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CCMG Statement on Direct-to-Consumer Genetic Testing

CCMG Ethics and Public Policy Committee
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PURPOSE: This document recommends minimum standards for medically significant genetic testing in Canada, and is designed particularly for consideration in the field of direct-to-consumer genetic testing (DTC-GT).

STATEMENT DEVELOPMENT: This statement was developed by the CCMG Ethics and Public Policy committee with input from members of the Molecular Genetics and Clinical Practice committees and the membership-at-large. This statement has been approved by the CCMG Board of Directors. The statement should be viewed as a living document, as it reflects current knowledge and experience which will change over time.

RECOMMENDATIONS: Given the potential for harm, Canadians are best served when standards and regulations are in place to maintain consumer safety around medically significant genetic tests. Standards and regulations require some concessions with respect to customer access, autonomy, and empowerment, but they protect the consumer. We accept this approach in the pharmaceutical industry, and we should accept it in business of medically significant genetic testing. Specifically, all medically significant genetic testing should comply with the following standards and regulations:

Genetic testing must be based on valid scientific evidence.

The utility of the test in assessing health should be clearly stated.

Testing laboratories and personnel must be accredited/certified by reputable accrediting/certifying bodies for the provision of clinical genetic testing and must participate in recognized proficiency testing programs.

Genetic tests should be accurately labelled as medically significant or not.

Providers of genetic testing must practice responsible marketing.

Orders for tests with medically significant implications should only be accepted from a medical professional on behalf of the individual to be tested.

Professional guidelines related to the practice of medical genetics should be adhered to, particularly with respect to genetic testing of children.

Privacy and confidentiality must be addressed and maintained.

Samples for tests with medically significant implications must be collected in a manner that limits the possibility of accidental or purposeful misidentification and contamination.

RATIONALE: Significant advances in our understanding of the genome and our approaches to studying it have reduced the cost of genetic testing and led to the expansion of genetic testing services outside of the realm of a relatively small number of controlled clinical laboratories into the commercial sphere. Direct-to-consumer genetic testing (DTC-GT) refers to tests ordered directly by the consumer without the involvement of a healthcare provider[1]. Currently, DTC-GT services are marketed for a variety of types of genetic testing including recreational, ancestry analysis, paternity testing, fetal gender determination, disease risk stratification, assessment of drug response, and pre-conception screening. For example, DTC-GT companies may report on characteristics such as earwax type or bitter taste perception that are generally irrelevant to well being, may provide a predicted risk of developing complex disease such as macular degeneration, or may provide carrier screening for single gene disorders.

Proponents of DTC-GT have stated that these services allow for increased access, increased autonomy, and empowerment of the consumer. It has also been suggested that DTC-GT might allow for increased privacy and confidentiality, and that this could decrease some forms of genetic discrimination. However, genetic testing has the potential to be harmful, and in a non-regulated environment has the potential to devastate individuals or families.

Quality of Information

Only scientifically valid tests should be offered. Both the actual genetic finding and the interpretation of the finding(s) should be reported. 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. References should be provided to the primary data upon which the test is based and, where applicable, to the method used for assessing risk.

It has been demonstrated that due to lack of standardization, an individual assessed by more than one DTC-GT service can receive significantly different predictions for future risk of a specific disease[2]. Furthermore, there are many genetic variants throughout the genome for which the interpretation of pathogenicity is not clear, particularly in the context of identification via carrier screening of an apparently healthy individual. The analytical approaches used by some DTC-GT companies will identify genetic variants of unclear significance, as well as benign polymorphisms known to have no impact on the individual's or future pregnancy's health. The premature provision of genetic assessments, especially in the context of non-standardized approach to risk assessment, has the potential to lead to inappropriate follow up testing, consumer anxiety, confusion, misinterpretation (falsely reassured or falsely increased concern)[3], and unnecessary reproductive interventions.

Quality of Service

The issue of quality management is rapidly evolving due to increased scrutiny by regulators, at least within the United States of America. However, depending on local requirements, DTC-GT companies may not conform to standard quality management systems that are in practice in clinical laboratories. For example, the laboratory may not be licensed or certified to perform genetic testing, may not participate in proficiency testing or other ways of monitoring data quality, and the laboratory staff performing testing, analysis and interpretation may not be trained and certified by appropriate governing bodies for the provision of medically-related genetic testing.

Marketing of Genetic Testing

Genetic testing should be marketed appropriately to reflect whether or not the results of testing are medically significant. Marketing should not include disclaimers that imply or state that testing is non-medical when the results are medically relevant. For example, hemochromatosis carrier testing, or risk for developing cancer, clearly have medical implications and should be labelled appropriately and responsibly.

Additionally, when providers receive direct benefit from an individual choosing to access downstream services, options should be presented without bias and alongside any risks and/or limitations. This is especially true in the context of reproductive decision making. Deferring discussion to the potential consumer's primary health care provider, as suggested by advertised recommendations stating "ask your doctor", is insufficient and inappropriate.

The information provided should be stated in language appropriate to the target market, with appropriate scientific references.

Public Health System

The Canadian health care system is based on the premise of "universal coverage for medically necessary health care services provided on the basis of need, rather than the ability to pay"[4]. Prior to implementation, programs established within the Canadian health care system are put through a stringent process that includes assessment of overall benefit to public health and associated follow-up funding. DTC–GT that identifies people at risk for disease, without full service and follow-up, results in a group of individuals who seek interpretation and further work up within the public system. When a subgroup of the Canadian population purchases a test that can make them eligible for public resources, the integrity of the system with just and universal access is threatened. Further, if the company providing the test has limited disclosure and substandard quality assurance, the results of DTC-GT may be difficult for even the medical experts to interpret, resulting in the medical system using limited resources to dismiss falsely identified "at-risk states". As such, without adherence to best practices, DTC-GT could introduce unjust access to publicly funded health services and become a net resource drain on the public health care system in Canada.

The Role of the Clinician

The medical professional ordering a test has an obligation to: obtain informed consent prior to ordering testing, interpret the result(s), support the individual with respect to the psychosocial and biological implications of the result(s), and access available health care to modify the natural history of a predisposition or a diagnosis. This includes, but is not limited to, educating an individual about the test and its various implications, such as what the results predict about the individual's future health and that of family members, the potential for insurance discrimination, the possibility of receiving difficult to interpret results or misinterpreted results. It is evident that genetic testing that provides a risk or probability of developing disease, rather than a definitive answer, can only be accurately interpreted within the context of the individual's personal and familial medical history. Other factors, for example obesity or smoking, may confer a higher risk for a given individual than the "genetic risk" provided by the test. Similarly, pre-conception risk prediction often requires assessment of the interaction of the two parental variants, and the resultant prediction is not always clear. Receiving a test result without interpretation within the clinical context may, therefore, falsely reassure or falsely alarm an individual with regards to their risk, or their future offspring's risk, for developing disease. In this context, it is naïve to expect that the consumer, whose perception of genetic risk may be influenced by their personal experiences rather than an understanding of the limitations of testing, can interpret these data in isolation.

Involving the professional in accessing medically significant testing also provides an important safeguard to our society. The professional assumes responsibility for ensuring that testing is sought autonomously. It is not appropriate for such tests to be purchased on behalf of others, for example a young child, or as a surprise gift to another.

Practice guidelines

Professional and ethical guidelines have been developed by professional organizations consisting of medical geneticists. These documents direct the standard of care for genetic testing including, but not limited to, predictive testing for late onset disorders, prenatal testing, and genetic testing in minors (which is generally considered inappropriate unless there is benefit to the minor in the immediate future i.e. while still a minor)[5,6,7]. These evidence-based guidelines are created to ensure the well-being and safety of individuals accessing genetic testing and are updated to reflect current scientific evidence. All physicians should be aware and understand these guidelines before advising patients/individuals or ordering genetic testing.

Privacy and Confidentiality

While privacy and confidentiality have been cited as possible advantages of testing through a private DTC-GT company as compared to the public medical system, in reality the safeguards offered by such companies are unregulated and could leave the client's private information and genetic sample vulnerable. Furthermore, genetic

privacy in the context of DTC-GT may be unrealistic given current technologies, the provision of DTC-GT through the internet, and the popularity of social networks[8].

Prior to DTC-GT, the consumer should be provided the following information: accessibility of test results (who and how), security of the results and the DNA sample, fate of the DNA samples upon test completion, mechanism to report breaches of privacy, and if applicable, fate of an individual's information and DNA sample in the event of a change in ownership or closure of a company or laboratory.

With respect to DTC-GT in particular, there is the risk that individuals may consent to the release of their information without understanding the potential ramifications of such consent or even being aware that they have provided consent, and without having considered the potential consequences of sharing their information with others[9]. Given the often lengthy and unwieldy language of software terms and conditions, privacy and license agreements it is common that individuals do not review the agreement prior to choosing "I agree". Therefore, it is reasonable to assume that if a consumer purchasing DTC-GT services is presented with information in a similar format, regardless of the simplicity of the document, an assumption may be made about the content and the consumer may consent (agree) without truly providing informed consent.

Sample collection and management

Ideally, samples should be collected by an accredited blood collection facility using the facility's established identification protocol(s). Alternatively, samples should be collected by a medical professional who assumes the responsibility for ensuring that identification protocols are followed. Self-collection allows for an increased risk of accidental or purposeful sample misidentification and contamination at the time of collection. Mistakes are dangerous for the individual, and abuse the limited resources of a public health system as they lead to costly downstream testing in order to prove/disprove the results of a genetic test.

References:

Evans JP and Green RC. Direct to consumer genetic testing: Avoiding a culture war. Genet. Med. 2009;11(8):568-569.

Kutz G. United States Government Accountability Office. Testimony. Direct-to-Consumer Genetics Tests. GAO-10-847T (22Jul10). http://www.gao.gov/new.items/d10847t.pdf

Caulfield T *et al*. Direct-to-consumer genetic testing: good, bad or benign? Clin. Genet. 2010;77(2):101-105.

http://www.hc-sc.gc.ca/hcs-sss/index-eng.php (date modified: 2009-05-20)

Borry P *et al*. Presymptomatic and predictive genetic testing in minors: a systematic review of guidelines and position papers. Clin. Genet. 2006;70(5):374-81.

Arbour L *et al*. Guidelines for genetic testing of health children. Paediatr Child Health 2003;8(1):42-45.

Arbour L *et al*. Guidelines for genetic testing of healthy children - addendum. Paediatr Child Health 2008;13(4):311.

Knoppers BM. Consent to 'personal' genomics and privacy. EMBO reports 2010;11(6):416-419.

Howard HC, Knoppers BM, Borry P. Blurring lines. EMBO reports 2010;11(8):579-582.