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## Position Statement on Genetic Testing 2012

### 1. Preamble

1.1 The AMA believes that genetic testing will increasingly play an important and vital role in mainstream health care in terms of preventive health and diagnosing and treating illness. Genetic tests might be undertaken for a variety of reasons including to confirm a diagnosis, to predict the risk of an individual developing a particular disease or disorder, to provide prognostic information or to provide a personalised diagnosis that can be used to tailor medical treatments to the individual.

1.2 While genetic testing benefits the individual for the reasons outlined above, genetic testing might also benefit the wider health care system through more cost effective treatment and improved patient outcomes. This might be attributed to earlier and more accurate diagnoses, greater efficiency in providing the correct treatment, reduced adverse events related to medication side effects and preventive health.

1.3 The shared nature of genetic information, however, poses ethical challenges to privacy and confidentiality that, along with the risk of genetic discrimination, need to be recognised and appropriately addressed.

1.4 The AMA believes there should be a national approach to the provision and regulation of genetic testing to ensure equitable and efficient access to safe, evidence-based genetic testing services throughout Australia.

1.5 The AMA advocates that there should be a national approach to raising public awareness of the risks and benefits of genetic testing including promoting reasonable expectations of the benefits of genetic testing as well as the shared nature of genetic information.

1.6 The AMA advocates that the medical profession and other health care professions must be appropriately educated and trained in relation to the clinical and social issues related to genetic testing.

1.7 The AMA advocates that there must be a sufficient health care workforce and infrastructure to support the use of genetic testing in mainstream health care.

1.8 The AMA advocates for an evidence-based approach to establish the accuracy and clinical utility of new genetic tests.

1.9 Genetic testing might be undertaken for a variety of reasons such as paternity testing, ancestry determination, forensic, and health care issues. This position statement focuses on genetic testing for health care issues.

### 2. Types of genetic tests

2.1 Genetic tests may be undertaken for non-familial, known as non-heritable, conditions and for familial, known as heritable, conditions.

2.2 A non-familial condition is one where the genetic variant associated with the condition is not heritable; therefore, the existence of the genetic variant in an individual has no particular implications for genetic relatives and future offspring. Most genetic testing of cancer is testing for non-heritable mutations in cancer cells. These tests assist with diagnosis, prognosis, and treatment of the cancer.

2.3 In relation to a familial condition, the identification of a genetic variant in one individual has implications for their genetic relatives and future offspring. This sort of genetic testing poses particular ethical challenges in relation to the right of genetic relatives to know, or not to know, the individual's genetic status.

2.4 There are a variety of genetic tests that are used in different contexts; for example:

#### 2.4.1 Genetic testing for a non-heritable mutation:

- This is also known as somatic genetic testing. This tests for mutations acquired in the DNA of somatic cells after conception (ie. non-heritable). Somatic genetic testing might be used in the management of cancer patients to confirm a diagnosis, assist in prognosis, and/or in treatment selection.

#### 2.4.2 Genetic testing for a heritable mutation:

- This is also known as germline genetic testing. This tests for mutations in the DNA of germ cells (ova and sperm), which are heritable;
- Carrier genetic tests might be used to determine if an individual carries a particular heritable mutation that might affect the health of future offspring. The presence of the mutation will not affect the health of the carrier but the mutation might be passed on to the carrier's offspring, which might result in the offspring developing the relevant condition. Carrier testing is often undertaken to assist individuals in making reproductive decisions;
- Diagnostic genetic tests might be used to confirm the diagnosis of a heritable condition in an affected individual;
- Pre-symptomatic genetic tests might be used to determine if an individual has inherited a mutation for a particular heritable disorder that the individual is highly likely to develop;
- Predictive genetic tests might be used to determine if an individual has inherited a mutation that increases the risk of an individual developing a particular disorder or disease;
- An individual's inherited genes are only one factor that contributes to their health. Other factors include environment, lifestyle, complex interactions between inherited genes, spontaneous mutations occurring during life, and chance. Genetic information is often about possibilities rather than certainties. Whilst an individual with an inherited mutation might be at higher risk for developing a particular disorder, that does not mean the disorder is certain to develop. The individual with the mutation can use that information to make lifestyle choices that might reduce the chance the disorder will develop, or allow for earlier detection of the disorder.

#### 2.4.3 Pharmacogenomic testing

- Pharmacogenomics involves determining how an individual's genetic makeup affects their response to a particular medication. Because many of the enzymes involved in drug metabolism have multiple genetic variants (alleles), pharmacogenomic testing can guide the prescription of drugs. It can allow selection of individuals who will benefit from a particular treatment and exclude individuals who will not benefit from that treatment or who are at high risk of adverse side effects. This allows individuals to receive the correct treatment sooner and reduces the chance of adverse side effects.

### 3. Access to genetic testing

3.1 There should be a national approach to the provision of genetic testing to ensure equitable access to safe, evidence-based genetic testing services throughout Australia.

3.2 There should be appropriate infrastructure to ensure that individuals have access to genetic testing, relevant health care professionals, pathology services, and counselling services regardless of where they live.

3.3 The current balance and mix of public and private services ensures Australia has a diversity of pathology providers with different areas of expertise that can provide a wide range of services. Neither the public nor private sectors have sufficient capacity to meet demand alone. To ensure equitable and affordable patient access, genetic testing services should be available from public and private pathology providers. This requires appropriate funding under the National Health Reform Agreement for public services and appropriate rebates for patients under the Medicare Benefits Schedule for private services.

#### 4. Consent and genetic counselling

4.1 As for all medical testing, genetic testing should only be undertaken with the consent of the individual or, in appropriate circumstances, the individual's surrogate decision-maker. There is no requirement for specific consent procedures for the somatic genetic tests (eg., for cancer diagnosis) or many of the diagnostic tests used to confirm clinical suspicion of heritable conditions (eg., for haemochromatosis). There may need to be written consent, however, where the genetic test has the potential to lead to complex clinical issues. This is the case for a severe disorder, particularly if the test is presymptomatic or prenatal and professional genetic counselling should precede and accompany the test. The provision of information should be test-specific and unhurried. This might involve explaining the intrinsic risks and clinical significance of a test; for example, whether a particular genetic test will be used to confirm a diagnosis, facilitate a prognosis, determine the risk of developing a particular disease; and/or assist in determining the most appropriate form of treatment. The provision of information should also include the possible social, psychological, physical and reproductive consequences of, genetic testing for the individual and, where appropriate, their genetic relatives.<sup>1</sup>

4.2 Unless specific disease intervention or prophylaxis is available, or it would assist the parents' role as the child's guardian, children should not normally undergo predictive genetic testing until they have reached the age of consent and so are able to request the test on their own behalf. Similarly, children should not normally undergo carrier testing until they reach the age of consent. A person consenting on behalf of a minor needs to consider the minor's right to know or not to know relevant genetic information if it relates to a disease likely to cause significant suffering.

4.3 Unless required by law, consent by living persons should normally be sought for incidental genotyping on their stored identifiable tissue and must be specific, contemporaneous and, where possible, in writing.

4.4 In certain circumstances, such as where an individual is considering undertaking presymptomatic genetic testing for a significant heritable disorder, pre-test and post-test genetic counselling should be offered. Genetic counselling involves supporting the individual (and family, where appropriate) through the decision-making process and through understanding the outcome and clinical and social implications of the test results, enabling the individual to reflect on their situation. As part of genetic counselling, it is important for individuals to understand how their information might be used and provided with information regarding the risks and benefits of testing or not testing based on fears of discrimination.

4.5 Genetic support groups and related community organisations play an important role in the community. They promote the interests and well-being of those affected by genetic conditions and provide information, resource materials, and other support services to affected individuals and their families as well as to health care professionals. Genetic support groups reflect the cultural sensitivities associated with many genetic conditions and the role of these groups should be respected.

4.6 It is now evident that incidental genetic information may be revealed by modern genetic testing technologies. Discussion of this information should occur with appropriate disclosure and referral for counselling, where indicated. As information on the genome and disease grows, it is likely that information considered unremarkable today could prove significant and clinicians and pathologists will need to put in place systems for recall of patients.

4.7 An individual has the right to withdraw from genetic testing and to require that their sample be destroyed at any time before the genetic test results are relayed to them. An individual also has the right to ask for a second opinion on the genetic test results.

4.8 Unless required by law, there should be no compulsion on any person to undertake or refuse any genetic test.

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<sup>1</sup> National Pathology Accreditation Advisory Council. *Classification of Human Genetic Testing (2007 Edition)*. A supplementary guide to Laboratory Accreditation Standards and Guidelines for Nucleic Acid Detection and Analysis.

## 5. Privacy of genetic information

5.1 Everyone is entitled to privacy and confidentiality of their personal health information; however, genetic information relevant to one member of a family might have implications for the health of other genetic relatives.

5.2 Genetic information acquired in the context of the doctor-patient relationship should not be disclosed to a third party without the patient's specific, and where possible, written consent unless undertaken in accordance with relevant privacy legislation. In the case of a positive test result that might have implications for third parties such as close genetic relatives, the individual tested should be encouraged to discuss the results of the test with relevant third parties. Privacy legislation allows health practitioners to use or disclose patients' genetic information, whether or not they give consent, in circumstances where there is reasonable belief that doing so is necessary to lessen or prevent a serious threat to the life, health or safety of their genetic relatives.<sup>2</sup>

5.3 Unless required by law, there should be no compulsion on, or coercion of, the person, the attending doctors or the staff of a genetics laboratory or register to acknowledge or in any other way to reveal that a genetic test has been undertaken, or to divulge the results of any test which might have been undertaken.

5.4 Governments, the criminal justice system, employers and insurers should not be authorised to compel patients to provide samples which would disclose genetic traits or disorders, unless legitimate public health interests outweigh an individual's right to privacy.

5.5 Unless required by law, consent should normally be sought from the executor or appointed administrator for genotyping the stored identifiable tissue of a deceased individual.

5.6 Genetic information about a deceased individual might have implications for the health, confidentiality, and privacy of living genetic relatives; however, there are special ethical and legal issues that arise in relation to the disclosure of the deceased individual's genetic information to a third party. Currently, the *Privacy Act* applies to living persons only. Genetic information about a deceased person might be subject to legal duties of confidentiality.

## 6. Genetic discrimination

6.1 Along with the benefits of genetic testing comes the possibility of genetic discrimination, where an individual, their family members, or community is treated differently, possibly unfairly, based on their real or perceived genetic makeup. Discrimination on the grounds of genetic characteristics is unacceptable. It is not consistent with the *AMA Code of Ethics* and with the ethical principles of the doctor-patient relationship for a patient's genetic characteristics to determine their access to medical care.

6.2 In order for the community to accept and embrace genetic testing, it's important to minimise the real and perceived risk of genetic discrimination; otherwise, individuals might forego beneficial genetic testing due to fear of discrimination.

## 7. Direct to consumer (DTC) genetic tests

7.1 DTC genetic tests are offered on a commercial basis directly to consumers, without referral or consultation with a doctor or other health care professional. DTC tests might cover testing for paternity, ancestry, and health matters.

7.2 DTC tests for health matters are particularly problematic as individuals might undertake a genetic test without fully understanding and appreciating the purpose and implications of the test. Further, the

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<sup>2</sup> National Health and Medical Research Council. *Use and disclosure of genetic information to a patient's genetic relatives under Section 95AA of the Privacy Act 1988 (Cth). Guidelines for health practitioners in the private sector.* 2009.

individual might receive test results that are inaccurate, contradictory, misleading, taken out of context and open to misinterpretation.

7.3 In the context of providing health care, genetic testing should only be undertaken with a referral from a medical practitioner. The AMA strongly encourages any patients considering using a DTC genetic test to discuss the risks and benefits of DTC genetic testing with their General Practitioner (GP) first. If a patient has already undertaken a DTC genetic test, the AMA encourages them to discuss the test results with their GP.

7.4 Many companies offering DTC genetic tests use overseas laboratories where it is difficult to assess the quality framework. Furthermore, these laboratories are often not subject to local clinical quality accreditation agencies, as they may not be legally deemed to be offering “medical tests”. The AMA supports the current system of accreditation (NATA/RCPA) as an essential part of ensuring quality standards across the industry.

7.5 In order to alleviate the potential negative impacts of DTC genetic testing, the AMA advocates that consumer awareness campaigns are used to highlight the risks of DTC genetic tests. Education initiatives should also be targeted at health care professionals, providing information on the risks of DTC testing and support for advising patients on the use and interpretation of DTC genetic tests.

## **8. Public education and awareness of the risks and benefits of genetic testing**

8.1 There should be public education and awareness campaigns that highlight the risks and benefits of genetic testing. Such campaigns should be comprehensive and objective.

## **9. Professional education and training in clinical and social implications of genetic testing**

9.1 There must be an adequate workforce and health care infrastructure to meet the demands for genetic testing.

9.2 All doctors should continue their education and training in genetic issues throughout their career, recognising that this cultural awareness is a rapidly moving field. Education and training should include the clinical, ethical, legal, and social implications of genetic testing, skills in genetic counselling, and innovations in genetic testing.

## **10. Workforce and infrastructure to support genetic testing**

10.1 Genetic testing and associated counselling in health care may require a multidisciplinary approach with a range of health professionals and community support groups.

10.2 There should be an appropriate spread of pathology services sufficient to provide safe, affordable, and timely access to quality genetic testing for all patients in Australia.

10.3 There should be appropriate workforce planning and investment in training and development to ensure an ongoing high quality genetic pathology-related workforce.

## **11. Research**

11.1 Research into genetic testing for health care issues:

- has the potential to benefit humanity;
- should be conducted under appropriate ethical guidelines;
- should be conducted in secure environments according to recognised international standards;
- should be applied to the prevention, management and treatment of disease;
- might be conducted in the absence of consent, if testing is part of a research project approved by a human research ethics committee on stored genetic material in a fully de-identified form.

## 12. Gene patents

12.1 The holding of patents should not infringe the principle that the human genome is the common heritage of humanity.

12.2 The holding of patents must not present an obstacle to the diagnosis, prevention, management and treatment of disease.

## 13. Genetic selection

13.1 The practice of eugenics violates human rights. In the absence of effective therapy, genetic testing of pre-implantation embryos or of a foetus should be restricted to fatal or seriously and permanently disabling diseases.

13.2 Genetic selection should not be undertaken on the basis of sex (except in order to avoid hereditary sex-related disease) or on the basis of characteristics or traits that are unrelated to disease.