Direct-to-consumer genetic testing: driving choice?


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Direct-to-consumer genetic testing (DTC-GT), which includes both the advertising and offer of genetic tests directly to the public [1], existed for many years before the, now famous, companies such as deCODE and 23andme made their appearance on the market [2]. However, the sharp increase in companies offering DTC-GT over the last 3 years has sparked much regulatory and ethical debate [3–5]. The former, rightly concerned with quality and safety, and the latter, with consumer understanding and the absence of medical oversight. Even more speculation surrounds the larger socio-political issue of whether this is an enabling, democratization of medical information [6] and so just another example of modern, social networking, with genetic information as the common currency.

A short summary of DTC-GT, of the socio-ethical parameters and of recent regulatory approaches reveals that the commercialization of genetic testing is not likely to disappear. The same approach to privacy that sees individuals posting their personal genomes on the web characterizes the social networking potential of these commercial sites [6], where clients can not only receive their probabilistic genetic futures (however inaccurate and misunderstood) but also share this lottery with other consumers if they so choose. As with patients [101], asymptomatic genetic voyeurs can connect to like-minded consumers. Will such informed consumers spell the end of medical paternalism, or will the associated ethical and regulatory concerns be transferred to health professionals who, in the future, may well themselves become the conduits for such tests?

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The sheer number of genetic tests now on offer from over 30 companies with a large range of purposes (from health-related for both monogenic and complex traits, to preconceptional, to measuring athletic performance, intelligence and ancestry) [102], has contributed to the complex debate. With respect to the clinical validity and utility of tests, this number makes an adequate analysis of each of the tests practically impossible within the near future. Furthermore, issues can rapidly become confusing with such a large variety of tests; for example, would it be wise to have the same regulations for ancestry testing as for health-related genetic tests? Furthermore, even within the category of health-related tests, the concerns are not the same for all health conditions (e.g., cancer vs gluten sensitivity) and as such, the imposition of a one-size-fits-all approach may not necessarily be the best path to follow.

Socio-ethical

The most common socio-ethical concerns include the lack of both genetic counseling and health professional involvement [7,8].

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The sample banking and data privacy aspects are equally vague in terms of ownership, duration, confidentiality, bankruptcy, sale of the company [9] and the sharing of information with friends and family [10]. The impact on the traditional medical system with the worried healthy population asking for needless procedures is difficult to estimate [5], particularly for universal healthcare systems. Even if one day, clinically significant and accurate information were to become available, how would this be communicated and by whom? If information is familial, what about minors and children, where the general rule is no predictive genetic testing of minors in the absence of direct benefit, that is, treatment or prevention [11]? Furthermore, little is known concerning the extent of research conducted by DTC-GT companies and what ethics review, consent and monitoring are in place [9]. In short, while medical and research ethics have generally become well defined and integrated into biomedicine, a Klondike mentality still prevails in the private DTC-GT sector. Nowhere is this more evident than in the patchwork of regulatory approaches.

Regulation
At the national level, consumer protection laws would generally apply and specific prohibitions of DTC are still relatively rare. Indirectly, the 1997 Oviedo Convention of the Council of Europe (while not envisioning DTC-GT per se) limits predictive genetic testing to health purposes or for scientific research linked to health purposes, and subject to genetic counseling (art. 12; emphasis added) [103]. In its 2008 additional Protocol on genetic testing for health purposes, the requirements of scientific validity and clinical utility were added, as well as the need for individualized medical supervision [104]. Moreover, if considered low risk, genetic tests are not subject to premarket review [105], as per the In Vitro Medical Devices Directive 98/79/EC of the EU [106]. The Human Genetics Commission of the UK has recently finalized its ‘framework of principles’ on DTC-GT. This document has the value of having been written with the input of many stakeholders, including industry, policy makers and scientists, and it aims to: ‘promote high standards and consistency in the provision of genetic tests amongst commercial providers at an international level...’ [107]. The European Society of Human Genetics (ESHG) also recently issued a guidance document on DTC-GT for health purposes, in which it recommends ensuring the quality of the testing services, the provision of pre-test information and genetic counseling, a face-to-face consultation, absence of conflicts of interest and oversight of this industry [108].

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In the USA, 25 states and Washington, DC allow DTC-GT, 13 states categorically prohibit DTC-GT, while 12 states only allow it for some types of tests [109]. That being said, 2010 has seen an unprecedented interest in DTC-GT by US regulatory agencies. Between May and July 2010, the US FDA sent letters to at least 20 DTC-GT companies telling them that their genetic tests are considered medical devices and, as such, the companies would need US FDA approval before being able to continue to market their products [12, 10, 111]. Moreover, in mid-July 2010, the FDA held a public meeting on the oversight of laboratory-developed tests (LDTs) [112]. The issue of (lack of) oversight of LDTs or ‘home brews’ is closely related to that of DTC-GT since many of the tests offered by DTC-GT companies could be considered LDTs. Until now, the FDA did not require that most LDTs be reviewed for clinical validity, the exception being those genetic tests that produce a result "for the purpose of diagnosing, treating, or preventing disease" (such as breast cancer and prostate cancer) [113]. Immediately after this FDA meeting, the Committee on Energy and Commerce held a hearing – titled ‘Direct-To-Consumer Genetic Testing and the Consequences to the Public Health’ on 22 July 2010 [114], during which a report was presented by the US Government Accountability Office entitled ‘Misleading Test Results Are Further Complicated by Deceptive Marketing and Other Questionable Practices’ [115]. Indeed, all these events are suggestive of a forthcoming increase in regulatory oversight of DTC-GT in the USA.

Health professionals
Even if this ‘democratization’ of genetic testing were to be properly framed in the future so as to offer consumer protection in the market place, little attention has been paid to another venue of facilitation of access by citizens. What if DTC-GT companies were to continue to focus their marketing efforts on consumers while also increasing direct-to-physician advertising? This way, even if regulations were put into place whereby GT must be administered via a physician, by advertising to both groups, companies could potentially assure consumer demand and obtain physician collaboration. Take, for example, Counsyl, which advertises as prominently to physicians as to consumers on its principal website page [116]. This company offers preconceptional carrier testing for more than 100 genetic conditions [117]. Although gaining access to consumers via physicians may appear on the surface to ‘fix’ the problem of the lack of healthcare supervision or gate-keeping, the fact is that companies could still, in essence, be driving consumers toward their products by creating a needless demand. Consumers would then demand these genetic tests from their physicians who, based on studies of advertising of pharmaceuticals, are likely to deliver them [13, 14]. Another way companies could increase their market share would be to establish collaborations with individual clinician–researchers or entire research-clinics, such that the companies would offer genotyping services in exchange for the ‘communication’ of their genetic tests to patients in an invitation letter from their physicians, thereby automatically gaining the trust relationship already established between the patient and the doctor. A parallel can be drawn here with current constraints on drug marketing to physicians [15], so prospective policy making and regulation may be a welcome route. Furthermore, with the advent of whole-genome sequencing (soon to be affordable), the entire genome is out of the bottle [16]. The issue then is that of personal autonomy within a regulated environment that offers safe, quality testing whatever the approach will be: open market, regulations or integration into medical practice.

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Both physician paternalism and personal privacy may well be dead, the former due to the accessibility of information and the latter due to social networking. What we have is the potential for a curious hybrid context: the market-‘informed’ consumer in a physician–patient relationship governed by traditional deontological norms (if not laws) that presume an ongoing relationship of privileged, confidential exchange and communication but where the physician is still presumed to be an ‘expert’. Are physicians sufficiently genetic–genomic savvy? While there is no doubt that the physician–patient relationship is changing to become more consensual and communicative, the advent of whole-genome sequencing in the hands of the private sector (for now) does not bode well for the integration of genomics into medicine and public health. At the heart of this issue on the translation of genetic tests from the laboratory to the consumer market to the physician–patient relationship, is the fact that there is no such control or oversight in the USA, Canada and many European countries for the transfer of genetic tests from the laboratory to the traditional clinical setting. There are guidelines, such as the ACCE framework [108], but these do not make up for the lack of a real review of genetic tests performed before they are introduced in the clinic [109].

Conclusion
Calls for a statutory framework for genetic technologies are not new; in 2001, Zimmern and coauthors stated that: “All countries must consider introducing statutory frameworks to regulate genetic technologies. These frameworks should be robust enough to protect patient and public interests but balanced enough to allow new developments, including predictive tests, gene therapies and reproductive technologies” [17]. More recently, the Foundation for Genomics and Population Health published an independent response to the House of Lords Science and Technology Committee report in the UK [120], which also emphasizes the need for increased oversight of genetic testing and serves as a reminder that the illusion of expanded ‘choices’ for consumers may just be a mirage:

“Proven clinical utility and cost–effectiveness should drive the strategy for implementing genomics in the NHS [and] an appropriate infrastructure urgently needs to be established to ensure that the clinical benefits of genomic information can be realised. Specifically, a system is needed to prioritise, commission, generate and evaluate data on the clinical validity, clinical utility and cost–effectiveness of different tests and technologies. We cannot overemphasise the importance of, and urgent need for, evidence relating to molecular tests and analyses; the lack of a system for generating data and evaluating tests not only prohibits evidence-based decision making within the NHS, but also presents a barrier to innovation and realising investment within the diagnostics industry”[120].

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