



Mitochondrial and Autoimmune Neurological Disorders Laboratory
Department of Clinical Neurosciences & Neurological Research

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Contact the laboratory or visit webpage for the specimen handling protocols and requisition/consent forms. Doctors must complete the clinical features sections to indicate specific tests. *Patients or next of kin must read, sign the consent and fill in billing details for testing to proceed.*

<https://svhm.org.au/home/health-professionals/diagnostic-services/clinical-neuroscience>

Mitochondrial diseases and some neurological disorders are hereditary which may have implications for family members. All genetic testing must be accompanied by genetic counselling.

Director of Diagnostic Lab: Professor Steve Collins
Scientists: Dr Rosetta Marotta
Ms Judy Chin

Laboratory Hours: 9am – 5pm
Mon-Fri (except public holidays).
No urgent testing unless specified.

**Mitochondrial disease
mtDNA variant screening**

Mitochondrial Encephalomyopathy Lactic Acidosis and Stroke like episodes (MELAS)	MT-TL1 m.3243A>G MT-TL1 m.3271T>C \$240.00** MT-ND5 m.13513G>A \$240.00** Sanger Sequencing PCR/RFLP*
Myoclonic Epilepsy and Ragged Red Fibres (MERRF)	MT-TK m.8344A>G Sanger Sequencing \$240.00
Neuropathy Ataxia and Retinitis Pigmentosa (NARP) Leigh's Syndrome	MT-ATP6 m.8993T>C/G Sequencing PCR/RFLP* \$240.00**
Leber's Hereditary Optic Neuropathy (LHON)	MT-ND1 m.3460A>G MT-ND4 m.11778G>A MT-ND6 m.14484T>C Multiplex PCR/RFLP* Sanger Sequencing \$240.00**
Patients with specific phenotypic/histopathologic evidence of mitochondrial disease	mtDNA gene mutations Targeted Sanger sequencing \$240.00 per gene**

Specimens:

Variant screening: any two of blood (2x 3ml EDTA), hair (10-20 follicles attached), urine (50ml -**first morning** void sent immediately), or muscle.

Muscle must be frozen immediately and sent on dry ice.

* Polymerase Chain Reaction / Restriction Fragment Length Polymorphism

Prices + GST **per specimen.

Autoimmune Encephalitis Testing

Anti-glutamate receptor (type NMDA) encephalitis	Anti-glutamate receptor (type NMDA) N-methyl-D-aspartate Indirect immunofluorescence
Limbic encephalitis Atypical psychosis	Anti-glutamate receptor (type AMPA) α -amino-3-hydroxy-5-methyl-4-isoxazol-propion acid Indirect immunofluorescence
Limbic encephalitis	Anti-GABA _B receptor γ -amino-butyric acid Indirect immunofluorescence
Limbic encephalitis	Anti-LGI 1 Leucine –rich glioma-inactivated protein 1 Indirect immunofluorescence
Neuromyotonia Morvan's Syndrome Limbic encephalitis	Anti-CASPR2 Contactin-associated protein 2 Indirect immunofluorescence
Autoimmune encephalitis	Anti-DPPX Dipeptidyl amiopeptidase-like protein Indirect immunofluorescence

Specimens: Panel of six autoantibodies screened in Serum - >1ml SST tubes or > 60 μ l of CSF sent immediately, refrigerated (4°C) or frozen
Price: **\$240.00 + GST** per specimen



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