Guidance for Health Professionals Obtaining Consent for Clinical Genomic Testing

Overview

The national model for consent to clinical genomic testing ('the model') has been developed to help health professionals and patients to understand the important issues that should be considered during the consent process.

The model applies to obtaining consent to genomic testing by an accredited laboratory for clinical purposes.

This guidance reflects current clinic and laboratory practice and allows sufficient flexibility for Australia wide adoption.

Note on terminology

Genomics: Consistent with the National Health Genomics Policy Framework, the term 'genomics' is used in these materials to refer to both the study of single genes (genetics) and the study of an individual's entire genetic makeup (genome) and how it interacts with environmental or non-genetic factors.

Patient: For brevity, the term 'patient' is used in these materials to refer to any person receiving health care. It also includes the relevant decision maker where the patient does not have capacity to consent.

Consent requirements

Consent for genomic testing follows the same established ethical and legal principles of consent for other clinical purposes.

Consent should be obtained by a suitably experienced health professional who understands the specific complexities and implications of the genomic test.

Consideration should be given as to when and how consent is sought from patients. Processes for obtaining consent to genomic testing must allow them sufficient time and opportunity for deliberation over the information provided and discussion of any questions arising.

Consent is to be given voluntarily, in the absence of coercion.

Clinical consent for genomic testing must be specific to the patient and to the genomic test/s at the time that consent is sought.

Consent to genomic testing must be given by a person with the capacity to consent, that is, a person with the ability to understand the implications of having the genomic test. This may be the patient or an authorised person in accordance with applicable legislation and policies.

Consent forms

Written documentation of consent, such as by completion of a consent form, provides a record of consent for genomic testing.

Provision of a consent form does not discharge the duty of the health practitioner to ascertain whether an individual understands the nature of, risks involved in, and potential outcomes of the genomic test.

Consent forms may be in both digital and non-digital modes.

A written consent form for genomic testing assists in maintaining accurate clinical records and supports practitioners in providing appropriate information to patients under their care, in line with community expectations and legal requirements.

A copy of the consent form should be provided to the patient and retained in the patient's clinical record in accordance with relevant legislation and guidelines, as applicable to the organisation.

Records of specific written consent may be requested by accredited Australian laboratories for tests where potential complex issues are associated with testing for the individual and/or other family members. Inadequate records of consent may lead to unnecessary delays in sample processing and return of results.

Information on laboratory requirements, including for tests with potential complex issues, is within "*Requirements for the Medical Testing of Human Nucleic Acid (Second Edition 2013)"* available on the Department's website at: www.health.gov.au and use the search term 'testing nucleic acids'

Supporting patient information

Pre-prepared material about the genomic test may be useful if given to the patient as a means of stimulating discussion and for guiding the health professional when informing them.

It may be useful if materials are provided prior to the appointment and made available again following the appointment. Materials may be presented in digital, non-digital or accessible multimodal format.

The national model for consent to clinical genomic testing includes a supporting patient information video and factsheet. The materials are multimodal and designed for broad utility for a range of clinical applications of genomic testing and may be used to complement specific patient resources. To access the patient information video and supporting factsheet, please visit the NSW Health Website at www.health.nsw.gov.au and use the search term "Patient materials for Clinical Genomic Testing".

Pre-prepared material does not substitute for the health professional discussing the nature, risks and potential outcomes of the genomic test with the patient.

Information provided to patients needs to be tailored to the individual. This includes consideration of that individual's Health Literacy, so that they can make an informed decision.

The health professional should assist the patient to understand the material and explain any information that they find unclear.

The health professional must give the patient sufficient time and opportunity to read the material and raise any specific issues or concerns, either at the time the information is given to them or subsequently.

Cultural and linguistic diversity

Consideration must be given to cultural and linguistic diversity, and the relevant implications for consent. Health practitioners should follow local policies and guidelines relevant to the delivery of health services to culturally and linguistically diverse populations.

An interpreter should be considered to communicate information to a non-English speaking patient. Health practitioners should follow relevant local policies and guidelines regarding the use of interpreters.

Aboriginal and Torres Strait Islander peoples

An Aboriginal liaison officer or other appropriate health worker should be considered to assist in consent processes for Aboriginal and Torres Strait Islander peoples. Health professionals should follow local policies and guidelines relevant to the delivery of health services to Aboriginal and Torres Strait Islander peoples.

Genetically, the Aboriginal and Torres Strait Islander population is the oldest and the most diverse in the world. The past colonial experience for Aboriginal and Torres Strait Islander people is marked with racism and discrimination, the stealing of children, loss of identity, knowledge, culture and land.

The storage, use and disclosure of genomics clinical data and information may uniquely impact Aboriginal and Torres Strait Islander Peoples and have possible impacts on the broader Aboriginal and Torres Strait Islander community.

Specific consideration must be given to the limited availability of genomic reference data for Aboriginal and Torres Strait Islander Peoples and the significant implications this has on the degree of certainty of results and the management of incidental (also called unsolicited / unexpected) findings.

The approach to obtaining consent should consider cultural practice, belief and support systems of Aboriginal and Torres Strait Islander Peoples. This will facilitate the right people to be part of the decision-making process, whilst maintaining patient autonomy and dignity.

Other communication difficulties (sensory and communication disability)

Where a patient has deafness, visual and/or hearing impairment or other special communication needs, appropriate actions must be taken to assist them to understand the nature of, risks involved in, and potential outcomes of the genomic test.

Health practitioners should follow relevant policies and guidelines specific to the delivery of health services for patients with disability or other communication needs.

Disclosure of results

Consent processes for genomic testing must include mechanisms to contact the patient with information regarding the findings. Health professionals must advise the patient of how results will be disclosed, particularly in the event of an adverse result.

Consent processes must include protocols for disclosure of sensitive genomic information, including familial risk and unexpected family relationships. This should also include consideration of the return of test results in the event of a patient's death.

Health professionals should inform the patient of possible results. This includes that the testing may find an answer to the clinical question, may not find an answer, could find results of uncertain significance, or could reveal additional (e.g. incidental/ unsolicited/ unexpected) findings that are not related to the primary reason for testing.

Health professionals should be aware of the testing laboratory's policy on the reporting of incidental/ unsolicited/ unexpected findings and results of uncertain significance. This will enable the health professional to inform the patient of the types of findings that may be reported, if any, and the likelihood of receiving such results. Where laboratory policy permits, health professionals may be able to offer patients a choice whether or not to receive such results.

NPAAC requires accredited laboratories to limit the reporting of incidental findings to variants that are unequivocally classified as pathogenic or likely pathogenic. Laboratories are also required to have a policy for variant reporting that considers the strength of evidence supporting the association between the variant and the clinical outcome of interest.

Information on laboratory requirements for reporting of results of uncertain significance and the management of incidental findings is within *'Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies (First Edition 2017)'* available on the Department's website at: www.health.gov.au using the search term 'genome testing'.

Implications for family members

An individual's genomic test result may also be important for the care of their genetic relatives. Consent processes should include consideration of the release of information to genetic relatives.

Support and guidance should be offered to the patient to assist them in determining the appropriate disclosure of information to genetic relatives. The patient should be offered a supported opportunity to self-disclose in the first instance.

Health professionals should be aware of the relevant guidelines for the disclosure of genomic health information, as applicable to their organisation. This will enable the health practitioner to inform patients of circumstances in which lawful disclosure of genomic health information may occur without their consent, such as to prevent a serious threat to the health of a genetic relative.

Health professionals should advise the patient that if this occurs, efforts will be made not to disclose the identity of the person tested.

Further testing and analysis

Consideration should be given to potential further testing and/or re-analysis of a sample and/or genomic data. This should be incorporated into an ongoing and supported conversation between the patient and health professional.

Health professionals should be aware of the testing laboratory practices, as some current bioinformatics pipelines may include automated re-analysis.

Data management

Health professionals must adhere to local policies and guidelines relevant to privacy, confidentiality and data management. Information from the health service and testing laboratory should be accessible to the health professional and the patient. This will assist understanding how a patients genomic material and data is used, shared, stored and protected.

The health professional should consider whether additional consent is required, as applicable to specific clinical tools used to deliver care, such as sharing and comparing phenotypic data in public databases. For example, consent to Matchmaker Exchange, in which data may be shared internationally and may result in the commercialisation of discoveries.

Research

The potential utilisation of specimens and/or genomic data for the purposes of research may arise during clinical care. The use of clinical test specimens and/or genomic data for secondary research purposes contributes to the advancement of clinical and scientific knowledge.

Consideration is required as to how consent for research purposes is obtained separately to that for clinical consent. There must be no inadvertent effect on the patient's understanding of the nature, risks and potential clinical outcomes of the genomic test.

Patients should be informed they may be contacted in the future asking to participate in ethically approved research into the same or related condition. Patients should be aware they can decline any such request.

Specific requirements for genomics research are within the *National Statement on Ethical Conduct in Human Research (2007)* available on the NHMRC website at: www.nhmrc.gov.au using the search term 'ethical conduct'.

Insurance

Currently, genomic testing will not alter the patient's ability to get health insurance or the patient's health insurance premiums.

Genomic testing could affect how easy it is for a patient or their genetic relatives to get income protection, travel or life insurance; or the price of their premium.

The 'Moratorium on Genetic Tests in Life Insurance' was agreed by life insurance companies that are members of the Financial Services Council. The Moratorium started on 1 July 2019 and lasts until at least 30 June 2024 and allows people to access a level of life insurance without being asked about the result of a previous genomic test. It does not apply to existing life insurance policies. It is unknown how genomic test results may affect applications for life insurance when the Moratorium expires.

Further information on the '*Moratorium on Genetic Tests in Life Insurance'* is available on the Financial Services Council website at www.fsc.org.au using the search term "moratorium key facts".

Support

Consideration should be given to the range of supports that may be required for patients throughout the genomic testing process. This may include patient referral to relevant health services including genetic counselling, and mental health; and linkage to support organisations and groups.

A comprehensive list of support organisations and support groups is available on the Centre for Genetics Education Website, www.genetics.edu.au using the search term "support".