Position Statement

Title Direct to Consumer Genetic Tests Position Statement

Date October 2013

Purpose

The purpose of this position statement is to assist health professionals and consumers to consider the issues associated with direct-to-consumer genetic testing and to highlight the importance of genetic counselling in this process.

Background

This statement has been developed in response to growing public interest in genetic testing and an increase in online companies offering testing directly to consumers without the need for a medical referral or the involvement of a recognised health professional. It highlights issues and implications of such testing that consumers may need to consider.

Genetic tests provide individuals with information about their risk of developing certain conditions as well as their "carrier status" (the likelihood of passing these on to their children). These tests exist for a large number of conditions and the list is expanding.

In Australia, most genetic testing is offered in a healthcare setting such as a general practice or the clinic of a medical specialist or genetic service. Patients offered testing through a healthcare provider benefit from meeting with a clinical geneticist or genetic counsellor who will ensure that the test is appropriate and advise the patient of the implications that he or she — and his or her family — may face as a result of the information gained. The results of genetic tests can have significant implications for life insurance, employment, lifestyle and reproductive choices. They can also impact on family members found to be at risk of developing a condition.

Genetic tests usually require a sample of blood but can also be performed using samples of saliva or cheek cells, the latter taken by swab from the inside of the cheek. The sample — be it blood, saliva or cheek cells — is then analysed. This involves scanning for genetic markers that increase the individual's risk of developing a certain condition and/or their status as a carrier. Analysis can also predict an individual's likely response to certain medications.

In recent years an increasing number of web-based companies have been using the internet to market genetic tests directly to the public. The availability of these tests is growing rapidly. For less than $100, some will use saliva or cheek swab samples to test for more than 250 conditions. But the genetic markers these companies use are sometimes based on limited research, making the assessments inaccurate and unreliable.

Some companies offering direct-to-consumer tests make it a requirement of testing that the consumer have an over-the-telephone consultation with a certified genetic counsellor, either before or after the test. Others require that the test be administered in consultation with a

doctor. Most however, release results directly to the consumer, without counselling and without a doctor's consultation.

Often companies will store the samples and use them at a later date for further research, profiting in the process. Without appropriate counselling from a health professional consumers might not appreciate the full implications of undergoing such testing and being involved in research, an understanding needed to provide truly informed consent.

Issues

Numerous ethical issues arise when direct-to-consumer genetic testing is offered without the involvement of a health professional. These include that:

- individual consumers may not understand all the implications of testing and being involved in further research, such as on insurance, employment and their family
- the sheer number of conditions for which a test may check (in some cases more than 250) does not allow for informed consent
- direct-to-consumer companies fail to provide ongoing support and follow up
- the results of a test might have a significant impact on the consumer and necessitate clinical management (for example an individual who tests positive for the breast cancer gene will need to consult a physician)
- testing could have psychological implications for the consumer and members of his or her family arising from results indicating that they have an increased risk of developing a condition later in life, resulting in the "worried well"
- without proper counselling, individuals could be falsely reassured about their future and neglect important health checks
- most direct-to-consumer companies perform testing in laboratories outside of Australia, making it difficult to assess quality standards
- the accuracy and limitations of results might not be communicated effectively to consumers
- some tests could be based on limited research and provide misleading results
- some tests may be inappropriate for the individual
- testing could be provided to non-consenting individuals such as minors.

Recommendations

The Department of Health recommends that genetic testing be provided in a healthcare setting. Only in this way can consumers be assured appropriate counselling before and after the tests and, where necessary, the appropriate support, referral and clinical management. This will also ensure that all tests meet required standards of quality.