

Private Genetic Testing in Canada: A Summary

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A. Introduction

The last few years have seen a rapid growth in the discovery of genes associated with hereditary disease or susceptibility, and an accompanying rise in the number of genetic tests becoming available. While most Canadians likely still access genetic testing through the public system, there is also a small developing private market. As more tests become available for conditions of greater general incidence, e.g., heart disease or the common cancers, there is every likelihood that the market will expand.¹

Genetic testing will provide information about individual risk for disease that may permit people to make lifestyle changes to reduce risk, make career planning decisions, or take prophylactic measures, and for those already affected, may allow for individualized therapy.² However, private provision may lead to premature access to unproven technologies, while marketing might minimize the uncertainties associated with testing,³ support a reductionist view of disease and disability,⁴ and inflate public anxiety to increase demand.⁵ A private market also has serious implications for the Canadian public health care system.⁶ Private companies will profit from overhead services without contributing to funding or development,⁷ draw experienced professionals from the public to the private sector thereby reducing quality and lengthening waitlists, and increase demand for counselling and medical treatments. Despite these concerns, we currently do not have good mechanisms for controlling the development of a private market. More generally, we also lack good criteria for determining what health care services should or should not receive public funding – the determination of health care funding is the result of an *ad hoc* rather than a rational decision making process.

In 1999, a small interdisciplinary group of academics, clinicians, business representatives and a member of the public met to discuss the role of privately financed genetic

testing in Canada. Out of this workshop was developed a paper by Caulfield, Burgess, Williams-Jones, *et al.*,⁸ that outlined an analytic framework of six thresholds to help support more rationalized decision making in the funding of genetic services. The first four thresholds determine whether anyone should receive the service: whether the test morally acceptable; whether it identifies a genetic factor; whether the test useful; and whether the test harmful. The last two thresholds consider whether the service should be financed with public monies or be available for private purchase. The goal here is to provide a summary of the larger paper that highlights its key points and recommendations.

B. Thresholds 1-4: Which Tests are Acceptable?

At any point in the development of a genetic test, we may decide as a society that the goals or consequences are morally objectionable and thus restricted access or prohibition of the test is warranted. We must also consider the adverse implications, for example, on potentially useful research or on economic development. This first threshold evokes questions of which values will be considered relevant, and how and by whom they will be expressed. What is the commercial influence on the way economic, technology and public policy are developed? Is public input valued only if it is “educated” about the technical aspects of genetic testing? Adequately addressing these questions will require further research and development of effective public participation mechanisms. It should also be noted that regardless of whether a particular genetic test is prohibited locally, Canadians can access testing through international sources.⁹ But this does not eliminate the value of government regulation prohibiting specific tests. Such a stance can be an important statement of national values that can strongly influence consumer preferences.

Genetic tests should meet some minimum standard of efficacy, such that a test at least detects what it claims to



detect. This criteria ensures that tests that have no efficacy are not marketed to health care professionals and patients. Yet in Canada (and the U.S. as well), there is currently a lack of such regulations. While laboratory quality is governed by provincial accreditation schemes, and the development and marketing of test kits is overseen by federal agencies, the majority of genetic tests which are offered as in-house laboratory services are not formally reviewed.¹⁰ A regulatory hole exists which could be and needs to be filled, but given that patent law permits the establishment of single source providers (e.g., with a laboratory in another country), it may prove difficult for any one jurisdiction to comprehensively regulate quality.¹¹

Once a test is proven effective, the usefulness of the information it provides must be evaluated. Is there a clear association between the genetic variation and the phenotype for a given population, and what are the harms and benefits? Benefits may include directing treatment, avoiding unnecessary screening, preventative strategies, anxiety reduction, and information for personal and/or reproductive planning. Promoting autonomy and individual choice may further support access to any accurate information about genetic risk that can be provided without undue harm.¹²

The harms associated with a particular genetic test must be outweighed by the benefits. Knowledge of genetic risk when there is no prevention or treatment will be harmful to some, resulting in anxiety and depression, suicide, or inappropriate lifestyle changes or prophylactic surgery. Harm may also result from responses of third parties and social systems, as in the case of stigmatization, insurance and employment discrimination, or altered family relations. Some of these harms might be eliminated through regulatory initiatives – some tests may warrant prohibition via professional or government regulation, while others will be permissible so long as there is appropriate public education, gatekeeping and support for gatekeepers.

Given the amount of information available about genetics and the variety of sources of information (e.g., the Internet), it will also be crucial to improve quality and accessibility so that consumers receive the information they need to better evaluate the harms, benefits, and quality of genetic testing. Since accurate information is essential for both the public and clinicians, it is reasonable to regulate the marketing and advertising of genetic services to require truth in advertising,

not unlike current demands placed on the pharmaceutical industry.¹³ Thresholds one to four presume that a governance mechanism similar to drug and device regulatory agencies is required.

C. Thresholds 5 & 6: Public or Private Access?

Tests that have passed the previous thresholds need to be evaluated to see whether they merit public financing. The key ethical issue at this point is whether it is acceptable that people who want and can benefit from testing, but who cannot afford it, are denied access. Ideally, beneficial tests that can be provided at a reasonable cost should be

publicly funded. Equal access to effective health care independent of economic status is a central tenet of the Canadian public health insurance system. Unfortunately, health care budgets are restricted without consideration for what services should be covered. Furthermore, decisions about coverage are influenced by professional or institutional practices; public demand influenced by the media, interest groups, or marketing pressure; researcher interest and clinician enthusiasm.¹⁴ Some services will be insured without adequate demonstration of benefit, while others that are clearly beneficial will not be insured. Decisions about genetic testing will be made in this *ad hoc* fashion.

The issue of rationalized health resource allocation is unlikely to be soon resolved, but evaluating the public and private availability of genetic testing in a consistent and justified manner might nonetheless be helpful for the larger discussion. Thus, after determining that a test is ethical, useful, and relatively safe, we must consider the worth of the service relative to other health care goods. Public and provider consultation, and an evaluation of the wider social and structural impacts will be essential in determining the relative value of funding genetic tests. Tests that have passed the first four thresholds but not the fifth will require re-evaluation as understanding about their accuracy or benefits develops. In the current policy environment, however, some genetic tests at this stage will be rejected for public funding not because they lack accuracy or benefits,

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but because health care funding is inadequate to fund all cost-worthy services.

Assuming that some genetic tests will meet the first four thresholds but not receive public funding, useful and safe genetic tests may be available only through the private sector. Some publicly funded tests will be under-funded, resulting in lengthy wait lists that further develop a market for genetic testing, or some people may prefer private purchase to ensure confidentiality. However, there are significant economic and ethical problems associated with the co-existence of public and private testing. Private tests and the related profits are publicly subsidized through education, research and supportive services; the quality of the public system may be reduced if health professionals move to the private system;¹⁵ and private testing, by reducing pressure to increase access, may lengthen wait lists in the public system. Ideally, there would be more adequate health care funding of the public system and more rigorous evaluation of services for inclusion, but this is unlikely to happen in the near future. While equitable distribution of health care resources is high on the social agenda,¹⁶ Canada is also a country that places great importance on individual autonomy and consumer choice. The tension created by these two social norms is emphasized in the context of genetic testing.

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D. Gatekeepers to Genetic Testing

One mechanism for regulating genetic tests is to require access through an appropriate gatekeeper. The level of expertise needed for gatekeepers will vary between tests, so for some a family physician or genetic counsellor may be sufficient, others may require a medical specialist, and a few tests may be made privately available with the assurance of accurate information. The norms surrounding non-directive counselling and informed consent would apply to all gatekeepers, and would include a discussion of the social consequences of testing as well as health related information. However, there are concerns that in the private sector, gatekeepers may be unduly influenced by profit incentives to favour testing, thereby creating conflict of interest.¹⁷ It is therefore crucial to be explicit and transparent about the possible conflict, and to consider mechanisms (e.g., guidelines, consensus statements and professional standards of practice) that can enhance gatekeeper independence. A robust education program is also needed to support physicians in this role,¹⁸ but the rapid development

of genetic technologies will make it difficult for many to effectively weigh the benefits and harms of all new tests. A regulatory regime or oversight mechanism that supports health professionals' decision making abilities is therefore required to assess evidence of test accuracy and net benefit.

Another approach to controlling the potential harms associated with private access is to fund public genetic counselling. Genetic counselling may be an effective and economical means of addressing anxiety associated with the perception of genetic

risk. Regardless of whether the test is publicly or privately accessed, it might be reasonable to include partial or full support for counselling. However, it could be argued that funding genetic counselling would allow the private system to unjustly benefit from public system personnel and infrastructure. There might nonetheless be creative ways of ensuring access to pre- and post-test counselling, e.g., private genetic counselling services or requiring licensing agreements whereby private companies subsidize publicly funded counselling.¹⁹

If the goal is to ensure safe, respectful, and equitable access to genetic testing, it will be essential to engage in open and pragmatic discussions about the nature of genetic services and their role in health care. The issue of private genetic testing is part of a larger picture – the moral and political task of providing equitable access to health care services and promoting health in the context of reflections on “how best to bring capital, morality, and knowledge into a productive and ethical relationship.”²⁰

E Conclusion

Debates about public versus private funding of genetic testing raise a host of social, ethical and policy issues that tie into the larger question of health care resource allocation. Under a rational and well-funded health care system, all services proven to have beneficial health effects would be supported, and genetic tests would simply need to meet this criteria. But the current system in Canada is less than rational and is insufficiently funded to cover all effective services. There will be services that are either included but underfunded, or useful but not funded. It is within this complex context that Caulfield, Burgess, Williams-Jones, *et al.* propose a series of thresholds to help determine whether a genetic test is morally appropriate, effective, safe,

efficient, and appropriate for public funding, and whether private purchase poses special problems that require further regulation.²¹ The hope is that this approach, while highlighting areas in need of further research, helps to address some of the problems associated with new genetic technologies, without reducing the pressure to evaluate the nature and funding of the larger health care system.

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