HEALTH PROFESSIONAL GUIDE

OBTAINING CONSENT FOR GENETIC AND GENOMIC TESTING

This guide aims to support health professionals obtaining patient consent for genetic and/or genomic testing by an accredited laboratory for clinical purposes in Australia. This guide should be used in conjunction with the **Clinical Consent Form for Genetic and Genomic Testing** templates and **Patient Fact Sheet** developed by Australian Genomics (2022). These documents have been developed as flexible resources for broad adoption in clinical settings across Australia to standardise the consent process for all clinicians and patients.

If you have any questions about genetic or genomic testing,
please contact your local genetics clinical service.

genetics.edu.au/SitePages/Genetic-Services.aspx

This document discusses the information that is included in the documents named above, to assist health professionals in providing clear explanations of the purpose, potential benefits, risks and outcomes of genetic and genomic testing to patients in their care. This guide reflects current clinical and laboratory practice regarding obtaining consent, impact on personal insurance products, data sharing/storage/usage, caring for diverse communities, and using data for research purposes.

NOTE ON TERMINOLOGY:

The term '**genomic**' is used in this document to describe both genetic and genomic testing and types of information. There are separate consent form templates for genetic and genomic testing, and the difference is outlined in the **Patient Fact Sheet**.

The term '**patient**' is used to refer to any person receiving health care. It also includes the relevant decision maker where the patient does not have capacity to consent, e.g. their guardian.

CONSENT REQUIREMENTS

- Consent for genomic testing follows the same established ethical and legal principles of consent that govern all clinical practice in Australia.
- Consent should be obtained by a suitably experienced health professional who understands the specific complexities and implications of the genomic test that they are ordering.
- Consideration should be given as to when and how consent is sought from patients. Processes for obtaining consent for genomic testing should allow patients sufficient time and opportunity for deliberation over the purpose and possible outcomes of the test. Patients should be given the opportunity to ask questions and discuss any concerns with their health care provider and anyone else they deem necessary, such as family members.
- Consent is to be **informed**, and given **voluntarily**, in the absence of coercion.
- Clinical consent for genomic testing must be specific to the patient and to the genomic test/s being conducted.
- Consent to genomic testing must be given by a person with capacity, that is, a person with the ability to understand the implications of having the particular testing being offered. This may be the patient or an authorised person in accordance with applicable legislation and policies.

CONSENT FORM

Written consent forms for genomic testing assist in maintaining accurate clinical records, as well as supporting health professionals in providing appropriate information to patients under their care, in line with community expectations and legal requirements. They provide evidence that a consent process took place.

A copy of the consent form should be offered to the patient and retained in the patient's clinical record in accordance with relevant legislation and guidelines, as applicable to the organisation. Consent forms may be digital or non-digital. Inadequate records of consent may lead to unnecessary delays in sample processing and return of results.

Provision of a consent form does not discharge the duty of the health professional to ascertain whether an individual understands the purpose, risks, benefits, and potential outcomes of the genomic test.

The table below and on the following pages outlines the clauses of the Clinical Consent Form for Genomic and Genetic Testing templates to provide specific explanations for health professionals using these resources.

CLAUSE	EXPLANATION
The test does not detect all genetic changes or predict all possible health conditions.	Health professionals should inform the patient of the purpose of testing, the type of test and associated possible results. It is important for the health professional to set expectations about what the test may or may not reveal and to clarify why it is being offered. There are limitations to testing and all genetic variants may not be found. The **Patient Fact Sheet** provides a short definition for the different purposes of testing i.e., diagnostic, predictive etc.
Genomic Testing Consent Form Only The test may find a genetic change not related to the reason for testing ('incidental finding').	Health professionals should be aware that there is a possibility of an **incidental** finding. Where laboratory policy permits, health professionals may be able to offer patients a choice whether or not to receive such results. The possibility of incidental findings is explained in the Patient Fact Sheet. The National Pathology Accreditation Advisory Council (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.
The test may find a genetic change of uncertain significance.	Health professionals should be aware that there is a possibility of finding a variant of uncertain significance (VUS). A VUS means that at this point in time there is not enough evidence to say it is or is not the cause. Laboratory policies vary with regards to reporting VUS. Health professionals should be aware of these policies. The possibility of VUS is explained in the **Patient Fact Sheet.** Information on laboratory requirements for reporting of VUS 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: health.gov.au using the search term 'genome testing'.

CLAUSE	EXPLANATION
More tests or analysis may be needed to understand the results. This may include testing blood relatives.	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. While further testing may be suggested, it may or may not identify the cause. It is a possibility that the test may fail. This may require a new sample from the patient. You will be notified by the laboratory if this occurs.
The sample or results may be re-examined in the future using new knowledge of testing methods.	Patients should be made aware that the fast expansion of genomic knowledge allows for re-examination of data/samples when test results are inconclusive.
Results may have health implications for blood relatives.	An individual's genomic test result may also be important for the care of their blood relatives. Consent processes should include consideration of the release of information to blood relatives. Support and guidance should be offered to the patient to assist them in determining the appropriate disclosure of information to blood relatives. The patient should be offered a supported opportunity to self-disclose in the first instance. Patients should be made aware that blood relatives may be required to get testing to support their diagnosis.
Results may show unexpected family relationships.	Health professionals should make patients aware that unexpected family relationships, such as non-paternity, may be discovered in genomic testing.
Results may affect the ability to obtain some types of insurance.	Currently, genomic testing will not alter the patient's ability to get health insurance or the patient's health insurance premiums. It could affect how easy it is for patients and their genetic relatives to get income protection, travel insurance, or life insurance in the future (but not existing cover) and could impact the price of the premium for new products or different cover in the future. Until 2024, Australians can purchase new life insurance policies without the results of previous genetic tests being taken into account. However, an existing clinical diagnosis may affect an individual's ability to obtain these kinds of insurance. Industry regulation prevents insurers from asking relatives for genetic test results and from requesting people to have testing. Health professionals must not provide patient results to an insurance provider without patient/guardian consent. Further details can be found by searching for 'Moratorium' at fsc.org.au/resources
The sample will be stored and may be shared with other laboratories to assist with genomic testing.	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 in understanding how a patient's genomic material and data is used, shared, stored and protected, as well as destroyed once the legal storage period has ended. The patient should be made aware that their sample may be used for the purpose of quality control, internal validation, test improvement, and training purposes by the accredited laboratory that is organising the testing. This is part of usual testing processes and does not require additional consent.

CLAUSE	EXPLANATION
Results and related health information may be shared with genomic and medical databases that are used for patient care. All identifying information will be removed.	To enhance the understanding of human genetics, genomic information is shared with clinical databases and consent is not required for this. For example, ClinVar is a clinical database that collates information about genomic variation and its relationship to human health.
Results are confidential and will only be shared with consent, or as required or permitted by law.	Health professionals should be aware of relevant law, regulation, policy or guidelines regarding the disclosure of genomic health information, as applicable to them or their organisation. This will enable them to inform patients of circumstances in which lawful disclosure of genomic health information may occur without consent, such as to prevent a serious threat to the health of a genetic relative.
I can choose not to be told the results if I change my mind, but the report will remain in medical records.	The Patient Fact Sheet explains that results cannot be removed from patient medical records once documented. If the patient chooses not to be told results before the laboratory has recorded their findings, there may be a report documenting that testing had not been completed.

OPTIONAL CONSENT FORM CLAUSES:

CLAUSE	EXPLANATION
I consent to share the results and related information with health professionals to help with the genetic testing of blood relatives. I understand that identifying information will not be disclosed to the relative wherever possible.	Patients should be made aware that in some circumstances where there is the risk of serious and imminent harm to blood relatives, their genomic information can be shared with their blood relatives' health professionals without the patient's consent, under applicable law and regulations.
I consent to share the sample, genomic data, and related health information for ethically approved research into the same or related conditions. I understand identifying information will be removed and may be replaced with a unique code so that information can be returned to me in some situations.	De-identifying and re-identifying data for the purpose of research is explained in the Patient Fact Sheet. Patients should not expect to receive any personal results from such sharing as not all research projects return results to their participants.
I consent to genomic testing. I understand the reason for testing and the potential benefits, consequences, and limitations. I have been able to discuss the information with a health professional, ask questions and have any concerns addressed. I am satisfied with the explanations and answers to my questions.	Explanations of reasons for testing, potential benefits, consequences and limitations are outlined in the **Patient Fact Sheet**. The **Patient Fact Sheet** is not an exhaustive list of answers to the questions that patients might have and should not replace the pre-test discussion between a health professional and their patient.
Consent for Biological Parents Undergoing Duo/Trio Genomic Analysis	For Trio Analysis, each parent needs to sign a clinical consent form. This permits the parent's data and health information to be used for research and reanalysis only related to their child's condition.
Optional: Interpreter/Liaison Officer Signature	In some jurisdictions where an interpreter has been involved in the consent consultation, they are required to sign the consent form as well as the patient.

PATIENT FACT SHEET

The Patient Fact Sheet has been developed to assist patients in understanding the benefits, consequences, and limitations of genomic testing, as well as provide a resource to guide discussions with their health care providers and family members to decide whether to have testing or not. Health professionals must give patients sufficient time to read, understand and ask questions about this fact sheet before giving consent. Some patients may wish to delay giving consent until they have had time to give it further consideration.

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 that the result will impact the patient's health management.

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.

RESEARCH

The potential utilisation of sample 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 scientific knowledge. Health professionals should clearly differentiate obtaining consent for research from clinical purposes. There must be no (even 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 inviting them to participate in ethically approved research into the same or a related condition. Patients should be aware they can decline any such request.

CULTURAL, ETHNIC, AND LINGUISTIC DIVERSITY

Consideration must be given to cultural, ethnic and linguistic diversity, and the relevant implications for consent. Health professionals should follow local policies and guidelines relevant to the delivery of health services to culturally and linguistically diverse populations, including the use of interpreters for non-English speaking patients. Translations of the Consent Form templates, and the Patient Fact Sheet are available in multiple languages, including a basic English version.

PATIENTS WITH DISABILITIES

Where a patient has particular communication needs due to visual and/or hearing impairments, appropriate actions should be taken to assist them to understand the implications and benefits of genomic testing. Health professionals should follow the protocols of their particular organisation specific to the delivery of health care for patients with disabilities and/or communication difficulties and to ensure cultural safety and respect. People should be offered Easy Read versions of the materials

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 genomic 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 findings. Initiatives such as that of the Centre for Population Genomics in Australia are working to increase representation of First Nations peoples in reference databases.

The approach to obtaining consent should consider cultural practice, belief, and support systems of Aboriginal and Torres Strait Islander Peoples. This will facilitate the appropriate people to be part of the decision-making process, while supporting people to make a choice about testing that is consistent with their values, whether individual or cultural. This applies to all patients, regardless of ethnicity or ancestry.

PROFESSIONAL SUPPORT

If you have any questions about genetic or genomic testing, please contact your local genetics clinical service. genetics.edu.au/SitePages/Genetic-Services.aspx

For further reading, refer to this resource: Human Genetics Society of Australasia Position Statement: Use of Human Genetic and Genomic Information in Healthcare Settings - hgsa.org.au/documents/item/12121

PATIENT 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. There are many condition-specific support groups available. The organisations below may be able to direct patients to the most suitable support.

National

Rare Voices Australia
rarevoices.org.au

SWAN Australia
swanaus.org.au

Genetic Alliance Australia
geneticalliance.org.au
(02) 9295 8359

NSW

Centre for Genetics Education
genetics.edu.au

QLD

Support Groups Queensland
supportgroups.org.au

VIC

Genetic Support Network Victoria
gsnv.org.au
(03) 8341 6315

SA

Lived Experience Telephone Support Service
letss.org.au
1800 013 755

NT

Northern Territory Mental Health Line
1800 682 288

WA

Helping Minds
helpingminds.org.au
(08) 9427 7100

Mental Health Support

Lifeline Australia
24/7 Crisis Support
13 11 14

Beyond Blue
beyondblue.org.au
1300 224 636