



**SONIC
GENETICS**

Quality is in our DNA

Genetic Testing Guide

ACT

Capital Pathology

NSW

Douglass Hanly Moir Pathology
Barratt & Smith Pathology
Capital Pathology
Southern IML Pathology
Sullivan Nicolaides Pathology

QUEENSLAND / NORTHERN TERRITORY

Sullivan Nicolaides Pathology

SOUTH AUSTRALIA

Clinpath Laboratories

TASMANIA

Hobart Pathology
Launceston Pathology
North West Pathology

VICTORIA

Melbourne Pathology

WESTERN AUSTRALIA

Clinipath Pathology



Genetic testing

Genetic testing is changing the face of modern pathology. Rapid technological advances continue to impact all areas of clinical medicine, creating new opportunities to identify genetic disorders in infancy, and to confirm diagnoses that were not previously possible. Other genetic tests can be used to predict the clinical safety and efficacy of medications, or to guide best treatment for some cancers. Genetic testing can also be used to define the risk of certain diseases in members of families with serious genetic disorders, potentially allowing earlier intervention for those at risk.

Sonic Healthcare is at the forefront of this emerging field. With specialised genetic laboratories across Australia, UK, Europe and USA, Sonic Healthcare is committed to providing a comprehensive genetic testing service to support the optimal management of your patients.

Sonic Genetics

Sonic Genetics brings together the extensive range of genetic tests performed by Sonic Healthcare's specialised genetic laboratories, under the guidance and leadership of a group of eminent genetic pathologists. Through Sonic Genetics, Sonic Healthcare is able to provide a comprehensive genetic service for specialists and counsellors. With worldwide resources, Sonic Genetics offers a broad range of genetic tests, performed both in Sonic Healthcare laboratories as well as high-quality fully accredited referral laboratories.

Sonic Genetics tests – Australia

This brochure provides a guide to the current range of tests performed in Sonic's Australian laboratories. It does not include tests performed in our overseas laboratories. A full listing of genetic tests provided by Sonic Healthcare laboratories worldwide is available online at www.sonicgenetics.com.au

Sonic Genetics referral service

In order to provide a fully comprehensive genetic testing service and in addition to the wide range of tests performed in Sonic Healthcare laboratories, Sonic Genetics has established a referral service for highly esoteric tests, where volumes may be very low. These esoteric genetic tests are referred to accredited quality laboratories with specialised experience. Sonic Genetics is able to advise on quality laboratories and to arrange collection, transport, tracking and results delivery for these very rare tests. Please contact our genetic referral service for further information, on 1800 010 447 or at info@sonicgenetics.com.au

Ordering genetic tests

Routine genetic tests may be requested using Sonic Healthcare local laboratory referral forms – Sullivan Nicolaides Pathology, Douglass Hanly Moir Pathology and Barratt & Smith Pathology, Melbourne Pathology, Clinipath Pathology, Clinpath Laboratories, Hobart Pathology, Launceston Pathology, North West Pathology, Capital Pathology and Southern IML Pathology. Many genetic tests require patient counselling and consent, and some tests also require detailed patient information and/or special request forms. For more information about ordering genetic tests, please phone 1800 010 447 or email info@sonicgenetics.com.au

Cost of genetic tests

Genetic testing is a highly specialised field of medicine. Most genetic tests are extremely comprehensive and require significant involvement and interpretation from pathologists and scientists. They also rely on expensive new technology.

The costs for genetic tests are frequently not covered by Medicare or private health insurance, so patients may incur non-rebatable private fees for some tests.

Some tests attract a full or partial Medicare rebate, and these have been denoted with a hash (#) in the test listings. Clinical notes for all genetic tests are extremely important, especially for tests that only attract a Medicare rebate when certain criteria are met. Please ensure that clinical notes are included on all request forms, and that all criteria are clearly stated.

For further information on the costs of genetic tests, please contact your local laboratory or Sonic Genetics – phone 1800 010 447 or email info@sonicgenetics.com.au. Information is also available on our website www.sonicgenetics.com.au

Expert team

Sonic Genetics' expert team in Australia is led by Professor Graeme Suthers, together with genetic pathologists Dr James Harraway and Dr Scott Mead. Along with Dr Karl Baumgart and Dr Melanie Galea, they are available for advice and consultation, as well as recommendations on appropriate genetic counselling, which forms an integral part of genetic investigations. Our medical team is ably supported by senior scientists who specialise in different areas of genetic testing.

Internationally, Sonic Genetics harnesses the expertise of pathologists and scientists in laboratories in Germany, UK and USA. Professor Carsten Bergman (Germany) and Dr Lisa Levett (UK) add particular areas of expertise.

This global network allows us to provide referring clinicians with a comprehensive range of testing, including many esoteric tests not currently available in Australia.



Introducing Sonic Genetics

**Professor Graeme Suthers**

Director – Genetics, Sonic Healthcare (Australia)
BSc (Med), MBBS, PhD, FRACP, FRCPA, GAICD

Sonic Genetics is headed by Professor Graeme Suthers. A clinical geneticist and a genetic pathologist, Professor Suthers is one of Australia's most respected experts in the field of genetics. With an outstanding career and deep experience in genetics, healthcare and research, Graeme is nationally and internationally recognised for his expertise in genetic disorders, testing and clinical service provision. He has been closely involved in the development of software solutions for specialty genetic services and in the evaluation of healthcare interventions to promote genetic services of the highest quality and utility to patients, families and managing clinicians.

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**Dr James Harraway**

MB ChB, FRCPA, DPhil

Dr James Harraway completed his medical training in Christchurch, New Zealand. In 2005, James was awarded a Nuffield Medical Fellowship to undertake a DPhil at Oxford University, examining the molecular pathogenesis of Cockayne Syndrome. In 2008, he became the first pathologist from New Zealand to obtain a FRCPA in genetic pathology. Dr Harraway has an interest in both inherited and somatic (oncology) genetic testing.

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**Dr Scott Mead**

PhD, MB ChB, FRCPA (Genetics)

Dr Scott Mead graduated from the University of Auckland (New Zealand), where he also gained a PhD in Molecular Medicine. His genetic pathology training was undertaken at Christchurch School of Medicine (University of Otago, NZ) and the Royal Prince Alfred Hospital (Sydney). Dr Mead has a long-standing research interest in cancer genetics, which has included study in neuroblastoma at Great Ormond Street Hospital (UK), leukaemia at the University of Auckland (NZ) and the clinical application of next generation sequencing in pancreatic cancer at the Kinghorn Cancer Centre (Sydney). Dr Mead is a Conjoint Senior Lecturer at the School of Medical Sciences, University of NSW.

P: 02 9855 6276

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**Dr Karl Baumgart**

BSc (Med), MBBS, PhD, FRACP, FRCPA

Dr Karl Baumgart works partly in clinical practice in clinical immunology and allergy. His laboratory interests include paternity testing, tissue typing, pharmacogenomics and periodic fever syndromes.

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**Dr Melanie Galea**

FRCPA (Genetics), MBBS (Hons), BMedSc (Hons)

Dr Melanie Galea graduated in Medicine with Honours at the University of Sydney (Australia), where she had previously gained an Honours degree in Medical Science, specialising in molecular genetics. Her genetic pathology training involved exposure to a broad range of molecular and cytogenetic techniques in Australian diagnostic laboratories, including Prince of Wales Hospital (Sydney), Royal Brisbane & Women's Hospital (Brisbane) and Westmead Hospital (Sydney). Dr Galea has an interest in both constitutional and somatic genetics, medical education and quality assurance.

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DIAGNOSTIC TESTS

| Name of test | Disease/Syndrome/Indication | Test method or gene/s analysed |
|--|--|---|
| 1p36 FISH [#] | Diagnosis of a microdeletion syndrome | Genes at chromosome 1q36 |
| 22q11.2 FISH [#] | Diagnosis of DiGeorge syndrome | Genes at chromosome 22q11 |
| Alpha thalassaemia screen | Identifying carriers of alpha thalassaemia | Common mutations of the HBA1 and HBA2 genes |
| Angelman syndrome FISH [#] | Diagnosis of Angelman syndrome | Genes at chromosome 15q11.2 |
| APOE screen | Hyperlipoproteinaemia | Specific variants of the APOE gene |
| CFTR screen (cystic fibrosis) | Cystic fibrosis diagnosis and carrier status | Common mutations in CFTR gene |
| Coeliac disease HLA typing [#] | Exclusion of coeliac disease | Specific variants of the HLA-DQ and DR genes |
| Cri du chat syndrome FISH [#] | Diagnosis of a microdeletion syndrome | Genes at chromosome 5p15 |
| Factor V Leiden [#] | Deep vein thrombosis, APC resistance | Specific mutation of the F5 gene |
| Familial Hibernian Fever | TNF-receptor associated periodic syndrome (TRAPS) | TNFRSF1A gene |
| Familial Mediterranean Fever | Periodic peritonitis, benign paroxysmal peritonitis | MEFV gene |
| Fragile X syndrome [#] | Diagnosis of the fragile X syndrome (autism, developmental delay) and carrier status | FRAXA gene |
| Gilbert syndrome | Diagnosis of benign hyperbilirubinaemia | Specific variants of the UGT1A1 gene |
| Haemochromatosis type 1 [#] | Persistent elevation of ferritin, or evidence of iron overload | Specific variants of the HFE gene |
| Hereditary angioedema (I & II) | Hereditary angioedema type I and type II | SERPING1 gene |
| Hereditary angioedema type III | Hereditary angioedema type III | F12 gene |
| HLA-B27 [#] | Ankylosing spondylitis | Specific variants of the HLA-B gene |
| HLA-B51 | Behcet disease | Specific variants of the HLA-B gene |
| HLA-DR, HLA-DQ | Sjögren syndrome susceptibility, narcolepsy, rheumatoid arthritis | Specific variants of various HLA genes |
| KAL1 FISH, FGFR1 FISH [#] | Diagnosis of a microdeletion syndrome | Deletions of the KAL1 and FGFR1 genes |
| Karyotype (blood) [#] | Multiple indications | Number and structure of all chromosomes |
| Karyotype with rapid FISH [#] | Prenatal screen on amniotic fluid, CVS, or fetal blood | Aneuploidies of chromosomes 13, 18, 21, X, and Y and chromosome studies |
| Microarray [#] | Developmental delay, autism, or multiple congenital malformations | Sub-microscopic deletions and duplications of all chromosomes |
| Miller-Dieker syndrome FISH [#] | Diagnosis of a microdeletion syndrome | LIS1 gene deletion |
| Prader-Willi syndrome FISH [#] | Diagnosis of a microdeletion syndrome | Genes at chromosome 15q |
| Prothrombin gene [#] | Deep vein thrombosis | Specific variant of the F2 gene |
| SHOX FISH [#] | Diagnosis of a microdeletion syndrome | SHOX gene deletion |
| Smith-Magenis syndrome FISH [#] | Diagnosis of a microdeletion syndrome | Genes at chromosome 17p11 |
| Sotos syndrome FISH [#] | Diagnosis of a microdeletion syndrome | NSD1 gene deletion |
| Williams syndrome FISH [#] | Diagnosis of a microdeletion syndrome | ELN gene deletion |
| Wolf-Hirschhorn syndrome FISH [#] | Diagnosis of a microdeletion syndrome | Genes at chromosome 4p |

[#] Partial or full rebate by Medicare (subject to MBS criteria being met)

FISH – fluorescent *in situ* hybridisation

Aneuploidy – abnormal number of specific chromosome

PCR – polymerase chain reaction

ONCOLOGY TESTS

| Name of test | Disease/Syndrome/Indication | Test method or gene/s analysed |
|---------------------------------|---|---|
| LEUKAEMIA / LYMPHOMA | | |
| 13q FISH | Mature B-cell lymphoid neoplasms | Genes at chromosome 13q |
| 20q FISH | Myeloid neoplasms | Genes at chromosome 20q |
| 5q FISH | Myeloid neoplasms | Genes at chromosome 5q |
| 6q21 FISH | Mature B-cell neoplasms | Genes at chromosome 6q21 |
| 7q FISH | Myeloid neoplasms | Genes at chromosome 7q |
| ALK FISH (lymphoma) | Lymphoma | ALK gene fusions |
| ATM FISH | Chronic lymphocytic leukaemia | ATM gene deletion |
| BCL6 FISH | B-cell non-Hodgkin lymphomas | BCL6 gene fusions |
| BCR/ABL1 FISH [#] | Leukaemias | BCR/ABL1 gene fusion |
| BCR/ABL1 PCR [#] | Leukaemias | BCR/ABL1 gene fusion |
| CBFB/MYH11 FISH [#] | Acute myeloid leukaemia | CBFB/MYH11 gene fusion |
| Chimerism FISH | Post-bone marrow transplantation | Presence of X and Y chromosomes |
| Chronic lymphocytic leuk FISH | Chronic lymphocytic leukaemia | Numerous genes |
| Eosinophilia FISH panel | Eosinophilic neoplasms | Numerous genes |
| ETV6/RUNX1 FISH | Acute lymphoblastic leukaemia | ETV6/RUNX1 (i.e. TEL/AML1) gene fusion |
| FGFR1 FISH | Eosinophilic neoplasms | FGFR1 gene fusions |
| IGH/BCL2 FISH | Lymphoma | BCL2 gene fusions |
| IGH/CCND1 FISH | Mantle cell lymphoma, myeloma | IGH/CCND1 gene fusion |
| IGH/MAF FISH | Multiple myeloma | IGH/MAF gene fusion |
| IGH/MALT1 FISH | Lymphoma | IGH/MALT1 gene fusions |
| IGH/MYC FISH | Lymphoma | IGH/MYC gene fusion |
| JAK2 screen [#] | Myeloid neoplasms | Specific mutations of the JAK2 gene |
| Karyotype (tumour) [#] | Haematological malignancy | Number and structure of all chromosomes |
| KMT2A FISH [#] | Acute leukaemia | KMT2A (i.e. MLL) gene fusions |
| Lymphoma FISH panel 1 | Lymphoma | Various genes |
| Lymphoma FISH panel 2 | Lymphoma | Various genes |
| MLLT3/KMT2A FISH [#] | Acute myeloid leukaemia | MLLT3/KMT2A (i.e. MLLT3/MLL) gene fusion |
| Multiple hit lymphoma FISH | B-cell lymphoma | Various genes |
| Multiple myeloma panel | Multiple myeloma | Various genes |
| MYC FISH | Non-Hodgkin lymphomas | MYC gene fusion |
| Myelodysplastic syndrome FISH | Myelodysplastic syndrome | Various genes |
| PDGFRA/FIP1L1 FISH [#] | Myeloid and lymphoid neoplasms | PDGFRA/FIP1L1 gene fusion |
| PDGFRB FISH | Myeloid neoplasms | PDGFRB gene fusions |
| PML/RARA FISH [#] | Acute promyelocytic leukaemia | PML/RARA gene fusions |
| RPN1/MECOM FISH [#] | Acute myeloid leukaemia, myelodysplastic syndrome | RPN1/MECOM (i.e. RPN1/EVI1) gene fusion |
| RUNX1/RUNX1T1 FISH [#] | Acute myeloid leukaemia | RUNX1/RUNX1T1 (i.e. AML1/ETO) gene fusion |

ONCOLOGY TESTS

| Name of test | Disease/Syndrome/Indication | Test method or gene/s analysed |
|-----------------------------------|---|---|
| LEUKAEMIA / LYMPHOMA | | |
| TCR & IGH rearrangements | Lymphocyte proliferation | Rearrangements of TCR and IGH genes |
| TP53 FISH | Chronic lymphocytic leukaemia | TP53 gene deletions |
| Trisomy 12 FISH | Chronic lymphocytic leukaemia | Genes on chromosome 12 |
| Trisomy 8 FISH | Acute myeloid leukaemia, myelodysplastic syndrome | Genes on chromosome 8 |
| SOLID TUMOURS | | |
| ALK FISH (lung cancer) | Non-small cell lung cancer | ALK gene fusions |
| BRAF screen [#] | Melanoma, early-onset colorectal cancer | Specific mutation of the BRAF gene |
| EGFR FISH | Glial tumours | Amplification of the EGFR gene |
| EGFR screen [#] | Non-small cell lung cancer | Specific mutations of the EGFR gene |
| Ewing sarcoma FISH | Ewing sarcoma | EWSR1 gene |
| FKHR FISH | Alveolar rhabdomyosarcoma | FKHR (i.e. FOXO1) gene fusions |
| FUS/DDIT3 FISH | Liposarcoma | FUS/DDIT3 gene fusion |
| KRAS and NRAS screen [#] | Colorectal cancer | Specific mutations of the KRAS and NRAS genes |
| MDM2 FISH | Various tumours | MDM2 gene amplification |
| MYCN FISH | Various tumours | MYCN (i.e. N-MYC) gene amplification |
| PTEN FISH | Gliomas | PTEN gene deletion |
| SS18 FISH | Sarcoma | SS18 gene deletion |
| t(1;19) FISH | Brain tumours | Genes on 1p and 19q |
| USP6 FISH | Bone tumours | USP6 gene rearrangements |

PHARMACOGENETIC TESTS

| Name of test | Medication/Indication | Test method or gene/s analysed |
|------------------------|--|----------------------------------|
| CYP2C19 | Clopidogrel, tricyclic antidepressants, proton pump inhibitors | CYP2C19 gene |
| CYP2C9 | Tricyclic antidepressants | CYP2C9 gene |
| CYP2C9, VKORC1 | Warfarin | CYP2C9 and VKORC1 genes |
| CYP2D6 | Tamoxifen, codeine and other medications | CYP2D6 gene |
| CYP2D6, CYP3A4, CYP3A5 | Fentanyl, methadone and other medications | CYP2D6, CYP3A4, and CYP3A5 genes |
| DPYD | 5-fluorouracil, capecitabine, tegafur | DPYD gene |
| HLA-B1502 [#] | Carbamazepine hypersensitivity | HLA-B1502 analysis |
| HLA-B5701 [#] | Abacavir hypersensitivity | HLA-B5701 analysis |
| HLA-B5801 | Allopurinol hypersensitivity | HLA-B5801 analysis |
| TPMT [#] | Azathioprine and 6-mercaptopurine | TPMT gene |
| UGT1A1 screen | Irinotecan | UGT1A1 gene |

RELATIONSHIP TESTS

| Name of test | Indication | Test method or gene/s analysed |
|-------------------------------|---------------------------|--|
| Immigration relationship test | Relationship confirmation | Various markers on different chromosomes |
| Prenatal paternity | Relationship confirmation | Various markers on different chromosomes |
| Relationship testing | Relationship confirmation | Various markers on different chromosomes |
| Specimen matching | Relationship confirmation | Various markers on different chromosomes |

REPRODUCTIVE TESTS

| Name of test | Disease/Syndrome/Indication | Test method or gene/s analysed |
|---|---|---|
| CFTR screen (CBAVD) | Male infertility (congenital absence of the vas deferens) | Common mutations of the CFTR gene |
| DAZ deletion PCR | Male infertility | DAZ gene |
| Fragile X (reproductive) [#] | Premature ovarian failure | FRAXA gene |
| Karyotype (prenatal) [#] | Various indications | Number and structure of all chromosomes |
| Karyotype (prod of conception) [#] | Miscarriage | Number and structure of all chromosomes |
| Karyotype (reproductive) [#] | Recurrent miscarriages, infertility | Number and structure of all chromosomes |
| Non-invasive prenatal test | Prenatal screening for major chromosome abnormalities of fetus | Analysis of cell-free DNA in maternal blood for aneuploidies of chromosomes 13, 18, 21, X and Y |
| Rapid interphase FISH [#] | Various indications for urgent prenatal assessment of fetal chromosomes | Aneuploidies of chromosomes 13, 18, 21, X and Y and chromosome studies |
| Single cell FISH | IVF pre-implantation diagnosis of selected aneuploidies | Aneuploidies of chromosomes 13, 16, 18, 21,22, X and Y |
| SRY FISH | Sex reversal or ambiguous genitalia | Male determining factor of the Y chromosome |
| Tissue FISH studies [#] | Miscarriage | Aneuploidies of chromosomes 13, 16, 18, 21,22, X and Y |
| Y chromosome FISH | Y chromosome variant analysis | Genes on the Y chromosome |

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FISH – fluorescent *in situ* hybridisation

Aneuploidy – abnormal number of specific chromosome

PCR – polymerase chain reaction

For further information about ordering genetic tests, including collection requirements and costs, please contact your local laboratory or Sonic Genetics.
Phone 1800 010 447 or Email info@sonicgenetics.com.au

Sonic Healthcare

Sonic Healthcare is one of the world's largest medical diagnostic companies, providing comprehensive laboratory and radiology services to medical practitioners, hospitals, community health services, governments and industries. Our expertise in diagnostic medicine has led to a deeper involvement in other healthcare services, including general practice, occupational medicine, community nursing services and clinical trials.

Sonic Healthcare's success is underpinned by our commitment to medical sovereignty and independence. All healthcare divisions are medically-led and dedicated to providing outstanding quality and service to the doctors and patients that they serve.

Sonic Healthcare is an Australian company listed on the Australian Securities Exchange (ASX). We employ more than 27,000 people globally, including Australia, New Zealand, the United Kingdom, Germany, Switzerland, the USA, Belgium and Ireland.



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