



ST VINCENT'S
HOSPITAL
MELBOURNE

A FACILITY OF ST VINCENT'S HEALTH AUSTRALIA



Accredited for compliance with
NPAAC standards and ISO 15189
Accreditation No. 13786



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Mitochondrial and Autoimmune Neurological Disorders Laboratory

SPECIMEN HANDLING:

All incoming patient samples are to be accompanied by a requisition and out of pocket consent forms provided by the abovementioned laboratory. Forms can be downloaded from the website:

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

The requisition form is designed to obtain all relevant information in relation to the patient to carry out the appropriate genetic tests. Ensure all information is completed. There are no Medicare or private health care rebates for genetic testing carried out by the laboratory. Please indicate if patient or institution is to be invoiced. All Victorian, interstate and overseas referred patients will be invoiced.

Genetic testing may have implications for family members and must be accompanied by genetic counselling.

Clinicians are to obtain the genetic consent upon consultation and discuss billing with the patient. The patient must sign the consent and understand the billing arrangement in order to proceed with genetic testing.

No specimens are collected by the scientific staff in the abovementioned laboratory. Please direct your patient to the nearest pathology collection centre. Please note that the requisition form is not a referral slip for specimen collection. The referring clinician must also complete centre specific pathology collection slips for the collection of any specimen. (e.g. St. Vincent's Pathology, Melbourne Pathology.)

For muscle biopsies please ensure that referral letters are sent to appropriate clinicians to book in biopsies. It is recommended that muscle samples are sent for histopathological staining before genetic testing.

Clinicians and pathology services forwarding samples to us are to be advised that it is preferable to contact our laboratory prior to dispatch. Laboratory hours are from 9.00 am to 5.00 pm Monday to Friday. The laboratory is not open on local government and federal public holidays, and there is no urgent testing without prior arrangement.

Samples Required for genetic testing:

The laboratory requisition form with the patient's signed consent for genetic testing and out of pocket billing consent form must accompany the specimen/s or the forms can be faxed or e-mailed to the laboratory.

Please also complete a designated pathology collection slip for two or more separate specimens. See below.

Suitable for genetic variant screening:

Blood: request for 2x 3 ml of whole blood (*not serum only*) in an EDTA-treated tube (usually a pink/purple capped tube) to be delivered immediately following collection. Samples can be refrigerated at 4°C, if unable to be delivered within 24 hours.

All blood sent in the wrong tube e.g. Lithium/Heparin, clot activator tubes will be rejected and a recollection requested. Blood collected in these tubes inhibit the testing.

Hair: request 10-20 hair follicles pulled from several regions of the scalp to be delivered at room temperature in a sealed sterile specimen jar. Do not cut hair or stick the follicles to adherent paper or sticky tapes as the hair follicles contain the cells with DNA required in the testing.

Urine: request 50 mls of first morning void in sterile specimen jar. Store at 4°C for no longer than 24 hours. For samples unable to be delivered on the day of collection: Centrifuge at 2000g (3000rpm) for 10 min. Carefully remove all except 200-400 µl of urine to leave behind sediment pellet and freeze -20°C and deliver on ice.

Please note that collection of urine other than the first morning void do not have sufficient cells for mtDNA extraction. For the collection of first morning void urine, the patient can be sent home with the sterile specimen jar and return to the pathology collection centre on the day of collection. All urine collected at times other than the first morning void, stored incorrectly or not delivered within the day will be rejected and a recollection requested. Micro-organisms allowed to grow in the urine inhibit the testing.

Muscle: Delivery of muscle should be pre-arranged.

Request greater than 50 mg of fresh biopsies. From within the Melbourne metropolitan area biopsies are to be delivered immediately on ice; deliveries from all other hospitals to be on dry ice. Samples not available for delivery following collection are to be stored at -70°C and delivered to the laboratory on dry ice.