



PRICE LIST FOR OUT-OF-STATE PATIENTS (JUL 2017 – DEC 2017)

What methods of testing do we employ?

Available Methods

- PCR and/or Sanger DNA Sequencing for predictive testing and familial cascade screening.
- Targeted Massive Parallel Sequencing (MPS) panels and Sanger sequencing to analyse large genes.
- MLPA to detect larger deletions and duplications.
- MS-MLPA to detect methylation changes in addition to deletions and duplications.

If you are unsure which method is appropriate for your patient, please contact us by phone on 08 6383 4223 or email on DiagnosticGenomicsQE.PathWest@health.wa.gov.au.

Who do we accept testing requests from?

Requesting Clinicians

Diagnostic testing can only be requested by a suitably qualified clinician – we do not provide a service direct to the public. For some tests, we will only accept requests once the patient has undergone genetic counselling from a recognised genetic counsellor, due to the clinical sensitivity of these tests.

What types of sample(s) are required for testing?

Sample requirements for each test are listed below.

EDTA Samples

Most tests will require a single 2-4mls sample of blood collected with an EDTA preservative. EDTA samples must arrive at our lab within 5 days of phlebotomy, and must be sent at room temperature.

Tissue

10-50mg of tissue is required for DNA extraction

DNA

1-5µg of extracted DNA (depending on test request) in place of EDTA blood

Predictive Testing

We recommend testing two separate EDTA blood samples collected from the patient at least 10 minutes apart.

Familial Cancer and Confirmation Testing

We recommend testing a second EDTA blood sample in cases where a pathogenic variant is found.

Maternal Contamination for prenatal samples

A sample of maternal blood collected in EDTA is required.

How long will it take to receive a result?

Estimated turn-around-times (TAT) are listed next to the respective test and are shown in weeks.

Prenatal Testing

To assist us in prioritising prenatal testing, please ensure you advise us well ahead of time of pending samples. Patients undergoing prenatal testing will be anxious to receive their results as soon as possible so we prioritise prenatal testing and will endeavour to report results within a week of receiving a sample.

How much do we charge for testing?

Prices are listed below (excluding 10% GST) in Australian Dollars (\$AUD) and include DNA extraction costs.

Medicare Rebates

Testing is not covered by Medicare, so please do not request Bulk Billing. The nominated party will be charged for the full cost of the test as listed below.

Payment Responsibility

Please clearly indicate on the request form the clinician, health institution or pathology laboratory who will be paying for the cost of testing. We will not charge patients without their express written consent and acknowledgement of cost. A copy of our payment consent form can be found on page 7. Where the party responsible for payment is not clear, we will request a signed payment consent form prior to commencing testing.

Where do you need to send your sample?

Ensure you advise us well ahead of time of urgent samples including prenatal and overseas samples.

All samples should be sent to the address below accompanied by a request form outlining the test(s) required and billing information. Please ensure you mark all urgent samples accordingly to ensure they are given priority.

Delivery Address:

Diagnostic Genomics, Level 2
c/- Specimen Reception
Ground Floor, PP Block
QEII Medical Centre
Hospital Avenue
Nedlands WA 6009
AUSTRALIA

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Predictive Testing	Gene	\$AU	TAT(w)	Sample Type
Dominant / Single Amplicon	(Select Exons)	\$300	8	Various
Recessive / Dual Amplicon	(Select Exons)	\$300	8	Various

Prenatal Testing	Gene	\$AU	TAT(w)	Sample Type
Prenatal Diagnosis including Maternal Contamination	(Select Exons + MCC)	\$1000	1	Amniotic Fluid <u>or</u> CVS
Prenatal Diagnosis <u>without</u> Maternal Contamination	(Select Exons)	\$800	1	Amniotic Fluid <u>or</u> CVS

Inherited Cancer Gene Panel Testing

Inherited cancer gene testing is performed by Massively Parallel Sequencing (MPS) using the Illumina TruSight Cancer Panel™. This panel contains 94 inherited cancer genes. Bioinformatic filtering is used to analyse a single gene or multiple genes as requested. The standard cost for the panel is \$1000. If required, additional MLPA testing is charged at \$300 per probe mix (NB: Some genes need more than one MLPA test). **Reported variants are confirmed by Sanger sequencing and will incur an additional \$200 cost.** Please contact the laboratory with enquiries concerning testing of other genes on this panel.

	Gene (s)	\$AU	TAT(w)	Method
Ataxia Telangiectasia	ATM	\$1300	12	MPS + MLPA
Birt-Hogg-Dubé Syndrome	FLCN	\$1300	12	MPS + MLPA
Breast Cancer	BRCA1, BRCA2, TP53, PTEN, PALB2 (truncating variants)	\$1000	12	MPS + MLPA (BRCA1 and BRCA2)
Cowdens	PTEN	\$1300	12	MPS + MLPA
Familial Adenomatous Polyposis	APC	\$1300	12	MPS + MLPA
	MUTYH	\$1300	12	MPS + MLPA
Lynch Syndrome/Hereditary Nonpolyposis Colorectal Cancer	MLH1, MSH2, MSH6	\$1600	12	MPS + MLPA
	PMS2	\$1300	12	MPS, Sanger Sequencing + MLPA
Hyperparathyroidism-2	HRPT2/CDC73	\$1000	12	MPS
Legius Syndrome	SPRED1	\$600	12	Sanger Sequencing
Li-Fraumeni Syndrome	TP53	\$1300	12	MPS + MLPA
Multiple Endocrine Neoplasia	MEN1 (Type 1)	\$1300	12	MPS + MLPA
	RET (Type 2A/2B)	\$1000	12	MPS
Neurofibromatosis	NF1	\$1600	12	MPS + MLPA
	NF2	\$1300	12	MPS + MLPA
Hereditary Paraganglioma-Pheochromocytoma Syndromes	SDHA, SDHB, SDHC, SDHD, SDHAF2	\$1300	12	MPS + MLPA
Peutz-Jeghers Syndrome	STK11	\$1300	12	MPS + MLPA
Proteus Syndrome	PTEN	\$1300	12	MPS + MLPA
Retinoblastoma	RB1	\$1300	12	MPS + MLPA
Tuberous Sclerosis	TSC1, TSC2	\$1600	12	MPS + MLPA
Von Hippel Lindau Disease (VHL)	VHL	\$1300	12	MPS + MLPA

Other available genes on the Cancer Panel:

AIP, ALK, BAP1, BLM, BMP1A, BRIP1, BUB1B, CDK4, CDKN1C, CDKN2A, CEBP1, CEP57, CHEK2, CYLD, DDB2, DICER1, DIS3L2, EGFR, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCM, FH, GATA2, GPC3, HNFF1A, HRAS, KIT, MAX, MET, NBN, PALB2, PHOX2B, PRF1, PRKAR1A, PTCH1, RAD51C, RAD51D, RECQL4, RHBDF2, RUNX1, SBDS, SDHAF2, SLX4, SMAD4, SMARCB1, SUFU, TMEM127, WRN, WT1, XPA, XPC

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Inherited Cardiac Panel Testing

Inherited cardiac gene testing is performed by MPS using the Illumina TruSight Cardio Sequencing Kit™. This panel contains 174 genes associated with inherited cardiac conditions. Bioinformatic filtering is used to analyse multiple genes as subpanels depending on phenotypic information provided. **Reported variants are confirmed by Sanger sequencing and will incur an additional \$200 cost per variant.** Please contact the laboratory with enquiries concerning testing of other genes on this panel.

	Genes (s)	\$AU	TAT (w)	Method
Aortopathy subpanel	ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, MYH11, MYLK, NOTCH1, SLC2A10, SMAD3, SMAD4, TGFB2, TGFB1, TGFB2	\$1000	16	MPS panel
Cardiac conduction subpanel	AKAP9, ABCC9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CASQ2, CAV3, DPP6, GJA5, GPD1L, HCN4, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, NPPA, RANGRF, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SNTA1, TRDN, TRPM4	\$1000	16	MPS panel
Cardiomyopathy gene subpanel	ABCC9, ACTA2, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, CALR3, COL3A1, CRYAB, CSRP3, CTF1, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FBN1, FHL2, FKTN, GAA, GATAD1, GLA, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MIB1, MURC, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYOZ2, MYPN, NEXN, NKX2-5, NOTCH1, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RYR2, SCN5A, SDHA, SGCD, SLC25A4, SLC2A10, SMAD3, SMAD4, TAZ, TCAP, TGFB2, TGFB3, TGFB1, TGFB2, TMEM43, TMPO, TNNC1, TNNT3, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL	\$1000	16	MPS panel
Marfan Syndrome subpanel	FBN1, TGFB1, TGFB2	\$1600	16	MPS + MLPA (FBN1)
Rasopathy Subpanel	BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SHOC2, SOS1	\$1000	16	MPS panel

Other available genes on the Cardiac Panel:

ABCG5, ABCG8, ACTA1, APOA4, APOA5, APOB, APOC2, APOE, CETP, COX15, CREB3L3, CRELD1, ELN, FHL1, FKRP, FXN, GCKR, GPIHBP1, HADHA, HFE, HSPB8, ILK, KLF10, LAMA2, LDLR, LDLRAP1, LMF1, LPL, LTBP2, MYO6, NODAL, PCSK9, PRKAR1A, RYR1, SALL4, SCO2, SEPN1, SGCB, SGCG, SREBF2, TBX20, TBX3, TBX5, TRIM63, ZBTB17, ZHX3, ZIC3

Please contact the laboratory.

Other Genetic Testing (A-Z)

	Gene	\$AU	TAT	Method Type
Where testing is performed by MPS, reported variants are confirmed by Sanger sequencing and will incur an additional \$200 cost per variant.				
5alpha-Reductase (5ARD)	SRD5A2	\$600	8	Sanger Sequencing
Achondroplasia (Short Stature) (ACH)	FGFR3 (Select Exons)	\$800	8	Sanger Sequencing
Acyl-Coenzyme A Dehydrogenase (ACADM)	ACADM	\$500	8	PCR
	ACADM (Select Exons)	\$200	4	Sanger Sequencing
Alagille Syndrome (AGS)	JAG1	\$1300	16	*MPS Panel + MLPA
Alexander Disease (ALX)	GFAP	\$800	12	Sanger Sequencing
Alpers Syndrome (AHS)	POLG (Select Exons)	\$600	8	Sanger Sequencing
Alpha-Thalassemia X-Linked Intellectual Disability (ATRX)	ATRX (Select Exons)	\$1000	12	Sanger Sequencing
Androgen Receptor (AR)	AR	\$1000	12	Sanger Sequencing
Angelman Syndrome (AS)	15q11-13 (Methylation)	\$400	8	MS-MLPA
Ataxia	66 genes	\$1100	16	†MPS Panel

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Axenfeld-Rieger Syndrome (RIEG)	PITX2	\$400	12	Sanger Sequencing
Becker Muscular Dystrophy (BMD)	DMD (MLPA)	\$600	8	MLPA
	DMD (cDNA-Frozen Muscle)	\$2000	16	Sanger Sequencing
Beckwith Wiedemann Syndrome (BWS)	11p15 (Methylation)	\$400	8	MS-MLPA
Blepharophimosis-Ptosis-Epicanthus (BPES)	FOXL2	\$600	8	Sanger Sequencing + MLPA
CADASIL	NOTCH3	\$1100	8	†MPS Panel
Cerebral Cavernous Malformation (CCM)	KRIT1, CCM2, PDCD10	\$1900	12	**MPS Panel + MLPA
Charcot-Marie-Tooth Neuropathy (CMT)	92 genes	\$1100	16	†MPS Panel
	PMP22	\$300	8	MLPA
CHARGE Syndrome	CHD7	\$1600	12	**MPS Panel + MLPA
Congenital Adrenal Hyperplasia (CAH)	CYP21A2	\$900	12	Sanger Sequencing + MLPA
Congenital Muscular Dystrophy (CMD)	37 genes	\$1100	16	†MPS Panel
Congenital Myasthenic Syndrome (CMS)	24 genes	\$1100	16	†MPS Panel
Cri du Chat Syndrome (CDC)	5p15	\$300	8	MLPA
Cystic Fibrosis (CF)	CFTR (CF29 Screen)	\$200	4	Multiplex PCR
	CFTR	\$1600	16	**MPS + MLPA
Deafness / Connexin (CX)	GJB2 / CX26	\$300	8	Sanger sequencing
	GJB6 / CX30	\$200	8	Deletion PCR
Deafness / Aminoglycoside Hearing Loss (AHL)	MT-RNR1	\$200	8	Sanger sequencing
Dentatorubral-Pallidolusian Atrophy (DRPLA)	ATN1	\$200	8	PCR
DiGeorge Syndrome (DGS)	22q11	\$300	8	MLPA
Distal Arthrogyrosis (DA2B)	77 genes	\$1100	16	†MPS Panel
Duchenne Muscular Dystrophy (DMD)	DMD (MLPA)	\$600	8	MLPA
	DMD (cDNA) Frozen Muscle	\$2000	16	Sanger Sequencing
Dystonia	TOR1A (Common Mutation)	\$200	8	Sanger Sequencing
	25 genes	\$1100	16	†MPS Panel
Familial Cerebral Angiopathy (HCHWA-D)	APP	\$200	8	Sanger Sequencing
Familial Visceral Myopathy / Berdon Syndrome (FVM)	ACTG2	\$900	12	Sanger sequencing
Friedreich Ataxia (FRDA)	FXN	\$300	8	PCR
Glycogen Storage Disease/Rhabdomyolysis (GSD/Rhabdo)	21 genes	\$1100	16	†MPS Panel
Haemophilia A	Factor VIII (INV22 Screen)	\$500	8	PCR
Hereditary Pancreatitis (PCTT)	PRSS1	\$600	8	Sanger Sequencing
Hereditary Spastic Paraparesis (HSP)	92 genes	\$1100	16	†MPS Panel
	SPAST (SPG4 MLPA)	\$300	12	Sanger Sequencing
HNPP	PMP22	\$300	8	MLPA
Hypokalemic Periodic Paralysis (HypoPP)	CACNA1S + SCN4A	\$400	8	Sanger Sequencing
Hypothyroidism (HT-H)	SLC26A4	\$2000	12	Sanger sequencing
Incontinentia Pigmenti / Bloch-Sulzberger Syndrome (IP)	IKBKG	\$1000	12	PCR and Sanger sequencing
Kallmann Syndrome (KS)	KAL1	\$1900	12	**MPS Panel + MLPA
Kearns-Sayre Syndrome (KSS)	Mitochondrial DNA	\$1000	16	PCR
Kennedy Disease / SBMA	AR	\$200	8	PCR
Langer-Giedion Syndrome (LGS)	8q24.12	\$300	8	MLPA
Leber Hereditary Optic Neuropathy (LHON)	MT-ND1/-ND4/-ND4L/-ND6	\$600	8	Sanger sequencing
Leukodystrophy	34 genes	\$1100	16	†MPS Panel

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Lissencephaly /Microcephaly (LIS)	74 genes	\$1100	16	†MPS Panel
	LIS1, DCX	\$600	12	MLPA
Lymphedema-Distichiasis Syndrome (LDS)	FOXC2	\$500	8	Sanger Sequencing
Malignant Hyperthermia (MH)	RYR1 (<i>cdNA</i>)	\$2000	16	Sanger Sequencing
Maternally-Inherited Leigh Syndrome (MILS)	MT-ATP6	\$300	8	Sanger Sequencing
McArdle Disease (GSDV)	PYGM (<i>Common Mutation</i>)	\$200	8	Sanger Sequencing
MELAS	MT-TL1 (<i>Common Mutation</i>)	\$300	8	Sanger Sequencing
Melnick-Needles Syndrome (MNS)	FLNA	\$1300	16	*MPS Panel
MERRF	MT-TK (<i>Common Mutation</i>)	\$300	8	PCR
Microdeletion of the Y Chromosome	AZF (<i>Males Only</i>)	\$200	4	Deletion PCR
Microdeletions	1p36	\$300	8	MLPA
	2p16	\$300	8	MLPA
	3q29	\$300	8	MLPA
	9q22.3	\$300	8	MLPA
	15q24	\$300	8	MLPA
	17q21	\$300	8	MLPA
Miller-Dieker Syndrome (MDS)	17p13.3	\$300	8	MLPA
Mitochondrial Myopathy (Nuclear)	33 genes	\$1100	16	†MPS Panel
Motor Neurone Disease/Spinal Muscular Atrophy (MND/SMA)	41 genes	\$1100	16	†MPS Panel
	C9orf72	\$400	12	PCR
Moyamoya Disease (MYMY)	ACTA2	\$900	12	Sanger Sequencing
Myotonic Dystrophy (DM)	DMPK (<i>Type 1</i>)	\$400	8	PCR
	CNBP / ZNF9 (<i>Type 2</i>)	\$400	8	PCR
Myopathy	89 genes	\$1100	16	†MPS Panel
Muscle Channelopathy	5 genes	\$1100	16	†MPS Panel
Nemaline Myopathy (NEM)	NEB (<i>Type 2 Exon 55 Deletion</i>)	\$200	8	PCR
Neurogenic Ataxia <u>and</u> Retinitis Pigmentosa (NARP)	MT-ATP6 (<i>Common Mutation</i>)	\$300	8	Sanger Sequencing
Oculocutaneous Albinism (OCA)	TYR (<i>Type 1</i>)	\$800	12	Sanger Sequencing
Oculopharyngeal Muscular Dystrophy (OPMD)	PABPN1	\$200	8	Sanger Sequencing
Phelan-McDermid Syndrome (PMS)	22q13	\$300	8	MLPA
Potocki-Lupski Syndrome (PTLS)	17p11.2	\$300	8	MLPA
Prader-Willi Syndrome (PWS)	11q11-13	\$400	8	MS-MLPA
Prion Disease (TSE)	PRNP	\$300	8	Sanger Sequencing
Progressive External Ophthalmoplegia (CPEO)	Mitochondrial DNA	\$1000	16	PCR
Rett Syndrome (RTT)	MECP2 (<i>MLPA</i>)	\$300	8	MLPA
	MECP2	\$600	8	Sanger Sequencing
Rippling Muscle Disease (RMD)	CAV3	\$300	8	Sanger Sequencing
Rubinstein-Taybi Syndrome (RSTS)	16p13.3	\$300	8	MLPA
Russell Silver Syndrome (RSS)	11p15 (<i>Methylation</i>)	\$400	8	MS-MLPA
Shwachman-Diamond Syndrome (SDS)	SBDS	\$1000	8	***MPS Panel
Simpson-Golabi-Behmel Syndrome	GCP3	\$300	8	MLPA
Smith-Magenis Syndrome (SMS)	17p11.2	\$300	8	MLPA
SOTOS Syndrome	NSD1	\$1000	16	***MPS Panel
Spinal Muscular Atrophy (SMA)	SMN1	\$300	8	Deletion PCR

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Spinocerebellar Ataxia (SCA)	ATX1 (Type 1)	\$200	8	PCR
	ATX2 (Type 2)	\$200	8	PCR
	ATX3 (Type 3)	\$200	8	PCR
	CACNA1A (Type 6)	\$200	8	PCR
	ATX7 (Type 7)	\$200	8	PCR
Analysis of all of the above 5 genes	(All of the Above)	\$500	8	PCR
	TBP (Type 17)	\$200	8	PCR
Tauopathies	MAPT (Select Exons)	\$600	12	Sanger Sequencing
Uniparental Disomy (UPD)	UPD 14	\$400	8	MS-MLPA
	UPD 7	\$400	8	MS-MLPA
WAGR Syndrome	11p13-14	\$300	8	MLPA
Williams-Beuren Syndrome (WBS)	7q11.23	\$300	8	MLPA
Wilson Disease (WND)	ATP7B	\$1300	12	**MPS Panel
Wiskott-Aldrich Syndrome (WAS)	WAS	\$1300	12	**MPS Panel
Wolf-Hirschhorn Syndrome (WHS)	4p16	\$300	8	MLPA
X Inactivation Studies (AR)	AR	\$400	8	Methylation PCR
X-Linked Agammaglobulinemia (XLA / AGMX1)	BTK	\$1500	8	Sanger Sequencing
X-Linked Anhidrotic Ectodermal Dysplasia (EDA1 / XHED)	EDA	\$1600	12	**MPS Panel + MLPA
X-Linked Hydrocephalus (HYCX)	L1CAM	\$1300	16	**MPS Panel
X-Linked Mental Retardation (MRX)	ATRX (Select Exons)	\$1000	12	Sanger Sequencing
	ARX (Select Exons)	\$200	8	Sanger Sequencing

A number of genes that are not listed above are available for testing using MPS technology. Please contact the laboratory with other gene testing enquiries.

- * MPS via Illumina TruSight Cardio Sequencing Kit
- **MPS via Illumina TruSight One Sequencing Panel
- ***MPS via Illumina TruSight Inherited Cancer Panel
- †MPS via Custom Neuromuscular gene panel



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Payment Consent Form **(FIELDS MARKED * ARE MANDATORY)**

Return via Fax: Att: Clerical Officer Fax: +61 (0)8 6457 4029 Phone: +61 (0)8 6383 4234 Return via Email: DiagnosticGenomicsQE.PathWest@health.wa.gov.au	Return via Post: Department of Diagnostic Genomics PathWest Laboratory Medicine WA Level 2, PP Block, QEII MC Locked Bag 2009 Nedlands WA 6909
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Date Returned: _____ Returned By: _____ Institution: _____	Pages (if Faxed): _____ <i>(Including Attachments)</i> Phone Number: _____ Fax Number: _____
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TESTING DETAILS

Patient Surname:* _____ Patient Forename:* _____ Referring Sample No.: _____ Referring Laboratory:* _____ Test(s) Requested:* _____ <i>(list one per line)</i>	Patient DOB:* _____ Patient Gender:* _____ Requesting Clinician:* _____ Clinician Institution:* _____ Test Cost(s):* _____ \$AU <i>(incl. GST if required)</i> \$AU _____ \$AU _____ \$AU Total Cost of Above Test(s):* _____ \$AU
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BILLING DETAILS

DO NOT PROCEED WITH TESTING *(Please provide details of the party responsible for cancelling the above testing)*

Authorising Officer:* _____ **Institution:*** _____

HEALTH INSTITUTION / CLINICIAN TO BE INVOICED *(Please provide details of the party to be invoiced for the above testing)*

Please note the full contact details of the party to be invoiced for the above testing:

Contact Name:* _____ Phone Number:* _____ Fax Number:* _____ Email Address:* _____	Billing Institution:* _____ Postal Address:* _____ Suburb & Postcode:* _____ State & Country:* _____
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PATIENT/GUARDIAN TO BE INVOICED *(Testing CANNOT be claimed via Medicare. Your patient must pay for the above testing in full)*

I*, _____ *(PRINT PATIENT/GUARDIAN NAME)* consent to pay the total cost of the requested test(s) listed above, up to but not exceeding a total amount of \$AU* _____ *(TOTAL COST)* I am aware that this amount cannot be claimed via Medicare.

Signature of Patient/Guardian*: _____ *(PATIENT/GUARDIAN SIGNATURE)*

Contact Name:* _____ Phone Number:* _____ Fax Number:* _____ Email Address:* _____	Postal Address:* _____ Suburb & Postcode:* _____ State & Country:* _____
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