and make informed decisions related to their care. They then turn to health care providers to interpret test results, many who also have limited knowledge of genetics. This may lead to decisions regarding further testing, treatment and referrals that do not benefit the patient and waste health care resources. Using interviews of stakeholders in the fields of health, genetics, education and ethics, I performed a thematic analysis to identify the key issues facing the health system in relation to DTC genetic testing. I then identified policy options available to the health system to aid physician decision-making. I evaluated the tradeoffs between the two most feasible policy options: promoting the Medical Genetics P&P telephone support line, and expanding HealthLink BC.

Keywords: Direct-to-consumer genetic testing; physician education; genetic counsellors; Medical Genetics P&P line; HealthLink BC.
Dedication

In remembrance of my mom and sis. The memory of their strength gave me strength.

“I think there is a popular myth that information is value neutral and that . . . more information is necessarily a good thing. But with information comes responsibility.”

C. Ben Mitchell, Trinity Evangelical Divinity School
(Genetics & Public Policy Center, Reproductive Genetic Testing: Issues and Options for Policy Makers, 2004)
Acknowledgements

I am deeply grateful for the overwhelming support and the invaluable contributions I obtained throughout this entire process. Thank you…

– to the many interview participants, for their generosity when sharing their time and their openness when sharing their insights. Their energy and passion for their work was motivating. Their drive and desire for improvement was inspiring.

– to my supervisor Olena Hankivsky, for sharing her wisdom, support and advice. Also for giving me the confidence to believe that I had the strength to accomplish my academic goals while dealing with my personal challenges.

– to my husband, for supporting me in every possible way. I am truly blessed.

— to my friends and family, for their patience, understanding and encouragement. With a special shout-out to those that could commiserate.
# Table of Contents

Approval ................................................................................................................................. ii
Abstract ................................................................................................................................... iii
Dedication ............................................................................................................................... iv
Acknowledgements .............................................................................................................. v
Table of Contents ................................................................................................................ vi
List of Tables ........................................................................................................................ viii
List of Acronyms .................................................................................................................... ix
Executive Summary .............................................................................................................. x

1. Introduction: .......................................................................................................................... 1
   1.1. The Genetics Revolution ............................................................................................... 2
   1.2. Direct-to-Consumer Genetic Testing ............................................................................. 4
   1.3. Risks and Benefits of DTC Genetic Tests ................................................................. 7
   1.4. Current State of Affairs ............................................................................................... 10
       1.4.1. Use of DTC Genetic Tests ................................................................................ 11
   1.5. What next? .................................................................................................................. 13

2. Methodology .......................................................................................................................... 15
   2.1. Interviews ................................................................................................................... 16
   2.2. Thematic Analysis of Interviews .............................................................................. 17

3. Thematic Analysis .................................................................................................................. 20
   3.1. Technology Theme ...................................................................................................... 20
   3.2. System Theme ............................................................................................................ 24
   3.3. Physician Theme ........................................................................................................ 26
   3.4. Genetic Counselor Theme ......................................................................................... 28
   3.5. Patient Theme ............................................................................................................ 29
   3.6. Summary .................................................................................................................... 30

4. Policy Options ....................................................................................................................... 32
   4.1. Status Quo .................................................................................................................... 32
   4.2. Basic Education .......................................................................................................... 34
   4.3. Guidelines ................................................................................................................... 36
   4.4. Electronic Tools ........................................................................................................... 37
   4.5. Compensation System ............................................................................................... 38
   4.6. Access to Genetic Specialists .................................................................................... 38
   4.7. Summary .................................................................................................................... 40

5. Analysis ................................................................................................................................ 41
   5.1. Preliminary Assessment of Policy Options: Criteria Matrix ..................................... 41
       5.1.1. Summary of the Preliminary Assessment ............................................................ 44
   5.2. Further Development of Leading Options ................................................................. 45
       5.2.1. Electronic Tools: HealthLink BC ........................................................................ 45
       5.2.2. Access to Genetic Specialists: P&P line .......................................................... 47
   5.3. Evaluation of Leading Options: Multiple Account Benefit-Cost Analysis ............... 49
<table>
<thead>
<tr>
<th>Section</th>
<th>Title</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>5.3.1.</td>
<td>Summary of the Leading Options</td>
<td>52</td>
</tr>
<tr>
<td>5.4.</td>
<td>Recommendation</td>
<td>52</td>
</tr>
<tr>
<td>6.</td>
<td>Areas for further research</td>
<td>54</td>
</tr>
<tr>
<td>6.1.</td>
<td>The Public's Role</td>
<td>54</td>
</tr>
<tr>
<td>6.2.</td>
<td>Personalised Medicine in BC</td>
<td>54</td>
</tr>
<tr>
<td>6.3.</td>
<td>The Role of the Federal Government</td>
<td>55</td>
</tr>
<tr>
<td>6.4.</td>
<td>The Role of Other Health Care Professionals</td>
<td>55</td>
</tr>
<tr>
<td>6.5.</td>
<td>Dysfunctional System?</td>
<td>56</td>
</tr>
<tr>
<td>7.</td>
<td>Conclusion</td>
<td>57</td>
</tr>
<tr>
<td></td>
<td>References</td>
<td>58</td>
</tr>
<tr>
<td></td>
<td>Appendices</td>
<td>63</td>
</tr>
<tr>
<td></td>
<td>Appendix A. Risks and Benefits of DTC Genetic Testing</td>
<td>64</td>
</tr>
</tbody>
</table>
List of Tables

Table 1. Risks and benefits of DTC genetic testing ................................................................. 8
Table 2. Description of criteria used in the evaluation matrix .............................................. 42
Table 3. Criteria matrix of the preliminary evaluation of policy options .............................. 43
Table 4. Policy option evaluation: multiple account benefit-cost analysis......................... 51
# List of Acronyms

<table>
<thead>
<tr>
<th>Acronym</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>AHRQ</td>
<td>Agency for Healthcare Research and Quality</td>
</tr>
<tr>
<td>BCCGN</td>
<td>British Columbia Clinical Genomics Network</td>
</tr>
<tr>
<td>BCMA</td>
<td>British Columbia Medical Association</td>
</tr>
<tr>
<td>BCMJ</td>
<td>British Columbia Medical Journal</td>
</tr>
<tr>
<td>CHARD</td>
<td>Community Health Access Resource Database</td>
</tr>
<tr>
<td>CHSRF</td>
<td>Canadian Health Service Research Foundation</td>
</tr>
<tr>
<td>CME</td>
<td>Continuing Medical Education</td>
</tr>
<tr>
<td>CPD</td>
<td>Continuing Professional Development</td>
</tr>
<tr>
<td>DTC</td>
<td>Direct-to-consumer</td>
</tr>
<tr>
<td>EGAPP</td>
<td>Evaluation of Genomic Application in Practice and Prevention</td>
</tr>
<tr>
<td>FDA</td>
<td>Federal Drug Administration</td>
</tr>
<tr>
<td>GAO</td>
<td>Government Accountability Office</td>
</tr>
<tr>
<td>GPAC</td>
<td>Guidelines and Protocols Advisory Committee</td>
</tr>
<tr>
<td>GPPC</td>
<td>Genetics &amp; Public Policy Center</td>
</tr>
<tr>
<td>GTR</td>
<td>Genetic Testing Registry</td>
</tr>
<tr>
<td>GWAS</td>
<td>Genome-wide Association Studies</td>
</tr>
<tr>
<td>HGP</td>
<td>Human Genome Project</td>
</tr>
<tr>
<td>IVDDs</td>
<td><em>In vitro</em> Diagnostic Devises</td>
</tr>
<tr>
<td>LDT</td>
<td>Laboratory Developed Test</td>
</tr>
<tr>
<td>MSP</td>
<td>Medical Services Plan</td>
</tr>
<tr>
<td>NCBI</td>
<td>National Center for Biotechnology Information</td>
</tr>
<tr>
<td>NCHPEG</td>
<td>National Coalition for Health Professional Education in Genetics</td>
</tr>
<tr>
<td>P&amp;P</td>
<td>Phones and Postals</td>
</tr>
<tr>
<td>RACE</td>
<td>Rapid Access to Consultative Expertise</td>
</tr>
<tr>
<td>REB</td>
<td>Research Ethics Board</td>
</tr>
<tr>
<td>SACGHS</td>
<td>Secretary’s Advisory Committee on Genetics, Health, and Society</td>
</tr>
<tr>
<td>SFU</td>
<td>Simon Fraser University</td>
</tr>
<tr>
<td>UBC</td>
<td>University of British Columbia</td>
</tr>
</tbody>
</table>
Executive Summary

Introduction

The desire for personalized medicine, where health interventions are tailored to each individual, has driven the development and demand for technologies that offer information about an individual's unique biology. One such technology is genetic testing, which can now be cost-effectively advertised and sold directly to the public without the involvement of health care providers. Companies that offer direct-to-consumer (DTC) genetic tests provide a service they claim will empower and educate customers, and will ultimately help predict, prevent and treat common conditions such as cardiovascular disease, diabetes, Alzheimer’s Disease. However, a number of stakeholders have called for tighter regulation of the DTC genetic testing market over concerns that these tests lack clinical validity, accuracy and utility. In addition, they fear that a lack of genetics knowledge in the health care system may lead to sub-optimal patient management and inappropriate use of health care system resources, such as physician time, specialist services and follow-up tests.

Acknowledging the regulation of the DTC genetic testing industry is uncertain, provincial health care systems would be wise to monitor and manage how health care providers handle these tests. As such, the problem I identified is that **BC lacks a strategy for the management of direct-to-consumer genetic tests to ensure consumers get accurate information while minimizing the use of health care resources.** The province is in an optimal position to proactively manage this emerging technology before unintended consequences to the health care system are felt. In order to capitalize on this opportunity to gather information, increase skill sets, improve care and plan for the future, I investigated the management of genetic tests in the health care system. I identified key actors and explored their roles in order to identify what is required to ensure patient questions regarding DTC genetic tests are managed appropriately.
Methodology

Initial research utilized academic journals, grey literature and informal exploratory interviews to determine the focus of the project as it related to the management of DTC genetic tests in the health care system. Through this, I identified the lack of physician education and knowledge of genetics as an impediment to optimal decision-making. Since family physicians are considered the “first line of defence”, their lack of knowledge poses major risks to the system if and when consumers of DTC genetic services seek advice. My research then centered on how to address this concern in BC. I undertook semi-structured interviews (n=26) using purposive and snowball sampling to explore this topic among stakeholders representing different interests within the health system or genetics industry. This included geneticists, genetic counsellors, family physicians, physician/genetics educators, and academics specializing in genomics policy. A thematic analysis was then performed to identify common themes among the interviews.

Thematic Analysis

Five main themes arose from the analysis of the interviews. The Technology theme focused on genetic testing technologies and the opportunities and challenges they bring, the System theme explored how genetic testing is managed in BC, the Physician theme looked at the strengths, weaknesses and needs of physicians in the area of genetics, the Genetic Counsellor theme explored their strengths and weaknesses and how they can be optimally utilized, and the Patient theme looked at what patients want from the health system and genetic testing. A common thread throughout all themes and conversations with interview participants was the central role of the family physician as front line responders. DTC genetic testing for susceptibility to common diseases was seen as the purview of family physicians. Unfortunately, it was also felt that physicians currently lack the knowledge and confidence to manage these tests appropriately. As such, the relevant policy question then became: **how do physicians get the education and/or resources they need to identify and fulfill the obligations within their scope of practice, while ensuring resources are used most effectively?**
Participants provided a number of suggestions for how to improve physicians’ knowledge and skills in DTC genetic tests management. The main strategies include: basic physician education in genetics via medical school or Continuing Professionals Development (CPD), the development of professional guidelines for physicians, the provision of electronic tools for physicians that provide relevant information, the expansion of the compensation system to include billing codes for activities related to DTC genetic testing, and improving physician access to genetic specialists (specifically genetic counsellors) to aid in patient management. To narrow down these broad policy options, a criteria matrix was used to identify the most optimal options for implementation in BC. While options are not mutually exclusive and all have potential utility, based on political and administrative feasibility, efficiency, and effectiveness two leading options emerged: the provision of electronic tools and access to genetic specialists.

Additional interviews and online research was performed to present a more detailed description of the options and explore their suitability for implementation in BC. For the development of electronic tools, I suggested the expansion of the HealthLink BC website to include information relating to DTC genetic testing. It is a public website, however it contains a number of additional components targeted directly to health care providers, namely Health Files fact sheets and the CHARD referral service. To provide access to genetic specialists, I suggested the promotion of the existing Medical Genetics department consultative service telephone line, the “P&P line”. This option involves increasing the awareness among family physicians that the phone line exists for their use, and that the genetic counsellors involved are able to address questions regarding DTC genetic testing. In order to compare these options a multiple account benefit-cost analysis was performed. This analysis was used to highlight the trade-offs inherent in each policy option (the broad and general HealthLink BC and the targeted and specific P&P line), compared to the status quo. It looked at the benefits and costs of the policies from the perspective of those impacted through three accounts: the Government Account (Ministry of Health), the User Account (family physicians), and the Social Account (patients).
Recommendation

In the short term, I recommend the expansion of the HealthLink BC website. Implementation of this option would provide information to both physicians and patients, it would begin engagement between the Ministry of Health and other stakeholders, and it can also serve as a platform to promote the P&P line. In the medium term, data from the website analytics and Medical Genetics department can be analysed to determine what content is being used and what issues are making their way into the health care system. This can aid in health system planning. In the long term, the system can capitalize on the increased dialogue between the Ministry and those involved in genetic services to improve the way all genetic services are managed in BC.

Areas for Future Research & Conclusions

A number of other relevant areas of interest came out of the research. This includes a broader analysis of genetic testing and the role of the public, how it fits in with personalized medicine efforts in BC, the role of the federal government, the role of other health care providers, and the functionality of the current system in BC. However, the focus of this capstone was how decision-makers can avoid the unintended consequences of DTC genetic testing on the health care system. In particular, it explored the role of the physician in managing DTC genetic tests. The capstone explored how physician skills can be improved through the provision of education and resources. Ideally, this would help physicians provide effective care without needlessly utilizing health care resources, such as specialist services and follow-up tests. As an up-and-coming health care issue, decision-makers in other jurisdictions who want to take a proactive approach to the management of new technologies may benefit from this work.
1. Introduction:

Technology continues to evolve at a rapid pace, and the tools that make personalized medicine a realizable dream are within grasp. Personalized medicine tailors health interventions to the individual, based on their susceptibility to a disease or responsiveness to a treatment, with the aim to achieve optimal health outcomes by helping physicians choose a disease management approach most likely to succeed based on the patient’s unique biology (Constant et al., 2011). As individuals begin to take personal control of their health and well being, they are beginning to expect customized service in their health care management and treatment. It is a desire for this personalized, technologically advanced type of service that makes direct-to-consumer (DTC) genetic testing alluring to the public.

Acting outside of the traditional health care system, companies offering DTC genetic testing services use new technologies and advances in genetic knowledge to provide a service that they claim will help their customers predict, prevent and treat common diseases and conditions the public has come to fear. These services have the potential to be invaluable and vital to health management. It is for this reason why on January 31st of this year the federal government announced a $67.5 million dollar investment to support funding of research teams in the area of Personalized Health (CIHR, January 31, 2012). However, this was shortly followed by a Globe and Mail article that highlighted the debates around whether we have enough information for genetic-profiling to be useful and how this information may affect individual behaviour (Weeks, February 10, 2012). In addition, in a 2011 report on cost drivers in health care, the Canadian Health Services Research Foundation (CHSRF) identified personalized medicine and direct-to-consumer advertising of genetic tests as potential emerging factors that may exert additional pressures on health care utilization and increased health expenditure in the future (Constant et al., 2011).

In light of these developments, the field of DTC genetic testing is evolving quickly and warrants attention. Genetic testing offered through the DTC business model,
characterized by companies offering genetic services directly to customers online, is in its early stages. The public health system is still in a position to manage this new technology proactively; ensuring health care resources are appropriately and efficiently used. In that sense, DTC genetic testing provides the health care system an opportunity to gather information, increase skill sets, improve quality of care, develop strategies for the future and ultimately improve health outcomes. If this is done, some of the potential risks of this new technology can arguably be avoided, such as growing numbers of patients purchasing confusing services, increasing physician visits, rising referrals for genetic specialists, requests for potentially unnecessary tests, and a gradual drain of health care resources.

The problem I aim to address is that BC lacks a strategy for the management of direct-to-consumer genetic tests to ensure consumers get accurate information while minimizing the use of health care resources. My goal is to illustrate some of the issues and opportunities that will arise as personalized medicine evolves and DTC genetic testing becomes more prevalent. In particular, I will explore the ramifications of predictive DTC genetic tests, ones that calculate the risk of developing complex conditions, because the costs and benefits of these tests are most controversial. I will undertake an exploration of whether the health care system, and health care providers within it, are prepared to manage patient questions and concerns regarding genetic results obtained from a DTC genetic testing company. My aim is to identify who the key actors are, explore what roles they could or should be playing, and identify the resources required to execute on those roles. From this I present policy recommendations that focus on educating and equipping family physicians on the subject, as they were identified as the “gatekeeper” and “first line of defence” in managing this patient-driven technology. I will also highlight some of the principles and values that informed the analysis. But first, I will provide background information on the state of affairs of DTC genetic testing.

1.1. The Genetics Revolution

In 2000 Craig Venter and Francis Collins stood by President Clinton at the White House to accept recognition for one of the greatest scientific endeavours since the moon
landing – the sequencing of the human genome. At the time, it was predicted that the genome would reveal the causes of most common diseases and open the door for a variety of treatments and cures. Individuals would be prescribed specific drugs and therapies based on the mutations that led to their illness. Better yet, individuals could learn what diseases they were at risk for and engage in preventative measures before symptoms even appear. It was the beginning of personalized medicine.

While this model still serves as the ideal that the medical and scientific communities strive towards, there has also been a keen awareness that the Human Genome Project did not lead to the medical revolution that was expected (see Box 1). Genetic testing, while common in medical practice for a variety of diagnostic purposes, has had limited use therapeutically. This is due in part to the inability of scientists to connect genetic variations to the nature and occurrence of complex diseases (Box 1), but also due systemic barriers to the integration of this new technology (PMC, 2012). Despite the lack of initial success, there is still hope that genetic information will allow individuals to personalize and improve their health care choices.
1.2. Direct-to-Consumer Genetic Testing

This hope has led to a new phenomenon – direct-to-consumer (DTC) genetic testing. It is a new business model for genetic testing in which private companies advertise and sell genetic testing services directly to the public, often without the involvement of a health care practitioner. Customers provide biological samples (saliva or blood) directly to a company (in person or through the mail) and receive the subsequent genetic results from the company, usually on-line. Perhaps the most well known (and used) DTC genetic testing company is 23andMe, popular for its low price tag ($99 per kit plus a $9 per month service subscription). Another industry leader is Navigenics, which makes its genetic analysis services available through physicians and corporate wellness programs (at a higher price). Another company that received a great amount of media attention in 2010 is Pathway Genomics. The company made headlines when its partnership to sell its saliva collection kits at Walgreens nationwide was
abandoned due to Federal Drug Administration (FDA) concerns that regulatory approval for the product had not been obtained. Presently, they only sell their services through physicians.

There are currently at least 21 companies offering DTC genetic services (Box 2). The genetic testing services offered by these companies vary. Some offer non-health related personal information services, such as ancestry testing or the identification of personal traits (ranging from baldness to artistic ability). Others offer reproductive related services, such as paternity testing or carrier testing of prospective parents to determine their chances of having a child with a genetic disorder. Pharmacogenomic testing services are also available, which provide information on an individual’s genetic response to pharmaceutical drugs. But the most common genetic testing services are those used for the assessment of disease and other health related conditions. This would include tests for things such as nutritional status (nutrigenomics), migraines, diabetes, schizophrenia, cancer, HIV or Huntington’s Disease. What becomes apparent from this overview is that while some of the genetic tests can be seen as a form of entertaining self-exploration, others can have serious repercussions – test results may be used to plan whether one has a child, undertakes preventative surgeries, or continues disease monitoring (Williams-Jones & Ozdemir, 2008). The consequences of inaccurate information may be severe.

The DTC genetic testing business model has been made possible by technological innovations in two key areas. First, there has been tremendous progress in genetic science technology that has made sequencing the genome relatively quick and cost effective. This has allowed scientists to undertake numerous studies exploring the relationship between genes and disease, and allowed companies to provide genetic testing services at consumer-friendly prices. Second, advances in computer technology, most importantly the Internet, have made a wealth of genetic information efficient to generate and publically available. This has provided companies with the scientific data they need to correlate an individual’s genetic markers with specific diseases, and a tool by which they can access consumers – who increasingly rely on the Internet for health information and to purchase services and products (McBride et al., 2010).
Genetic tests offered directly to consumers present a new paradigm in medical testing in several ways. First, many of the tests sold over the Internet have not undergone any form of clinical evaluation. Second, tests can be obtained without having a health care professional involved in ordering or interpreting the results. Third, the health value and personal ramifications of these tests are unclear. In relation to this last point, many of the tests offered are not generally indicative that a disease or condition exists, but are more prospective in predicting the likelihood of a disease or condition occurring (Caulfield et al., 2009; Gniady, 2009). Even when genetic links are clear, a 1995 report by the National Human Genome Task Force on Genetic Testing highlighted three important points about genetic testing results: 1. There is often a therapeutic gap between testing and treatment (no medical interventions available for many genetic diseases), 2. Negative results do not always rule out the possibility of getting a disease or condition, and 3. The positive presence of a gene does not guarantee that a disease or condition will occur in that individual (Gniady, 2009). These issues raise clinical and ethical concerns regarding how genetic testing is used and whether non-predictive genetic services provide any legitimate benefit to consumers.

My research will focus on one key aspect within the field of genetic testing – genetic tests used to provide consumers with health information related to common disease risk. It is thought that tests for common diseases offer large market potential but little clinical relevance. To understand why, it is important to point out the difference between a diagnostic test and a predictive test. Diagnostic tests are conventionally used in medicine to define something about a patient’s current condition, for example blood counts or imaging tests. Predictive tests provide information about a future condition that may or may not develop. For some tests the predictive nature of the risk is high, but always contains a component of uncertainty about whether a condition will develop, when it may appear, or how severe it will be (Evans et al. 2001). Predictive genetic tests can be further differentiated into (a) predictive tests that evaluate alteration in a single gene for few genes strongly associated with disease onset (e.g. Huntington’s Disease, Tay Sachs, some familial inherited cancers), and (b) susceptibility tests that test for multiple genetic variants associated with low to modest increased risk of developing health conditions (e.g. diabetes, many cancers, cardiovascular conditions) (McBride et al., 2010). It is this latter form of test that I am most interested in, because for these tests many of the risks and opportunities for personalized medicine are uncertain.
1.3. Risks and Benefits of DTC Genetic Tests

A number of stakeholders, including company owners, geneticists, doctors, scientists, ethicists and regulators, have publicly debated the risks and benefits of DTC genetic testing and its potential consequences for consumers and health care systems. Table 1 summarizes the main arguments, each of which is discussed in detail in Appendix A. When identifying the main risks, of particular importance to health care providers is the lack of clinical validity, accuracy and utility of the tests. Of particular importance to this report, is the void of genetics knowledge in some parts of the system and the effect this may have on the potential drain on the medical commons. The principle benefits include enabling individual rights and empowerment, and the educational opportunities.
### Table 1. Risks and benefits of DTC genetic testing

<table>
<thead>
<tr>
<th>Risks</th>
<th>Benefits</th>
</tr>
</thead>
</table>
| **Lack of Validity, Accuracy and Utility**<br>**Analytic Validity** – ability to provide the right answer as to whether a genetic variant is present or absent.<br>There are currently no regulatory measures in place to ensure DTC genetic testing companies use analytically valid methodologies. Even if regulatory measures are put in place, the technologies used, limitations in scientific knowledge and diversity in individual genetic make-up can result in erroneous results.<br>**Clinical Validity** – the genetic variant must correlate with a disease or condition.<br>The clinical validity of DTC genetic tests, especially susceptibility tests, has been seriously questioned in the scientific and health care communities.<br>**Clinical Utility** – the genetic variant must provide information that is helpful to the individual tested.<br>Utility can be interpreted very subjectively, and can range from providing someone peace of mind to informing treatment options. Since utility can be defined differently from one individual to another and may vary from one genetic variant to the next, there are serious difficulties in evaluating the utility of DTC genetic tests.<br>**Misleading Advertising**<br>Since the involvement of a health care provider is not required when purchasing a DTC genetic test, customers often rely on information provided by the company itself to determine if a test is right for them. An investigation of 15 DTC genetic testing companies found that 10 of the 15 engaged in some form of fraudulent, deceptive or otherwise questionable marketing practices. Many companies use deterministic language to exploit pre-existing fears about disease. They also used simplified language that tended to broaden the class of person for whom testing was indicated.<br>**Unintended Health Consequences**<br>Individuals with results indicating low risk of<br>**Individual Rights and Empowerment**<br>It is proposed that individuals should have the right to the information in their own DNA, without the involvement of a health care professional. DTC services have the potential to increase access to genetic testing by removing intermediaries who could serve as a barrier. From this perspective, the goal should be to improve accuracy and transparency of the interpretation of genetic information in order to integrate patients into the management of their own health, rather than to regulate and limit patients’ access to personal health information.<br>**Education**<br>DTC genetic tests have the potential to educate patients and health care providers, increasing awareness of genomic advances and applications, and having a positive impact on the patient-provider relationship. Through genetic testing, consumers can learn more about the health conditions they have, the conditions they may develop, and what they can do to improve their overall health status. Searching for health related information on the Internet is common, and physicians have become accustomed to talking to patients about health information they obtain through various media outlets.<br>**Positive Health Impacts**<br>It is hoped that as patients and health care
Disease may reduce healthy preventative behaviours. Individuals who are given a high risk of disease may experience psychological distress or make risky shortsighted lifestyle choices. Others may choose to undergo unnecessary treatments or procedures that have side-effects and risks. Many of these concerns may be based on worst-case scenarios, but because the benefit of genetic risk information available at this time is questionable, many medical professionals feel it is reasonable to be cautious.

<table>
<thead>
<tr>
<th>Drain on the Medical Commons</th>
<th>Health Care System Savings</th>
</tr>
</thead>
<tbody>
<tr>
<td>The first area of concern is the potential waste of physician time on non-essential service provision. The second area of concern is that genetic testing could lead to a cascade effect in which genetic results lead to further work up that creates anxiety, cost and potential harm.</td>
<td>If individual health outcomes can improve with increased education and knowledge of their health risks, this should translate into savings for the health care system in the long run. Cost savings could come from lower screening costs for low-risk populations and earlier detection and prevention for high-risk populations. As such, genetic risk testing can be seen as aiding the movement towards cost-effective personalized medicine.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Void of Genetics Knowledge in the System</th>
<th>Innovation, Access, Uptake</th>
</tr>
</thead>
<tbody>
<tr>
<td>Studies show that physicians lack the knowledge and confidence about how to counsel or when to refer patients to genetic services. In 2004 the Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS) reported that “insufficient education and training in genetics and genomics has led, and may continue to lead, to inaccurate or delayed disease diagnosis, misguided disease management…and unnecessary costs” (Gnaidy, 2009).</td>
<td>Instead of costing the health care system, innovation from the consumer genetics industry may benefit the medical system at the cost of the consumers who value it. Examples include the development of infrastructure for the storing and analysis of genetic information, and the creation of user-friendly interfaces for exploring genetic results. DTC genetic testing also ensures that new technologies will be accessible to individuals and that uptake is unrestricted.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Privacy and Genetic Discrimination</th>
<th>Privacy and Convenience</th>
</tr>
</thead>
<tbody>
<tr>
<td>In Canada there is no anti-genetic discrimination legislation. Some companies offering DTC genetic testing services are developing biobanks for research purposes, and it is unclear whether proper informed consent is obtained from customers for the research use of their DNA.</td>
<td>Selling tests directly to consumers may be the best way to ensure privacy is maintained, as it enables individuals to control who can access their information by reducing the likelihood that the tests will appear in medical records. DTC sales also provide convenience to consumers, eliminating the hassle of scheduling a doctor’s appointment.</td>
</tr>
</tbody>
</table>
1.4. Current State of Affairs

As is the case with new technologies, the fears and concerns are abundant and the advantages and benefits are still unproven. While DTC genetic testing has the potential to improve the health and well-being of consumers, the public cannot rely on DTC genetic service companies to ensure that customers have the information they need to make informed decisions. There is a growing awareness that the current completely unregulated DTC genetic testing market can put consumers and health care resources at risk (McGuire & Burke, 2008; Gnaidy, 2009; Magnus et al., 2009, Hogarth et al., 2010). The FDA has been pushed to address this issue by the large number of apprehensive stakeholders and the concerning results of a Government Accountability Office (GAO) investigation of several DTC genetic testing companies (see Appendix A, pages 65-66). As mentioned previously, the regulatory landscape for DTC genetic tests has begun to change. Letters sent by the FDA to twenty popular personal genomics companies in June and July of 2010 made it clear that regulating DTC genetic tests is within their mandate, and that the FDA has the authority to require premarket approval to ensure analytical and clinical validity of their products (Shuren, 2010). They have opened the door to discussions with DTC genetic testing companies over how to best regulate the market.

The federal government in Canada has recognized that DTC genetic tests fall in a regulatory gap that may leave the government without the authority to regulate the DTC tests currently on the market (Box 3). Health Canada currently has no official position on how they will proceed, but according to a Health Canada official they are assessing the situation via a working group looking at different aspects of personalized medicine (including regulatory issues related to genetic testing). Representatives from Health Canada were also among many international experts and relevant stakeholders who participated in an event hosted by Genome Canada in Ottawa that explored the issue. In a resulting policy brief the policy options proposed included: (1) control access to genetic tests for more serious conditions, (2) allocate resources to enforce existing laws, (3) enhance information availability, and (4) actively monitor the situation (Ries & Einsiedel, 2010). Regardless, a plan to regulate the industry is far from clear and it will likely take some time before quality in the DTC genetic test market is assured.
As long as customers can purchase DTC genetic tests in an unregulated market many of the concerns that critics raise must be considered. Without a defined national strategy, provincial governments and those working in the health care system are strategically placed and play a central role in ensuring customers have the information they need to make informed decisions. At the same time, they must safeguard the health care system itself, ensuring it is not put at risk by the overutilization of health care resources by those who are asymptomatic and simply curious or ‘covering their bases’.

1.4.1. **Use of DTC Genetic Tests**

Before determining how to deal with DTC genetic tests, it is important to determine to what extent DTC genetic testing is impacting the health care system in BC. This will help determine the degree of intervention required. Because DTC genetic testing companies do not publicize their sales numbers, it is difficult to know how many consumers have purchased DTC genetic tests with any certainty. It is also hard to know...
how many of those customers have discussed such tests with health care practitioners because specific records of DTC genetic test consultations are not kept.

Surveys of potential DTC genetic test consumers in the United States indicate that approximately 60% of the population would use genetic tests to obtain information about their health (Cogent, January 2011; McGuire et al., 2009). In an online survey of users or would-be users of DTC genetic tests, the most cited reasons for testing were curiosity about genetic make-up and to see if a specific disease runs in the family. The most cited reason for not using a test is that they do not think the information would be useful (McGuire et al., 2009). In a study of Canadians’ interest in genetic testing, it was found that obtaining information that can inform decisions and actions related to health was an important factor in willingness to pay for such tests. With that said, approximately 51% would still be willing to pay to test their risk for serious and unpreventable diseases (Ries et al., 2009). This study also found that 70% of respondents were unaware that genetic tests could be purchased directly from private companies. However, the level of awareness could increase with targeted advertising campaigns by DTC genetic testing companies. For example, in a study of select U.S. cities, an advertising campaign for the BRCA1/2 breast cancer genetic test doubled the number of people who have heard of the test (MMWR, 2004).

Estimates of the number of individuals who have purchased DTC genetic tests vary, with numbers ranging from 0.3% (Kolor et al., 2009) to 5.8% (McGuire et al., 2009) of survey respondents (although the latter may be skewed by self-selection of survey participants). According to the Statistics Canada population clock for BC the current population is approximately 4.6 million (http://www.statcan.gc.ca/ig-gi/pop-bc-eng.htm). If just 0.1% of the population purchased a DTC genetic test that would be 4,600 tests. With Canada’s population at 34.6 million (Statistics Canada population clock) and the US population at 312 million (http://www.census.gov/main/www/popclock.html), BC’s population accounts for 1.3% of the combined population. If 4,600 tests represents 1.3% of tests sold, that would mean over 350,000 tests have been sold in both countries. 23andMe, the most popular DTC genetic testing company that receives 78% of the Internet traffic of the top three companies (Wright & Gregory-Jones, 2010), announced in June of 2011 that it had the DNA data of 100,000 people in its database (23andMe, June 15, 2011). If 23andMe in fact has a dominant share of the market, the estimate of
350,000 tests may be high and the 4,600 tests sold in BC would be a worst-case scenario (presuming that the more tests sold, the greater potential risk to health care resources).

According to the literature, anywhere from approximately 30% (Leachman et al., 2011) to 53% (McGuire et al., 2009) of DTC genetic test customers discuss their results with physicians (Appendix A, pages 68-69). Using the estimated 4,600 tests sold in BC as a guide, and assuming that at most 50% discussed results with a health care provider, it is possible to estimate 2,300 physician visits could have been used to discuss DTC genetic tests in a worst-case scenario. Looking at it another way, in 2009 there were 5,282 family physicians in BC (CIHI, 2010). One U.S. study found that 42% of physicians are aware of DTC genetic testing services (n=2218), of those 42% have answered questions about them with a patient (n=932) and 15% have helped a patient interpret results (n=333) (Kolor et al., 2009; Appendix A, pages 68-69). Even considering that a physician can have multiple patients requesting information about DTC genetic tests, there is discordance between the estimated 2,300 physician visits and the 932 physician consults. To narrow this down, a BC Clinical Genomics Network survey of physicians indicated that 24% of respondents have had at least one patient use DTC genetic testing and come to their office and ask them about it. Extrapolating this to all family physicians in BC (acknowledging survey respondents may be self-selected for encounters with genetic tests), that would be 1050 family physicians. So it would appear reasonable to say there could have been 1,000 physician visits regarding DTC genetic tests in BC (likely over the past three years).

1.5. What next?

The possibility that 1,000 doctors visits may have been used to discuss DTC genetic tests in BC, while not indicative of an emergency situation, does indicate the

---

1 The statistic comes from currently unpublished data provided by the BCCGN. Data will be submitted and published at a later date.
start of trend driven by a new and growing industry. Previous experience with DTC advertising in the pharmaceutical industry gives us reason to be cautious. While touted as a tool to open the door to conversation between a patient and their doctor, DTC pharmaceutical advertising has led to more requests for advertised medicines, more prescriptions, lengthened clinical encounters and changes to prescribing practices – despite physician ambivalence about treatment choice (Mintzes et al., 2003; Robinson et al., 2004). So while the system may not currently have a major problem, what it does have is an opportunity. An opportunity to plan for the future of personalized medicine. An opportunity to think about what part genetic testing is going to play in health care. An opportunity to develop a strategy for the management of DTC genetic tests before use grows and unintended consequences are felt. An opportunity to gather information and learn about DTC genetic testing – who uses what and why. And, an opportunity to enhance the skills sets of our health care providers.

The current situation necessitates consideration of how DTC genetic tests for complex disease should be managed and interpreted in the health system once a patient requests information about them. What services should be provided and who should provide them? Should doctors be allowed to continue to make decisions as to when tests results should be incorporated into care management decisions? Is the education of doctors required so they can perform this task more effectively? Should the role of genetic counsellors be expanded so they can manage patient requests? Should the use of health care providers be avoided, instead utilizing online resources to address patient questions and concerns? What principles should inform decision making around these options? It is these questions my research attempted to address. As Caulfield (2011) notes, much of the literature addressing DTC genetic testing policy concentrates on regulatory oversight. Recommendations are made to provide regulation of industry activity and legal response to ethical issues (Hogarth et al., 2008; Magnus et al., 2009; Hogarth, 2010; Caulfield & McGuire, 2012). As such, many jurisdictions are focusing their efforts in this area (Borry et al., 2012; SACGHS, 2010). However, other commentators also recommend education of health care providers as another policy approach to address concerns around DTC genetic testing (Caulfield et al., 2009; Gnaidy, 2009), and some jurisdictions have begun to act in this area (SACGHS, 2011; HGSG, 2012). Recognizing the importance of health care providers in DTC genetic test management, it is this policy approach through which I frame my research.
2. Methodology

Academic journals, grey literature and informal exploratory phone interviews were used to determine the focus of the project as it related to DTC genetic testing. Through this research I identified the lack of physician education and knowledge of genetics as an impediment to genetics-related decision-making. Since family physicians are considered the “first line of defence”, their lack of knowledge poses major risks to the system if and when consumers of DTC genetic services seek advice. As such, my research then centred on how to address the concerns around inappropriate decision making by physicians. This included exploring ways to provide physicians with the skills and resources needed for effective patient management and health system utilization.

I undertook further interviews to determine if and how this should be done in BC. I used a qualitative methodology because I did not know in advance what all of the main issues were going to be. The openness of qualitative methods allowed me to explore the topic and adapt the objectives of the research question to the realities of the BC context throughout the research process. As such, it provided the ability to obtain greater substance, depth and understanding; and allowed the participants to have a greater voice in the research.

I took a pragmatic approach to the research. This paradigm allows for the use of mixed methodology, with tools ultimately selected based on usefulness and workability for the particular research question. There is an emphasis on researcher behaviour; specifically how shared meanings between researcher and participant are formed in the interview process. The goal is to get transferability in data obtained, focusing on context and generalizability, in order apply research findings. This paradigm also acknowledges values play a role in results interpretation.

Two key values informed my analysis throughout this process. I avoided assuming that the integration of genetic tests for complex diseases is either inevitable or desirable. New technologies should be adopted based on their utility and effectiveness,
which has yet to be illustrated. As such, I do not assume that action should be taken to improve the environment for DTC genetic testing to occur. Rather I explore cost-effective ways to create an environment in which health care resources are protected, in particular the prevention of challenging doctors visits, inappropriate specialist referrals and unnecessary follow-up tests. To minimize the costs and commitment, I focused on policy options that contain components already within the health care system. The second value relates to the patient-physician relationship, specifically, I recognized that (1) patients have autonomy and that (2) health care providers are considered experts. This makes for a complex relationship and interesting power dynamics. This relationship will influence the information that will be transmitted to patients and how the management of care moves forward. As such, the goal of my research is not to dictate a particular course of action in regards to DTC genetic testing, but rather to improve the environment in which decision making can occur.

2.1. Interviews

The interviews were semi-structured, consisting of several key questions and prompts as a guide but allowing for follow-up questions and exploration of topics the participants identified as relevant. Most of the formal interviews were face-to-face, which allowed for rapport to develop and easier sharing and exploration of opinions. This also allowed for recording of the interview (with consent). However, due to scheduling or geographic constraints, telephone interviews were also conducted. Informal interviews conducted early in the research or to obtain specific information from specific sources were also conducted by telephone. A total of 26 participants were interviewed.

Interviews were conducted with stakeholders representing different interests within the health system or genetics industry. The interview methodology included purposive sampling (participants selected based on their role in specific organizations identified online) and snowball sampling (participants recommended by other participants). Participants were selected based on their professional involvement in the provision of care related to genetic testing, and their familiarity with DTC genetic test use in the system. The majority of interview participants can be broken down into several often-overlapping categories, in accordance with their role in genetics and health care.
1. Geneticists – medical doctors with a genetics specialty, who primary work through the Medical Genetics Program at BC Women’s Hospital.

2. Genetic Counselors – Master’s degree health professionals specializing in genetics and genetic counseling. Most genetic counselors work in conjunction with geneticists in the Medical Genetics department or other genetic specialty areas such as the Hereditary Cancer Program at the BC Cancer Agency or the Alzheimer’s and Multiple Sclerosis programs at the University of British Columbia (UBC).

3. Family Physicians – members of the Society of General Practitioners.

4. Educators – geneticists, physicians, genetic counselors and educators who are involved in physician education at the undergraduate, medical school, residency and in practice levels. In BC the primary education providers are UBC, the professional Colleges, and the BC Clinical Genomics Network (BCCGN).

5. Academics – specialists in the field of law, ethics, and genomics policy working from universities across the country.

In general participants were a source of province-specific information regarding the current state of genetics in the system, the resources that are required to effectively manage patient requests related to DTC genetic testing, and the role of various health care professionals. However, many other topics were addressed and explored.

2.2. Thematic Analysis of Interviews

Thematic analysis is a method for identifying, analyzing and reporting patterns (themes) in data, and for organizing, describing, and interpreting the data sets (Braun and Clarke, 2006). Typically, as in this research, themes are repeating patterns identified across an entire data set (in this case, multiple interviews). According to Braun and Clarke (2006), thematic analysis has many advantages over other forms of qualitative research. Of particular importance to this work are: it is flexible, accessible to both the researcher and public, can summarize key features of a large body of data and/or offer a ‘thick’ description of the data set, can generate unanticipated insights and can be useful for producing qualitative analyses suited to informing policy development.
Braun and Clarke identify a series of choices that need to be considered and reflected upon by the researcher undertaking a thematic analysis. They are as follows.

What counts as a pattern or theme? Flexibility is required when identifying themes, but consistency within the analysis is essential. I defined a theme to be a topic that was addressed by the majority of the participants and consisted of a series of “sub-themes” or topics that were also addressed by at least several of the participants.

Is the aim to provide a rich description of the entire data set or one particular theme across the data set? I used the thematic analysis to provide an overview of the entire data set.

Was an inductive (“bottom up”) or deductive/theoretical (“top down”) approach used to identifying themes? Since I did not have a theoretical interest and my aim was to identify issues considered relevant to the participants, I used an inductive approach. Themes identified were strongly linked to the data itself and were not fit to a pre-existing coding or analytic framework.

At what level were themes identified: at a semantic/explicit level, or at a latent/interpretive level? The thematic analysis began primarily at the semantic level (the explicit meaning of the data), however as analysis evolved into the development of policy options and evaluation a more interpretive approach developed.

What is the epistemological approach: essentialist/realist or constructionist? Since I analyzed themes at the semantic level a realist approach was taken based on the belief that meanings can be taken from the participant’s descriptions and
experiences (I did not theorize the sociocultural contexts that underlie the accounts provided by the participants)² (Braun & Clarke, 2006, pages 81-86).

I began the thematic analysis process by reviewing transcripts and making descriptive notes of the content. I generated initial codes and applied them to the notes. The initial codes that arose were: system, physician, genetic counselor, patient, and education. While deriving themes from the coded data I divided the system theme into two: system and technology. I then grouped specific points from the notes into under each theme in order to reanalyze and identify sub-themes. These sub-themes described the key elements of each theme and became the foundation of my analysis.

² The research epistemology guides what can be said about the data, and informs how meaning is theorized. With an essentialist/realist approach, a simple, largely unidirectional relationship is assumed between meaning and experience and language. As such, motivations, experience and meaning can be theorized in a straightforward way. In contrast, from a constructionist perspective, meaning and experience are socially produced and reproduced. Therefore, thematic analysis conducted within a constructionist framework seeks to theorize the sociocultural contexts, and structural conditions, that enable the individual accounts that are provided (Braun & Clarke, 2006, p. 85).
3. **Thematic Analysis**

In analyzing interview notes and transcripts, I grouped participant comments into five thematic areas that are relevant to the discussion of DTC genetic testing in the health care system.

1. The Technology – genetic testing and its characteristics, opportunities and challenges.

2. The System – how genetic testing is managed in the health care system in BC and some of the issues that arise.

3. Physicians – what their strengths and weaknesses are in the area of genetics and what they need to be successful in their role.

4. Genetic Counselors – what their strength and weaknesses are and how they can be optimally utilized.

5. Patients – what they want from the system and how they approach genetic testing.

Each theme is discussed in greater detail below.

### 3.1. Technology Theme

It is common in the media and at time in the literature to describe advances in genetic technology as driving a revolution in medicine and health care delivery. In asking participants about where they felt the future of genetics in medicine is going in the next several years, I received a more cautious approach to the rate of change. According to most participants, while they expected continued progress and impact on patient care made in key areas, they were skeptical that genetics would have a large impact on the care of most individuals. As one participant stated:
So I think that in the next two to five years genetic testing in patient care I think will remain, in my view, still very focused to better identify patients with genetic disorders… But I don’t personally feel that all this genomics and genetic sequencing and whole genome sequencing is going to have a big impact on—or maybe I should say it may have an impact but maybe not have great benefit to just, you know, sort of the healthy individual and prediction of risks and, you know, there is a lot of hype about that but I personally don’t see that making a big difference in patient care in the next, you said, three to five years. (#22)

The participants expected that new genetic information would continue to give insight into rare genetic conditions. For example, participants indicated that advances in genetic testing would play a role in prenatal testing and hereditary cancers, providing more accurate diagnosis. These are areas where genetic testing already plays a role in health care, and participants see genetic testing increase in importance. A more novel area of growth that participants identified was the area of pharmacogenomics. This was considered “a large area where there are becoming testable items” (#2) which could be used to prevent adverse drug outcomes. Participants took a much more cautious view of the application of genetic research in the area of susceptibility testing and risk prediction for common diseases, the focus of many DTC genetic testing companies. Here participants expected integration into health care to be slow, giving estimates of 5, 10 or 15 years before access to genetic testing for common disease would be available. As one participant said: “the gatekeepers to offering those sort of services are probably going to make that transition quite slowly because we would want to be very sure that we’re offering something of value and that we’re not just opening Pandora’s box.” (#4)

From a technological standpoint, participants noted that genome-sequencing technology will continue to become faster and cheaper. This was expected to increase how many genetic tests can be performed and how many patients can access and benefit from this technology. In addition several participants expected that the introduction of whole genome sequencing would soon eliminate the need to do tests of specific genes. As one participant mentioned when discussing the recent attainment of
the thousand-dollar genome\textsuperscript{3}: “it (a genome scan) is cheaper, as it is now, than sequencing for the mutations of BRC 1 and 2” (#21).

While participants noted the benefit of cheaper and faster technology, namely improved patient access to testing, they were also quick to note the potential drawbacks of collecting more genetic data from patients. Recent experience in the use of microarray technology and in the Hereditary Cancer Program (which does genetic testing to assess risk for cancer) illustrates this. Participants stated that while test users have found an increase in data obtained can be helpful, it can also be quite frustrating to get results that cannot be interpreted because the technology has outpaced the research. In addition, participants noted that questions regarding how these large amounts of data are to be stored and whether it should be provided to patients have yet to be formally addressed. So while there was a sense among participants that when it comes to the integration of new technology the system needs to “play some catch-up” (#3) because it is “already behind” (#1), there was also the acknowledgement that progress must be made slowly and cautiously. As one participant said:

A large challenge that I see is what happens when it becomes more cost effective to get more data, then just to test those individual 10 genes. And then what do you do with the rest of the data? That I think is a huge question. Because then you’re back down to this ill defined risk. (#2)

Looking at DTC genetic testing technology specifically, participants varied in their perspectives on how the market for this technology may evolve. One participant heard suggestions that the DTC business model is unsustainable and will eventually fail (however, he also notes that providing information over the internet is cheap, so this might be a questionable assertion). Another participant noted that DTC genetic testing companies (23andMe in particular) are transitioning to whole genome sequencing.

\textsuperscript{3} In January of this year a U.S. company announced the release of its genome sequencer capable of sequencing the human genome for $1000, a much-anticipated technological milestone. However, this price only covers the cost of the consumables, not the instrument, labour or overhead. [Oh, G. (2011). Beyond the thousand-dollar human genome. Medill Reports Chicago, January 12, 2012.]
services. In general, the degree of patient uptake of DTC genetic tests, now and in the future, was unknown. Direct experience with DTC genetic technologies varied among participants. Several participants cited encounters with individuals (patients, colleagues, or acquaintances) who have purchased DTC genetic testing services. The Medical Genetics department in particular has noticed the use of these services by their patients; as one participant shared:

And then we have already begun to receive telephone calls, questions about direct to consumer testing and would we help interpret the results. And secondly we’ve had patients come, I can cite an example where there’s an abnormal prenatal ultrasound and the parents have said ‘by the way I’ve had 23andMe screening would that be helpful to you?’.

Most participants felt that there is high public interest, but believe that the majority of Canadians are unlikely to purchase DTC genetic testing services because they are unaccustomed to paying for health services. If there were to be increase in customer demand, there are deep concerns among participants that these customers will end up in their doctor’s office asking questions and requesting services, potentially wasting health care resources.

In summary, many participants acknowledged genetic technology, including DTC genetic testing, might provide opportunities to improve patient health and increase patient autonomy. However, most were quick to point out the challenges associated with this technology: questions regarding the relevance and reliability of genetic tests performed, difficulties in data interpretation and management, by-passing the “checks and balances” (#4) of our system that ensure fair resource utilization, and the lack of guidelines on how health care providers are to incorporate these technologies into patient care. There were also concerns about the “cascade of misinformation” (#15) that may arise as patients receive genetic information without understanding its relevance then pass that information along to family members. In addition there was the fear of the “cascade” of intervention (#22) that may occur once a potentially healthy patient has received a genetic result, which then triggers further tests and interventions that may cause harm. These were some of the issues that participants felt the health care system will need to consider.
3.2. System Theme

When discussing the role of the health care system in managing genetic tests, there was a sense among participants that it was important to strike the right balance. As one participant noted, DTC technology (at its current price point) is used by a tiny market of white, educated, high income, technologically savvy individuals, so “is the policy fix worth it?” – and then notes that the level of concern is “enough to do something” (#6). There was a solid level of concern that DTC genetic testing is part of a growing and larger issue that needs to be addressed, namely how genetic technology is handled in the health care system. There was a keen awareness among participants that resources are limited and need to be managed effectively. However, there was no indication from participants that the lack of resources justifies inaction. The majority of participants felt there were activities that should start now to ensure the system is properly managed into the future. There was also an acknowledgement that every player has a role to ensure this occurs. As one participant stated:

(regulations) got to be governmental, but on the other hand the practice guidelines obviously should come through the medical associations, and then the implementation needs to be in conjunction with the provincial health authorities and the providers. So it is more complex that just who’s responsible. And it’s not that it’s all bad, some of it’s going to offer benefits to people. And then going back to the term recreational genomics, I mean some of it will just be of interest to people – people doing ancestry searches, so I don’t think you can look at it all as a health care issue. (#2)

A common sentiment among participants was that no one seems to be pushing the genetics agenda in the Ministry of Health, or perhaps even acknowledging that such an agenda should exist. Participants questioned why the Ministry has not begun a dialogue with genetics professionals to identify and address the issues. Attempts by these health care providers to initiate this process or propose solutions to delivery of care problems have largely been ignored. It was suggested the reasons for this include a lack of awareness of the issues at hand and their importance now (not the distant future), and the lack of knowledge of genetics that serves as a barrier to engagement. It was also suggested that the Ministry simply does not trust the health professionals managing medical genetics with the budget. This has created a funding arrangement for genetic tests in BC that participants indicate is an endless source of frustration for the
genetic specialists providing patient care (see Areas for Further Research, page 56). It has also created a system that was described as “narrow” and “slow to change”, which raised questions as to whether the system is planning enough or flexible enough to deal with the inevitable changes genetic technology will bring.

On the other hand, the Medical Genetic Program in BC appears to be a key player in identifying and responding to genetic testing issues in the system. As one participant also noted, the Medical Genetics department is “small and close enough that…things get discussed and policies get developed as soon as there is a problem that we’ve seen…a number of times” (#17). Representatives of the Medical Genetics department see the department as having the responsibility to manage these queries and play a role in education. As one participant stated:

So I think that we need to take on responsibility to, within our own department, develop the expertise to … help with those requests. I see that we have a role in educating, first, I think, the physicians of the province…. And I think that I would say somehow figuring out, or to be helpful with education of the public, but I see for us, if I have to use resources available to us that physician education is important. (#3)

What the above quote illustrates is the acknowledgement of participants that physicians have a key role. While genetic specialists have the skills to manage this issue, participants identified that it is actually family physicians that will be the first line of defense in the system and must be prepared to act accordingly. As one participant said, physicians are the “front line in having these options available, and then being confronted with people bringing in their own information, need to understand it at a higher level” (#2). There was general consensus among participants that they currently do not have the knowledge or resources required to do so. Most participants voiced the need for physician education. But as one participant noted, it may be too soon to educate until the province has a system in place to give guidelines (#13).

Developing guidelines for physicians is just one example of some of the system issues that participants stated need to be addressed. On a related note, several participants mentioned they would like to see the system ensure that national or international standards are utilized in the development of guidelines to determine what genetic data is relevant (#10) and avoid the duplication of efforts (#2). Several
participants also requested that the system ensure that there is regional equity (#3) and patient equity in service provision to, for example, limit attempts at queue-jumping (#15) and ensure equal access to care and preventative information (#21). If two patients have identical clinical symptoms but one has privately-obtained genetic test results that may inform care and the other does not, should these cases be managed the same way? (#21). These issues are in addition to questions that arose around the “discourse of rational resource decision making”, namely how to get the best value for money in genetic service provision, who should be in what roles to ensure optimal service provision, and then how to measure success or impact of policies?

3.3. Physician Theme

As previously mentioned, much of the decision making around the response to DTC genetic testing will be the responsibility of the family physician. Participants indicated family physicians are positioned as “gate-keepers” (#16) who play a role in protecting specialist resources. They are the first point of contact with patients, utilized as a source of advice and information. Participants believed many physicians want to maintain this role when dealing with genetic tests, realizing that not all these patients can or should be referred to genetic specialists, especially in “rural communities” (#1). Several participants felt that family physicians have the skills needed to adapt to changes in practice that will arise as genetic research evolves, and pointed out that physicians already manage and counsel patients on issues of risk for common disease. Many reject the “genetic determinism” (#10) mindset that may lead some to believe that genetic information is fundamentally different than any other piece of data that a doctor may be asked about or test for.

However, there are still some reasons for concern that participants felt may limit the ability of physicians to provide the optimal level of care in this area. Historically genetic tests have been managed differently than other tests in that doctors are unable to order some tests and have limited experience with many of them. As one participant noted: “I think they’re a little bit nervous around it because they don’t do it a lot. I also think there are also certain genetic tests that they haven’t been able to order, that they aren’t able to order, and so it puts it automatically in a different class.” (#1). This is in
part because, as one participant described, some genetic tests involve unique ethical challenges – confidentiality, impact on family members, insurance eligibility, and potential for discrimination (#16) – issues which are relevant for DTC genetic tests as well. In addition, participants indicated many physicians have very little knowledge of genetics – both the basic biology and technological components. According to the research of one group interviewed, physicians themselves rate their knowledge quite poorly (#1b). A number of factors contribute to this; several participants noted physician have a lack of education (knowledge gap) and a lack of time (competing demands).

Finally, there was the concern that this lack of knowledge has translated into a lack of awareness about the relevance and clinical value of genetics to patient care. As one participant stated: “They don’t know what they don’t know” (#11).

Another important point that many participants raised in relation to the role of physicians in managing genetics related patient care, was the importance of family histories. A thorough family history came up again and again as a cheaper and more informative way to identify disease risk and inform patient care than the genetic testing offered by DTC services. As one participant said:

You know, if you do enough of them (tests) you will find mutations in breast cancer genes that are predictive; you will find some things but whether or not you will change the management of that patient anyway and whether or not you wouldn’t have known about that increased risk based on her history—I’m kind of a believer that, you know, your best tool is still a detailed family history. (#22)

Another participant indicated that in order to act in their role as gatekeepers, general practitioners could improve their family history skills. Once a disease that runs in the family is identified, the standard system of referral to a genetic specialist can be used. This is a system that is believed to work well. According to this participant, what needs to be “tweaked” is the better identification of at risk patients (#16). A current barrier this that was identified by participants was the lack of time physicians have to take a thorough family history (#5, #22).

In summary, most participants felt that family physicians should be responsible for managing patient queries related to DTC genetic tests, but that they would need additional education, skills and/or resources to do it with confidence. This will be
explored in greater detail in the *policy options* section, but suggestions included basic education, guidelines, electronic tools, compensation system (billing codes), and access to genetic specialists.

### 3.4. Genetic Counselor Theme

Despite support for increased education and resources for physicians, participants also spoke of the need for more support and integration of genetic specialists. In particular, many identified genetic counselors as an excellent and potentially underutilized resource. Currently genetic counselors work in teams, typically under the supervision of clinical geneticists, and are therefore concentrated in areas where traditional medical genetics applies (prenatal testing, rare conditions). As genetic testing becomes more relevant to other disease areas (hereditary cancers, Alzheimer’s Disease, Multiple Sclerosis), their role in the system is expanding. However, as several participants pointed out, despite their Master’s education and a rigorous certification system for practice, without a regulatory college and provincial recognition of their skills they are limited in scope of practice and have restricted employment opportunities. Another barrier that several participants mentioned was that some doctors and most members of the public do not know what genetic counselors do and what their capabilities are.

Participants envisioned that genetic counselors could be used to counsel patients and help them interpret DTC genetic test results, educate patients and physicians, evaluate new developments in genetic technologies and tests, and serve as a bridge between groups of health professionals (roles they already perform). In interviews genetic counselors were said to be knowledgeable genetics educators capable of providing high quality care in a variety of clinical situations. Their principle characteristics, as described by one participant, are that they know how to look up the information needed, and then have the ability to communicate that information (#11). Several participants also noted their time is “cheaper” to the system than physicians (although they spend much more time with patients). One participant summarized genetic counselors as follows:
They are at a level with all of our other graduate students in terms of genetic knowledge. So they come with a very good knowledge base. They are also trained in counselling. So they have a unique perspective in being able to deal with stressful situations, grief, how to deal with new information, what should be on the table if they are ever asked prior to direct-to-consumer genetic testing, and they are educators. (#3)

Despite their skills, there were also concerns that genetic counselors are not as familiar with common diseases (#16) (of which much DTC genetic testing would focus), and their non-directive approach would be unsuitable for this area (#18 P1). Some participants believed that further training in this area might be required. While there was some disagreement among participants as to the validity of this concern, it was generally recognized that risk management of common diseases should be the purview of family physicians. As one participant put it: “what’s extremely important…is the move in the direction of testing for chronic disease, and that’s the bailiwick of family physicians” (#3). Another concern among participants regarding the use of genetic counselors (and other genetic specialist) in patient care for DTC genetic tests, was whether specialist resources would be wasted on clinically irrelevant cases should referrals increase. As one participated stated: “I don’t know that that is really appropriate to waste the medical geneticist when they have a…waiting list for other medically appropriate things” (#18 P2).

3.5. Patient Theme

One participant noted that DTC genetic testing has the potential provide consumers the opportunity to learn about their health risks and gives people autonomy (#16). The primary concern among participants regarding DTC genetic testing was that after testing customers “run to their doctors” and there is no benefit to the system in this, only cost (#14). While participants indicated the DTC movement could have positive impact on care, more often it was found to have a negative influence on the doctor-patient relationship (#18 P1, 2, 3). As such, it is important to consider what patients may want from testing and expect from the system. Participants identified several characteristics of patients and DTC consumers, namely:

- Patients want control of their genetic information, but they are altruistic with it.
Patients want help in the form of interpretation, direction and advice.

Patient interest in genetic testing decreases when they are informed of its limitations.

Patients want the government to pay for, regulate and vet the tests they use.

3.6. Summary

Within the technology theme, participants discussed how advances in genetics and genomics bring faster and cheaper technologies that expand the potential for changes to heath care delivery and patient management. These technologies have also opened the door for DTC genetic testing services. Without guidelines, these DTC tests may inappropriately impact patient management and the utilization of health care resources. Participants indicated (in the patient theme) they had concerns that consumers of these tests will end up in their physicians’ office, and that these tests will have a negative impact on the doctor-patient relationship. This leads to costs in the system, such as inappropriate referrals and tests, which have little benefit. In order to integrate the benefits of technological advances without experiencing the negative consequences, the research participants pointed out (in the system theme) the need for engagement from the BC Ministry of Health. Ideally the Ministry should take a leadership role in collaborating with genetic specialists and physicians in planning for the future. As participants indicated, this includes establishing guidelines and ensuring the system functions in a balanced, rational and equitable way. Here again, a key point was the need to support family physicians through the development of guidelines and education.

As the technology expands and interest in DTC genetic testing grows, the majority of participants noted it is family physicians that will be the front line responders. Family physicians have many of the basic skills required to address general patient concerns arising from DTC genetic tests. In the area of susceptibility testing for common diseases in particular, they see the counseling of patients as their role to take. Unfortunately, as many of the participants highlighted, at the moment many physicians lack the knowledge and the confidence to step into this role comfortably. Many also lack the time and resources to counsel patients effectively. Genetic counselors, on the other
hand, were said to have the knowledge and skills sets required to counsel patients on the implications of DTC genetic tests. However, they were considered a specialist resource that will not replace the central role of the family physician in the health care system. In managing DTC genetic tests, none of the participants talked about changing the current health system model: a patient sees their family physician, the physician addresses issues within their scope, and the physician refers to a specialist if issues fall outside their scope.

And yet, as this research reveals and existing literature supports, the view that physicians should be the first line of defense, despite their lack of knowledge in relation to DTC genetic testing, may be an impediment to optimal decision making (McGuire et al., 2009; Gulcher & Stefansson, 2010). The resulting question for advancing policy in this field is: how do physicians get the education and/or resources they need to identify and fulfill the obligations within their scope of practice, while ensuring resources are used most effectively?

Participants provided a number of suggestions to address this core challenge, namely: basic education, guidelines, electronic tools, compensation system, and access to genetic specialists; each described below in the Policy Options section.
4. Policy Options

I will begin my exploration of policy options emerging from the research findings. Each option arose out of discussions of the themes and addressed the challenges of the technology, the opportunities and gaps in the system, the responsibilities of physicians and the role of genetic specialists in supporting them. The policy options themselves are inter-related and can be combined to obtain synergies. While they are presented as stand alone options, participants were hesitant to take a “one or the other” approach to change. All options centre on the need to provide physicians with skills and resources required to manage the changing landscape of genetic testing.

4.1. Status Quo

The status quo policy option is a description of the current situation with no additional action taken. None of the participants interviewed were content with status quo management of genetic testing services and physician education, but none believed large, wholesale change was necessary. It was felt that improvements could be made to existing systems using minimal resources and with more open communication between the parties involved.

There are a number of basic characteristics and components of the status quo. It begins with physician education in genetics. In medical school, using UBC as a guide, participants indicated students receive approximately three weeks of genetics education, plus any organ specific information that may have a relevant genetics component scattered throughout the curriculum. According to one participant (#7), the trend is to have horizontal integration of topics embedded into coursework, which makes curriculum evaluation difficult, and the curriculum is still targeted to passing exams, which means there needs to be an exam component on an area for it to be a priority. Lastly, participants acknowledged that the medical school curriculum is “crowded” (#7) because there are so many topics that students need to learn. As one participant asked: what do
you cut out to educate on genetics (#10)? From this perspective, it is not difficult to understand why students in medical school learn only basic genetics information.

Once they are practicing, physicians have the opportunity to select from a range of Continuing Professional Development (CPD), also known as Continuing Medical Education (CME), course offerings. As on participant notes: “we can tackle the medical students now… but there is probably another 8,000… practicing physicians… so I think we have to—and we’ve started—to… educate them through… CME initiatives.” (#22). The CPD program through UBC, which provides most CPD for family physicians in the province, has identified, through needs assessment, there is a need for more partnerships with the Medical Genetics program (#5). Genetics-related CPD currently addresses the effects of genetic information on patients (e.g. impact of genetic testing on insurance coverage) and keeping doctors up to date (e.g. alerting them to new tests). The most well recognized source of genetics-specific education among participants is through the BC Clinical Genomics Network (BCCGN). The BCCGN, established to connect clinicians to genomic resources with the goal to improve understanding of disease, has adopted an education component to its activities. Education is done via website, outreach, workshops, conferences, factsheets, newsletters, and research awards; and some of the programs offer CME credits (#1). However, several participants noted that participation in CPD is voluntary, and there is a concern that many physicians consider genetics education to fall into the “not a perceived need” category (#5).

Less formally, the Medical Genetics department at BC Women’s Hospital provides a telephone consultation service to provide information about patient care and whether a referral is appropriate. Known as the “P&P line” for “phones and postals” (a remnant of the days of letter writing), through it staff receive calls from health care professionals for genetics-related questions. This helps ensure medical genetics services are utilized appropriately. The department also disseminates information to the physicians of BC when required, and has begun to explore expanding their educational conferences beyond their department as a way to “get everyone up to speed” on how genomics can be applied (#11).

There is little collaboration between genetics specialists and the Ministry regarding the development and dissemination of guidelines to physicians. Several
participants were concerned that genetics-related information was not disseminated effectively to all physicians that may benefit from it. Similarly, while there are websites that provide genetics-related information (Medical Genetics, BCCGN), in the medical community there is limited awareness of these sites and what they can be used for. The same can also be said for the use of genetic specialists as a source of information. While many physicians are comfortable with collaborating with genetic specialists in more common areas of practice (prenatal testing, rare conditions, hereditary cancers), as mentioned previously, several participants suggested physicians might think it is inappropriate to “waste” genetic specialists time on other issues such as DTC genetic testing and chronic disease. This is especially true given the belief of some participants (and perhaps physicians in general) that “the wait lists right now are ridiculous” to see a genetic specialist (#1). Finally, even if a physician wanted to invest the time to improve their knowledge base and spent time addressing genetics-related issues with patients, there is little incentive to do so. Since DTC genetic tests are not insured services under BC’s Medical Service Plan (MSP), physicians are unable to bill for the time spent counselling patients in this area (#18 P2).

4.2. Basic Education

There was general agreement among participants that the education and training physicians receive in genetics is insufficient to effectively manage patients in this area, now and in the future. As such, increasing basic education in genetics for physicians was commonly recommended. The desired characteristics of this education are summarized in Box 4.

As previously mentioned, in their needs assessment and strategic planning process the UBC CPD department has identified disease areas where knowledge in genetics is low but physician interest is high (which was described as the education “sweet spot”), and has recognized the need to partner with Medical Genetics (#5). In addition, as the only genetics-specific educator in BC, BCCGN is looking to expand its online CME offerings (#1). The BCCGN is respected, liked by users, and collects information via physician surveys. When it comes to training in medical school, participants were torn as to whether it would be advantageous to increase the amount of
time spent on genetics specifically. One participant pointed out the dilemma that educators face in that they must decide what should be cut out of the curriculum in order to make room for increased genetics education (#10). And even if a change in curriculum did occur, there were questions as to how helpful it would be (#10), especially considering the rapidly evolving nature of the subject. While participants promoted the concept of improving genetics education, there were questions regarding how this education should be structured and what should be incorporated. As one participant stated:

I’m very into education so I really value that and I think that is going to be a big help for any system like this where people are trying to deal with new bits of information. So I think that education starting at the, you know, medical school undergrad level and working through the residencies and people in practice are looking to professional development as we call it—those are all going to be areas that need to be, you know, targeted, but I think it is going to need to be a careful balance to make sure that we don’t jump the gun and try to put all this information in that might not be quite relevant yet. Like I think there is a lot of chomping at the bit when maybe there is not such a clear need in as many people’s views and so we might in a sense go too fast too far and damage the possibility for making effective headway in the future when the need is greater. I think that a careful needs assessment and a careful watch of what is actually happening so that it’s all very relevant is going to be really important because this is sort of an area that people get a bit tired of if there is too much and it is not relevant. (#17)

Box 4. Characteristics of good education.

Participants indicated there are several key attributes of successful education programs. Programs must:

1. Be multi-modal – need variety. People learn in different ways.
2. Involve group learning – peer review processes and a community of practice.
3. Be relevant – need to see impact on patients. In genetics doctors respond better to a focus on disease or risk assessment (not “genetics”).
4.3. Guidelines

The creation of guidelines for physicians was considered a key element in both physician education and management of their practice. It was even suggested that until a system of developing guidelines is in place, directing resources to physician education might be premature (#13). While many participants may feel it is never too soon to begin basic education, they would also agree that the creation and maintenance of up to date guidelines is important. Topics that participants felt should be addressed in guidelines include: identification of actionable tests, risk stratification of tests available, when to refer, and legal liabilities associated with genetic testing. In BC, guidelines are developed by the Guidelines and Protocols Advisory Committee (GPAC) – a joint committee of the BC Medical Association (BCMA) and the Ministry of Health. However, guidelines for genetics-related topics are often created and disseminated by Medical Genetics. As mentioned previously, the effectiveness of this system has been questioned. It was suggested that a provincial system of guideline development is required, involving all relevant stakeholders in genetics and health care.

Several participants recommended linking to national or international standards to avoid duplicating efforts. For example, in the US, the Evaluation of Genomic Application in Practice and Prevention (EGAPP) and the Agency for Healthcare Research and Quality (AHRQ) release clinical practice guidelines related to genetics. It was also mentioned that Washington State has a genetic counselor in place to research developments in genetic technology and feed that information back into the system (#13). Regardless of how the relevant guidelines are developed, a key element is how they are then disseminated to physicians who must apply them. Participants indicated that there is no one right way to do this. Physicians have personal preferences for how they receive information, whether it is online resources, e-mail notifications, mail-outs, journals, or point-of-care tools. What is key is that there is a central place where the guidelines “live”, so after they have been notified physicians know where to retrieve the information when needed (#18 P1, 2, 3).
4.4. Electronic Tools

A major component of educational efforts and guideline development is the use of online resources and tools. This may include websites that serve as a home for genetics related information in a jurisdiction, or point-of-care tools that provide a searchable, evidence-based synthesis of best practice for patient care that can be used during consults. Participants indicated that online resources are big “bang for the buck” (#5), but current efforts exist in silos related to specialities (#6) and it is difficult to identify which are up-to-date, evidence based and user-friendly.

In BC a popular website available to all citizens for health related information is HealthLink BC. It currently provides information in some areas of genetics, such as tests for select conditions, genetic influences on weight, ethical and legal issues, some basic biology, and where testing can be performed. In addition the Medical Genetics program has a website that provides a range of information to patients and physicians. And information can also be found on websites run by genetics focused organizations, like the BCCGN. However, none of these sites are specifically designed to offer point-of-care information custom tailored for physician-patient consults. Participants indicated that information for physicians must address clinical questions in a succinct and practical way – one or two key sentences that address the patient question (#6, #18). In Canada, Dr. June Carroll has led the way into the research and development of genetics resources for family physicians. The GenetiKit project developed tools and resources to “enhance the delivery of genetic services by family physicians”, and more educational projects are underway. In the US, the National Coalition for Health Professional Education in Genetics (NCHPEG) has developed GeneFacts, “a point-of-care, decision support system for non geneticists” that consists of Fact Sheets related to specific “Mendelian

4 Samples of research include:
disorders, common diseases and pharmacogenomic associations”. In addition the National Center for Biotechnology Information (NCBI) hosts GeneReviews, “expert-authored, peer-reviewed, current disease descriptions that apply genetic testing to the diagnosis, management, and genetic counselling of patients and families with specific inherited conditions”. The NCBI also recently launched a Genetic Testing Registry (GTR) that “provides a central location for voluntary submission of genetic test information by providers”. While none of these sites has focused on developing content specifically related to DTC genetic tests, it is possible that as they evolve these sites could address these broader issues.

4.5. Compensation System

As mentioned previously, there is little incentive for family physicians to invest the time and effort into educating themselves, and their patients, about DTC genetic tests when they are unable to bill for providing this service. Even if the parties involved agree that these conversations should be happening in a family physician’s office first, it has been made clear to some of participants I spoke to that there is “zero money for billing codes” (#18 P2). In concept, many participants felt that counselling patients about genetic tests for common diseases is a natural extension of conversations that relate to risk management and heredity. As such, it is not unreasonable for physicians to be compensated for their time (i.e. have an appropriate billing code). Considering that participants felt that questions about DTC genetic tests will often be brought up in relation to other health issues being discussed, and that physicians will find it difficult to avoid addressing the issue entirely, it is most likely that a degree of creative compliance to billing rules will take place in the meantime.

4.6. Access to Genetic Specialists

Participants acknowledged that, considering the infrequent number of requests for genetic testing services and interpretation (prenatal testing aside) (#18 P1, 2, 3) and the competing demands on their practice (#16), it may be unreasonable to expect family physicians to become (or want to become) more well-versed in genetics. While
participants did feel it would be wise for them to know the pros and cons of such testing, it was also suggested that it might be more important for them to know what resource to contact if and when these questions arise. A common suggestion was to provide family physicians with a hotline to access a genetic specialist, specifically a genetic counsellor, for informal consultations and questions about DTC genetic tests. One participant stated:

…that might be the simplest way of dealing with all of this, is that, just make sure the family doctors know that they have help, that there is somebody that can help navigate how to handle this, and it might just be even a phone call with a genetic counsellor to say ‘this is what my patient brought in, my interpretation of this is x, y, z, this is how I would talk about it with my patient, is there anything I’m not thinking of, is there anything else that I should bring up, is this something that you think I should refer over to you’ – just to have that phone call might be the simplest way of approaching it. (#4)

As mentioned previously, genetic counsellors are skilled at researching and communicating genetic information. They are utilized by the Medical Genetics Program telephone consultation service (P&P line) to deliver information to health care providers regarding patient care and whether a referral is appropriate. Members of the department have also discussed the possibility of developing a pilot project in which they create a genetic counsellor role with two functions: 1. To research the clinical implications of genetic abnormalities found in test results, and 2. To answer physician and patient calls about DTC genetic test results (#3).

A similar service that is offered in BC is Vancouver Coastal Health’s Rapid Access to Consultative Expertise (RACE) hotline which provides “guidance and advice regarding assessment, management and treatment of patients” in speciality areas such as cardiology and psychiatry. The phone line is currently focussed on addressing urgent issues; an estimated 70-80% of physician calls are responded to within 10 minutes (#23). What it provides is a specialist contact, a collegial relationship and an educational component to practical advice (#23). The program encourages physicians to then use the experience to apply for “Linking Learning to Practice” CME credits. While this service was considered more than is needed by some participants (#18 P1, 2, 3), many felt expanding this service to include the genetics specialty would be a good way to let general practitioners know that “help is out there”.

39
4.7. Summary

The policy options presented above are a collection of ideas and suggestions that arose from the research relating to the delivery of genetics-related primary care. They all address the core issue previously described: providing physicians the skills and tools they need to assist patients with questions and concerns about DTC genetic test results. The next step undertaken was to identify the most effective and practical ways in which to do this.
5. Analysis

Due to the large number and wide variety of policy options that arose from the research, I chose to undertake a two-step analysis. The first step utilized a criteria matrix (described below) to broadly compare the five general categories of options (described above). The criteria of administrative feasibility, political feasibility, equity, efficiency and effectiveness were used to determine (a) if any of the policy options had any barriers to implementation that made them unsuitable to pursue in BC at this time, or (b) if any of the options were unlikely to meet their objective. After identifying two policy options that appeared most suitable for implementation, I undertook further research and analysis of them through interviews and website searches. This allowed me to present a more detailed description of the options and do more thorough exploration of their suitability for implementation. I then used the framework of a multiple account benefit-cost analysis to compare the two options to the status quo. This framework highlighted the trade-offs between the two policy options and status quo, and allowed me to more directly assess the cost and benefits on those most impacted by DTC genetic testing: the Ministry of Health, family physicians and patients.

5.1. Preliminary Assessment of Policy Options: Criteria Matrix

A criteria matrix is a decision making tool used to evaluate and choose from several possible alternatives based on specific factors (criteria). I first identified and defined key criteria that could be used to assess the ability of each option to achieve the goals and objectives of the research problem. This was an essentially subjective process, informed by the data obtained in the research. The benefit of a criteria matrix is that it is transparent, and thus could be employed to clearly indicate the considerations utilized in my decision making process. Table 2 describes in detail the initial criteria used to select the most feasible and appropriate policy options. Table 3 summarizes the
results of the preliminary analysis. Red text indicates potential barriers to implementation and drawbacks of pursing the policy option.

Table 2. Description of criteria used in the evaluation matrix.

<table>
<thead>
<tr>
<th>Criteria</th>
<th>Description</th>
<th>Aim is to identify administrative barriers to implementation of the policy option.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Administrative Feasibility:</td>
<td>Is it feasible to implement?</td>
<td></td>
</tr>
<tr>
<td>Political Feasibility:</td>
<td>Will there be broad stakeholder support?</td>
<td>Aim is to identify options that may be met with resistance.</td>
</tr>
<tr>
<td>Equity:</td>
<td>Will the knowledge be required of all physicians?</td>
<td>Aim is to identify options that will be utilized by all physicians, and in turn whether the benefits of the option will be available to all patients.</td>
</tr>
<tr>
<td>Efficiency:</td>
<td>Will resources be easily available to physicians in a cost effective manner?</td>
<td>Aim is to identify options where the resource is in a centralized location (easy to find), available to physicians when needed, and provided in a cost effective manner.</td>
</tr>
<tr>
<td>Effectiveness (of clinical encounter):</td>
<td>Will it lead to better decision-making and protect health care resources?</td>
<td>Aim is to identify options that improve patient care and resource utilization during patient visits.</td>
</tr>
<tr>
<td>Effectiveness (system-wide):</td>
<td>Does user information feed back to decision makers in the health care system?</td>
<td>Aim is to identify options that provide information that can be used for future health resource planning.</td>
</tr>
<tr>
<td>Criteria</td>
<td>Basic Education</td>
<td>Guidelines</td>
</tr>
<tr>
<td>----------------------------------------------</td>
<td>---------------------------------------------------------------------------------</td>
<td>----------------------------------------------------------------------------</td>
</tr>
<tr>
<td><strong>Administrative Feasibility:</strong></td>
<td>No: medical schools slow to change.</td>
<td>No: leadership required from Ministry hard to get.</td>
</tr>
<tr>
<td><em>Is it feasible to implement?</em></td>
<td>Yes: CPD designed to grow as needed (determined via evaluations &amp; assessments).</td>
<td>Yes: participation from various stakeholders is attainable.</td>
</tr>
<tr>
<td><strong>Political Feasibility:</strong></td>
<td>Unlikely: medical school curriculum change will draw out competing interests.</td>
<td>Unlikely: Ministry support challenging (must be seen as a priority first).</td>
</tr>
<tr>
<td><em>Will there be broad stakeholder support?</em></td>
<td>Likely: CPD expansion supported.</td>
<td>Likely: genetics community recognizes need.</td>
</tr>
<tr>
<td><em>Will the knowledge be required of all physicians?</em></td>
<td>Yes: CPD voluntary.</td>
<td>Yes: recommended to all.</td>
</tr>
<tr>
<td><strong>Efficiency:</strong></td>
<td>Yes: but knowledge will only be available when needed if educators provide long term support/resources to physicians. Costs were not identified as a barrier.</td>
<td>Yes: guideline repository accessible (exists online). Guideline development can be costly, cost effectiveness would depend on the expected benefits.</td>
</tr>
<tr>
<td><em>Will resources be easily available to physicians in a cost effective manner?</em></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Effectiveness (of clinical encounter):</strong></td>
<td>Yes.</td>
<td>Yes.</td>
</tr>
<tr>
<td><em>Will it lead to better decision-making and protect health care resources?</em></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Effectiveness (system-wide):</strong></td>
<td>No: Evaluations are performed but the data does not feed back to decision makers in the system.</td>
<td>No: information not collected on doctor use (unless additional procedures/studies are in place).</td>
</tr>
<tr>
<td><em>Does user information feed back to decision makers in the health system?</em></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
5.1.1. Summary of the Preliminary Assessment

Based on the criteria selected, the creation of electronic tools and access to genetic specialists are the best policy options to pursue at this time. They both build on systems already in place, making implementation less difficult and costly. There is little indication that there would be any resistance from stakeholders, and both provide information to decision makers that can be used in future planning. Both options offer resources through clear centralized locations that physicians can go to when needed, making it easier to achieve the ultimate aim of better decision making. However, both options would be used by physicians on purely a voluntary basis, potentially making the knowledge acquisition unequally distributed. While this may lead to inequity of care for the patient, the trade-off is that only physicians who need the information would have to invest time and resources to obtain it. Since DTC genetic testing is still uncommon, this is a reasonable trade-off.

Providing basic education via CPD is also a good option to pursue. As an option in my analysis, its main drawback was that the information obtained does not feed directly back to decision makers in the health care system in a manner that will inform resource utilization. Since the population of physicians that will commit to genetics-related CPD is still small, most physicians would still be left without resources and unrepresented in data collection. Genetics related CPD will grow through the efforts of groups like UBC CPD and the BCCGN, and as a policy option my best recommendation is that they continue with their efforts. The UBC CPD program has systems in place to identify knowledge gaps and collaborate to fill them. Organizations like the BCCGN can leverage their resources in the scientific community to provide educational services. Beyond further promotion and support for these efforts, I have little to add to them as policy options.

The development of guidelines is an important part of the health care system’s genetics strategy. This option has several barriers; including questions regarding costs and the lack of a system to feed information back to decision makers. However, the key element here is leadership, which, based on my research, appears to be lacking. The Ministry needs to initiate a dialogue with key stakeholders and start planning for the future. It needs to ensure that genetic technologies and research are used – when
appropriate – to save health care resources, not waste them. I believe that these discussions should take place immediately because they should be the origin of all system efforts, however, it is beyond the scope of this project to address how that should occur or who in particular should lead it.

The most difficult policy options to implement would be the improvement of genetics education in medical school and the creation of a compensation system (billing codes that would relate specifically to DTC genetic testing and discussions of genetic risk for conditions where genetic tests are not currently insured). Medical schools have been slow to change the genetics curriculum, and any attempt to change the curriculum will likely spur a flurry of demands for competing interests. The compensation system is not politically feasible, and in and of itself would not provide physicians the knowledge and resources needed to make better decisions. Despite these potential barriers, all policy options can act synergistically and are not mutually exclusive. Although not selected for analysis in this project, they are all potential areas for future work.

5.2. Further Development of Leading Options

5.2.1. **Electronic Tools: HealthLink BC**

A web-based resource should serve as a central location for succinct evidence-based clinical guidelines for physicians. Participants indicated that HealthLink BC, while it is not geared toward physicians specifically, is respected and can serve both patients and physicians. As one participant noted:

HealthLink is, I think, a potentially credible location for a lot of stuff…. And they need to be the source of the trusted links. But I think that is the obvious place for it to reside because it is so much easier if you can just say to people, “Call 811 or go to….”. (#18 P2).

HealthLink BC consists of several different components. There is the public-facing HealthLink BC website and 8-1-1 phone line where “you can learn about health topics, check your symptoms, or find health services and resources near you”. There is HealthLink BC Health Files, “easy-to-understand fact sheets on a range of public health and safety topics” which are geared towards health care providers and that comes with a
HealthLink BC also has Clinical Decision Support Tools, which is an internal service for practitioners. Most recently, the HealthLink BC operations team has been hired by the BCMA to develop CHARD: Community Health Access Resource Database, a “web-based directory that provides BC healthcare providers with accurate, timely and relevant information to assist in making appropriate referrals” (also a registered service).

The Ministry wants HealthLink BC to be the main provincial resource for health information, so HealthLink BC directors are able to invest resources into improving and adding to the site (#24). When a new topic is suggested, it gets added to a queue and staff do due diligence to ensure it is a worthwhile project. They assess what the item is, how it relates to the Ministry, Health Authority and service delivery priorities, the ease of development, and whether there is well-established evidence. If the information obtained from the external vendor who manages the site is inappropriate for the BC context, they review the material, work with stakeholders to reformulate it, and then work with the vendor to relaunch the new content. The review cycle is every 1-3 years for web content (Health Files are reviewed more often), so the site is designed to provide general knowledge, not provide an in depth level of detail. In addition, user statistics from the website and postal codes from the HealthLink BC 8-1-1 phone line are collected to monitor use.

The biggest expense in this option is the time that would need to be committed to determining what the content should be and what to link to. This will likely require input from genetic specialists in conjunction with family physicians, or someone with the expertise in the needs of both fields. For information on DTC genetic testing, a specific HealthLink page could be developed that briefly describes the pros and cons of testing and what to look out for. It could even specifically state, “There are no clinical benefits to personal genome testing at this time”, the message many participants felt should get across. Additional information targeted specifically to physicians and other health care providers could also be developed and presented through Health Files.

However, even the best site is of little use if no one knows it is there. Physicians would need to be notified, likely through Health Files, that the site has added content related to genetic testing. Since it is a familiar site, it is more likely physicians will
remember it and integrate it into their practice. The key here is that physicians must know the site exists and it must be user friendly. Barriers to this may include the fact that a segment of practicing physicians do not use the Internet as a part of their practice and are not registered subscribers to Health Files.

So what could be the expected benefits from this effort? Physicians would have a trusted source for genetic information that they could use to educate themselves and their patients. Ideally this would help physicians counsel their patients to make better health care decisions and use health care resources more appropriately. This would hopefully improve patient satisfaction with the experience of talking to their family physician about DTC genetic testing. Guidelines and content can be added as they are developed. Links to other sites, such as the Medical Genetics website, GenetiKit or GeneFacts, could provide physician specific resources, tools and information. A website would also allow decision-makers to track usage of the site and specific links and content. This information could help them determine whether there is interest in the content and monitor how it changes over time.

5.2.2. **Access to Genetic Specialists: P&P line**

Consultative services with specialists are an important resource for family physicians, who are required to know a large breadth of information but cannot be expected to know everything in depth. While popular, in the short term the RACE hotline is not the right fit for the types of queries likely to result from encounters with patients with DTC genetic test results. The sense of urgency, as some participants mentioned, is simply not there. Expansion of the services provided by the program is driven by physician requests, and genetics has not come up (#23). However, the idea of a “warm line” has been discussed and this may be more suitable for genetics consultations. If the RACE steering committee decides to head this direction in the longer term, this may be a cost effective way to broaden the scope of exposure to genetic specialists. The current system, managed by Providence Health, runs for $80 a month and consists of a couple phone lines where physicians can leave messages for select specialists and a phone tree. A flat rate is then billed to MSP, $60 by specialists and $40 by general practitioners.

In the short term I recommend better promotion of the Medical Genetics Program telephone service, the P&P line. As mentioned previously, they see themselves as
having a role in the education of physicians. The service is already in place, the program has been proactive in adapting the service to what is needed, and a segment of family physicians use it. However, some participants indicated it might be that a select group of physicians connect and refer to genetic specialists regularly, while a number of physicians do not. It is possible that some family physicians are not familiar with the genetic services available to them, some may not be not sure how genetics plays a role in patient care, or for some genetic services may not be relevant to their practice. As such, I think the key is to get the message out that Medical Genetics is more than prenatal and can assist physician with any aspect of genetics. This would let the program know if lack of awareness is a main barrier to consultation regarding issues like DTC genetic testing. Like web-based services, consultative services are only useful if users know they are available.

Unfortunately there is no one easy way to do this. The RACE line promotes itself through departments of family practice, the BC Medical Journal ( BCMJ ), and practice support groups, but evaluation indicates that awareness has been driven primarily through word of mouth. Interestingly, in an evaluation of the billing codes used by the RACE line, marketing is one key implementation-related improvement opportunity for billing code use. The evaluation gave two recommendations: identify best avenues to reach general practitioners to market the phone fee initiatives, and to clearly articulate fee program objectives ( MNP LLP, 2011 ). The P&P line is currently promoted by word of mouth and is listed on the Red Book Online (based on the BC 2-1-1 resource database) and provides “information about community, social, and government services in the Metro Vancouver, Fraser Valley and Squamish-Lillooet Regional Districts”. Most of the services listed in the Red Book Online are provided by non-profit, community-based or government organizations that provide a direct service to the public.

A common sentiment shared by participants in the area of education and awareness was that multiple approaches are required to get information out to physicians. Options include notices in the BCMJ, submissions to groups/organizations related to family practice, and utilization of various social media sites ( BCMJ blog/Facebook page, BCMA e-news, UBC CPD Facebook page, etc.). The hope would be that these initial efforts would stimulate word of mouth recommendations. The main expense to the Medical Genetics Program would be the staff time invested in developing
the message it would like to deliver, and the strategy for how to deliver it. This would have to be done within the existing Medical Genetics budget constraints. When it comes to fielding the calls, staff are paid on salary. As such, the department would have to monitor call volume and if it were to increase substantially they would need to put forward a business case for an increase in staff.

So what could be the expected benefits from this effort? For family physicians, consultative services through the telephone service would be easy to use and would have a strong education component. Again, it is hoped that physicians would pass this information on to their patients, resulting in better health care decisions and use of health care resources. Information can be custom tailored to the question and case the physician is presented with. This also begins a broader conversation between family physicians and genetic specialists, and would improve the quality of referrals received by Medical Genetics.

5.3. Evaluation of Leading Options: Multiple Account Benefit-Cost Analysis

The purpose of a benefit-cost analysis is to identify and compare the advantages and disadvantages of alternative policies from the point of view of society as a whole (Shaffer, 2010). It is intended to take into account the impacts on all those (with standing) who are affected. According to Shaffer, the main drawback of a traditional benefit-cost analysis is that it does not take into consideration the distributional effects of policy options, choosing instead to aggregates values (measured in dollars) into an overall bottom line which is used to identify a single best option. A multiple account benefit-cost analysis, on the other hand, does not assume all values and preferences can or should be measured in dollar terms. It also recognizes that it is not necessary or beneficial in all cases to aggregate values into a bottom line summary of net benefit. The role, instead, is to focus and inform (not resolve) debates.

The multiple account benefit-cost analysis framework enables the identification and assessment of the different types of costs and benefits arising from policy alternatives. As Shaffer describes, the framework consists of a set of evaluation accounts, with money or other indicators used to assess the relative value of
significance for the affected parties. The measures used may be dollar estimates or other physical or qualitative indicators. In my analysis the accounts used include:

(1) Government Account (also known as Taxpayer Account) – this account evaluates costs and benefits of the program from the perspective of the Ministry of Health,

(2) User Account – this account evaluates costs and benefits of the program from the perspective of the physicians who will access the program and use the information to help patients in their practice, and

(3) Social Account – this is typically a “catch-all” account for perspectives not captured in other accounts. In this case it was used to evaluate costs and benefits of the program from the perspective of patients.

Due to the lack of monetary valuation in my analysis, I relied on qualitative descriptions as measures. The result is a matrix summary to describe the advantages and disadvantages of the different alternatives being investigated. The goal was to inform in a clear and consistent way the implications and relative merits of the different options, and the nature of the trade-offs entailed. An important aspect to keep in mind is that multiple account benefit-cost analysis looks at marginal costs and benefits – those costs and benefits associated with the program above and beyond what would otherwise have been spent. The policy option evaluation using the multiple account benefit-cost framework is presented in Table 4.
**Table 4. Policy option evaluation: multiple account benefit-cost analysis.**

<table>
<thead>
<tr>
<th>Status Quo</th>
<th>Electronic tool: HealthLink BC</th>
<th>Access to specialists: P&amp;P Line</th>
</tr>
</thead>
<tbody>
<tr>
<td>Government Account (Ministry of Health)</td>
<td>No increase in current costs for services offered. No benefits from education and better-managed health system utilization.</td>
<td>Cost of time to develop content with stakeholders. Cost of launching and maintaining web page, links &amp; fact sheets (HealthLink budget). (Assumes physicians would not bill for time above what is billed for the patient visit) Benefit from education and better-managed health system utilization. Benefit of data obtained from site analytics on future decision-making.</td>
</tr>
<tr>
<td>User Account (family physicians)</td>
<td>No increase in current costs to the practice. No benefits from education.</td>
<td>Cost in time spent with patient researching and addressing the question. (Assumes no information would have been provided otherwise). Benefit of access to guidelines and tools for better decision-making and patient care. Benefit of having a tool to use with patients during the visit.</td>
</tr>
<tr>
<td>Social Account (patients)</td>
<td>No increase in current cost in time with physician. No benefits from education or improved care.</td>
<td>Costs in more time with physician. Benefits from education (through physician and independently) and improved care.</td>
</tr>
</tbody>
</table>
5.3.1. **Summary of the Leading Options**

The analysis of the status quo highlights the consequences of inaction. While there will be no increase in the costs associated with running the programs as they are now, there will also be no benefit from education and better decision-making. Without this benefit, there is the potential to incur greater indirect cost from inappropriate resource management (in the form of increased difficult patient visits, inappropriate specialist referrals, and unnecessary follow-up tests).

In comparing the two policy options, the government has a choice between investing in HealthLink BC and the stakeholder engagement it will require, or the Medical Genetics program and the increase in staff time it will require. The resulting benefits that all accounts will obtain will be either be broad and general (in the case of HealthLink BC) or targeted and specific (in the case of the P&P line). The government will either receive general information on website use from the analytics, or detailed information on specific physician requests from phone calls. Physicians will either get general information via HealthLink BC that any patient can also be directed to, or custom-tailored information via the P&P line that can be shared with the patient in question. Patients will then benefit either through a shared resource with their physician (HealthLink BC) or directly from their physician’s education (P&P line).

5.4. **Recommendation**

Each policy option evaluated has its advantages. An advantage of HealthLink BC is that it has several components targeting different groups that work synergistically and are well known. In particular, while the aim is to provide physicians with a resource, HealthLink BC simultaneously offers information that is accessible to patients. Patient education is discussed in the areas for further research (page 54) and was a common suggestion among participants. The promotion of the Medical Genetics Program P&P line, as described, would target physicians specifically. The Program does receive calls from the public, and it may choose to expand on that service. However, I do not recommend a public promotion campaign for the service at this time. This was not suggested in my research, this would detract from the family physician being the primary
point of care, and I would suspect that there might be unintended consequences (such as high volumes of inappropriate calls).

An advantage of the telephone service is that it may be more simple and efficient to manage, and can respond in detail to any types of genetics questions. The genetic counsellors managing the requests keep up to date with the technologies and guidelines as a part of their job, and would only need to research specific questions when they are posed. A website can only address an issue generally, content would need to be added to address new issues, and it needs to be maintained regardless of whether someone is using it or not.

In the short term, I recommend that HealthLink BC collaborate with the Medical Genetics department, genetics educators, family physicians and other relevant stakeholders to discuss expanding the genetics related content and provide specific advice in regard to DTC genetic testing. Not only would it provide information to both physicians and patients (hopefully before a DTC genetic test is purchased), but it would also begin the process of stakeholder engagement in genetics policy planning. In addition, HealthLink BC can serve as a platform to promote the P&P line through CHARD, essentially allowing both policy options to proceed. In the medium term it will be important to analyze the data obtained from the site analytics and the Medical Genetics department regarding what content is being used and what issues are making their way into the health care system. This will help identify priorities and provide evidence to the Ministry for further planning, if needed. In the long term, the hope is that the system can capitalize on the much-needed dialogue between the Ministry and groups involved in genetics services. This could lead to broader changes in the way the system is managed.

This policy choice is feasible to implement. The main barrier is that, as an up-and-coming policy issue, there is currently little pressure to address the issue and take leadership of it. It is my hope that decision makers in government and health care recognize the opportunity that preparing for DTC genetic testing can provide.
6. **Areas for further research**

6.1. **The Public’s Role**

As mentioned previously, many participants brought-up the need for public education around DTC genetic testing. While increasing health care system capacity and providing resources to physicians will serve to educate patients indirectly, as one participant noted: “Health care is the most expensive way to educate”. The recommendations in this report do not replace public education on genomics and DTC genetic testing.

However, public education was not the only focus. Some participants felt that public discourse around broader issues is required. One area of concern was the social and ethics impacts of the use of genetic technologies. Another area of interest was a discourse on values of the health care system, and the idea that the public needs to make hard choices about what kind of services the health care system should and should not provide.

6.2. **Personalised Medicine in BC**

The other area that needs to be explored is how DTC genetic testing fits in to the concept of personalized medicine. On the surface, DTC genetic testing is a logical part of personalized care, and the system needs to address issues around how this data is going to be stored, utilized and monitored. Decision makers must also determine how physicians are going to be trained to manage this type of medicine.

The Personalized Medicine Initiative in BC has taken a different approach to this issue. Instead of trying to fit the technology available into the health system and patient management, it is trying to figure out what clinical problems practitioners encounter.
where a personalized approach may be helpful, and then identifying the technology that can be used to solve the problem. They also have taken the approach that personalized medicine should be utilized in conjunction with a social determinants of health approach to patient health and well-being. With the genomics assets we have in BC, we are positioned to potentially lead the way in this approach to health care.

6.3. The Role of the Federal Government

Most participants wanted to ensure that potential customers had the opportunity to explore the issues around DTC genetic testing before they purchased the test. Many individuals purchase these tests as curiosities and do not discuss them with a health care provider prior to purchase. One way to address this is restrict the types of tests being sold through regulation, as described in Box 3. But there is always a question of how effective these regulations will be in the age of the Internet. There are other ways the Federal government can aid the provinces and the public. Areas that have been discussed include requiring a health professional to order the test, national guidelines, and public education campaigns. It may be useful to connect with Health Canada to discuss how they are going to address this issue and potentially aid the provinces.

6.4. The Role of Other Health Care Professionals

The problem addressed in this capstone is narrow and specific in order to allow for an in-depth analysis, and focuses on the perspective of family physicians and genetic specialists. It should not be overlooked that questions regarding DTC genetic testing will be asked in other areas of health care. For example, nurses are often a source of information for patients and their families in hospitals, nursing homes and other settings, and are therefore well positioned to field questions about DTC genetic testing (Schutte, 2006). In addition, the argument has been made that in allowing nurses to take family histories and undertake genetic testing can reduce referrals to specialist genetic services by up to 75% - a type of “pre-genetics triage” (Ford, 2011). A basic knowledge of genetics will be advantageous for all health care professionals who interact with the public. The level of education required may be debateable but the National Coalition for
Health Professional Education in Genetics (NCHPEG) core competencies are a good place to start.

6.5. Dysfunctional System?

In the discussion of the System theme, I mentioned that the “funding arrangement for genetic tests in BC is an endless source of frustration for the genetic specialists providing patient care”. Concerns about the way genetic testing services are divided and managed are a key finding of my research. Although it is outside the scope of this report, the Ministry of Health may want to further investigate claims that the way out-of-country testing services are provided are not in the interest of patient care, and that genetic testing services provided under the PHSA are not transparent (which impedes system planning capacity).
7. Conclusion

The arrival and steady growth of DTC genetic testing in the health care landscape should make health care providers and decision makers pause to take note. This new technology fits within the broader movement of personalized medicine, which is touted as a prescription for more effective patient care, so it comes with a degree of support. The issue is, as with many new technologies, that the risks and unintended consequences that result may increase costs to the health care system without significant benefit. This capstone explores the current landscape of DTC genetic testing in BC. In particular, it investigates the role physician education in genetics can play to ensure decisions are made in the best interest of both the patient and the health care system. It analyses how the improvement of skills and resources will help physicians to provide effective care without needlessly utilizing health care resources, such as specialist services and follow-up tests. This capstone also proposes how this can be done cost-effectively and to the benefit of the government, health care providers and patients. Perhaps just as important, this capstone illustrates how DTC genetic testing fits within the broader context of genetic testing, and highlights the issues and opportunities that exist in BC that should be addressed. It notes the importance of leadership, collaboration and information sharing in driving and managing change in the health care system. Lastly, this capstone provides a different perspective from which to approach DTC genetic testing policy, offering a complement or alternative to regulatory efforts. These are insights that may also be important to other jurisdictions that are beginning to explore these issues and want to take a proactive approach to the management of new genetic technologies.
References


Caulfield, T. (2011). Direct-to-consumer testing: If consumers are not anxious, why are policymakers? Human Genetics, 130, 23.


Ries, N. M. (2010). *Analysis of privacy policies and practices of direct-to-consumer genetic testing companies: Private sector databanks and privacy protection norms*. Funded by the Office of the Privacy Commissioner of Canada Health Law Institute, University of Alberta.


Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS). (2010). Direct-to-consumer genetic testing.


Appendices
Appendix A.

Risks and Benefits of DTC Genetic Testing

*Risks of DTC Genetic Testing*

*Lack of Validity, Accuracy and Utility*

For a genetic test to benefit an individual the test must have three characteristics (Hogarth et al., 2008):

1. Analytic Validity – the laboratory performing the test must the right answer as to whether a genetic variant is present or absent. Analytic validity is a function of methodology employed, quality assurance systems in place, and training of staff.

Analytic validity is a fundamental concern because it can result in customers receiving the wrong information regarding their risk for a particular disease or condition. A 1999 study found 36 of 245 molecular genetics labs scored lower than 70% on a quality control scale, indicating the need for improved personnel qualifications and laboratory practice standards (Andrews, 2001). It can be argued that this is a regulatory issue, and that proper oversight and certification of genetic testing laboratories and staff should provide consumer protection. Some commentators have argued that market forces offer sufficient protection, since consumers can take legal action against any company that acts negligently (Gnaidy, 2009). However, in addition to traditional barriers to consumer complaints (Hogarth et al., 2008), it is difficult to see how a consumer is supposed to know whether their genetic test results are accurate, especially when the “proof” – the diseases or conditions tested for – are not expected to present themselves for decades into the future. Even if regulatory measures are put in place to control what is probably the most common source of error, sample handling, it is important to note that the technologies used may still be a source of error. Even small error rates, when magnified in an entire genome, can result in misclassified variants for any individual consumer (Hunter et al., 2008). In addition to differences in laboratory procedures, limitations in scientific knowledge and diversity in individual genetic make-up can result
tests that are false positives (a person does not have the disease variant but the test suggests they do), false negatives (a person has the disease variant but the test suggests they do not), or uninformative (a person has a previously unidentified mutation) (Williams-Jones & Ozdemir, 2008).

2. Clinical Validity – the genetic variant must correlate with a disease or condition. It is evaluated through the sensitivity, specificity and positive predictive value of the test (Wade & Wilfond, 2006). This is usually based upon the rapidly evolving scientific literature that compares the genetic profiles of populations with or without the condition of interest.

The clinical validity of DTC genetic tests, especially susceptibility tests, has been seriously questioned in the scientific and health care communities. There are concerns that the genetic variants used to correlate with disease (called single nucleotide polymorphisms, or SNPs) are inaccurate (Leachman et al., 2011; Ransohoff & Khoury, 2010). Data shows that the variants currently associated with disease only explain a small portion of overall disease risk, with variants typically associated with an odds ratio of 1.5 or lower. For example, a variant associated with coronary artery disease (the leading cause of death in men and women) increased the risk of acquiring the disease from 1% to 1.6% (Magnus et al., 2009). Also, risk predictions may change as new variants are found. Furthermore, variants currently identified have mostly been studied on European populations and may not be extrapolated to other ethnicities. In addition genetic predictions from DTC genetic tests do not take environmental factors into account when calculating risk. Lastly, a number of variations between firms occur because they each use different algorithms based on different variants to calculate risk (CMAJ, 2010; Janssens et al., 2008). They also use different criteria for integrating variants into their calculations, and in how they define the population from which risks are based (Ng et al., 2009).

These concerns have practical implications. Between 2009 and 2010 the US Government Accountability Office (GAO) investigated four DTC genetic testing companies to assess their advertising methods and validity of their services. Five test subjects sent two samples to each company, one with their factual profile and one with a fictitious profile containing inaccurate information related to age, race or ethnicity, and
medical history. In the analysis of disease risk for 15 selected conditions, they found different companies often provided different disease risks for identical DNA – each of the five donors had contradictory results for between 9 and 12 of the 15 diseases. In addition, four of the five subjects received risk predictions that conflicted with diagnosed medical conditions or family history. When confronted, the companies pointed out that tests are not meant to be diagnostic and acknowledged that using different variants would lead to different results. Of course, each company claimed that their own company’s tests were better than their competitors (Kutz, 2010).

3. Clinical Utility – the genetic variant must provide information that is helpful to the individual being tested. Utility can be interpreted very subjectively, and can range from providing someone peace of mind to informing treatment options.

Many things can detract from a test’s clinical utility: a lack of population-based evidence, questions about test reliability, or a lack of clarity of how a patient’s management should be changed as a result. But since utility is defined differently from one individual to another and may vary from one genetic variant to the next, there are serious difficulties in evaluating the utility of DTC genetic tests (Foster et al., 2009). The GAO investigation effectively illustrated the concerns that some detractors have with regard to the clinical utility of DTC genetic tests. All of the companies provided the test subjects generally accepted health information related to the conditions tested (including symptoms, treatments, and methods of prevention). All subjects received the same information, regardless of the risk profile they received. Three companies provided follow-up consultations over the phone, but company representatives provided little guidance beyond what was contained in the test reports. Two companies also conceded that the subject’s doctors would probably not know what to do with the results (Kutz, 2010). This is interesting, considering companies often encourage customers to speak with a health care practitioner regarding the results. This begs the question at the heart of clinical utility: if a patient is found to be at risk for a disease, what can be done about it (Hunter et al., 2008)?

Misleading Advertising

Since the involvement of a health care provider is not required when purchasing a DTC genetic test, customers often rely on information provided by the company itself
to determine if a test is right for them. As such, misleading or fraudulent advertising has serious consequences to the consumer. The GAO’s investigation into misleading advertising found several reasons for concern. In the initial assessment of the four companies sent subject samples, they found that when race was listed as Asian or African American the subject did not receive complete test results. Many of the disease predictions apply to only those of European ancestry, but this was never disclosed prior to purchase (even though a lengthy consent form is required). A further investigation of 15 DTC genetic testing companies found that 10 of the 15 companies engaged in some form of fraudulent, deceptive or otherwise questionable marketing practices. Some companies made misleading representations about the reliability of the tests and the ability of health care practitioners to use the results to treat patients. Others made claims that DNA could be used to create supplements that could cure disease; or that tests could predict which sports children would excel in. And some companies condoned the potentially illegal practice of testing someone’s DNA without their consent (Kutz, 2010).

In general, companies offer an unbalanced view of the utility of their service. One study found that while 90% of websites present information on the benefits of DTC genetic testing, only 55% give any information on the limitations (and information on limitations is harder to find on the website) (Leachman, 2011). Another study found only four of 13 websites mentioned any kind of risk associated with their tests, and only one mentioned risks other than privacy (such as the predictive nature of the test, impact on family, informed consent, or emotional response) (Berg & Fryer-Edwards, 2008). Many companies use deterministic language to exploit pre-existing fears about disease, focussing on the causal role of genetics to the exclusion of other social or environmental factors. And since the segment of the population that would benefit from genetic testing is often small, they also used simplified language that tended to broaden the class of person for whom testing was indicated (Gollust et al., 2002; Williams-Jones & Ozdemir, 2008; Berg & Fryer-Edwards, 2008; Goddard et al., 2009). It is questionable whether customers know what they are paying for. And since individuals may approach web-based information with scepticism, companies use strategies to build trust and gain credibility in the eyes of the perspective consumer (Einsiedel & Geransar, 2009).

**Unintended Health Consequences**
Concerns have been raised that DTC genetic tests may cause more harm than good, in that they may lead to unintended consequences. Individuals with results indicating low risk of disease may reduce healthy preventative behaviours (Caulfield et al., 2009). Individuals who are given a high risk of disease may experience psychological distress that extends to family members (McBride et al., 2010), or make risky shortsighted lifestyle choices (Gnaidy, 2009). Others may choose to undergo unnecessary treatments or procedures that have side-effects and risks. And there are indications that simply “labelling” subjects may have negative consequences, like an increase in lost days from work (Ransohoff & Khoury, 2010). Yet others are concerned that DTC genetic testing may exacerbate existing health disparities, as those who cannot afford or understand DTC advertising will fail to benefit from this new information (Kontos & Viswanath, 2011).

Many of these concerns may be based on worst-case scenarios, as other studies indicate there is minimal negative impact (Ransohoff & Khoury, 2010). Yet medical professions feel there is good reason to be hesitant. Many have seen the emotional effects that genetic information has had on their patients. They understand how difficult it is for patients to grasp probabilities. They experienced situations of misunderstandings. They know the cost of unnecessary tests and procedures. And because the benefit of genetic risk information available at this time is questionable, many medical professionals feel it is reasonable to be cautious (Evans & Green, 2009).

**Drain on the Medical Commons**

“Canadian patients who send their data to a US firm end up in Canadian health clinics” (CMAJ, 2010). This quote from an article in the Canadian Medical Association Journal illustrates the concern that many commentators have regarding private DTC genetic testing services utilizing the limited resources of a public health care system. The first area of concern is the potential waste of physician time on non-essential service provision. Surveys of potential and current DTC genetic test users have found that 61% of people believe that physicians have the professional obligation to help individuals to interpret DTC genetic testing results, 78% who considered using these services would ask their physician for help interpreting the results (McGuire et al., 2009), and when asked to imagine that a genetic test told them they be at risk for cancer, 84% said they
would speak to their doctor as a next step (Cogent, March 2011). Moving from hypothetical scenarios, one study found that 53% of people who have used a DTC genetic test discussed their results with their physician (McGuire et al., 2009), while other studies indicate that up to 30% of people do (Leachman et al., 2011). A survey of US health practitioners found of the 42% of physicians who were aware of DTC genetic tests that 42% had at least one patient who asked questions about having such a test and 15% had at least one patient who brought DTC genetic test results to them for discussion in the past year (Kolor et al., 2009).

This same survey found that of those practitioners who discussed test results with their patients, 75% indicated the results changed some aspect of the patient’s care, such as screening tests offered, medications or dosages prescribed, lifestyle changes recommended, frequency of follow-up appointments, or diagnosis made. This leads us to the second area of concern, that genetic test results could lead to a cascade effect in which genetic results lead to further work up that creates anxiety, cost and potential harm (McGuire & Burke, 2008). The cascade effect is often initiated by physician anxiety, fear of litigation and poor training in the implications of tests, but is facilitated by the attitude that ‘more is better’ and the patient’s assumption that the only reason why tests aren’t done is financial (Deyo, 2002). However not all follow up tests are wasteful. Individuals found to be at high risk for a disease could benefit from more regular screening tests. But caution should still be used. One study found that for the 22 potential screening tests available for the genetic conditions tested, 13 had no clinical benefit to asymptomatic individuals (Bloss et al., 2011).

**Void of Genetics Knowledge in the System**

Physicians are often the front line of defence in the health care system. They ensure that patients get the tests and services they require, while protecting the limited resources available in the system. Their ability to manage this effectively is dependent on their ability to gauge the most appropriate course of action. This ability, however, is undermined when physicians do not have the required knowledge of the issue at hand. This is often the situation they face when confronted with DTC genetic test results. Studies show that physicians lack the knowledge and confidence about how to counsel or when to refer patients to genetic services. In one study 90% of physicians acted
against evidence-based guidelines and inappropriately referred the patient to BRCA1/2 genetic counselling (for breast cancer) when it was not warranted; another found only 30% of clinicians correctly identified appropriate scenarios for referring patients to genetic counselling (McBride et al., 2010). In 2004 the Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS) reported that “insufficient education and training in genetics and genomics has led, and may continue to lead, to inaccurate or delayed disease diagnosis, misguided disease management…and unnecessary costs” (Gnaidy, 2009).

**Privacy and Genetic Discrimination Concerns**

Despite the passing of the Genetic Information Nondiscrimination Act (GINA) in the US in 2008, a study found that 79% of physicians were concerned that genetic information may be misused by insurance companies, government or employers; and 71% of Americans have concerns about who can access their genetic information and how that information will be stored (Cogent, January 2011). Some companies offering DTC genetic testing services are developing biobanks for research purposes, and it is unclear whether proper informed consent is obtained from customers for the research use of their DNA (Hogarth et al., 2008). While most companies have policies to protect their customer’s genetic information, it is unclear how adequate they are in practice (McBride et al., 2010). The Office of the Privacy Commissioner of Canada funded a report that undertook and analysis of the privacy policies of DTC genetic testing companies. It found wide variation in the comprehensiveness and quality of privacy policies among companies; and identified deficiencies in specific privacy policies, policy assertions that were simply advertising claims, and internal inconsistencies in policies (Ries, 2010).

In Canada there is no anti-genetic discrimination legislation equivalent to GINA, raising the importance of privacy and discrimination concerns (Cassels, 2011). Article 3 of the Canadian Human Rights Act prohibits discrimination based on disability (real or perceived ailments), which may provide a legal basis for anti-genetic discrimination claims (Lemmens et al., 2010). While there is little pressure to adopt genetic discrimination legislation, there was a private members bill (C-508) introduced in April 2010 to amend the Human Rights Act to specify genetic characteristics as a prohibited
ground for discrimination (Bonter, 2011). The bill did not become law (http://openparliament.ca/bills/40-3/C-508/).

**Benefits of DTC Genetic Testing**

**Individual Rights and Empowerment**

One argument put forward by proponents of DTC genetic testing is that individuals should have the right to the information in their own DNA, without the involvement of a health care professional, because it is a fundamental part of them (Magnus et al., 2009). It is legitimate to question whether the information obtained from a DTC genetic test can be considered “a part of you” when it was obtained using an algorithm that has not been validated and is proprietary to the company. But it is also legitimate to question whether the involvement of a health care professional is necessary. DTC services have the potential to increase access to genetic testing by removing intermediaries who could serve as a barrier. And in a culture where more information is typically views as “better”, restricting access to one’s own health information would appear inappropriate (Leachman et al., 2011; Gulcher & Stefansson, 2010). From this perspective, the goal should be to improve accuracy and transparency of the interpretation of genetic information in order to integrate patients into the management of their own health, rather than to regulate and limit patients’ access to personal health information (Foster et al., 2009).

**Education**

A large component of patient empowerment is patient education. DTC genetic tests have the potential to educate patients and health care providers, increasing awareness of genomic advances and applications and having a positive impact on the patient-provider relationship (Leachman et al., 2011). Through genetic testing, consumers can learn more about the health conditions they have, the conditions they may develop, and what they can do to improve their overall health status. Searching for health related information on the Internet is common, and physicians have become accustomed to talking to patients about health information they obtain through various
media outlets (McGuire & Burke, 2008). In a study of people who considered or sought DTC genetic testing, researchers found those who opted to get tested had the same balanced view of the genetic contribution to disease as those who chose not to, but were more likely to believe that learning about genetics is important (McBride et al., 2009).

**Positive Health Impacts**

It is hoped that as patients and health care providers become more knowledgeable in the factors that influence health, more can be done to prevent, screen and treat. As such, identifying individuals at high risk of certain disorders can lead to informed decision-making and better health outcomes. Identifying individuals who are low risk can give them a sense of relief and spare them from unnecessary tests and interventions (Berg & Fryer-Edwards, 2008). Early studies suggest that consumers of DTC genetic tests do not act rashly on the basis of what they learn. And many of the initial concerns that recipients of DTC genetic information will experience psychological distress appear to be unfounded, as most studies indicate that learning of an increased genetic risk of a disease does not lead to adverse emotional impact (Leachman et al., 2011; Bloss et al., 2011). One study even found that when patients shared DTC genetic test results with their physician it was associated with lower fat intake and increased exercise activity (Bloss et al., 2011). The same study also found an increase in the intention of undergoing more routine screening tests in the future, but it is unknown if this will translate into action. Whether it is possible to achieve long term changes in behaviour based on risk information has not been determined (Mihaescu et al., 2009). In addition, it is unknown whether self-selection plays a role in existing studies – those who may have been psychologically harmed may have declined to participate (Bloss et al., 2011).

**Health Care System Savings**

If individual health outcomes can improve with increased education and knowledge of their health risks, this should translate into savings for the health care system in the long run. If the health system was to use genetic screening of patients more proactively, this could have significant impacts. For example, based on the current genetic variants identified for prostate cancer, the upper 15<sup>th</sup> percentile of the population who average of 2.1-fold risk of prostate cancer can be identified (an increase from a
17% lifetime risk to a 34% lifetime risk of contracting the disease in white individuals). Along with the 5% of the population who have two-fold risk based on family history, this identifies a high risk population comprising of 20% of the population and that would account for 40% of the total prostate cancer burden. Increased screening of this group could lead to a decrease in prostate cancer morbidity, mortality and cost in later stage care (Gulcher & Stefansson, 2010).

Proponents of DTC genetic testing, or genetic risk testing for common diseases in general, do not see these tests as determinative or diagnostic. They propose that these tests should be used together with other conventional risk factors to help make more accurate and informed estimates of overall risk. The field of medicine already uses risk-based guidelines for certain conditions, where patients who exceed certain risk thresholds receive earlier screening or preventative strategies. The integration of genetic risk into the decision making process would appear advantageous. Rather than leading to an overutilization of health care resources, genetic testing could lead to a more appropriate use of resources. Cost savings could come from lower screening costs for low-risk populations and earlier detection and prevention for high-risk populations. As such, genetic risk testing can be seen as aiding the movement towards cost-effective personalized medicine (Gulcher & Stefansson, 2010).

**Innovation, Access, and Uptake**

The movement towards personalized medicine will require innovation. Instead of costing the health care system, innovation from the consumer genetics industry may benefit the medical system at the cost of the consumers who value it. The development of infrastructure for the storing and analysis of genetic information, and the creation of user-friendly interfaces for exploring genetic results, are example of innovations that the DTC genetic testing industry has developed which could be utilized by the health care sector to manage and communicate patient data (Leachman et al., 2011). As more information is gained about the connection between genes, their disorders and potential therapies, the clinical utility of genetic tests is expected to increase (Gnaidy, 2009). So it is not a stretch to see how the expansion of DTC genetic testing can provide useful information to the field of medicine, which can then apply new risk factors as they are discovered and validated (Gulcher & Stefansson, 2010).
However, the information obtained from new technologies will be limited if access to those technologies is restricted. To date, genetic tests have not been integrated as a standard part of primary care, despite the fact that genetic tests have been developed for over 1,100 diseases. A number of reasons have been cited for this, including (a) limited evidence about how genetic test results will lead to improved patient health, (b) complexity of interpretation of genetic results for common diseases, (c) lack of physician knowledge about genetics, and (d) potential for genetic discrimination (Berg & Fryer-Edwards, 2008). While some of these concerns have been addressed (e.g. through the GINA legislation), it is physicians that still seem to be a barrier to access. Physicians may be uncomfortable providing access to genetic tests or unaware of the existence of some tests, inhibiting their desire or ability to offer genetic testing to interested individuals (Berg & Fryer-Edwards, 2008). Selling genetic tests directly to consumers overcomes these barriers. And with the increasing ubiquity of the Internet and the relatively low cost of some DTC genetic test services ($99), there could be an increase in the uptake of this new technology among segments of the population that typically could not take advantage of innovations (McBride et al., 2010). It has also been recognized that the medical profession actively resists certain new technologies, such as electronic medical records, in order to preserve professional autonomy (Evans & Green, 2009; Walter & Lopez, 2008). As such, there may be resistance to DTC genetic testing driven by a perceived threat to the autonomy of medical professionals.

Privacy and Convenience

While opponents to DTC genetic testing propose that protection of privacy among DTC service companies is a concern, proponents argue that selling tests directly to consumers is the best way to ensure privacy is maintained. DTC sales enable individuals to control who can access their information by reducing the likelihood that the tests will appear in medical records, thereby reducing access for employers and insurance companies (Berg & Fryer-Edwards, 2008). This is of particular importance since genetic discrimination legislation does not cover life or extended care insurance (McBride et al., 2010). DTC sales also provide convenience to consumers, eliminating the need and hassle of scheduling a doctor’s appointment (Berg & Fryer-Edwards, 2008).