# National Model of Consent for Clinical Genomic Testing

Final report

January 2021

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The National Model of Consent for Clinical Genomic Testing was prepared under the auspices of the Australian Health Ministers' Advisory Council.

The report and accompanying resources can be obtained from the NSW Health website at www.health.nsw.gov.au

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## Acknowledgements

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# Introduction

The New South Wales Ministry of Health (NSW Health) on behalf of the Australian Health Minister's Advisory Council (AHMAC) Project Reference Group on Health Genomics, developed a national model for clinical consent to genomic testing.

Genomic technologies are rapidly advancing, so too are their application in clinical practice. The national model aims to facilitate a consistent and standardised approach to optimise the consent process for genomic testing.

This program of work was delivered in accordance to priority 4A [*to build on existing work to develop and promote nationally consistent templates and guidance for consent*] of the National Implementation Plan for the National Health Genomics Policy Framework.

This project included extensive stakeholder consultation with Federal, State, and Territory health agencies, clinicians, researchers and consumers to ensure that national views are incorporated in the model.

## Note on terminology

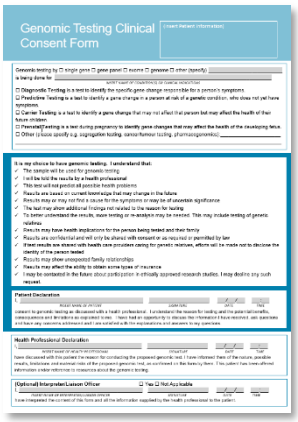
*Genomics*: Consistent with the National Health Genomics Policy Framework, the term 'genomics' is used in these materials to refer to both the study of single genes (genetics) and the study of an individual's entire genetic makeup (genome) and how it interacts with environmental or non-genetic factors. While genetic testing for clinical purposes is already embedded in the health system, the term genomics is used for brevity and to acknowledge the cross-over of issues between genetics and genomics, other than where it is necessary to differentiate between genetics and genomics.

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

# 1. Deliverables

This section includes the materials developed to support the national model.

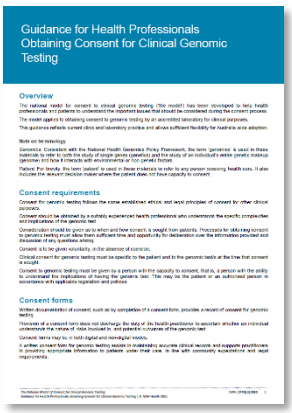
## 1.1 Template consent for clinical genomic testing


Genomic Testing Clinical Consent Form

The template consent for clinical genomic testing has been developed as a flexible tool for adoption and adaptation across all jurisdictions. While it does not make specific provision for third party consent due to variation in jurisdictional regulations, it is intended to be readily adaptable for this purpose.

To access the consent form template, please visit the NSW Health website at www.health.nsw.gov.au and use the search term "Template for Clinical Genomic Testing".

## 1.2 Guidance for health professionals obtaining consent for clinical genomic testing


Guidance for Health Professionals Obtaining Consent for Clinical Genomic Testing

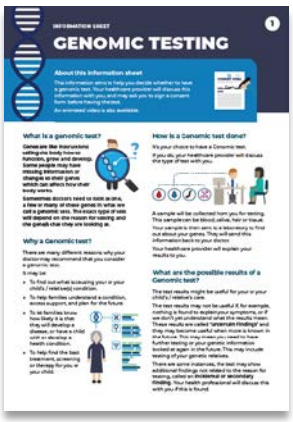
The guidance for health professionals is aligned to the consent form template. It highlights the important issues that should be considered during the consent process and reflects current clinical and laboratory practice in Australia.

To access the guidance, please visit the NSW Health website at www.health.nsw.gov.au and use the search term "Guidance for Clinical Genomic Testing".

## 1.3 Patient information materials

The supporting materials for patients are multimodal and pitched at a level to support broad understanding for a range of clinical applications of genomic testing.

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".



# 2. Methodology

An expert advisory group was established to oversee project milestones, consultation and communications with key stakeholders and to provide expert guidance on the development of a consent form template and other deliverables.

## 2.1 Phase one

The initial consultation phase used a range of distinct methods to reach a variety of stakeholders. This report presents the findings from a broad sector survey, targeted interviews with stakeholder organisations, and a focus group of consumer representatives with supplementary submissions, conducted as part of this project.

NSW Health acknowledges the Australian Genomics Health Alliance and the NSW Agency for Clinical Innovation, Clinical Genetics Executive Committee for their collective contributions to the consent form template utilised for the consultation.

NSW Health acknowledges Nous Group (Nous) who were engaged to provide support to the initial phase of consultation.

### Broad-based survey of genomics sector stakeholders (130 responses)

The survey sought to understand stakeholders' views on whether they agreed or disagreed with the inclusion of specific fields and information in the consent form and the guidance material. NSW Health led the development and administration of the survey. Under guidance from NSW Health, Nous analysed specific elements of the survey responses to produce the detail in this report.

There were 130 survey responses collected. 48 per cent (62) of these responses were from genetic health practitioners [genetic counsellors (31), clinical geneticists (26) or genetic pathologists (5)]. Non-genetic health practitioners accounted for nine responses, whilst non-health practitioners provided six responses. The remaining 41 per cent of responses (53) were completed by 'other' participants which included researchers, scientists and laboratory staff.

### Focus group and submissions representing nine consumer groups

A focus group was facilitated by Nous to determine the views of consumers, specifically to understand participant views on what factors are important and relevant for inclusion in a nationally consistent consent model for genomic testing and how effective consent for genomic testing can be obtained.

Nine participants attended, or made a submission following the focus group. The attendees represented a mix of organisations by size and condition specificity and included individual consumers.

### Stakeholder interviews and submissions representing 15 organisations

NSW Health conducted several interviews with stakeholder organisations. The interviews sought to understand stakeholders' views on issues associated with consent to genomic testing and whether they agreed or disagreed with the inclusion of specific fields and information in the consent form. NSW Health led the interviews and captured transcripts and key discussion points.

In addition to the stakeholder interviews, submissions in writing were received by NSW Health.

Submissions and interview transcripts were produced, with stakeholders representing medical colleges (2), research institutions (4), private pathology companies (1), insurance companies (1) and States and Territories (7). Nous analysed this information to produce the detail in this report.

## 2.2 Phase Two

### Broad-based consultation on draft final report

Consultation was sought on the draft final report, inclusive of the second phase consent form template, guidance for health professionals and relevant inclusions for patient materials.

19 submissions were received from stakeholders, representing individuals, organisations, or the collated responses of stakeholder networks within an organisation. Submissions including detailed feedback were received from medical colleges (2), genetic counsellors (2), genetic pathologist (1), education centres (1), Aboriginal and Torres Strait Islander Health representative (1), support group bodies (4), consumers (2), industry (1), contract research organisations (1), research institutes (2), Commonwealth Department of Health (1) (including National Health and Medical Research Council) and the Human Genetics Society of Australasia (1) inclusive of submissions collated from the Education, Ethics and Social Issues Committee and Clinical Geneticist Special Interest Group).

**Patient information working group**

A working group was established to further guide development of the patient information. Members included clinical, pathology, education, research and consumer representatives.

An initial workshop was held in March 2020 to determine the relevant inclusions in a 3 minute animation for patients consenting to a genomic test. 10 participants attended or made a submission following the focus group.

The working group members reviewed the draft script, story board and final animated video resource, and the draft and final factsheet.

NSW Health acknowledges Limelight Media Creative who were commissioned to develop the patient information materials.

# 3. Summary of consultation findings

Consultations highlighted a range of perspectives on the approach to consent for genomic testing, however there were common themes amongst this feedback. This section outlines the key themes related to the supplied consent form template and guidance for health professionals, with a summary of the suggested changes to these documents. This section also includes suggestions for supporting information for patients to assist them in understanding the consent process and the implications of testing. Detailed consultation findings are provided in Appendix A.

## 3.1 The consent form template

Two broad themes were identified in stakeholder comments on the consent form template. The first is a need to review the structure and content of the template and the second is to clarify the process identified within or accompanying the consent form.

### Structure and content of the consent form template

Consent for research: There were mixed comments from participants on the inclusion of consent for research in the template. Some suggested that this should be removed as obtaining consent for research should not be part of a clinical consent form. One participant commented that 'Research using patient samples requires specific consent from the research group and is not necessary to discuss on a genetic testing consent form'.

Participants suggested that in some instances it may be unethical to obtain consent for research in a clinical scenario, for example, when patients are under immense stress and concerned for their health. One commented that 'It's traumatic enough to be going through a process that has so many unknowns. Let's not make the process more traumatic by adding research consent. Let the patient have the test and get the results first before requesting consent for research'.

Some stakeholders commented that this field should be mandatory rather than optional as it is important to familiarise patients with genomics data sharing and research. One commented that 'Clinical practice and research are so intertwined within the genomics space at the moment. It is detrimental to the family to have to go back and re-consent for every research opportunity that may arise. To further advance our knowledge in the field of genetics and genomics we have to be able to share data with other researchers'.

Others indicated that the provisions of the National Statement on Ethical Conduct in Human Research (2007, updated 2018) make it unlikely that Human Research Ethics Committees (HREC) would accept unspecified or extended consent for research using identifiable genomic data, although in some circumstances will approve research using deidentified data without requiring patient consent. Stakeholders also noted that generally no mechanism exists to enable researchers to identify patients who have consented to participate in research as part of the clinical consent process.

Highlight the importance of the health professional's role: Participants suggested that the patient's consent should state that they choose to have genomic testing 'as discussed with my health care provider', to reference the role of the health professional in ensuring that this is an informed decision. Stakeholders supported inclusion of the health care provider's declaration at the end of the form, to acknowledge that they have gained consent from the individual in proceeding with the completion of the declaration.

Describing types of genomic testing: Consultation participants commented that the test examples provided should be clearly explained and ordered to reflect how commonly they are conducted. Following considerable feedback and discussion with stakeholders it was agreed that important but less common tests, such as segregation testing, cancer/tumour testing and pharmacogenomic tests, should be included as examples for the 'other' category.

### Clarify the processes outlined in the consent form template

Unsolicited/ unexpected/ incidental and unknown/ uncertain findings: Feedback suggested that the term 'incidental' is outdated and should be replaced with 'unsolicited' or 'unexpected' findings. Participants suggested defining and including examples of incidental findings and to consider giving patients the option to learn about any incidental findings that may arise through their genomic testing. One participant stated that 'The definition of incidental findings is a bit ambiguous'.

Participants discussed the variation in processes for reporting and informing patients of incidental findings between the States and Territories and suggested adding 'laboratories may differ in their policies on variant reporting' as a potential outcome of testing. One participant said, 'the point on incidental findings would require amendment for use in WA, as we would not seek nor report incidental findings'.

Participants discussed amending the use of 'unknown' to 'uncertain' in the template and suggested adding the possibility that results from the test may not reveal a diagnosis. One participant stated that 'Uncertain clinical significance is better than unknown, which implies that we have no knowledge at all, which is usually not the case'.

To support an accessible reading level, it was agreed that the template would include general statements about these concepts in plain English, to prompt further explanation and discussion with the health care provider.

Release of results to family members: Participants suggested amending the template to inform the patient that their results may be released to health care providers treating their genetic relatives as legally permitted or required (e.g. to prevent a serious threat to another's health) but that this would be done without revealing their identity if possible.

Withdrawing consent: Participants suggested further clarifying the process to withdraw consent, specifically what can practically be done if a patient changes their mind about having the test. Other stakeholders suggested removing this statement as it may not be feasible to action it once the testing process has been initiated.

## 3.2 Guidance for health professionals

The consultation on the guidance material for health professionals highlighted key aspects to consider when obtaining consent, including the sharing of genomic data.

Informed consent: Consent should be obtained by a health professional who has a good understanding or knowledge of genomics. The implications and interpretation of genomic test results have a layer of complexity that doesn't exist in broader clinical circumstances. Practitioners should ensure that an appropriate amount of time is set aside to obtain informed consent and use plain language. Information supporting the consent form may be useful if given before the appointment and made available following the appointment, and health professionals should document the supporting information that they share with patients. Guidance is required on when a written consent form is to be used.

Cultural and linguistic diversity: Health professionals should be culturally and linguistically sensitive when obtaining consent from people with diverse backgrounds. This includes noting that many cultures may not have a specific phrase or term for genomics, and that some cultures may have specific practices for the treatment of tissue. Therefore, participants also suggested amending this section to include the use of telephone interpreters, non-professional interpreters and liaison officers' roles (in addition to professional interpreters) to extend the range of individuals available to ensure that patients understand the proposed test.

Aboriginal and Torres Strait Islander peoples: There are specific considerations relevant to first nations Australians in consenting to genomic testing. Clinical testing, collecting, storing, disseminating and using genomic clinical data and information will uniquely impact on first nations people, because of their cultural practice, belief and support systems; and the possible effects of the genomics information of an individual could have on the broader Aboriginal and Torres Strait Islander communities.

Communication difficulties: Distinct guidance is required for people who have hearing and/or visual impairment among other special communication needs.

Modes of obtaining consent: Consent can be obtained using many modes, including digital tools. Simple informative videos can also be provided to patients in advance of pre-test consultations to help them understand the process and prepare for the discussion.

Clarification of data management: Participants commented that data sharing and ownership must be transparent to the patient, and they should be comfortable with how their sample and data are being treated and accessed. Participants commented on the need to understand the authority and scenarios in which their test results can be shared with and without their consent for insurance, research and diagnostic purposes. Participants also

discussed a need for a national policy on data sharing. It is considered that information and data sharing without individual patient benefit are beyond the scope of this project and are being addressed through other national initiatives.

Consultations highlighted that further information should be included to reassure patients that appropriate measures will be in place to respect their decision if they choose not to be informed of any incidental/ unsolicited/ unexpected findings.

Obtaining consent for re-testing and re-analysis: Participants commented that they did not see it necessary for formal (i.e. written) consent to be reobtained for re-testing and re-analysis, particularly if the purpose of the test/clinical indication is the same. It was also noted that any considerations for further testing and analysis should be incorporated into ongoing and supported conversation between the patient and their health professional.

Research: Participants overwhelmingly recognised the value of research in genomics. However, as with the consent form template, there were mixed comments from participants on the degree to which research should be addressed in the guidance. Some participants suggested research consent as 'a completely separate issue' to clinical consent, highlighting ethical considerations and practical limitations for including research in a clinical consent model. Other participants proposed research consent as a necessity and prescribed detailed information including definitions applicable for data sharing for research purposes. It was noted the guidance must align to the consent form template and include any information about research of relevance in a clinical consent process. Suggestions considered future participation, ethical conduct in human research and the role of HRECs.

## 3.3 Patient information

The types of information that could be provided to patients to help them to understand the consent process and the implications of testing were discussed during the participant interviews, focus groups and workshops, and refined through stakeholder feedback.

Several clear themes arose from this feedback on what information can best support patients. These themes are consistent with those discussed in the patient guidance material and the consent form template feedback, and include:

Genomic testing implications for genetic relatives: Consultations highlighted that information should be provided to patients to help them understand why their test results could have implications for their genetic relatives and what these implications could be.

Participants discussed that further information should be provided to help patients understand the process that health providers and organisations use to notify genetic relatives of genomic test results.

Genomic testing process: Consultations highlighted the need to help patients distinguish between the different types of genomic tests. Participants discussed providing further information to help patients understand their test results, including that the test may return incidental findings. Participants also sought advice on how to navigate the decision to receive these results, or not.

Consultations also highlighted that information should be provided to ensure patients understand the test may return uncertain findings and there may be a need for further re-analysis and re-testing. Participants also suggested providing further guidance on engaging with support groups and patient reference groups when referred for testing and after receiving test results.

- Sharing genomic data: Participants discussed the need to provide further information to help patients understand other issues related to testing. These include scenarios in which their information can be shared, both with and without their consent, and the legislative basis for this. Participants commented that further information is needed regarding the application of data, including if it will be used ethically, whether their data will be shared for diagnostic purposes and if used for research (locally and internationally). Participants also suggested additional information is needed to define 'advance scientific knowledge' and 'other scientific purposes' if used within the consent form template. Participants noted variation in data management by jurisdiction and organisation, and such information should be made available by individual organisations to both health practitioner and the patient.

For insurance purposes: Participants discussed the need to provide further clarity to patients on how their genomic testing results will be used for insurance purposes. Participants recognised published advice currently available regarding the Moratorium on Genetic Tests in Life insurance that patients could be linked to. There was mixed discussion on the inclusion of detailed information on insurance implications, as these would be best advised by a financial consultant.

Principles: Resources to be developed should apply to any genomic test and include information relevant to patients consenting to a test in all Australian jurisdictions. To accommodate broad utility and avoid duplication of already existent materials, the resources will support patients consenting to a genomic test through multimodal access to foundational information.

The patient information working group considered the patient information resources should be an easy first port of entry, and utilise user friendly language, avoiding repetition and complex terminology. The resources should provide balanced information to support informed consent with alignment to the consent form inclusions. They must include outcomes of testing and next steps, addressing support groups. The resources should also address incidental/ unsolicited/ unexpected findings, and implications for genetic relatives, and direct to further information and discussion regarding complex considerations (e.g. insurance).

# 4. Future Considerations

The consultation highlighted potential areas for further consideration that were noted as beyond the scope of the existing project. Areas included:

- insurance consequences – the implications of the use of genetic tests for insurance purposes are unknown post moratorium.
- sharing of deidentified genomic information – data sharing without individual patient benefit should be addressed through other national initiatives.
- approach to incidental/ unsolicited/ unexpected findings – principles for managing the return of incidental findings is an identified area for future national consideration.

Additionally, considerations for implementation were raised during consultation, and included:

- Aboriginal and Torres Strait Islander consumers to be included during local implementation of the model. This may include development of additional resources in partnership with consumers to appropriately convey the diversity of Aboriginal and Torres Strait Islander communities.
- Provision in accessible formats such as through an Easy Read/Easy English documents, document readers or form readers.

# Appendix A – Detailed consultation findings

**Table 1 | Summary of changes suggested by stakeholders to consent form templates**

| Template Field | Phase 1 consultation (Version 1 template) | Phase 2 consultation (Version 2 template) | Comments/Other |
|---|---|---|---|
| **1 Clinical indication for and type of test**<br><br>**(Field 2C**<br>**Field 2D**<br>**in Version 1)** | • Consider amending language to 'Genetic/genomic testing by [ ] single gene [ ] gene panel [ ] exome [ ] genome [ ] other – specify _____is being conducted for _____(insert name of condition(s) or clinical indication(s)'<br>• Provide further information to distinguish between the different types of genomic technologies that the test may use<br>• Suggest editing language used in test definitions:<br>• Predictive and pre-symptomatic testing is a genetic test to identify a gene change in a person at risk of a genetic condition, who does not usually have symptoms. (The current definition does not apply to patients without a family history)<br>• Carrier testing is a genetic test to identify a gene change that is unlikely to be of relevance for that person but may be of relevance for their child. (The current definition applies to every form of testing)<br>• Diagnostic testing is a genetic test to identify the specific gene change responsible for a person's diagnosis. (The current definition presumes that the patient has already had a specific genetic condition diagnosed on clinical grounds)<br>• Pre-natal: a genetic test of a mother or the developing baby to identify gene changes that would affect the baby<br>• Consider using pre-symptomatic or predictive testing<br>• Reorder testing to reflect commonly ordered tests: diagnostic, predictive then carrier and then remaining options<br>• Consider inclusion of 'segregation' testing<br>• Suggest editing language used in test definitions:<br>• Predictive testing is a genetic test to identify a gene change in a person at risk of a genetic condition, who does not usually have | • Consider reviewing this field in future when clinical genomics is more widespread<br>• It can be problematic to specify the type of test as plans may change or testing reflexed to a larger test if the initial result was negative, requiring a new consent although the purpose and implications remain the same<br>• Specification of the type of test helps laboratories to distinguish Level 1 and 2 tests<br>• The health practitioner's statement is useful to laboratories as it confirms that information on the testing consequences and further resources have been discussed<br>• Consider deleting family studies as it is confusing<br>• Consider including a tick box for diagnostic testing of cancer tissue to inform treatment regimes<br>• Clarify where pharmacogenomics fits and explain in the supporting information<br>• Consider suggestions to simplify language and clarify terminology<br>• Consider specifying who should complete which section of the form | • Delete family studies from list<br>• Cancer/tumour testing to go under Other<br>• Pharmacogenomic testing to go under Other<br>• Explain cancer/tumour and pharmacogenomic testing in supporting information<br>• Explain that interpretation of carrier testing is done with the results of the reproductive partner in supporting information |

| Template Field | Phase 1 consultation (Version 1 template) | Phase 2 consultation (Version 2 template) | Comments/Other |
|---|---|---|---|
| | symptoms. (The current definition does not apply to patients without a family history) | | |
| **2 Patient consent**<br><br>(Field 3A<br>Field 3B<br>Field 3C<br>Field 3D<br>in Version 1) | • Suggest amending to 'After discussion with my doctor/health professional, it is my choice to have genetic testing. I understand and acknowledge that: _____'<br>• Consider removing 'acknowledge'<br>• Consider amending to genetic/genomic for consistency within the form<br>• Consider removing 'having the test performed'<br>• Consider amending the first point to 'My biological sample will be used for genetic and genomic testing'<br>• Consider amending the second point to state 'I can change my mind and ask for the testing process to be stopped and I can choose not to be told the result'<br>• Consider providing further guidance on the process to withdraw consent, in the scenario where the health practitioner that obtained the consent is no longer contactable<br>• Consider adding that 'Laboratories may differ in their policies on variant reporting'<br>• Consider amending point four to 'Further testing and information sharing may be needed now and into the future to finalise my result'<br>• Consider changing first point to 'results are based on current knowledge and technology' and consider leaving out 'that may change in the future'<br>• In the second point, consider changing 'unknown' to 'uncertain'<br>• Add a new point that communicates that results may not reveal a medical diagnosis<br>• In the last point, suggest changing the wording to '…could find information about other medical conditions (incidental findings)'<br>• In the fourth point consider changing 'finalise' to 'understand'<br>• Consider using word 'unrelated' in definition of incidental findings<br>• Suggest adding to this field that the results of the test could reveal a serious condition | • Consider changing initial statement to: "It is my choice **whether or not** to have genomic testing" to avoid any assumption that consent will be given<br>• Information that the test will not predict all future health problems is important, particularly for exomes where clinicians often explain it as reading 'all' genes<br>• Consider advising that test results will be available to health professionals involved in the patient's care<br>• Consider further clarification of the insurance question and 5 year moratorium on risk profiling<br>• Consider a section to allow the genomic data and related information to be shared and stored in an untraceable form to help advance scientific knowledge<br>• Consider including consent for release of results to a designated person<br>• Consider adding more detail about the type of sample used - saliva, tissue, biopsy; what happens to the sample, how long is it retained, and by whom<br>• There are mixed reactions to the use of the term 'told' vs 'explained' - 'explained' is more conversational but 'told' will lower the reading age<br>• Consider removing reference to 'my' sample / results to permit adaptation of the form for use by guardians for testing on minors<br>• Consider changing the term 'incidental findings' to 'unsolicited findings' or 'unexpected findings'<br>• Consider adding to the consent form this point that "Re-testing and/or re-analysis may be undertaken in future".<br>• Consider adding advice about the ability to withdraw consent | • Information on insurance, such as types of insurance affected and the moratorium, to be included in supporting information<br>• Information on data storage and sharing to be included in supporting information<br>• Information about research use of data to be included in supporting information |

| Template Field | Phase 1 consultation (Version 1 template) | Phase 2 consultation (Version 2 template) | Comments/Other |
|---|---|---|---|
| | • Consider amending the first point to indicate that the test may not return any immediate results but may in the future through re-analysis of genetic/genomic data<br>• Provide further information and examples to define incidental findings and consider including the option to learn about any incidental findings<br>• Simplify point four to state 'My results are confidential and will only be released with my consent or as required by law'<br>• Consider changing the use of the word 'family' to 'genetic relatives'<br>• Consider amending the first point to add that the practitioner delivering the results may be different to the practitioner obtaining consent<br>• Suggest providing further information about the availability of referrals to other services and health professionals | | |
| **3 Optional consent**<br><br>**(Field 3F**<br>**Field 3G**<br>**in Version 1)** | • Consider revising first point to state: 'My test results can be shared with relevant health practitioners involved in my care, as well as those involved in the care of my family members (if clinically relevant)<br>• Add further information to describe efforts that health practitioners will go through to contact individual<br>• Consider amending point two to include a yes/no box so patients have an option to not nominate an individual<br>• Consider adding to the end of the first point '…though my identity will not be revealed to any other family members'<br>• Provide further information to allow individuals to express their preferences on how they receive their results<br>• Consider amending the second point to include that a notification to collect test results will be sent to the individual if the first contact attempt is unsuccessful, prior to any results being released to the appropriate person that is requested in this point<br>• Consider including that this field applies to the release of this information beyond the laboratory for purposes that are not related to the purpose for which the sample and information were collected | • Consider using yes/no boxes instead of tick boxes to avoid ambiguity if the box is not ticked<br>• Consider removing 'if clinically relevant' as too open to interpretation<br>• Consider removing the word 'optional' as discussion of these options should not be optional<br>• Consider adding 'subject to any legal requirements' for sharing information with relatives or their treating health practitioners; and this may be done without disclosing the patient's identity<br>• Consider provision for disclosure to a nominated relative<br>• Consider an additional option of sharing traceable genomic data for future ethically approved research, so the patient can benefit from any clinically relevant findings<br>• Note that Human Research Ethics Committee (HREC) approval is highly likely to require specific patient consent for research that uses identifiable patient information | • Information about research consent to be included in supporting information, including the role of HRECs and the National Statement on Ethical Conduct in Human Research (2007) updated 2018<br>• Suggest supporting information consider disclosure of results not requiring consent (e.g. to prevent a serious threat to another's health) but that disclosure may occur without revealing their identity |

| Template Field | Phase 1 consultation (Version 1 template) | Phase 2 consultation (Version 2 template) | Comments/Other |
|---|---|---|---|
| | • Consider including that the scientific research is performed in Australia and with oversight by a suitably accredited research ethics committee<br>• Consider amending the section to include a dot point on who to contact if an individual chooses to withdraw their consent to their data being used in research<br>• Consider separating consent for research<br>• Amend each point to include a yes/no box<br>• Consider separating consent for research from clinical form<br>• Reflect on making this section mandatory to complete<br>• It is currently unclear which points the yes/no box applies to – this section may need reformatting to indicate which points in the field the yes/no box applies to<br>• Consider increasing the font size as it appears smaller than the rest of the form | • Consider an additional option of sharing untraceable genomic data for future ethically approved research<br>• Consider an additional option for storing a sample in a biobank for future ethically approved research<br>• Note that the requirements of the National Statement on Ethical Conduct in Human Research (2007) updated 2018, include extended or unspecified consent and permission to enter the original data or tissue into a databank or biobank/tissue bank<br>• Clinical geneticists request leaving out the research option as they never recruit patients on this basis; the form is for clinical use only<br>• Consider an additional option for inclusion of results in MyHealth Record<br>• Consider an additional tick box regarding whether the patient wishes incidental/secondary findings to be reported<br>• Consider just one option, 'Sharing my test results with health practitioners involved in the care of my genetic relatives (your identity will not be revealed)' | |
| **4 Patient declaration**<br>(Field 2A in Version 1) | • Distinguish between genetic and genomic testing<br>• Consider moving this to the end of the form before the patient's consent is obtained and consider simplifying to 'I agree that the issues listed in this form have been discussed and that the patient agrees to have the test under these conditions'<br>• Survey participants suggested change the date to include forward slashes from ______ to __/__/__<br>• Consider moving this to the end of the form<br>• Consider simplifying<br>• Consider removing 'written information' as it may not always be required, noting clinicians and patients should work together to select appropriate information based on the clinical evidence and the patient's informed preferences<br>• Consider presenting each component in this field separately to allow each aspect to be individually reflected upon | • Consider including that the form is for adult patients with capacity to give consent<br>• Consider including nomination of a person to receive results if the patient cannot be contacted or is deceased<br>• The term 'consequences' may need explanation, including possible outcomes of the test<br>• Consider including patient acknowledgement that they were given information (in addition to explanation)<br>• Consider replacing 'Patient Declaration' with 'Consent'<br>• Consider removing the time of consent<br>• Consider including reference to patient understanding the discussion and information provided | |

| Template Field | Phase 1 consultation (Version 1 template) | Phase 2 consultation (Version 2 template) | Comments/Other |
|---|---|---|---|
| **5 Health practitioner declaration**<br><br>(Field 3H<br>Field 3E<br>in Version 1) | • Consider removing health practitioners name from this section<br>• Survey participants suggested change the date to include forward slashes from ______ to __/__/__<br>• Consider removing 'in a way I understand'<br>• Consider further simplification<br>• Consider repositioning this field towards the end of the form to enable all fields to be considered prior | • The placement of this declaration after the patient declaration was well received<br>• Consider omitting 'additional' information being offered as this is not done routinely at this time | |
| **6 Interpreter / liaison officer**<br><br>(Field 2B<br>in Version 1) | • Consider amending to seek further information about the interpreter, for example, if they are professionally qualified or not<br>• Consider amending to accommodate for telephone interpreters, non-professional interpreters and liaison officers' roles in supporting consent for Culturally and Linguistically Diverse populations<br>• Survey participants suggested change the date to include forward slashes from ______ to __/__/__ | • Consider using consistent plain English language throughout the consent form and patient information<br>• Consider providing materials in an accessible format for people with disabilities rather than assuming language (e.g. Easy Read/Easy English for people with intellectual disability); to be trialled and tested before being adopted<br>• Consider emphasising the importance of proactive engagement of an interpreter with time during consultation to obtain informed consent. If an interpreter is not available, this should be documented<br>• Consider including which language was interpreted as important from a risk management perspective | • While it is beyond the scope of this project to develop materials for people with disabilities it should be considered for future development<br>• Suggest emphasising the importance of allowing enough time during consultation to obtain informed consent in supporting materials |
| **Additional comment** | | • Consider including consent to sharing deidentified genomic data and information with international databases such as ClinVar to help advance scientific knowledge, even though there may be no direct benefit to the patient. This fits with international (GA4GH) and NPAAC recommendations on data sharing for clinical benefit<br>• Consider restoring research statements from version 1 of the form, as NPAAC requirements that consent for research be "distinct" from the consent process for clinical testing need not mean a separate appointment or separate materials<br>• Consider that for an existing research project, research consent could be obtained at the time of consent for diagnostic testing, saving clinical time and resources to seek separate consent if a potential research finding is there, or be forced to ignore alleles that with some | • Suggest that information and data sharing without individual patient benefit are beyond the scope of this project and are being addressed through other national initiatives<br>• Note that while research consent may be appropriate and efficient for a clinician researcher who has an established relationship with their patient, this is not the case for most clinicians or patients. Feedback on version 1 suggested that seeking research consent may pose a barrier to obtaining clinical consent for some cultures<br>• Note that the submitted extract from NPAAC requirements is selective |

| Template Field | Phase 1 consultation (Version 1 template) | Phase 2 consultation (Version 2 template) | Comments/Other |
|---|---|---|---|
| | | translational work will rapidly become diagnostic findings | and ignores provisions that the informed consent process should clearly state the protocol for re-contacting the patient about findings, including incidental findings, identified during subsequent research projects.<br>• Note also the NPAAC requirement that all requests from researchers for the release of de-identified data from samples submitted for diagnostic testing and of associated laboratory data to biobanks or databases will require approval by the Human Research Ethics Committee with responsibility for oversight of activities of the pathology service |

**Table 2 | Summary of changes suggested by stakeholders to guidance for health professionals**

| Themes | Phase 1 consultation | Phase 2 consultation | Comments/Other |
|---|---|---|---|
| **Informed consent** | • Add that consent must be obtained by a health professional with genetics specific experience<br>• Consider added that ongoing training for health professionals around obtaining genetic consent<br>• Consider adding that health professionals ensure there is adequate time during consultations to obtain informed consent for a genetic/genomic test<br>• Consider adding that health professionals use plain language to obtain consent for a test and that they distinguish between consent for a test and consent for the use of results | • Consider clearer advice on when a consent form should be completed. For instance, consider adding that if testing of more than one or two genes i.e. panel, exome or genome sequencing a consent form should always be completed. There is the potential for confusion without clear definition of complexity.<br>• Consider relevant consent laws, age of consent for example.<br>• Consider application for a person without capacity.<br>• Feedback highlighted differing views on the requirement for written consent.<br>• Consider highlighting the implications and interpretation of genetic / genomic test results has a layer of complexity that doesn't exist in broader clinical circumstances. | • Provide further information within 'written consent form' section, including link to the NPAAC requirements that offer further explanation of complexity.<br>• Amend first statement to highlight the importance of consent in the context of genomics.<br>• Note, there is no set age at which a child or young person can give consent. Provide guidance in 'written consent form' section that different forms may be required for minors and substitute consent. |
| **Written consent forms** | • Amend last point to state that issues in the consent form should serve as a prompt for health professionals to discuss them at a more in-depth level with patients<br>• Consider amending to include digital and non-digital modes of 'written' consent | • Suggest including requirements when laboratories performing testing need to confirm evidence of consent; including examples of complex issues resulting from genetic testing could be provided, or direction to places where different levels of genetic testing are explained.<br>• Consider that different forms may be required for minors and substitute consent.<br>• Clarify that a consent form is a tool that can be used to help guide the discussion with the patient during the consent process and should not be used merely to 'tick a box'.<br>• Consider addressing storage of consent forms i.e. provision of a copy to the patient and retaining within the patients' medical record. | • Provide further information including link to the NPAAC requirements that offer further explanation of complexity.<br>• Amend wording regarding NPAAC requirements to clarify the laboratory requirements<br>• Consider different forms may be required for minors and substitute consent. |
| **Patient information** | • Amend the first point to specify that information may be useful if given before the appointment and made available following the appointment<br>• Consider simple informative videos to provide in advance of pre-test consultations | • Consider links to available resources for health practitioners o guide discussion<br>• Consider the time that should be allowed for the individual to process the information provided prior to the decision to do or not to do the testing? Consider | • Suggest additional resources should be developed in the future in partnership with Aboriginal consumers and communities.<br>• Include reference to Health literacy |

| Themes | Phase 1 consultation | Phase 2 consultation | Comments/Other |
|---|---|---|---|
| | • Avoid being prescriptive in the approach as there are many new and evolving ways to assist the patient in ways the patient prefers<br>• Consider adding that health professionals should prepare patients to receive inconclusive or potentially life changing results, as these are both possible test outcomes<br>• Consider adding that health professionals should refer patients to support groups and patient reference groups when referred for testing and when they receive their test results<br>• Consider adding that health professionals should ensure that patients have time between receiving the consent form or supporting materials, and providing consent | a flow chart of steps and processes required available for the practitioner and participant.<br>• Include reference to the patient information that accompanies the clinical consent form.<br>• Suggest including the word accessible before multi-modal<br>• Note the option to receive the genomic information and consent form should be given with enough time for patients to read it and process the information so they can discuss their concerns at their genomics appointment. Also, to take material away to consider in privacy as well as to seek other advice.<br>• Note additional resources to be developed in partnership with consumers to appropriately convey the diversity of the Aboriginal community<br>• | |
| **Aboriginal and Torres Strait Islander peoples** | • A separate section on Aboriginal and Torres Strait Islanders is required. | • Note, trained aboriginal Liaison and health workers in genetics should be made available, by telehealth, as needed.<br>• Amend to include 'limited' or 'lack of' genetic reference data/literature for Aboriginal and Torres Strait Islander Peoples to assist in diagnosis, which needs to be discussed with participants.<br>• Health professionals should receive training on how to support aboriginal and Torres Strait islanders<br>• Aboriginal and Torres Strait Island Peoples Community need to be included in consumer feedback and consultation groups.<br>• Genetically, the Aboriginal population is the oldest and the most diverse in the world. The past colonial experience for Aboriginal people marked with racism and discrimination, the stealing of children, loss of identity, knowledge, culture and land. This has led to the mistrust of government services by Aboriginal and Torres Strait Islander peoples, and the ongoing concern around data and information sovereignty.<br>• Clinical testing, collecting, storing, disseminating and using of genomics clinical data and information will | • Relevant local guidance to be followed in response to address feedback that is not specific to genomics and/or consent, but applicable to the provision of healthcare to Aboriginal and Torres Strait Islander peoples.<br>• Further address that cultural practice, belief and support systems may generate unique impacts of genomic testing for Aboriginal peoples. |

| Themes | Phase 1 consultation | Phase 2 consultation | Comments/Other |
|---|---|---|---|
| | | uniquely impact on Aboriginal people, because of Aboriginal people's cultural practice, belief and support systems; and the possible effects of the genomics information of an individual could have on the broader Aboriginal communities.<br>• It may be necessary for the health practitioner to also seek informed consent, consult with, and/or support from trusted representatives in the community prior relevant clinical genomic testings.<br>• Noting the role of interpreters, it is important to recognise that in some Aboriginal languages there may not be a specific word or description to communicate the process. | |
| **Cultural and linguistic diversity** | • Consider amending this section to extend to the use of telephone interpreters, non-professional interpreters and liaison officers' roles in supporting consent for Culturally and Linguistically Diverse populations<br>• Consider adding that health professionals should be mindful when communicating with people from culturally and linguistically diverse backgrounds as many cultures may not have a specific phrase or term for genomics, and some cultures may have specific guidelines on the treatment of tissue | • Noting the role of interpreters, there are examples where there is no equivalent word in the non-English language.<br>• Health professionals should be guided on how to be supportive of an individual culture and backgrounds when working with patients. | • Relevant local guidance to be followed in response to address feedback that is not specific to genomics and/or consent, but applicable to the provision of healthcare to culturally and linguistically diverse populations |
| **Other communication difficulties (sensory and communication disability)** | • | • Deafness or other special communication needs is quite distinct from those who are not fluent in English or have cultural considerations.<br>• Consideration of other needs including intellectual disability.<br>• Amend to include 'hearing and/or visual impairment' rather than just 'deafness'.<br>• Accessible information Easy Read/Easy English genomic information and consent forms need to be made available to patients. Any website which has genomic information should have accessible features incorporated into them to meet the needs of people with disabilities. | • Suggest separate section for other communication difficulties<br>• While an Easy Read/Easy English version of the materials for people with intellectual disabilities is beyond the scope of this project it should be considered for future development |

| Themes | Phase 1 consultation | Phase 2 consultation | Comments/Other |
|---|---|---|---|
| **Research** | • Amend the last point to make it explicit that in some instances it may not be appropriate to obtain consent for research from the patient<br>• Consider including examples to demonstrate use in research<br>• Consider obtaining consent for research in a separate consent form | • Consider simplifying.<br>• The principal purpose of the "Research" section is to highlight that this form is not for research consent. This should be clearly stated as the very first point of the section.<br>• Consider clarification of consenting to be recontacted, not consenting to further research.<br>• Consider further clarification and safeguards to ensure a patient is aware they can choose not to agree; and aware of the difference between clinical testing and research testing.<br>• Consider further clarification of 'clinically appropriate'. Examples requiring sensitive management of requests include bereaved parents concerning children's DNA, adult offspring concerning parent's DNA and siblings concerning other sibling's DNA.<br>• Broadly, clients and families participate willingly and with altruistic motives for future improvements, in the hope it assists more patients be diagnosed or leads to treatment options and better clinical care or possibly even a cure for their child's genetic condition.<br>• The research component is important but a completely separate issue to genomics testing.<br>• There is a need to include consent to research. For example, in some circumstances genomic test results may reveal a novel or very rare finding which needs to be corroborated by further studies, including research either nationally or internationally.<br>• Further comments on research data management, are captured under 'data management' section. | • Information about research, as relevant to clinical consent to be included in supporting information.<br>• Suggest that information and data sharing without individual patient benefit are beyond the scope of this project and are being addressed through other national initiatives<br>• Note that while research consent may be appropriate and efficient for a clinician researcher who has an established relationship with their patient, this is not the case for most clinicians or patients. Feedback on version 1 suggested that seeking research consent may pose a barrier to obtaining clinical consent for some cultures<br>• Note that the submitted extract from NPAAC requirements is selective and ignores provisions that the informed consent process should clearly state the protocol for re-contacting the patient about findings, including incidental findings, identified during subsequent research projects.<br>• Note also the NPAAC requirement that all requests from researchers for the release of de-identified data from samples submitted for diagnostic testing and of associated laboratory data to biobanks or databases will require approval by the Human Research Ethics Committee with responsibility for oversight of activities of the pathology service |
| **Disclosure of results** | • Consider removing the second sentence in point four<br>• Add that health professionals should document in writing if a patient declines to be notified about incidental findings<br>• Consider amending the last point to reassure patients that appropriate measures will be in place to respect | • Note different laboratory protocols nationally and internationally on return of incidental findings.<br>• This section should include expectations about the timing and mode of result deliver so patient expectations and anxieties are managed.<br>• Consideration of test results in the event of a patient's death or loss of capacity. Future | • While identifying an approach for the return of incidental findings is beyond the scope of this project, it should be considered in the future.<br>• Consider timing, mode and protocols for return of incidental findings to manage patient expectations.<br>• Consider circumstances in which disclosure of results does not require consent (e.g. to prevent a serious |

| Themes | Phase 1 consultation | Phase 2 consultation | Comments/Other |
|---|---|---|---|
| | their decision if they choose to not be informed of any incidental findings<br>• Consider rewording incidental findings point five to give patients and option to find out about these or not | generations should have the choice to obtain this information or not.<br>• Consider here using term 'additional' findings instead of 'incidental' findings.<br>• Note, there may be an expectation from patients that they are getting more than what the clinical question is asking.<br>• Consider disclosure of results in the future. | threat to another's health) but that disclosure may occur without revealing their identity |
| **Data management** | • Consider adding further information on how data is stored and where it is shared, specifically noting that for diagnostic purposes an individual's data may be shared, including internationally<br>• Consider clarifying if this section covers data used in research | • The relevant data management policy held by individual organisations should be made available to both health practitioner and the patient.<br>• Details about information security should not be added in any detail to the guide as this opens the health care practitioner to questions they are not qualified to answer.<br>• Consider the retention and destruction of genomic material and data, includes consideration of the potential benefits to the patient, their family and community in the future of the retention of genomic material and data.<br>• Consider the return of tissue to patients for religious purposes.<br>• Note that Aboriginal and Torres Strait Islander data sovereignty and ownership, collective and individual privacy, IP, storage remains an ongoing discussion.<br>• Note, there needs to be a clear understanding on how data will be shared for research and consent forms for specific research projects should be given when the time arises for a patient to be considered for a research project.<br>• Note, additional considerations would be required for consent to research, including data descriptions, re-identification processes, withdrawal, management of blood/saliva/tissue samples, how and where data will be stored, shared and used for research projects. | • Suggest that information and data sharing without individual patient benefit are beyond the scope of this project and are being addressed through other national initiatives |
| **Re-testing and re-analysis** | • Consider amending guidance material to state that consent does not need to be reobtained for re- | • Considerations for further testing and analysis is should be incorporated into ongoing and supported | |

| Themes | Phase 1 consultation | Phase 2 consultation | Comments/Other |
|---|---|---|---|
| | analysis and re-testing if the purpose of the test/clinical indication is the same<br>• Consider amending to state that that consent should be reobtained for re-testing or re-analysis if the clinical indication changes | conversation between the patient and health professional.<br>• Health professionals should explain other mechanisms in place to search for a diagnosis. | |
| **Other** | • Encourage doctors to document what information materials were provided to patients, for liability purposes<br>• Consider online consent processes that involve animation and record of consent such as those already supported by NSW Health | • Consent information needs to be provided in accessible format such as through an Easy Read/Easy English documents, document readers or form readers. There is no reference to give people the option to ask for information in other formats and we wonder how many health professionals would think to offer information in accessible formats.<br>• Address how a consumer can withdraw their consent, specifically what can be done if they change their mind about undertaking a test.<br>• Costs of all testing (including re-testing) should be disclosed prior to consent.<br>• Terminology Clinic | |

**Table 3 | Patient information and other consultation notes**

| Patient information | Phase 1 consultation | Phase 2 consultation | Patient information working group |
|---|---|---|---|
| | • Consider providing information about the Moratorium on Genetic Tests in Life Insurance to ensure that patients are not deterred from testing because of the possible insurance implications<br>• Provide further information to ensure patients understand how their information will be used to 'advance scientific knowledge' and for 'other scientific purposes'<br>• Ensure that supporting information is provided with the consent form<br>• Ensure that further detailed supporting information is available upon request<br>• Provide further guidance on the implications for the family if a family member chooses to undergo genetic or genomic testing, what circumstances can information be released by law, and insurance consequences including what information an insurance company can request and what information they need to be informed of<br>• Provide more information on how results will be shared to care for family (relatives)<br>• Participants commented that further information is needed on the application of data including if it will be used ethically, used internationally for research, why samples cannot be returned and that in some instance's labs share internationally as part of the diagnostic process<br>• Provide information to patients to inform them of the safeguards in place to ensure that incidental findings will not be released to them if they do not wish to find out about this information<br>• Information for patients on how their data will be managed<br>• Supporting information should be provided to help individuals understand when their data can be | • Keep simple.<br>• Explain the outcomes of testing, the benefits and the risks.<br>• Consider in include detail around the definitions of the types of testing as shown on the consent form<br>• Include relevant links to information where patients and HCPs can find out more.<br>• Include relevant links to support groups.<br>• Recommend plain English, including the option for document readers and form readers<br>• Recommend use of visuals/graphics/pictures<br>• Recommend different mediums – Easy read, Easy English, visuals, videos, interactive options.<br>• Should be given in advance of their health appointment.<br>• Recommend a glossary of terms<br>• Mention of patient decision/choice whether to undergo genomic testing<br>• Genetic vs genomic - This difference is already being dropped by clinicians and specialist groups so why labour it.<br>• The focus should be what genomics testing means for the patient. Include topics such as why the test is being considered, what it is testing.<br>• Consider information on sample management 'How is the sample collected; 'What can I expect when I have my samples collected? 'What happens to the sample/s?'.<br>• Consider results management. 'What is the process and how long does it take to receive results?' 'What is the likelihood of each type of result", "What are | • The written resource will align with the content inclusions within the video, to offer multimodal access to foundational information to support patients consenting to a genomic test.<br>• User friendly language<br>• Easy first port of entry<br>• Direct to further information and discussion regarding complex considerations (e.g. insurance).<br>• Avoid repetition<br>• Avoid complex terminology<br>• MUST include next steps; addressing support groups<br>• MUST include the 3 potential outcomes – find an answer, not find an answer, uncertain findings<br>• Should address incidental findings, and implications for family members<br>• Should apply to ANY genetic/genomic test<br>• Should provide balanced information to support informed consent<br>• Should align to the consent form inclusions |

| Patient information | Phase 1 consultation | Phase 2 consultation | Patient information working group |
|---|---|---|---|
| | shared (both with and without their consent) and the enabling legislative instruments and policy directives<br>• Supporting information should be provided to patients about their obligations to notify insurers about their genetic/genomic testing results and the impacts of the Moratorium on Genetic Tests in Life Insurance on the use of genetic tests in insurance<br>• Supporting information should be provided to patients to guide individuals to decide whether to receive incidental findings<br>• Consultations highlighted that participants would like to understand the process that health providers and organisations use to notify family members of genetic/genomic testing results | incidental findings, and does the patient get to choose whether or not to receive them?".<br>• Consider "What are some of the possible next steps?" "When may retesting and reanalysis be needed?"<br>• Consider data management; privacy and confidentiality. Clients need to be informed of the criteria for personal and genetic data sharing for diagnostic purposes, for example which health professionals' data can be shared with.<br>• This should have clear explanation of the implications to genetic relatives, the importance of sharing information with other family members, and the questions associated.<br>• Feedback highlighted that if research is included, additional information is required, including data definitions, storage, risks and benefits, what is being shared e.g. blood/saliva/tissue sample, phenotypical information, family history, behavioural, cognitive, physical etc.<br>• Consider what types of insurance. Consider the level of focus on insurance. This issue is less pertinent to a patient who already has a medical condition, however, remains important for their relatives. | |

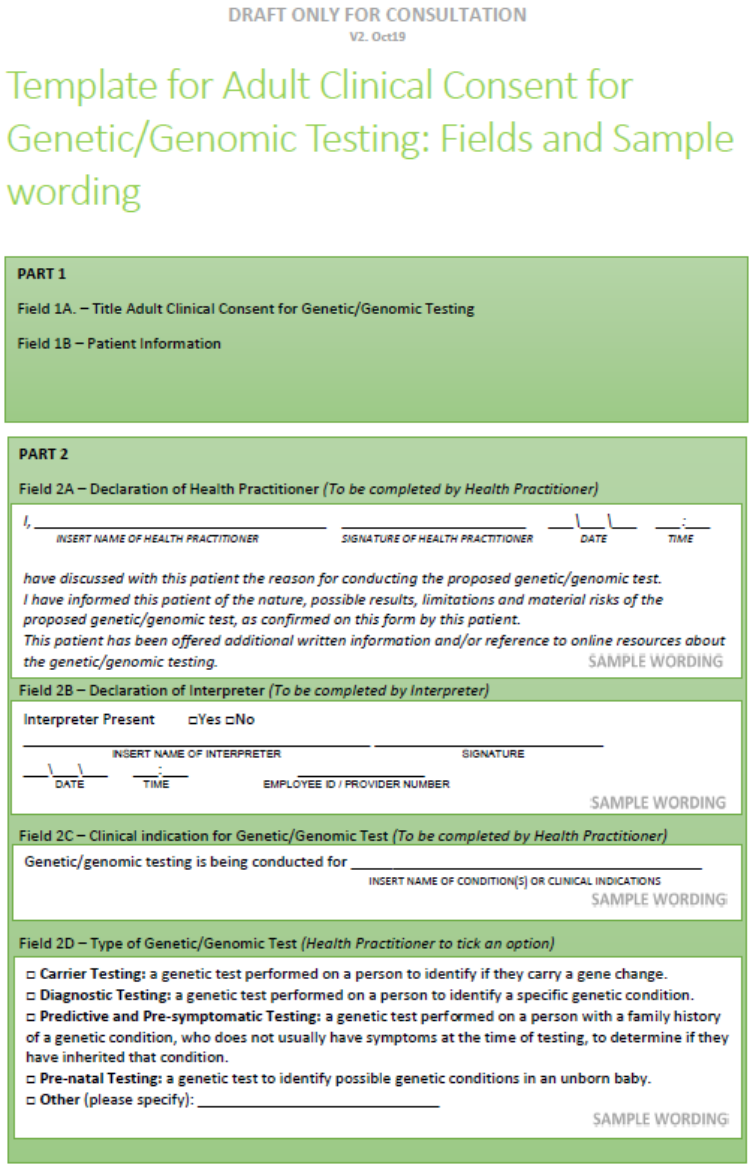
# Appendix B – Phase 1 consultation

This appendix provides detailed consultation findings on the consent form template, the guidance for health professionals and the suggested patient information from the consumer focus group, the survey and the interviews.

## B.1 Consent form template version 1

This section includes the draft template utilised for the first phase of consultation. Contributions to the inclusions and sample wording within the template by the NSW Agency for Clinical Innovation and Australian Genomics are gratefully acknowledged.

**Figure 1 | Consent form template version 1**



DRAFT ONLY FOR CONSULTATION
V2. Oct19

# Template for Adult Clinical Consent for Genetic/Genomic Testing: Fields and Sample wording

**PART 1**

Field 1A. – Title Adult Clinical Consent for Genetic/Genomic Testing

Field 1B – Patient Information

**PART 2**

Field 2A – Declaration of Health Practitioner *(To be completed by Health Practitioner)*

I, _____ _____ _________ __:__
INSERT NAME OF HEALTH PRACTITIONER | SIGNATURE OF HEALTH PRACTITIONER | DATE | TIME

*have discussed with this patient the reason for conducting the proposed genetic/genomic test.*
*I have informed this patient of the nature, possible results, limitations and material risks of the proposed genetic/genomic test, as confirmed on this form by this patient.*
*This patient has been offered additional written information and/or reference to online resources about the genetic/genomic testing.*
SAMPLE WORDING

Field 2B – Declaration of Interpreter *(To be completed by Interpreter)*

Interpreter Present ☐Yes ☐No

_____ _____
INSERT NAME OF INTERPRETER | SIGNATURE

_________ __:__ _____
DATE | TIME | EMPLOYEE ID / PROVIDER NUMBER

SAMPLE WORDING

Field 2C – Clinical indication for Genetic/Genomic Test *(To be completed by Health Practitioner)*

Genetic/genomic testing is being conducted for _____
INSERT NAME OF CONDITION(S) OR CLINICAL INDICATIONS

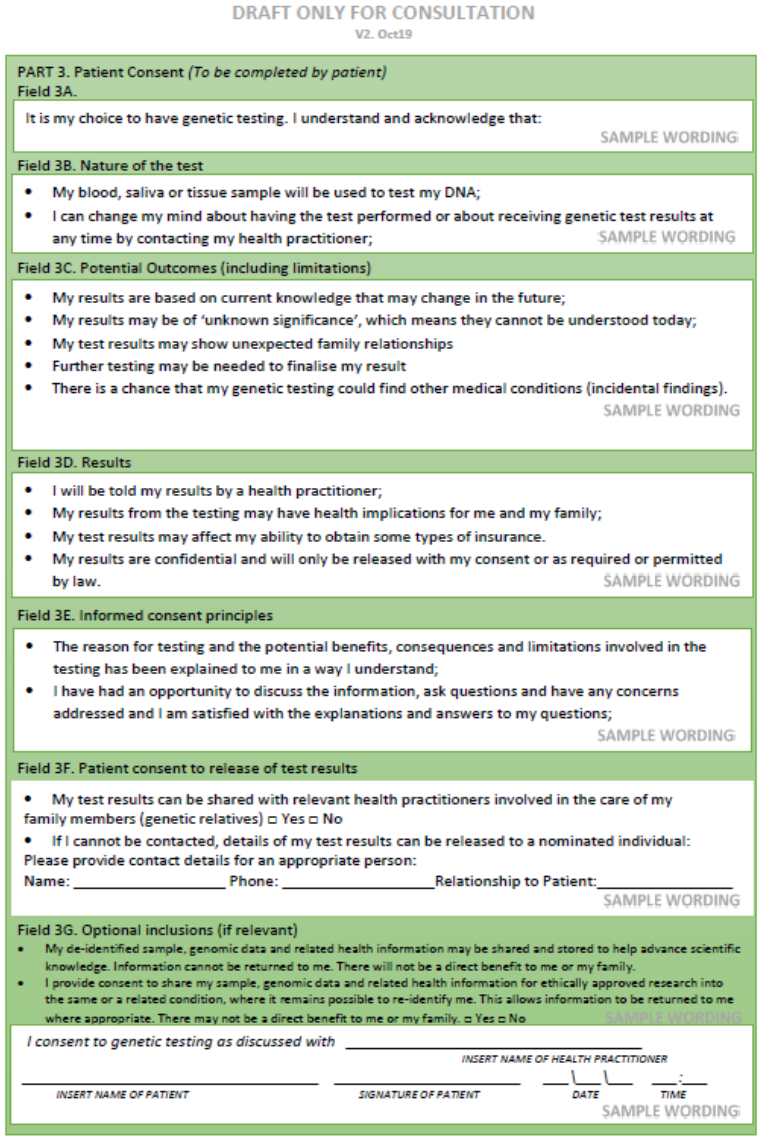
SAMPLE WORDING

Field 2D – Type of Genetic/Genomic Test *(Health Practitioner to tick an option)*

☐ **Carrier Testing:** a genetic test performed on a person to identify if they carry a gene change.
☐ **Diagnostic Testing:** a genetic test performed on a person to identify a specific genetic condition.
☐ **Predictive and Pre-symptomatic Testing:** a genetic test performed on a person with a family history of a genetic condition, who does not usually have symptoms at the time of testing, to determine if they have inherited that condition.
☐ **Pre-natal Testing:** a genetic test to identify possible genetic conditions in an unborn baby.
☐ **Other** (please specify): _____

SAMPLE WORDING

Acknowledgements to the NSW ACI Clinical Genetics Executive Committee; and the Australian Genomics Health Alliance.



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V2. Oct19

PART 3. Patient Consent *(To be completed by patient)*
Field 3A.

It is my choice to have genetic testing. I understand and acknowledge that:
SAMPLE WORDING

Field 3B. Nature of the test

- My blood, saliva or tissue sample will be used to test my DNA;
- I can change my mind about having the test performed or about receiving genetic test results at any time by contacting my health practitioner;

SAMPLE WORDING

Field 3C. Potential Outcomes (including limitations)

- My results are based on current knowledge that may change in the future;
- My results may be of 'unknown significance', which means they cannot be understood today;
- My test results may show unexpected family relationships
- Further testing may be needed to finalise my result
- There is a chance that my genetic testing could find other medical conditions (incidental findings).

SAMPLE WORDING

Field 3D. Results

- I will be told my results by a health practitioner;
- My results from the testing may have health implications for me and my family;
- My test results may affect my ability to obtain some types of insurance.
- My results are confidential and will only be released with my consent or as required or permitted by law.

SAMPLE WORDING

Field 3E. Informed consent principles

- The reason for testing and the potential benefits, consequences and limitations involved in the testing has been explained to me in a way I understand;
- I have had an opportunity to discuss the information, ask questions and have any concerns addressed and I am satisfied with the explanations and answers to my questions;

SAMPLE WORDING

Field 3F. Patient consent to release of test results

- My test results can be shared with relevant health practitioners involved in the care of my family members (genetic relatives) ☐ Yes ☐ No
- If I cannot be contacted, details of my test results can be released to a nominated individual:

Please provide contact details for an appropriate person:
Name: _____ Phone: _____Relationship to Patient:_____
SAMPLE WORDING

Field 3G. Optional inclusions (if relevant)

- My de-identified sample, genomic data and related health information may be shared and stored to help advance scientific knowledge. Information cannot be returned to me. There will not be a direct benefit to me or my family.
- I provide consent to share my sample, genomic data and related health information for ethically approved research into the same or a related condition, where it remains possible to re-identify me. This allows information to be returned to me where appropriate. There may not be a direct benefit to me or my family. ☐ Yes ☐ No

SAMPLE WORDING

*I consent to genetic testing as discussed with* _____
INSERT NAME OF HEALTH PRACTITIONER

_____ _____ _________ __:__
INSERT NAME OF PATIENT | SIGNATURE OF PATIENT | DATE | TIME

SAMPLE WORDING

Acknowledgements to the NSW ACI Clinical Genetics Executive Committee; and the Australian Genomics Health Alliance.

## B.2 Consumer focus group

This section is structured to reflect the discussion during the focus group. Consumers had the opportunity to comment on the consent form template, however a large proportion of their feedback was also on supporting information provided with the template.

### B.2.1 Information to support patients

**Patients need more guidance throughout the consent process**

Focus group consultations highlighted that individuals could benefit from further information to support their understanding of key concepts discussed during the consent process. These areas include:

Further information on genomic data sharing: Consultations highlighted that individuals' perceptions of how their data will be used is dependent on their experience. For example, some individuals with a rare condition may expect that their data will be contributed to a pool of information to advance health care. Supporting information should be provided to help individuals understand when their data can be shared, both with and without their consent, and the enabling legislative instruments and policy directives.

Improved understanding of information used for insurance purposes: Focus group participants discussed varying levels of awareness amongst individuals of their obligation to notify insurers about the results of any genetic/genomic testing they undergo. Supporting information should be provided to patients about their obligations to notify insurers about their genomic testing results and the impacts of the moratorium on the use of genetic tests in insurance.

Clarification on incidental findings process: Participants suggested that the consent process should be used to help individuals understand the concept of incidental findings, to provide guidance to decide whether to receive these results and offer support if they chose to. Supporting information should be provided to patients to guide individuals to decide whether to receive incidental findings.

Understanding of the process for notifying family members: There may be implications for family members if an individual who has undergone genomic testing receives results that indicate an inherited disease. Consultations highlighted that participants would like to understand the process that health providers and organisations use to notify family members of genomic testing results. Participants discussed alternative models whereby patients have a certain period to notify their family members, prior to the respective health body notifying the relevant family members.

### B.2.2 Guidance for health professionals

**Stakeholders noted specific areas for support and upskilling of health practitioners to undertake the genomic test consent process**

The focus group noted these specific skills and knowledge as important for front line staff, and thought it was an opportunity for development. The consent process is very information dense and requires a good knowledge base to sufficiently explain. The following are suggested areas to focus training and support for health professionals, that should also be reflected in consent materials:

Managing patient expectations: Participants discussed that individuals should be prepared to receive inconclusive or potentially life changing results, as these are both possible test outcomes. Participants suggested that clinicians should use the consent process to prepare individuals for both scenarios.

Being culturally and linguistically sensitive: Consultations highlighted that many cultures may not have a specific phrase or term for genomics, and that some cultures may have specific guidelines on the treatment of tissue. Participants discussed that clinicians should be mindful of this when communicating with people from culturally and linguistically diverse backgrounds.

Referring to appropriate health professionals and support groups: Participants discussed engaging with support groups and patient reference groups when referred for testing and when they receive their test results.

Consultations highlighted that clinicians should consider if there are support groups that patients should be referred to, including if there are groups that culturally and linguistically diverse people could be referred to.

Ensure that an appropriate amount of time is set aside to obtain informed consent: Consent is often collected in scenarios where there is little time for clinicians to explain in detail the implications of genomic testing. Participants discussed the need for permitting more time during consultations to obtain informed consent for a genomic test.

**Participants discussed their views on how consent should be obtained in a clinical setting:**

Use a range of mediums to obtain consent: Participants expressed that the preferred mode to be consented is dependent on an individual's experiences. A range of modes including paper based and digital forms and videos should be available to for an individual to select their preferred mode to give consent and to receive further supporting information about the process.

Give individuals the opportunity to reflect on the implications of the test: The implications of genomic testing extend beyond the individual that is being tested and to their family. Participants discussed having time between receiving the consent form or supporting materials and providing consent.

One participant highlighted that they would prefer to receive the consent form and supporting materials prior to their medical appointment so they are able to reflect on the implications of the test and use the appointment to ask clarifying questions. Participants highlighted that this would only be suitable in scenarios where the type of test being conducted is not urgent. In urgent situations this information would need to be provided during the consultation, with an adequate amount of time for the individual to consider the implications.

## B.3 Stakeholder surveys

This section outlines the results of the closed survey questions, as well as the responses to the open questions, where participants chose to leave a comment about the consent form template.

### B.3.1 Feedback on the consent form template

Participants were asked if the following fields in the consent form template and the guidance material should be included, Table 4 below.

**Table 4 | Survey responses to the inclusion of fields in the consent form template**

| Field | Agree | Disagree |
|---|---|---|
| Field 2A – Declaration of health practitioner | 91% | 9% |
| Field 2B – Declaration of interpreter | 89% | 11% |
| Field 2C – Clinical indication for genetic/genomic test | 94% | 6% |
| Field 2D – Type of genetic/genomic test | 84% | 16% |
| Field 3A – Patient consent | 92% | 8% |
| Field 3B – Nature of the test | 90% | 10% |
| Field 3C – Potential outcomes (including limitations) | 89% | 11% |
| Field 3D – Results | 88% | 12% |
| Field 3E – Informed consent principles | 93% | 7% |
| Field 3F – Patient consent to release of test results | 89% | 11% |
| Field 3G – Optional inclusions (if relevant) | 79% | 21% |
| Field 3H – Health practitioner and patient signatures | 93% | 7% |

Participants also made specific comments about fields within the consent form template.

These include:

**Field 2A – Declaration of Health Practitioner**

Survey participants suggested change the date to include forward slashes from ______ to __/__/__.

Consider moving this to the end of the form and simplifying.

Consider removing 'written information' as it may not always be required, noting clinicians and patients should work together to select appropriate information based on the clinical evidence and the patient's informed preferences.

**Field 2B – Declaration of Interpreter**

Consider amending this to accommodate for telephone interpreters, non-professional interpreters and liaison officers' roles in supporting consent for Culturally and Linguistically Diverse populations.

Survey participants suggested change the date to include forward slashes from ______ to __/__/__.

**Field 2C – Clinical indication for Genetic/ Genomic Test**

Consider amending language to 'Genetic/genomic testing by [ ] single gene [ ] gene panel [ ] exome [ ] genome [ ] other – specify _____ is being conducted for _____ insert name of condition(s) or clinical indication(s)'.

**Field 2D – Type of genetic/genomic test**

Consider including ‘segregation’ testing.

Suggested editing language used in test definitions:

Predictive testing: is a genetic test to identify a gene change in a person at risk of a genetic condition, who does not usually have symptoms. (The current definition does not apply to patients without a family history).

**Field 3A – Patient Consent**

Consider removing ‘acknowledge’.

**Field 3B – Nature of the test**

The participants that agreed wondered if it was possible or practical to change your mind, and that the use of the wording ‘DNA’ and or ‘sample’ is too restrictive.

The participants that disagreed thought this section was unnecessary.

**Field 3C – Potential Outcomes**

The participants that disagreed commented that the results may not identify a medical issue. The participants that agreed also commented that the results may not identify a medical issue or that they may be of unknown significance or incidental.

Participants also suggested amending the use of certain words in this field to align with current terminology:

Consider changing first point to ‘results are based on current knowledge and technology’ and consider removing ‘that may change in the future’.

In the second point, consider changing ‘unknown’ to ‘uncertain’.

In the fifth point consider changing ‘finalise’ to ‘understand’.

Consider using word ‘unrelated’ in definition of incidental findings.

**Field 3D – Results**

The participants that agreed commented about needing greater clarification on the intent of the use of the word ‘family’ and what the health implications for the family are.

Participants also commented that further clarification of the legal permissions to release information including what circumstances information can be released by law, and of insurance consequences including what information an insurance company can request and information that they are required to know.

Participants also commented on the communication of results, specifically that the use of ‘health practitioner’ is too generic and that someone with genetic experience is more appropriate.

The participants that disagreed commented that ‘health practitioner’ should be more specific for example a doctor, genetics councillor or genetics specialist.

Patient information: Further guidance on the implications for the family, if a family member chooses to undergo genetic or genomic testing, what circumstances can information be released by law, and what are the insurance consequences (including what information an insurance company can request and what information they need to be informed of).

**Field 3E – Informed Consent Principles**

Consider further simplification.

**Field 3F – Patient consent to release of test results**

The participants that agreed commented on ensuring each point is separately agreed to as this field has two separate issues that should be clearly differentiated. This is because there may be cases where a person consents to family use but not to the release to another person. The participants that disagreed also commented about this issue.

Participants also commented that ‘nominated individual’ may need to be updated periodically.

Participants commented that the first dot point should be amended to reflect that their information can be released for care of their genetic relatives without revealing their identity. There were also comments that further information should be provided on how a patient's information will be used for the care of their family members.

Patient guidance: Provide more information on how results that will be shared to care for family (relatives).

**Field 3G – Optional inclusions (data sharing/research)**

Of the participants that agreed, most commented on the need to clarify where the yes/no box applies, specifically that each point should have a yes/no box. They also commented that this is a valuable section.

Of those that disagreed, they perceive that consent for research should be included in a separate form. Participants who disagreed also desired further clarification on the intention and application of this field specifically around the sharing of their data.

Patient guidance: Participants commented that further information is needed on the application of data including if it will be used ethically, used internationally for research, why samples cannot be returned and that in some instance's labs share internationally as part of the diagnostic process.

**Field 3H – Health practitioner and patient signatures**

Survey participants suggested changing the date to include forward slashes ______ to __/__/__.

**Overarching**

Review the use of semi-colons.

Review the use of 'genetic/genomic' testing.

### B.3.2 Feedback on the guidance for health professionals

This section outlines the results of the closed survey questions, as well as the responses to the open questions, where participants chose to leave a comment about the guidance for health professionals.

Participants were asked if the following fields in the consent form template and the guidance material should be included. Table  overleaf presents these results.

**Table 5 | Survey responses to the Guidance for Health Professionals**

| | Agreement | Strongly agree | Agree | Neither agree nor disagree | Disagree | Strongly disagree |
|---|---|---|---|---|---|---|
| *Informed consent* | | | | | | |
| Processes for obtaining clinical consent for genetic/genomic testing must provide the patient sufficient time and opportunity for consideration. | 95% | 68% | 27% | 5% | 0% | 0% |
| Clinical consent for genetic/genomics testing must be voluntary (i.e. without coercion). | 97% | 81% | 16% | 3% | 0% | 0% |
| Clinical consent for genetic/genomic testing must be specific to the patient and the genetic/genomic test at the time which consent is sought. | 89% | 53% | 36% | 6% | 3% | 2% |
| Consent for genetic/genomic testing must be obtained by a patient with the capacity to consent; that is, a person with the ability to understand the implications of having the genetic/genomic test. | 90% | 64% | 26% | 4% | 4% | 2% |
| *Written consent form* | | | | | | |
| A written form is best practice for supporting informed consent processes for all genetic and genomic testing. | 71% | 33% | 38% | 17% | 11% | 2% |
| Written consent is only required for those tests with potential complex issues associated with testing (i.e. Level 2 DNA tests as per NPAAC guidelines). | 41% | 9% | 32% | 25% | 27% | 7% |
| An adequate written consent form assists in maintaining accurate patient records and supports practitioners in providing appropriate information to their patients. | 85% | 45% | 40% | 8% | 7% | 1% |
| A consent form does not discharge the duty of the health practitioner to ascertain whether a person understands the nature of, risks involved in and potential outcomes of the genetic/genomic test. | 95% | 55% | 40% | 4% | 1% | 0% |
| *Patient information sheets* | | | | | | |
| Pre-prepared material about the genetic/genomic test may be useful if given to the patient as a means of stimulating discussion and for guiding the health practitioner when informing the patient. | 94% | 40% | 54% | 4% | 1% | 1% |
| Pre-prepared material should not substitute for the health practitioner discussing the nature of, risks involved in and potential outcomes of the genetic/genomic test with the patient. | 98% | 62% | 36% | 2% | 0% | 0% |
| *Cultural and linguistic diversity* | | | | | | |
| Consideration must be given to cultural and linguistic diversity and the relevant implications for obtaining consent. | 100% | 59% | 41% | 0% | 0% | 0% |

| | Agreement | Strongly agree | Agree | Neither agree nor disagree | Disagree | Strongly disagree |
|---|---|---|---|---|---|---|
| Where a patient is not fluent in English or has other communication needs, appropriate action must be taken to assist the patient to understand the nature of, risks involved in and potential outcomes of the genetic/genomic test. | 100% | 71% | 29% | 0% | 0% | 0% |
| A professional interpreter must be involved to communicate information to a non-English speaking patient. | 84% | 50% | 34% | 12% | 4% | 0% |
| The consent form signed by a non-English speaking patient should contain a statement signed by the Interpreter that he/she has interpreted the content of the form and information supplied by the treating practitioner to the patient. | 83% | 45% | 38% | 12% | 6% | 0% |
| *Research* | | | | | | |
| The potential utilisation of specimens and/or genetic/genomic data for the purposes of research, may arise during clinical care. | 91% | 41% | 50% | 4% | 3% | 2% |
| Consent to research should occur in accordance with NHMRC guidelines. | 92% | 53% | 39% | 7% | 1% | 0% |
| Consideration is required as to how consent to research is collected in relation to clinical consent. This is to ensure there is no inadvertent effect on patient understanding of the nature of, risks involved in and potential outcomes of the clinical genetic/genomic test. | 94% | 52% | 42% | 5% | 1% | 0% |
| *Disclosure of results* | | | | | | |
| Consent processes for genetic/genomic testing must include mechanisms to contact patients with information of the findings. | 94% | 62% | 32% | 6% | 0% | 0% |
| Consent processes must include protocols for disclosure of sensitive genetic information including familial risk and unexpected family relationships. | 93% | 51% | 42% | 6% | 0% | 1% |
| Processes should include consent to release of information to genetic relatives. | 86% | 48% | 38% | 13% | 0% | 1% |
| Management of incidental findings should occur in accordance with relevant jurisdictional and/or laboratory policies. This is likely to require consultation between the laboratory and the referrer to determine if the finding is clinically actionable prior to laboratory reporting. | 90% | 44% | 46% | 9% | 0% | 1% |
| Consideration should be given to an individual's decision not to be informed of specified genetic/genomic information, including results of unknown significance and incidental findings. | 91% | 42% | 49% | 5% | 4% | 0% |

| | Agreement | Strongly agree | Agree | Neither agree nor disagree | Disagree | Strongly disagree |
|---|---|---|---|---|---|---|
| *Data management* | | | | | | |
| Consent processes should consider the retention and destruction of genomic material and data. | 83% | 36% | 47% | 11% | 5% | 1% |
| Data access and management should consider the cultural beliefs of patients. | 75% | 34% | 41% | 24% | 0% | 1% |
| Consideration should be given to the ownership of data and genetic/genomic material, including the interests and rights of Aboriginal and Torres Strait Islander peoples. | 84% | 46% | 38% | 16% | 0% | 0% |
| Processes should consider consent to data sharing as applicable to clinical tools utilised by health practitioners in delivery of care. For example, sharing and comparing phenotypic data. | 85% | 43% | 42% | 9% | 5% | 1% |
| There are Commonwealth, State and Territory laws that specify how patients' personal information, including health information, can be collected, used, stored and disclosed. Consent processes must consider variation across jurisdictions. | 82% | 39% | 43% | 14% | 2% | 2% |
| *Re-analysis and re-testing* | | | | | | |
| Consent should be reobtained for re-testing due to advances in genomic technologies and/or changes to patient circumstances over time. This would enable consent to be specific to the nature of, risks involved in and potential outcomes of the genetic/genomic test. | 68% | 33% | 35% | 15% | 13% | 4% |
| Consent should be reobtained for re-analysis of an existing sample due to advances in genomic technologies and/or changes to patient circumstances over time. This would enable consent to be specific to the nature of, risks involved in and potential outcomes of the genetic/genomic test. | 52% | 25% | 27% | 22% | 19% | 7% |

Participants also made specific comments about the guidance for health professional. These include:

**Written consent form**

Consider amending to include digital and non-digital modes of 'written' consent.

**Patient information sheets**

Feedback:

Consider simple informative videos to provide in advance of pre-test consultations.

Avoid being prescriptive in the approach as there are many new and evolving ways to assist the patient in ways the patient prefers.

**Cultural and linguistic diversity**

Consider amending this section to extend to the use of telephone interpreters, non-professional interpreters and liaison officers' roles in supporting consent for culturally and linguistically diverse populations.

### Research

The participants who agreed or strongly agreed noted that consent for research should be obtained in a separate consent form.

Participants also commented on including examples to demonstrate how a patient's genetic and genomic information would be used in research.

### Disclosure of results

The participants that agreed, or strongly agreed, commented on the practical implications of ensuring an individual that does not wish to be informed of incidental findings, especially if the results will remain in the patients record. Those who disagreed also commented on this.

Individuals also noted that health practitioners have an important role in discussing incidental findings, to empower an individual to make an informed decision.

Patient information: Provide information to patients to inform them of the safeguards to ensure that any incidental findings are not released to them should they wish to not know about this information.

### Data management

Those who strongly agreed or disagreed with this information commented that data sharing and ownership must be transparent to the patient, and all patients should know that their sample and data is being treated/accessed in a way that they are comfortable with. Those who agreed also wanted clarification on whether this applies to clinical and research data.

Others also commented on the need for a national policy on data sharing, those who neither agreed nor disagreed also commented on this.

Patient information: Information for patients on how their data will be managed.

### Re-testing and re-analysis

Those who agreed or strongly agreed commented that there is no need to reobtain consent if the clinical indication is unchanged.

Those who disagreed commented that there is no need to reobtain consent for re-testing or re-analysis as patients may be happy for this to occur anyway. Others who disagreed commented that consent may only be required for re-analysis and re-testing if the clinical indication changes.

## B.4 Stakeholder interviews

This section summarises the discussion about the consent form template and the guidance for health professionals from the stakeholder interviews.

### B.4.1 Feedback on the consent form template

**Field 2A**

Interview participants agreed that this field should be included in the consent form but commented that it would benefit from clarification of the distinction between genetics and genomics. This could be done through adding that incidental findings are a key distinguishing factor of genomic testing that doesn't apply to single gene testing.

Other participants also commented that the field could benefit from being simplified and may be better suited at the end of the document prior to obtaining the patients consent.

**Field 2B**

Interviewees agreed that this field should be included. They also discussed amending the field to include further information on whether the interpreter is professionally qualified or not.

**Field 2C**

Interviewees did not have any suggested changes to this field. They agreed that this field should be included.

**Field 2D**

Interviewees commented on revising the test definitions and the order that the tests were listed. Interviewees suggested that the test should be ordered to reflect how common they were.

Interviewees suggested clarifying whether whole genome sequencing would be consented to through this form.

**Field 3A**

Interviewees agreed that this field should be included. They also cautioned that it is important the template not oversimplify complex concepts, as how people make decisions about healthcare is complicated.

Interviewees commented on adding that the first sentence should reference that the patient decides to have testing after discussion with their doctor.

**Field 3B**

Interviewees commented that they agreed on this section being included. They also commented about amending the second point to reflect that it may not be possible and out of the health professionals' sphere of control to stop the test being performed if a patient changes their mind.

**Field 3C**

Interviewees commented about adding that the test may not find the cause of the condition.

They also noted that laboratories may have different policies on reporting variants and that in WA laboratories do not report on incidental findings.

Interviewees commented that the content in point four needs to be expanded upon to add more information about obtaining a result.

**Field 3D**

Interviewees agreed with the inclusion of this field. They also suggested simplifying point four and adding further information about the Moratorium on Genetic Tests in Life Insurance in the guidance material to ensure patients are not deterred from testing by the insurance implications.

Patient information: Consider providing information about Moratorium on Genetic Tests in Life Insurance to ensure no patients are deterred from testing because of the possible insurance implications.

**Field 3E**

Interviewees also agreed with the inclusion of this field. They commented on considering the removal of the phrase 'in a way I understand' in the first point. This change was suggested as the interviewee perceived that it is not possible to ensure the patient has an adequate level of understanding, and that it is the duty of the healthcare practitioner to provide a level of information to ensure that they are persuaded the patient understands the test and its implications.

**Field 3F**

Interviewees agreed to this field being included in the form.

Interviewees discussed revising the first point to reflect that most requests are from family members themselves and not health professionals. Others also discussed revising the first point to reflect that the results will also need to be shared with other health professionals involved in their care.

Others discussed that issues may be encountered if the contact information for the nominated individual needs to be stored, as it would need to be easily accessible by other health services. Interviewees also suggested clarifying or adding more information to describe the efforts that will be undertaken to contact the individual before their information is shared with the nominated individual.

**Field 3G**

Interviewees agreed with this field, but also suggested editions to its current state.

Interviewees discussed that consent for research should be obtained for each individual project.

Others discussed the importance of research and that it should not be optional. They also discussed including a new point on the release of information beyond the laboratory for purposes that are not related to the purpose for which the sample and information were collected.

Interviewees also discussed that the field should add that the scientific research is performed in Australia and with oversight by a suitably accredited research ethics committee.

They also discussed amending the section to include a dot point on who to contact if an individual chooses to withdraw their consent to their data being used in research.

Other interviewees discussed clarifying what 'advance scientific knowledge' and 'other scientific purposes' means.

Patient information: Provide further information to ensure patients understand how their information will be used to 'advance scientific knowledge' and for 'other scientific purposes'.

**Field 3H**

Interviewees agreed with the inclusion of this field. They suggested removing the health practitioners name from this section as it is already requested in the first section.

### B.4.2 Feedback on the guidance for health professionals

**Informed consent**

Interviewees discussed that a strong and supportive consent process is necessary to ensure that a patient understands the test and its implications. Part of ensuring this is through providing supporting information with the consent form.

Interviewees discussed the importance of health professionals with genetics specific experience, in ensuring patients understand the testing process and implications. Interviewees specifically mentioned that they found gaps in patients' knowledge when consent for genomic testing was obtained by a non-genetic specialist.

Interviewees also discussed that informed consent is the best tool that is used to enable and facilitate choice, and that it is embedded in legal and organisational structures, but that its implementation in clinical practice can be problematic.

Patient information: Ensure that supporting information is provided with the consent form.

### Written consent form

Interviewees commented that the consent form should include all important information that a patient should be aware of and should serve as a prompt for health professionals to discuss with their patients.

Others also discussed that there are a range of patients, each with different preferences about the level of information they desire. These interviewees suggested the form include a minimal level of information, with further detailed information available and provided upon request.

Interviewees also discussed that information on the consent form should be reviewed to consider if it constitutes a Reasonable Patient Standard (the doctor must disclose all information which would influence the patient, or any other reasonable person, in deciding whether to go ahead).

Patient information: Ensure that further detailed supporting information is available upon request.

### Patient information

Interviewees discussed providing patients with information about issues discussed in the consent form prior to their appointment. Information should also be available following the appointment and to clinicians for their use and to support discussion with their patients

### Cultural and linguistic diversity

Interviewees noted there may be situations in which genetic testing is authorised at the level of the community rather than the individual. They also discussed that Aboriginal and Torres Strait Islander people's cultural sensitivity in genomics is well documented and may require more community involvement.

### Research

Interviewees discussed the inclusion of consent to research in a clinical consent form and suggested separating consent for research and clinical consent.

Interviewees noted that clinicians have limited time to talk through implications of research in a clinical context. They also discussed that often patients consenting to clinical testing are under immense stress and concern for their health, and that obtaining consent to research under these circumstances may be unethical.

Others were not comfortable about obtaining consent for research where research details are not available, and some discussed consent being obtained for each project.

### Disclosure of results

Interviewees commented on incidental findings, particularly that consenting to testing in a clinical setting is usually for a particular purpose and not for additional findings. Others also commented on the need for a national policy on incidental findings.

An interviewee also commented about point four, that the laboratory should be able to determine the clinical validity of a variant without further discussion with the clinician, and that it is for the clinician to decide whether a pathogenic finding is clinically actionable.

Interviewees also commented that it should be documented in writing if a patient declines to be notified about incidental findings.

### Data management

Interviewees noted that data sharing with international databases is part of the diagnostic process. They also discussed adding information to demonstrate how the data might be stored.

One interviewee mentioned that data sharing beyond the testing laboratory should be mandatory.

### Re-testing and re-analysis

Most interviewees mentioned that there is no need to reobtain consent for re-testing and re-analysis if the purpose is unchanged.

An interviewee also mentioned that consent would need to be reobtained for re-testing only if the test has changed and that it is difficult to anticipate the need for re-analysis at the time of obtaining consent; therefore, re-analysis

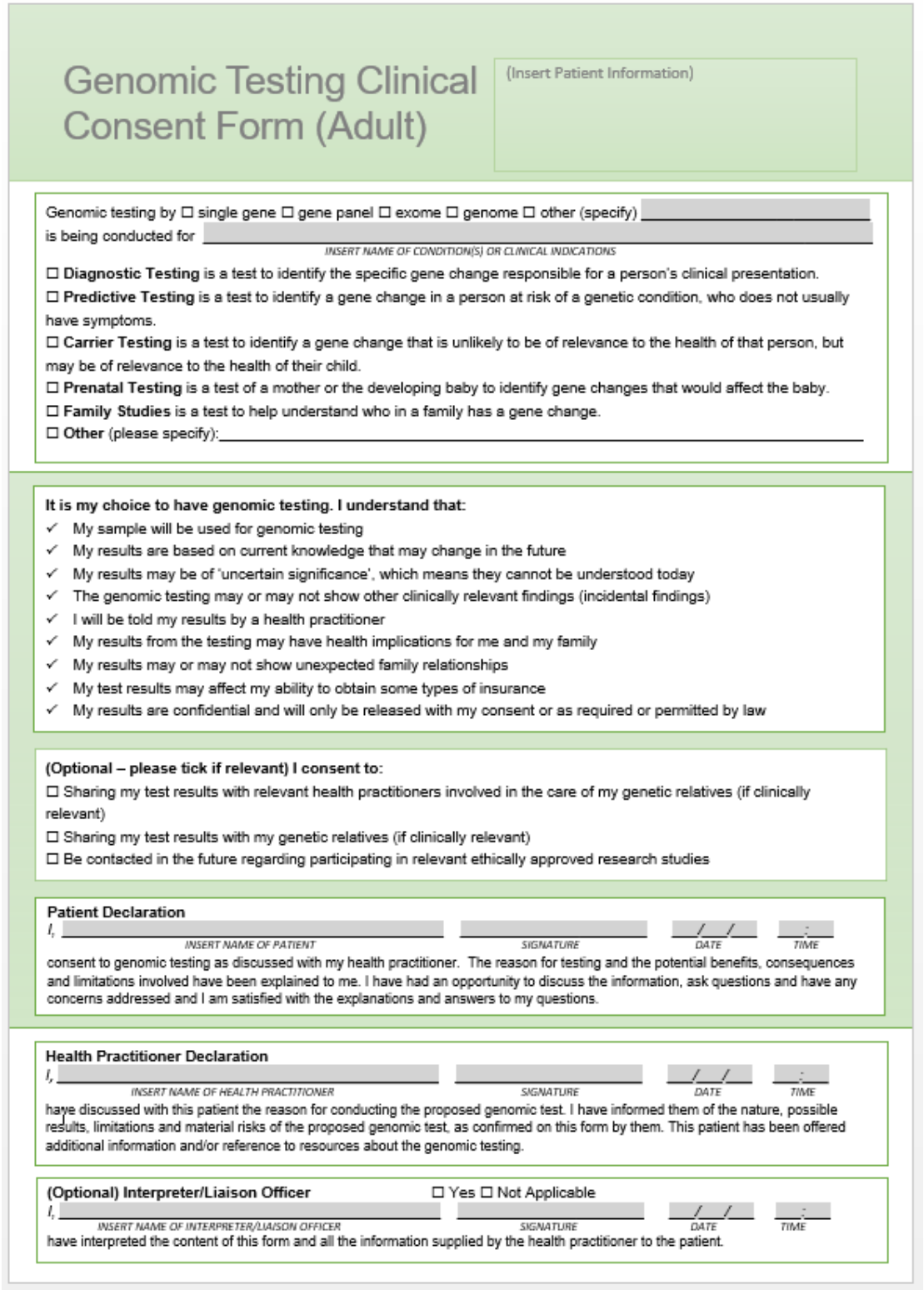
should be initiated by the laboratory without obtaining consent. Another interviewee also mentioned that it should be part of the standard clinical process and that re-analysis may already be conducted on an automated basis.

Interviewees also noted that consent processes are specific to a point in time. The factors at this time may change when the information is reanalysed, which questions the validity of the consent.

# Appendix C – Phase 2 consultation

## C.1 Consent form template version 2

This section includes the draft template utilised for the second phase of consultation. Broad stakeholder input from the phase one consultation on the inclusions and wording within the template are gratefully acknowledged.



Genomic Testing Clinical Consent Form (Adult)

(Insert Patient Information)

Genomic testing by ☐ single gene ☐ gene panel ☐ exome ☐ genome ☐ other (specify) _____
is being conducted for _____
INSERT NAME OF CONDITION(S) OR CLINICAL INDICATIONS

☐ **Diagnostic Testing** is a test to identify the specific gene change responsible for a person's clinical presentation.
☐ **Predictive Testing** is a test to identify a gene change in a person at risk of a genetic condition, who does not usually have symptoms.
☐ **Carrier Testing** is a test to identify a gene change that is unlikely to be of relevance to the health of that person, but may be of relevance to the health of their child.
☐ **Prenatal Testing** is a test of a mother or the developing baby to identify gene changes that would affect the baby.
☐ **Family Studies** is a test to help understand who in a family has a gene change.
☐ **Other** (please specify):_____

**It is my choice to have genomic testing. I understand that:**

- ✓ My sample will be used for genomic testing
- ✓ My results are based on current knowledge that may change in the future
- ✓ My results may be of 'uncertain significance', which means they cannot be understood today
- ✓ The genomic testing may or may not show other clinically relevant findings (incidental findings)
- ✓ I will be told my results by a health practitioner
- ✓ My results from the testing may have health implications for me and my family
- ✓ My results may or may not show unexpected family relationships
- ✓ My test results may affect my ability to obtain some types of insurance
- ✓ My results are confidential and will only be released with my consent or as required or permitted by law

**(Optional – please tick if relevant) I consent to:**

☐ Sharing my test results with relevant health practitioners involved in the care of my genetic relatives (if clinically relevant)
☐ Sharing my test results with my genetic relatives (if clinically relevant)
☐ Be contacted in the future regarding participating in relevant ethically approved research studies

**Patient Declaration**

*I,* _____ _____ __/__/__ __:__
INSERT NAME OF PATIENT SIGNATURE DATE TIME

consent to genomic testing as discussed with my health practitioner. The reason for testing and the potential benefits, consequences and limitations involved have been explained to me. I have had an opportunity to discuss the information, ask questions and have any concerns addressed and I am satisfied with the explanations and answers to my questions.

**Health Practitioner Declaration**

*I,* _____ _____ __/__/__ __:__
INSERT NAME OF HEALTH PRACTITIONER SIGNATURE DATE TIME

have discussed with this patient the reason for conducting the proposed genomic test. I have informed them of the nature, possible results, limitations and material risks of the proposed genomic test, as confirmed on this form by them. This patient has been offered additional information and/or reference to resources about the genomic testing.

**(Optional) Interpreter/Liaison Officer** ☐ Yes ☐ Not Applicable

*I,* _____ _____ __/__/__ __:__
INSERT NAME OF INTERPRETER/LIAISON OFFICER SIGNATURE DATE TIME

have interpreted the content of this form and all the information supplied by the health practitioner to the patient.