Sonic Genetics

Information for Doctors

Genetic testing overview

SONIC PATHOLOGY AUSTRALIA

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.

This brochure details the broad range of genetic tests currently available through Sonic Genetics. Testing is performed both in Sonic Healthcare pathology laboratories, as well as high-quality fully accredited referral laboratories.

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, 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
Cancer		
Hereditary breast and ovarian cancer (germline)	Familial breast cancer, BRCA, familial ovarian cancer, serous cancer, hereditary breast and ovarian cancer (HBOC)	ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53
Cardiac		
APOE screen	Apolipoprotein E genotyping, hyperlipoproteinaemia	APOE
Cardiac gene panels	Multiple panels, including cardiomyopathies, arrhythmias, congenital malformations, vasculopathies and hyperlipidaemias	For details, please refer to the Sonic Genetics website, sonicgenetics.com.au/cardiac-genetics
Familial hypercholesterolaemia	FH, hyperlipidaemia	LDLR, APOB, PCSK9, LDLRAP1
Gastrointestinal		
Coeliac disease HLA typing	Coeliac tissue typing, variants of HLA-DQ and DR	HLA-DQ2, DQ8, DQA1*02, DQA1*05, DR4
Lactose intolerance	Lactose test, lactase metabolism, lactase persistence	LCT, MCM6
Haematological		
Alpha thalassaemia screen	α-thalassaemia	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
Hepatic		
Haemochromatosis type 1	HFE, HH, hereditary haemochromatosis, bronzed cirrhosis, iron storage disorder	HFE
UGT1A1 screen	Gilbert syndrome, irinotecan	UGT1A1
Hyperlipidaemia		
APOE screen	Apolipoprotein E genotyping, hyperlipoproteinaemia	APOE
Familial hypercholesterolaemia	FH, hyperlipidaemia	LDLR, APOB, PCSK9, LDLRAP1
Immunological		
Hereditary angioedema (I, II & III)	HAE type I, HAE type II, HAE type III, SERPING1, F12	F12, SERPING1
Neurological		
Fragile X syndrome	FMR1, FRAXA, FXS, Marker X syndrome, Martin-Bell syndrome, X-linked mental retardation	FMR1, FRAXA
Recurrent fever screen		
Periodic fever syndromes	Familial Mediterranean fever screen, MEFV, Familial Hibernian fever, TRAPS screen, mevalonate kinase deficiency, cryopyrin-associated periodic fevers syndromes	MEFV, MVK, NLRP3, TNFRSF1A
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 & III)	HAE type I, HAE type II, HAE type III, SERPING1, F12	F12, 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
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
Gastrointestinal		
Coeliac disease HLA typing	Coeliac tissue typing, variants of HLA-DQ and DR	HLA-DQ2, DQ8, DQA1*02, DQA1*05, DR4
Lactose intolerance	Lactose test, lactase metabolism, lactase persistence	LCT, MCM6
Recurrent fever screen		
Periodic fever syndromes	Familial Mediterranean fever screen, MEFV, Familial Hibernian fever, TRAPS screen, mevalonate kinase deficiency, cryopyrin-associated periodic fevers syndromes	MEFV, MVK, NLRP3, TNFRSF1A

Oncology

NAME OF TEST	TEST ALIAS	GENES
HAEMATOLOGICAL MALIGNANCIES		
Chimerism		
Sex mismatch FISH	Sex mismatch FISH, chimerism FISH	
Myeloid neoplasms - Karyotype and panels		
Acute myeloid leukaemia FISH panel	AML FISH	CBFB, DEK, ETV6, EVI1, KMT2A, MYH11, NUP214, PML, RARA, RUNX1, RUNX1T1
Eosinophilia FISH panel (myeloid/lymphoid neoplasms)	PDGFRA, PDGFRB, FGFR1	
Karyotype (leukaemia/lymphoma)	Chromosome studies, karyotype (oncology)	
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		
CALR screen	CALR gene test, calreticulin gene test	CALR
FGFR1 FISH	Fibroblast growth factor receptor 1	FGFR1
JAK2 screen	JAK2 gene test, myeloid neoplasms	JAK2 V617F
MPL screen	MPL gene test	MPL
PDGFRA/FIP1L1 FISH	PDGFRA FISH, FIP1L1-PDGFRA FISH	PDGFRA, FIP1L1
PDGFRB FISH	5q33 FISH, PDGFRB FISH	PDGFRB
Lymphoid neoplasms - Karyotype and panels		
Chronic lymphocytic leukaemia FISH panel	CLL FISH	
Chronic lymphocytic leukaemia microarray	CLL microarray	
IGH/FGFR3 FISH	t(4;14) FISH	IGH, FGFR3
IGH/MAF FISH	t(14;16) FISH	IGH, MAF
Karyotype (leukaemia/lymphoma)	Chromosome studies, karyotype (oncology)	
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		
13q deletion FISH	13q FISH, mature B-cell lymphoid neoplasms	
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	

Oncology

NAME OF TEST	TEST ALIAS	GENES
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
Anaplastic large cell lymphoma FISH	ALCL FISH, DUSP22 FISH, TP63 FISH	TP63
BCL6 FISH	3q27 FISH, B-cell non-Hodgkin lymphomas	BCL6
Burkitt lymphoma FISH	t(8;14) FISH, IGH-MYC FISH, MYC-IGH FISH	IGH, MYC
CCND2 FISH	B-cell lymphoma	CCND2
Hepatosplenic T-cell lymphoma FISH	Isochromosome 7q, i(7q), i(7)(q10)	Isochromosome 7q
IGH/BCL2 FISH	t(14;18) FISH	IGH, BCL2
IGH/MALT1 FISH	MALT FISH, t(11;18) FISH	IGH, MALT1
IGK, IGL FISH	IGKC FISH, IGLC1 FISH	IGKC, IGLC1
IRF4/DUSP22 FISH	Interferon regulatory factor 4, dual-specificity phosphatase 22	IRF4/DUSP22
Mantle cell lymphoma FISH	t(11;14) FISH, MCL FISH, CCND1 FISH, CCND2 FISH	IGH, CCND1
MYC FISH	8q24 FISH, c-myc FISH	MYC
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
Glioma focused gene panel	Glioma panel	BRAF, IDH1, IDH2, TP53
PTEN FISH	PTEN deletion FISH, gliomas	PTEN
Breast cancer		
Breast cancer focused gene panel	Breast panel	AKT1, ERBB2 (HER2), ESR1, PIK3CA
Breast cancer gene expression prognostic assay	Prosigna®, PAM50	
Hereditary breast and ovarian cancer (germline)	Familial breast cancer, BRCA, familial ovarian cancer, serous cancer, hereditary breast and ovarian cancer (HBOC)	ATM, BRCA1, BRCA2, BRIP1, CHEK2, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53
Secretory carcinoma of the breast FISH	Gene fusions involving NTRK3 and ETV6	NTRK3
Colorectal cancer		
BRAF V600 screen	BRAF gene test, melanoma, colorectal cancer	BRAF
Colorectal cancer focused gene panel	Colorectal panel	BRAF, KRAS, NRAS, PIK3CA
KRAS and NRAS screen	RAS panel, KRAS and NRAS gene test, colorectal cancer	KRAS, NRAS
Endometrial cancer		
Endometrial cancer focused gene panel	Endometrial panel	CTNNB1, PIK3CA, POLE, TP53
Gastrointestinal stromal cancer		
GIST focused gene panel	GIST, gastrointestinal stromal panel	BRAF, KIT, PDGFRA
Head and neck cancer		
Mammary analogue secretory carcinoma FISH	Gene fusions involving NTRK3 and ETV6	NTRK3
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, MET, 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
Renal cancer		
MiT family translocation RCC FISH	Papillary renal cell carcinoma FISH, alveolar soft part sarcoma FISH	TFE3
Sarcoma		
Alveolar rhabdomyosarcoma FISH	FOXO1 FISH, FKHR FISH, FOXO1-PAX3, FOXO1-PAX7	FOXO1, PAX3, PAX7
Alveolar soft part sarcoma FISH	TFE3 FISH, MiT family translocation renal cell carcinoma FISH	TFE3
Aneurysmal bone cyst FISH	USP6 17p13 FISH, bone tumours	USP6
Dermatofibrosarcoma protuberans (DFSP)	COL1A/PDGFB FISH	COL1A, PDGFB
Desmoplastic small round cell tumour FISH	EWSR1-WT1 FISH	EWSR1, WT1

Oncology

NAME OF TEST	TEST ALIAS	GENES
Ewing-like sarcoma FISH	t(4;19) FISH, CIC-DUX4 FISH, undifferentiated small blue round cell sarcoma FISH, primitive round cell sarcoma FISH	CIC, DUX4
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
FUS/DDIT3 FISH	t(12;16) FISH, liposarcoma	FUS, DDIT3
Liposarcoma FISH	MDM2 amplification, MDM2 FISH	MDM2
Myxoid liposarcoma FISH	t(12;16) FISH, CHOP-FUS FISH, FUS-DDIT3 FISH	FUS, DDIT3 (formally known as CHOP)
Nodular fasciitis FISH	USP6 17p13FISH, USP6 FISH	USP6
Synovial sarcoma FISH	t(X;18) FISH, SS18 FISH	SS18
Solid tumour		
Foundation Medicine	Pan-cancer comprehensive genomic profile assay	>300 genes
Karyotype (tumour)	Chromosome studies, karyotype (oncology)	
NTRK1, NTRK2, NTRK3 FISH	Oncogenes	
Solid tumour panel (Find It)	Find It cancer hotspot panel, full Find It panel, NSCLC/melanoma/colorectal focused panel	≥30 genes

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	
Microarray (constitutional)	SNP array, microarray testing, SNP microarray, CGH microarray, CGH array	
Microarray (parental)	Follow-up microarray, parental SNP array, parental CGH microarray, parental microarray, parental molecular karyotype	
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
Gastrointestinal		
Coeliac disease HLA typing	Coeliac tissue typing, variants of HLA-DQ and DR	HLA-DQ2, DQ8, DQA1*02, DQA1*05, DR4
Lactose intolerance test	Lactose test, lactase metabolism, lactase persistence	LCT, MCM6
Respiratory		
CFTR mutation panel	Cystic fibrosis, cystic fibrosis-related disease	CFTR

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
Medication guidance		
Pharmacogenomic (PGx) screen	Pharmaco profile, PGx, Sonic PGx panel, pharmacogenomics, pharmacogenetics, Cytochrome P450	ABCB1, CYP1A2, CYP2C19, CYP2C9, CYP2D6, CYP3A4, CYP3A5, OPRM1, SLCO1B1, VKORC1
Toxicity		
DPYD screen	DHP, DPD, DHPDHASE, dihydropyrimidine dehydrogenase, 5-FU, capecitabine, tegafur	DPYD
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	HBA1, HBA2
CFTR mutation panel	Cystic fibrosis, cystic fibrosis-related disease, congenital bilateral absence of vas deferens (CBAVD)	CFTR
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
Reproductive carrier screen (expanded)	Beacon carrier screen, expanded preconception carrier screen	>400 genes
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)	
Microarray (prenatal)	Prenatal SNP array, amniotic fluid microarray testing, CVS microarray testing, SNP microarray, CGH microarray, CGH array	
Prenatal single gene testing	Detect mutations found in carrier screening	
Rapid prenatal FISH	Prenatal interphase FISH, rapid FISH, aneuploidy FISH	
Rapid interphase FISH	Urgent neonatal FISH	
Infertility/miscarriage		
CFTR mutation panel	Cystic fibrosis, cystic fibrosis-related disease, congenital bilateral absence of vas deferens (CBAVD)	CFTR
Factor V Leiden	Factor 5 gene, thrombophilia	F5
Fragile X syndrome	FMR1, FRAXA, FXS, Marker X syndrome, Martin-Bell syndrome, X-linked mental retardation	FMR1, FRAXA
Fragile X premutation screen	Premature ovarian failure	FMR1, FRAXA
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	
MTHFR screen	5,10-methylenetetrahydrofolate reductase	MTHFR
POC FISH studies	POC FISH	
Prothrombin gene	Prothrombin gene mutation, F2, thrombophilia	F2
Y chromosome FISH	Y FISH, Y chromosome FISH panel	
Y microdeletion (DAZ gene)	DAZ deletion PCR, AZF	DAZ
IVF		
Preimplantation FISH	IVF FISH, PGD FISH, single cell FISH	
Prenatal screen		
Non-invasive prenatal test	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, sonicgenetics.com.au/nipt

SONIC PATHOLOGY AUSTRALIA

Sonic Genetics is part of Sonic Pathology Australia, the largest pathology provider in the country. Our state-of-the-art laboratories and extensive network of collection centres serve each state and territory capital, as well as regional and rural Australia.

Douglass Hanly Moir Pathology
Sullivan Nicolaides Pathology
Melbourne Pathology
Barratt & Smith Pathology
Capital Pathology
Clinipath Pathology
Bunbury Pathology
Clinpath Pathology
Hobart Pathology
Launceston Pathology
North West Pathology
Southern.IML Pathology

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