Clinical Consent for Genomic Testing

Project Lead:
Prof Julie McGaughran Director, Genetic Health Queensland

Project Coordinators:
Keri Finlay Project Coordinator, Australian Genomics
Isabella Sherburn Project Officer, Australian Genomics

Working Group Members Tissue-Targeted Genomic Testing Consent:
Dr Dimitar Azmanov Head of Department, PathWest
A/Prof Mandy Ballinger Head of Cohorts, Australian Genomic Cancer Medicine Centre (Omico)
Dr Anna Brown Head of Molecular Oncology, SA Pathology
Nicole Den Elzen Genetic Counsellor, Peter MacCallum Cancer Centre
Dr Helen MarFan Medical oncologist and cancer geneticist, Genetic Health Queensland
Dr Adayapalam Nandini Principal Chief Scientist, Queensland Health
Dr Niles Nelson Haematologist, The Royal Hobart Hospital
Dr Sinead O'Sullivan Clinical Geneticist, Genetic Health Queensland
Grace Pendlebury Shariant Project Officer, Australian Genomics
Dr Chris Richmond Clinical Geneticist, Genetic Health Queensland
A/Prof Alison Trainor Clinical Geneticist, Peter MacCallum Cancer Centre
Mary-Anne Young Head of Clinical Engagement and Translational Platform, Garvan Institute of Medical Research

Working Group Members Online Consent Tool:
John Cannings Patient Advocate, Australian Genomics Community Advisory Group
Liana Cross Digital Communications Manager, Australian Genomics
Ivan Macciocca Principal Genetic Counsellor, Victorian Clinical Genetic Service
Dr Nicola Poplawski Clinical Geneticist & Head of Unit, Royal Adelaide Hospital
A/Prof Danya Vears Principal Research Fellow, Murdoch Children's Research Institute

1.1 Project Description

Tissue-Targeted (Somatic) Genomic Testing Consent Package:

Currently there are no clear consent processes or supporting materials available for tissue-targeted (somatic) genomic testing. It was brought to the attention of Australian Genomics that with an increase in germline variants being detected through tissue-targeted (somatic) testing, clinicians felt they needed to ensure that patients are properly informed about the implications of testing. There has been no national initiative with the remit to develop consent materials for tissue-targeted (somatic) testing.

Australian Genomics convened an expert working group to develop a clinical tissue-targeted (somatic) genomic testing consent package, which meets current national standards and current clinical and laboratory practices. The consent package comprises a clinical consent form for tissue-targeted (somatic) genomic testing, a patient fact sheet and a health professional guide. This

package underwent a public consultation to ensure it met the needs of both health professionals and patients. The package was released in June 2025.

Online Consent Tool:

Through the development of the clinical genomic testing consent package, it was recognised that the delivery of patient information in a fact sheet format is not suitable for all audiences. Efforts were made to improve the patient experience through the development of an online tool which supports patients through the consent process. Development began in November 2023 with an expert working group formed to drive the sub-project. The online tool was launched in December 2024.

1.2 Progress Report

Tissue-Targeted (Somatic) Genomic Testing Consent Package:

Project Aims

1. Review existing consent materials
2. Draft consent materials based on current national standards and practices
3. Undertake a targeted consultation on the consent package and amend materials based on feedback
4. Provide materials for clinical use

Project outcomes

1. *Review of existing consent materials*

A search for existing clinical tissue-targeted and somatic consent materials found there were currently none in use. The absence of consent material has led to several challenges, such as patients being unaware of possible familial implications of tissue-targeted testing, inconsistencies in the patient experience and a lack of guidance for health professionals who are consenting patients for this test.

The project reviewed broader consent materials that were available, and a decision was made to draw from materials used for research purposes and germline genomic testing.

2. *Drafting the consent materials based on current national standards and practices*

The expert working group drafted a consent package, which comprises:

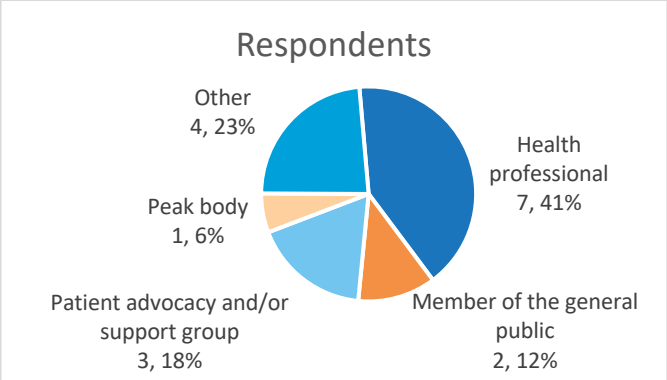
- Clinical consent form for tissue-targeted (somatic) genomic testing
- Patient fact sheet (PFS)
- Health professional guide (HPG)

This consent package is largely based on the *Australian Genomics Clinical Consent Package for Genetic and Genomics Testing* (2024). Decisions to omit, refine or add clauses were made following thorough discussions of current clinical and laboratory processes, ensuring alignment with national standards.

These materials were developed and refined iteratively over a six-month period amongst working group members. An early review of the consent form was carried out by the Australian Alliance for Indigenous Genomics and the form was revised based on feedback.

3. *Targeted consultation process on the consent package and amend materials based on feedback*

A single public consultation was undertaken. Draft materials were made available on the Australian Genomics website in April 2025 and were promoted via Australian Genomics social media platforms. A targeted list of experts including patient support and advocacy groups and peak bodies (oncology and genetics/genomics focused) were also specifically invited to participate in the consultation.



In total there were 17 respondents to the consultation, 16 via the consultation survey and one via email. Of the respondents, 41% were health professionals and 30% were patient groups or members of the public. The "other" category included a collaborative network, researcher, project coordinator and ethicist.

In the consultation survey respondents were asked to rate the appropriateness of each clause. Most clauses rated quite highly with over 80% of respondents indicating that they found five of the eight clauses appropriate.

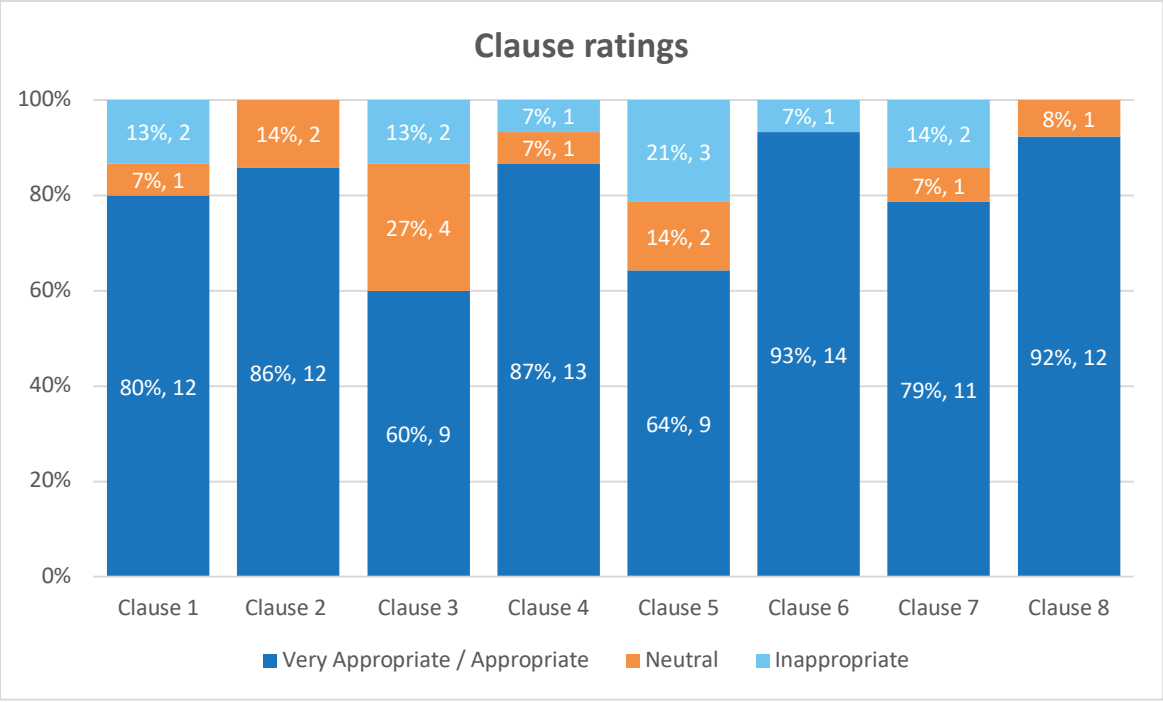


Figure 1: Clause ratings from consultation feedback

Clause 3 *"Although not intended, this test may find a result that is unrelated to the current condition and/or could have implications for blood relatives"* and clause 5 *"Results and related health information may be shared with genomic and medical databases that are used for patient care. All identifying information will be removed"* rated the poorest. Overarching feedback and key concerns with each clause are detailed in Tables 1 and 2. Within the survey, respondents were able to provide suggested re wording for the clauses and the content in the HPG and PFS.

Table 1: General feedback

Feedback	Response
Not all patients will have an alternate contact	The addition of an alternate contact has been made optional.
The flow chart is confusing in its current position	The flow chart has been moved to lower in the Health Professional Guide.
Addition of Aboriginal and Torres Strait Islander genomic resources and the development of specific resources to support Aboriginal and Torres Strait Islanders	Links to resources to better support health professionals when working with Aboriginal and Torres Strait Islander people have been included in the health professional guide.
Level of language is too complex for the general public	The language in the PFS has been simplified.
There is a lack of consistency between the consent form and patient fact sheet	Clauses have now been referenced within the PFS to help guide patients.
Addition of support services available and referring genetic services in the PFS	Given the range of conditions that undergo somatic testing listing all support services is not possible. Also, as the focus of this consent form is somatic testing, referral to genetic services is not appropriate at this point. If a germline genetic test is required further information will be given at that stage (through the genomic testing package).
Inclusion of further information on results process, implications for patients and family members, potential benefits and limitations, and what happens if no result is identified	Further information has been included, however more specific information is challenging given the diversity of conditions that use somatic testing.

Table 2: Key concerns for each clause

Clause	Feedback	Response
This test aims to look for genetic changes in the tissue sample that may be related to the condition.	Too much of a focus on diagnosis, the main aim of somatic testing is treatment selection	This has been edited in the PFS to reflect other purposes of testing
The test does not detect all genetic changes or all genetic conditions.	Too much of a focus on diagnosis, the main aim of somatic testing is treatment selection	This has been edited in the PFS to reflect other purposes of testing
	Highlight the limitations of current knowledge and the evolution of genomic knowledge over time, in the HPG and PFS	Further information has been provided within the HPG
	Some of the explanatory material in the 'Aboriginal and Torres Strait Islander Peoples' clause under 'Other Points for Consideration' would be better placed in the main 'explanatory' section	Information has been integrated into the main table within the HPG
Although not intended, this test may find a result that is unrelated to the current condition and/or could have implications for blood relatives.	No information provided on paired testing	Paired testing was discussed within the working group, and it was felt that paired testing is not yet integrated into the clinical setting in a consistent manner, so was not included in the current version of the consent form. This was noted as a possible addition in the next iteration of the tissue-targeted (somatic) testing consent form.

	Include incidental findings information in the PFS	Information on incidental findings has been strengthened in the PFS
	Clarification of the difference between germline and somatic variants in the PFS	This has now been included in the PFS
To better understand the test results, more testing, another tissue sample or re-examination may be needed.	Include information on re-testing in the PFS	This has now been included in the PFS
	Clarify referral processes for germline testing	Further information has been included in the HPG
Results and related health information may be shared with genomic and medical databases that are used for patient care. All identifying information will be removed.	Multiple concerns about the use of clinical data for research purposes. Respondents were unclear about whether this clause related to research databases.	Sharing with clinical databases, e.g. ClinVar, is for clinical purposes only and is not used for research. This is standard practice in all lab services. Wording in the consent form has been edited to clarify this.
	Additional information about data sharing should be included in the PFS	This has now been included in the PFS
Results are confidential and will only be shared with my consent, or as required or permitted by law.	Information about this clause should be included on the PFS	This has now been included in the PFS
	Add links to laws in the HPG	Laws, regulations and policy differ in each jurisdiction. Given this form has a national remit, we believe it is not appropriate to include links for every jurisdiction.
I can change my mind about testing and choose not to be told the results, but if testing has started a report will remain in patient medical records.	Further information about withdrawal should be included on the PFS	This has now been included in the PFS
	Information about costs of testing should be made clear	The discussion of cost of testing was felt to be variable across conditions and a separate issue to consent for testing and requires a specific financial consent discussion. Health professionals should

		have a clear discussion with patients regarding cost prior to the test.
If a relevant genetic change has been found, test results and related information can be shared with health professionals to help with the genetic testing of blood relative/s. I understand that identifying information will not be shared with relative/s wherever possible.	Further information about sharing results for the care of blood relatives should be included on the PFS	This has now been included in the PFS
	Clarify that this sharing only refers to germline variants	This has now been included in the PFS

4. *Provide materials for clinical use*

The final tissue-targeted (somatic) consent package materials were uploaded to the Australian Genomics website and distributed to the wider Australian Genomics network in June 2025. The package was also publicised via Australian Genomics social media channels. Working group members were also encouraged to distribute the final package to their own networks.

Online Consent Tool:

Project Aims

This sub-project aimed to create a friendly, informative online tool which allowed patients and their families to learn about genomic testing and its implications at their own pace, with the hope that this would better prepare patients when consenting to genomic testing.

Development

The website content was derived from the information found in the clinical genomic testing patient fact sheet. The wording was edited for an online audience while retaining the overall messaging. The website was structured so that key information such as how the test is carried out, outcomes of testing, benefits and risks, were presented early in the tool. Information which was considered “nice to know” was presented on subsequent webpages. This structure meant that if individuals accessing the tool disengaged, they were still obtaining the most important information needed to make an informed decision about undergoing genomic testing.

Each section of the tool featured a summary of each topic and a drop-down tab to empower the viewer to decide whether they want more detailed information about that topic. An accessibility tab was included, featuring tools to enlarge text, adjust colour contrasting and change text to a more readable font. As a further measure to support accessibility, a speaker function was included in all sections of the tool. This allows individuals to decided which section they wanted to hear rather than only being able to hear the full page in its entirety as offered through the accessibility tab.

Stoke Design Co were enlisted to in January 2024 to develop the tool, with design beginning in March 2024. Design and build of the tool took eight months with a significant proportion of that time dedicated to resolving technical challenges predominantly with the speaker function. The tool was launched in December 2024.

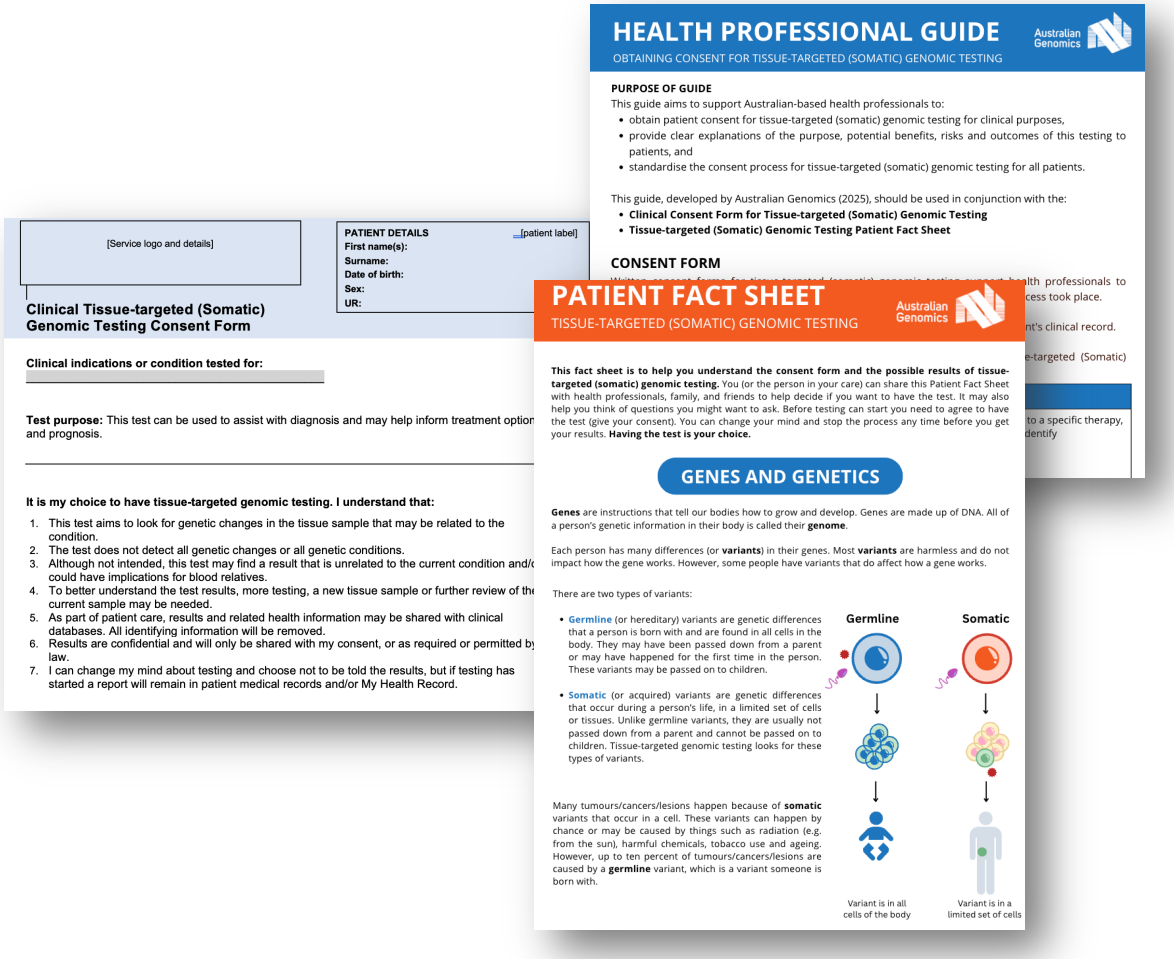
Dissemination

The tool was promoted to clinical genetics services. We suggest clinicians provide it to patients before their first appointment, so they attend the session with some initial genomic testing knowledge. Other uses suggested included sharing the online tool with family members or reviewing the tool post-appointment to help refresh one's memory. The tool was also shared via Australian Genomics social media platforms and newsletter.

1.3 Resources Generated

Tissue-Targeted (Somatic) Genomic Testing Consent Package:

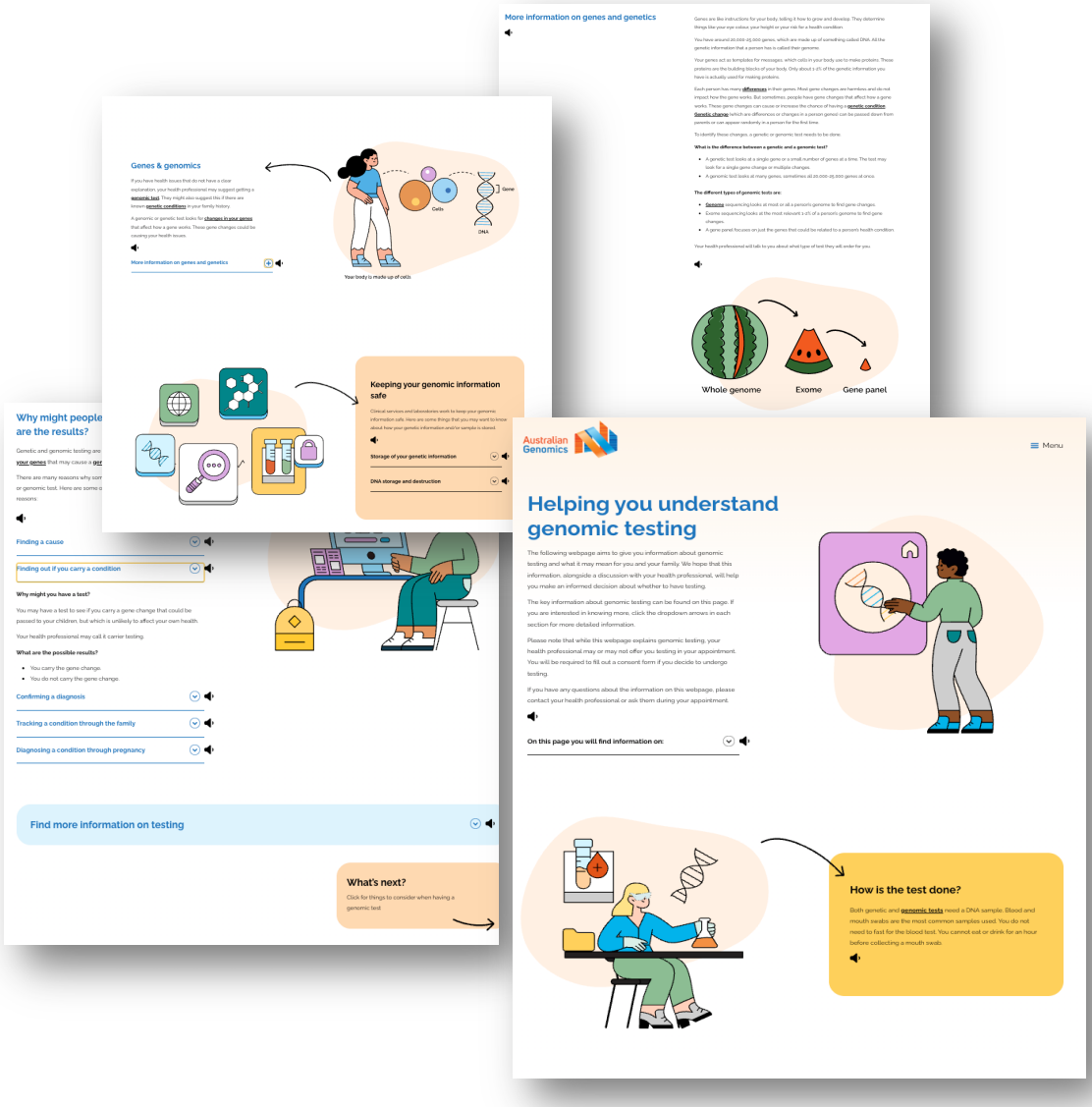
The Tissue-Targeted (Somatic) Genomic Testing Consent Package is located on the Australian Genomics website and is accessible via this link: [Tissue-Targeted (Somatic) Genomic Testing Consent Package]



Online Consent Tool:

The online consent tool is located on the Australian Genomics website and is accessible via this link: Helping you understand genomic testing.

Images from the website can be found below.



1.4 Key Impacts

Tissue-Targeted (Somatic) Genomic Testing Consent Package:

The projected impacts of this project include:

- the use of this form leading to patients having a more consistent experience with undergoing tissue-targeted (somatic) testing
- non-genetic health professionals have reliable guidance when discussing this testing with patients and are aware of the need to refer to clinical genetic services should germline variants be found.

Online Consent Tool:

The key impact of the Online Consent Tool is the delivery of patient information in a clear, friendly manner which empowers the individual to decide how much information they would like on each topic.

1.5 Challenges and Barriers

Tissue-Targeted (Somatic) Genomic Testing Consent Package:

- Given the tight timeframe to conduct this project there was limited opportunity to promote and evaluate the implementation of the consent package. This means that all interested parties may not be aware that the form exists.
- Again, due to time constraints, and evaluation of the package's use in practice was not possible. However, thought the consultation we identified and aim to address the needs of stakeholders.
- Within the consultation some respondents identified issues that were outside the scope of somatic testing which may indicate some of the implementation challenges moving forward.

Online Consent Tool:

During the development of the online tool there were significant impacts to the timeline. This was primarily due to the challenges implementing a speaker function to enhance accessibility. This posed a technical challenge to the development team however was eventually rectified.

1.6 Recommendations

Tissue-Targeted (Somatic) Genomic Testing Consent Package:

1. As indicated though the consultation, efforts should be made to develop resources that support First Nations people undergoing tissue-targeted (somatic) testing, including a specific health professional guide and patient fact sheet. There are groups with interest and expertise in this area such as the Australian Alliance for Indigenous Genomics that should be commissioned to carry out development of these resources.
2. Further implementation activities should be undertaken such as liaising with pathology services at testing sites for integration of the consent clauses into test request forms.

Online Consent Tool:

1. The tool should be made more accessible to people from diverse backgrounds. The speaker function could integrate different languages, or the written content could be translated.
2. An Easy Read function should be added to increase the accessibility of the tool.