Ethics series — 2

Direct-to-consumer genetic testing — where should we focus the policy debate?

What are the implications for health systems, children and informed public debate?

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Until recently, human genetic tests were usually performed in clinical genetics centres. In this context, tests are provided under specific protocols that often include medical supervision, counselling and quality assurance schemes that assess the value of the genetic testing services. Direct-to-consumer (DTC) genetic testing companies operate outside such schemes, as noted by Trent in this issue of the Journal.[1] While the uptake of DTC genetic testing has been relatively modest, the number of DTC genetic testing services continues to grow.[2] Although the market continues to evolve,[3] it seems likely that the DTC genetic testing industry is here to stay.

This reality has led to calls for regulation, with some jurisdictions going so far as to ban public access to genetic tests outside the clinical setting.[4,5] In Australia, as Nicol and Hagger observe, the regulatory situation is still ambiguous;[6] regulation is further complicated by the activity of internet-accessible companies that lie outside Australia's jurisdiction. In general, the numerous policy documents that have emanated from governments and scientific and professional organisations cast DTC services in a negative light, seeing more harms than benefits, and, in some jurisdictions, governments have tried to regulate their services and products accordingly.[7,8] Policy debates have focused on the possibility that DTC tests could lead to anxiety and inappropriate health decisions due to misinterpretation of the results. But are these concerns justified? Might they be driven by the hype that has surrounded the field of genetics in general. If so, what policy measures are actually needed and appropriate?

Time for a hype-free assessment of the issues?

Driven in part by the scientific excitement associated with the Human Genome Project, high expectations and a degree of popular culture hype have attracted both public research funds and venture capital to support the development of disease risk-prediction tests.[3] This hype — which, to be fair, is created by a range of complex social and commercial forces[9] — likely contributed to both the initial interest in the clinical potential of genetic testing *and* the initial concerns about possible harms. Both are tied to the perceived — and largely exaggerated — predictive power of genetic risk information, especially in the context of common diseases. There are numerous ironies to this state of affairs, including the fact that the call for tight regulation of genetic testing services may have been the result, at least in part, of the hype created by the both the research community and the private sector around the utility of genetic technologies.[9] This enthusiasm helped to create a perception that genetic information is unique, powerful and highly sensitive, and specifically that, as a result, the genetic testing market warrants careful oversight.

Now that research on both the impact and utility of genetic information is starting to emerge, a more dispassionate assessment can be made about risks and the need for regulation. Are the concerns commonly found in policy reports justified? Where should we direct our policymaking energy?

It may be true that consumers of genetic information — and, for that matter, physicians — have difficulty understanding probabilistic risk information. However, the currently available evidence does not show that the information received from DTC companies causes significant individual harm, such as increased anxiety or worry.[10,11] In addition, there is little empirical support for the idea that genetic susceptibility information results in unhealthy behavioural changes (eg, the adoption of a fatalistic attitude).[5]

The concerns about consumer anxiety and unhealthy behaviour change have driven much of the policy discussion surrounding DTC testing. As such, the research could be interpreted as suggesting that there is no need for regulation or further ethical analysis. This is not the case. We suggest that the emerging research invites us to focus our policy attention on issues that reach *beyond* the potential harms to the individual adult consumer — where, one could argue, there seems to be little empirical evidence to support the idea that the individual choice to use DTC testing should be curtailed — to consideration of the implications of DTC testing for health systems, children and informed public debate.

Health system costs

Although genetic testing is often promoted as a way of making health care more efficient and effective by enabling personalised medical treatment, it has been suggested that the growth in genetic testing will increase health system costs. A recent survey of 1254 United States physicians reported that 56% believed new genetic tests will increase overall health care spending.[12]

Will DTC testing exacerbate these health system issues by increasing costs and, perhaps, the incidence of iatrogenic injuries due to unnecessary follow-up? This seems a reasonable concern given that studies have consistently shown that DTC consumers view the provided data as health information that should be brought to a physician for interpretation. One study, for example, found that 87% of the general public would seek more information about test results from their doctor.[13] The degree to which these stated intentions translate into actual physician visits is unclear. But for health systems striving to contain costs,

even a small increase in use is a potential health policy issue, particularly given the questionable clinical utility of most tests offered by DTC companies. It seems likely that there will be an increase in costs with limited offsetting health benefits — although more research is needed on both these possible outcomes.

Compounding the health system concerns is the fact that few primary care physicians are equipped to respond to inquiries about DTC tests. A recent US study found that only 38% of the surveyed physicians were aware of DTC testing and even fewer (15%) felt prepared to answer questions.[14] As Trent notes, even specialists can encounter difficulties in interpreting DTC genetic tests.[1] This raises interesting questions about how primary care physicians will react to DTC test results. Will they, for example, order unnecessary follow-up tests or referrals, thus amplifying the concerns about the impact of DTC testing on costs?

Testing of children

While there is currently little evidence of harm caused by DTC genetic testing, most of the research has been done in the context of the adult population. The issues associated with the testing of minors are more complicated, involving children's individual autonomy and their right to control information about themselves. Many DTC genetic testing companies include tests for adult-onset diseases or carrier status. Testing children for such traits contravenes professional guidelines. Nevertheless, research indicates that only a few DTC companies have addressed this concern. A study of 29 DTC companies found that 13 did not have policies on the issue and eight allowed testing if requested by a parent.[15] While it is hard to prevent parents from submitting samples from minors to genetic testing companies, this calls for an important policy debate on whether there are limits on parental rights to access the genetic information of their children. Current paediatric genetic guidelines recommend delaying testing in minors unless it is in their best interests, but these are not enforceable and not actively monitored.[16]

In addition, unique policy challenges remain with regard to the submission of DNA samples in a DTC setting. It is difficult for DTC companies to check whether the sample received is from the person claiming to be the sample donor. Policymakers should consider strategies, such as sanctions, that eliminate the ordering of tests without the consent of the tested person.

Truth in advertising

The DTC industry is largely based on reaching consumers via the internet. Research has shown that the company websites — which, in many ways, represent the face of the industry — contain a range of untrue or exaggerated claims of value.[17] Advertisements for tests that have no or limited clinical value have a higher risk of misleading consumers, because the claims needed to promote these services are likely to be exaggerated. It is no surprise that stopping the dissemination of false or misleading statements about the predictive power of genetics has emerged as one of the most agreed policy priorities.[8] While evidence of actual harm caused by this trend is far from robust, it is hard to argue against the development of policies that encourage truth in advertising and the promotion of more informed consumers. Moreover, the claims found on these websites may add to the general misinformation about value and risks associated with genetic information that now permeates popular culture. Taking steps to correct this phenomenon is likely to help public debate and policy deliberations. For example, this might include a coordinated and international push by national consumer protection agencies to ensure that, at a minimum, the information provided by DTC companies is accurate.[18]

Conclusion

These are not the only social and ethical issues associated with DTC genetic testing. Others, like the use of DTC data for research and the implications of cheap whole genome sequencing, also need to be considered. But they stand as examples of issues worthy of immediate policy attention, regardless of what the evidence says about a lack of harm to individual adult users. We must seek policies that, on the one hand, allow legitimate commercial development in genomics and, on the other, achieve appropriate and evidence-based consumer protection. In finding this balance, we should not be distracted by hype or unsupported assertions of either harm or benefit.

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