

Home-based genetic testing: a risky business?

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Introduction

Genetic tests are powerful medical tools that appraise the probability of disease¹. *Presymptomatic* tests identify gene mutations that lead to inevitable hereditary conditions such as Huntington's disease, while *predispositional* tests locate gene expressions that are risk factors assisting in the early identification of diseases such as cancer, heart disease, and Alzheimer's disease^{1,2}. Best practice guidelines for the medical provision of genetic testing state that genetic tests must be accompanied by patient education and support or counselling¹⁻³. Education prior to genetic testing ensures that the patient is aware of the risks and benefits and can subsequently provide informed consent¹⁻⁴. Genetic counselling is the process through which health professionals help and support patients as they cope with and adapt to having, or being at risk of developing, a genetic condition³. Genetic education and counselling are critical to minimize negative effects of, and help patients cope with, test results.

Psychosocial Impact of Genetic Testing

A positive DNA test result can cause prolonged feelings of anxiety and worrying as well as worsened psychological functioning⁵. It may also influence an individual's reproductive choices, particularly when it is determined that they are carriers of incurable hereditary diseases such as Huntington's disease or cystic fibrosis^{3,6}. The risks of genetic tests extend beyond the individual, affecting their social environments. For example, results of genetic testing can lead to disruption of familial relationships and other social consequences including loss of life insurance or employment^{1,2}. Despite these serious implications, scientific and technological advances in the field of genetics are making genetic testing increasingly accessible to the general public². Today, Canadians can purchase simple home-based genetic tests (HBGTs) online for the price of

\$199. Once purchased, consumers submit a small sample of saliva to identify over 200 genetic markers which provide genealogical health information on more than 100 health conditions that the customer can interpret however they see fit without the guidance of a healthcare professional⁷. Therefore, the authors of this paper explore the potential repercussions of widespread merchandizing of HBGTs and offer recommendations to ameliorate current practices and develop policies that protect consumer health.

Current Practice of HBGT

Alarming, Health Canada currently has no policies in place to regulate quality, reliability, or validity of HBGTs and without government regulation and standardization of these measures, the likelihood of erroneous tests results increases¹. Both positive and negative genetic test results can precipitate monumental life changes; therefore, inaccurate results from HBGTs may cause individuals to make fallacious and irreversible decisions about their lifestyle, relationships, and employment^{4,6}. For example, false negative HBGT results may lead people to avoid necessary medical advice or treatment¹. In contrast, if a HBGT overstates the genetic basis of a disease or provides inconclusive results, an individual may overestimate their risk of disease and subsequently seek unwarranted screening services and clinical examinations^{1,8}. This increased demand for medical attention congests doctors' offices, squanders health care dollars, and increases the burden on an already strained health care system⁸.

Furthermore, unlike medically administered genetic testing, businesses selling HBGTs are not obligated to obtain informed consent, provide education or offer counselling for genetic tests.¹ Without adequate education and informed consent, individuals may not be aware of the implications of their HBGT results, which may increase the risk and severity of confusion, anxiety, and psychological distress

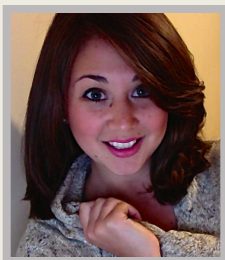
over test results. Therefore, lack of government regulation and medical oversight of HBGTs can exacerbate the existing risks of genetic testing, causing greater implications for individuals and society as a whole^{1,2,9}.

Recommendations and Conclusions

In accordance with the Canadian College of Medical Geneticist position on HBGT, it is recommended that only scientifically valid tests be offered, and the technical and clinical limitations of the testing including sensitivity, specificity, and utility in assessing health must be clearly stated in a manner understandable to the target market.¹⁰ Furthermore, the authors of this paper strongly recommend the implementation of obligatory policies that require HBGT services to: 1) provide information about the psychosocial risks of genetic testing, 2) disclose who has access and ownership to genetic sample and test results, and 3) obtain informed consent from consumers. Finally, since HBGTs are already marketed and distributed in Canada, the authors call for further research to be conducted in order to establish quality standards that ensure the accuracy of these tests. The Canadian health care system has best practices in place including patient education and counselling to mitigate negative consequences and protect the health and well being of patients that undergo genetic testing. Home-based genetic tests blur the line between patient and consumer. In light of this, private companies selling HBGTs for profit must also be held accountable for the implications of their products. ■

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Kristen Reilly obtained her Master of Public Health (MPH) from the University of Guelph and is currently a Ph.D. student in the Department of Health and Rehabilitation Sciences at Western University. Kristen is passionate about health promotion, health policy, and advocacy. The primary objective of her doctoral research is to develop and implement a parent-focused advocacy network (C.H.A.M.P. Families) to engage, inform, and unite parents in London, Ontario. She hopes that this network will empower parents to advocate for the health of their families and communities by providing them with the necessary tools and resources to do so. Kristen is a recipient of the Canadian Institutes for Health Research Frederick Banting and Charles Best Canada Graduate Scholarships Doctoral Award.



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Shawn Slade is PhD student in Health Promotion at Western University in London, Ontario. His Master's thesis examined the perceived barriers, facilitators, and health benefits of sustaining a fitness facility membership amongst active and inactive adult women. Shawn's area of research for his dissertation will focus on family physicians and physical activity counselling/prescription in the primary care setting. Shawn is returning to graduate education after years as a certified exercise professional to be a professor within the health sciences. He is eager to continue developing his knowledge on the use of theory in qualitative health promotion research.