

Online DTC genetic testing: Implications of this new business model

A genetic counsellor's perspective

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Canadian Association of Genetic Counsellors

Association Canadienne des Conseillers en Génétique

Genetic Counselling in Canada

- 250 GCs belong to the Canadian Association of Genetic Counsellors (CAGC)
 - ~2/3 of workforce
- Majority of GCs are Masters-trained and work with Medical Geneticists
- Certification is via examination
 - CCGC (Canadian certification)
 - CGC (American certification)

What does a genetic counsellor do?

- Helps people to understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.

The classic GC encounter

- Face-to-face consultation for ~1 hour to:
 - Take a detailed family history
 - Make risk assessment + help **contextualize** risk
 - Present testing options, if available (via provincial or out-of-country labs), including discussion of
 - Clinical utility of test
 - Potential results: positive, negative, unclear
 - Potential implications for health, insurability, discrimination and impact for family members
- Follow-up appointment to communicate results, if patient has chosen to proceed with testing

Three primary concerns about the DTC genetic testing model

1-Pressure on existing Genetics resources and the healthcare system

- Potential for an increase in volume of referrals to medical genetics clinics (appropriate or not)
- Queue-jumping
- Potential for repeat genetic testing

2-Quality assurance in DTC labs

- Validity and utility of results
 - Depending upon the DTC company used, an individual may receive a completely different risk profile
 - Laboratory accreditation
- Lab director and personnel qualifications
 - Often not specified
- Concern about erosion of public confidence in the value of genetic testing and Medical Genetics.

3-Results interpretation

- Who is responsible for explaining the results and for follow up care resulting from the findings?
 - Potential for inappropriate or disproportionate reaction to DTC results
- Dissemination of DTC genetic test information among family members—increased opportunity for a cascade of misinformation?

3-Results interpretation cont'd

- “INTERPRETABLE”
 - e.g. BRCA result
- (Although even well-known genes contain variants of unknown significance)
- “ UNINTERPRETABLE”
 - e.g. genome-wide scan
- Bioinformatics outpacing understanding of clinical significance

Policy option 1:

- *Tighter regulatory oversight, such as mandatory **involvement** of a **health professional** independent from the company, may be useful for “more **serious**” tests.*
 - Challenge to define these terms
- The need for counselling is not always proportionate to the seriousness of the condition tested.
- Ideally, a genetics health professional should be involved
- Regulatory oversight is desirable.

Policy option 2:

- Enforcement of existing consumer protection laws is important
- This option is not likely to help genetics health professionals confronted with DTC test results in practice

Policy option 3:

- *“...consumers require...clear, evidence-based information about the predictive value of the test and the **meaningfulness** of test results, and information about the company’s policies and practices for handling genetic samples and personal information.”*
 - Meaningfulness is highly subjective, not something easily disclosed on a website
 - Helping people to determine the meaningfulness of a test result is a reason for genetic counselling.

Policy option 3 cont'd:

- Information campaign desirable
 - In line with CAGC's own policy and public awareness objectives
- Support for practical tools
 - publicly accessible genetic test registries
 - any tools that would equip genetics health professionals to assess DTC genetic test results

Policy option 4:

- Action is preferable to “wait and see”

Future research questions

- *How are consumers using their DTC genetic test information?*
 - What are the factors driving the demand for DTC genetic tests?
 - Are there gaps that the Medical Genetics community could address by providing alternatives?
- *Incorporating genetic testing into public healthcare system*
 - Even when tests are available clinically, they are likely to be desirable to the consumer
- *Impact of DTC genetic testing on health care costs*
- Genetics health services research in personalized medicine
- Do current genetic counselling approaches and outcome measures fit the DTC business model?

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The mission of the Canadian Association of Genetic Counsellors is to promote high standards of practice, encourage professional growth and increase public awareness of the genetic counselling profession in Canada.