HEALTH PROFESSIONAL GUIDE

OBTAINING CONSENT FOR GENETIC AND GENOMIC TESTING

Australian Genomics

This guide aims to support Australian based health professionals to obtain patient consent for genetic and/or genomic testing by an accredited laboratory for clinical purposes. 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 (2023). 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 using the Centre for Genetics Education's local service finder here.

Or contact the Human Genetics Society of Australasia (HGSA):
P: +61 (02) 9669 6602 | E: secretariat@hgsa.org.au

This document discusses the information that is included in the consent forms and patient fact sheet named above, to assist you in providing clear explanations of the purpose, potential benefits, risks and outcomes of genetic and genomic testing to patients in your 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.
- To obtain consent you should be suitably experienced and understand the specific complexities and implications of the genomic test that you 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 you 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 your 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 to provide 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 hardcopy. Inadequate records of consent may lead to unnecessary delays in genomic testing and return of results.

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

The following table outlines the clauses of the **Clinical Consent Form for Genomic and Genetic Testing** templates to provide specific explanations for you, when using these resources:

CLAUSE	EXPLANATION
The test does not detect all genetic changes or predict all possible health conditions.	You should inform your patient of the purpose of testing, the type of test and associated possible results. It is important for you 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. A variant may not be found for a number of reasons, including: • the variant/s causing the condition cannot be found by the test, • the gene causing the condition was not tested, • the gene causing the condition is not yet known, • the condition is polygenic, or • the condition is not genetic. The **Patient Fact Sheet** provides a short definition for the different purposes of testing (diagnostic, predictive etc).
Genomic Testing Consent Form Only **The test may find a genetic change not related to the reason for testing ('incidental finding').**	In genomic testing and analysis, there is a possibility of **incidental** findings. Where laboratory policy permits, health professionals may be able to offer patients a choice whether or not to receive such results. It is best to check local laboratory policies for guidance on the reporting of incidental findings as laboratories may have different policies. An example of an incidental finding is if a person has a genomic test for kidney disease and the test finds they are a carrier of a gene variant that causes cystic fibrosis (CF). Although CF is a genetic condition it is not a kidney condition. It is not related to the reason for testing. Finding the person is a carrier for CF was an incidental finding. 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.

CLAUSE	EXPLANATION
The test may find a genetic change of uncertain significance.	The genetic or genomic test may report a variant of uncertain significance (VUS). A VUS means that at this point in time there is not enough evidence to indicate it is causative of the clinical purpose of testing. Laboratory policies vary with regards to reporting VUS. You should be aware of these policies. The possibility of VUS is explained in the Patient Fact Sheet. Given the uncertainty of this result, it should be acknowledged that VUS can have an emotional impact on your patient. Adequate support should be provided if this is the case. 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 Australian Government Department of Health website.
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. The testing of blood relatives helps identify patterns of inheritance of the variant. The possibility of further testing should be incorporated into an ongoing and supported conversation between you and your patient. You should be aware of the testing laboratory practices. While further testing may be suggested, it may or may not identify the cause. It is also possible 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 rapid expansion of genomic knowledge allows for re-examination of data/samples when test results are inconclusive. Currently there is no standard process for the re-analysis of genomic data or samples. This process can be driven by the clinician, through re-referral or by the patient.
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. Refer to the first optional tick box (sharing of results for the healthcare of blood relatives). Patients should be made aware that blood relatives may be required to get testing to support their diagnosis. In some circumstances such as if there is imminent harm to their blood relative, you may need to disclose your patient's result, without their consent, to a blood relative. You should be aware of laws regarding this in your jurisdiction. These laws vary in different jurisdictions and differ between private and public settings.

CLAUSE	EXPLANATION
Results may show unexpected family relationships.	You should make your patients aware that unexpected family relationships, such as misattributed parentage, may be discovered in genomic testing.
Results may affect the ability to obtain some types of insurance.	Health insurance Currently, genomic testing should not alter a patient's ability to get private health insurance or how much the patient will pay for health insurance. However, a diagnosis of condition may mean that health insurance companies may apply waiting periods for treatment of the condition. Income protection, life insurance or travel insurance Genomic test results do not affect existing insurance cover. If a patient has pre-existing signs or symptoms, their presence may affect how easy it is and/or the cost for patients to get new income protection, life insurance or travel insurance cover in the future. In some circumstances, the genomic test results may also impact how easy it is and/or the cost for patients (and genetic parents, if undergoing trio testing) to get these types of insurances. Under the current industry-led moratorium, Australians can purchase new life insurance policies or increase their current level of cover without the results of previous genomic tests being taken into account, up to certain financial limits. Those limits are $500,000 for life cover and $4000/month for income protection. For cover over that amount (on an aggregate basis, including cover that is included in superannuation), individuals can be required to disclose, and insurers can use in making underwriting decisions, genomic test results that show increased risk. The moratorium prevents insurers from asking for the genomic test results of an applicant's relatives, and from requiring people to have testing. However, under the moratorium insurers can still use clinical diagnoses (whether made through a genomic test or otherwise) in underwriting. Patients can be encouraged to think about whether they want to organise life insurance cover before consenting to have their genomic test. More information on life insurance can be found on the Centre for Genetics Education website, **here**. See **here** for the Human Genetics Society of Australasia's Position Statement on genetic testing and insurance. It is the patient's obligation to provide genetic test information to an insurer where required, not the health professional's obligation. Health professionals must not provide patient results to an insurance provider without patient/guardian consent.

CLAUSE	EXPLANATION
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 you and your 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 storage period (as suggested by NPAAC guidelines) has ended. Refer to local guidelines and policies if a patient asks to have their sample destroyed earlier. 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. The sample may also be used as a positive control for the testing of blood relatives. This is part of usual testing processes and does not require additional consent.
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 an international clinical database that collates information about genomic variation and its relationship to human health and **Shariant Australasia** shares such information between laboratories and clinical services.
Results are confidential and will only be shared with my consent, or as required or permitted by law.	You should be aware of relevant law, regulation, policy, or guidelines regarding the disclosure of genomic health information, as applicable to you or your organisation. This will enable you to inform your 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 change my mind about testing and choose not to be told the results, but if testing has started, a report will remain in medical records.	The testing process can be stopped at any time, and patients can choose not to be told the results of the test. However, the report may remain in medical records depending on at what stage they make this decision. 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. If the laboratory has started analysing the DNA sample and your patient decides not to be told the results of the test, the laboratory's analysis will be entered into laboratory records, but the result does not need to be disclosed to your patient. Results cannot be removed if they have already been entered into the medical record. You should refer to local policies and guidelines in the case that the patient withdraws consent, which can happen at any time.

If your patient is 14 years or older and they have decision-making capacity, they can be considered a mature minor and can give their consent for a genetic or genomic test.

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.	When discussing the sharing of results for the healthcare of blood relatives, the following point should be communicated to your patient: if your patient's result shows that genetic testing is available to other family members, you should recommend that patients advise their relatives of this. Explain that health professionals will generally not contact relatives without patient permission. This option allows your patient to consent for their result to be released to health professionals caring for their relatives so they can access testing. 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**. Providing consent to share data for research purposes does not benefit the patient. 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.**	Further 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 you and your patient.
Consent for Parents Undergoing Duo/Trio Genomic Analysis	For Trio Analysis, each genetic parent needs to sign a clinical consent form. This permits the parents' 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. As the requesting healthcare professional, it is your responsibility to 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. You must advise your patient of how results will be disclosed, particularly in the event that the result will impact your 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. If you choose to discuss consent for research with your patient, you must 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 and that choosing not to participate in research will in no way impact their relationship with you, their health professional, nor in their clinical genetic testing. The discussion of research is at your discretion. You may feel that it is in the patient's best interests to discuss research post results delivery.

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** will be available in multiple languages in the near future via the Australian Genomics website.

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. You should follow the protocols of your 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 resources to ensure they have the opportunity to provide fully informed consent. Gene Equal has developed a toolkit for health professionals that can be found **here**.

ABORIGINAL AND TORRES STRAIT ISLANDER PEOPLES

You should follow local policies and guidelines relevant to the delivery of health services to 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.

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, and 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 Australian Alliance for Indigenous Genomics (ALIGN) and 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. Search for your local service **here**.

Further online information about genetic or genomics testing can be found here:

- **Centre for Genetics Education**
  - **Education for Paediatricians**
- **Melbourne Genomics Health Alliance**
- **The Royal Australasian College of Physicians**

Gene Equal have co-designed a toolkit for genetic health professionals caring for patients with intellectual disability. This can be found on their website, **here.**

For further reading, refer to this resource: **Human Genetics Society of Australasia Position Statement: Use of Human Genetic and Genomic Information in Healthcare Settings.**

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
0497 003 104

SWAN Australia
swanaus.org.au
0404 280 441

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

NSW

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

QLD

Support Groups Queensland
supportgroups.org.au
(07) 3344 6919

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

National Mental Health Support Services

Lifeline Australia
24/7 Crisis Support
13 11 14

Beyond Blue
beyondblue.org.au
1300 224 636

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