ST VINCENT'S HOSPITAL
MELBOURNE

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

5th Floor Daly Wing,
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35 Victoria Pde Fitzroy Vic 3065
Telephone: 9231 3366
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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
Ms Maria Chiotis

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

Mitochondrial disease mtDNA mutation screening

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

Specimens:
Point mutation screening: any two of blood (2x 3ml EDTA), hair (10-20 follicles attached), urine (50ml -first morning void sent immediately), or muscle.
mtDNA deletions: 50mg muscle frozen immediately and sent on dry ice.
* Polymerase Chain Reaction / Restriction Fragment Length Polymorphism

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-hydrosy-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: Serum - 2x 8ml SST tubes or > 60 µl of CSF sent immediately, refrigerated or frozen

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Accreditation No. 13786