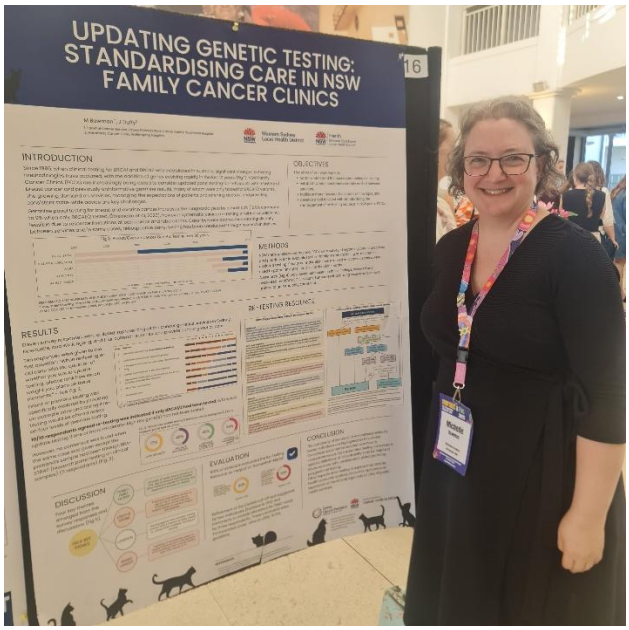
Name: Michelle Bowman
Position & Affiliation: Senior Genetic Counsellor, Familial Cancer Service, Westmead Hospital (WSLHD)
Full Reference: Updating genetic testing: standardising care in NSW Family Cancer Clinics; M Bowman, J Duffy
Conference/Meeting Name: International Meeting of the Psychosocial Aspects of Hereditary Cancer and Familial Aspects of Cancer Research and Practice Meeting (combined event)
Location: Kingscliff, NSW, Australia
Dates: August 25-28 2025
Presentation Type: Poster



International Meeting of the Psychosocial Aspects of Hereditary Cancer (IMPAHC) is a biennial event held in different locations internationally. I had the privilege of attending and presenting in 2019 when it was held in the Netherlands, and it was wonderful to reconnect with several presenters here in my own “back yard”. The Familial Aspects of Cancer (FAC) meeting occurs in Kingscliff annually, and the combined national and international events were an excellent opportunity for continuing education in the rapidly evolving field of cancer genetics.

IMPAHC presentations covered a wide range of topics, including: polygenic risk scores and comprehensive risk assessments across different tumour groups, psychological impacts of surveillance in those at increased risk of pancreatic cancer (Australian research) and in Li Fraumeni syndrome, development of resources such as decision aids, communication about genetic risk within families including minors, as well as service models such as mainstreaming and the role of general practice in risk assessment.

An excellent presentation by a Sydney-based colleague raised the occurrence of vicarious trauma in cancer genetics health professionals. It was one of the first times I have seen a conference turn the lens onto ourselves as practitioners, and prompted several conversations during the break. I look forward to her preliminary work being expanded and published in due course, as doing the best we can for our patients is not possible if we are not also looking after our own mental and physical health.

Another stand-out was an international invited speaker who presented at both meetings on a widely used risk modelling platform, CanRisk. Her talks were accessible and insightful, and also prompted many conversations outside of the sessions. As I am particularly interested in the implementation and use of technology, including integration with patient systems, data privacy and governance, it was very helpful to discuss these aspects with her. We may be able to collaborate in the future in relation to integrating their platform with the clinical genetics database used by NSW.

IMPAHC delegates enjoyed an excellent evening of networking and socialising at a local venue (Husk Distillery) for dinner. The poster session was well attended and was a very good opportunity to share experiences and challenges of service delivery when people visited my poster.

The FAC meeting focussed on scientific/laboratory and research topics. Several presentations addressed advances in technology/analysis for genetic/genomic testing, such as long-read sequencing and copy number analysis. Tumour/somatic testing, variant classification, and treatment trials were also featured topics. A standout for me was the presentation about translating TRACEBACK into clinical practice. Testing for germline variants on stored ovarian cancer tissue has been proven to be feasible in their research setting and offers an important avenue to identify and reduce cancer risk in relatives. The research team is working on further feasibility studies to demonstrate how it can be implemented into clinics, while also trying to address barriers to genetic testing in ovarian cancer so prospectively there are far fewer cases where family members need to seek testing on tumour samples because testing has been undertaken appropriately at the time of diagnosis.

Thank you to Sydney Cancer Partners for enabling my attendance at this combined conference.

The next IMPAHC meeting will be held in Vancouver in 2027, which I hope to also attend.

UPDATING GENETIC TESTING: STANDARDISING CARE IN NSW FAMILY CANCER CLINICS

M Bowman[1*], J Duffy[2]

1. Familial Cancer Service, Crown Princess Mary Cancer Centre, Westmead Hospital
2. Hereditary Cancer Clinic, Wollongong Hospital

INTRODUCTION

Since 1995, when clinical testing for *BRCA1* and *BRCA2* was established in Australia, significant changes to testing methodologies have occurred, with the addition of genes evolving rapidly in the last 10 years (Fig 1). NSW Family Cancer Clinics (FCCs) are increasingly being asked to consider updated panel testing for individuals with a history of breast cancer and previously uninformative germline results, many of whom were only tested for *BRCA1/2* variants. The growing demand on services, managing the expectations of patients and referring doctors, and providing consistent state-wide advice are key challenges.

Germline panel testing for breast and ovarian cancer increases the diagnostic yield by almost 50% (16.6% compared to 9% when only *BRCA1/2* tested, Öfverholm et al, 2023), however systematic clinical re-testing of all individuals is not feasible due to resource limitations at both clinics and laboratories. Case-by-case approaches vary significantly between services and in some cases, retrospective panel testing has been conducted through research initiatives.

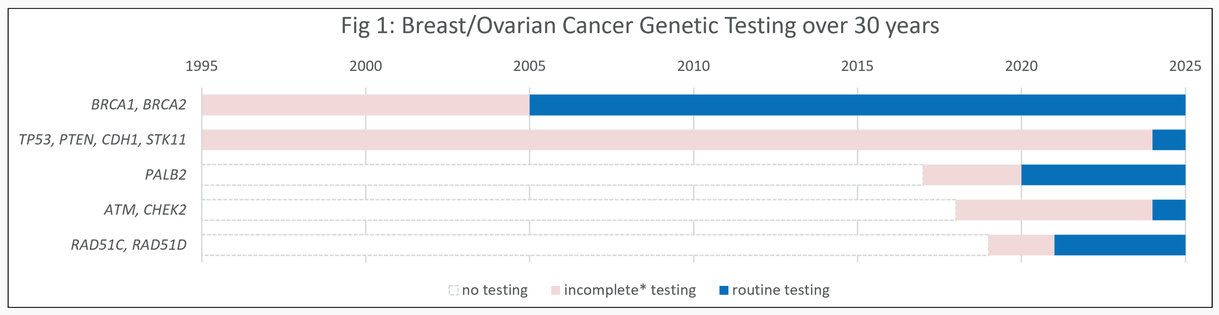


FIG 1: GENETIC TESTING FROM 1995 INCLUDED SINGLE GENES AND MOVED TO PANELS AROUND 2017.
* 'Incomplete testing' may refer to incomplete sequencing, and/or MLPA, and/or single variant testing (e.g. *CHEK2* c.1100del, *ATM* c.7271T>G), or inconsistent inclusion of gene(s) on panels

OBJECTIVES

The aims of our work were to:

- better understand the issues surrounding re-testing,
- establish where consistency already existed between services,
- facilitate more focused discussions on the topic, and
- develop a tool to assist with standardising the management of re-testing requests in NSW public FCCs.

METHODS

NSW metropolitan and regional FCCs were surveyed regarding current practices and priorities for individuals (or their family members) being re-referred for updated testing. Priority considerations were ranked and case scenarios were used to gather insights into clinical decision-making.

A resource (Fig 4) was developed based on these findings, reviewed and evaluated by NSW FCC clinicians. Further feedback using case examples was gained to gain majority consensus.

RESULTS

Eleven survey responses were collated, representing all the cancer genetics services in Sydney, Newcastle, and Wollongong, and four outreach services who provide cancer genetics care.

Ten responses were given to the first question: "When reviewing an old case with the question of whether you would update testing, please rank how much weight you place on these elements:" - see Fig. 2.

Extent of previous testing was specifically explored by providing an example case and asking if re-testing would be offered based on four levels of previous testing.

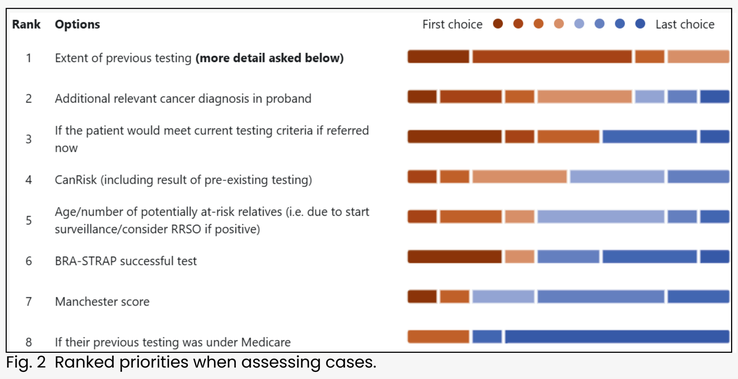
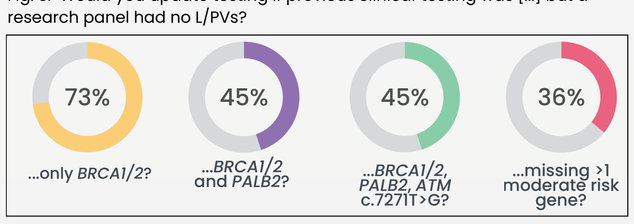


Fig. 2 Ranked priorities when assessing cases.

10/10 respondants agreed re-testing was indicated if only *BRCA1/2* had been tested, 8/10 would update testing if one or more moderate-high risk gene(s) had not been tested.

However, no consensus was found when the same case was given except the proband's sample had been through BRA-STRAP (research panel testing on clinical samples) (11 respondants) (Fig. 3).

Fig. 3. Would you update testing if previous *clinical* testing was [...] but a research panel had no L/PVs?



RE-TESTING RESOURCE

NSW FCC guide to updating testing in breast cancer families

Background:
The goal of this guide is to assist decision-making regarding re-testing of previously uninformative breast cancer germline samples. Past cases may come to attention through a variety of means, including referral of a family member (+/- new cancer diagnoses), re-discussion at an MDT, routine VUS reviews, or other circumstances. The FCCs acknowledge that a standardised, equitable review process is preferred but resourcing makes this impractical at present. This guide was developed with input from NSW FCCs in 2025.

New cancer diagnoses:
If there is a more appropriate affected individual available for testing, this should be the first step. If testing is not possible or declined by the family member, consider re-testing the historical proband according to the below process.
A **new diagnosis in the proband may prompt re-testing** regardless of below process, given the potential treatment implications.

Deceased proband:
Where the proband is deceased, case by case consideration should be given to the terms of the previous consent, who is seeking advice/is available to re-contact (in collaboration with other FCCs if needed), along with the below decision-aid.

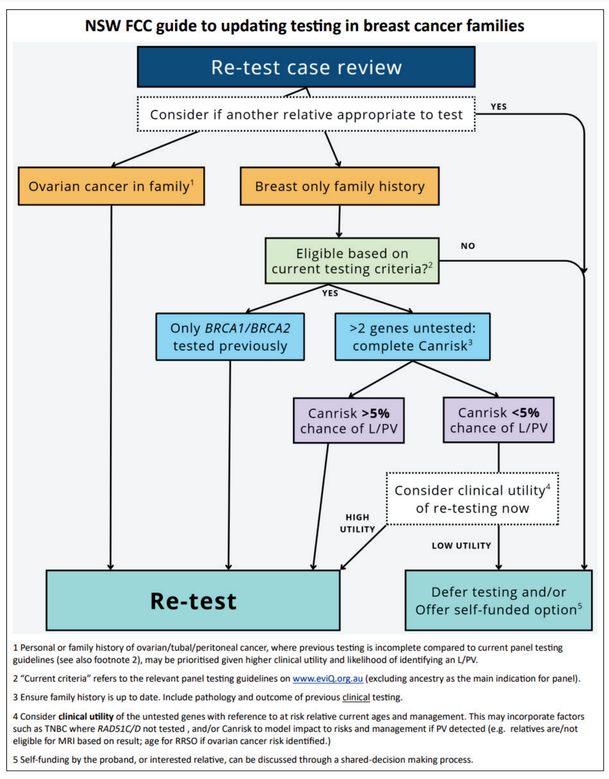
Re-referral:
Individual FCCs will have their own requirements in relation to requiring re-referral for access to MBS billing for consultations and/or testing. Resource limits may also impact their ability to action requests without a new referral, such as being able to reach out to a proband as a "cold call".

Consent:
Wherever possible, the proband should be re-consented to updated testing even when past consents included a statement about future testing. Verbal consent may be sufficient where appropriate written consent is held on file. Ensure "person to contact" has been confirmed or updated.

Research:
Some probands/families will have had samples included in research projects. While this reduces the chance of finding a reportable variant in a clinical test, caution must be given to potential misses of large CNVs, the presence of suspicious VUS not being reported, along with limitations associated with not testing in a NATA accredited lab. FCCs did not reach consensus regarding how much weight to give to uninformative research results once clinical testing has covered high-risk genes.

Rare genes/phenotypes:
Inclusion of genes associated with a clear phenotype (e.g. *PTEN*, *STK11*), including consideration of *TP53* testing falls outside the scope of this guide.

Future testing:
Clinically available PRS +/- other moderate risk genes is anticipated, although the timeframe is unknown. Deferring updating testing in some situations is appropriate, where re-analysis in the (near) future would likely have greater utility for guiding the care of family members.



1 Personal or family history of ovarian/tubal/peritoneal cancer, where previous testing is incomplete compared to current panel testing guidelines (see also footnote 2), may be prioritised given higher clinical utility and likelihood of identifying an L/PV.

2 "Current criteria" refers to the relevant panel testing guidelines on www.eviQ.org.au (excluding ancestry as the main indication for panel).

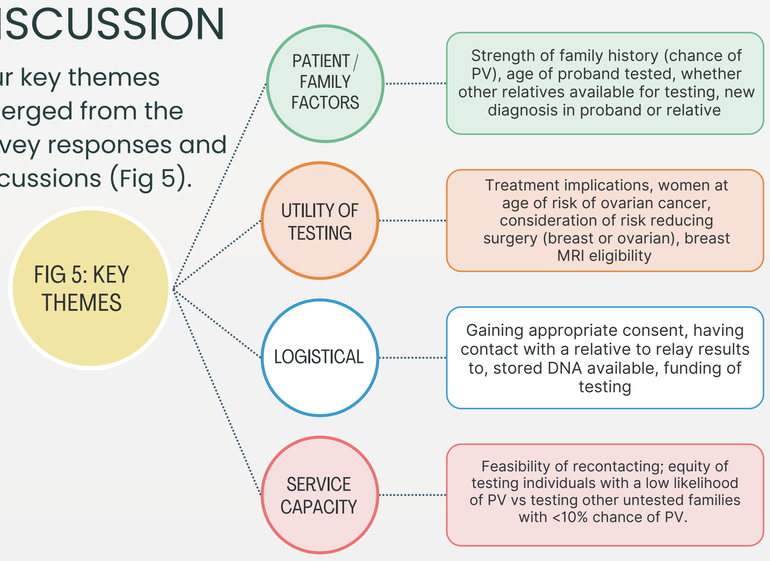
3 Ensure family history is up to date. Include pathology and outcome of previous clinical testing.

4 Consider **clinical utility** of the untested genes with reference to at risk relative current ages and management. This may incorporate factors such as TNBC where *RAD51C/D* not tested , and/or Canrisk to model impact to risks and management if PV detected (e.g. relatives are/not eligible for MRI based on result; age for RRSO if ovarian cancer risk identified.)

5 Self-funding by the proband, or interested relative, can be discussed through a shared-decision making process.

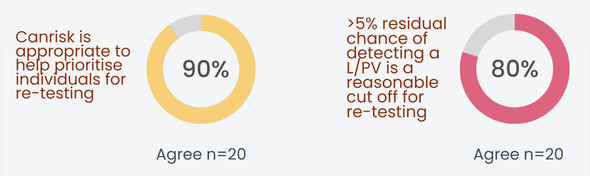
DISCUSSION

Four key themes emerged from the survey responses and discussions (Fig 5).



EVALUATION

100% of clinicians evaluated the Re-Testing Resource as 'Helpful' or 'Somewhat Helpful'.



Refinement of the Canrisk cut-off was suggested by two individuals (increase to 10%) and comments to include allowance for TNBC noted by three respondents. These comments were incorporated under 'clinical utility' in the guideline.

CONCLUSION

The complexity of decisions to re-analyse previously tested individuals are highlighted by this review. Consensus could not be reached over how to incorporate results from expanded testing via research. Specific definitions of 'clinical utility' may be helpful in future versions, with consideration of incorporating polygenic risk testing.
Other aspects of re-testing to explore may include health economic analysis, consumer opinions and exploration of the ethical/legal duty of care of public health services.

REFERENCES

1. Öfverholm, A., Törngren, T., Rosén, A. et al. Extended genetic analysis and tumor characteristics in over 4600 women with suspected hereditary breast and ovarian cancer. BMC Cancer 23, 738 (2023).

Proudly supported by Cancer Institute NSW

M Bowman thanks Sydney Cancer Partners and the Cancer Institute NSW for their support to travel and present this work.