A NATIONAL CLINICAL GENOMIC CONSENT PROCESS

Development of standardised consent materials for clinical genomic testing in Australia

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Australian Genomics Health Alliance

A National Clinical Genomic Consent Process

Summary

Challenge: currently in Australia, there is no single, national approach to obtaining patient consent to undertake genetic or genomic testing. Standards and practices are fragmented, and consent forms differ not only between states, but also between genetic service providers. This creates inconsistency in the experience, outcomes and management of patients across Australia, and is a barrier to the flow of health information between jurisdictions.

In response to this need, in 2017 Australian Genomics embarked on a project to develop a clinical genomics consent form that can be adopted nationally, for use across all genetic conditions. The working group (see 'Investigators, above) undertook a systematic review of existing consent forms, developed guidelines for content of consent materials, and drafted a form with supporting information. Professional and public consultation was undertaken against these materials. The finalised National Clinical Genomic Consent Form and Supporting Information was piloted in different jurisdictions. The evaluation of the pilot permitted finalisation of the materials that are now available for adaptation and adoption by interested health services in Australia, and beyond.

In 2019, the Australian Health Ministers' Advisory Council (AHMAC) launched a project led by the NSW Ministry of Health: "Nationally Consistent Templates and Guidance for Consent to Genetic and Genomic Testing", which reflects Action 4A of The Implementation Plan for the National Health Genomics Policy Framework administered by the Project Reference Group on Health Genomics (PRG). We are collaborating with this project to share the outcomes of our work to date, so that the learnings of our consultation and pilot can inform this process.

These will be living documents, undergoing ongoing assessment and refinement to retain currency and reflect regulatory evolution as genomics is mainstreamed throughout the Australian healthcare system.

This report details the process and outcomes of this project.

Project aims and methodology:

1. **To review existing consent materials**
   - (i) Systematically evaluate forms currently in use in Australia, with representative examples from different geographic jurisdictions
   - (ii) Identify characteristics (good and bad) to inform the development of recommendations on content of consent materials.
2. **To develop recommendations and guidelines around genomic consent materials**
   - (i) Determine mandatory content and information
   - (ii) Describe key features of structure, voice and delivery of the consent materials
3. **To draft consent materials based upon the recommendations developed in (2)**
   - (i) Prepare a consent form
   - (ii) Prepare supporting information
4. **Undertake a broad consultation on the materials**, inviting comment and input from the Australian public and professionals
   - (i) Develop a consultation form, to support standardised and systematic evaluation of the materials developed

(ii) Request / invite review from the Australian public and professional groups / individuals

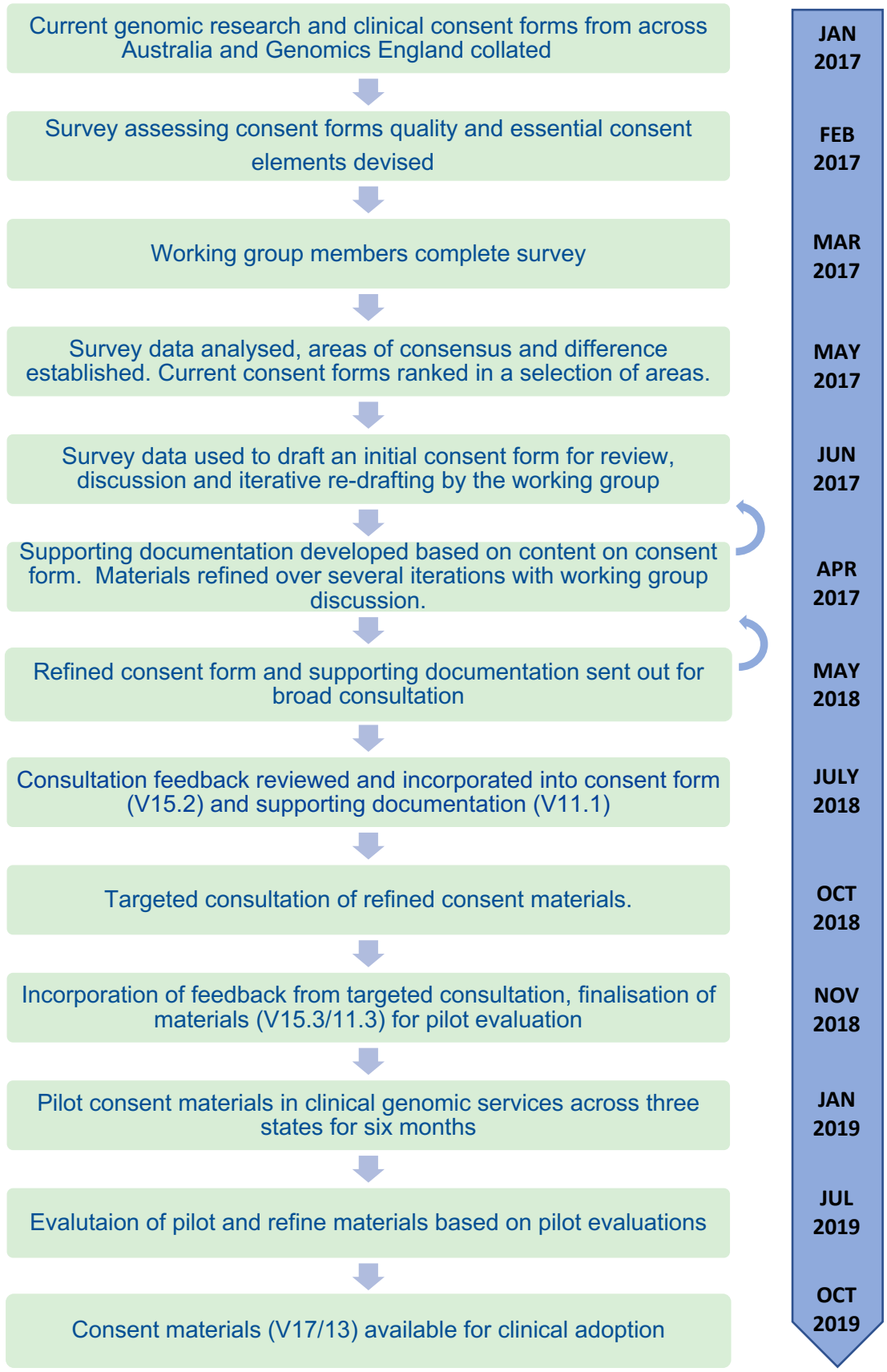
(iii) Assimilate and evaluate responses

5. **Re-draft consent materials based upon the consultation in (4)**
6. **Undertake a trial roll-out of the consent materials in different states**
   (i) Pilot the use of the materials,
   (ii) Evaluate the experiences of health professionals to the materials
7. **Re-draft consent materials based upon the pilot evaluation**
   (i) Based on a six-month trial, the materials will be reviewed, modified and finalised.
8. **Contribute to the nationally consistent consent project of the AHMAC PRG under the National Health Genomics Policy Framework**
   (i) Collaborate with the NSW led project team to develop nationally consistent guidance and template/s for clinical consent for genetic/genomic testing, for Australia-wide adoption.

The Consent Form and Supporting Information Material are provided in the appendices

Project overview

Figure 1. Process Summary



Project Outcomes

1. Review and evaluation of existing consent materials

- There is **no common national stance on management of issues** relating to genetic/genomic consent process:
    - Return of actionable and non-actionable disease-causing variants;
    - Incidental findings (which inform genetic risk for conditions not related to the purpose of the test: e.g. a BRCA1 mutation identified in a child being sequenced for epilepsy);
    - Analysis and gene lists;
    - Further use of samples & genomic data in research & whether re-contact is required / best-practice;
    - Storage and use of the samples by the diagnostic laboratory;
    - Data sharing;
    - Risks of genomic testing; and
    - Implications for relatives and insurance.
- This lack of national consistency may be a **barrier to the flow of health information** across states or jurisdictional borders as families are dispersed across the country.
- A **landscape analysis** and evaluation of genomic testing consent forms from different states permitted the group to 'cherry pick' the best elements of each:
    - SA Pathology (clinical)
    - Melbourne Genomics Health Alliance (clinical and research)
    - New South Wales Health (clinical)
    - The University of Western Australia (research)
    - PathWest (clinical)
    - Genomics England (research)
- The **barriers to genomic clinical consent** were discussed, and the **essential elements of a consent form** were determined.
- The **legal, ethical and social issues** relevant to the project were discussed. Balancing the need to convey concepts adequately to support informed consent, without overburdening the reader, must be a deliberate and carefully considered process. The principal of autonomy in the context of consent, and the information provided, was also explored.

2. Guidelines and Recommendations

1/ **Brevity -** The National Clinical Genomic Consent form must be in plain language, and brief. While accompanying information material can support the consent process, the consent form itself must be **no longer than one double-sided page**.

2/ **The patient voice** – The development of consent forms must engage the target audience: the patient. Australian Genomics consulted our Community Advisory Group, patient community and patient advocacy groups from around Australia in development of these materials.

3/ **Review** - Given the changing landscape around key issues associated with genomic testing, it is expected that these documents will need to be reviewed on an annual basis.

4/ **Specialised consent forms** – Australian Genomics will seek broad input on the demand for specialised consent forms (e.g. people with intellectual disabilities). We will develop these

variations on the national clinical genomic consent form with specialised input from the involved clinical specialists and patient groups.

> ***"It will be easier to get consensus about consent for return of incidental findings when we have achieved consensus on what the definition of clinically actionable findings should be for the Australian population."***

3. Drafting the National Clinical Genomic Consent Materials

The Clinical Genomic Consent form and Supporting Information Material were drafted based upon extensive review and discussion of example materials in use, and aligned with the agreed Guidelines and Recommendations for structure and content.

These materials underwent iterative development and refinement over an 11-month period amongst working group members.

4. Consultation Process

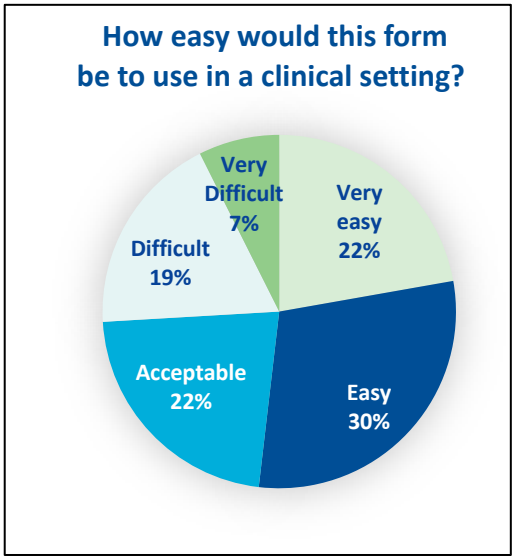
The working group agreed that the National Clinical Genomic Consent form agnostic of testing purpose (V15.1) was ready for broad consultation in May 2018. A consultation framework was developed to support the collection of standardised, comprehensive feedback (Appendix 3).

In the initial round of consultation, invitations were sent to 65 individuals and organisations (Appendix 4). The documents were also made available to the public on the Australian Genomics website. Responses were submitted by 42 individuals and organisations, including Committees of the Human Genetics Society of Australasia; Commonwealth and State Departments of Health; Clinical Geneticists; Genetic Counsellors; Research Institutions; Community Advisory Groups; Genomic Alliances; and Medical Specialist Colleges.

Over 80% of respondents found all elements in consent form and supporting documentation very good, good or acceptable.

Seventy-four per cent of respondents believed that it would be at least acceptably easy to implement the consent form in the clinic.

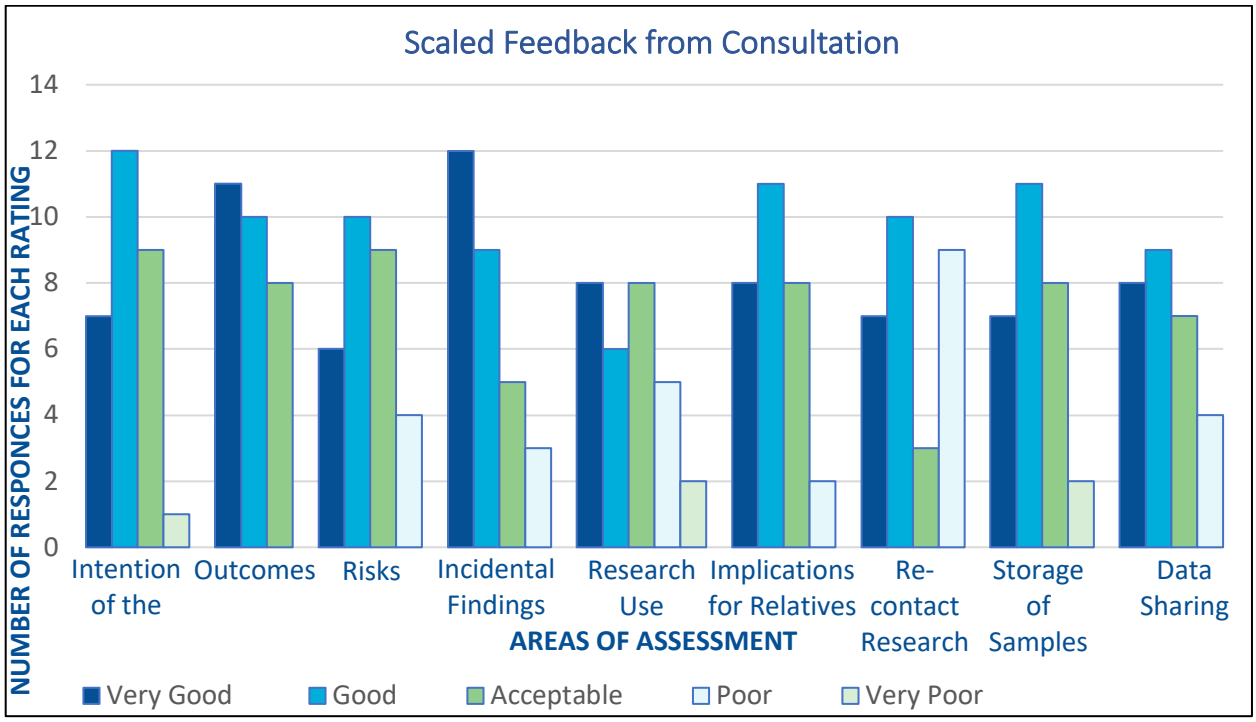
Inclusion of consent for research: The 're-contact for future studies' and 'use of sample for research', seemed to be a key point of differentiation in perceived acceptance of the form: more as a philosophical segregator of the community consulted, rather than the structure of these specific materials.



Some groups remain convinced that an option for research should <u>not</u> be incorporated into clinical genomic consent forms, despite strong support from the majority of the genomics community who provided feedback.

There were major recurring themes from the consultation feedback, which are addressed in the table below:

Major Recurring Themes	Specific Feedback	How they have been addressed
Need for parent/guardian (P/G) consent and broader consent for other testing processes	• P/G consent form needed or change wording of current form • Predictive testing, segregation, carrier testing etc. not covered	• P/G consent form will be developed as next phase of work • This form is for genomic testing specifically. Other types of testing will use genetic testing consents.
My Health Record (MyHR)	• MyHR is not mentioned on the consent form	• Advice was sought from the Digital Health Agency on management of MyHR information on a national document, given differences in state policy around genomic reports in MyHR. • Check box inserted, per their recommendations
Risks	• Risks are not well defined • Some risks are understated where as others are overstated	• Wording to be changed to better define risks of genomic testing
Anonymous versus coding data sharing for clinical and research purposes	• Confusion between what data will be shared anonymously versus coded • Confusion regarding what data will be shared for research • Concern with the terminology	• Data sharing and research section has been re-worded for clarity • Descriptions for anonymous and coded data simplified, and accompanied with a figure
Use of results for the healthcare of family members	• Optional sharing of results for the healthcare of family members should be removed	• Familial sharing option will remain on the consent form, with the understanding that majority of individuals will select 'Yes'. However in rare cases 'No' must be an option, given jurisdictional differences in management.



"…consider how clinicians undertake a process of consent; and to use the form as a way to summarise that process."

5. Incorporation of consultation outcomes, and secondary targeted consultation.

After aggregation and analysis of the consultation data, the working group convened a face-to-face meeting to discuss and resolve conflicts or discrepancies in the responses, and agreed to content for amendment and inclusion.

A secondary consultation was undertaken with multiple Patient Advocacy Groups and the Regulatory and Ethics Workstream of the Global Alliance for Genomics and Health (GA4GH) (Appendix 3).

The responses of this consultation informed further refinement of the materials, and they were approved for release and evaluation in clinical practice.

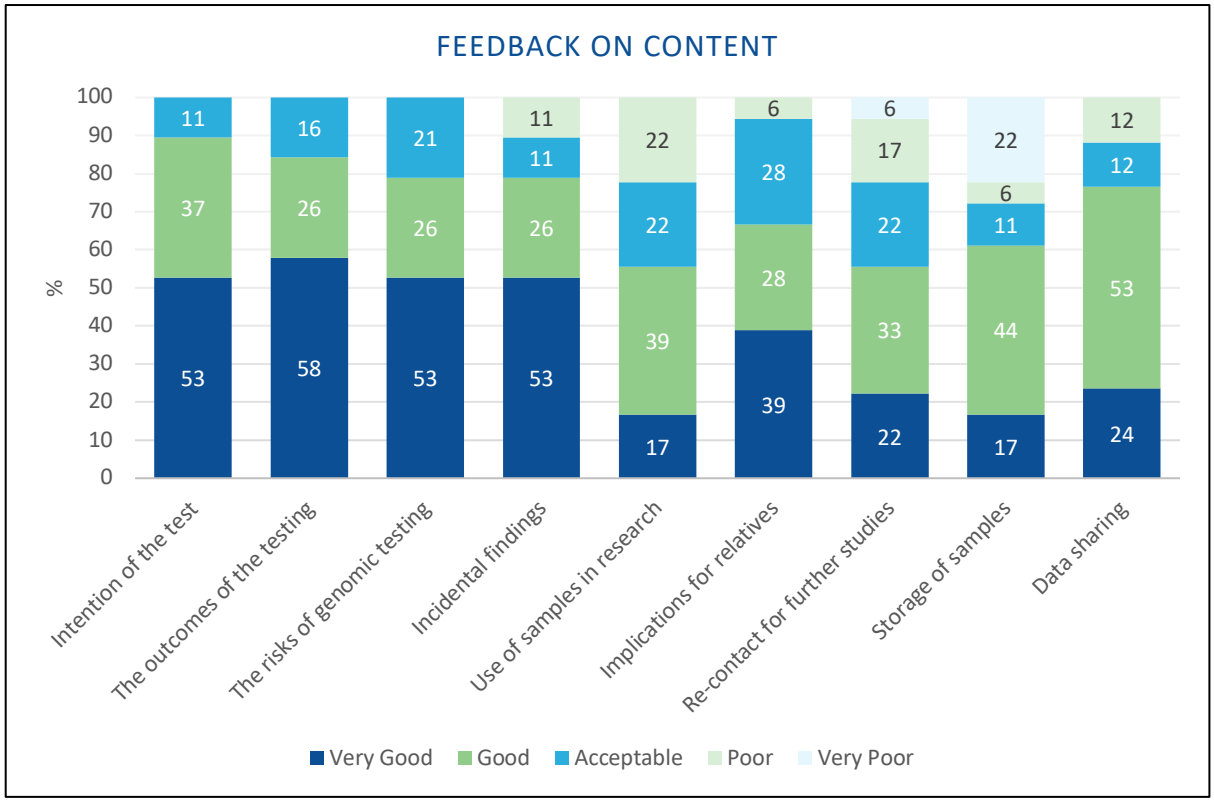
The resulting National Clinical Genomic Consent Form and Supporting Information Material are provided in the appendices.

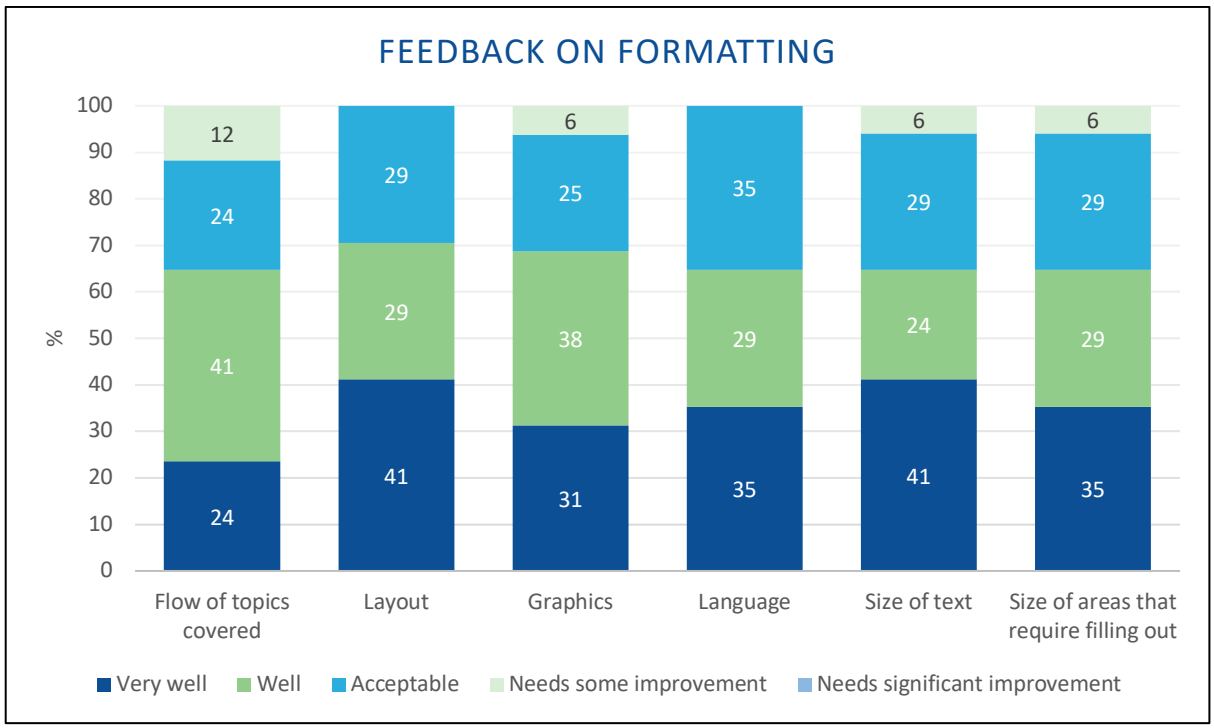
6. Trial use and evaluation

The developed Clinical Genomic Consent Materials (V15.3 and V11.2) underwent a six-month pilot in Victoria (Victorian Clinical Genetics Services) and Queensland (Genetic Health Queensland) January – June 2019, where the forms were used in place of the services' own genomics consent forms. The development of a paediatric form was also carried out, to allow for trialling of the package's in a larger cohort. Structured evaluations which included surveys of clinicians using the package and a focus group with lead site clinicians were conducted partway and at the conclusion of the pilot. Data was gathered on the general ease of use of the materials, the adequacy and completeness of the information

included, the simplicity and clarity of the language, and the degree to which the materials support the consent process for health professionals in the genomics community.

Feedback (N=19) from the two surveys (conducted partway through and at the conclusion of the pilot) are summarised in the graph below:





Comments from surveys and the focus group included:

- Removal of statements that were not currently possible by either the clinical or laboratory services:
    - Optional release of results to My Health Record
    - Destruction of samples after required timeframe
- Clarification of language in research statements
- Removal of optional tick box for sharing of results for family members' healthcare
- Clarification of statement regarding withdrawal from testing
- Structural changes to the supporting document

Patient opinion on the materials was sought through the Australian Genomics Community Advisory Group.

Feedback received led to further refinement and improvement of the materials, which were finalised and published 18 October 2019.

We are contributing to the PRG "Nationally Consistent Templates and Guidance for Consent to Genetic and Genomic Testing" project, led by the NSW Ministry of Health, to harmonise terminology, content and approach where possible between these projects. , We are also working with colleagues internationally who plan to adapt and adopt the materials for use.

Australian Genomics envisages that the National Clinical Genomic Consent Form and Supplementary material will be living documents, and we would recommend a process of structured annual review.

7. Conclusions

The development of a National Clinical Consent form is challenging. There is jurisdictional variation in policies and procedures in clinical care; there are limitations in the resourcing, flexibility and infrastructure in some genetic laboratories and services; and there is inconsistency in use of terminology across Australian genomic practice.

The development and consultation process of these clinical genomic consent materials was a resource-intensive, lengthy process, but an extremely valuable one.

There is a significant amount of information to digest when consenting to a genomic test: consent should be considered as a process, and the consent form and supporting documentation aims to better support this process.

Development of the National Clinical Genomic Consent Materials has identified several key areas that will need to be targeted in support of mainstreaming genomics in a standardised fashion across the Australian healthcare system:

- Seek consensus on what the definition of clinically actionable findings should be for the Australian population;
- Develop common policies around management of incidental / secondary findings;
- Harmonise approaches to release of results to family members, for the benefit their healthcare;
- Consultation on the relative benefits and risks of including consent for research as part of a clinical consent process – and a calculation as to the impact this has on patients and families tested;

- Seeking jurisdictional input and agreement on approaches to identified challenges and differences would make significant gains toward equitable application of genomic testing across Australia.
- Consider consent requirements and preferences of Aboriginal and Torres Strait Islanders, minority groups and culturally and linguistically diverse communities in the ongoing refinement of clinical consent materials, by involvement and collaboration with these groups.

Australian Genomics looks forward to addressing these issues in partnership with State and Commonwealth Departments of Health, particularly the Project Reference Group on Health Genomics; our colleagues at the Human Genetics Society of Australasia and the Royal College of Pathologists Australasia; and patient communities.

Australian Genomics gratefully acknowledges the considerable in-kind investment in time and effort of the expert members of the National Clinical Genomic Consent working group.

APPENDIX 1 – NATIONAL CLINICAL GENOMIC CONSENT FORM

Affix identifier information here

Clinical Consent Form for Genomic Testing

It is my choice to have genomic testing.

I, _____ (patient name), understand that my DNA will be tested by panel/exome/genome to look for changes in genes that may be associated with: _____ (condition or clinical indication)

About the Test

- Genomic test results are based on current knowledge, which may change in the future.
- If I change my mind, I can choose not to be told about the result.

Potential Outcomes

- This test might find a cause for the condition(s).
- This test might not find a cause for the condition(s).
- The result might be of *'unknown significance*', which means it cannot be understood today.
- There is a chance that genomic testing could find other medical conditions (incidental findings).
- Genomic testing may show unexpected family relationships.
- Further tests or information sharing may be needed to finalise the result.

Results

- I will be told the results by a health professional.
- Results may have implications for the health/genetic risks of my family members.
- Results can be used to inform counselling and testing of family members, though my identity will not be revealed to them.
- Results from these tests may affect my ability to obtain some types of insurance.
- The results will be available to health professionals involved in my care.
- Results are confidential and may not be released without my consent, unless allowed by law.
- The following individual can be given my results, if I am unable to be contacted:

  Name _____ Contact_____

Data and Sample Sharing

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.

Research

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**

I have had enough time to consider the information in this consent form and have:

- Had the opportunity to discuss genomic testing and its implications with a health professional
- Been given access to information about genomic testing.
- Been able to ask questions until I am satisfied with the answers.
- Been offered a copy of this consent form.

I provide consent to have genomic testing as summarised in this form.

Signature_____Date _____

Print Name_____

Date of Birth_____

Email/ Address_____

Health Professional Signature _____Date _____

Health Professional Print Name_____

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APPENDIX 2 – Genomic Testing Fact Sheet

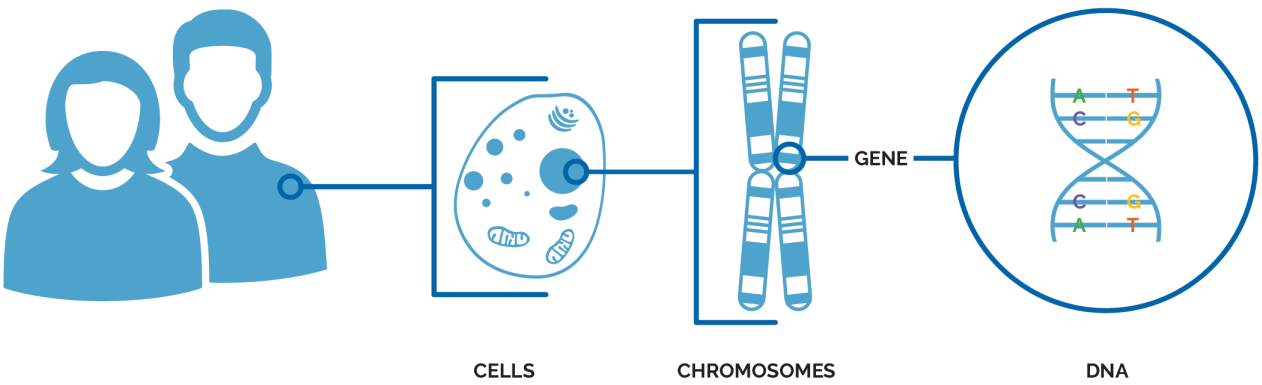
This fact sheet aims to help you discuss the **consent form for genomic testing** with your health professional. The consent form needs to be signed before genomic testing is started. *It is your choice whether to have this test*.

Questions to ask your doctor/genetic counsellor

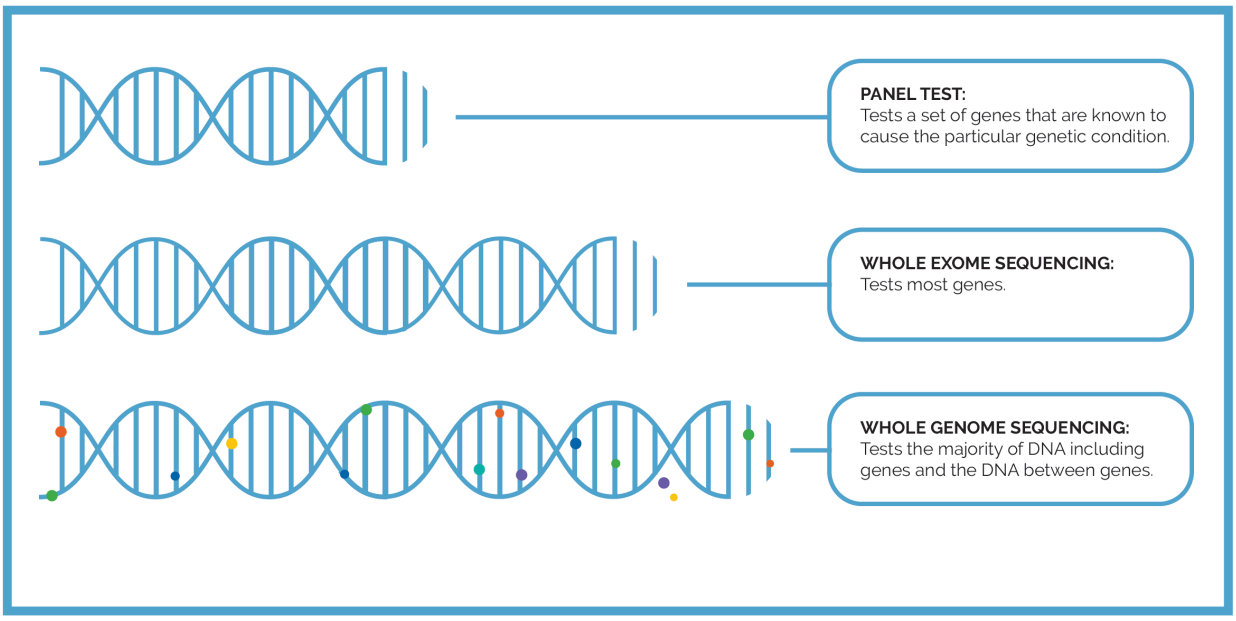
- What is the chance that the genomic test will identify the cause of my/my child's condition?
- How long will it take to get a result?
- Who will give me the result and how?
- Where will my genomic test be done?
- What is the cost to me (if any) of my genomic test?
- *What can this mean for other members of my family if I have this test?*
- What is the chance of this test finding something that is unrelated to my/my child's current health condition?

What is genomic testing?
Our bodies are made up of billions of cells. In most of our cells we have a complete copy of our genetic information (genome). We all have about 20,000 genes in our genomes. Our genes are made of DNA and contain the instructions for growth and development of the body. Until recently, doctors and scientists were only able to test one gene at a time. Genomic technology allows us to test many of our genes at once (genomic testing).

DNA for testing comes from blood, tissue, saliva, or mouth swab.



DNA can be taken from your body cells to identify changes that contribute to or cause disease.



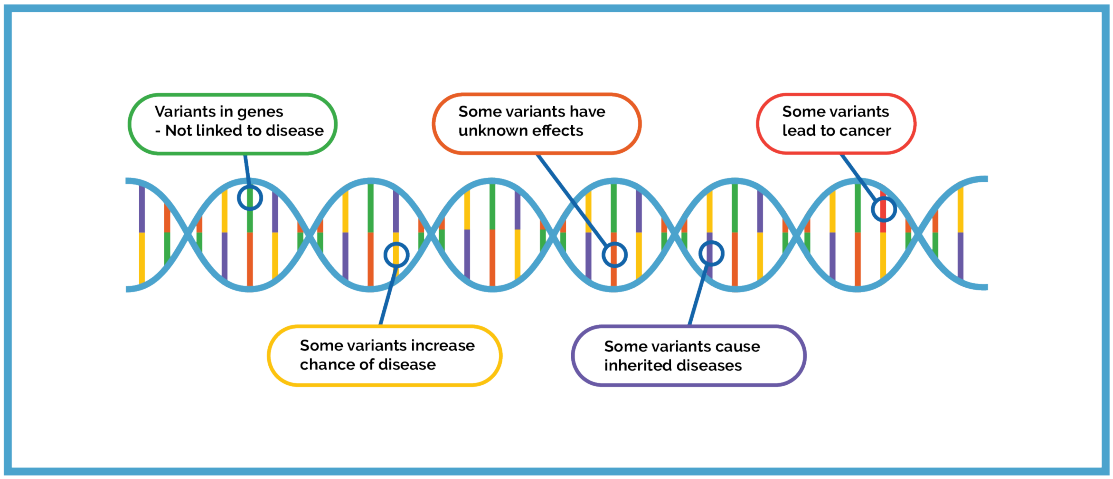
What are genetic variants?
Each person's genome contains many genetic differences (variants). Most of these are harmless and do not change how the gene works in the body. Genomic testing is done to find variants that *do* change how a gene works and therefore cause genetic conditions.

Example:

Think of a gene as like a sentence in an instruction manual.

Consider the sentence:

MUM CUT THE HOT DOG	The gene works as it should.
MOM CUT THE HOT DOG	Some spelling changes look different but *don't* change the meaning of the sentence. Therefore, the gene still works.
MXM CUT THE HOT DOG	Other spelling changes look different and *do* change the meaning. Therefore, the gene doesn't work.
MEM CUT THE HOT DOG	Other spelling changes look different but we do not know if the meaning changes. Therefore, we are uncertain what the effect on the gene might be.



Different types of DNA variants have different effects on the genetic code and body functions.

Potential <u>outcomes</u> of genomic testing

The test may:

- find a cause of the condition.
- not find a cause that could explain the condition.
- find a result of '*variant of unknown significance'* (VUS), which means that it cannot be understood today. Sometimes testing in other family members for the VUS may help to understand if it could be the cause of a condition. The understanding of VUS may change over time. Future testing may help clarify this.

A cause for the condition may not be found for a number of reasons, including:

- the variant causing the condition cannot be found by the test;
- the gene causing the condition was not tested;
- the gene causing the condition is not yet known.

Why genomic testing?

Some reasons people have genomic testing are:

- to find a genetic diagnosis for their or their child's condition.
- to help families understand the condition, access support they need, and plan for the future.
- to help health professionals manage a condition.
- to provide families with information about the chance of having another child with the same condition.

Sometimes, the genomic test result in one person may also be important for the care of their relatives.

If a diagnosis does not happen today, the genomic test result could be looked at in the future as our understanding improves.

It is important to remember that genomic testing is not a general health test and will not identify *all* gene changes that could contribute to health problems that may develop in the future.

Other things to think about

Incidental findings
As this test looks at many genes at once, there is a small chance a variant may be found in a gene that is not related to your health condition. Such '*incidental findings'* could be important to know about for your health. If your doctor thinks that an incidental finding may be important for you or other family members, they will raise it with you.

Insurance
In Australia, genomic testing will not alter your ability to get health insurance or your health insurance premiums. Genomic testing in you or your child could affect how easy it is for you or other family members to get income protection, travel or life insurance; or the price of your premium. From July 2019 to June 2024 the life insurance industry has put in place a moratorium to allow people to access a level of life insurance without being asked about the result of a previously taken genetic test. An existing diagnosis may already affect your ability to obtain these kinds of insurance. Industry regulation prevents insurers from asking relatives for your genetic test results, and you cannot be requested to have a test by an insurer. Your healthcare provider will not provide your results to an insurance provider without your permission.

Further details can be found at: http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/FactSheet23A and by searching for 'Moratorium' at https://www.fsc.org.au/resources

Withdrawal from testing
You can change your mind about having genomic testing or being told the results. You can cancel the test at any time before the laboratory finishes the test. You can also choose not to be told the result after the test is finished, but the test result will be placed in your medical record.

Sharing results to help family members
Genetic services will not usually contact your family members. But, your relatives may be referred for genetic testing, when they find out there is a genetic condition in the family. Your test results may be released to another genetics service to help with the care of other family members, because genetic changes run in families. Genetic services need to share information, so that the correct testing can be offered to others. All efforts will be made to ensure that your identity is not revealed to those family members.

Data and Sample Sharing
Your results and genomic data will be stored securely in databases that meet Australian/international security standards and laboratory guidelines.

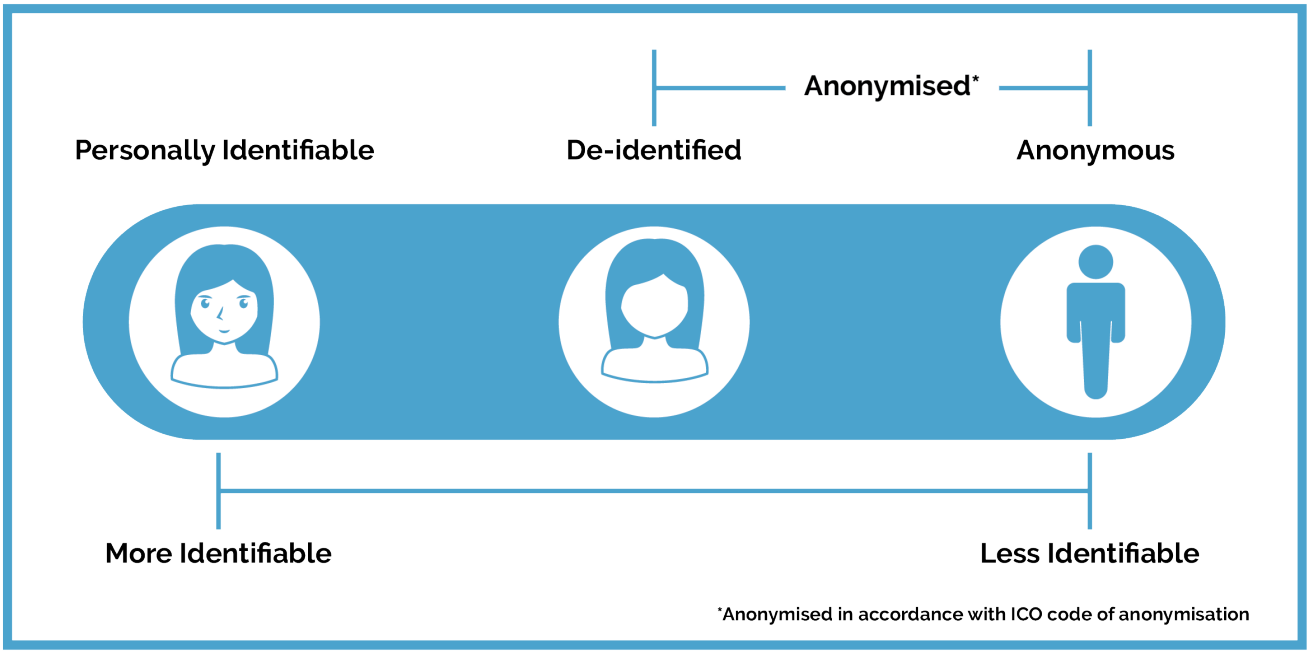
The health professionals involved in your care may order further testing of your sample or share your genomic data with each other to help work out what your test results mean. However, your results, genomic data and identified sample, or the fact that you have had genomic testing, will not be used or disclosed outside of your care without your consent, unless required or allowed by law.

Sharing of your genomic data and health information can advance scientific knowledge. This includes sharing gene variant information with large databases to help improve our understanding e.g. by comparing your results to those of other people.

When your data is shared there are safeguards in place to help protect your privacy, such as:

- Personal identifiers (information) will be removed (such as your name and address)
- Security measures that help prevent unauthorised access or misuse.

However, there is always a very small chance that you might be re-identified. Given that the potential to identify you is significantly reduced, you are unlikely to directly benefit from this sharing.



So why share my data?
You can also provide consent to sharing data for research into the **same or a related condition** in a way that may be linked back to you. The personal identifiers can be removed from your information or sample, and replaced by a code. If there are findings from this research that have implications for your clinical care, it may be possible to re-identify you, so that your results can be returned. However, participating in research does not guarantee direct benefits to you.

All researchers are bound by the law and ethical guidelines. This research will only happen for projects approved by a human research ethics committee. You can choose not to consent for research.

Questions?
This information sheet provides general information about genomic testing. It supports the information you should receive from a medical specialist or genetic counsellor. If you have any questions about the test or any of the information in this sheet, please contact your doctor or genetic counsellor.

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APPENDIX 3

National Clinical Consent Form for Genomic Testing and Supporting Information

Consultation document

1. **How would rate the consent form and the supporting documentation regarding:**
   (please mark with an x)

	Very Good	Good	Acceptable	Poor	Very Poor
Intention of the test					
The outcomes of the testing					
The risks of genomic testing					
Incidental findings					
Use of samples in research					
Implications for relatives					
Re-contact for further studies					
Storage of samples					
Data sharing					

2. **What do you like about this form and supporting documentation?**

3. **What would you change about this form and supporting documentation?**

4. **How easy would this form be to use in a clinical setting?**
   (please mark with an x)

Very Easy	Easy	Acceptable	Difficult	Very Difficult

5. **What can we do to help ensure uptake in your state or institution?**

6. **Do you have any further comments on this form and supporting documentation?**

OPTIONAL FURTHER QUESTIONS:

7. **The intention of the test:**

   a. Do you agree with the content regarding intention? ☐ Yes ☐ No

   b. If you selected 'No' or 'Undecided', what were your reasons why?

   c. Do you have any suggestions for how this section in either the consent form or the supporting documentation, could be better expressed?

   d. Is there any further information regarding the intention of the test that you think should be included in the consent form or supporting documentation? Please describe these below.

8. **The outcomes of the testing:**

   a. Do you agree with the content regarding outcomes of the testing? ☐ Yes ☐ No

   b. If you selected 'No' or 'Undecided', what were your reasons why?

c. Do you have any suggestions for how this section in either the consent form or the supporting documentation, could be better expressed?

d. Is there any further information regarding the outcomes of the testing that you think should be included in the consent form or supporting documentation? Please describe these below.

9. Risks of genomic testing:

a. Do you agree with the content regarding risks of genomic testing? ☐ Yes ☐ No

b. If you selected 'No' or 'Undecided', what were your reasons why?

c. Do you have any suggestions for how this section in either the consent form or the supporting documentation, could be better expressed?

d. Is there any further information regarding the risks of genomic testing that you think should be included in the consent form or supporting documentation? Please describe these below.

10. Incidental findings:

a. Do you agree with the content regarding incidental findings? ☐ Yes ☐ No

b. If you selected 'No' or 'Undecided', what were your reasons why?

c. Do you have any suggestions for how this section in either the consent form or the supporting documentation, could be better expressed?

d. Is there any further information regarding incidental findings that you think should be included in the consent form or supporting documentation? Please describe these below.

11. Use of samples in research:

a. Do you agree with the content regarding use of samples in research? ☐ Yes ☐ No

b. If you selected 'No' or 'Undecided', what were your reasons why?

c. Do you have any suggestions for how this section in either the consent form or the supporting documentation, could be better expressed?

d. Is there any further information regarding the use of samples in research that you think should be included in the consent form or supporting documentation? Please describe these below.

12. Implications for relatives:

a. Do you agree with the content regarding implications for relatives? ☐ Yes ☐ No

b. If you selected 'No' or 'Undecided', what were your reasons why?

c. Do you have any suggestions for how this section in either the consent form or the supporting documentation, could be better expressed?

d. Is there any further information regarding the implications for relatives that you think should be included in the consent form or supporting documentation? Please describe these below.

13. Re-contact for further studies:

a. Do you agree with the content regarding re-contact for further studies? ☐ Yes ☐ No

b. If you selected 'No' or 'Undecided', what were your reasons why?

c. Do you have any suggestions for how this section in either the consent form or the supporting documentation, could be better expressed?

d. Is there any further information regarding re-contact for further studies that you think should be included in the consent form or supporting documentation? Please describe these below.

14. Storage of samples:

a. Do you agree with the content regarding the storage of samples? ☐ Yes ☐ No

b. If you selected 'No' or 'Undecided', what were your reasons why?

c. Do you have any suggestions for how this section in either the consent form or the supporting documentation, could be better expressed?

d. Is there any further information regarding the storage of samples that you think should be included in the consent form or supporting documentation? Please describe these below.

15. Data sharing:

a. Do you agree with the content regarding data sharing? ☐ Yes ☐ No

b. If you selected 'No' or 'Undecided', what were your reasons why?

c. Do you have any suggestions for how section in either the consent form or the supporting documentation, could be better expressed?

d. Is there any further information regarding data sharing that you think should be included in the consent form or supporting documentation? Please describe these below.

APPENDIX 4

LIST OF INDIVIDUALS / ORGANISATIONS APPROACHED IN THE CONSULTATION PROCESS

	Broad Consultation - May 2018	
	Name	Organisation
1	Vanessa Clements	Department of Health (NSW)
2	Eva Pilowsky	Department of Health (NSW)
3	Paul Fennessy	Department of Health and Human Services (Vic)
4	Shane Porter/ Moira Campbell	Department of Health
5	Edwina Middleton	Department of Health (NSW)
6	Sally Howard	Department of Health (NSW)
7	Keith McNeil	Queensland Health
8	Hugh Dawkins	Department of Health (WA)
9	Jan Williamson	Department of Health and Human Services (TAS)
10	Dr Bruce Latham - President	The Royal College of Pathologists
11	Dr Michael Gannon	Australian Medical Association
12	Dr Zena Burgess	The Royal Australian College of General Practitioners
13	Mr John Batten	ROYAL AUSTRALASIAN COLLEGE OF SURGEONS
14	Professor Steve Robson	Royal Australian and New Zealand College of Obstetricians and Gynaecologists
15	Dr Catherine Yelland	The Royal Australasian College of Physicians
16	Dr Michael Buckley	Human Genetics Society of Australasia
17	Kevin Forsyth	Academy of Child and Adolescent Health
18	Ivan Macciocca	Australasian Society of Genetic Counsellors
19	A/Prof Michael Gabbett	Australasian Association of Clinical Geneticists
20	Kate Dunlop	Centre of Genetic Education
21	Australian Genomics Community Advisory Group	Australian Genomics
22	Melbourne Genomics Community Advisory Group	Melbourne Genomics
23	Queensland Genomics Community Advisory Group	Queensland Genomics
24	Monica Ferrie	Genetic Support Network of Victoria
25	Amanda Samanek	Genetic and Rare Disease Network (WA)
26	Steffani-Jade McDonagh	Genetic Alliance Australia (NSW)
27	Nicole Millis	Rare voices Australia
28	Julia Overton, Chief Executive	Health consumer Alliance (SA)
29	Norah Grewal	Research Associate in Law and Ethics
30	Emma Tudini	QIMR Berghofer
31	Amanda (Mandy) Spurdle	QIMR Berghofer
32	Asra Gholami	Executive Officer, SCHN Research Ethics Office
33	Kaye Hewson	Genetic Testing Working Group, GHQ Service Plan Implementation
34	Jodi Johnson-Glading	Department of Health and Human Services (TAS)
35	Mathew Wallis	Austin Hospital, Clinical Genetics Service
36	Matthew Hunter	Monash Medical Centre Genetics Clinic
37	Ingrid Winship	Royal Melbourne Hospital Genetic Medicine
38	Martin Delatycki	Victorian Clinical Genetics Service (VCGS)
39	Melissa Gratz	Mercy Women Genetics service
40	Susan Fawcett	Royal Women's Hospital
41	Paul James	Peter Mac familial cancer
42	Monash Health Familial Cancer Centre	Monash Health Familial Cancer Centre
43	Geoff Lindeman	The Royal Melbourne Hospital Familial Cancer Centre

	Name	Organisation
44	Trinity Mahede	Department of Health (WA), Office of Population Health Genomics
45	Alicia Bauskis	Department of Health (WA), Office of Population Health Genomics
46	Dr Nicholas Pachter	Genetic Services WA
47	Dr Mimi Berman	Royal North Shore Hospital: General genetics
48	Dr Mike Field	Royal North Shore Hospital: Cancer genetics and GOLD
49	Dr Felicity Collins	Royal Prince Alfred: General Genetics
50	Dr Michel Tchan	Westmead Adult Hospital: General Genetics
51	Dr Judy Kirk	Westmead Adult Hospital: Cancer Genetics
52	Dr Linda Goodwin	Nepean Hospital
53	Dr Alison Colley	Liverpool Hospital: General Genetics
54	Dr Annabelle Goodwin	Liverpool Hospital: Cancer Genetics / Royal Prince Alfred: Cancer Genetics
55	Dr David Mowat	Sydney Children's Hospital Randwick
56	Dr Kathy Tucker	Prince of Wales Cancer Service
57	Dr Kathy Wu	St Vincent's Genomic Centre
58	Bronwyn Burgess	Hunter Genetics
59	Julie White	Genetic Helath QLD
60	Karin Kassahn	SAPathology
61	Chris Barnett	SAPathology
62	Eric Haan	SAPathology
63	Nicola Poplawski	SAPathology
64	Skye Jakobi	Department of Health (SA)
	Targeted Consultation - October 2018	
65	Adrian Thorogood	Global Alliance regulatory ethics working group (REWG)
66	Tara Morrison	HCU Network Australia
67	Megan Donnell	Sanfilippo Children's foundation
68	Bronwyn Byrne	Leukodystrophy Australia
69	Adam Walczak	CanTeen
70	Deidre Gorrie	Cystic Fibrosis Victoria
71	Glenda Colburn	Lung Foundation Australia
72	Jan Mumford	GAA
73	Kate Vines	Rare Cancers Australia
74	Paul Bennett	CanTeen
75	Rebecca Davis	AMDF
76	Amanda Samanek	GARDN
77	Christine Walker	CIA
78	Kathy Wells	BCNA
79	Monica Ferrie	GSNV
80	Nicole Millis	RVA
81	Shannyn Floyd	KHA
82	Steve Roach	Myeloma
83	Sharon Gavioli	lungfoundation
84	Snezana Djordjevic	lukemia Foundation
85	Cindy Van Rooy	CFV

References

D Avard, & E Harmsen. *Informed Consent and Multiplex Genetic Screening.* In: Encyclopedia of Life Sciences (ELS) (2010). DOI: 10.1002/9780470015902.a0005642.pub2

A Cohen, A Hoon, AM Wilms Floet. *Providing family guidance in rapidly shifting sand: informed consent for genetic testing.* Developmental Medicine & Child Neurology, 55: 766–769 (2013). DOI: 10.1111/dmcn.12102

R Sharp. *Downsizing genomic medicine: Approaching the ethical complexity of whole-genome sequencing by starting small.* Genetics IN Medicine Vol 13, Number 3 (2011). DOI: 10.1097/GIM.0b013e31820f603f

S Elias & G Annas. *Sounding Board: Generic Consent for Genetic Screening.* NEJM 330:22, 1611-1612 (1994).

F Facio, J Sapp, A Linn, L Biesecker. *Approaches to informed consent for hypothesis-testing and hypothesis-generating clinical genomics research.* BMC Medical Genomics, 5:45 (2012) http://www.biomedcentral.com/1755-8794/5/45

B Koenig. *Have we asked too much of consent?* Hastings Center Report 44, no. 4 (2014): 33-34. DOI: 10.1002/hast.329

W Foulkes, BM Knoppers, C Turnbull. *Population genetic testing for cancer susceptibility: founder mutations to genomes*. Nat Reviews Clin Oncol 13: 41-54 (2016) doi:10.1038/nrclinonc.2015.173

K Burke, A Clarke. *The challenge of consent in clinical genome-wide testing*. Arch Dis Child (2016) doi:10.1136/archdischild-2013-304109

C Benjamin, M Boudioni, H Ward et al. NHS Genomic Medicine Centres National Service Evaluation of the Consent Process and Participant Materials used in the 100,000 Genomes Project. Research Gate (2016) https://www.researchgate.net/publication/311440245

National Pathology Accreditation Advisory Council. Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies (2017)

Consent and information materials:
University of Western Australia. DNA (Genetic) Testing & Storing (2017)

GA4GH Consent Policy & Consent Tools (2015)

NSW Health Pathology, Genetic Testing Including DNA Diagnostic Testing, DNA testing for mutation carriers & DNA predictive and pre-symptomatic testing (2015)

SA Pathology – Informed Consent (2017)

Melbourne Genomics PICF V8 (2016) & Exome Information Sheet V18 (2015)

100,000 Genomes Project Participant Consent Form for adults with a rare genetic condition, and their adult family members. V2.3 (2017) REC Reference Number 14/EE/1112.