



DIRECT-TO-CONSUMER GENETIC TESTING

See also [CMA Policy PD17-05 Direct-to-Consumer Genetic Testing](#)

Some direct-to-consumer (DTC) genetic tests, such as “compatibility testing” for online dating, are purely recreational. Other tests, however, are marketed both as being for recreational use and as producing results that are useful to the management of one’s health. This document concerns this second category of tests. The characteristics of these tests differ widely, and some of the companies that offer them clearly state that they do not guarantee the validity and reliability of their tests. As of January 2016, 246 companies offered some form of DNA test online.¹ Many DTC genetic tests have started to penetrate the Canadian market, especially after the U.S. Food and Drug Administration issued a warning letter instructing some companies in the U.S. to cease providing unreliable health information that could potentially lead consumers to make misinformed decisions about their health, which caused some of these companies to seek out alternative markets.²

The increasing availability of DTC genetic tests in Canada presents several challenges, as the predictive value of most of the DTC genetic tests currently on the market is very low. Moreover, there is no standard model for the delivery and interpretation of the results of these tests. Greater regulatory guidance and protection is needed to ensure that individuals who choose to submit samples to DTC genetic testing companies are not adversely affected by information that is not necessarily predictive or even accurate.

Survey research indicates that the general public is overwhelmingly interested in genetic testing technologies.³ Researchers predict that an increasing number of individuals will use DTC genetic testing as testing technologies continue to become more affordable and efficient.³ Since genetic issues tend to cross medical specialties, it often falls on primary care physicians to understand the role of genetics in clinical care.⁴ In fact, genetic testing companies often direct patients to discuss their results with their primary care physician.⁵ Patients not only seek out their primary care providers to discuss their genetic test results and obtain appropriate follow-up but also expect them to be able to answer questions about personal genome test results.⁶ Despite these expectations, health professionals’ awareness and knowledge of DTC genetic tests remains low.⁷

Although DTC genetic tests are marketed under similar names, the genetic tests available in Canada have very different characteristics. Three types of tests are offered: (1) single-nucleotide polymorphism (SNPs) analysis, which assesses an individual’s risk for common

multifactorial diseases (e.g., diabetes, myocardial infarction), (2) targeted mutation analysis and (3) sequencing. Some are ordered directly by the consumer while others are pre-ordered by the consumer and the order is co-signed by a physician (the patient's physician or a physician who has never met the patient and whose services are provided by the company). SNP testing assesses for a number of genetic variants that are common in the general population and that have been identified in association studies to modify (increase or decrease) the risk of a given disease.

Some DTC genetic testing companies explicitly state in their terms of service that they do not guarantee the accuracy or reliability of the test. This is due in part to deficiencies in the science underlying the tests and their interpretation. For example, the interpretation of SNPs analysis for common multifactorial diseases can only be as good as the science behind it. The scientific community has a long way to go before it will have identified all of the significant genetic risk factors and protective factors for these diseases. Because of this, a given consumer could receive greatly divergent risk interpretations.³ In the case of targeted mutation analysis and sequencing, the specific panels offered by DTC genetic testing companies may not include all of the clinically relevant genes and mutations. This could result in a consumer receiving harmful false reassurance. Test results may include information on genetic changes that are only weakly associated with disease, leading to undue anxiety. As such, the clinical and health value of DTC genetic testing continues to be debated despite consumer uptake of, and enthusiasm for, DTC genetic testing offered online.

Currently, most DTC genetic testing services exist in regulatory limbo, benefiting from laws that tend to lag behind technological innovation. Questions about access to the information yielded by these tests have emerged as a particular concern. For some companies, an important part of the business model is to sell consumers' DNA along with the clinical information that the consumers provide via their interactive websites. Most Canadians are unaware of this: they pay for a test and do not expect that their data will later be sold.

ISSUES ARISING IN CLINICAL CONTEXTS

Studies have shown that physicians see a number of benefits with DTC genetic testing, but they also have concerns. The benefits physicians have identified include convenience, promotion of preventive medicine and the provision of personalized services.⁵ They are concerned about the reliability of test results, the provision of adequate information/counselling, patient anxiety if the results are misunderstood, inappropriateness of advertising, discrimination with respect to employment and insurance, the possible spread of beliefs such as genetic determinism, and the inappropriate disclosure of patients' genetic information.⁵

The following sections will address primary concerns identified by research and in practice.

1. Patient privacy

Privacy is one of the top concerns of the general public about genetic testing.⁸ According to a 2010 report commissioned by the Office of the Privacy Commissioner of Canada, approximately

60% of patients indicated that privacy and discrimination fears would influence their decision to undergo genetic testing.⁹ The average Canadian consumer is not adequately informed that an important part of the business model of many DTC genetic testing companies is to build and sell their database of phenotypic information and DNA accumulated from their clients to third parties, such as biobanks or pharmaceutical companies.

1.1 Informed consent

The increasing quantity, complexity and diversity of DTC genetic testing services pose challenges for informed consent because both specific and generic models do not meet ethical standards when applied to this type of service.¹⁰ Many companies bind their consumers to contracts that are activated once the website is viewed, a practice that challenges the adequacy of consent, as it is common for people to view a website without reading or even seeing its terms of reference.¹ Consumers who present to genetic clinics tend not to question the validity of the results they have received from DTC genetic testing,¹¹ which can be interpreted as an indication that consumers give their consent without reading or understanding the disclaimers made by the companies.¹¹ Physicians are concerned that this lack of informed consent could compromise the confidentiality of personal health information, encourage requests for unnecessary medical tests and potentially cause distress to patients.

1.2 Insurance

The insurance industry is of particular concern in the context of privacy and DTC genetic tests. A study of patients' perceptions of DTC genetic tests found that participants were concerned that genetic results could affect their health insurance premiums or lead to denial of coverage.¹² Private insurance is fundamentally rooted in the practice of discriminating between clients on the basis of risk. While insurers have generally been entitled to request genetic information in the form of family history, to access medical files and to conduct medical tests,¹³ consumers have expressed the view that the rules governing access to genetic information should be stricter than for access to other forms of personal information.³

While there are studies that report cases of genetic discrimination, it is often unclear whether such treatment is perceived or actual.¹⁴ Thus, the consequences of genetic testing remain uncertain. Of particular concern is the potential for discrimination on the basis of results that may not be accurate and/or reliable. Although there is presently no evidence of widespread use of genetic testing by insurance companies,¹⁴ experts agree that in the next 10 years public acceptance of the use of information from genetic testing will increase and it will become possible to more accurately interpret data from genetic tests (K. Boycott, J. Davies and K. Morin, CIHR Café Scientifique, unpublished remarks), threatening to alter the currently limited role that genetic testing plays in insurance company decision-making. Before policy-makers tackle the potential issues related to the use of DTC genetic testing, it is imperative that they start at ground level and explore options to regulate insurance companies' access to such tests.

2. Patient response

2.1 Interpretation of results and changes in behaviour

Proponents of DTC genetic testing point to the potential for patients to make positive changes to

their health as a result of learning about their genetic susceptibility to certain diseases. Findings of studies in this area, however, are inconsistent to date. While some studies have reported that there are some behaviour changes, it is important to keep in mind that early adopters of these services are likely to also be among those most motivated to make health-related changes.¹⁵ Recent evidence suggests the opposite response: the general population has a tendency to decrease healthy practices upon learning about a lower health risk, and they do not increase healthy practices when they learn that they have an increased health risk.¹⁷ Indeed, patients may make poorer health decisions if they are under the impression that they are not at risk for developing a certain disease; for example, they may avoid routine screening for breast or prostate cancer, or they may not follow exercise and diet advice.¹⁶

These variations in behaviour can be largely attributed to the fact that there is an overarching risk that patients will misinterpret the data they receive from the testing companies. The problem with susceptibility tests in the context of DTC genetic testing is not only that the test results may cause psychological or physical harm but also that there is a possibility that patients will over-interpret their disease risk.¹⁰ Without expert guidance, the patient may not be able to evaluate their test results accurately enough to make informed health decisions.¹⁶ There is very little evidence to suggest that receipt of a DTC genetic test result produces sustained behavioural change.¹⁷ In fact, studies on psychological theories related to motivation do not consider disease risk information a useful tool for motivating patients to change their behaviour.¹⁷ Therefore, while receipt of DTC genetic test results may encourage patients to see their family physician and possibly undergo further consultation, the health care resources invested in interpreting results with limited clinical validity may not produce sustained behavioural changes, good or bad.

3. Resource allocation

One of the stated goals of personalized medicine is to save health care systems money by facilitating the use of fewer but more effective treatments.¹⁸ However, greater demand for genetic testing, whether public or private, could produce the opposite effect: consumption of health care resources may increase as patients consult with their regular physician about results they obtained through a DTC company.¹⁸ Furthermore, physicians who are presented with DTC genetic test results by their patients have a legal and ethical obligation to do their due diligence and carry out a complete, clinically valid investigation, which may ultimately negate the cost savings that personalized medicine is expected to produce.¹⁸

Patients who participate in DTC genetic testing are likely to drive up the utilization of health care providers, as they seek out their primary care provider to discuss their results and they obtain follow-up care from a genetic counsellor.^{19,5} At least one study has suggested that there is an expectation that physicians will help patients to interpret their DTC genetic test results, and DTC genetic testing companies frequently direct patients to discuss their results with their physicians before acting upon their testing information.⁵ Consequently, the responsibility falls on primary care providers to discuss this technology with their patients.⁵

Primary care providers, however, believe that genetic specialists are the most appropriate providers of counselling for DTC genetic tests.¹⁶ While they acknowledge the benefits of DTC

genetic tests, including the potential for test results to encourage patients to be more involved in their care and take responsibility for their health, they also agree that test results may encourage patients to seek unnecessary and potentially expensive follow-up tests.¹⁶ As a result, additional health care resources may be required to cope with the increased demand for medical follow-up.²⁰

4. Physician education

Although DTC genetic testing companies have been around since the early 2000s, levels of awareness among health care professionals vary, and knowledge and understanding of the services generally remain low.²¹ Research suggests that few physicians feel they have the necessary training and knowledge in genomics to provide adequate care in this area.¹⁹ A perceived lack of clinical utility appears to be a barrier to learning more about DTC genetic testing.⁶ Increased genetics training and awareness may allow physicians to better appreciate the complex issues involved and help them to better counsel patients who seek follow-up for their DTC genetic test results.

4.1 Topics that physicians want to learn about

Most physicians are concerned about the privacy implications of DTC genetic testing, specifically health insurance and employment discrimination, which may affect their patients who present with a DTC genetic test.⁵ Therefore, important discussion points to include in a physician education program would be information on the risks of insurance and employment discrimination, legislation currently in place to protect against genetic discrimination, and guidelines for managing risk.⁶ Given the ease with which patients can access DTC genetic testing, it is essential to provide health professionals with appropriate education on the potential benefits and risks of DTC genetic testing and help them develop an approach to interpreting the results of such testing, so that they can protect their patients from harm and arrange follow-up appropriately.²¹

5. Legislative landscape in Canada

Before May 2017, Canada did not have a law to specifically protect against genetic discrimination. Existing human rights and privacy law could only be ambiguously and tenuously applied to DTC genetic testing issues, including genetic discrimination and information collection, use and disclosure.¹⁴ The laws that regulate medical devices, such as the *Food and Drugs Act*, did not clearly apply in the context of DTC genetic tests either,² because consumers are not purchasing genetic testing kits but rather they are purchasing testing services, which fall outside the scope of that legislation.²² As a result, there was limited evidence to form the regulations necessary to ensure the validity and utility of these tests.

Fortunately, on May 4, 2017, Bill S-201 (hereinafter termed S-201), *An Act to prohibit and prevent genetic discrimination*,²³ received Royal Assent and will soon become law. S-201 provides a basis for the creation of regulations concerning the validity and utility of DTC genetic tests. The bill prohibits the requirement that an individual submit to genetic testing or disclose the results of genetic tests in order to receive goods or services or in order to enter into or continue a

contract or agreement, and it prohibits submission to genetic testing or disclosure of test results from being used as the basis of any specific conditions in a contract or agreement. S-201 amends the *Canada Labour Code* to protect employees from being required to undergo or disclose the results of genetic testing and amends the *Canadian Human Rights Act* to prohibit discrimination on the grounds of genetic characteristics.²³ Legislation at a provincial level, however, may still be required. Private Member's Bill 127, *An Act to amend the Human Rights Code with respect to genetic characteristics*,²⁴ was presented to the Legislative Assembly of the Province of Ontario in 2013 but did not move past the first reading.

Federal and provincial privacy legislation (such as the federal *Personal Information Protection and Electronic Documents Act*, or PIPEDA) also plays a role in protecting against genetic discrimination by requiring an individual to consent to the collection, use or disclosure of personal information.²⁵ Currently, the Office of the Privacy Commissioner of Canada does not support amending the *Privacy Act* or PIPEDA, on the grounds that these laws sufficiently apply to genetic information.²⁵ While this legislative framework might provide some protection against genetic discrimination, there is a lack of clarity as to whether it strikes the appropriate balance between consumers' rights to privacy and the interests of insurers. Furthermore, the courts have yet to provide an opinion regarding the constitutionality of S-201 or to assist in the interpretation of privacy legislation in the context of DTC testing, because of the novelty of the service.

It is uncertain *if* and *how* Bill S-201 will inform future regulations placed upon employers and insurers. Significant gaps in the legislative framework remain; in particular, privacy protection in Canada has yet to counterbalance the lack of consumer protection in Canadian insurance laws.²⁴ While existing legislation may offer some protection, the absence of legal precedents creates uncertainty and leaves consumers to engage in DTC testing services at their own risk.

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See also [CMA Policy PD17-05 Direct-to-Consumer Genetic Testing](#)

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