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## DIRECT-TO-CONSUMER GENETIC TESTING

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See also [Background to CMA Policy on Direct-to-Consumer Genetic Testing](#)

### RATIONALE

While genetic testing is typically provided in a clinical setting through the referral of a health care professional (HCP) or a regulated research project, a number of private companies now offer genetic testing services directly to consumers over the Internet. Direct-to-consumer (DTC) genetic testing is distinguished from clinical genetic testing ordered by a HCP in several ways:

1. DTC genetic tests are not regulated in Canada. The clinical validity and reliability of these tests varies widely, but DTC genetic testing companies make them available to consumers without distinguishing between those that may be useful to the management of one's health, those that have some limited health value, and those that are meant purely for recreational use.
2. Many of the tests advertised and sold via the Internet have not undergone clinical evaluation.
3. Marketing materials for these tests often imply that they have health value, but the terms of reference of some of the companies that offer them state that the tests are to be used for recreational purposes and many vendors do not guarantee the validity or reliability of their results.
4. Resale of personal health information and/or DNA samples is often an important part of the business model of companies that offer DTC genetic testing, raising concerns about patient privacy and insufficient or unclear disclosure of privacy terms.
5. Unlike genetic tests ordered and administered by HCPs, DTC genetic tests are ordered directly by the consumer, who most often has not consulted with a HCP as part of a clinical assessment, and the testing may not be clinically indicated. Some companies only agree to do testing if it has been ordered by a physician, but they will provide a phone conversation with one of their physicians (not based in Canada) if a consumer does not have access to a physician. When the testing is ordered by a physician, it will sometimes be ordered by the patient's personal physician. In such cases, this does not truly represent DTC genetic testing.

6. Without appropriate pre- and post-testing counselling by a HCP, consumers are left to interpret and act upon their results on their own. They might suffer psychological consequences if they overestimate their disease risk as a result of DTC.
7. As access to DTC genetic testing increases, Canadian HCPs (specifically primary care physicians) are faced with the challenge of appropriately counselling patients when they receive their test results. However, few physicians feel they have the necessary training and knowledge in genomics to provide adequate care in this area. Furthermore, these tests may have no clinical indication, produce uncertain results with ambiguous clinical applicability and have tenuous legal status, but they can potentially influence a patient's sense of well-being.

## GENERAL PRINCIPLES

1. The CMA is concerned with understanding, raising awareness of, and mitigating potential patient and societal harms that may arise from DTC genetic testing.
2. The CMA emphasizes the importance of the principle of protection of patient privacy and supports the right of Canadians to understand how their health information is being used by third parties, including insurance and DTC genetic testing companies.
3. The CMA believes that patients have the right to be fully informed about what a DTC genetic test can and cannot say about their health and that the scientific evidence on which a test is based should be clearly stated and easy to understand.
4. The CMA recommends regulation of both DTC genetic tests and the marketing of these tests through the development of a national framework that would include a combination of government and industry regulation with input from medical experts.
5. The CMA believes that unnecessary genetic testing should be avoided to ensure more appropriate use of health care resources. Even if a consumer pays directly for testing, any test result, even an incidental finding from a DTC genetic testing laboratory without clinical certification, may trigger a cascade of clinical investigations and lead to further unnecessary testing and inappropriate use of resources.
6. The CMA supports educational initiatives on DTC genetic testing for physicians practising in all specialties so that they can respond to patient queries about these tests and, when necessary, their results.

## PROTECTION OF PRIVACY

- Privacy and confidentiality of patients' personal health information must be maintained.
- Before a patient submits a sample to a DTC genetic testing company, the company should obtain express informed consent from the patient concerning the way in which their data will be collected and used, who will have access to the data and the interpreted results, what safeguards are in place to protect it, and how it will be disposed of in the event of a company/laboratory closure.

- Patients have the right to a clear understanding of who owns the sample and the generated data, in particular whether their data will be sold or shared with third parties. If resale of personal health information and/or DNA samples is an important part of the business model of DTC-GC companies, this should be stated explicitly in terms understandable by the consumer.
- DTC-GC companies that solicit Canadian consumers should be subject to the *Personal Information Protection and Electronic Documents Act (PIPEDA)*.
- The CMA encourages physicians to become familiar with privacy legislation affecting the use of DTC genetic tests by insurance companies and employers.

## ROLE OF PHYSICIAN

- Physicians should generally avoid using DTC genetic tests unless they have been clinically and empirically validated.
- Physicians who are presented with a patient's DTC genetic test results should take the following actions:
  - They should explain to their patient the limits of the specific test the patient used. If a physician does not know this information he/she should discuss with the patient the fact that DTC genetic test results are not necessarily obtained from an accredited laboratory or interpreted in a standardized way; therefore, the validity and clinical utility of the results may be highly variable for certain tests.
  - They should disclose their level of comfort in providing an accurate interpretation of the results.
  - They should assess whether the test results are clinically significant in the context of that patient's symptoms, signs, medical history and family history before deciding whether it is appropriate to formally consult a specialty provider such as a medical geneticist.
  - If a physician wishes to use the results of a test in their clinical assessment, they should ensure that the laboratory performing the test guarantees analytical reliability and validity.
- Physicians should adhere to the following principles related to medically indicated genetic testing:
  - Physicians should generally avoid recommending and/or ordering DTC genetic tests if they do not have a clear understanding of the validity and limitations of the tests they select.
  - Physicians should follow best practice guidelines and make use of clinically valid tests, accredited laboratories and specialist referral(s), when appropriate.
  - Physicians must obtain informed consent from the patient before ordering any genetic test, assist the patient in interpreting the results, support the individual with respect to

psychological and biological implications of the results, and refer the patient to appropriate resources.

- o Many genetic tests require pre- and post-test counselling, particularly (but not limited to) tests involving children, tests establishing carrier status or tests considered to be predictive. If a provider decides to order such testing, they also accept the responsibility for facilitating access to pre- and post-test counselling.

## ROLE OF GOVERNMENT

- The CMA calls on the government to enact regulations based on Bill S-201 (*An Act to prohibit and prevent genetic discrimination*) that establish clear boundaries for the marketing, distribution, accreditation and third-party use of DTC genetic tests.
- The CMA believes that it is the government's responsibility to ensure that Canadians are only offered reliable, accurate and medically relevant genetic testing services.
- The CMA encourages the development of national standards for the reliability and validity of DTC genetic tests by relevant federal government agencies, in conjunction with interested stakeholders (e.g., geneticists and laboratory scientists, genetic counsellors, physicians, private and public laboratories, industry, and patient groups).
- The CMA encourages the government to enact standards that can keep pace with the rapid development of technological innovation in genetic testing and genetics more generally.
- The CMA encourages the government to enact standards that hold companies accountable for being transparent about their uses of data/DNA and the potential resale of such material.
- The CMA encourages the government to enact standards that mandate that the type of testing (e.g., single-nucleotide polymorphism [SNP] analysis, targeted mutation testing, sequencing) be clearly labelled and that a clear explanation be provided of the type of information that can (or cannot) be obtained from such testing.

## SYSTEMS INFRASTRUCTURE

- Genetic testing and the interpretation of the results of such testing are highly technical and complex processes. For this reason, the CMA believes that clinical testing laboratories that are used by DTC genetic testing companies must be accredited if the companies are to claim that their testing is valid.
- The CMA believes that scientific evidence describing the validity and utility of a DTC genetic test should be clearly stated in language that is easy to understand. This information should include a clear statement of what a test can or cannot diagnose or infer, and statements about the validity of a specific test should be supported with references. A company that does not guarantee the reliability or validity of its test should

not be allowed to make any (implicit or explicit) claims about the potential medical utility of its test and/or its potential to improve health.

## EDUCATION AND PUBLIC ENGAGEMENT

- The CMA supports public education initiatives to increase patient awareness of the potential implications and limitations of DTC genetic testing for health purposes. The CMA supports increased genetics training for physicians to help them to further appreciate the complex issues involved and keep pace with the rapid changes in molecular genetics. Such training would support physicians to counsel patients who seek follow-up for their DTC genetic test results.

Approved by the CMA Board of Directors May 2017

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