National Clinical Consent Package for Genetic and Genomic Testing Development Report

Revision of standardised consent materials for clinical genetic and genomic testing in Australia

Australian Genomics

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Working group

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Summary

Currently there is no genetic / genomic consent form that is used nationally, which leads to issues such as inconsistencies in patient experience, different terms and conditions for testing, and difficulties in sharing health data across jurisdictional borders.

A national expert working group facilitated by Australian Genomics has developed a revised clinical consent package, which meets current national standards and current clinical and laboratory practices. The consent package comprises a clinical consent form for genomic testing, a clinical consent form for genetic testing, a patient fact sheet and a health professional guide. The consent package looked to harmonise and update the National Model of Consent for Clinical Genomic Testing (dated 2021) and the consent form developed by Australian Genomics (dated 2020). This revised package underwent three consultations, including consumer review and Aboriginal and Torres Strait Islander expert review.

Key updates to the National Model of Consent for Clinical Genomic Testing include amendments to clauses for sharing of results for the healthcare of family members, future re-analysis of data and sharing of data for clinical purposes. Additional clauses for inclusion are withdrawal from testing, sharing results to My Heath Record, and data sharing for research purposes.

This report details the process and outcome of this project.

Project aims

1. **Review existing consent materials**
2. **Draft consent materials based upon current national standards and practices**
3. **Undertake a broad consultation on the consent package and re-draft materials based upon feedback**
4. **Seek approval/endorsement from jurisdictions across Australia**

The final consent package is provided alongside this report.

Project outcomes

Review of existing consent materials

In the last five years, two projects were undertaken to develop a national clinical consent form and supporting materials: an Australian Genomics project whose consent package was released in 2020 and an AHMAC-approved National Model of Consent for Clinical Genomic Testing, led by the NSW Ministry of Health and released in 2021.

Since both these forms have been released there have been amendments to national standards, specifically the National Pathology Accreditation Advisory Council (NPAAC) standards, and changes to current clinical and laboratory practices.

Amendments to NPAAC standards - *Requirements for medical testing or human genetic variation* (3rd Edition, dated February 2023) which have the greatest implication for the content of genomic and genetic consent forms is the following clause:

"For Level 3 tests, the laboratory must document and act in accordance with the patients' decisions regarding the items below:

a. *return of unsolicited findings, including unexpected familial relationships*
b. *data sharing of potentially re-identifiable data for clinical care*
c. *data sharing for ethically approved research*
d. *opt out if results are not to be included in the My Health Record"*

Clinical and laboratory practice advances include:

- Increasing opportunities for re-analysis of data
- Data sharing for clinical purposes becoming standard practice
- Changes to My Health Record processes and requirements

The project reviewed current consent materials and found that the changes above mean that available forms and supporting materials no longer meet the needs of clinical services, laboratories, and patients.

Although more consistent than previous years, there is still no consent package that is used nationally, leading to challenges such as inconsistencies in the patient experience, different terms and conditions for testing, and difficulties in sharing health data across jurisdictional borders.

Drafting the consent materials

Due to the aforementioned challenges, the expert working group drafted a consent package which comprises:

- clinical consent form for genomic testing
- clinical consent form for genetic testing
- patient fact sheet (PFS)
- health professional guide (HPG)

This consent package is largely based on the National Model of Consent for Clinical Genomic Testing and used the consent form as a template. Updates to clauses were made upon extensive discussion of current clinical and laboratory processes and to align with national standards.

These materials underwent iterative development and refinement over a 12-month period amongst working group members.

Consultation processes

During the development of the consent package three consultations were undertaken:

1. Consumer consultation
2. Aboriginal and Torres Strait expert consultation
3. Public consultation

Each consultation and the subsequent actions undertaken have been detailed below.

Consumer consultation

The consumer consultation was undertaken in July 2022, where six consumers were invited to review the drafted consent package. Broad consumer input was sought from representatives with rare diseases, cancers, and from Aboriginal and Torres Strait Islander communities.

Consumers were provided the consent package and asked to provide comment directly on the materials. Table 1 below outline key concerns and the measures taken to resolve them.

Table 1: Key concerns from consumer reviews

Consumer comments	Resulting amendment to document	Reason suggestion was not adopted
General feedback		
Concern about how the consent package will be delivered, e.g. what time in the patient journey will patient be given this information and will patients view the form independently.		This consent package will be viewed in discussion with a health professional. Consumer comments relating to this issue are not applicable.
Explanation of some terminology required, such as segregation testing, confirmation testing, blood relative, reanalysis, and family implications.	A glossary has been added to the Patient Fact Sheet (PFS) featuring these and others relevant terms.	
The clause table included in the Health Professional Guidance (HPG) should be included in the PFS.	Clause table added to PFS and the PFS re-structured to align better with the HPG.	
Some inconsistencies in terminology, e.g. genetic and blood relative used interchangeably.	All documents reviewed for consistency.	
Specific concerns raised by Aboriginal and Torres Strait Islander representative: • literacy and language barriers experienced by sections of the Aboriginal and Torres Strait Islander community • Aboriginal and Torres Strait Islander people may have different family structures. This in part is due to a history of trauma such as having a family member who is a part of the Stolen Generations or children who are in care and not connected with relatives or community. Discussions and information relating to family history and impact of testing on relatives must take this into account.	A specific Aboriginal and Torres Strait Islander expert review was undertaken.	

Consumer comments	Resulting amendment to document	Reason suggestion was not adopted
Consent form		
Inclusion of clearer information about legal obligations regarding sharing results with health professionals caring for family members.	This information was included in the PFS	
The term 'uncertain significance' is not clear.	This is a term commonly used in clinical genetics. It has been defined in the PFS and the health professional will guide patients through possible results that could be obtained through testing.	
Some concerns that the clause regarding health implications for relatives may "scare" patients, specifically parents worrying about passing conditions on to children.		Given the nature of genetic testing, heritability is a valid concern in many cases so the purpose of the statement is it inform patients of this issue. Health professionals will guide and support patients through these concerns.
Clarity around re-analysis processes.		Re-analysis processes are not standardised across Australia so the inclusion of further information on a national form is challenging. Health professionals will discuss opportunities for re-analysis with patients.
Concern that the phrase 'unexpected family relationships' might "scare" patients.		Further information about unexpected family relationships is provided in the PFS. Health professionals will guide and support patients through these concerns.
Request for further information to be provided about data storage.		As this is a national form, providing standardised statements about data storage (e.g. where data is stored and how long for) is difficult due to differences in laboratory policies. Information has been provided about pathways for samples to be destroyed.

Consumer comments	Resulting amendment to document	Reason suggestion was not adopted
Request for further information about sharing information with medical database.		As this is a national standardised statement about sharing with specific database, it is difficult to predict as different services have different policies.
Information about sharing with My Health Record should be included.	A tick box has been added to the consent form regarding results being shared to My Health Record.	
Health Professional Guide		
Information about the possibility of the test failing should be included on the HPG.	A statement indicating that a test may fail and a new sample may be needed has been included on the HPG.	
Clarification requested regarding sharing of information with insurance providers.	Statement indicating that results cannot be shared with insurance providers without patient permission was included.	
Further information about sharing results for the care of family members to be included.	Further information about what points need to be discussed with patients regarding sharing of results has been included on the HPS.	
Highlight that patients may need more time to consider whether testing is the right approach for them.	Additional statement included which suggests health professionals should allow patient to take forms and PFS home to have time to consider the information if needed.	
Include a separate section on people living with disability.	Further information on consent processes for people with disability has been included.	
Suggestion to include resources on avenues for professional support.	Professional support section with resources has been added.	
Suggestion to be more directive and request health professionals tell their patients that support groups are available.		It is acknowledged that support groups play a vital role in supporting people with genetic and undiagnosed conditions however support group information should be shared with patients at a time that best suits them. While health

Consumer comments	Resulting amendment to document	Reason suggestion was not adopted
		professionals are encouraged to share information about support groups, it is up to them to decide whether the timing is right for their patient.
Formatting suggestion: clearer table heading and numbering.	Changes made so table is easier to use.	
Patient Fact Sheet		
Addition of the following to the 'questions to ask' section: "How will the test be performed? Will it be invasive and carry risks? Will I be awake or asleep? How many days will I need off work?".		This information is covered in the "How is the test done?" section.
Suggestion to add *Choosing Wisely Australia* questions.	Questions were added to the PFS.	
Suggestion to simplify language.	Suggestions incorporated in materials.	
Not enough information provided on trio testing.	Further information on trio testing added to the clause table in both the HPS and PFS.	
Not enough information provided on withdrawal of testing.	Further information on withdrawal and change of mind included in both the HPS and PFS.	
Suggestions for more mental health supports such a Beyond Blue and state specific support services.	Suggested support services added.	
Suggestions for more detailed information about support services.	Further information on the role of support services added to the PFS.	
Formatting suggestions such as the addition of headings to aid clarity, shifting of information to improve flow.	Suggested changes made to the PFS.	

Aboriginal and Torres Strait Islander expert consultation

Due to feedback received from the Aboriginal and Torres Strait Islander representative in the consumer consultation, it was decided that a second, more comprehensive review by Indigenous researchers working in the genomics space was required. The aim of this consultation was to determine the suitability of this package for Aboriginal and Torres Strait Islander people engaging with genetic services. In August 2022, seven researchers were invited to comment on the consent package. Table 2 outlines their key concerns.

Table 2: Key concerns from Indigenous reviewers

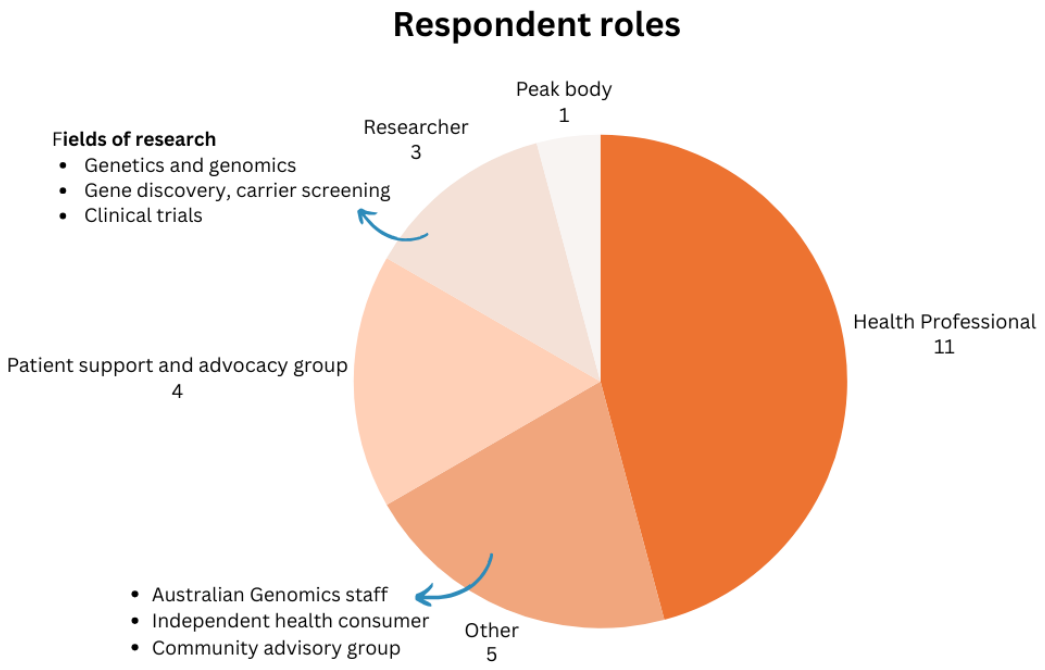
Concern	Details
Optional research clause	Concerns that the inclusion of the research clause blurs the line between medical diagnostic testing and research. Concerns that the samples are used in perpetuity for non-disclosed research. Unclear where samples are retained and what governance mechanisms will ensure patients are able to make informed decisions about how and for what research their samples are used. No mechanisms to support Aboriginal and Torres Strait Islander data sovereignty. Research consent information should be separated from the clinical testing consent to avoid people ticking boxes without really knowing what they are consenting to.
Lack of opt out pathways	Opt in clause does not clearly offer an opt out mechanism. No opt in mechanisms for other components of the consent e.g. storage of samples.
Literacy level of the documents	Consent processes are often tricky for participants with strong English literacy skills, more so for vulnerable individuals and families. These documents will be challenging for people to understand.
Unavailability of information in other formats	Multi-modal/multi-format consent protocols are essential for Indigenous peoples (e.g. visual cues, audio files in local dialects), as well as access to independent interpreters with experience in research and medical consent.
Lack of support for non- genetics health professionals	The guidance to health professionals is not enough to guidance for non-genetic health professionals to support ethical genetic consenting processes, especially to those patients from other cultural backgrounds.

Based on the feedback acquired, the working group engaged with experts from the National Centre of Indigenous Genomics to collaborate on the development of resources specifically designed to support Aboriginal and Torres Strait Islander people through the consent process. This project is currently ongoing, and is expected to be completed in early 2025. An Australian Genomics supported project being led by Indigenous genomics researchers is also working to build genomics knowledge in Aboriginal and Torres Strait Islander communities.

Public consultation

Between December 2022 and January 2023, the consent package was made available for public consultation. A consultation survey was developed to support the collection of standardised, comprehensive feedback. Participants had the option to complete a brief survey or a more detailed version. Invitations to take part in the consultation were sent to 85 individuals and organisations, representing a broad number of stakeholders including genetic health professionals, peak bodies, patient support and advocacy groups, and researchers. The consent package was also made available to the public on the Australian Genomics website.

Responses were submitted by 24 individuals and organisations, with health professionals providing the greatest number of responses. Eight participants carried out the brief survey and 16 carried out the detailed survey.



Over 80% of respondents found all clauses appropriate or very appropriate, except for the following clauses '*More tests or analysis may be needed to understand the results. This may include testing blood relatives'* and *'Results may show unexpected family relationships'*.

Table 3 summarises feedback provided for select clauses. Some feedback was deemed to be out of scope for this project. Specific examples of this are provided in the table.

Table 3: Example feedback from public consultation

Where noted, amendments suggested were in relation to a specific document. This is indicated by Patient Fact Sheet (PFS) or Health Professional Guide (HPG).

Clause	Suggested amendment	Action taken
Clauses where >80% of respondents rated very appropriate or appropriate*		
The test does not detect all genetic changes or predict all possible health conditions.	**PFS:** Contradiction of 'test only looks for changes related to your condition' and test may find incidental findings.	Clearer information about incidental findings added.
	PFS: Add section highlighting role of genetic counsellors and clinical geneticists.	Additional section on genetic counsellors and clinical geneticists and their roles has been included.
	HPG: Give reasons for no variant detected.	Reason for why no variant may be detected has been included on the HGP.
The test may find a genetic change not related to the reason for testing ('incidental finding').	There is no option on the consent form to express patient preference of receiving incidental findings.	There are currently no clinical, laboratory, or regulatory mechanisms in place to support this suggestion. It was therefore deemed out of scope.
	PFS: Include examples of what an incidental finding could be.	Example of incidental findings added.
	PFS: Suggestion to provide link for incidental finding support service.	There are no such services available.
The test may find a genetic change of uncertain significance.	**PFS:** Use HPG wording explaining VUS in PFS. Suggestions to state how often more testing occurs to clarify VUS.	The language used in the HPG is complex. A simplified version has been included in the PFS.
	PFS: Suggestions to state how often more testing occurs to clarify VUS.	There is too much variability to include this information.
	HPG: Mention the potential emotional impact of VUS.	Additional information on the emotional impact has been added to the HPG.

The sample or results may be re-examined in the future using new knowledge or testing methods.	**Consent forms:** Add clause "I agree/do not agree to reanalysis of the genomic data".	It is a risk to the patient if they select 'no' to re-analysis as this could potentially lead to a diagnosis in the future, should one not be obtained at the time of testing, therefore this statement was not included.
	PFS: Add information about how and when re-examination of samples may occur.	Currently there are no standardised processes for re-analysis. This process can be driven by the clinician, through re-referral or by the patient. The level of variability in processes makes it challenging to include this level of information on the PFS.
	Storage and sharing of samples should articulate the needs of CALD and Indigenous communities and their data sovereignty.	There are currently no standardise guidelines on data sovereignty. This makes it challenging to include this level of information on the PFS.
Results may have health implications for blood relatives	**PFS:** Copy what is in the HPG explanation to this section.	The language use in the HPG is complex. A simplified version has been included in the PFS.
Results may affect the ability to obtain some types of insurance.	**PFS:** Change "premium" to "how much you pay".	Language simplified in the insurance section.
The sample will be stored and may be shared with other laboratories to assist with genomic testing.	**PFS and HPG:** Both documents refer to 'legal' storage period, but these are accreditation/NPAAC guideline requirements, not legal requirements.	Information changed to state "as suggested by NPAAC standards".
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.	**PFS:** More relevant to say advance medical knowledge rather than scientific knowledge.	Suggested amendment included in the PFS.
	HPG: As examples use ClinVar as an international clinical database and Shariant as an Australian database.	Examples provided in the HPG.
I can choose not to be told the results if I change my mind, but the report will remain in medical records.	**Consent form:** Clause needs clarity – rewording suggested.	Clause reworded to make withdrawal and change of mind implications clearer.

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.	**PFS:** Explain the points of difference between this and the non-optional data sharing would help understanding.	Further information has been added to the clause table to make this clearer.
	This needs a supporting consent framework for ongoing (dynamic) consent.	This comment was deemed as out of scope as dynamic consent mechanisms are not available in the in the clinical setting nationally.
Clauses where 70-79% of respondents rated very appropriate or appropriate*		
More tests or analysis may be needed to understand the results. This may include testing blood relatives.	**PFS:** amend information to include that further testing may or may not identify a cause.	Amendment made to clause table.
	PFS and HPG: Explain in PFS and HPG why testing of blood relatives is included in this clause.	Further information added to the clause table.
Results may show unexpected family relationships.	**PFS:** Include a statement suggesting to speak to the health professional if there is a chance that testing may show unexpected relationships.	Statement included in the PFS.

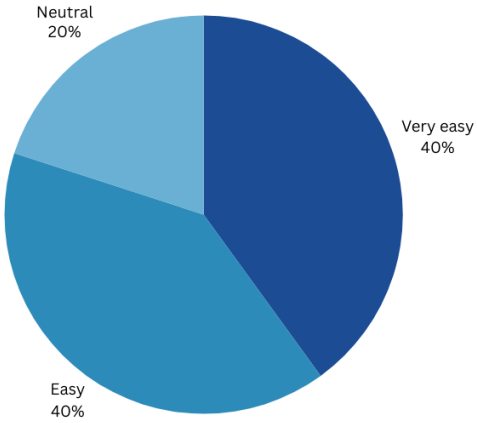
*combined brief and detailed survey ratings

A large majority (80%) of health professional surveyed believed that it would be very easy or easy to implement the consent package in the clinical setting.

Interestingly, there was no feedback in the public consultation which suggested that the research clause was concerning to respondents.

After aggregation and analysis of the consultation data, the working group convened a meeting to discuss and resolve discrepancies in responses and agree to content for amendment and inclusion. Table 3 indicates the actions taken to address the suggestions provided by respondents.

How easy would it be to implement this consent form at you clinical service?



Jurisdiction approval processes and outcome

Once feedback from the three consultations was incorporated, the consent package was sent to relevant representatives from the Australian Capital Territory, Northern Territory, Queensland, South Australia, Tasmania, Western Australia and Victoria for approval in July 2023. New South Wales was not approached for approval as it was indicated that the existing state-wide genomic consent form would continue to be used.

On advice from the working group and Government officials, the consent package was sent to either the directors of the clinical genetic service within the state or territory, or the State Department of Health. In Queensland and Victoria, State Departments of Health were approached, whereas clinical genetics services were approached in all other states and territories. All jurisdictions were given the opportunity to comment on the consent package.

Amendments made to the consent package based on feedback from the states and territories is summarised below:

Consent forms
• ‘Misattributed parentage’ was removed from Clause 7 (i.e., about the possibility finding of unexpected family relationships). To avoid confusing patients, all examples were removed as clinicians can provide verbal examples to patients. This is indicated in the HPG. • Removed the option to specify relatives and leave as a ‘yes or no’ answer.
Patient Fact Sheet
• Additional questions added about results delivery and changing consent to ‘questions to ask your health professional’. • Minor formatting changes. • Edited wording to emphasise the options of withdrawing consent. • Additional text to explain the role of health professionals. • Additional definitions in glossary.
Health Professional Guide
• Additional statement about test requestors communicating with laboratories about test types and associated possible results to keep their patients informed. • Additional statements suggesting health professionals discuss the possibility of further testing and sample destruction with patients.

- Additional statement indicating health professionals should provide patients with examples of unexpected family relationships, if appropriate.

The following states have endorsed the consent package: Australian Capital Territory, Northern Territory, Queensland, South Australia, Tasmania, Western Australia. The consent package has not been approved in Victoria, however on the suggestion of the Victorian Department of Health, the consent package will be submitted to the Health Technology Genomics Collaboration for consideration and to seek endorsement.

Conclusion

The development of a National Clinical Consent Package continues to be a challenging endeavour. There is jurisdictional variation in policies and procedures in clinical care, limitations in the resourcing, and there is inconsistency in use of terminology across Australian genomic practice which makes national consensus difficult. However, a unified approach will reduce issues currently seen, such as inconsistencies in the patient experience and difficulties in sharing health data across jurisdictional borders.

Developing a single form that meets the needs of Australia's diverse communities has also proved to be a challenge. Australian Genomics will look to experts to aid in the development of target resources to better support underrepresented groups such as Aboriginal and Torres Strait Islander people, people with disability and migrant and refugee communities.

With endorsement from a national perspective, there is likely to be a greater uptake in the use of the National Clinical Consent Package which will benefit all Australians undergoing genomic testing in the future.