Direct-to-consumer genetic testing in Canada

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The goal of this article is to discuss DTC genetic testing in terms of the process of testing, the current regulation of the industry in Canada and the effect the resulting information can have on managing individual and populations in healthcare. This wide-ranging and rapidly evolving topic extends into the fields of genetics, ethics, law and business, each of which have been addressed in the following fashion: the accuracy of testing, the predictive value of the results, the possible effects that the test will have on the consumer and finally the effect that the personal genetics industry will have on physician practice and society. Due to the rapid evolution of the field, testing kits can now be purchased for as little as three hundred dollars, making personal genetic testing highly affordable and accessible.

THE ACCURACY OF THE ASSAY

DTC gene testing companies such as 23andme, deCODEME, Navigenics and Knome (the ‘main players’ in the current market) are private labs governed by the laws in their jurisdiction. In Ontario that licensing body is the Laboratory and Specimen Collection Centre Licensing Act (LSCCLA). However, the majority of these corporations are American and are hence regulated by the Centers for Medicare & Medicaid Services (CMS) under the Clinical Laboratory Improvement Amendments (CLIA). Recently there has been concerns about “faulty lab data analyses, exaggerated clinical claims, fraudulent data, poor clinical study design and a lack of traceability”. The United States Government Accountability Office recently reported that when samples of DNA were sent to four different DTC testing companies “different companies often provide different results for identical DNA”. Interestingly, the testing kits themselves are not considered medical devices and are hence not regulated by the Food and Drugs Act. Additionally, the majority of genetic tests are offered as in-house laboratory services and are similarly not regulated by the federal government through the Health Protection Branch or Therapeutic Products Program.

THE LINK BETWEEN TESTED GENES AND DISEASE

In contrast to genetic testing in a clinical context, private genetic testing also examines more “personal” genetics traits. According to the website for deCODEME you can “discover your genetic risk for 47 diseases and traits ranging from Heart Attack and Diabetes to Alcohol Flush Reaction and Male Pattern Baldness”. However disclaimers also state the results may have poor predictive value and are not considered diagnostic, leading customers to question the financial value of such test results. Nonetheless, these companies give their customers information about the research that links these polymorphisms to increased probability of developing a given disease.

THE EFFECT OF THE TEST RESULT ON THE CONSUMER

While some DTC companies offer over-the- phone genetic counselling with the results, it is important that all customers understand the information in a contextualized manner. This requires understanding of concepts such as relative and lifetime risks of disease. Physicians have also expressed concern over so called ‘genetic determinism’, where the individual negates environmental risk factors which are contributory to the disease in favour of the test results. For example, a patients’ risk of heart disease is more reflective of their weight, blood pressure and/or cholesterol than genetic factors. Another ethical concern is testing for conditions where there is no viable treatment or preventative measures. Advocates of DTC testing point to studies such as the REVEAL study, which showed that disclosure of a genetic trait predisposing patients to Alzheimer’s disease “did not result in significant short-term psychological risks” compared to those that were not tested.

Privacy concerns also exist about third party disclosure of data, especially in regards to health and life insurance, where this detailed information could lead to genetic discrimination. While company policy is generally not to disclose information without explicit consent of the customer, the information itself is not protected to the same standard as information covered by the US Health Insurance Portability and Accountability Act (HIPAA). If a customer discovers that they are positive for a mutation and fails to disclose it to their insurance company, it may serve as grounds for voiding policy if the insurance company finds a way to access the information. Similarly, the Canadian Human Rights Act does not contain a clause protecting against genetic discrimination, though an amendment is currently under consideration in the House of Commons (Bill C-536). On the other hand, Americans are protected from such discrimination by the Genetic Information Non-discrimination Act (GINA) which does not apply to Canadians using the US based companies.

THE ROLE OF THE HEALTHCARE SYSTEM

Ontario’s Regulated Health Professions Act stipulates that a healthcare professional is responsible for “communicating to the individual or his or her personal representative a diagnosis identifying a disease or disorder as the cause of symptoms of the individual in circumstances in which it is reasonably foreseeable that the individual or his or her personal representative will rely on the diagnosis”. Does the genetic risk assessment offered by these privately owned companies count as a diagnosis? The companies explicitly state that their products are not diagnostic or are substitute for medical advice and that all concerns arising from the test should be referred to your healthcare provider. The onus then lies on the
family practitioner to interpret the information, a burden that could become substantial according to a study by McGuire et al where 78% of respondents said they would consult their family doctor about results of commercial genetic test\(^4\). This increased burden on the healthcare system is further complicated by the fact that family physicians are often not trained in genetics counselling and may not be equipped to deal with this extensive genetic information.

**CONCLUSION**

In 2010, the FDA sent regulation letters to five American commercial genetic testing companies stating that their device is regulated “…under section 201(h) of the Federal Food, Drug, and Cosmetic Act (the Act), 21 U.S.C. 321(h) because it is intended for use in the diagnosis of disease or other conditions or in the cure, mitigation, treatment, or prevention of disease, or is intended to affect the structure or function of the body,”\(^1\); it went on to state that the product is classified as a medical device under the Medical Device Act and as such require premarket approval\(^13\). Increasing regulation has been occurring in both the United States and many European countries out of concern for inaccurate or misleading test results\(^14\).

However, suppose the tests are accurate and relatively specific the next logical question is whether the knowledge of risk factors will translate into better outcomes for patients: reducing risk, earlier screening, prophylactic measures and, in some cases, influence on family planning\(^6\). It is difficult to assess this as the industry is relatively new and there is no follow up after the company discloses the results to the customer. Without counselling it may be difficult to put the risk into context for the individual patient and an over or underestimation could be deleterious to a patients’ health\(^4\).

Proponents argue these tests champion patient autonomy and to limit access to one’s own genome is paternalistic while detractors are concerned the patchwork regulation does not protect customers purchasing these products. With expanding use of these services the effects of this plethora of information, both positive and negative, will be become more apparent.

**REFERENCES**