Sonic Genetics

Quality is in our DNA

Your comprehensive genetic testing service

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 and is committed to providing a comprehensive genetic testing service to support the optimal management of your patients.

Sonic Genetics offers a broad range of genetic tests, performed both in Sonic Healthcare pathology laboratories, as well as high-quality fully accredited referral laboratories.

This brochure details our current range, and with the ever-increasing expansion of genetics and genomics across all facets of medicine, we are constantly reviewing available test options and building on our assay menu.

Arranging genetic testing

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

Clinical notes

Clinical notes for all genetic tests are important, as the interpretation of a genetic result may be determined by the clinical context.

Costs

Genetic testing is a highly specialised field of medicine, requiring significant involvement and interpretation from pathologists and scientists. Genetic tests are frequently not covered by Medicare or private health insurance, so patients may incur non-rebatable private fees.

Clinical notes should be included, especially for tests that only attract a Medicare rebate when certain criteria are met.

Please refer to the Sonic Genetics website, www.sonicgenetics.com.au, for current pricing.

DNA relationship

NAME OF TEST	TEST ALIAS	GENES
Relatedness		
DNA matching	Tissue specimen matching	
Immigration relationship test	Immigration DNA testing	
Prenatal paternity	Paternity of unborn child	
Relationship testing	Paternity/maternity/other relationships	

Familial

NAME OF TEST	TEST ALIAS	GENES
Cardiac		
Aortopathy panel	Thoracic aortic aneurysm, Lois-Dietz syndrome, Marfan syndrome, Ehlers-Danlos syndrome type 4, EDS type 4	For details, please refer to the Sonic Genetics website, www.sonicgenetics.com.au/cardiac-genetics
Arrhythmogenic right ventricular cardiomyopathy panel	ARVC, ARVD, arrhythmogenic right ventricular dysplasia	
Bicuspid aortic valve	BAV	
Brugada syndrome panel	Sudden unexplained death syndrome, Pokkuri death syndrome, idiopathic ventricular fibrillation	
Dilated cardiomyopathy panel	Dilated cardiomyopathy, DCM	
Hypertrophic cardiomyopathy panel	Hypertrophic cardiomyopathy, HCM, HOCM	
Left ventricular non-compaction panel	Isolated ventricular non-compaction, LVNC	

NAME OF TEST	TEST ALIAS	GENES
Long QTS panel	LQT syndrome, LQTS, Romano-Ward syndrome, Jervell Lange-Nielsen syndrome	For details, please refer to the Sonic Genetics website, www.sonicgenetics.com.au/cardiac-genetics
Pulmonary hypertension panel	Pulmonary veno-occlusive disease, hereditary haemorrhagic telangiectasia	
Vasculopathy panel	Hereditary haemorrhagic telangiectasia, aortic valve disease, polycystic kidney disease, tortuosity of retinal arteries, Adams-Oliver syndrome, cutis laxa, arterial tortuosity syndrome	
Cancer		
Familial breast/ovarian cancer	BRCA1, BRCA2, familial breast cancer, BRCA, familial ovarian cancer, serous cancer	BRCA1, BRCA2, PALB2
Gastrointestinal		
Coeliac disease HLA typing	Coeliac tissue typing, variants of HLA-DQ and DR	HLA-DQA1, HLA-DQB1
Lactose intolerance	Lactose test, lactase metabolism, lactase persistence	LCT, MCM6
Hepatic		
Haemochromatosis type 1	HFE, HH, hereditary haemochromatosis, bronzed cirrhosis, iron storage disorder	HFE
UGT1A1 screen	Gilbert syndrome, irinotecan	UGT1A1
Haematological		
Alpha thalassaemia screen	α-thalassaemia, mutations of HBA1 and HBA2	HBA1, HBA2
Factor V Leiden	Factor 5 gene, thrombophilia	F5
MTHFR screen	5,10-methylenetetrahydrofolate reductase	MTHFR
Prothrombin gene	Prothrombin gene mutation, F2, thrombophilia	F2
Hyperlipidaemia		
APOE screen	Apolipoprotein E genotyping, hyperlipoproteinaemia	APOE
Hypercholesterolaemia panel	Familial hypercholesterolaemia, familial combined hyperlipidaemia, sitosterolaemia	
Hypertriglyceridaemia panel	Familial hypertriglyceridaemia	
Immunological		
Hereditary angioedema (I & II)	HAE type I, HAE type II, SERPING1	SERPING1
Periodic fever		
FMF screen	Familial Mediterranean fever, MEFV	MEFV
TRAPS screen	Familial Hibernian fever, TNF-receptor associated periodic syndrome, TNFRSF1A	TNFRSF1A
Neurological		
Fragile X syndrome	FMR1, FRAXA, FXS, Marker X syndrome, Martin-Bell syndrome, X-linked mental retardation	FMR1, FRAXA
Respiratory		
CFTR mutation panel	Cystic fibrosis, cystic fibrosis-related disease, congenital bilateral absence of vas deferens (CBAVD)	CFTR

Immunology

NAME OF TEST	TEST ALIAS	GENES
Angioedema		
Hereditary angioedema (I & II)	HAE type I, HAE type II, SERPING1	SERPING1
Autoimmune		
HLA panel screen (DR and DQ)	HLA-DR, HLA-DQ, Sjögren syndrome susceptibility, narcolepsy, rheumatoid arthritis	HLA-DR, HLA-DQ
HLA-B27	B27, human leucocyte antigen B, ankylosing spondylitis	HLA-B
HLA-B51	B51, human leucocyte antigen B, Behçet disease	HLA-B
Gastrointestinal		
Coeliac disease HLA typing	Coeliac tissue typing, variants of HLA-DQ and DR	HLA-DQA1, HLA-DQB1
HLA-B*15:02	B*15:02 , human leucocyte antigen B, carbamazepine hypersensitivity	HLA-B
HLA-B*57:01	B*57:01 , human leucocyte antigen B, abacavir hypersensitivity	HLA-B
HLA-B*58:01	B*58:01, human leucocyte antigen B, allopurinol hypersensitivity	HLA-B
Periodic fever		
FMF screen	Familial Mediterranean fever, MEFV	MEFV
TRAPS screen	Familial Hibernian fever, TNF-receptor associated periodic syndrome, TNFRSF1A	TNFRSF1A

Oncology

NAME OF TEST	TEST ALIAS	GENES
HAEMATOLOGICAL MALIGNANCIES		
Chimerism		
Sex mismatch FISH	Sex mismatch FISH, chimerism FISH	
Myeloid neoplasms - Karyotype and panels		
Karyotype (leukaemia/lymphoma)	Chromosome studies, karyotype (oncology)	
Acute myeloid leukaemia FISH panel	AML FISH	CBFB, DEK, ETV6, EVI1, KMT2A, MYH11, NUP214, PML, RARA, RUNX1, RUNX1T1
Myeloid/lymphoid neoplasms with eosinophilia FISH panel	PDGFRA, PDGFRB, FGFR1	
Myelodysplastic syndrome FISH	Deletions in the long arms of chromosomes 5, 7 and 20	
Myeloid neoplasms - Targeted assays		
Myeloid leukaemias		
BCR/ABL1 FISH	BCR-ABL FISH, t(9;22) FISH	BCR, ABL1
BCR/ABL1 PCR	t(9;22)PCR, BCR-ABL PCR	BCR, ABL1
CBFB/MYH11 FISH	inv(16) FISH, AML	CBFB, MYH11
DEK/NUP FISH	t(6;9) FISH	DEK, NUP
KMT2A FISH	MLL FISH	KMT2A
MLLT3/KMT2A FISH	t(9;11) FISH, MLLT3-MLL FISH	MLLT3, KMT2A
PML/RARA FISH	t(15;17) FISH	PML, RARA
RPN1/MECOM FISH	RPN1-EVI1 FISH, inv(3) FISH	
RUNX1/RUNX1T1 FISH	t(8;21) FISH, AML1/ETO FISH	RUNX1, RUNX1T1
Trisomy 8 FISH	Tri(8) FISH	
Myelodysplastic disorders		
5q deletion FISH	Myelodysplastic syndrome associated with isolated deletion (5q) chromosome abnormality, myeloid neoplasms	NPM1, APC, CTNNA1
7q deletion FISH	Myelodysplasia and leukaemia syndrome with monosomy 7, MLSM7, myeloid neoplasms	
20q deletion FISH	Deletion of part of the long arm of chromosome 20 (20q), myeloid neoplasms	
RPN1/MECOM FISH	RPN1-EVI1 FISH, inv(3) FISH	
Trisomy 8 FISH	Tri(8) FISH	
Myeloproliferative disorders and myeloid/ lymphoid neoplasms with eosinophilia		
JAK2 screen	JAK2 gene test, myeloid neoplasms	JAK2 V617F
CALR screen	CALR gene test, calreticulin gene test	CALR
MPL screen	MPL gene test	MPL
FGFR1 FISH	Fibroblast growth factor receptor 1	FGFR1
PDGFRA/FIP1L1 FISH	PDGFRA FISH, FIP1L1-PDGFRA FISH	PDGFRA, FIP1L1
PDGFRB FISH	5q33 FISH, PDGFRB FISH	PDGFRB
Lymphoid neoplasms - Karyotype and panels		
Karyotype (leukaemia/lymphoma)	Chromosome studies, karyotype (oncology)	
Chronic lymphocytic leukaemia FISH panel	CLL FISH	
Lymphoma panel	ATM deletion, 13q deletion, TP53 deletion, MALT1 rearrangements and IGH rearrangements	
Multiple hit lymphoma FISH	Double/triple hit lymphoma panel	
Myeloid/lymphoid neoplasms with eosinophilia FISH panel	PDGFRA, PDGFRB, FGFR1	
Myeloma panel	IGH rearrangement, TP53 deletion, 13q deletion and 1q trisomy	
TCR/IGH rearrangements	T-cell & B-cell gene rearrangement PCR	TCR, IGH
Lymphoid neoplasms - Targeted assays		
Lymphocytic and lymphoblastic leukaemias		
ATM deletion FISH	Deletion of the ATM gene at 11q22, CLL	ATM
CDKN2A FISH	Cyclin-dependent kinase inhibitor 2A	CDKN2A
ETV6/RUNX1 FISH	TEL/AML1 FISH, t(12;21) FISH, ALL	ETV6, RUNX1
KMT2A FISH	MLL FISH	KMT2A
TP53 FISH	p53 FISH, 17p deletion FISH	TP53
Trisomy 12 FISH	Tri(12) FISH	
13q deletion FISH	13q FISH, mature B-cell lymphoid neoplasms	

NAME OF TEST	TEST ALIAS	GENES
Lymphoid neoplasms - Targeted assays		
Lymphomas		
6q21 deletion FISH	Deletion affecting the long arm of chromosome 6, mature B-cell lymphoid neoplasm	
13q deletion FISH	13q FISH, mature B-cell lymphoid neoplasms	
ALK FISH (lymphoma)	ALCL FISH (lymphoma), ALK gene fusions	ALK
BCL6 FISH	3q27 FISH, B-cell non-Hodgkin lymphomas	BCL6
CCND2 FISH	B-cell lymphoma	CCND2
IGH/BCL2 FISH	t(14;18) FISH	IGH, BCL2
IGH/CCND1 FISH	t(11;14) FISH, Mantle cell lymphoma FISH	IGH, CCND1
IGH/MALT1 FISH	MALT FISH, t(11;18) FISH	IGH, MALT1
IGH/MYC FISH	t(8;14) FISH	IGH, MYC
IGK, IGL FISH	IGKC FISH, IGLC1 FISH	IGKC, IGLC1
IRF4/DUSP22 FISH	Interferon regulatory factor 4, dual-specificity phosphatase 22	IRF4/DUSP22
MYC FISH	8q24 FISH, c-myc FISH	MYC
TP63 FISH	Anaplastic large cell lymphoma FISH, ALCL FISH	TP63
SOLID MALIGNANCIES		
Brain cancer		
EGFR FISH	EGFR amplification FISH, glial tumours	EGFR
Glioma FISH panel	1p/19q co-deletion FISH, brain tumours	EGFR, 1/19q co-deletion
PTEN FISH	PTEN deletion FISH, gliomas	PTEN
Breast cancer		
Breast cancer gene expression prognostic assay	Prosigna®, PAM50	
Colorectal cancer		
Colorectal cancer focused gene panel	Colorectal panel, KRAS, NRAS, BRAF, PIK3CA	KRAS, NRAS, BRAF, PIK3CA
KRAS and NRAS screen	RAS panel, KRAS and NRAS gene test, colorectal cancer	KRAS, NRAS
Lung cancer		
ALK screen (lung cancer)	ALK IHC; ALK FISH (lung cancer), non-small cell lung cancer	ALK
EGFR screen	EGFR gene test, non-small cell lung cancer	EGFR
Lung cancer focused gene panel	Lung panel	ALK, BRAF, EGFR, ERBB2 (HER2),
KRAS, ROS1		
ROS1 FISH	ROS1 IHC, ROS1 FISH (lung cancer), non-small cell lung cancer	ROS1
Melanoma		
BRAF V600 screen	BRAF gene test, melanoma, colorectal cancer	BRAF
Melanoma focused gene panel	Melanoma panel	BRAF, KIT , NRAS
Neuroblastoma		
MYCN FISH	N-MYC FISH, 2p23 FISH	MYCN
Sarcoma		
CIC DUX4 FISH	t(4;19) FISH, Ewing-like sarcoma FISH, undifferentiated small blue round cell sarcoma FISH, primitive round cell sarcoma FISH	CIC, DUX4
Dermatofibrosarcoma protuberans (DFSP)	COL1A/PDGFB FISH	COL1A, PDGFB
Ewing sarcoma FISH	Ewing FISH, t(11;22) FISH, primitive neurectodermal tumour, PNET, clear cell sarcoma FISH, desmoplastic small round cell tumour FISH, extraskeletal myxoid sarcoma FISH	EWSR1
EWSR1/FLI1 FISH	Ewing FISH, t(11;22) FISH, primitive neurectodermal tumour, PNET	EWSR1, FLI1
EWSR1/WT1 FISH	Desmoplastic small round cell tumour FISH	EWSR1, WT1
FOXO1 FISH	FKHR FISH, alveolar rhabdomyosarcoma FISH	FOXO1
FOXO1/PAX3 FISH	FKHR FISH, alveolar rhabdomyosarcoma FISH	FOXO1/PAX3
FOXO1/PAX7 FISH	FKHR FISH, alveolar rhabdomyosarcoma FISH	FOXO1/PAX7
FUS/DDIT3 FISH	t(12;16) FISH, liposarcoma	FUS, DDIT3
MDM2 FISH	MDM2 amplification	MDM2
Myxoid liposarcoma FISH	t(12;16) FISH, CHOP-FUS FISH, FUS-DDIT3 FISH	FUS, DDIT3 (formally known as CHOP)
SS18 FISH	t(X;18) FISH	SS18
USP6 FISH	USP6 17p13 FISH, bone tumours	USP6
Solid tumour		
Karyotype (tumour)	Chromosome studies, karyotype (oncology)	
FIND IT® panel	FIND IT cancer hotspot panel, full FIND IT panel, NSCLC/melanoma/colorectal focused panel	≥30 genes
Renal cancer		
TFE3 FISH	Papillary renal cell carcinoma FISH, alveolar soft part sarcoma FISH	TFE3

Paediatric

NAME OF TEST	TEST ALIAS	GENES
Congenital disorder		
1p36 FISH	Monosomy 1p36 syndrome	
22q11.2 FISH	22q FISH, 22q11.2 deletion syndrome, DiGeorge syndrome, velocardiofacial syndrome (VCFS), conotruncal anomaly face syndrome (CTAF), Opitz G/BBB syndrome, Cayler cardiofacial syndrome, Shprintzen syndrome, CATCH22	TBX1
Angelman syndrome FISH	15q11.2 FISH	UBE3A
Cri du Chat syndrome FISH	5p FISH	
Fragile X syndrome	FMR1, FRAXA, FXS, Marker X syndrome, Martin-Bell syndrome, X-linked mental retardation	FMR1, FRAXA
Karyotype (blood)	Chromosome studies, karyotype	
Lactose intolerance test	Lactose test, lactase metabolism, lactase persistence	LCT, MCM6
Microarray (constitutional)	SNP array, microarray testing, SNP microarray, CGH microarray, CGH array	
Miller-Dieker syndrome FISH	LIS1 FISH	LIS1
Phelan-McDermid syndrome FISH	22q13 deletion FISH	SHANK3
Prader-Willi syndrome FISH	15q11 microdeletion	SNRPN, NDN
SHOX FISH	Xp22 FISH	SHOX
Smith-Magenis syndrome FISH	17p11.2 FISH	RAI1
Sotos syndrome FISH	Sotos FISH, cerebral gigantism FISH, NSD1 FISH	NSD1
Spinal muscular atrophy	SMN1, 5q SMA, spinal muscle wasting, spinal amyotrophy	SMN1
SRY FISH	Swyer syndrome, 46,XX testicular disorder, sex determining region on Y	SRY
Williams syndrome FISH	7q11.23 FISH, ELN deletion	ELN
Wolf-Hirschhorn syndrome FISH	del(4p) FISH	
Y chromosome FISH	Y FISH, Y chromosome FISH panel	
Endocrine		
KAL1 FISH, FGFR1 FISH	Kallman syndrome	KAL1, FGFR1
Respiratory		
CFTR mutation panel (cystic fibrosis and CBAVD)	Cystic fibrosis, CBAVD	CFTR
Gastrointestinal		
Coeliac disease HLA typing	Coeliac tissue typing, variants of HLA-DQ and DR	HLA-DQA1, HLA-DQB1

Pharmacogenomics

NAME OF TEST	TEST ALIAS	GENES
Hypersensitivity		
HLA-B*15:02	B*15:02, human leucocyte antigen B, carbamazepine hypersensitivity	HLA-B
HLA-B*57:01	B*57:01, human leucocyte antigen B, abacavir hypersensitivity	HLA-B
HLA-B*58:01	B*58:01, human leucocyte antigen B, allopurinol hypersensitivity	HLA-B
Toxicity		
DPYD screen	DHP, DPD, DHPDHASE, dihydropyrimidine dehydrogenase, 5-FU, capecitabine, tegafur	DPYD
Sonic PGx	Pharmacogenomic screen, pharmacogenetic screen, PGx test, PGx panel, pharmacogenetic screens for antidepressants/pain relief/warfarin sensitivity/proton pump inhibitors	ABCB1, CYP1A2, CYP2C19, CYP2C9, CYP2D6, CYP3A4, CYP3A5, OPRM1, SLCO1B1, VKORC1
TPMT screen	Thiopurine methyltransferase, azathiopurine, 6-mercaptopurine	TPMT
UGT1A1 screen	Gilbert syndrome, irinotecan	UGT1A1

Reproductive

NAME OF TEST	TEST ALIAS	GENES
Carrier screen		
Alpha thalassaemia screen	α-thalassaemia, mutations of HBA1 and HBA2	HBA1, HBA2
CFTR mutation panel	Cystic fibrosis, cystic fibrosis-related disease, congenital bilateral absence of vas deferens (CBAVD)	CFTR
Expanded reproductive carrier screen	Beacon carrier screen, expanded preconception carrier screen	>400 genes
Fragile X syndrome	FMR1, FRAXA, FXS, Marker X syndrome, Martin-Bell syndrome, X-linked mental retardation	FMR1, FRAXA
Reproductive carrier screen (CF, SMA and Fragile X)	Preconception carrier screen	CFTR, FMR1, SMN1
Spinal muscular atrophy	SMN1, 5q SMA, spinal muscle wasting, spinal amyotrophy	SMN1
Fetal diagnosis		
Karyotype (amniocentesis)	Chromosome studies, karyotype (prenatal)	
Karyotype (CVS)	Chromosome studies, karyotype (prenatal)	
Rapid prenatal FISH	Prenatal interphase FISH, rapid FISH, aneuploidy FISH	
Rapid interphase FISH	Urgent neonatal FISH	
Microarray (prenatal)	Prenatal SNP array, amniotic fluid microarray testing, CVS microarray testing, SNP microarray, CGH microarray, CGH array	Chromosomes 13, 18, 21, X and Y, plus specific microdeletions For further details, please refer to the Sonic Genetics website, www.sonicgenetics.com.au/nipt
Haematological		
Prothrombin gene	Prothrombin gene mutation, F2, thrombophilia	F2
Infertility/miscarriage		
DAZ gene (Y microdeletion)	DAZ deletion PCR, AZF	DAZ
Factor V Leiden	Factor 5 gene, thrombophilia	F5
Karyotype (products of conception)	Chromosome studies, karyotype (tissues), karyotype (miscarriage)	
Karyotype (reproductive)	Chromosome studies, karyotype (reproductive)	
Microarray (products of conception)	SNP array, microarray testing, SNP microarray, CGH microarray, CGH array	
Tissue FISH studies	POC FISH	
Y chromosome FISH	Y FISH, Y chromosome FISH panel	
Prenatal screen		
Non-invasive prenatal test	Harmony®, NIPT, NIPS, cfDNA, T21, T18, T13, Trisomy, Turner syndrome, Klinefelter syndrome, 22q11.2 deletion	Chromosomes 13, 18, 21, X and Y, plus specific microdeletions For further details, please refer to the Sonic Genetics website, www.sonicgenetics.com.au/nipt

Our Genetic Pathologists

Led by Professor Graeme Suthers, our expert Australian team of genetic pathologists is 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 supported by senior scientists who specialise in different areas of genetic testing.

Dr Kym Mina
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Director – Genetics
Douglass Hanly Moir Pathology

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If you would like to speak with one of our pathologists, please call 1800 010 447 or email info@sonicgenetics.com.au

Sonic Genetics

How to access

Sonic Genetics is a member of the Sonic Healthcare group of companies.

Supported by Sonic Healthcare's extensive network of state and regional laboratories, we benefit from interdisciplinary collaboration between many pathologists and scientists working across all specialties.

Please contact your local Sonic Healthcare laboratory listed below:

ACT	Capital Pathology
NSW	Douglass Hanly Moir Pathology Barratt & Smith Pathology Southern.IML Pathology
QLD/NT	Sullivan Nicolaides Pathology
SA	Clinpath Pathology
TAS	Hobart Pathology Launceston Pathology North West Pathology
VIC	Melbourne Pathology
WA	Clinipath Pathology

For further information please refer to our website, www.sonicgenetics.com.au or call us on 1800 010 447

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