Perspectives

DTC genetic testing: pendulum swings and policy paradoxes


After decades of optimistic portrayals, there has been a shift in the way that the popular press represents genomic research. A skeptical view has become more common. The central reason for this pendulum swing away from popular support is the harsh truth that most genetic risk information just isn’t that predictive. This reality has created a fascinating policy paradox. If, as many in the scientific community are now saying, genetic information is not the oracle of our future health as we were once led to believe, and if access does not, for most, cause harm, why regulate the area? Why worry about shoddy direct-to-consumer (DTC) genetic testing companies? One primary justification, and one endorsed by the recent Canadian College of Medical Geneticists (CCMG) Policy Statement on DTC Genetics Testing, is that information that is conveyed to the public about genetics via marketing and to those who access DTC tests should, at a minimum, be accurate.

Conflict of interest
The author has no conflicts of interest to declare.

In 1983, The New York Times ran an article with the headline: ‘Keeping Up with the Genetic Revolution’ (1). The story outlined the exciting, near future, clinical breakthroughs that were likely to emerge as a result of genetic research. After the Human Genome Project (HGP) took off in the early 1990s, so did the number of hyperbolic headlines and magazine covers that asserted that a revolution was either already here (2) or just around the corner (3, 4).

But after decades of this constant and consistent cheerleading, we have seen a shift in how popular culture references genetics. Over the past couple of years, the tone of the headlines has changed (Table 1). The new, HGP-informed genetics is now framed as a ‘Revolution Postponed’ (5), as being ‘oversold’ (6), as failing to produce the expected payoffs (7), as providing ‘few new cures’ (8) and, most despairingly, as a complete failure (9).

The central reason for this pendulum swing away from popular support is the harsh truth that, as it turns out, most genetic risk information just is not that predictive. As noted in a 2011 commentary from The Guardian, ‘the genetic variation confidently expected by medical geneticists to explain common diseases, cannot be found’ (9). This commentator goes on to suggest that ‘[t]he failure to find meaningful inherited genetic pre-dispositions is likely to become the most profound crisis that science has faced’ (9). While some of the critiques found in the popular press are probably unfairly grandiose and pessimistic – translating basic research into clinical benefic takes time, after all – similarly themed commentary can be found in the scientific literature (10–12). The bottom line: from a health perspective, most genetic risk information is not very useful.

This reality has created a fascinating policy paradox. Just as the benefits of genetic testing may have been oversold, so have the potential harms. Much of the justification for the regulation of direct-to-consumer (DTC) genetics tests, for example, is based on the idea that genetic information is powerful and profound and, as a result, access to that information requires careful oversight. But just as emerging research is illuminating the predictive limits of genetic risk information, it is also suggesting that the public’s response to DTC results is much more complex and, in general, more muted than anticipated. For instance, recent research has shown...
that, contrary to conventional wisdom, receiving genetic risk information does not cause anxiety (13, 14). In fact, it does not do much of anything (15). It does not motivate healthy behaviour change (16). It does not cause a fatalistic response (17). It does not even cause an increase in the use of screening tests (14).

If, as many in the scientific community are now saying, genetic information is not the oracle of our future health as we were once led to believe, and if access does not, for most, cause harm, why regulate the area? Why worry about shoddy direct-to-consumer genetic testing companies? Why not let DTC companies sell, say and do whatever they want?

The Canadian College of Medical Geneticists (CCMG) Policy Statement on DTC Genetics Testing (18) does not specifically engage these questions, but the answer (the correct one, I think) emerges from the Statement’s central message: The information that is conveyed to the public about genetics via marketing and to those who access DTC tests should, at a minimum, be accurate. While there is as yet little evidence to support many of the speculative harms outlined in the Statement (such as the concerns about anxiety), all can agree that the distribution of misinformation about the value and clinical utility of genetic test results is a bad thing. Indeed, this is a theme in almost every policy that has tackled the DTC testing issues (19–21).

There are, of course, many reasons to promote the provision of accurate tests and accurate information, including facilitating autonomy, consumer choice and informed public debates about the benefits, harms and limits of this emerging technology. There is also reason to believe that the circulation of inaccurate information could exacerbate the possible health policy problems implicated with DTC testing. Research tells us that the individuals that utilize DTC tests view the results as health information (22). Consequently, the users of these services may take the results to a healthcare professional for interpretation. This seems particularly likely if DTC marketing exaggerates the health value and downplays the limits of the tests, which, research tells us, is the norm (23). As noted in the CCMG Statement, this could create a drain on the public system, resulting in little health benefits and, even, the possibility of follow-up-induced iatrogenic injuries. These problems are compounded if the test results are inaccurate or if the primary healthcare provider is not equipped with the skills necessary to interpret results. Unfortunately, both of these scenarios seem all too common (21, 24).

Ironically, the CCMG recommendations are needed not because genetic information is of a highly sensitive and harm-inducing nature, but because the value of genetic risk information has been exaggerated by decades of promotional rhetoric (25). The purveyors of DTC genetic testing services have leveraged this hype to create an industry built on services of dubious worth and, for many DTC tests, questionable scientific legitimacy (26). The implementation of CCMG recommendations, such as those calling for high quality information and responsible marketing, would help to moderate the hyperbolic messaging.

There are three important caveats to my claim that the recommendations are needed largely because DTC

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Table 1. Examples of ‘genetic revolution’ newspaper headlines

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<tr>
<th>Past optimism</th>
<th>Present pessimism</th>
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<tr>
<td>• ‘On the Frontier of the Genetic Revolution/How DNA breakthroughs are creating a new level of medicine’, San Francisco Chronicle, 21 May 1995</td>
<td>• ‘Genome yet to deliver golden era of medicine’, Townsville Bulletin, 3 April 2010</td>
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<td>• ‘Scientists pore over ‘Book of Life’ secrets; Deciphering code will pave way for genetic revolution’, The Toronto Star, 12 February 2001</td>
<td>• ‘Ten years on, the benefits of decoding the human genome still lie ahead’, The Independent, 26 June 2010</td>
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<tr>
<td>• ‘Mapping genome may revolutionize treatment for diseases, addiction’, Kitchener-Waterloo Record, 12 February 2001</td>
<td>• ‘The unveiling of the first draft of our genetic code in 2000 was a milestone – but has medicine changed as I predicted it would?’, The Daily Telegraph, 21 September 2010</td>
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<td>• ‘Genetics: The Future Is Now’, Time Magazine, 17 January 2004</td>
<td>• ‘Did they oversell the genomic revolution? A decade after the first published results of the human genome project, the hoped-for flood of new therapies is still a trickle’, Globe and Mail, 18 February 2011</td>
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<td>• ‘Genetics called key to medical revolution’, Arizona Daily Star, 8 April 2004</td>
<td>• ‘Why the gene revolution has been postponed; It costs $1bn to develop a new drug, so don’t expect personalised treatments. But the genome project is still worthwhile’, The Times (UK), 25 August 2011</td>
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tests have little health value. First, some genetic risk information is highly predictive, of course. There are monogenic diseases, for example, for which a genetic test result is relatively definitive. Second, though research tells us that access to DTC results do not, in general, cause undue anxiety, there may be a cohort of individuals that do have an adverse and unhealthy reaction. And third, it seems inevitable that extremely cheap DTC whole-genome sequencing will soon be available, thus providing consumers with even more genetic information of varying degrees of predictive value. These are three additional reasons to call for oversight or, at least, additional research on the impact of the DTC industry (e.g. how many Canadians are learning about monogenic disease predisposition or carrier status via DTC testing?).

Still, the need for a clearheaded assessment of the limited health value of DTC testing remains the core reason that the CCMG recommendations are timely and appropriate. The recommendations may have been triggered by concerns attached to a now fading belief in a genetic revolution, but, somewhat paradoxically, the recommendations still hit all the right notes. At this moment in time, when the pendulum is swinging (sensibly) away from overly optimistic portrayals, a call for truth, accuracy and moderation seems just right.

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References

24. Sample I. Genetics tests flawed and inaccurate, say Dutch scientists, investigation found they gave wildly different results and arrived at predictions that were no better than flipping a coin. The Guardian, 30 May 2011. Available at: http://www.guardian.co.uk/science/2011/may/30/genetics-tests-flawed-dutch-scientists?INTCMP=SRCH. Accessed on October 31, 2011.