BMJ

BMJ 2011;342:d2317 doi: 10.1136/bmj.d2317

EDITORIALS

Direct to consumer genetic testing

Regulations cannot guarantee responsible use; an international industry certificate is needed

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Genetic tests sold on the internet often claim to profile a person's risks for a wide range of diseases, including diabetes, Parkinson's disease, and certain cancers.[1] The DNA sample is usually taken at home by the consumer swabbing her or his cheek and sent to the laboratory by mail. Results may be communicated over the telephone, by post, or electronically. In 2009, the *BMJ* published a clinical review that recommended caution when clinicians are asked to interpret test results in patient consultations.[2] The recent widening of applications and easy access to direct to consumer genetic tests is likely to increase the number of patients who see their doctor because of a test result that indicates increased risk of disease.[3]

Urgent calls for regulation of direct to consumer genetic tests have come from a wide variety of international policy arenas as the industry offering these services expands.[4 5] In 2010 the Australian National Health and Medical Research Council published a report on medical genetic testing for health professionals, which acknowledged many problems with direct to consumer tests, but also noted that as market products they cannot be banned.[6] In the United Kingdom, the Human Genetics Commission has published a framework of ethical guidance that tackles informed consent, marketing, risk communication, availability of counselling, and data protection for direct to consumer genetic testing.[7] In the United States, the Food and Drugs Administration has looked into the practices of US based direct to consumer companies, and, at its most recent meeting in March 2011, its advisory committee on molecular and clinical genetics recommended involvement of doctors and that "specific tests should be offered solely on prescription." The problem with these regulatory approaches is enforcement. A globally acting, internet based industry cannot be forced to comply with laws or regulations that are binding only country by country.[8]

The validity and clinical utility of the tests currently available varies greatly.[9] Although results given by some tests may be well founded, other genetic testing products have been described as scientifically meaningless.[5] Many of the tests have not been evaluated clinically.[10] Most genetic variants included in direct to consumer test panels are associated with multifactorial complex conditions of low predictive value.[8 11] Serious criticisms have been made about dubious usage of secondary data without informed consent, as well as the lack of professional advice on how to interpret the genetic test outcomes and how to integrate them meaningfully into daily life and the image of the past and future self.[5 12]

However, it would be unhelpful to dismiss these tests out of hand, because consumers will continue to use them and they can help to identify some risk factors, particularly monogenetic variations. In some cases the tests have alerted patients and doctors to certain conditions. In a US study from 2010, 133 specialists in clinical genetics reported on patients who came to them as a consequence of direct to consumer genetic testing, either self referred or physician referred. Over half of the professionals judged the tests as potentially clinically useful, in particular tests for *BRCA1* and *BCRA2* (87%), which were far ahead of all other genetic tests (37%).[3]

Clear criteria are needed for health professionals and consumers to identify clinically useful tests of good ethical standard. Quality assurance of direct to consumer testing requires a type of rigorous control of these genetic tests that is not bound to national laws or regulations. An international product quality certificate that controls for compliance with ethical standards, provisions for counselling, and stringent standards of scientific validity could fulfil this task. Obtaining such a product quality certificate would be voluntary, yet should bring market advantages. International Standards Organisation (ISO) certificates, for example, are well established instruments for product quality. A similar modus of standard setting for direct to consumer genetic tests would benefit consumers and the health sector.

The availability of certified products would enable consumers to choose tests that they know meet high scientific and high ethical standards. An ISO-type quality assurance certificate should present a clear market advantage for the companies holding it.

However, product accreditation would not solve all the problems arising from the development of direct to consumer genetic tests. The coexistence of a second tier market of uncertified products of low quality cannot be prevented, and products and practices that are outlawed in some countries—such as genetic testing for prenatal sex selection or secret paternity tests—may boom in this second tier market. Yet, for healthcare professionals

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an easy system of recognising uncertified genetic testing products would render low quality tests a marginal problem. The certification would present a viable method to decide whether the test a patient used can be taken seriously. Such practice in the medical sector would align with the practice in judicial and forensic systems of only accepting genetic test results from certified laboratories, but it would also exceed their local limitation by using a globally recognised identifier for quality.

Ultimately, the means to discipline the use of genetic testing within the bounds of respect for human rights, especially of individual and group self determination, can only come from a global debate on values and rights. Regulations, laws, and certificates cannot guarantee responsible use, and thus a quality certificate for direct to consumer products seems the most helpful instrument, in lieu of such political debates.

Competing interests: The author has completed the Unified Competing Interest form at www.icmje.org/coi_disclosure.pdf (available on request from the corresponding author) and declares: no support from any organisation for the submitted work; no financial relationships with any organisations that might have an interest in the submitted work in the previous three years; no other relationships or activities that could appear to have influenced the submitted work.

Provenance and peer review: Commissioned; not externally peer reviewed.

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Cite this as: *BMJ* 2011;342:d2317