

## VCGS MOLECULAR GENETICS LABORATORY TESTS, CHARGES AND REPORTING TIME

- Current as of June 2010
- These charges are relevant to Non Victorian patients and non funded tests
- If sample is urgent and requires a quick turnaround time, please contact VCGS Pathology, Molecular Genetics Laboratory on 03 8341 6275 to discuss.

Disorder	Cost	Multiple Testing	Time taken
Ashkenazi Jewish Screening [Tay Sachs disease, Canavan disease, Niemann-Pick disease, CF, Bloom Syndrome, Fanconi anemia, Familial Dysautonomia]		Full screen (7 conditions) \$500 Cascade testing (only 1 condition reported) \$275	4 - 6 weeks
Collagen Type 2 Diagnostic Family History	\$1950 \$250		TBD
Connexin 26 [full sequence] Carrier Testing for known mutation	\$350 \$250	Cx26 sequencing and Cx30 \$495	4 - 8 weeks 4 - 8 weeks
Connexin 30 deletion [2 mutations] Prenatal	\$170 \$600	The Children's Hospital at Westmead - Cx26 +Cx30 \$350	4 - 8 weeks 4 days- 3 weeks
Cystic Fibrosis Population Screen Standard Screen Extended Screen Prenatal	\$220 \$220 \$220 \$600		5 - 7 working days 2 - 3 weeks 4 weeks 4 days- 3 weeks
DRPLA	\$250		3 - 6 months
DNA Extraction Automated extraction (10-20 µg) Manual extraction Prenatal sample for sending	\$60 \$120 \$120		2 - 4 weeks 2 - 4 weeks 4 days- 3 weeks
Duchenne/Becker Muscular Dystrophy MLPA Prenatal	\$450 \$600		4 - 6 weeks 4 days- 3 weeks
Emery Dreifuss Muscular Dystrophy Lamin gene sequencing Emerin gene sequencing	\$750 \$450	Lamin + emerin \$1,000	6 - 9 months 6 - 9 months

VCGS Pathology / Laboratory: Molecular Genetics	Prepared by: Melanie Smith	Doc No: MG-W-212
Doc Name: VCGS Molecular Genetics Laboratory Test Table	Authorised by: Desiree du Sart	Version: 6
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VCGS Molecular Genetics Laboratory Tests, Charges and Reporting Time

Predictive testing	\$250		4 - 6 weeks
FAP			
Mutation Detection (sequencing)	\$1700		8 - 12 weeks
APC MLPA	\$250		6 - 8 weeks
Predictive testing	\$250		4 - 6 weeks
Friedreich's Ataxia	\$250		4 - 6 weeks
Sequencing	\$500		6 - 12 months
Prenatal	\$600		4 days- 3 weeks
HNPCC			
Sequencing [per gene]	\$850	Multiple genes	8 - 12 weeks
MLPA screening [per kit]	\$250	MLPA + Sequencing	6 - 8 weeks
Predictive testing	\$250	\$1000 per gene	4 - 6 weeks
Huntington Disease	\$250		4 - 6 weeks
Prenatal	\$600		4 days- 3 weeks
Long QT Syndrome (LQT)			
Mutation Detection [6 genes]	\$3500 (Seq+MLPA)	6 genes including MLPA for LQT 1, 2, 3, 5 & 6.	3 - 4 months
Mutation Detection [6 genes]	\$2950 (Seq only)		3 - 4 months
LQT1 (KCNQ1)	\$890 (Seq only)		3 - 4 months
LQT2 (KCNH2)	\$1000 (Seq only)		3 - 4 months
LQT3 (SCN5A) - Brugada	\$1650 (Seq only)		3 - 4 months
LQT3 (SCN5A) - Brugada	\$1800 (Seq+MLPA)		3 - 4 months
LQT5 (KCNE1)	\$300 (Seq only)		3 - 4 months
LQT6 (KCNE2)	\$300 (Seq only)		3 - 4 months
LQT7 (KCNJ2)	\$450 (Seq only)		3 - 4 months
MLPA only (2 kits)	\$550		3 - 4 months
Predictive or confirmatory	\$250		4 - 6 weeks
Maternal Cell contamination in prenatal sample	\$600	Standard 6 markers	4 days- 3 weeks
MCAD	\$225		4 - 8 weeks
Menkes Disease			
Mutation Validation	\$600	[testing family trio]	Up to 6 months
Carrier test (known mutation)	\$250	Primers to be designed for sequencing.	TBD
Prenatal	\$600		4 days- 3 weeks
Mitochondrial Disorders			
Common Mutations		All common mutations \$250	6 - 8 weeks
Rare Mutations		All rare mutations \$250	6 - 8 weeks
Pol G ( 3 common mutations)	\$220		6 - 8 weeks

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Deletion Screening	\$400		6- 8 weeks
Mutant Load	\$275	\$170 if combined with other testing	8 - 10 weeks
Prenatal	\$660 [incl mutant load]		4 days- 3 weeks
Myotonic Dystrophy			
PCR	\$220		4 - 6 weeks
PCR + Southern	\$460		6 - 8 weeks
Prenatal [PCR+Southern]	\$660		2 - 4 weeks
MYH			
Predictive testing	\$250		4 - 6 weeks
Full gene sequencing	\$750		8 - 12 weeks
NF2			
Sequencing	\$850	MLPA + Sequencing \$1200	4 - 6 months
MLPA screening	\$250		6 - 8 weeks
Predictive testing	\$250		4 - 6 weeks
PKU (Common mutations)			
Diagnostic	[1 to 3 mutations] \$350		4 - 8 weeks
Prenatal	\$600		4 days- 3 weeks
PDH			
Carrier testing [known mutations]	\$250		4 - 8 weeks
Prenatal	\$600		4 days- 3 weeks
PMP22 (4 exons)			
Sequencing	\$450		6 - 12 months
Retinoblastoma			
Sequencing + MLPA	\$1200		12 - 16 weeks
MLPA screening	\$250		8 - 12 weeks
Predictive testing	\$250		4 - 6 weeks
Prenatal	\$600		4 days- 3 weeks
Spinal Muscular Atrophy			
Diagnostic (PCR)	\$225		4 - 6 weeks
Carrier testing (SMN1 Quantitation)	\$350		4 - 6 weeks
[parents of an affected child]	\$175/parent		
SMN2 Quantitation	\$350		TBD
SMN Mutation Analysis			
Diagnostic (Sequencing)	\$550		TBD
Carrier Testing (Sequencing)	\$250		
Prenatal [gene test + linkage]	\$600	Linkage, depending on the number of markers \$250 -\$600	1 - 3 weeks
Linkage			TBD

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Spinocerebellar Ataxia each SCA type Cascade testing Prenatal	\$250 \$250 \$600	Screen for SCA 1, 2, 3, 6, 7 Total \$500	4 - 6 weeks 4 days- 3 weeks
Setting up a specific new gene testing and validation of a research lab findings Carrier testing Prenatal	\$600 [includes testing of family trio] \$250 \$600		Up to 6 months 4 - 8 weeks 4 days- 3 weeks
SURF-1 Diagnostic Carrier Testing Prenatal	\$275 \$250 \$600		4 - 8 weeks 4 - 8 weeks 4 days- 3 weeks
Validation of research lab finding on <b>gene already tested in lab</b> (PCR primers already in lab)	\$375		4 - 6 weeks

**REFERENCE**

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