Jreissati Pancreatic Centre | Epworth

Understanding genetic and molecular testing

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Acknowledgements

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The Jreissati Pancreatic Centre at Epworth acknowledges the Traditional Custodians of Country across Australia. We recognise their continuing connection to land, waters, communities and culture.

We pay our respects to Aboriginal and Torres Strait Islander Peoples and to Elders past, present and emerging.

Note to readers

This booklet provides general information about pancreatic cancer. It is not intended to replace advice from your treating team or other qualified professionals. You should always speak with your doctor about decisions that may affect your health. If you require legal, financial or psychological support, we encourage you to seek advice from appropriately trained specialists.

Every effort has been made to ensure the information is accurate at the time of publication. However, evidence and clinical guidance relating to cancer care are continually evolving. The Jreissati Pancreatic Centre at Epworth is not responsible for any loss, injury or damage resulting from reliance on this material.

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VICTORIAN CANCER AGENCY

VICTORIA State Government

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About the Jreissati Pancreatic Centre at Epworth

Pancreatic cancer is the eighth most diagnosed cancer in Australia. In 2025, it was estimated that 4,825 people would receive a pancreatic cancer diagnosis. This would account for 2.8% of all cancer diagnoses in the country.[1]

The Jreissati family are all too familiar with the fear and uncertainty that follow a diagnosis of pancreatic cancer, having lived through the experience when Elias was diagnosed at age 54. Knowing the impact the disease has had on their family, Elias, his wife Colleen and the rest of the Jreissati family wanted to change the patient and family experience for others. This led to the establishment of the Jreissati Pancreatic Centre at Epworth in 2021. This Centre provides clinical care and support to patients with a diagnosis of pancreatic cancer, supports early diagnosis and better patient outcomes, advocates for patients and raises awareness and understanding of the disease. 100% of the funding it receives is directed into research, patient support and clinical care.

Our centre comprises three intersecting parts, each of which is integral to improving patient outcomes and experiences

- Comprehensive patient-centred clinical care
- Patient advocacy and support
- Clinical trials and research

We are a specialist centre offering a centralised model of care for pancreatic disease management. This means our patients have access to all healthcare professionals required for their care within a single centre and hospital network. International studies support the view that centralised care improves overall patient outcomes.[2] By managing patients through one main centre, case volume increases, which in turn enhances clinical expertise. This growing expertise drives innovation and leads to advances that will improve patient procedures and treatments in the future.

The **patient experience** is a central focus of our centre. Receiving a diagnosis of pancreatic cancer can be an overwhelming and emotional time, often bringing many questions and uncertainties. Knowing that you are not facing this alone, and understanding where to turn for support, is very important. Once you are referred to the centre, you will meet our pancreatic nurse coordinator, who will be there to guide and support you throughout your care at Epworth. They will be your first point of contact and are always available to answer any questions or concerns you or your family may have.

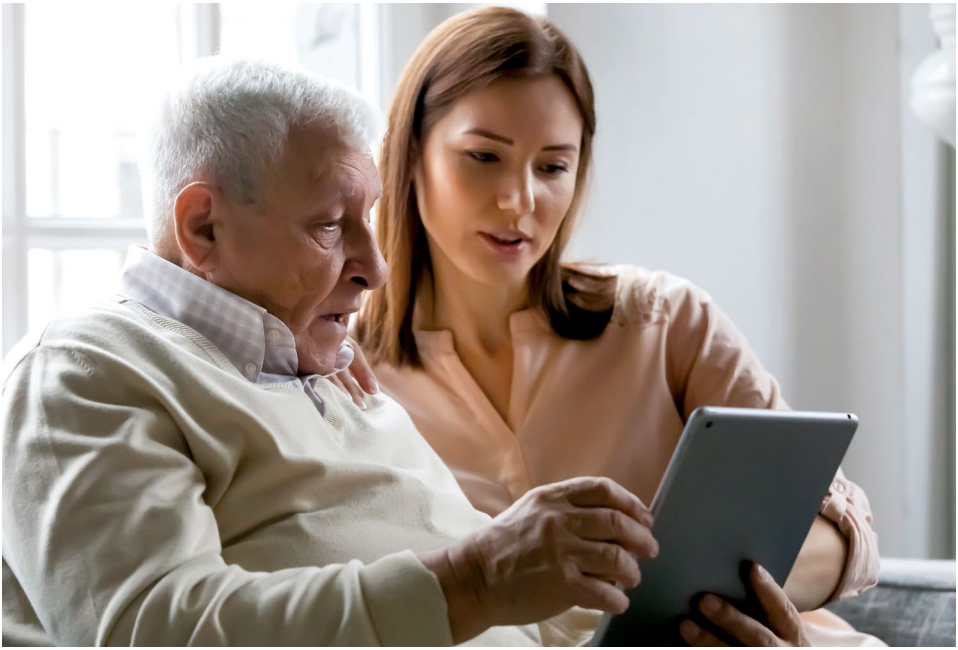
Understanding genetic and molecular testing

Genetic and molecular testing can provide important information about your cancer and how best to treat it. In some cases, testing may also help identify whether your family members could be at increased risk.

This booklet explains the different types of testing used in pancreatic cancer. It outlines what the results may show, how they can guide treatment or clinical trials, and what they may mean for your family.

Not everyone will need genetic testing. Your care team will explain whether it is appropriate in your situation and discuss any next steps

This resource does not replace medical advice. Always speak with your healthcare team about decisions that may affect your care.



Notes

What is genetic and molecular testing?

Genetic and molecular testing looks for changes in your DNA that may affect how your cancer behaves or how it responds to treatment. These tests may be done on a blood sample, a saliva sample or a tumour biopsy.

There are two main types of changes:

- **Inherited (germline) changes** (also called genetic testing): These are changes passed down from a parent. They are present in all cells of the body and may increase the risk of certain cancers, including pancreatic cancer.
- **Tumour-specific (somatic) changes** (also called molecular testing): These changes occur only in the cancer cells. They are not inherited and are not passed on to family members.

Inherited testing is often done through blood or saliva. It may look for mutations in genes such as *BRCA1*, *BRCA2*, or *PALB2*, which are linked to breast, ovarian and pancreatic cancers.

Tumour testing is done using a sample of the cancer itself, often taken during a biopsy or surgery. It can identify changes that may help guide treatment, including access to targeted therapies or clinical trials. Your care team will explain which type of testing is recommended and why.

When and why these tests are offered

Genetic and molecular testing is not routine for everyone with pancreatic cancer, but it is increasingly used to support personalised care.

Your care team may recommend one or both of the following types of testing, depending on your diagnosis and treatment options.

Inherited (germline) testing may be offered when:

- You are diagnosed with pancreatic cancer, especially at a younger age or with a family history
- You have a personal or family history of other cancers, such as breast, ovarian or prostate
- A gene change has already been found in a family member
- Your care team wants to understand whether inherited risk factors are involved

Inherited testing may also help family members understand their own cancer risk and consider further testing, screening or prevention strategies.

Tumour (molecular or somatic) testing is often performed when:

- You are newly diagnosed with advanced or metastatic pancreatic cancer
- Standard treatments are not working, and new options are being considered
- Your oncologist is looking for targeted therapies or clinical trials
- Testing may influence the choice or timing of chemotherapy

Tumour testing looks for specific changes in the cancer that may respond to newer treatments, including targeted therapies or immunotherapy.

Testing is optional. Your care team will explain whether testing is appropriate in your case and how the results may support your care or inform family members.

Understanding germline testing and inherited risk

Germline testing looks for inherited changes in your genes that may increase the risk of developing certain cancers. These changes are present from birth and can be passed from parent to child.

In pancreatic cancer, inherited mutations are uncommon but may be found in genes such as:

- *BRCA1* and *BRCA2*, which are also associated with breast, ovarian and prostate cancers
- *PALB2, ATM, CDKN2A* and others, which are linked to increased pancreatic cancer risk

If an inherited mutation is found, this may have important implications for your care and for your family.

What the results may mean for you

- May help explain why the cancer developed, particularly in the absence of other risk factors
- May guide follow-up care or screening for other types of cancer
- May influence treatment options in some cases

What the results may mean for your family

- Close relatives (such as siblings or children) may be offered genetic testing
- Family members who carry the same mutation can access screening or prevention programs
- Those who do not carry the mutation may avoid unnecessary monitoring

Support with decision-making

If germline testing is being considered, your care team may refer you to a genetic counsellor. These health professionals are trained to help you understand how inherited conditions may affect you and your family.

The genetic counsellor will:

- Explain what the test looks for and what the results may mean
- Discuss how the results could affect your family
- Support you in deciding whether to proceed with testing
- Help you plan how to share information with family members if needed

You can take time to consider your options. Testing is voluntary, and support is available at every stage.

Understanding tumour testing and targeted treatment

Tumour testing, also called somatic or molecular testing, looks for changes in the DNA of cancer cells. These changes are not inherited and are found only in the tumour.

Testing is done on a sample of tumour tissue, often collected during a biopsy or surgery.

What the results may show

Tumour testing may identify:

- Specific mutations or molecular features that explain how the cancer developed
- Changes that suggest the cancer may respond to targeted therapies or immunotherapy
- Eligibility for clinical trials investigating new treatment options

This information can help your oncologist select treatments that are more likely to be effective.

Limitations of tumour testing

Not all cancers have a known or targetable mutation. In some cases, the test may not find any changes that influence treatment. In others, there may not be enough tumour tissue to complete the test.

Your care team will explain whether tumour testing is recommended in your situation and how the results may support decision-making.

What to expect from the testing process

If your care team recommends genetic or molecular testing, they will explain the steps involved and help you decide whether to proceed. The process may vary slightly depending on the type of test.

Genetic and molecular testing may be covered by Medicare, or there may be out-of-pocket costs. Your healthcare team can provide information about any expected costs to help you make an informed decision.

Inherited (germline) testing

- Typically involves a blood or saliva sample
- Often arranged through referral to a genetic counsellor or familial cancer centre
- You will receive pre-test information about the purpose of the test, possible results, and how they may affect you and your family
- Results are usually available within several weeks

Tumour (molecular) testing

- Usually performed on a tissue sample collected during your biopsy or surgery
- No additional procedure is required unless there is not enough tissue available
- Results may take several weeks
- Your oncologist will explain the findings and how they may influence treatment

You will be supported throughout the testing process. This may include:

- Pre-test counselling to help you understand your options
- Post-test discussions to explain the results and next steps
- Support to share relevant information with family members, if an inherited risk is identified

All testing is optional. You are encouraged to ask questions and take time to make an informed decision. You can decline testing if it does not align with your preferences.

Understanding your results and what they mean

Test results are usually available within a few weeks. Your care team will let you know when to expect them and arrange a time to discuss the findings.

The results may help guide your treatment or identify options such as targeted therapy or clinical trials. They may also provide information about inherited cancer risk that could be relevant to your family. In some cases, results may not lead to any immediate changes in your care but may still help inform future decisions.

Common types of results	What it means
Pathogenic or likely pathogenic variant	A gene change was found that is believed to increase cancer risk or influence treatment. This may help guide your care and could have implications for your family.
Variant of uncertain significance (VUS)	A gene change was found, but it is not yet clear whether it increases cancer risk. These results may be updated in future as more information becomes available.
No significant finding	No gene changes of clinical relevance were identified. This does not rule out cancer risk but means no action is required based on current knowledge.

What happens next

- Your oncologist or genetic counsellor will explain the results and what they may mean for your care
- If an inherited mutation is found, your family may be offered genetic counselling or testing
- You may be referred for further follow-up, additional screening or support
- If the result has no immediate impact, it will still be recorded in your medical history

Your team will also discuss how your results will be handled, whether they need to be shared with other health professionals or family members, and how your privacy will be protected.

How your genetic information is used and protected

If you choose to have genetic or molecular testing, your results will be treated as confidential medical information. They will only be shared with members of your healthcare team who are directly involved in your care.

Your results may also be:

- Used to help plan your treatment
- Discussed in multidisciplinary meetings (with your privacy protected)
- Stored in your medical record for future reference

If an inherited (germline) gene change is found, you will be asked whether you would like your family members to be informed. This is always your choice, and support is available to help you with this process.

What about insurance?

In Australia, your genetic results cannot be used to deny you access to Medicare or private health insurance. However, they may be relevant for life insurance, income protection or travel insurance. If you are concerned, your genetic counsellor can explain your rights and how to seek advice before making any new insurance applications.

Support, questions and next steps

If genetic or molecular testing is recommended, you do not need to decide straight away. These are complex topics, and it is normal to have questions.

Support is available through:

- Your care team, who can explain why testing is being considered and how it may affect your treatment
- Genetic counsellors, who can help you understand the benefits and implications for you and your family
- Cancer support services, which may offer emotional support and practical advice

You may wish to ask:

- Why is this test being recommended for me?
- What could the results show, and how might they affect my care?
- Will my results be shared with anyone else?
- Should my family members also consider testing?
- What are my options if I choose not to proceed?

You are encouraged to ask questions and take the time you need. If you decide to go ahead, your care team will guide you through each step and help you understand what the results mean.

The APRISE pancreatic screening program

The Australian Pancreatic high-Risk ScrEening (APRISE) Program is a national research study led by the Jreissati Pancreatic Centre at Epworth in partnership with Pankind: Pancreatic Cancer Australia. The study is assessing whether regular screening can help detect pancreatic cancer earlier in people with a higher-than-average risk due to a strong family history of pancreatic cancer or genetic factors.

Screening in this study includes:

- A yearly MRI (magnetic resonance imaging) or EUS (endoscopic ultrasound) scan
- A brief yearly questionnaire about your health and medical history

To learn more about the APRISE Program, scan the QR code below.

Key takeaways

Genetic and molecular testing can offer important insights about pancreatic cancer and help guide treatment, clinical trial options, or planning for family members. Not everyone will need testing, but for some, it can play a key role in shaping personalised care.

If testing is offered, take your time, ask questions, and make a decision that feels right for you. Your care team and genetic counsellors are there to support you at each step.

Contact us

Jreissati Pancreatic Centre at Epworth

Phone 03 9426 8880
Email EH-PancreaticCentre@epworth.org.au

pancreaticcentre.org.au

Glossary

Biopsy: A procedure that removes a small sample of tissue to examine under a microscope or test for cancer-related changes.

BRCA1 / BRCA2: Genes that help repair DNA. Inherited changes in these genes can increase the risk of breast, ovarian, prostate and pancreatic cancers.

DNA: The material in your cells that carries genetic information. Changes in DNA can affect how cells grow and behave.

Familial cancer centre: A specialised service that provides support and advice for people with a personal or family history of inherited cancer.

Germline testing: A test that looks for inherited changes in your genes. These changes are present from birth and may be passed on to family members.

Gene mutation: A change in a gene that may affect how it works. Some mutations are inherited and others occur only in cancer cells.

Genetic counsellor: A trained health professional who helps you understand genetic test results and what they may mean for you and your family.

Genetic testing: A test that looks for inherited gene changes that may increase cancer risk.

Inherited mutation: A gene change passed down from a parent. These changes may increase the risk of certain cancers.

Molecular testing: A test that looks for gene changes in cancer cells. These changes are not inherited but may help guide treatment.

Multidisciplinary team: A group of health professionals who work together to plan and deliver your cancer care.

Pathogenic variant: A gene change known to increase the risk of cancer or affect treatment decisions.

Saliva test: A method for collecting DNA using a saliva sample. Often used for genetic testing.

Somatic mutation: A gene change found only in cancer cells. It is not inherited and cannot be passed to family members.

Targeted therapy: A treatment that uses drugs to target specific changes in cancer cells.

Tumour testing: A test that examines cancer tissue for gene changes. Also called molecular or somatic testing.

Variant of uncertain significance (VUS): A gene change that has been found, but it is not yet clear whether it increases cancer risk.

Additional resources

General

Cancer Australia

Cancer Australia is the Australian Government's national cancer control agency. The website explains how genetic and molecular testing can support cancer diagnosis, treatment planning and family care. It also provides information about inherited risk and screening.

www.canceraustralia.gov.au

Cancer Council Australia

Cancer Council offers plain-English resources on inherited cancer risk, genetic testing and what to consider before and after testing. It also provides telephone support, counselling and links to local services.

www.cancer.org.au/cancer-information

Centre for Genetics Education (NSW Health)

The Centre for Genetics Education is Australia's leading source of public information on genetic testing and hereditary cancer.

The website includes factsheets, videos and decision aids to help individuals and families understand their options.

www.genetics.edu.au

eviQ – Genetics and Familial Cancer Information Sheets

eviQ is a national cancer information resource widely used in Australian hospitals. It includes patient handouts on hereditary cancer syndromes, testing options and how different results may be used in care planning.

www.eviq.org.au

Inherited Cancers Australia

Inherited Cancers Australia is a national charity that supports people affected by inherited cancer risk. The website offers practical information, peer support, downloadable guides and tools to help individuals and families navigate testing and make informed decisions.

www.inheritedcancers.org.au

Financial and legal advice

Cancer Council's Financial Assistance Program

Provides advice and support on a variety of topics including legal issues, financial planning, small business accounting, workplace (HR or recruitment) advice.

13 11 20

cancer.org.au/support-and-services/practical-and-financial-assistance

Community Legal Centres Australia

Provides links to local community legal centres across Australia.

clcs.org.au/legal-help

Cancer Council's Dealing with Debt factsheet

Information for people who are having trouble paying their debts.

cancer.org.au/assets/pdf/dealing-with-debts

Services Australia's Financial Information Service

Information on employment, disability, sickness and carer payments.

132 850

servicesaustralia.gov.au

Type 'Financial Information Service' in the search box

Cancer Council's Help with Bills factsheet

Explains options to help you pay your utility bills, including rebates, discounts, and concessions

cancer.org.au/assets/pdf/help-with-bills-factsheet

Money Smart

Financial tips from the Australian Securities and Investments Commission (ASIC). Includes the Financial Advisers Register.

1300 300 630

moneysmart.gov.au

State Trustees

Public Trustees who can help you prepare a will and manage your finances. Contains links to public trustees by state and territory.

willshub.com.au/succession-law-resources/public-trustees/

Cancer Council's Superannuation and Cancer factsheet

Advice on finding and accessing super, tax implications of accessing super, and super-related insurance.

cancercouncil.com.au/wp-content/uploads/2022/09/Superannuation-and-Cancer.pdf

Counselling and mentoring services

Canteen

Free counselling and emotional support to young people and their families who are impacted by cancer.

1800 226 833

canteen.org.au

Care Search

Australian Government website that provides palliative care information and links to services for people affected by cancer.

caresearch.com.au

Kids Helpline

Telephone and online counselling service and crisis support for young people aged 5–25.

1800 55 1800

kidshelpline.com.au

Pancare support groups

Online and face-to-face support groups for patients and carers.

1300 881 698

pancare.org.au/living-well/support-groups/upper-gastrointestinal-cancer-support-groups

Pankind Support Navigator

(02) 7207 6970

support@pankind.org.au

Telephone support groups

The below telephone support groups are free, professionally facilitated support groups. They are open groups and you can join in whenever you feel well enough, on an ongoing basis, from anywhere in Australia.

Members are telephoned by Cancer Council so there are no costs to participate. You can be called on a landline or a mobile.

The Pancreatic Telephone Support Group meets the first and third Wednesdays of the month from 1.30pm–2.30pm (EST) and accepts new members at any time.

The Advanced Cancer Telephone Support Group meets the first and third Tuesday of the month from 1.30pm–2.30pm (EST) and is for people with a variety of cancers, including pancreatic cancer.

The Carers' Telephone Support Group meets the second and fourth Wednesdays of the month from 1.30pm–2.30pm (EST) and is for people caring for someone with any cancer, including pancreatic cancer.

To find out more about any of these telephone support groups, 1300 755 632 or email tsg@nswcc.org.au

Medicare Safety Nets thresholds

Medicare Safety Nets have thresholds. When you spend certain amounts in gap and out-of-pocket costs, you may reach these thresholds. Once you've reached the thresholds, you'll begin to receive higher Medicare benefits. This means you'll receive more money back for certain Medicare-funded services.

servicesaustralia.gov.au/what-are-medicare-safety-nets-thresholds

Please make sure you register so that your payments are counted towards the threshold.

Information for carers

Carer Gateway

Links carers to local support services such as emergency respite care, counselling and peer support groups. Offers practical information, resources and support packages for carers.

1800 422 737

carergateway.gov.au

Carers Australia

National body working with state and territory Carers Associations to provide information and counselling for carers.

1800 242 636

carersaustralia.com.au

Advance care planning

Advance Care Planning Australia

Information about planning for your future healthcare, including advance care directives.

1300 208 582

advancecareplanning.org.au

My Values

A set of specially constructed statements designed to help you identify, consider and communicate your wishes about the medical treatment you would want in the later stages of life.

myvalues.org.au

Helpful guides and factsheets

Cancer Council's Employment and Cancer factsheet

This fact sheet answers some common questions people have about their rights. It explains federal employment laws and protections under anti-discrimination laws.

cancer.org.au/assets/pdf/employment-and-cancer

Cancer Council's Getting your Affairs in Order factsheet

Instructions on making a will, preparing documents that will help others to make decisions for you if you're not able to make them yourself, nominating a beneficiary for your superannuation and insurance, and sorting out legal and financial paperwork.

cancer.org.au/assets/pdf/getting-your-affairs-in-order-factsheet

My Aged Care

Established by the Australian Government to help you find and access the right government-funded aged care services.

You can call the My Aged Care Contact Centre on 1800 200 422.

Monday to Friday 8am to 8pm and Saturdays 10am to 2pm

myagedcare.gov.au

Notes

References

1. Cancer Australia, *Pancreatic cancer in Australia statistics*, Cancer Australia, 2025, accessed 1 December 2025.
2. OO Faluyi, JL Connor, M Chatterjee, C Ikin, H Wong and DH Palmer, 'Advanced pancreatic adenocarcinoma outcomes with transition from devolved to centralised care in a regional Cancer Centre', British Journal of Cancer, 2017, 116:424-431.

Epworth Medical Foundation

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epworth.org.au/giving

Contact us

Jreissati Pancreatic Centre at Epworth

Phone 03 9426 8880
Email EH-PancreaticCentre@epworth.org.au

pancreaticcentre.org.au

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CONSUMER REVIEWED

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