SUSTAINABLE FUNDING FOR GENETIC TESTING IN THE PROVINCE OF BRITISH COLUMBIA

by

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ABSTRACT

Funding strategies for providing genetic testing in the province of British Columbia are examined, including alternate forms of publicly funding the B.C. Molecular Genetics Laboratory (MGL), privatization of the MGL, and outsourcing of testing to private laboratories. Demand scenarios are constructed and costs of supplying genetic testing estimated for different scenarios. Public funding of MGL is more efficient at providing genetic tests in all scenarios except the most optimistic private funding scenario.

Funding MGL through public funds using at least a partial per-test funding strategy provides B.C. with reasonably priced genetic testing for the near future, an internal source of genetic test provision and development, and capacity to expand and provide genetic testing to clients inside and outside B.C. All of these contribute to the benefit of the people and province of British Columbia while operating in a fiscally and medically prudent manner.

EXECUTIVE SUMMARY

Project Overview & Aim

British Columbia's Molecular Genetics Laboratory (MGL) develops and provides diagnostic testing for congenital anomalies and genetic diseases in foetuses, children, and adults. MGL has a history of constrained funding and a consequent provincial outsourcing of samples resulting in reduced benefits and higher costs to the province.

The aim of this project is threefold: to establish a balanced five-year funding strategy for MGL; to reduce out-of-province expenses; and to present and justify a strategy to sustainably fund genetic testing in B.C. for the future.

MGL Funding and Demand

MGL is currently funded via hospital-level global funding, capped by strategic plan at 2002/3 levels. With no adjustment to funding since 2003, available funds for laboratory supplies and reagents have been severely reduced by increasing labour costs while test demand has risen steadily. MGL has succeeded in meeting rising demand despite reduced supply funding via operational efficiencies and repurposing of available funds, but MGL's Director feels the limit of operational efficiencies has been reached.

With future demand expected to rise, constant global funding is unsustainable over the next five years. I explore the costs and scalability of different funding models including per-test funding, privatization of MGL, and outsourcing genetic testing to private laboratories.

Conclusions

The province is best served by MGL through a per-test funding strategy in which MGL's budget is at least partially proportional to the number and cost of tests performed. Per-test funding for MGL provides cheaper access to services than either privatizing MGL or outsourcing genetic testing to private companies.

Instituting per-test funding for MGL allows expansion of services through addition of new tests. New tests proposed by Director of the MGL Dr. Brett Casey and approved in June 2008 will provide a 45% demand increase and increase referred-in revenue. By developing in-house versions of tests often sent out-of-country, OOC payments will drop significantly.

Per-test funding for MGL ensures that MGL will be able to supply expanded services within the province and to out-of-province clients for the near-term future. By 2013, MGL will be performing 2 to 7 times as many tests annually as in fiscal 2008 at a total cost of $1.9 - $6M.

Recommendations

1. Transition MGL to a funding strategy in which salaries and maintenance contracts are provided through hospital global funding and increase annually with step increases and promotions. Provide supply funding on a per-test basis starting at $50/test. Update per-test payments rapidly as test type, test frequency, and test cost information become available.
2. Centralize MSP's tracking, administration, and approval of genetic testing inside and outside B.C. to collect test type, test frequency, and test cost tracking information to improve decision-making about tests to develop and provide in-province. MGL would be a logical place to perform this tracking.

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GLOSSARY

Alzheimer's Disease	Degenerative brain disease, the most common form of dementia.
Amyotrophic Lateral Sclerosis (ALS), Sporadic	Progressive degeneration of motor neurons. "Sporadic" indicates random or isolated cases
Babesia	Tick-borne protozoan parasites.
Bordetella	Aerobic bacteria, including *Bordetella pertussis*, the cause of whooping cough
Borellia	*Borellia burgdorferi* is a spirochete bacteria, the cause of Lyme Disease
C&W	B.C. Children's & Women's Health Centre
Charcot-Marie-Tooth	Progressive genetic peripheral neuropathy, characterized by degeneration of nerves' myelin sheath
Connexin 26	Genetic cause of hearing loss due to disrupted potassium flow.
Craniosynostosis	Birth defect in which the bones of the skull join before full brain growth, resulting in abnormal skull shape and brain growth.
Crohn's Disease	Inflammatory gastric disease
Cystic Fibrosis	Genetic disorder characterized by excess mucus production in the respiratory tract and dysfunction of the exocrine glands
CYP2D6	Gene position for Cytochrome P-450 2D6 enzyme, responsible for metabolizing certain drugs.
Cytochrome P-450	Family of oxidizing enzymes present in many organisms. Some of the enzymes metabolize or clear drugs.

DNA	*D*eoxyribo*N*ucleic *A*cid. The molecular coding mechanism for genetic information, held in the nucleus of cells. A double-stranded molecule joined by weak connections.
DNA Extraction	Process by which DNA is collected and purified from a sample.
Ehrlichia	Small bacteria that attack leukocytes (white blood cells) and cause Ehrlichiosis, a disease with symptoms including fever, headache, malaise, and muscle aches.
Exon	Separated DNA portion of a gene with multiple physical sections
Fragile X Syndrome	Most frequent cause of mental retardation. Often characterized by macroorchidism, large, prominent ears, and a long narrow face.
FTE	*F*ull *T*ime *E*mployee or *F*ull *T*ime *E*quivalent
Gene	The physical unit of heredity. A specific region (or separated regions) of a chromosome. Composed of DNA
Genome	Complete genetic information for an individual
Genomics	The study and use (science) of genomes
Genotype	The genetic information for an individual or cell
Haemoglobin (C, S)	Haemoglobin is the oxygen-transporting molecule in blood. C and S refer to abnormalities. S is the most common haemoglobin abnormality and is the basis of sickle cell trait and anaemia.
Haemophilia A	X-linked disorder, deficiency of coagulation factor VIII. Characterized by haemorrhages and failure of blood to clot
Hereditary Hemorrhagic Telangiectasia (HHT)	Genetic disorder characterized by abnormal blood vessels.
HLA-DRB1	Part of the major histocompatibility complex, responsible for portions of the human immune system and autoimmune disorders.
Ichthyosis, X-Linked	Genetic disorder characterized by scaling of the skin.

Likely Cases (LCs)	Statistically calculated number of cases of a disorder, determined by multiplying the size of a population and prevalence of the disorder in that population
Long QT Syndrome (LQTS)	Cardiac syndrome characterized by fainting (syncope), a long QT interval in the EKG, and risk of sudden death by ventricular arrhythmia. Several genetic markers exist for LQTS.
Lou Gehrig's Disease	See *Amyotrophic Lateral Sclerosis (ALS), Sporadic*
Marfan Syndrome	Hereditary disorder of connective tissue.
MGL	B.C. Molecular Genetics Laboratory at B.C. Children's & Women's Health Centre
MoH	B.C. Ministry of Health
MSP	B.C. Medical Services Plan
Mycobacterium	Family of bacteria, including *Mycobacterium tuberculosis,* which causes tuberculosis, and *Mycobacterium leprae*, which causes Hansen's Disease
Mycoplasma	Group of bacteria lacking a cell wall, conferring resistance to some antibiotics that attack the cell wall. Includes *Mycoplasma pneumoniae*, which causes respiratory diseases.
Neurofibromatosis (Type 1, Type 2)	Two common genetic disorders. Spots, freckling, and fibrous tumours on nerve cells characterize Type 1. Type 2 is usually limited to the acoustic nerve and central nervous system.
OOC	Out-of-Country
OOP	Out-of-Province
Pharmacogenomics	Designing and developing drugs targeted to individuals with specific genetic (genomic) characteristics.
Phenotype	Characteristics displayed by an individual
PHSA	Provincial Health Services Authority of B.C.

Prevalence	Frequency of disease in a population
Sequencing	Processing DNA to determine the order (sequence) of base pairs. Reading the information in the DNA.
SNP	*S*ingle *N*ucleotide *P*olymorphism
Sporadic Amyotrophic Lateral Sclerosis (ALS)	See *Amyotrophic Lateral Sclerosis (ALS), Sporadic*
Thalassemia (α, β)	Inherited haemoglobin disorders.
X-Linked Ichthyosis	See *Ichthyosis, X-Linked*
Zygosity	Determining whether siblings of a multiple birth are identical (monozygotic or from a single fertilized egg) or fraternal (di- or poly-zygotic or from multiple fertilized eggs).

1 PROJECT OVERVIEW

1.1 Context

The Molecular Genetics Laboratory (MGL) at the British Columbia Children's & Women's Health Centre is British Columbia's primary laboratory for gene based testing. MGL develops and provides diagnostic testing for congenital anomalies and genetic diseases in foetuses, children, and adults.[1]

MGL is cost-constrained and has been "chronically underfunded despite rapidly increasing demand".[2] A one-time proposal for increased funding was approved in 2008[3] but no provision has been made for sustainable long-term funding.

According to Dr. Brett Casey, Director of the MGL, 200 samples were referred out-of-province for testing in fiscal 2007. Without the recent funding increase, an estimated 400 samples, costing $300K, would have been referred out in fiscal 2007/8.[4] The long-run trend is an increasing shortfall of internal funding and an increasing number of samples referred out.

1.2 Aim

This analysis will address three goals:

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[1] B.C. Children's Hospital. (2007). B.C. Children's Hospital Strategic Plan 2007. Retrieved 2008.08.10, from http://www.bcchildrens.ca/AboutUs/default.htm

[2] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services, pg 4

[3] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services, pg 4

[4] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services, pg 4

- Establish a balanced five-year funding strategy for MGL
- Reduce out-of-province (referred out) expenses
- Present and justify a sustainable approach to genetic testing funding in British Columbia

1.3 Scope

Options and recommendations will be limited to:

- Genetic testing for inherited diseases
- Testing for the benefit of the people and province of British Columbia
- Organizational characteristics of the MGL, including funding and ownership
- Capabilities of the MGL
- A five-year time horizon

2 FUNDING THE B.C. MOLECULAR GENETICS LABORATORY

In order to exploit the benefits of genetic and genomic medicine, the province of B.C. must have the capability to do DNA-based testing. The MGL at C&W is the province's primary lab for doing DNA-based testing. Choices made about funding MGL will affect the efficacy of genetic testing and treatment in B.C. for the foreseeable future.

2.1 MGL Services

MGL offers DNA-based genetic tests for hereditary diseases to residents of B.C. through the Ministry of Health's Medical Services Plan. In addition, MGL facilitates out-of-country (OOC) testing for tests that MGL cannot perform for capacity reasons, supply reasons, or through not yet having developed a test. All of MGL's testing is diagnostic, meaning that it directly affects the treatment of a patient. Predictive testing (tests which indicate the likelihood of having or acquiring a condition) is not performed at MGL because predictive testing is not covered under the provincial MSP.

2.1.1 Mission

MGL lacks a formal mission statement, but the Provincial Medical Genetics Program "…is committed to providing high quality genetic health care to residents of

B.C., while participating in and contributing actively to research and education in the field of Medical Genetics."[5]

MGL's participation in this mission is the creation and execution of DNA-based diagnostic tests, and the facilitation of out-of-province testing. Research into the identifying of genes and their connections to diseases is *not* part of MGL's participation. MGL takes in research as an input and outputs a test.

2.1.2 Tests

MGL offers a menu of 41 DNA-based diagnostic tests for a variety of hereditary genetic diseases and 2 generic DNA-based tests (DNA extraction and zygosity testing). As of June 2008, MGL is working on adding ten new DNA-based diagnostic tests for hereditary genetic diseases. These new tests are responsible for the most out-of-province requests and the highest out-of-province costs and represent approximately 10% of all tests performed in fiscal 2008[6].

2.1.3 Benefits

Genetic testing provides diagnostic confirmation (or differential diagnosis) of many conditions requiring treatment. The Medical Services Commission specifies that "Genetic testing is an insured service in British Columbia only when it is medically necessary to the medical management of the beneficiary's condition"[7], which specifically

5 B.C. Children's Hospital. (2007). B.C. Children's Hospital Strategic Plan 2007. Retrieved 2008.08.10, 2008, from http://www.bcchildrens.ca/AboutUs/default.htm

6 See Appendix B: C&W Molecular Genetics Test Menu on page 63 for a table of currently offered tests and Appendix F: June 2008 Tests to be Added, on page 70 for a table of new tests planned.

7 Medical Services Commission. (2006). Medical Services Commission Out of Province and Out of Country Medical Care Guidelines for Funding Approval. Retrieved 2008.07.05, from http://www.health.gov.bc.ca/msp/infoben/ooc_funding_guidelines.pdf, pg 5

excludes predictive testing and limits covered tests to those which assist a clinician in determining treatment. Every test performed by MGL on a sample from B.C. directly affects treatment of a patient in B.C.

2.2 MGL Funding

MGL is funded through the Ministry of Health as part of the Children's & Women's Health Centre of British Columbia (C&W). The 2007 strategic plan for C&W assumes a continuing funding cap at fiscal 2002/2003 levels[8].

2.3 Mismatch

MGL's budget for laboratory supplies (chemical reagents and other consumables) has been declining sharply over the last five years. The available supply budget projected for fiscal 2007/8 is only 27% of fiscal 2004. Partly as a result of decreased laboratory supplies, the number of samples sent out-of-country (OOC) has risen from an estimated 90 in 2005/6 to a projected 400 in 2007/8. The cost of OOC samples has risen from approximately $70K to nearly $300K.[9]

[8] B.C. Children's Hospital. (2007). B.C. Children's Hospital Strategic Plan 2007. Retrieved 2008.08.10, from http://www.bcchildrens.ca/AboutUs/default.htm

[9] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services

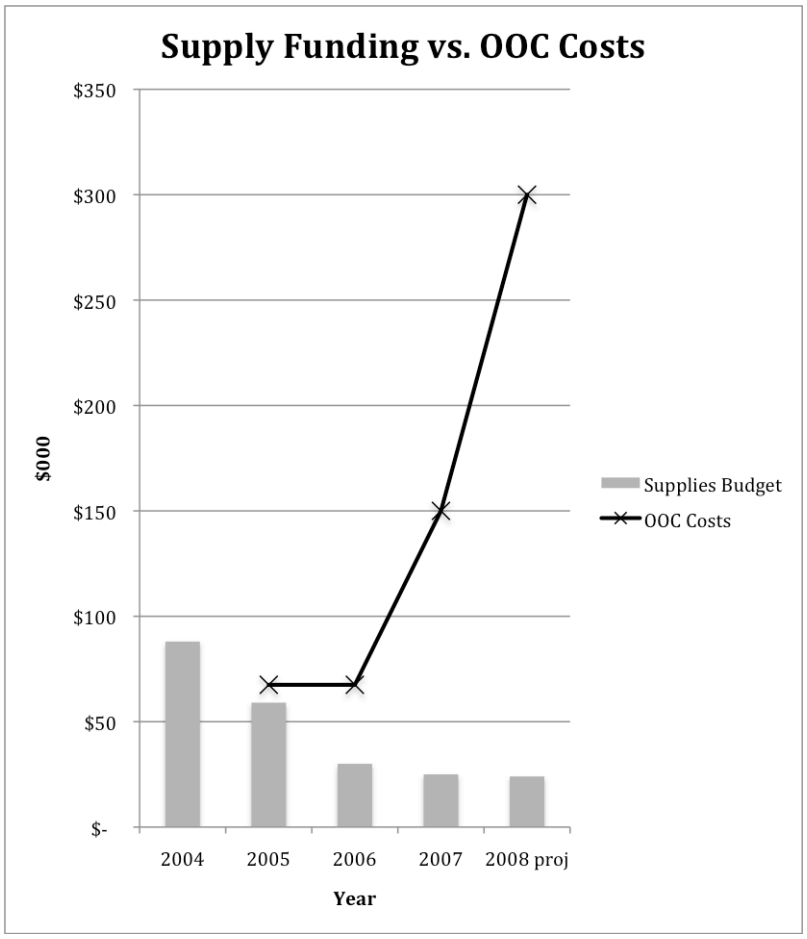

Figure 1: Supply Funding vs. Out-of-Country Costs

3 STAKEHOLDERS

Stakeholders are defined as "[p]eople who will be affected by the project or can influence it but who are not directly involved with doing the project work."[10] Organizations or people who potentially will be affected by or can influence MGL's services and funding include:

- Funding organizations (C&W, MSP, potential future clients);
- Test providers (competing and complementary laboratories);
- Test consumers (doctors, laboratories, individuals);
- Test developers (testing firms, laboratories)

In order to determine which stakeholders are likely to be affected by MGL's funding and which can influence MGL's funding strategy, I will evaluate each category of stakeholder for influence in two directions: the organization or individual's influence upon MGL and MGL's funding strategy; and MGL's influence upon the organization or individual. By finding the high influence stakeholders, recommendations can be evaluated from their point of view and in terms of their interests as well as MGL's interests and point of view.

3.1 Funding Organizations

In accordance with The Golden Rule of Economics, "[h]e that has the gold makes the rules"[11], funding organizations usually have great influence upon decision-making.

[10] Olson, D. & samagaio. (2003). Term Definition: Stakeholder. Retrieved 2008.07.05

[11] "He that has the gold makes the rules" is in common usage, attributed at least once to Lyndon Foreman. There are many similar variants.

The negative form "the power to tax involves the power to destroy"[12] more clearly expresses the power of monetary influence. The organizations providing budget funds to the MGL now and in the future are those most likely to influence organizational decisions.

Whether those organizations are tightly cohesive will dramatically affect their influence. Current funding comes from two major locations: C&W global funding and MSP out of country (OOC) funding. The separation of these two sources (even under the overall umbrella of the B.C. Ministry of Health) reduces the cohesiveness of the funding plan and almost certainly introduces inefficiencies. For example, capacity at the MGL is presently artificially limited by a declining supply budget (provided by C&W global funding) while out-of-country genetic testing costs rise dramatically (provided by MSP OOC funding). Since OOC costs rise considerably more than the cost of doing those tests at MGL[13], the Ministry of Health spends more money on genetic testing than necessary. Despite this, the Ministry of Health has enormous organizational and market power over MGL.

Potential future funding sources include organizations and individuals outside B.C. Categorized collectively as "potential future clients", they have almost no overall organizational cohesion and little market power over MGL. Future clients represent the largest opportunity[14] to expand MGL's market, but that market is only indirectly related

[12] Webster, D. & Marshall, J. (1989). 1798: Daniel Webster (1782 - 1852). Retrieved 2008.07.05, Daniel Webster, arguing before the US Supreme Court in *McCulloch* v. *Maryland*: "An unlimited power to tax involves, necessarily, a power to destroy," 17 U.S. 327 (1819). Chief Justice John Marshall's decision in *McCulloch* v. *Maryland*: "That the power to tax involves the power to destroy … [is] not to be denied".

[13] See Section 5.2, Current and Future Test Costs by Funding Method on page 46 for detailed cost and price information.

[14] See Section 4.5, Detailed Examination of Demand on page 37 for demand scenarios.

to the Provincial Medical Genetics Program's mission "to provid[e] high quality genetic health care to *residents of B.C.*..." [italics mine].

3.1.1 B.C. Children's & Women's Health Centre

B.C. Children's & Women's Health Centre (C&W) is operated by the Provincial Health Services Authority (PHSA) under the auspices of the Ministry of Health. MGL is physically co-located with C&W and currently depends upon C&W for primary funding to cover facilities, personnel, and supplies.

C&W represents a primary barrier to organizational change at MGL. As a practical matter, any changes to MGL funding will have to be supported by C&W. Any proposed changes to MGL funding will have to provide benefits to C&W or, at worst, be neutral to C&W. MGL performs only about ¼ of 1% of the tests done annually at C&W[15] and is therefore below the radar for most decisions at C&W. Considering their respective sizes, MGL might do well to present changes that will *decrease* C&W's involvement in decisions about MGL to reduce MGL's effect upon C&W's attention overhead.

C&W's influence over MGL decision-making is high. The effect of changes at MGL on C&W is likely to be low.

[15] B.C. Children's Hospital. (2007). B.C. Children's Hospital Strategic Plan 2007. Retrieved 2008.08.10, http://www.bcchildrens.ca/AboutUs/default.htm, pg 5; and Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services.

3.1.2 B.C. Ministry of Health

The B.C. Ministry of Health is responsible for delivering healthcare services to the residents of British Columbia. With a $13 billion budget[16] (fiscal 2007/8 estimate), the Ministry of Health represents nearly 35% of the total expenditures by the provincial government of B.C.[17] The Ministry of Health provides funding for MGL through two distinct pathways: Out-of-Country funding via the Medical Services Plan (MSP) and hospital global funding through Children's & Women's Health Centre (C&W).

3.1.2.1 Out-of-Country funding

The B.C. Medical Services Plan (MSP) offers limited coverage of expenses incurred out-of-province or out-of-country, generically referred to as Out of Country (OOC) care. The Medical Services Commission (MSC) publishes guidelines for out-of-country services.[18] The guidelines make provision for non-emergency out-of-country care including "Laboratory and Medical Imaging Tests" and "Genetic Tests"[19], which establish three conditions before OOC expenses for testing are covered:

1. The tests must not be experimental. This is roughly equivalent to requiring that only drugs in the B.C. PharmaCare formulary be covered.
2. Diagnostic alternatives within B.C. and Canada must be exhausted.
3. The test must significantly alter management of the patient's condition. This condition specifies that the test must provide information necessary for

[16] B.C. Ministry of Health. (2007). 2007/08 - 2009/10 Service Plan: Resource Summary. Retrieved 2008.07.22, http://www.bcbudget.gov.bc.ca/2007/sp/hlth/default.html#9

[17] Public Affairs Bureau. (2008). Balanced Budget 2008 Backgrounder. Retrieved 2008.08.10, http://www.bcbudget.gov.bc.ca/2008/backgrounders/backgrounder_fiscal_plan.htm

[18] Medical Services Commission. (2006). Medical Services Commission Out of Province and Out of Country Medical Care Guidelines for Funding Approval. Retrieved 2008.07.05, http://www.health.gov.bc.ca/msp/infoben/ooc_funding_guidelines.pdf

[19] See Appendix H: MSC Guidelines on Genetic Tests on page 73

diagnosis of a treatable syndrome or information necessary to establish which of several treatments are preferable.

In fiscal 2006/7, the MSP approved roughly 200 out-of-country samples for payment. Before the approval of expanded funding in 2007/8, the estimate for fiscal 2007/8 was 400 samples outsourced at a cost of approximately $300K. Although the cost seems high in relation to MGL's budget, the OOC cost is a very small fraction[20] of MSPs annual budget, and is apparently below the radar for improving efficiency. As genetic medicine becomes more mainstream and more diagnostic (as opposed to predictive), it's likely that these costs will rise, possibly dramatically and quickly. MSP has the ability to control whether or not they fund OOC testing, but the acceptability and value of genetic testing is largely out of MSP's control and thus represents a real budgetary risk, as the cost of an OOC test averages several times that of a test performed at MGL[21].

MSP's influence on MGL's funding is high from MGL's point of view. Obtaining access to the OOC expenses currently predicted would add approximately 50% to MGL's funding. From MSP's point of view, MGL's funding requirements are low and MGL's influence upon funding decisions is low to none. This low influence from MGL provides a reason for the long delay[22] in reassessing MGL's declining supply budget.

[20] According to the B.C. Ministry of Health Resource Summary for 2007/8, MGL's expanded budget represents approximately 1/10 of 1% of MSP funding and personnel. B.C. Ministry of Health. (2007). 2007/08 - 2009/10 Service Plan: Resource Summary. Retrieved 2008.07.22, http://www.bcbudget.gov.bc.ca/2007/sp/hlth/default.html#9

[21] See Section 5.2, Current and Future Test Costs by Funding Method on page 46 for detailed information on MGL costs and pricing of tests from outside laboratories.

[22] Anecdotally, funding changes at the MGL have been pursued for roughly four years, and only achieved in the summer of 2008.

3.1.2.2 MSP

Blood tests in B.C. are either on an inpatient or outpatient basis. Inpatient tests are funded through hospital global funding. Outpatient tests are conducted either by public hospitals or private laboratories. Approximately 1/3 of outpatient tests are performed by public hospital labs, with private labs accounting for the remaining 2/3. Outpatient blood tests by hospital or private labs are funded by MSP on a per-test basis.[23]

If testing at MGL were to be funded as outpatient testing instead of inpatient testing, MSP's influence on MGL funding would remain high. The effect of changes at MGL on MSP is likely to be low to none so long as MGL remains owned by the government of B.C. If MGL were privatized or genetic testing outsourced, the cost of testing to the Ministry of Health would almost certainly rise. The influence upon the Ministry would depend upon the rise in cost, but would start as low to none.

In an ideal organization, there would be no difference between the MSP and the MSP Out-of-Country payment program. Practically speaking, genetic testing costs are a minimal drain upon the MSP and the OOC program and there is little urgency to curtail them[24]. Interdepartmental inefficiency raises the cost of providing genetic testing because the budget spent on OOC genetic tests can buy far more tests done by MGL in-province. A coordinating office for genetic testing (probably at MGL, which already facilitates OOC genetic tests) would be better able to track test types, frequency, and

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[23] Bayne, L. (2003). BC Laboratory Services Review. Retrieved 2008.07.05, http://www.health.gov.bc.ca/library/publications/year/2003/lab_review.pdf, pg 20

[24] According to the B.C. Ministry of Health Resource Summary for 2007/2008, MGL's expanded budget represents approximately 1/10 of 1% of MSP funding and personnel. Any changes at MGL are unlikely to have a significant effect upon the MSP or the OOC program. B.C. Ministry of Health. (2007). 2007/08 - 2009/10 Service Plan: Resource Summary. Retrieved 2008.07.22, http://www.bcbudget.gov.bc.ca/2007/sp/hlth/default.html#9

costs, as well as reducing inefficiencies introduced by the current dual-agency funding of genetic tests depending upon the location of the lab doing the test.

3.1.3 Potential future clients

Potential future clients include individuals, laboratories, doctors, and organizations providing healthcare, such as provincial health authorities, insurance companies, hospitals, etc. Potential products include existing tests, newly developed tests, and licenses to perform tests developed at MGL.

Unless large organizations come to dominate MGL's future clientele (unlikely, since the B.C. Ministry of Health is likely to always be MGL's largest source of funds), potential future clients outside B.C. will have low to no influence upon MGL's funding or decision-making.[25] Potential future clients within B.C. may have considerable influence as genetic medicine becomes mainstream and genomics becomes affordable. However, the aggregate influence of demand within B.C. should already be represented by the influence of MSP (and through MSP, the Ministry of Health) and MGL's mission. The effect of changes at MGL on potential future clients will depend on what new services MGL brings on-line and whether those services are available elsewhere. If new services are merely substitutes for ones available elsewhere, MGL's effect upon potential future clients will be low to none. If new services offered by MGL are unique, the effect upon potential future clients could be anything up to high.

[25] MGL's mission is to provide genetic testing services *to the residents of B.C.* (my emphasis), according to B.C. Women's Hospital & Health Centre. (2008). Medical Genetics. Retrieved 2008.06.27

3.2 Test Providers (Competitors and Complementors)

Test providers include publicly and privately owned laboratories. Several publicly owned facilities in B.C. have labs that offer services similar to (and potentially overlapping) MGL's:

- Biochemical and Cytogenetics Laboratories at C&W
- Molecular and Cytogenetics Laboratory at B.C Cancer Agency
- PHSA Laboratories

While these labs use various technologies for testing, it is the diagnostic effect rather than the technology that makes them (in some cases) viable substitutes. Because all the public labs are (officially) cooperative with each other, it is generally a combination of minimal cost and clinician choice as to which lab is used. B.C. Cancer Agency labs concentrate on cancer (e.g. the breast cancer susceptibility genes BCRA1 and BCRA2) and not generalized genetic testing. The Biochemical and Cytogenetics Labs at C&W extensively use non-DNA-based testing to achieve similar results. Many current and future genetic tests have no non-DNA-based alternatives.

Privately owned labs, regardless of location, are in competition with MGL to the extent that they offer similar services. The influence of other labs on MGL funding is the extent to which stakeholders believe private labs represent a cost saving to MSP. The effect of changes at MGL upon competing labs will be dependent upon the increased volume of testing MGL performs. That effect is likely to be low.

3.3 Test Consumers

Individually, test consumers have little market power or influence upon MGL funding. Collectively, doctors are MGL's primary customer and will have the greatest

influence, laboratories are secondary, and individual patients will be tertiary. Overall, I expect consumers to have much lower influence upon MGL than funding organizations. MGL's services will have a medium to high effect upon consumers, particularly patients.

3.3.1 Clinicians

Doctors order genetic tests for diagnostic purposes. In keeping with the MSP guidelines, genetic tests are only covered (and thus generally only ordered by doctors) "when it is medically necessary to the medical management of the beneficiary's condition".[26] Genetic testing remains relatively rare in B.C., and changes to the system will have low to no effect upon clinicians. Clinicians have little effect upon MGL,[27] via aggregated test demand[28].

3.3.2 Labs

Currently labs (inside or outside the province) are unlikely to order genetic tests from MGL. In the future, as MGL brings more tests on-line and offers those tests to non-MSP organizations on a for-profit basis, outside labs may become a significant portion of MGL's clientele. Labs may represent two different revenue streams: direct orders for testing and requests to license tests developed by MGL.

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[26] Medical Services Commission. (2006). Medical Services Commission Out of Province and Out of Country Medical Care Guidelines for Funding Approval. Retrieved 2008.07.05, http://www.health.gov.bc.ca/msp/infoben/ooc_funding_guidelines.pdf, pg 5

[27] It's a valid point to argue that the real influence upon MGL is not the clinicians, but the genetic disorders, and their frequencies, but it is the clinicians who (properly instructed by medical schools and medical literature) recognize the diagnostic need for genetic tests.

[28] Demand is a very indirect influence in a government organization, especially a small portion of a popular governmental institution. More direct is MGL's mission to provide genetic testing for the benefit of residents in B.C. Demand would be a far more direct influence to a privately owned laboratory.

MGL's effect upon labs will be low to none, rising as MGL offers more tests to outside providers. Labs' effects upon MGL will be volume dependent — as volume of tests delivered to any given lab increases, so will the Lab's effect upon MGL.

3.3.3 Patients

Assuming a change in policy regarding patient requests for genetic tests, some test requests will undoubtedly come directly from patients. The volume of such requests (and thus the influence upon MGL) is likely to be low. The effect of such testing upon individual patients will likely be high or at least medium (or they wouldn't be going to the effort of ordering tests themselves). If MSP policy regarding predictive testing were to change, direct test demand from patients would increase.

3.4 Test Developers

Test Developers have almost no influence upon MGL. MGL chooses to develop tests for two reasons: 1) because the test is demanded by clinicians in the province and must be referred-out or doesn't exist; or 2) because the test is widely in demand and would result in referred-in revenue. In the first case the presence or absence of the test is irrelevant. When developing a test exclusively to generate referred-in revenue, level of demand is more important than existence (that is, an additional test for a very prevalent condition might be more in demand than a first test for a low prevalence condition). MGL similarly has quite low influence upon other Test Developers. Only under special conditions might a newly developed test significantly affect another laboratory.

3.5 Summary

It's clear from Table 1 on page 18 that the Funding Organizations (C&W, MSP OOC, & MSP) are the critical decision-makers for MGL funding. They are the only high influence organizations and MGL has no significant reciprocal influence upon any of them, which explains why increasing MGL's funding took four years. The primary influence of all these organizations is to increase the quality and efficiency (and decrease the cost) of health care in B.C. When a clear win (better service for about the same money) was properly presented, it was accepted.

Because the MSP and MSP OOC have the same influences and ultimate goals, I will consider them under the umbrella of MSP from now on. C&W represents the most significant barrier to organizational change at MGL. C&W must be consulted about and approving of changes in MGL's funding strategy.

Stakeholder(s)	Effect upon MGL	MGL's Effect Upon
Funding Organizations		
C & W	High	Low
MSP OOC	High	Low → None
MSP	High	Low → None
Potential Future Clients	Low → None	High → None[29]
Test Providers		
Publicly Owned	Low	Low
Competitive	Low	Volume Dependent[30]
Test Consumers		
Clinicians	Low (Demand Dependent)	Low → None
Laboratories	Volume Dependent	Low → None
Patients	Low	Medium → High
Non-patient Consumers within B.C.	Demand Dependent[31]	Low → Medium
Non-patient Consumers outside B.C.	Low → None[32]	Low → Medium
Test Developers	Low	Low[33]

Table 1: Stakeholder Influence (Summary)

[29] Depends upon (and is positively correlated with) uniqueness of services offered.

[30] Depends upon the relationship between volume of new tests at MGL and volume of competing laboratory.

[31] If (when) genome sequencing becomes commonplace, there will be an enormous change in demand.

[32] MGL's mission is limited to supporting genetic testing for the residents of B.C.

[33] Depends upon the relationship between tests developed at MGL and provided by other Test Developers.

4 DETAILED EXAMINATION OF MGL

The Molecular Genetic Laboratory (MGL) at B.C. Children's & Women's Health Centre is the province's primary lab for DNA-based testing for hereditary diseases. MGL performed approximately 5000 tests in fiscal 2007/8 and facilitated out-of-province testing for about 400 additional samples. MGL's budget has been constant since fiscal 2002/3 but demand has more than doubled since then. Operational efficiencies have allowed MGL to satisfy rising demand but Dr. Casey (Director, MGL) believes that opportunities for operational efficiencies have been exhausted without additional personnel and equipment[34]. MGL must expand capacity to meet the province's rising demand for DNA-based testing.

In order to determine what funding strategy is best for MGL and the province, I examine MGL's position in the provincial health care system, the services MGL provides, how MGL is currently funded, a variety of alternate funding options, and demand for MGL's services.

4.1 Overview of MGL

MGL operates as a stand-alone unit within the B.C. Children's & Women's Health Centre in Vancouver, B.C., and performs a variety of tests for genetic disorders. MGL contributes to the Ministry of Health's overall mission of providing sustainable, affordable health care for the residents of B.C. while operating within the requirements of the Medical Services Committee's requirements for non-experimental testing. Because

[34] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services

genetic testing is a fast-changing field of study, requirements for additions to MGL's menu of tests may happen quickly. Using mission statements and published goals, I will establish a set of criteria by which future decisions can be evaluated.

4.1.1 Mission

MGL has and supports missions, goals, and objectives for several levels of organization in the province[35]. It's important to compare alternate scenarios with organizational visions, missions, goals, and objectives firmly in mind. MGL's primary roles in these missions, goals, and objectives are creating and performing DNA-based diagnostic tests and facilitating out-of-province genetic testing. Decision criteria for alternatives thus include whether or not (or to what extent) the alternatives contribute to:

- High quality genetic health care
- Diagnostic consultation and services on which vital decisions are made in the care of patients
- [Helping individuals] make healthy lifestyle choices
- Protection of the public from preventable disease, illness, and injury
- Sustainable, affordable, publicly funded health system
- Sound business practices

These criteria break down into two categories:

- Health Care Related: High quality genetic health care consists of diagnostic testing which allow clinicians to make decisions affecting the care of patients or patients to make healthy lifestyle choices to protect themselves (the public) from preventable disease, illness, and injury.
- Business Related: High quality genetic health care uses sound business practices to contribute to a sustainable, affordable, publicly funded health system.

[35] Appendix G: Mission Statements and Goals on page 71 lists specific missions, goals, and objectives for organizations to which MGL is responsible.

4.1.1.1 Decision Criteria

While there are many possible criteria derivable from the above mission and goal statements, I present mine for this project. Each element is evaluated by asking:

1. Does it promote diagnostic testing?
2. Does it allow…
    a. clinicians to make decisions affecting the care of patients? or
    b. patients to make healthy lifestyle choices?
3. Does it protect the public from preventable disease, illness, and injury?
4. Is it a sound business practice?
5. Is it sustainable and affordable in the short and long-term?
6. Does it promote a publicly funded health system?

The first three are health care related and generally fall outside this project's scope. It is sufficient to note that the initial selection of a test must meet these criteria: be diagnostic (not predictive); make a difference to care or lifestyle choices of a patient; and protect the public (patients) from preventable disease, illness, or injury.

4.1.1.2 Decision Criteria and Stakeholders

How do the important stakeholders (funding organizations) consider these criteria? Only C&W is in the day-to-day business of directly providing healthcare, so they will regard Criteria 1 – 3 as more important, while the MSP will have at least some concern for business issues.

Criteria	C&W	MSP	Sum
1. Diagnostic Testing	++	+ (OOC)	+++
2a. Clinical Decisions	++	+ (OOC)	+++
2b. Patient Decisions	+	O	+
3. Protect Public	+	+	++
4. Sound Business Practice	O	+	+
5. Sustainable & Affordable	O	++	++
6. Public Funding	O	++	++

Table 2: Importance of decision criteria to current funding organization stakeholders

As a rule, the bureaucratic organization will be more concerned with business practices, but MSP OOC involves itself in questions of whether tests are diagnostic and whether they affect clinical decisions. C&W, being closer to the patient, will concern itself less with long-term questions of business and sustainability than effective treatment. Between them, the two organizations (C&W and MSP) consider all the decision criteria at least somewhat important. I will address the business issues (Criteria 4 – 6) and leave the medical criteria to the medical experts.

4.1.2 Location

MGL is located at the B.C. Children's Hospital, 4480 Oak Street, Vancouver, B.C. Vancouver B.C. is a biotech cluster with multiple research institutions, life science associations, bioinformatics firms, biopharmaceutical firms, and supporting infrastructure including consultants, contract laboratories, and manufacturers.[36] MGL is co-located with B.C. Children's Hospital because of history and the number of pre-natal and childhood tests performed.

[36] LifeSciencesBC. (2008). LifeSciences BC Member Profiles by Sector. Retrieved 2008.06.29, http://www.lifesciencesbc.ca/Members/Member_Company_Profiles/Members_by_Sector2.asp

4.1.3 Staff

Dr. Brett Casey has been Director of the MGL since 2001, and is also head of the Program in Laboratory Genetics at B.C. Children's Hospital. Dr. Casey performed his residency in Anatomic and Clinical Pathology at the University of California at San Diego and fellowships in Human Molecular Genetics and Paediatric Pathology at Baylor College of Medicine and Texas Children's Hospital in Houston, Texas.

Dr. Casey's molecular genetics experience covers a wide range of applications, and his research has focused on malformation syndromes in humans. He is a Diplomate of the American Boards of Pathology and Medical Genetics and Pathology and is a Fellow of both the Canadian and American Colleges of Medical Genetics.[37]

The functional staff of the MGL consists of 3.5 FTE technologists. With the recently approved funding, additional functional staff will be hired to 5 FTE technologists.

Employee turnover in the MGL is very low.

4.1.4 Funding History

MGL was first established in 1990, funded by the provincial Medical Service Plan (MSP) via the Alternative Payments Branch. This funding continued through 2002. In B.C.'s fiscal 2004, apparently because genetic testing was becoming mainstream, MGL's budget allocation was transferred to the Children's & Women's Health Centre (C&W).[38]

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[37] Casey, B. (2008). Re: [Sustainable Funding Analysis] Collected Questions

[38] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services

MGL's total operating budget is divided into professional salaries and supplies and operating expenses. Professional salaries rise over time and a constant overall budget thus leads to a declining supplies budget. Since fiscal 2003/4, MGL's supplies budget has declined from nearly $90K to near $25K, a reduction of almost 2/3. In fiscal 2007/8, MGL's supplies budget was returned to $100K and supplemented by $93K in referred-in revenue.

4.2 Detailed Examination of Services

MGL's services are all related to diagnostic tests for hereditary diseases. More and more DNA markers for diseases are being found as a result of the Human Genome Project. Because of Medical Services Commission rules on genetic testing, no predictive tests are performed, only diagnostic tests. These tests show definitively that a patient does or does not have a specific genetic abnormality directly associated with a disease.

MGL is expanding their menu of services as the result of the approval of Dr. Casey's *Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services*[39] in June 2008. The detailed examination of MGL services will look at pre-approval tests, post-approval tests, and speculative tests for the future.

4.2.1 Current

MGL offers 43 distinct genetic tests[40], including:

- Charcot-Marie-Tooth — hereditary neuropathy

[39] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services.

[40] See Appendix B: C&W Molecular Genetics Test Menu on page 63 for the complete menu

- Connexin 26 — deafness
- Cystic Fibrosis — lung tissue & small intestine mucosal disease
- Fragile X — inherited mental retardation
- Haemoglobin S, C — blood disorders
- Haemophilia A — blood clotting disorder
- Thalassemia α and β — anaemia
- X-Linked Ichthyosis — STS Deficiency

In fiscal 2006/7, MGL tested more than 4000 samples. Estimated growth for fiscal 2007/8 is over 20% to more than 5000 samples. With the tests added in June 2008, projected demand in fiscal 2008/9 is estimated at approximately 7500 samples.

4.2.2 Possible Future

MGL has already proposed developing new tests "in demand by the B.C. healthcare community but presently unavailable in the province".[41] The proposal was approved in June 2008 to add 10 additional tests[42] including:

- Craniosynostosis syndromes — skull deformation
- Hereditary hemorrhagic telangiectasia (HHT) — malformation of blood vessels
- Long QT syndrome (LQTS) — cardiac rhythm disorder
- Marfan syndrome — connective tissue disorder
- Neurofibromatosis Type 1 and 2 — fibroid tumors on nerves

All future services are speculative. Those proposed by MGL are less speculative than others. Other possible future tests are representative placeholders — that is, testing may be done for these conditions or others of roughly similar prevalence. I present scenarios for comparison, using these and other potential tests as guides for market size

[41] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services, pg 5

[42] See Appendix F: June 2008 Tests to be Added on page 70 for a complete list.

and demand estimation[43]. It is not likely that any specific predicted scenario will be correct, because genomic medicine and pharmacogenomics are fast moving fields.

Other potential tests include BRCA1 and 2, but testing is already available in province from the B.C. Cancer Agency.[44] As more diseases are linked to genetic markers, testing demand is likely to rise. Recent publications suggest genetic links in the following conditions:

- Alzheimer's disease[45] — common form of dementia
- Crohn's disease[46] — autoimmune gastric disorder
- Sporadic amyotrophic lateral sclerosis (ALS or Lou Gehrig's disease)[47] — neurodegenerative disease

More speculative genomic medicine tests include:

- Detection of patients susceptible to adverse drug reactions[48] — including Cytochrome P-450 testing for drug metabolizing variations.
- Detection of autoimmune susceptibility markers — including HLA-DRB1 testing.
- Identifying so-called "responders" — finding genetic markers for those in trials or treatment who benefit from a drug.[49]

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[43] See Section 4.5, Detailed Examination of Demand on page 37 for demand scenarios.

[44] B.C. Cancer Agency. (2005). Genetic Counselling and Testing. Retrieved 2008.07.04, http://www.bccancer.bc.ca/PPI/Prevention/Hereditary/GeneticcounsellingandtestingHtm#testing

[45] Paddock, C. (2008). Scientists Find New Gene Link To Alzheimer's Disease. Retrieved 2008.07.04, http://www.medicalnewstoday.com/articles/112803.php; and Marambaud, P., Dreses-Werringloer, U., Lambert, J.-C., Vingtdeux, V., Zhao, H., Vias, H. et al. (2008). A Polymorphism in CALHM1 Influences Ca2+ Homeostatis, Aβ Levels, and Alzheimer's Disease Risk. Cell, 133, 1149-1161

[46] Henderson, M. (2008). Gene discovery will help to fight Crohn's disease. Retrieved 2008.07.04, http://www.timesonline.co.uk/tol/news/uk/science/article4238020.ece

[47] Science Daily. (2008). New Gene Responsible For Lou Gehrig's Disease Identified. Retrieved 2008.07.04, http://www.sciencedaily.com/releases/2008/03/080331122528.htm

[48] Daly, A. K., King, B. P., & Leathart, J. B. S. (2005). *Cytochrome P450 Protocols* (320). Secaucus, NJ: Springer Science & Business Media; and Ingelman-Sundberg, M. (2001). Implications of Polymorphic Cytochrome P450-Dependent Drug Metabolism For Drug Development. *Drug Metabolism and Disposition*, *29*(4), 570-573; and Ingelman-Sundberg, M. (2008). Pharmacogenomic Biomarkers for Prediction of Severe Adverse Drug Reactions. New England Journal of Medicine, 358;6

[49] Connor, S. (2003). Glaxo chief: Our drugs do not work on most patients. Retrieved 2008.07.04

These two previous categories all fail my Criterion 1: these tests are predictive rather than diagnostic. However, they serve as quantitative examples of the *kind of test* that might be added in the future.

DNA based testing can also affect treatment choice when used to conclusively identify disease-causing organisms including:

- *Babesia microti* and *WA1* (babesiosis)[50]
- *Bordetella pertussis* (pertussis or whooping cough)[51]
- *Borrelia burgdorferi* (chronic Lyme disease)[52]
- *Ehrlichia chaffeensis* (human monocytic ehrlichiosis or HME)[53]
- *Ehrlichia phagocytophilia* (human granulocytic ehrlichiosis or HGE)[54]
- *Mycobacterium*[55]
- *Mycoplasma incognitus*[56] and *pneumoniae*[57]

As more genetic markers are discovered for more medical conditions (hereditary, epigenetic, and infectious) the field of potential tests becomes larger. Should genomic medicine become a reality, full genome sequencing will become commonplace.[58]

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[50] IGeneX, Inc. Babesiosis. Retrieved 2008.07.04, http://www.igenex.com/tickset1.htm

[51] Division of Health Surveillance - Epidemiology. (2007). Pertussis PCR testing at the Vermont Department of Health Laboratory. Infectious Disease Bulletin Retrieved 2008.07.04, http://healthvermont.gov/pubs/IDB/documents/IDB12_07.pdf; and Kirchner, J. T. (1999). Infectious Cause of Chronic Cough in Adults. American Family Physician.

[52] IGeneX, Inc. Detection of *Borrelia Burgdorferi* by PCR -- Overview. Retrieved 2008.07.04, http://www.igenex.com/lymeset5.htm

[53] IGeneX, Inc. Ehrlichiosis. Retrieved 2008.07.04, http://www.igenex.com/tickset2.htm

[54] IGeneX, Inc. Ehrlichiosis. Retrieved 2008.07.04, http://www.igenex.com/tickset2.htm

[55] MacReady, N. (2006). PCR Test Hastens Identification of Mycobacteria Species. Retrieved 2008.07.04, http://www.medscape.com/viewarticle/546427

[56] Vojdani, A. & Choppa, P. C. Sensitive Method for the Quantitative Detection of Mycoplasma Infections. Retrieved 2008.07.04, http://www.immuno-sci-lab.com/html/pcr.html

[57] Kempsell, K. E., Cox, C. J., McColm, A. A., Bagshaw, J. A., Reece, R., Veale, D. J. et al. (2001). Detection of *Mycobacterium tuberculosis* Group Organisms in Human and Mouse Joint Tissue by Reverse Transcriptase PCR: Prevalence in Diseased Synovial Tissue Suggests Lack of Specific Association with Rheumatoid Arthritis. *Infection and Immunity*, *69*(3), 1821 - 1831

[58] This is perhaps a reversal of the causative statement: if full genome sequencing becomes cost effective (and therefore commonplace), genomic medicine will become a reality.

4.3 Detailed Examination of Funding 2007-2008

Table 3 provides basic financial information for MGL's fiscal 2007/8.[59] Referred-in revenue comes from tests referred to MGL from outside the B.C. healthcare system — the inverse of OOC payments. Compensation refers to total labour costs (excluding the Director). Supplies provides for chemical reagents, laboratory consumables, and maintenance contracts.

	2007-2008 Budget			2007-2008 Actual		
	Expenses	Revenue	Totals	Expenses	Revenue	Totals
Referred-In Revenue		$25,000			$93,297	
Total Revenue			$25,000			$93,297
Compensation	$(399,726)			$(363,038)		
Supplies	$(99,624)			$(213,908)		
Total Expenses			$(499,350)			$(576,946)
Budget Total			$(474,350)			$(483,649)

Table 3: 2007-2008 MGL Budget vs. 2007-2008 MGL Expenditures

4.3.1 Funding Sources

Ninety-five percent of MGL's budget in 2007-2008 came from C&W funding, with the remaining five percent budgeted as referred-in revenue. As budgeted, C&W funding is divided 80% for Compensation and 20% for Supplies. As shown in the right half of Table 3, the actual division of money between salaries and supplies is closer to 65%-35% than 80%-20%. The differential comes largely from increasing referred-in revenue and using that revenue for supplies. A small diversion of funds from compensation (via an unfilled .5 FTE position) makes up most of the difference.

[59] Casey, B. (2008). 07-08 Genetics Fiscal Position.xls

4.3.2 Cost Sources

MGL's continuing costs come from two primary sources:

- Employee salaries
- Supply funding

A third, minor, source of costs is maintenance agreements on equipment, but cost tracking at the laboratory level does not appear to capture this as a separate category. For fiscal 2007/8, maintenance agreements are estimated at $25K[60].

4.3.2.1 Employee Salaries

Labour expense is by far MGL's most significant cost source. Step increases and reclassification cause labour expenses to rise annually, regardless of test volume.

MGL currently has 3.5 FTE technologists, soon to expand to 5 FTE technologists. The Director's salary is not included in MGL's global budget from C&W.

Labour costs are now 63% of MGL's costs.

4.3.2.2 Supply funding

Funding for reagents and other laboratory consumables is the most variable of MGL's budget categories. Over the fiscal years from 2004/5 to 2007/8, real budgeted supply funding has dropped precipitously from nearly $90K to about $25K[61], more than a 70% reduction. During that same period, samples tested have risen over 45%. In 2007/8, supply funding rose back to $100K, which was still insufficient to cover necessary costs

[60] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services

[61] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services.

of nearly $215K[62]. The difference between $99K budgeted supply costs and the actual $214K supply expense can be accounted for by:

- $99K in referred-in revenue applied to the purchase of supplies
- Repurposing of other funds, including funding for an unfilled .5 FTE position

According to budget, the mean supply cost per test performed was budgeted to drop from $25 to $5 in five years. The mean supply cost reduction is almost certainly an accounting anomaly rather than a real reduction. However, 2008 actual expense figures show a mean supply cost per test of $42, 167% of the 2004 budgeted supply cost per test. So at a time when the supply budget was fixed, the number of tests increased and the mean per test cost also increased. Supply funding is 37% of MGL's costs.

4.3.2.3 Potential Future Technology Costs

MGL has no plans to upgrade equipment beyond that authorized by the 2008 funding change. According to Dr. Casey, the PHSA lab organization is currently discussing outsourcing DNA sequencing to the Genome Sequencing Centre. If implemented, this transfer of responsibility would "…cut [MGL's] costs dramatically and obviate the necessity of upgrading [MGL's] sequencing instrument(s)…"[63].

[62] Casey, B. (2008). 07-08 Genetics Fiscal Position.xls.
[63] Casey, B. (2008). Re: [Sustainable Funding Analysis] Collected Questions

4.3.3 Current Shortfall

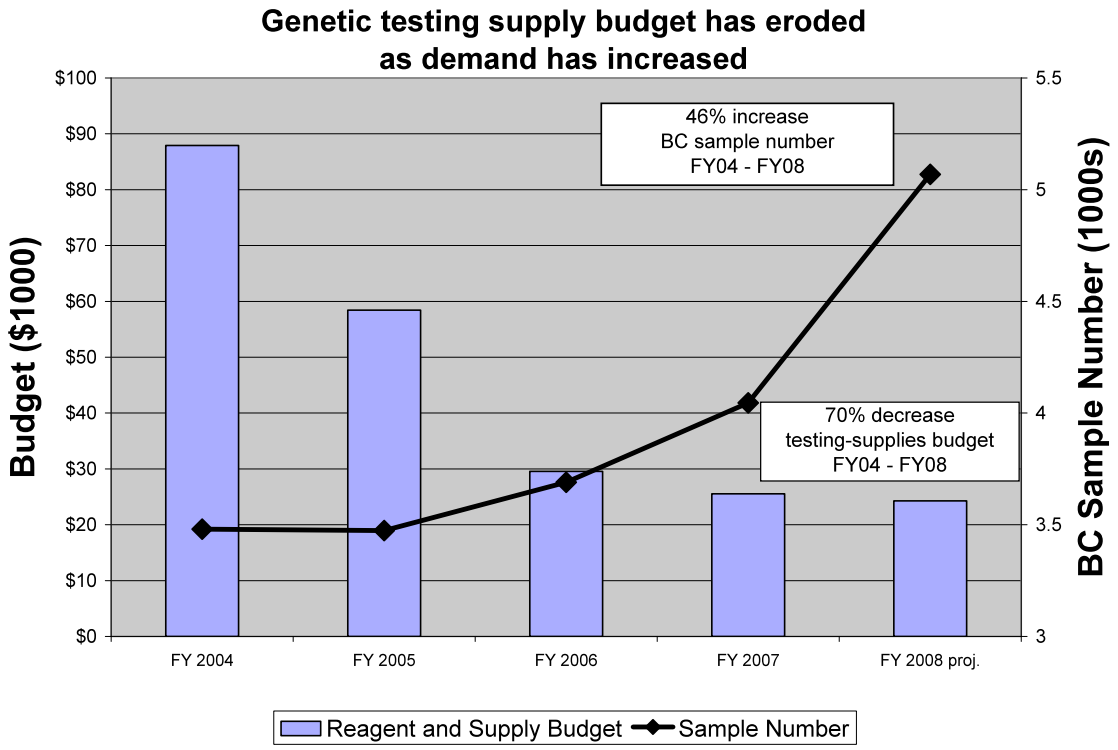

Figure 2: Supply Budget vs. Demand[64]

In fiscal 2008, MGL had a supply budget shortfall (compared to actual expenses) of $115K. MGL has successfully increased samples processed despite declining real supply budgets. By increasing efficiencies, repurposing funds, and increasing referred-in revenue from out-of-province tests, MGL has managed to maintain accelerating growth in test volumes.

Despite increased operational efficiency, MGL planned to send 400 samples out of country (OOC) for testing at a total cost of $300K ($750 per test mean cost) because of lack of capacity or lack of a required test.

[64] Graph from Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services

4.4 Alternative Funding Strategies

Current funding models within the B.C. Ministry of Health include global funding (as MGL is currently funded) and per-test funding (as MSP compensates private blood labs and out-of-province genetic test labs). Ownership can be public (e.g. MGL) or private (e.g. BC Biomedical Laboratories, Ltd.[65]). Combining forms of funding and ownership presents a short menu of possible funding strategies:

- Globally funding MGL as a publicly owned lab (current);
- Per-test funding MGL as a publicly owned lab;
- Fixed cost (global equivalent) funding a privately owned lab — can be discounted as unrealistic. No private corporation is likely to accept such an arrangement where more volume doesn't bring more income;
- Per-test funding MGL, Inc. as a privately owned lab (privatizing MGL);
- Per-test funding genetic tests through existing labs (outsourcing).

4.4.1 Current Global Funding for MGL as a Public Lab

MGL is currently funded as global funding from the Children's & Women's Health Centre of British Columbia (C&W), meaning that MGL receives a fixed annual budget from the C&W annual budget. In recent years, the MGL budget has been constant. Because of rising personnel costs, the available non-labour budget has decreased.

The B.C. Medical Services Plan pays for outsourced samples according to MSP's Out-of-Country Medical Care Guidelines.[66] These costs are not (technically) part of the

[65] http://www.bcbio.com/

[66] Medical Services Commission. (2006). Medical Services Commission Out of Province and Out of Country Medical Care Guidelines for Funding Approval. Retrieved 2008.07.05, http://www.health.gov.bc.ca/msp/infoben/ooc_funding_guidelines.pdf

MGL's budget but do represent real costs to the provincial Ministry of Health for genetic testing.

In 2007, Dr. Brett Casey proposed *Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services.*[67] In June 2008 the proposed budget increase was approved. This increased MGL's annual budget by $240K (to approximately $800K) and allocated a one-time investment of $350K for equipment. The purpose of this increase is to facilitate testing for Long QT Syndrome (LQTS) and other new tests.[68] This funding scenario maintains the current global funding model as modified in June 2008, and mirrors the provincial funding model for in-hospital laboratories.

4.4.2 Test-Based Funding MGL as a Public Lab

The B.C. Ministry of Health pays privately owned laboratories in B.C. on a per-test basis through the Medical Services Plan. The market price of a privately performed blood test, therefore, includes fixed costs as well as variable costs, and profit for the private laboratory.

4.4.2.1 Pure Per-Test Funding

Payments to MGL could be handled just as are payments to privately owned laboratories. On a total cost basis, the aggregate of per-test payments would roughly equal MGL's total budget as adjusted for test volume. For 2007/8, original budgeted cost per test would be roughly $115 (total budget of $577K divided by 5100 tests performed).

[67] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services

[68] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services

As modified in June 2008, cost per test would be roughly $160 (total budget of $817K divided by 5100 tests performed).

4.4.2.2 Mixed Per-Test Funding

MGL's budget could be split into fixed and semi-variable costs (salaries, facilities, maintenance contracts) and variable costs (reagents, other consumables, courier service, and postage). For fiscal 2007/8 as originally budgeted, semi-variable costs would be $363K and variable costs would be $214K or $42 per test up to the volume manageable by the current staff. The breakdown for fiscal 2008/9 as modified is unavailable until hiring is complete and salaries are established. An estimated breakdown for 2008/9 as modified in June might be semi-variable costs of $600K and variable costs of $215K. Semi-variable costs would rise according to step increases and promotions and staff additions[69], while variable costs would rise and fall according to the number and cost of tests performed and developed.

4.4.3 Privatization (Test-Based Funding of MGL, Inc. as a Private Lab)

Taking the MGL from public ownership (aka MGL) to private ownership (aka MGL, Inc.) and contracting with the resulting company to perform genetic testing at market rates. This funding scenario extends the current provincial model for other medical testing outside hospital laboratories, such as blood testing and radiology, to genetic testing. MGL, Inc. is free to offer services to other clients throughout the world, just as existing private laboratories do.

[69] Salaries are a semi-variable cost — fixed over a wide range of volume, and then increasing/decreasing by a relatively large increment as one or more FTEs are hired/laid off.

4.4.4 Outsourcing

Closing the MGL and using outside laboratories exclusively for genetic testing at market rates. This funding scenario relies entirely upon market forces for genetic testing.

4.4.5 Other Potential Sources of Revenue

MGL has two potential future sources of revenue: referred-in testing and licensing of established tests. MGL made over $90K through referred-in testing in fiscal 2007/8 and there is every reason to believe that revenue stream can increase if MGL has additional capacity. Tests are not yet being licensed, but any test developed at MGL could be licensed to other laboratories for additional revenue.

4.4.5.1 Tests for non-MSP clients

MGL could offer tests on the open market if they have excess testing capacity. MGL has spent as much as $5400 per test.[70] Other laboratories list test prices from $200 to $6000. The mean price of a test from Molecular Diagnostics Laboratories (MDL)[71], GeneDX[72], Massachusetts General[73], Johns Hopkins[74], and Tulane[75] is $880/test, with averages at the various laboratories ranging from $352/test at MDL to $1472/test at Tulane.[76]

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[70] Casey, B. (2008). Re: [Sustainable Funding Analysis] Collected Questions, pg 5.

[71] Fontaine, R. N. (2008). Customer Prices and CPT Codes 2006

[72] GeneDX. (2008). Current Price List. Retrieved 2008.07.14, http://www.genedx.com/pdf_files/current_price_list.pdf

[73] Massachusetts General Hospital. Neurogenetics DNA Diagnostic Laboratory Price List. Retrieved 2008.07.13, http://www.massgeneral.org/neuroDNAlab/price_list2_4-04.xls

[74] Johns Hopkins DNA Diagnostic Laboratory. Search for Tests by Syndrome Name. Retrieved 2008.07.14, http://www.hopkinsmedicine.org/dnadiagnostic/SyndromeSearch.htm

[75] Tulane Health Sciences Center. Matrix DNA Diagnostics. Retrieved 2008.07.11, http://www.som.tulane.edu/gene_therapy/matrix/matrix_dna_diagnostics.shtml

[76] Means calculated without access to frequency of tests.

Because tests from the open market will provide more revenue per test than B.C. internal testing, safeguards must be put in place to guarantee that internal tests have absolute priority over non-MSP tests. Additional revenue from non-MSP tests can be returned to the province as income, set aside for capacity expansion, or used for some other purpose as decided by the Ministry of Health.

4.4.5.2 Test Licensing

Upon developing useful tests, even if not unique, MGL could license those tests to other laboratories. Licensing fees are likely to be low, because tests are developed from publicly available (and unprotect-able[77]) information linking specific DNA locations and disease.

4.4.6 Summary

Table 4 shows summary information for each funding option, including whether costs are fixed or vary by volume, the funding agencies, existing organizations using the funding model, how the model scales up and down, whether the lab ownership is public or private, and whether the model is sustainable and affordable. Much of this information applies directly to the business criteria 4 – 6 on page 21. I will use it in combination with other data to form my recommendations.

[77] Wright, C. (2008). European recommendations on patenting and licensing genetic tests. Retrieved 2008.07.25, http://www.phgfoundation.org/news/4140/. Although this information is technically a "discovery" and not an invention, patents have been granted. Some jurisdictions choose to ignore these patents and allow use of the discovered information. This issue will eventually be litigated in both national and international courts.

Characteristics	Current Global Funding	Per-Test Funding	Privatization	Outsourcing
Fixed Costs	X			
Cost Varies by Volume		X	X	X
Funding Agency (-ies)	C&W + MSP	MSP; C&W + MSP	MSP	MSP
Organizations in B.C.'s health system using this funding model	Hospital Labs	Private Labs; None	Private Labs	Private Labs
Scales up	Poorly	Well	Very well	Very well
Scales down	Adequately	Adequately	Very well	Very well
Public/Private Ownership	Public	Public	Private	Private
Sustainable?	No	Yes	Yes	Yes
Affordable?	Yes	Volume Dependent	Volume Dependent	Volume Dependent

Table 4: Summary of Funding Options

4.5 Detailed Examination of Demand

MGL's primary potential market is the population of B.C. But that market is only reached if the Ministry of Health moves to fully genomic medicine. At present, MGL's end-user market is clinicians with:

- Patients at risk of hereditary disease
- Patients requiring diagnosis of syndromes where the differential diagnosis includes hereditary diseases

MGL directly serves only that portion of the market requiring tests MGL provides, while facilitating application by clinicians to the MSP OOC program to have

samples sent to out-of-province labs for testing[78]. MGL also hands some patients to other in-province laboratories[79].

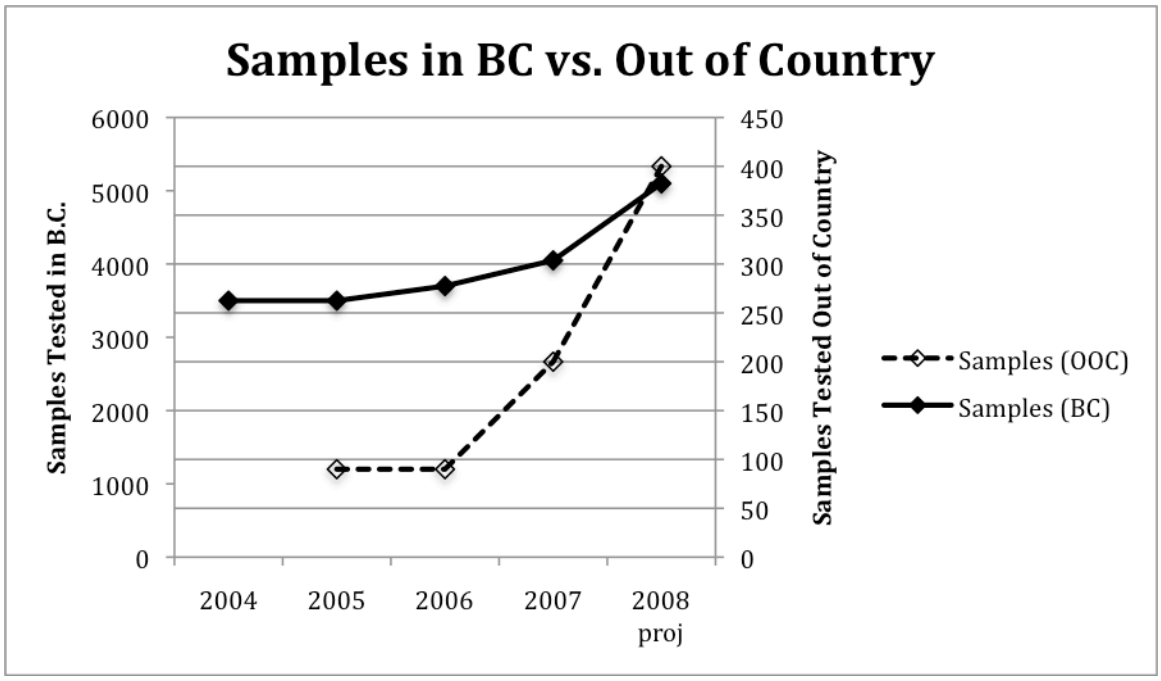

Figure 3: Comparison of Samples Processed Inside and Outside B.C.[80]

MGL has three ways to expand the market:

- Test at a higher rate
- Offer more tests
- Offer tests to a larger population

Since genetic disease prevalence is at least roughly known, I will estimate the number of likely cases (LCs) of each genetic disease based upon the B.C. population[81]. I

[78] This diversion of effort to paperwork for OOC samples represents a dead weight monetary loss to MGL and confirms that patient care is their primary driver. Consolidating genetic testing within the province and the management of OOC genetic testing into a single office will convert the dead weight loss to a cost of OOC testing, which is how it should be tracked.

[79] See Appendix C: C&W Cytogenetics Test Menu through Appendix E: C&W Biochemical Genetics Test Menu starting on page 63 for lists of tests provided by other C&W laboratories

[80] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services.

can estimate the number of tests MGL performs per likely case, allowing me to model demand based upon tests offered, the prevalence of those diseases, and the size of the target (market) population.

The rate of testing per 1000 LCs is 279 - 444 tests per 1000 LC or 28% to 44%, as shown in Table 5, below. Roughly speaking, each LC results in between ¼ and ½ of a genetic test at MGL. Without better information on the number of pre-natal tests, it's hard to estimate how many of these tests are confirming a diagnosis already made through other means, but .25 - .50 tests per LC seem low for effective coverage in the post-natal population. A change to 2 tests per LC would increase the within-B.C. market by between four and eight times. See Table 5 for estimates of testing volume at double the existing rate and at a rate of one test per LC.

Offering tests for additional genetic problems as a market expansion is already under way. The additional budget added in June 2008 addresses a proposal to add ten new tests to the 43 currently offered, increasing the test menu by 23% and the likely number of cases in B.C. by between 43% and 48%.

Offering tests throughout Canada or the US provides MGL with substantially larger markets. However, it's unclear what portion of those markets is underserved, and MGL might simply be entering a satisfied and competitive market. Competitive laboratory rates of testing and number of tests are not available.

[81] See Appendix A: Prevalence of Genetic Diseases on page 60 for more information on the prevalence of genetic diseases.

	Low LC Estimate	High LC Estimate
Tests Performed in B.C.	5,100[82]	5,100[82]
LCs (old tests) by known prevalence	5,343	8,506
known prevalence covers what % of tests?	47%	47%
LCs (old tests) estimate	11,488	18,289
Tests/1000 LCs	444	279
LCs (added tests) by known prevalence	5,567	7,855
Total LCs	17,055	26,143
% Increase in LCs with added tests	48%	43%
Projected Tests Performed with Added Tests	**7,571**	**7,290**
	Low LC Estimate	**High LC Estimate**
Projected Tests (Existing Tests in BC)	5,100[82]	5,100[82]
Projected Tests (Added Tests in BC)	7,571	7,290
Projected Tests at 2x Test Rate (Existing Tests in BC)	10,200	10,200
Projected Tests at 2x Test Rate (Existing + Added Tests in BC)	15,143	14,581
Projected Tests at 1 Test/LC (Existing Tests in BC)	11,488	18,289
Projected Tests at 1 Test/LC (Existing + Added Tests in BC)	17,055	26,143
%Increase in Tests at 1 Test/LC (All Tests in BC)	334%	513%

Table 5: Projected Test Volume with proposed added tests, June 2008 and at higher test rates

4.5.1 B.C. Consumers

B.C.'s present population of between 4 and 4.4 million people supported between 3500 and 5500 tests in the years 2004 – 2008. Precise information on the number of tests referred-in is unavailable, but $93K in referred-in revenue suggests anywhere from 46 to 175 tests are from out-of-province.[83]

[82] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services.

[83] $93K referred-in revenue divided by $526 (the lowest average price of a genetic test from the available test menus from outside laboratories) = 176 tests.

By researching the known prevalence of diseases[84] diagnosed by the existing tests, I estimate the number of Likely Cases (LCs) in B.C. in Table 5. Combined with the percentage of actual tests performed and the percentage of tests for which I have LCs, I obtain a ratio of tests done in B.C. per 1000 LCs.

Adding the LCs for the added tests provides a total number of LCs in B.C. and re-applying the ratio of tests done per 1000 LCs yields a projected number of tests that will be performed annually once the new tests are added to the menu. In addition, I provide estimates of the projected numbers of tests if the testing rate should increase, which might occur if MGL's services were more widely or forcefully presented to clinicians.

4.5.2 Out of Province Consumers

Using the same Tests/LC value as in B.C., projections for the potential number of tests to be performed in Canada and the US are:

Out of BC Demand	Canada Low	Canada High	US Low	US High
LCs (old tests)	86,188	137,209	782,458	1,245,644
LCs (added tests)	41,765	58,931	379,208	535,046
Total LCs	127,953	196,139	1,161,667	1,780,690
Total LCs/1000 * Tests/1000 LCs	**56,804**	**54,696**	**515,710**	**496,569**

Table 6: Projected Test Demand outside B.C.

Out of BC Demand	Canada Low	Canada High	US Low	US High
Projected Tests at 1 Test/LC (Existing Tests)	86,188	137,209	782,458	1,245,644
Projected Tests at 1 Test/LC (Existing + Added Tests)	127,953	196,139	1,161,667	1,780,690

Table 7: Projected Test Demand outside B.C. at higher testing rates

[84] See A: Prevalence of Genetic Diseases on page 60 for the prevalence of some genetic diseases.

5 COMPARISON OF FUNDING ALTERNATIVES & OUTCOMES

In order to determine the preferred funding strategy, I examine scenarios for test volume, testing rate, and test cost. Volume scenarios include linear and exponential projections based upon previous years actual test data. Testing rate scenarios include the addition of tests approved in June 2008 and more speculative scenarios including the addition of a widespread (15% of the population) test roughly equivalent to the CYP2D6 drug metabolism test and the adoption of widespread genome sequencing. Test cost projections are made based upon actual costs within the MGL, costs to the province in the OOC program, and average test costs published by other genetic testing laboratories.

By comparing the expected number of tests and the expected costs of tests in various scenarios, I can apply my business criteria 4 – 6 from Section 4.1.1.1 on page 21 to establish which funding strategy is preferred.

5.1 Assumptions

Testing rates are constant across B.C., Canada, and the US.

Drug metabolizing rate tests will take place at the same relative rate as other hereditary disease tests.

Fifteen percent of consumers will request full genome sequencing. This includes those who have reason to believe they suffer from a hereditary genetic disease and those who may believe they have a metabolizing rate defect. Fifteen percent is the bottom end

of the lowest range of testing interest among women surveyed concerning breast cancer testing.[85] Many people at risk for genetic disorders do not choose to be tested.[86]

Privatized and outsourced funding methods merge together because a private MGL, Inc. will either expand to become a significant player in the genetic testing market (and will thus enjoy larger markets, access to capital, and economies of scale) or MGL, Inc. will be acquired by a major player in the genetic testing market (and will thus enjoy larger markets, access to capital, and economies of scale). Collectively the privatized and outsourced funding models may thus be referred to as privately owned.

5.1.1 Test Volume Projections

Table 8, below, shows various scenarios for test volume based upon historical and projected levels. All scenarios begin with actual values for 2007 and 2008 with projected values for 2009 based upon 2008 volume plus the planned added tests. From 2010 on, all values are projected either linearly (based upon 2007 – 2008 growth or 2008 – 2009 growth) or exponentially (based upon a flat 10% growth rate, the 2007 – 2008 volume ratio or the 2008 – 2009 volume ratio).

Scenarios show five-year testing volume should range between 11,000 and 35,000 samples, or between 2x and 7x current test volume.

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[85] Bottorff, Joan L., Ratner, Pamela A., Balneaves, Lynda G., Richardson, Chris G., McCullum, Mary, Hack, Tom et al. (2002). Women's Interest in Genetic Testing for Breast Cancer Risk: The Influence of Sociodemographics and Knowledge. *Cancer Epidemiology, Biomarkers & Prevention*, *11*, 89-95., pg 5.

86 Quaid, K. A. & Morris, M. (1993). Reluctance to undergo predictive testing: the case of Huntington disease. American Journal of Medical Genetics, 45(1), 41 - 45; and van der Steenstraten, I. M., Tibben, A., Roos, R. A. C., van de Kamp, J. J. P., & Niermeijer, M. F. (1994). Predictive Testing for Huntington Disease: Nonparticipants Compared with Participants in the Dutch Program. American Journal of Human Genetics, 55, 618 - 625.

	Linear Test Growth		Exponential Test Growth		
Year	07/08 diff	08/09 diff	10%	07/08 ratio	08/09 ratio
2007	4,050[87]	4,050[87]	4,050[87]	4,050[87]	4,050[87]
2008	5,100[87]	5,100[87]	5,100[87]	5,100[87]	5,100[87]
2009	7,500	7,500	7,500	7,500	7,500
2010	8,550	9,900	8,250	9,444	11,029
2011	9,600	12,300	9,075	11,893	16,220
2012	10,650	14,700	9,983	14,976	23,853
2013	11,700	17,100	10,981	18,859	35,077
growth	1,050	2,400	110%	126%	147%

Table 8: Projected Test Volumes (various scenarios)

5.1.2 Testing Rate Growth

Cases for Testing Rate Growth (TRG) include:

- B.C. population growth
- New market growth (assumes provision of for-profit testing outside B.C.)

Testing growth much higher than population growth requires that either currently tested syndromes become more prevalent (unlikely) or that doctors increase their testing frequency for current tests.

New market growth requires expansion into non-B.C. markets or the development of new tests. Table 6 and Table 7 in Section 4.5.2 on page 41, show potential market sizes for existing tests in Canada and the United States at various testing rates. The market potential of Canada and the United States is between 7.5x and 68x B.C. at current testing rates and as much as 250x current B.C. at higher testing rates.

[87] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services.

5.1.3 Test Type Growth

Cases for Test Type Growth (TTG) include:

- Addition of proposed tests
- Addition of a CYP2D6 drug metabolizing test
- Addition of full genome sequencing

	Proposed Tests	CYP2D6	Full Genome
B.C. Only	Demand growth is projected at an immediate 45%. Future growth is at population growth rate	Potential 7x demand growth	Potential 3.5x – 55x demand growth for full genome sequence
New Markets	Market expands 7.5x (Canada) or 68x (US)	Market expands 5.5x – 220x (Canada) or 500x – 2000x (US)	Market expands 29x – 430x (Canada) or 260x – 3900x (US)

Table 9: Demand / Market Growth for Representative New Genetic Tests

5.1.4 Test Prices

Prices vary widely by test and laboratory, largely due to degree of test complexity (number of exons per gene and variations possible) and automation (capital investment in laboratory equipment). Good information on the frequency of various tests is not available. Five laboratories (Molecular Diagnostics Laboratories[88], GeneDX[89], Massachusetts General[90], Johns Hopkins[91], and Tulane[92]) publish price lists, suggesting a mean market price for the average genetic test at $880.

[88] Fontaine, R. N. (2008). Customer Prices and CPT Codes 2006

[89] GeneDX. (2008). Current Price List. Retrieved 2008.07.14, http://www.genedx.com/pdf_files/current_price_list.pdf

[90] Massachusetts General Hospital. Neurogenetics DNA Diagnostic Laboratory Price List. Retrieved 2008.07.13, http://www.massgeneral.org/neuroDNAlab/price_list2_4-04.xls

[91] Johns Hopkins DNA Diagnostic Laboratory. Search for Tests by Syndrome Name. Retrieved 2008.07.14, http://www.hopkinsmedicine.org/dnadiagnostic/SyndromeSearch.htm

[92] Tulane Health Sciences Center, Matrix DNA Diagnostics

Lab	Mean Price
MDL	$352
GeneDX	$1,420
Mass General	$525
Johns Hopkins	$626
Tulane	$1,472
Overall	$879
2008 Actual Cost	$750
Actual / Mean	85%

Table 10: Mean Test Price

5.2 Current and Future Test Costs by Funding Method

Cost to the Ministry of Health for the average genetic test done at MGL (while publicly owned) is $179, including fixed and variable costs as well as the five-year amortized cost of the new equipment budgeted of $18/test[93]. Estimated cost to the Ministry of Health of the average genetic test done on the public market is $879 compared with actual 2008 cost of $750/test (85% of the mean cost). The lowest price of any genetic test on the public market is $231, more than 25% higher than the mean cost per test at MGL.

Funding Method	Cost/Test	New Equipment	Total
Current	$160	$18	$179
Per-Test	$160	$18	$179
Privately Owned (Average)	$879	$0	$879
Privately Owned (Adjusted)	$750	$0	$750
Privately Owned (Minimum)	$231	$0	$231

Table 11: Current Test Costs by Funding Method

[93] Assuming 6% interest over five years, $350K has a future value of $468K or almost $94K/year, divided among 5,100 tests/year = $18.

Because genetic testing and genomics are so fast changing, I expect that costs will change over time. Economies of scale will benefit privately owned funding methods more than publicly owned methods. Future prices and costs are speculative, so several cases are provided as a sensitivity analysis.

Scale economies without major technological change may reduce the price of testing to half or even one-quarter at agencies that can take advantage of them (privately owned funding models or public funding on a per-test basis). Because future test requirements (number of genes and exons) and equipment capability (level of automation for handling samples, automatic extraction of DNA) and price are not entirely predictable, rough scenarios must suffice. If demand were to grow by 10x, a linearly projected investment of up to $5 million would add a maximum of $27 to the cost of each test[94]. If demand were to grow by 100x or more, an exponential equipment investment in automation of up to $100 million would add a maximum of $53 to the cost of each test[95]. These numbers are illustrative only because machine capability and cost are changing rapidly and future test specifics are, as yet, unknown. If full-genome sequencing becomes cost-viable at $1000, individual tests after sequencing have a marginal cost approaching $0.[96]

[94] Assuming 6% interest over five years, $5M has a future value of $6.7M or $1.3M/year, divided among 51,000 tests/year = $27. This is a sensitivity analysis, not a prediction that handing 10x the samples would require a $5 million capital increase.

[95] Assuming 6% interest over five years, $100M has a future value of $134M or $27M/year, divided among 510,000 tests/year = $53. This is a sensitivity analysis, not a prediction that handing 100x the samples would require a $100 million capital increase.

[96] Because once the genome sequence is stored, further testing consists of electronically searching data instead of extracting and sequencing DNA.

Funding	Scale Econ.	Huge Scale Econ.	10x demand	100x demand
Current	X	X	X	X
Per-Test	$80	$40	$66	$93
Privately Owned	$440	$220	$27	$27

Table 12: Potential Future Test Costs by Funding Method and Scenario

Current funding methods (hospital global funding plus MSP OOC funding) do not allow for the current annual rate of test growth. They are completely inadequate for the level of expansion required to achieve large economies of scale or to make the potential investments necessary to handle 10x or 100x demand growth. Per-test funding methods allow for the smaller economies of scale and suggest that costs will drop over time until a very large investment in equipment is required to meet demand. However, publicly funded laboratories face difficulty obtaining capital for larger investments necessary to manage very large demand increases. Privately owned labs have the advantages of per-test funding as well as an advantage in making large investments because of their access to capital and the larger markets they serve. However, the higher cost per test of private labs makes them considerably less affordable than MGL while demand is relatively low.

Characteristics	Current Global Funding	Per-Test Funding	Privatization	Outsourcing
Sustainable?	No	Yes	Yes	Yes
Affordable?	$180/test More	$180/test More	$750/test Less	$750/test Less

Table 13: Funding Models Sustainability and Affordability with Current Test Price Information

5.3 Projected Costs by Funding Method

Figure 4, Table 14, and Table 15 show projected cost ranges over five years by funding scenario. If the best-case private funding scenario (assuming 10x demand and attendant cost reductions to $48/test) is correct, it minimizes total five-year costs at $2.6M – $5M. That scenario is highly speculative. Per-test funding shows the best conservative scenario with a total five-year funding range of $10M – $18M and 2013 annual budget of $2M – $6.25M.

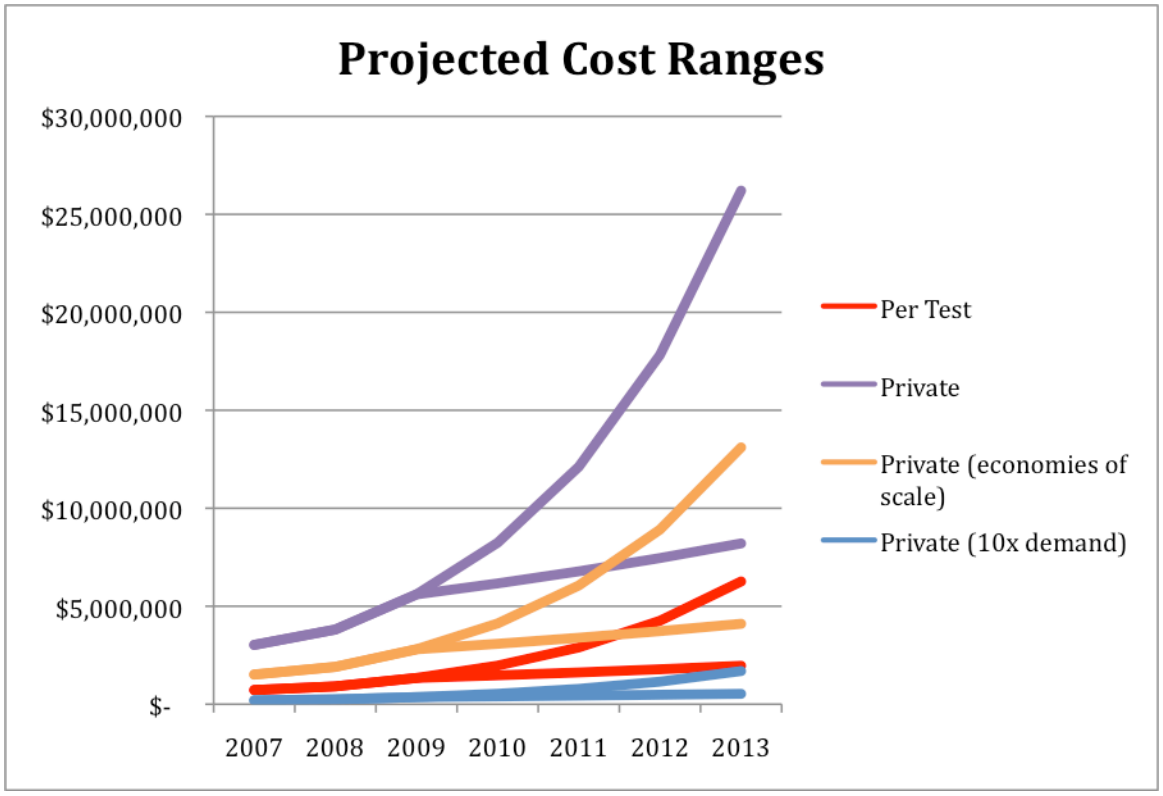

Figure 4: Projected Five-Year Cost Ranges by Funding Scenario

			# of Tests		
Total Cost by Scenario	**Low**	**High**	**low est.**	**high est.**	**Cost/Test**
Per-Test	$9,809	$18,360	54,938	102,829	$179
Private	$48,290	$90,386	54,938	102,829	$750
Private (scale)	$24,145	$45,193	54,938	102,829	$374
Private (10x)	$2,648	$4,957	54,938	102,829	$48

Table 14: Total Projected Five-Year Costs by Funding Scenario (costs in $000s)

		2007 (actual)	2008 (est)	2009	2010	2011	2012	2013
# of Tests	low est.	4050[97]	5100[97]	7500	8250	9075	9983	10981
	high est.	4050[97]	5100[97]	7500	11029	16220	23853	35077
Per-Test	low	$723	$911	$1,339	$1,473	$1,620	$1,782	$1,961
	high	$723	$911	$1,339	$1,969	$2,896	$4,259	$6,263
Privately Owned	low	$3,026	$3,810	$5,604	$6,164	$6,780	$7,458	$8,204
	high	$3,026	$3,810	$5,604	$8,241	$12,119	$17,821	$26,208
Privately Owned (scale economies)	low	$1,513	$1,905	$2,802	$3,082	$3,390	$3,729	$4,102
	high	$1,513	$1,905	$2,802	$4,120	$6,059	$8,911	$13,104
Privately Owned (10x demand)	low	$195	$246	$362	$398	$438	$481	$529
	high	$195	$246	$362	$532	$782	$1,150	$1,691

Table 15: Annual Testing Volume and Estimated Costs by Funding Method (costs in $000s)

5.4 Summary

All four funding strategies are sound business practices when properly applied. Criterion 4 thus provides no clear guidance for a decision.

[97] Casey, B. (2007). Toward a Sustainable Approach to Rare Genetic Disease Testing in British Columbia: Final Business Case for Clinical Services.

The funding strategies provide varying degrees of sustainability and affordability. Global funding is unsustainable in the long-term because of the increase in demand over time and the rapidly changing nature of genetic testing, both technologically and medically. Privatization and outsourcing are currently considerably less affordable than per-test funding and seem likely to remain so for the immediate future. Criterion 5 selects per-test funding as the most desirable.

Global funding and per-test funding maintain MGL as a publicly owned laboratory, while privatization and outsourcing do not. Keeping a useful resource in public hands better promotes a publicly funded health care system until and unless private ownership results in lower costs. Current costs are lower within the publicly owned system and seem likely to remain so until a large change in volume or capital investment is required. The future remains cloudy, but it seems unlikely that the $1000 genome will arrive in B.C. in the next five years, and so public ownership of MGL remains desirable and Criterion 6 selects global or per-test funding.

Per-test funding thus satisfies all three of the business criteria, while the other funding strategies satisfy roughly half (especially if global funding is considered unsound because of the rapidly changing nature of the genetic testing field). Private ownership fails primarily on grounds of affordability, and should be re-examined as prices change.

Criteria	Current Global Funding	Per-Test Funding	Privatization	Outsourcing
4. Sound Business Practice	+ (-)[98]	+	+	+
5. Sustainable & Affordable	- / +	+ / +	+ / -	+ / -
6. Public Funding	+	+	-	-
Total	(2 or 3) of 4	4 of 4	2 of 4	2 of 4

Table 16: Funding Strategies and Decision Criteria

[98] - if global funding is considered unsound business practice because of the rapid rate of change.

6 RECOMMENDATIONS

Recommendations flow from criteria and data. I have established a list of six criteria[99] and data on projected test volume and cost according by scenarios and funding strategies,[100] concluding with Table 16 showing which criteria are met by which funding strategy. Once data and analysis is presented in the right way, questions are answered at a glance.

6.1 Recommended Solution(s)

According to my business criteria 4 – 6, I am looking for a funding strategy that is a sound business practice, sustainable and affordable, and which promotes a publicly funded health care system. The differential between the various strategies is largely and most importantly in the areas of sustainability and affordability. It is by establishing which funding strategy is *both* sustainable and affordable that I find the preferred solution.

6.1.1 Budgeting and Funding

MGL should be funded by a per-test funding method on the basis of sustainability and current affordability along with the long-term benefit to the province of keeping a valuable medical resource in public hands. A per-test funding method balances flexibility and lower costs in the near future while maintaining the integrity of the publicly funded health system. When and if substantial ($5M or more) capital investment

[99] See Section 4.1.1.1 on page 21
[100] See Section 5, starting on page 42

is required (most likely to process expanded test volume), the province should reconsider the economics of privately owned solutions (privatizing MGL or outsourcing testing to private laboratories).

Whether MGL is funded by a pure per-test system or a mixed system where global funding is used for salaries (and adjusted upward annually to account for step increases and promotions) and the consumables budget is increased on a per-test basis depends upon the flexibility of the provincial accounting systems. Either will meet the basic requirement of ensuring appropriate funding for MGL through the near future. A mixed system is preferable from a psychological point of view.

A per-test budget over five years for MGL should total no more than $10M - $18M and will probably be less as per-test costs decline with learning curves and economies of scale.

6.1.2 Reducing OOC Payments

The 2008/9 budget adjustment for MGL begins the process of reducing OOC payments by developing new tests. The Ministry of Health should track tests sent out of province and develop in-province capability for the most common (and expensive) tests. To facilitate this process, some individual entity should be responsible for tracking all Ministry of Health funded genetic testing. The Director of MGL should receive monthly tracking data (type of tests, number of tests, and cost of tests) on all covered out-of-province genetic tests in order to plan test development.

6.2 Additional Steps

Although I have satisfied my basic project requirements to establish a balanced funding strategy for MGL over the next five or more years, to reduce referred out expenses, and to present and justify a sustainable and affordable approach to funding genetic testing in B.C., there are some additional items which have come to my attention as potential issues in the future implementation of this funding strategy. I present them here as issues to be explored, with prospective solutions where I have them.

6.2.1 Accounting and Tracking

MGL does not appear to have proper access to accounting and test tracking information. The Director should have instant access to basic financials (salaries, equipment costs, supply costs, etc.) and test cost (hours and supplies) and frequency, as well as information on number and frequency of referred-in tests and tests sent out of province (and whether they are sent out for reasons of capacity constraint or lack of an in-province test). This information is limited enough that a simple dashboard could almost certainly provide it.

To facilitate decisions about new tests and whether it is cost-effective to develop new tests and/or send tests out of province, all DNA based tests covered by MSP should be tracked and the information made available for analysis. A single storage database would be best, but aggregation is an acceptable alternative.

In order to adequately meet Criterion 4 (Sound Business Practices), better accounting and test tracking systems *must* be put in place. Because MGL is the primary

actor in B.C. genetic testing, it makes sense that the tracking function be added to MGL along with sufficient funding, equipment, and personnel to perform it.

6.2.2 Priorities

If MGL is allowed by the Ministry of Health to expand referred-in revenue by widely offering tests to out-of-province clients, priorities must be clearly established. Because MGL exists primarily for the benefit of the people and province of B.C., tests covered by the MSP must receive priority over uncovered or out-of-province tests. However, exceptions must be possible in cases where the uncovered or out-of-province tests are critical to immediate decisions about life-threatening conditions and the covered tests are not. The revenue from uncovered or out-of-province tests must not be allowed to displace covered tests, but there may be valid medical reasons to do so. A group of medical specialists and ethicists should be convened to establish priorities and the guidelines for exceptions.

6.2.3 Intellectual Property Issues

Because the value of genetic and genomic testing and treatment is represented by information, intellectual property law generally applies. Because we, as a society, consider ownership of some living things (humans, for example) to be morally repugnant, moral questions as to the legal ownership of genetic and genomic information arise. Because privacy, identity, and identity theft are increasingly interesting topics of legal and moral discussion, the ability of DNA to unequivocally identify a specific individual is both a legal and moral issue.

All these concerns are entwined with the questions of who owns genetic/genomic information and who should be allowed access to genetic/genomic information.

6.2.3.1 Patents on Genetic Discoveries

Although links between genome variations and disease are discoveries and not inventions, patents are nonetheless being issued by some jurisdictions covering the linkage between DNA variations and diseases. Individual jurisdictions are making *ad hoc* decisions about whether or not to honour those patents on either legal or humanitarian grounds. British Columbia should formally examine this question and resolve it so that clinicians, genetic counsellors, and the MGL have proper legal guidance.

6.2.3.2 Ownership of Genetic Information

Whether or not individuals own their genetic/genomic information remains unresolved. Once again, B.C. should formally examine this question and resolve it so that individuals and genetic researchers have proper legal guidance.

6.2.3.3 Privacy of Genomic Information

A full genome is the ultimate in personal information, but it cannot be easily used to identify an individual (that is, you cannot say, given a genome, that it belongs to X person). It can be used only to *confirm* the identity of an individual. As such, genome data deserves careful protection but not absolute protection. The question of exactly how much protection should be examined and established so that proper guidelines can be provided to laboratories and researchers.

6.2.3.4 Research Uses of Genomic Information

Genomic information has incredible potential for research. Predictive and diagnostic testing can be matured through data mining techniques if sufficient numbers of genomes and their accompanying phenotype information (in this case, diseases and syndromes) are made available for research. Full genome information can be used to conclusively identify an individual, which argues for careful protection of genomes. Aggregated genome information may open up new vistas in medical treatment, which argues for widespread research using genomic information. Genetic researchers, medical ethicists, legal experts, and computer security experts should determine how to balance protection with research.

6.2.4 MSP policy on Predictive genetic testing

MSP's policies currently specifically exclude predictive genetic testing for coverage. As genomic medicine matures, predictive testing will become more and more valuable. Knowing whether an individual has a heightened risk of diabetes, for instance, could save the province considerable expense by delaying the onset of disease through careful management. Such information will be available soon through predictive testing. B.C.'s Ministry of Health should examine the question of when predictive genetic testing will be cost-effective and reliable enough to cover, and put in place systems to approve specific predictive tests.

APPENDICES

Appendix A: Prevalence of Genetic Diseases

Establishing the prevalence or incidence of genetic diseases is not an exact science. Various sources report differing values, so all calculations have been done using a combination of low and high values. As appropriate, prevalence is relative to population, birth rate, or birth rate by sex. Many genetic disorders have varying prevalence by the geographic or genetic origin of the population. Wherever possible, I have used figures corresponding to Caucasian and Asian populations to reflect the genetic diversity of B.C. and especially Vancouver.

	Overall		Male Births		Female Births	
	Low	High	Low	High	Low	High
Original Menu						
□ Achondroplasia	1/40000	1/15000				
□ Angelman Syndrome	1/20000	1/10000				
□ Beta Thalassemia	1/114000	1/2600				
□ CADASIL	1/50000	1/50000				
□ Charcot Marie Tooth 1A	1/3300	1/3300				
□ Connexin 26-deafness	7/50000	7/50000				
□ Cystic Fibrosis	1/3900	1/3900				
□ Duchenne/Becker Muscular Dystrophy			1/5600			
□ Fragile X			1/8918	1/3600	1/6000	1/4000
□ Friedreich Ataxia	1/50000	1/25000				
□ Hemoglobin S,C	1/600	1/400				
□ Hemophilia A (F8)	1/10000	1/10000				
□ Hereditary Multiple Exostosis	1/100000	1/50000				
□ Hereditary Neuropathy w/Liability to Pressure Palsies	1/50000	1/20000				
□ Huntington Disease	1/1000000	7/100000				
□ Hypochondroplasia	1/28000	1/26000				
□ Myotonic Dystrophy I	1/100000	1/8000				
□ Prader-Willi Syndrome	1/15000	1/10000				
□ Spinalmuscular Atrophy	1/25000	1/25000				
□ X-Linked Ichthyosis-STS Deficiency	1/6000	1/6000				
Added June 2008						
Angelman	1/20000	1/10000				
Craniosynostosis	1/3000	1/2000				
HHT	1/5000	1/5000				
LQTS	1/10000	1/2500				
Marfan	1/5000	1/5000				
Neurofibromatosis 1	1/3000	1/3000				
Neurofibromatosis 2	1/25000	1/25000				
Rett	1/12500	1/12500				
Speculative Future Tests						
Crohn's	1/1000	1/544				
Sporadic ALS	3/100000	1/12500				
CYP2D6 Polymorphism	19/1000	77/1000				

Table 17: Known Prevalence of Genetic Diseases

Sources:

http://en.wikipedia.org/wiki/Achondroplasia#cite_note-pmid17879967-0

http://www.healthatoz.com/healthatoz/Atoz/common/standard/transform.jsp?requestURI=/healthatoz/Atoz/ency/thalassemia.jsp

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/AR_Sensorineural_Hearing_Loss_(DFNB1/Connexin_26)#Prevalence

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Cystic_Fibrosis_Carrier_Screening-2

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Duchenne_Muscular_Dystrophy-1

http://ovidsp.tx.ovid.com.proxy.lib.sfu.ca/spb/ovidweb.cgi?&S=GBMCFPFAMIDDMIMAMCHLAHOKEPPPAA00&Complete+Reference=S.sh.15.16.18|6|1

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Friedreich_Ataxia#Incidence_and_Carrier_Frequency

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Sickle_Cell_Anemia

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Hemophilia_and_Von_Willebrand_Disease#Hemophilia_A

http://en.wikibooks.org/wiki/Diagnostic_Radiology/Musculoskeletal_Imaging/Tumors_Basic/Hereditary_Multiple_Exostoses

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Hereditary_Neuropathy_with_Liability_to_Pressure_Palsies_(HNPP)_and_Lobular_Carcinoma_In_Situ_(LCIS)

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Achondroplasia-1#Incidence

http://www.myotonicdystrophy.org/General%20Information.htm

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Prader-Willi_Syndrome-2#Incidence

http://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Spinal_Muscular_Atrophy_(SMA1)#Incidence, http://en.wikipedia.org/wiki/Twin

Appendix B: C&W Molecular Genetics Test Menu

CW Molecular Genetics Test Menu
□ Achondroplasia
□ Alpha Thalassemia
□ Angelman Syndrome
□ Ashplex panel: tay sachs, canavan,
fanconi anemia, familial dysautonomia
□ Beta Thalassemia
□ CADASIL
□ Charcot Marie Tooth 1A
□ Chimerism-bone marrow
□ Connexin 26-deafness
□ Cystic Fibrosis
□ Duchenne/Becker Muscular Dystrophy
□ Dystonia (early-onset primary)
□ Familial Mediterranean Fever
□ Fascioscapulohumeral Dystrophy
□ Fragile X
□ Friedreich Ataxia
□ HFE 1 Hemochromatosis
□ Hemoglobin S,C
□ Hemolytic Disease Newborn - Rh, Kidd, Kell, Duffy
□ Hemophilia A (F8)
□ Hereditary Multiple Exostosis
□ Hereditary Neuropathy with Liability to Pressure Palsies
□ Heterotaxy (X-Linked)
□ Huntington Disease
□ Hyperkalemic Periodic Paralysis
□ Hypokalemic Periodic Paralysis
□ Hypochondroplasia
□ Linkage Analysis-CF, F8, DMD
□ Myotonic Dystrophy (Type 1)
□ Non-Ketotic Hyperglycinemia

□ Oculopharyngeal Muscular Dystrophy
□ Platelet-Specific Alloantigen (P1A1)
□ Prader-Willi Syndrome
□ Progressive Myoclonus Epilepsy
□ Spinal Muscular Atrophy
□ SpinoBulbar Muscular Atrophy
□ Spinocerebellar Ataxia
□ Thanatophoric Dysplasia
□ Transthyretin Amyloidosis
□ Uniparental Disomy 6, 7, 14, 15
□ X-Linked Ichthyosis-STS Deficiency
□ Zygosity
□ DNA extraction only

Table 18: C & W Molecular Genetics Test Menu

Appendix C: C&W Cytogenetics Test Menu

CW Cytogenetics Test Menu
Karyotype Analysis
□ Karyotype Analysis of Peripheral Blood
□ Karyotype Analysis of Amniotic Fluid
□ Karyotype Analysis of CVS
□ Karyotype Analysis of Pediatric Bone Marrow
□ Karyotype Analysis of Pediatric Tumours
□ Karyotype Analysis of Tissue
□ Karyotype Analysis of Products of Conception
□ Karyotype Analysis of malignant effusion
□ Karyotype Analysis of Fetal Blood
□ High Resolution Karyotype Analysis
□ Chromosomal Mosaicism
□ Chromosomal Breakage Studies
□ Chromosomal Special Banding
□ Chromosomal Special Staining
□ Culture for Biochemical or Molecular Analysis
FISH Analysis
□ Uncultured Amniocyte Aneuploidy Screen
□ XX/XY FISH Chimerism Testing
□ CGH
□ Interphase Chromosome Enumeration
□ Interphase Sex Chromosome Enumeration
□ MFISH
□ Sub-Telomeric Region Testing
Oncology Tests
□ 13q14 (RB1) Testing
□ AML1/ETO Fusion Testing
□ BCR/ABL Fusion Testing
□ CBFB Rearrangement Testing
□ Chromosomes 4,10 and 17 Enumeration
□ ETV6 Rearrangement Testing

□ EWSR1 Rearrangement Testing
□ FKHR Rearrangement Testing
□ Chromosome 5 Monosomy/ Deletion 5q Testing
□ Chromosome 7 Monosomy/Deletion Testing
□ MLL Rearrangement Testing
□ N-MYC Amplification Testing
□ p16 Gene Deletion Testing
□ TEL/AML1 Fusion Testing
Microduplication/deletion Testing
□ 21q22.2 Region Testing
□ 22q11.2 and 22q13.3 Region Testing
□ 1p36 Deletion Testing
□ Alagille Syndrome Testing
□ Angelman Syndrome Testing
□ Cri-du-Chat Syndrome Testing
□ Kallmann Syndrome Testing
□ Miller-Dieker Syndrome Testing
□ NF1 Gene Deletion Testing
□ PAX6 Gene DeletionTesting
□ Prader Willi Syndrome Testing
□ Smith-Magenis Syndrome Testing
□ SNRPN Deletion Testing
□ SNRPN Duplication/Deletion Testing
□ Sotos Syndrome Testing
□ SRY Gene Testing
□ Steroid Sulfatase Deficiency Testing
□ Willliams Syndrome Region Testing
□ Wolf-Hirschhorn Syndrome Testing

Table 19: C&W Cytogenetics Test Menu

Appendix D: C&W Biochemical Genetics Mitochondrial DNA Test Menu

Biochemical GeneticsTest Menu: Mitochondrial DNA Testing
□ LHON Leber's mtDNA Mutation (11778,14484,3460)
□ LHON-Dystonia mtDNA mutation (14459)
□ Long PCR mtDNA Deletion
□ MELAS mtDNA Mutation (3271 & 3243)
□ MERRF mtDNA Mutation (8344)
□ NARP mtDNA Mutation (8993)
□ Southern Blot mtDNA Deletion

Table 20: C&W Biochemical Genetics Mitochondrial DNA Test Menu

Appendix E: C&W Biochemical Genetics Test Menu

Biochemical GeneticsTest Menu: Inborn Errors in Metabolism Assays
□ Acylcarnitine Profile, Serum
□ Acylcarnitine Profile, blood dot card
□ Alpha-Galactosidase (Fabry),WBC
□ Alpha-Iduronidase (Hurler),WBC
□ Amino Acids, Urine
□ Amino Acids,CSF
□ Amino Acids,Plasma
□ Aryl Sulfatase A (Metachromatic leukodystrophy),WBC
□ Aryl Sulfatase C (X-linked ichthyosis),WBC
□ Beta-Galactosidase (GM1),WBC
□ Beta-Glucosidase (Gaucher),WBC
□ Biotinidase,Serum
□ Carnitine,Serum
□ Cell Culture (skin, fibroblast propagation, AFC)
□ CPT1a,P479L Variant, Blood Dot Card
□ DNA Extraction (blood, tissue, fibroblast)
□ Galactocerebrosidase (Krabbe) Enzyme,WBC
□ Hexosaminidase (Tay-Sachs disease/carrier status),Serum
□ Mitochondrial Respiratory Chain Enzymes, muscle (Complexes I, II, IV & citrate synthase)
□ Mucopolysaccharide,Urine
□ Oligosaccharides,urine
□ Quantitative organic Acids, Urine
□ Sphingomyelinase (Niemann-Pick A/B) ,WBC
□ Transferin Isoelectric focusing, serum
□ Urine Purines & Pyrimidines (includes creatine, creatinine and GAA to investigate for creatine synthetic and transporter defects)
□ WBC Preparation, whole blood
□ Alanine-Glyoxylate Aminotransferase (Hyperoxaluria type I),Liver
□ Alpha-Fucosidase,WBC
□ Alpha-Mannosidase,WBC
□ Aryl Sulfatase B (MPS VI),WBC
□ Beta-Glucuronidase (MPS VII),WBC

□ Beta-Mannosidase,WBC
□ Iduronate sulfatase (Hunter) enzyme, Serum
□ Hexosaminidase,WBC
□ Mycoplasma, culture supernatant
□ San Filippo Type B (MPS III),Serum
□ San Filippo Type C (MPS III) ,WBC
□ Sulfocysteine, Urine

Table 21: C&W Biochemical Genetics Test Menu

Appendix F: June 2008 Tests to be Added

- Angelman Syndrome
- Craniosyostosis Syndromes
- Hereditary Hemorrhagic Telangiectasia (HHT)
- Infantile Epilepsy
- Long QT Syndrome
- Margan Syndrome
- Neurofibromatosis Type 1
- Neurofibromatosis Type 2
- Pendred Syndrome
- Periodic Fever Syndrome
- Rett Syndromes
- RYR2-related Cardiac Disease

Appendix G: Mission Statements and Goals

At the program level:

The Provincial Medical Genetics Program is committed to providing high quality genetic health care to residents of B.C., while participating in and contributing actively to research and education in the field of Medical Genetics. We seek to fulfill this role in accordance with the missions of the Children's and Women's Heath Centre of B.C., the Provincial Health Services Authority and the University of British Columbia.[101]

At the department level:

The Department of Pathology & Laboratory Medicine at Children's & Women's Health Centre of British Columbia provides critical tertiary-care diagnostic consultation and services on which vital decisions are made in the care of patients. It is also dedicated to research and teaching in paediatric and obstetric laboratory medicine through the application of specialized consultative expertise in diagnosis, screening and monitoring.[102]

At the provincial level, the goals and objectives of the Ministry of Health include:

Goal 1: Improved Health and Wellness for British Columbians…

Objective 1: Individuals are supported in their efforts to stay healthy and make healthy lifestyle choices…

Objective 2: Protection of the public from preventable disease, illness and injury…

Goal 2: High Quality Patient Care…

Objective 2: Patient-centred care tailored to meet the specific health needs of patients and specific patient groups…

Goal 3: A Sustainable, Affordable, Publicly Funded Health System…

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[101] B.C. Women's Hospital & Health Centre. (2008). Medical Genetics. Retrieved 2008.06.27

[102] U.B.C. Department of Pathology & Laboratory Medicine. (2008). Research Programs. Retrieved 2008.07.04, http://www.pathology.ubc.ca/html/BCCH.html

Objective 4: Sound business practices to manage within the available budget while meeting the priority needs of the population.[103]

[103] B.C. Ministry of Health. (2006). 2006/7 - 2008/9 Service Plan. Retrieved 2008.06.27,, pp 20 - 33

Appendix H: MSC Guidelines on Genetic Tests

6. Laboratory and Medical Imaging Tests

a) Funding will not be provided for experimental or developmental laboratory and medical imaging tests where the efficacy of such services is not known.

b) In order for proven laboratory and medical imaging tests to be funded out of country, all diagnostic avenues in B.C. and Canada must have been exhausted.

c) If laboratory and medical imaging tests are not available in Canada, but are of proven value, prior approval will be given by MSP only if the result of the test would significantly alter the management of the beneficiary's condition. In limited circumstances, US Food and Drug Administration (FDA) approved laboratory and medical imaging processes may be deemed medically necessary if promising outcomes have been substantiated by reputable clinical trials, beyond Phase III published in peer reviewed medical literature.

d) It is the responsibility of the appropriate medical specialist making application on behalf of a beneficiary for prior approval of funding for out of country laboratory and medical imaging tests to provide MSP with peer reviewed medical literature about the laboratory and medical imaging tests requested.

7. Genetic Tests

Predictive genetic testing is not an insured service for beneficiaries of MSP in British Columbia, and is therefore not funded when performed outside of Canada. Genetic testing is an insured service in British Columbia only when it is medically necessary to the medical management of the beneficiary's condition.[104]

[104] Medical Services Commission. (2006). Medical Services Commission Out of Province and Out of Country Medical Care Guidelines for Funding Approval. Retrieved 2008.07.05, http://www.health.gov.bc.ca/msp/infoben/ooc_funding_guidelines.pdf, pg 5.

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