Ethics series — 2

Direct-to-consumer genetic testing — a regulatory nightmare?

Will the current framework protect consumers effectively?

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The age of personalised medicine has seen the rapid emergence of a direct-to-consumer (DTC) genetic testing industry.[1] While various forms of DTC testing have been available for many years, the emergence of DTC genetic testing is raising new concerns relating to the accuracy of predictions, and potential harms to consumers given there is typically no individualised genetic counselling.[2] DTC testing also has the capacity to increase pressure on an already overstretched health care system if confused consumers seek assistance from health practitioners in interpreting test results.[3]

In Australia, a number of companies advertise genetic testing directly to consumers. While some require that a health professional orders the tests and communicates results to the consumer, others offer unmediated services. Internationally, private companies are entering the DTC genetic testing market in increasing numbers. More likely than not, Australian consumers are responding to online advertising by these companies and sending their tissue samples for analysis overseas.

There is a growing body of academic commentary internationally calling for more stringent regulation of the industry.[4,5] In many countries, genetic tests are already included within regimens regulating therapeutic goods, in the form of in-vitro diagnostic medical devices (IVDs).[4] However, there is ongoing debate as to whether such regimens adequately regulate DTC testing, and a lack of consistency in regulatory approaches between countries, even within Europe.[6] In 2003, the Australian Law Reform Commission and Australian Health Ethics Committee concluded in *Essentially yours*, the report of their inquiry into the protection of genetic information, that there are "strong arguments for regulating the supply, directly to the public, of products used in some forms of genetic testing".[7] *Essentially yours* also canvassed the difficulties associated with regulating foreign companies offering DTC genetic testing through the internet.[7]

The Australian *Therapeutic Goods Act 1989* (Cwlth) (the Act) applies to all therapeutic goods imported into, supplied in and exported from Australia. In 2002 a new regulatory framework was established for medical devices through the *Therapeutic Goods (Medical Devices) Regulations 2002* (Cwlth) (the Regulations). Amendments to the Regulations in 2010 introduced an IVD regimen for genetic tests.

In this article we provide a brief overview of how this new regimen regulates IVDs, and some thoughts on its likely effect on DTC genetic testing. It is not our intention to add to the extensive debate surrounding the ethical and legal implications of DTC testing, or to take a position on the appropriate regulatory response.

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Therapeutic Goods Act and in-vitro medical devices

The new framework was designed to ensure that all IVDs supplied in Australia, with a few limited exceptions and exclusions, are registered on the Australian Register of Therapeutic Goods (ARTG). IVDs are defined broadly in the Regulations, embracing any medical device used to examine specimens derived from the human body for therapeutic purposes. This definition excludes IVDs used for testing parentage or for detecting the presence of drugs in samples from sportspeople.[8] However, genetic tests used for any health-related purposes fall within the definition, whether for detecting disease, predisposition to a particular condition or even for nutrigenomic purposes.[9]

IVDs are classified according to a four-tiered risk-based system, with Class 4 IVDs posing the greatest risk to public or individual health. All genetic tests are Class 3 IVDs and are required to comply with essential principles relating to quality, safety and performance. There are three categories of IVDs: all IVDs that are intended for therapeutic use, in-house IVDs and IVDs for self-testing.[8] In-house IVDs are for use specifically within laboratories. Reagents, calibrators and other equipment and materials used in DTC testing all seem likely to fit within this definition. Conversely, specimen receptacles provided to consumers fall outside the in-house category. As a general rule, these receptacles come within the broad low-risk category of Class 1 IVDs. However, receptacles used in DTC testing appear to also fall within the definition of self-testing IVDs, which includes IVDs intended to be used "in the collection of a sample by a lay person and, if the sample is tested by another person (eg, a laboratory) the results are returned directly to the person from whom the sample was taken".[10]

Regulatory developments, which we discuss in the next section, lend weight to the argument that the drafters intended to classify DTC receptacles as a form of self-testing IVD; however, whether this is the case remains uncertain.

Prohibition on self-testing in-vitro devices

Although the Act allows for certain self-testing IVDs to be included on the ARTG, the 2010 *Therapeutic Goods (Excluded Purposes) Specification* (the Specification) prohibits the supply of self-testing IVDs used for four specific purposes, including genetic testing for the presence of or susceptibility to serious diseases.[11]

This does not affect genetic testing mediated by health professionals, as devices used for this purpose do not come

within the definition of self-testing IVDs. However, the specific inclusion of genetic testing raises the question of whether the new regimen was intended to prohibit DTC testing in Australia. Unfortunately, the Explanatory Statement to the Specification is quite vague regarding its rationale and proposed effect.[12]

Prohibiting the supply of self-testing IVDs in Australia

Although the 2010 Specification prohibits registration of self-testing IVDs for genetic testing purposes, it does so only if the IVD is used *exclusively* for the listed purpose.[11] Many companies that supply IVDs to consumers for DTC genetic testing provide other related services, such as ancestry, parentage, nutrigenomic or dietary testing. Thus, one saliva sample can provide the customer with both disease susceptibility and ancestry information. Moreover, some DTC testing companies state that they do not test for disease susceptibility, but rather, their services are for informational purposes. It seems likely, then, that the prohibition in the Specification could be avoided relatively easily.

Prohibiting the import and export of self-testing IVDs

Under section 41MI of the Act, it is a criminal offence to import or export an IVD that has not been included in the ARTG. Potentially, this provision could make consumers of foreign DTC services liable on the basis that they are directly involved in the import and export of specimen collection kits. However, under item 1.1 in Schedule 4 of the amended Regulations, a medical device that is imported into Australia is an "exempt device" where it is "for use in the in vitro examination of a specimen obtained from the importer or a member of the importer's immediate family".[13] Item 1.2 in Schedule 4 of the amended Regulations provides that a medical device exported from Australia is similarly exempt provided *inter alia* that it "is not intended for commercial supply" or "for use for experimental purposes on humans".[13] These provisions seem to protect Australian consumers of overseas DTC genetic testing services from criminal liability. In France, in contrast, consumers face criminal liability for requesting genetic tests "outside the conditions laid by the law".[6]

Thus, it appears that the Specification will either have no effect on DTC companies, whether Australian or not, or it will apply discriminately to Australian DTC companies despite the absence of any relevant difference in the services offered by them and those offered by overseas companies.

Regulating in-house genetic testing

Come what may, the 2010 amendments to the Regulations will result in more stringent regulation of Australian companies offering genetic testing services because of the requirement that in-house IVDs be included on the ARTG (although this requirement does not come in force until 1 July 2014). Schedule 3 of the Regulations provides detailed information on the requirements imposed on manufacturers of certain classes of in-house IVDs.

As such, although it is unclear whether or not Australian-based companies are prohibited from offering DTC genetic testing, they will be scrutinised more closely come mid 2014. However, the issue of how to regulate foreign providers of DTC genetic testing services remains unresolved. One option that was canvassed in *Essentially yours* was the enactment of federal legislation, similar to that in place for offensive material and interactive gambling, to regulate advertising of DTC genetic testing on the internet. However, it was ultimately concluded that it would be premature to implement a similar regimen at this stage.[7]

Conclusion

The growing chorus of concerns about an unregulated DTC testing industry makes it increasingly difficult to argue against some form of regulation. It is unfortunate that the new Australian regimen for regulating IVDs tends to err on the side of opaqueness. Moreover, although the Australian regulatory regimen was intended to be compliant with international norms, until there is global harmonisation, the Australian regimen is likely to be as ineffective as the regimen established to deal with offensive content online. Short of restricting access to certain internet content in Australia, no further means for regulating offshore testing have been canvassed to date. It seems timely to explore the applicability of consumer protection and other laws more fully, and to encourage involvement of bodies such as the Australian Competition and Consumer Commission. In parallel, further work needs to be done to improve consumer education about genetic testing.

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