# Ovarian Cancer Genetic Testing Criteria

**Criteria for Mainstream Medicare Funded Testing**
**Germline Testing:**

- ❑ **MBS Item # 73296 – Personal History**
  - High-grade epithelial (non-mucinous) ovarian, fallopian tube or primary peritoneal cancer
- ❑ **MBS Item # 73295 – To determine PARP inhibitor Eligibility**
  - FIGO stage III-IV high-grade serous/epithelial ovarian, fallopian tube or primary peritoneal cancer for whom testing of tumour tissue is not feasible

**Criteria for Mainstream Medicare Funded Testing**
**Somatic (Tumour) Testing:**

- ❑ **MBS Item # 73307 – To determine PARP inhibitor Eligibility**
  - Tumour testing from a patient with FIGO stage III-IV high-grade serous/epithelial ovarian, fallopian tube or primary peritoneal cancer including BRCA1, BRCA2 and HRD status (homologous recombination deficiency)

If not eligible on above criteria, do not offer mainstream genetic testing.

Please note, the tumour (MSB item # 73307) and germline (MBS item # 73296) genetic testing criteria are NOT mutually exclusive. **We recommend every patient with high grade non-mucinous epithelial ovarian cancer have germline genetic testing.**

**Adult Genetics Unit**
Tel: (08) 7074 2697
Fax: (08) 8429 6112
Email: adultgenetics@sa.gov.au

Endorsed by the SA Cancer Clinical Network

Version 2; August 2024.

# Ovarian Cancer
# Mainstream Genetic Testing Checklist

Patient name:
DOB:
UR:
EMR Visit:
(or patient label)

- ❑ Patient's family history taken and documented
- ❑ Provide patient with genetic testing information leaflets
- ❑ Discuss genetic testing with patient
- ❑ Complete Consent to Genetic Testing form
- ❑ Provide patient with completed SA Pathology request form
  - 4mL blood in EDTA tube
  - Copy of histopathology report with lab number
    [for somatic (tumour) testing under MBS item #73307 only]
  - Cc: Responsible Consultant
  - Cc: Adult Genetics Unit, Royal Adelaide Hospital
- ❑ Place a copy of consent form and this checklist in patient notes (Paper or scan to EMR)
- ❑ Ensure patient follow-up appointment in 3 months to discuss results (date of appointment _ _ / _ _ / _ _ )

MO Signature: _____ Date _ _ / _ _ / _ _

Endorsed by the SA Cancer Clinical Network

Version 1; November 2022. Due for review December 2023.

Government of South Australia
SA Health

# Consent to Mainstream Cancer Genetic Testing

PATIENT LABEL (if available)

| | | |
|---|---|---|
| **Name of person to be tested** | | **DOB** |
| **Hospital** | | **UR** |
| **Sample to be collected** | ☐ Tumour Tissue (somatic) ☐ Blood (germline) ☐ Other (_____) | |

I consent to a genetic test for _____

The gene(s)/gene panel being tested is _____

I understand that:

1. The meaning of the result is based on what is known now. This could change in the future.
2. There are limitations to genetic testing:
   - We do not know all the genes that cause cancers.
   - Genetic variants may be found that cannot be interpreted. These are called variants of unknown significance or VUS. A VUS cannot be used to guide clinical care.
3. Rarely, there may be a technical problem with a genetic test. Further sample(s) may be needed.
4. Test results may have implications for both my treatment/cancer risks AND for my family members.

I am aware that:

1. Samples will be stored after testing for at least the period required by laboratory guidelines.
2. I can change my mind about testing at any point before a report is issued.

I consent to the genetic testing described above.
I have had the chance to ask questions and I am satisfied with the answers I have been given.

I give permission for this genetic test result to be retained confidentially by the Adult Genetics Unit and/or given to health care services looking after other members of my family: ☐ Yes ☐ No

Patient signature:..... Date:.....

If I am unable to receive my genetic test result, I nominate the following individual(s) to receive it on my behalf:

Name and Contact Information:.....

Person obtaining consent:..... Signature: .....

Position and specialty of person obtaining consent:.....

Responsible Consultant (please print in capitals): .....

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# PATHOLOGY REQUEST FORM

## Genetic Testing (V1 Nov 2022)

SA PATHOLOGY

AFFIX BARCODE HERE

### PERSON BEING TESTED *(all samples must include at least two patient identifiers)*

| Family Name | Date of Birth | Sex | Ethnicity *(if known)* | Your Ref |
|---|---|---|---|---|
| | | | | |
| Given Name(s) | Medicare No. | | | Telephone |
| | | | | |
| Address: *(Number, Street)* | Suburb | | | Postcode |
| | | | | |

Patient Status at the time of the service or when the specimen was collected:
- ☐ a private patient in a private hospital or approved day hospital facility
- ☐ a private patient in a private recognised hospital
- ☐ a public patient in a recognised hospital
- ☐ an outpatient public of in a recognised hospital

Medicare Assignment "Section 20A of the Health Insurance Act 1973"
I offer to assign my right to benefits to the approved pathology practitioner who will render the requested pathology service(s) and any eligible pathologist determinable services(s) established as necessary by the practitioner.

Patient Signature & Date _____

Your doctor has recommended that you use SA Pathology. You are free to choose your own pathology provider. However, if your doctor has specified a particular pathologist on clinical grounds a Medicare rebate will only be payable if that pathologist performs the service. You should discuss this with your doctor.

### REQUESTING DOCTOR DETAILS

**Requesting Clinician:**

**Responsible Consultant & provider number:**

### COPY REPORTS TO

**Adult Genetics Unit**
**Royal Adelaide Hospital**

### CLINICAL SETTING

- ☒ Diagnostic test ☐ Predictive test
- ☐ Carrier Test ☐ Prenatal *(please tick one)*
- ☒ Affected
- ☐ Unaffected *(please tick one)*

### TEST TYPE *(please tick)*

☐ Common mutation screen ☒ Full gene mutation analysis ☐ Known familial mutation(s)

### CLINICAL NOTES

**This is a <u>Medicare Funded</u> Mainstream Genetic Test**

Consent Obtained and Documented by ordering clinician ☐ YES ☐ NO

MBS Criteria:

- ☐ High Grade Epithelial Ovarian Cancer **(MBS item number 73296)**
- ☐ FIGO Stage III-IV High Grade Epithelial Ovarian Cancer to determine PARP inhibitor Eligibility where tumour testing is NOT feasible **(MBS item number 73295)**

### TESTS REQUESTED

| EDTA | BUCCAL SWAB | OTHER |
|---|---|---|
| | | |

**4ml blood in EDTA tube for:**

1. **Genetic testing: Breast and Ovarian Cancer gene panel analysis (sequencing and del/dup studies)**

### Doctor's Signature & Date

I have verified FULL NAME, DOB and URN on the sample label and request form verbally with the patient and/or checking the patient's ID band.

Collector's Signature: Specimen Collected: / / : Hrs

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Consumer Information Sheet

# Information about Genetic Testing and Cancer

This leaflet was written for people who are thinking about having a genetic test following a cancer diagnosis. This leaflet does not replace a discussion with your managing specialist. If you have any questions or concerns after reading this leaflet, please discuss them with your managing specialist or contact the Adult Genetics Unit.

## What are genes?

The human body is made up of millions of cells. Each cell contains DNA. DNA spells out the genetic instructions (genes) the cells need. Some genes tell cells how to grow, divide and work properly. Some genes help keep DNA healthy. Other genes tell worn-out cells when to self-destruct (die). These genes work together to control cell growth.

## What is cancer?

The DNA in our cells is continually damaged by the things we are exposed to in our environment, for example UV light or cigarette smoke, and the process of aging. This DNA damage is usually repaired but the repair process is not perfect. This means that damage can build up in our DNA. If a cell has too much DNA damage it normally dies.

Cancer occurs when abnormal cells do not die and start to grow in an uncontrolled way. These abnormal cells can damage or invade the nearby tissues or spread to other parts of the body; this is called a cancer.

## What is familial cancer?

Rarely, a person is born with a genetic error (called a variant or mutation) in a cell growth-control gene or a DNA-repair gene. These genetic errors increase the chance of developing a cancer. Usually, the genetic error has been inherited from the person's mother or father. If a genetic error is inherited, other blood relatives may also have an increased chance of developing cancer. This is called familial or hereditary cancer.

## What is a genetic test?

A genetic test involves collecting a sample, usually blood. Genetic material (DNA) is extracted from the sample and analysed looking for genetic errors or variants.

- Everyone's genes have differences or variants, this makes us each unique.
- Most genetic variants are harmless and do not cause problems.
- Some genetic variants change how a gene works and **do** cause a problem, like an increased risk of developing a cancer.
- The names for a variant that causes a medical problem include a disease-causing variant, mutation, genetic error or genetic fault.
- Most genetic tests analyse a number of genes that are all known to cause a particular health problem, like an increased risk of cancer. This is called a gene panel test.
- Genetic testing is part of the standard care for patients with certain types of cancer. This genetic testing is not research based or part of a clinical trial.

Endorsed by the SA Cancer Clinical Network

Version 3; September 2024. Due for review November 2025.

# Consumer Information Sheet

## Why have a genetic test?

There are many reasons a doctor may suggest having a genetic test in the setting of a cancer diagnosis, including:

- To help identify the best treatments for some types of cancer.
- To understand the chance of developing another cancer.
- To help family members understand their cancer risks.
- To help family members manage and reduce their cancer risks through early cancer screening tests and other management options.

## What are the possible outcomes of a genetic test?

1. No genetic variants are found. This is the most common result and is called a negative or uninformative test. This may mean that the cancer did not have an inherited genetic cause or that an inherited genetic cause cannot be found using the currently testing technology.
2. A genetic variant that explains the cancer is found. This is a less common result. This may influence cancer treatments. It also means that other family members may have the variant and can choose to have their own genetic test.
3. A variant that is not understood is found. This is an uncommon result and is called a variant of unknown significance or VUS. A VUS is neither good nor bad; its meaning is just not known yet. Sometimes more testing can help to understand the meaning of a VUS, or the meaning may become clearer overtime. A VUS cannot be used to influence cancer treatments or offer testing to other relatives.
4. An unexpected variant is found. This is a rare result called an incidental finding. It occurs when a genetic variant that causes a different medical problem is found.

## What do I tell my family about genetic testing?

A genetic variant found in you may be relevant for your blood relatives. Genetic variants can occur in both sexes and both sexes can pass a genetic variant down to their children. Telling your family members about a genetic variant can be difficult but may help them understand and manage or reduce their cancer risks.

## What about genetic tests and insurance?

A genetic test result is part of a person's health history. In Australia, premiums for private health insurance do **not** depend on health history. Previously, other types of insurance like income protection and life insurance could have been impacted by genetic testing. However, the laws have recently changed and there is now more protection from genetic discrimination by insurance companies. A genetic test should not impact insurance for a person with a cancer.

## Where can I get more information or support?

- **Watch Our Video** (scan the QR code or use the link) https://t2m.io/zr5HM4O0
- NSW Centre for Genetics Education https://www.genetics.edu.au/
- Inherited Cancers Australia https://www.inheritedcancers.org.au/
- Adult Genetics Unit, Royal Adelaide Hospital Tel: 08 7074 2697



Endorsed by the SA Cancer Clinical Network

Version 3; September 2024. Due for review November 2025.



Government of South Australia
SA Health

# PATHOLOGY REQUEST FORM

## Genetic Testing (V1 Nov 2022)

SA PATHOLOGY

AFFIX BARCODE HERE

### PERSON BEING TESTED *(all samples must include at least two patient identifiers)*

| Family Name | Date of Birth | Sex | Ethnicity *(if known)* | Your Ref |
|---|---|---|---|---|
| | | | | |
| Given Name(s) | Medicare No. | | | Telephone |
| | | | | |
| Address: *(Number, Street)* | Suburb | | | Postcode |
| | | | | |

Patient Status at the time of the service or when the specimen was collected:
- ☐ a private patient in a private hospital or approved day hospital facility
- ☐ a private patient in a private recognised hospital
- ☐ a public patient in a recognised hospital
- ☐ an outpatient public of in a recognised hospital

Medicare Assignment "Section 20A of the Health Insurance Act 1973"
I offer to assign my right to benefits to the approved pathology practitioner who will render the requested pathology service(s) and any eligible pathologist determinable services(s) established as necessary by the practitioner.

Patient Signature & Date _____

Your doctor has recommended that you use SA Pathology. You are free to choose your own pathology provider. However, if your doctor has specified a particular pathologist on clinical grounds a Medicare rebate will only be payable if that pathologist performs the service. You should discuss this with your doctor.

### REQUESTING DOCTOR DETAILS

**Requesting Clinician:**

**Responsible Consultant & provider number:**

### COPY REPORTS TO

**Adult Genetics Unit**
**Royal Adelaide Hospital**

### CLINICAL SETTING

| | | |
|---|---|---|
| ✕ Diagnostic test | ☐ Predictive test | ✕ Affected |
| ☐ Carrier Test | ☐ Prenatal *(please tick one)* | ☐ Unaffected *(please tick one)* |

### TEST TYPE *(please tick)*

☐ Common mutation screen ✕ Full gene mutation analysis ☐ Known familial mutation(s)

### CLINICAL NOTES

**This is a <u>Medicare Funded</u> Mainstream Genetic Test**

Consent Obtained and Documented by ordering clinician ☐ YES ☐ NO

MBS Criteria:

☐ Tumour Testing from a patient with FIGO Stage III-IV High Grade Epithelial Ovarian Cancer to determine PARP inhibitor Eligibility **(MBS Item 73307)**

### TESTS REQUESTED

| EDTA | BUCCAL SWAB | OTHER |
|---|---|---|
| | | |

**Please send away to Peter MacCallum Cancer Centre, 305 Grattan Street, Melbourne VIC 3052**

**Tumour testing for:**

**1. BRCA1 and BRCA2 gene sequencing AND HRD status from tumour block**

**Histopathology report # _____ (please include a copy of the report)**

### Doctor's Signature & Date

I have verified FULL NAME, DOB and URN on the sample label and request form verbally with the patient and/or checking the patient's ID band.

Collector's Signature: Specimen Collected: / / : Hrs

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# MOLECULAR ONCOLOGY TEST REQUEST FORM

Peter Mac
Peter MacCallum Cancer Centre
Victoria Australia

NATA & RCPA Accredited Laboratory Number 2465

RCPA The Royal College of Pathologists of Australasia

NATA

## PATIENT DETAILS – Or place Bradma label below

Surname: ..... ☐ Male ☐ Female

First name: ..... DOB: ...../...../.....

Address: .....

.....

Medicare Number: ☐☐☐☐☐ ( ☐ )

Peter Mac UR No (if known): .....

## REQUESTING CLINICIAN / PATHOLOGIST

Doctor name: .....

Address: .....

Provider No: .....

Email*: .....

Fax: ..... Phone: .....

Signature: ..... Date ...../...../.....

## REPORT COPY TO CLINICIAN/HEALTHCARE PROVIDER

Doctor name: .....

Address: .....

Email*: .....

Fax: ..... Phone: .....

Molecular reports are distributed by secure email. A legible institutional email address must be provided for this purpose. Report links remain active for 14 days and are accessible only to the recipients listed here.

## TISSUE FOR TESTING

Originating pathology lab: .....

Lab accession number: .....

Block ID: .....

Permission to exhaust block: ☐ Yes ☐ No If not selected, permission is assumed to be given

☐ Please tick if the specimen was collected after neoadjuvant chemotherapy was commenced

## SELECT PAYMENT OPTION

☐ **Hospital/Pathology Provider**

☐ **Other** - please specify: .....

☐ **Medicare -** Patient must sign in box on right. Non-rebatable components will be billed to the pathology provider unless otherwise specified). If a test is being requested through Medicare, the patient's hospital status at the time of the service or when the specimen was collected is required below.

- ☐ Private patient in a private hospital, or approved day hospital facility
- ☐ Public patient in a recognised hospital.
- ☐ Private patient in a recognised hospital.
- ☐ Outpatient of a recognised hospital.

☐ **Bill Patient** (For non-MBS items - must discuss with patient first. Complete separate financial consent form)

☐ Authority for Peter Mac to submit claim on behalf of claimant

I authorise the approved pathology practitioner who will render the requested pathology services, and any further pathology services which the practitioner determines to be necessary, to submit my unpaid account to Medicare, so that Medicare can assess my claim and issue me a cheque made payable to the practitioner, for the Medicare benefit. Verbal consent was provided by patient to submit unpaid account to Medicare (no signature available).

Your doctor has requested testing from Peter MacCallum Pathology. You are free to choose your own pathology provider however, if your doctor has specified a particular pathologist on clinical grounds, a Medicare rebate will only be payable if that pathologist performs the service. You should discuss this with your doctor. **Privacy Note:** The information provided will be used to assess any Medicare benefit payable for the services rendered and to facilitate the proper administration of government health programs, and may be used to update enrolment records. Its collection is authorised by provision of the Health Insurance Act 1973. The information may be disclosed to the Department of Health and Ageing or to a person in the medical practice associated with this claim, or as authorised/ required by law

## MOLECULAR TEST PANELS - see reverse for test details

Occasional FFPE samples may fail to yield sufficient high quality tumour DNA/RNA for molecular testing. These samples will be reported as insufficient. Details of regions covered by NGS panels are available on request. When selecting dual DNA/RNA-based tests, please select DNA analysis, RNA analysis, or both.

| TEST NAME / DESCRIPTION (See reverse for details) | | MBS ITEM OR COST |
|---|---|---|
| ☐ **Melanoma Panel –** *BRAF, NRAS, KIT, GNAQ, GNA11* | | 73336 |
| ☐ **Colorectal Panel –** *KRAS, NRAS, BRAF, PIK3CA* | | 73338 |
| ☐ **NSCLC Panel –** *EGFR, KRAS, BRAF, ERBB2, MET* exon 14 skipping *ALK, ROS1, RET, NTRK1, 2, and 3 fusions, MET* exon 14 skipping ☐ (**Select to include PDL1 IHC** 72814) | DNA ☐<br>RNA ☐<br>DNA & RNA ☐ | 73438<br>73439<br>73437 |
| ☐ **NSCLC EGFR -** *EGFR* only | | 73337 |
| ☐ **NSCLC MET -** *MET* exon 14 skipping only | | 73436 |
| ☐ **NSCLC T790M -** *EGFR* T790M only | | 73351 |
| ☐ **GIST Panel –** *KIT, PDGFRA, BRAF* followed by SDH and panTRK IHC if no variants detected | | $350 |
| ☐ **Breast Cancer –** *PIK3CA only* | | $200 |
| ☐ **Thyroid Panel –** *BRAF, NRAS, KRAS, HRAS, RET, TP53* OPA fusions and transcript variants | DNA ☐<br>RNA ☐<br>DNA & RNA ☐ | $350<br>$600<br>$885 |
| ☐ **Brain Panel –** Full OPA (see over for details) OPA fusions and transcript variants including *EGFR* vIII | DNA ☐<br>RNA ☐<br>DNA & RNA ☐ | $600<br>$600<br>$885 |
| ☐ **Oncomine Precision Assay (OPA) –** Variants in 50 genes & CNA Fusions and transcript variants (CNA= copy number alterations) | DNA ☐<br>RNA ☐<br>DNA & RNA ☐ | $600<br>$600<br>$885 |
| ☐ **Single Gene DNA Analysis –** Specify any single OPA gene (see reverse). Gene name: ..... (Please indicate clinical context in CLINICAL NOTES below) | | $350 |
| ☐ **Granulosa Cell Tumour of the Ovary –** *FOXL2* C134W only (Sanger sequencing) | | 73377 |
| ☑ **Ovarian HRD Panel –** HRD (Genomic Instability status), *BRCA1, BRCA2* | | 73307 |
| ☐ **Ovarian HRR Panel** - *BRCA1, BRCA2* | | 73301 |
| ☐ **Prostate HRR Panel –** *BRCA1, BRCA2* | | 73303 |
| ☐ **Sarcoma Panel –** 507 gene targeted RNA fusion panel (MBS items applicable – see over) | | $1000 |
| ☐ nexomics **TruSight Oncology 500 Panel** 523 gene DNA/RNA comprehensive genomic profiling | | $2670 |

## CLINICAL NOTES – Histopathology report must be provided

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.....

.....

.....

.....

**PLEASE NOTE:** THESE ASSAYS MAY DETECT GERMLINE VARIANTS WITH SIGNIFICANT IMPLICATIONS FOR BOTH THE PATIENT AND THEIR FAMILY. PLEASE ENSURE YOUR PATIENT IS APPROPRIATELY COUNSELLED PRIOR TO TESTING.

## ADDRESS & CONTACT DETAILS


Molecular Pathology (Level 4)
Peter MacCallum Cancer Centre VCCC Building
305 Grattan Street
Melbourne VIC 3000

Tel: +61 3 8559 5405
Fax: +61 3 8559 5409
Email: path_admin@petermac.org


## MEDICARE ASSIGNMENT FORM (Section 20A of the HIA 1973)

I offer to assign my right to benefits to the approved practitioner who will render the requested pathology service(s) and any eligible pathological determinable service(s) established necessary by the practitioner.

Patient's Signature:

Date ...../...../.....

# MOLECULAR ONCOLOGY TEST REQUEST FORM

Peter Mac
Peter MacCallum Cancer Centre
Victoria Australia

NATA & RCPA Accredited Laboratory
Number 2465

RCPA
The Royal College of Pathologists of Australasia

NATA

## SAMPLE REQUIREMENTS

Please send a formalin fixed paraffin embedded (FFPE) tissue block containing tumour, or 9 (14 if IHC listed) 4µm unstained sections on charged slides with a copy of the pathology report to the address on page 1. Blocks will be returned on completion of testing. Specimens with <20% tumour content, extensive necrosis, calcification, or those collected after initiation of chemotherapy should be avoided where possible.
**Please contact molecular.oncology@petermac.org for technical enquiries**

## TEST DETAILS

| | |
|---|---|
| **Melanoma Panel**<br>TAT ≤1 week (from specimen receipt) | NGS of clinically relevant variants in *BRAF, NRAS, KIT, GNAQ and GNA11*. Results are used to determine eligibility of stage III or stage IV metastatic cutaneous melanoma patients for treatment with dabrafenib, vemurafenib or encorafenib. Costs are covered by MBS item 73336. |
| **Colorectal Panel**<br>TAT ≤1 week (from specimen receipt) | NGS of clinically relevant variants in *KRAS, NRAS, BRAF and PIK3CA*. Results are used to determine eligibility of metastatic colorectal cancer (stage IV) patients for treatment with cetuximab, panitumumab or encorafenib. Costs are covered by MBS item 73338. |
| **NSCLC Panel**<br>OPA consists of a DNA and an RNA component, one or both components can be requested.<br>TAT ≤1 week (from specimen receipt) | NGS of clinically relevant DNA variants in *EGFR, KRAS, BRAF, ERBB2* and *MET* exon 14 skipping variants (including copy number alterations in *EGFR, KRAS, ERBB2, MET* and *CDKN2A*) and/or RNA alterations in *ALK, ROS1, RET, NTRK1, NTRK2, NTRK3*. Results are used to determine eligibility of newly diagnosed NSCLC patients for treatment with targeted kinase inhibitor therapies or immunotherapy. Costs are covered by MBS items 73437 includes the DNA and RNA component, 73438 includes the DNA component only, 73439 includes the RNA component only. |
| **NSCLC EGFR**<br>TAT ≤1 week (from specimen receipt) | NGS of clinically relevant variants in *EGFR*. Results are used to determine eligibility of NSCLC (non-squamous histology or not otherwise specified) patients for treatment with kinase inhibitors or immunotherapy. Costs are covered by MBS items 73337. |
| **NSCLC MET exon 14 skipping**<br>TAT ≤1 week (from specimen receipt) | NGS and/or RNASeq of *MET* to detect in-frame skipping of exon 14. Results are used to determine eligibility of locally advanced or metastatic non-small cell lung cancer patients for treatment with tepotinib. Costs are covered by MBS item 73436. |
| **NSCLC T790M**<br>TAT ≤1 week (from specimen receipt) | NGS analysis of *EGFR* T790M. Results are used to determine eligibility of NSCLC locally advanced (Stage IIIb) or metastatic (Stage IV), following progression on or after EGFR tyrosine kinase inhibitor (TKI) for treatment with osimertinib. Costs are covered by MBS item 73351. |
| **GIST Panel**<br>TAT ≤1 week (from specimen receipt) | NGS of clinically relevant variants in *KIT, PDGFRA* and *BRAF*. Results are used to determine treatment strategy with multikinase inhibitors for gastrointestinal stromal tumour (GIST) patients. If no variants are detected, SDH and panTRK IHC are also performed. No Medicare rebate available. |
| **PIK3CA for Breast Cancer**<br>TAT ≤1 week (from specimen receipt) | NGS of clinically relevant variants in *PIK3CA*. Results are used to determine eligibility of HR-pos, HER2-neg advanced breast cancer patients for treatment with alpelisib in combination with anti-hormone therapy. No Medicare rebate available. |
| **Thyroid Panel**<br>OPA consists of a DNA and an RNA component, one or both components can be requested<br>TAT ≤1 week (from specimen receipt) | NGS of clinically relevant variants in *BRAF, NRAS, KRAS, HRAS, RET, TP53** and OPA fusions which includes *ALK, BRAF, NTRK1, NTRK2, NTRK3* and *RET*.<br>No Medicare rebate available.<br>* Please note: *TP53* coverage on OPA is limited to the DNA binding domain only. |
| **Brain Panel**<br>OPA consists of a DNA and an RNA component, one or both components can be requested<br>TAT ≤2 weeks (from specimen receipt) | Full OPA DNA and RNA panel including *BRAF, EGFR, IDH1, IDH2, CDKN2A, EGFR*VIII and OPA fusions. See details of full panel below. No Medicare rebate available.<br>Please Note: OPA analysis does not include *TERT* promotor, 1p/19q co-deletion, *MGMT* methylation, *ATRX or H3F3A*. |
| **Oncomine Precision NGS Assay (OPA)**<br>OPA consists of a DNA and a RNA component, one or both components can be requested.<br>TAT ≤2 weeks (from specimen receipt) | Next generation sequencing (NGS) of hotspot variants in *AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CDKN2A, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNA11, GNAQ, GNAS, HRAS, IDH1, IDH2, KIT, KRAS, MAP2K1, MAP2K2, MET, MTOR, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, SMO, TP53**, copy number alterations in *ALK, AR, CD274, CDKN2A, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA, PTEN*, gene fusions in *ALK, BRAF, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3* and transcript variants in *AR, EGFR, MET*. No Medicare rebate available.<br>* Please note: *TP53* coverage on OPA is limited to the DNA binding domain only. |
| **Single gene DNA analysis selected from OPA**<br>≤1 week (from specimen receipt) | NGS of clinically relevant DNA variants in any single OPA gene (see list above) e.g., *KRAS* in pancreatic cancer, *IDH1* & *IDH2* in cholangiocarcinoma, *CTNNB1* for sporadic Desmoid-type fibromatosis (DTF) diagnosis. |
| **Granulosa Cell Tumour of the Ovary**<br>TAT ≤2 weeks (from specimen receipt) | Sanger sequencing of *FOXL2* c.402C>G p.C134W. Results are used to assist diagnosis of granulosa cell ovarian tumour. Costs are covered by MBS item 73377. |
| **Ovarian HRD Panel**<br>TAT ≤3 weeks (from specimen receipt) | Genomic Instability Index calculated from low-pass whole genome sequencing (WGS) combined with targeted NGS of *BRCA1* and *BRCA2*. Results are used to determine eligibility of patients with advanced (FIGO III-IV), high grade serous or high grade epithelial ovarian, fallopian tube or primary peritoneal cancer for treatment with poly-ADP ribose polymerase (PARP) inhibitors. Costs are covered by MBS item 73307. |
| **Ovarian HRR Panel**<br>TAT ≤ 3 weeks (from specimen receipt) | Targeted NGS of *BRCA1* and *BRCA2*. Results are used to determine eligibility of patients with relapsed high grade serous or high grade epithelial ovarian, fallopian tube or primary peritoneal cancer, for access to treatment with a poly-ADP-ribose polymerase (PARP) inhibitors. Costs are covered by MBS item 73301.<br>Note: Newly diagnosed patients are eligible for Ovarian HRD Panel above. |
| **Prostate HRR Panel**<br>TAT ≤3 weeks (from specimen receipt) | Targeted NGS of *BRCA1* and *BRCA2*. Results are used to determine eligibility of patients with metastatic castration-resistant prostate cancer for treatment with poly-ADP ribose polymerase (PARP) inhibitors. Costs are covered by MBS item 73303. |
| **Sarcoma Panel**<br>TAT 4 weeks (from specimen receipt) | Targeted RNAseq for 507 fusion genes relevant to sarcoma and rare soft tissue malignancies. Partial Medicare rebate is available. A gap fee may apply. Applicable MBS items: 73374, 73375, 73376, 73378, 73379, 73380, 73381, 73382, 73383. |
| nexomics<br>**TruSight Oncology 500 (TSO500)**<br>TAT 3 weeks (from specimen receipt) | Comprehensive Genomic Profiling of solid tumours for driver variants in 523 genes, 55 gene fusions, gene amplifications, tumour mutation burden (TMB), certain mutational signatures, and microsatellite instability (MSI). No Medicare rebate available. |

Consumer Information Sheet

# Information for people considering genetic testing of their tumour

This leaflet is for people who are thinking about having genetic testing done on their tumour or cancer tissue. It is intended to help people understand and make decisions about this testing. It does not replace a discussion with your managing specialist. If you have any further questions or concerns after reading this leaflet, ask your managing specialist or contact the Adult Genetics Unit.

## Genes and Genetic Testing

Genes are the instructions the body uses to grow, develop and work. Genes are written in DNA. Genetic testing involves collecting a sample (blood, hair, tumour tissue), extracting DNA from the sample, and testing the DNA to look for changes in the genes.

## Somatic Genetic Changes

A cancer forms when certain genetic changes develop in a cell. The genetic changes allow the cancer cells to grow and spread abnormally. This type of change is called a “somatic genetic change”

- Somatic genetic changes develop as you age; you are not born with these changes.
- Somatic genetic changes are only found in certain cells in the body, like tumour or cancer cells.
- Somatic genetic changes cannot be passed on to children.
- To find somatic genetic changes, testing is done on a sample of tumour or cancer, often from a biopsy or surgery.

## Somatic (Tumour) Genetic Testing

This is usually done in consultation with a cancer specialist (i.e. your oncologist), as part of your clinical care, or possibly as part of research or a clinical trial.

- Tumour testing looks for somatic genetic changes in tumour or cancer cells.
- Tumour testing can sometimes help make decisions about the best treatment for a cancer.

## Germline Genetic Changes

We are all born with genetic changes that are in all the cells of our body. This type of change is called a “germline genetic change”.

- Germline genetic changes make us each unique.
- Germline genetic changes are usually passed down from a parent, and can be passed down to a child (inherited).

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## Germline Genetic Changes and Health

- Most germline genetic changes are harmless and do **not** affect your health, they are called normal variants.
- Some germline genetic changes can affect your health or cause a health problem.
- Germline genetic changes that cause a health problem are called disease-causing variants, mutations, or genetic faults/errors.

## The Overlap Between Somatic and Germline Changes

- Germline and somatic (tumour) genetic testing sometimes overlap.
- Because germline genetic changes are present in all the cells of the body, they are also present in the cells of a cancer or tumour.
- This means tumour genetic testing can sometimes find germline genetic changes that are important for both the person with cancer **and** for other family members.
- This sort of genetic test result is uncommon and often unexpected.

## Emotions, Family and Tumour Genetic Testing

Genetic test results can have emotional impacts for both the person having the test and their family. If you are having any sort of genetic test, think about:

- How you might feel receiving the results, including unexpected results.
- How you will share the results with your family members, if required.
- How to be open with, and supportive and respectful of other family members' responses to a genetic test result.

## More Information

If you have questions or worries about a genetic test, you can talk to your specialist. In some situations, your specialist may refer you to a Clinical Genetics Service.

**Other places to get information:**
**Seattle Children's Hospital leaflet** (Somatic and Germline Cancer Testing)
www.seattlechildrens.org/globalassets/documents/for-patients-and-families/pfe/pe2960.pdf
**Centre for Genetics Education**
http://www.genetics.edu.au/

**If you have further questions or concerns, you can speak to your cancer specialist or contact:**

**The Adult Genetics Unit**
Royal Adelaide Hospital (8F401.52, MDP 63)
Port Road, ADELAIDE, SA 5000
Telephone: 08 7074 2697 Fax: 08 8429 6112



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