

MITOCHONDRIAL DIAGNOSTIC TESTS & FEE STRUCTURE

(CURRENT AT JANUARY 2011)

MITOCHONDRIAL LABORATORY

1. Respiratory Chain (Electron Transport Chain, Oxidative Phosphorylation) Enzymes

Complex I	NADH-CoQ ₁ Oxidoreductase
Complex II	Succinate-CoQ ₁ Oxidoreductase
Complex II+III	Succinate-cytochrome c Oxidoreductase
Complex III	Decylbenzylquinol-cytochrome c reductase
Complex IV	Cytochrome c Oxidase
(plus Citrate Synthase assayed as a marker enzyme)	

Preferred sample: skeletal muscle, liver, cardiac muscle (depending on clinical history). Complex V cannot be assayed in frozen tissues. Skin fibroblasts and EBV-transformed lymphoblasts are suitable only in certain circumstances. All cases, particularly those involving cell lines require prior confirmation from the laboratory that we are willing to accept and process the sample. Our Tissue Culture Lab charges us \$200 for processing and storing each cell line received. If we have not agreed to perform diagnostic testing on a patient cell line, we will have to bill this to the sender, even if we do not proceed with testing. For cell lines that we have agreed to test, this charge is built into the fee structure.

Fees

Tissues (per patient): one tissue \$1075, two tissues \$2,150.

Cultured cells : Assay of established cell line, skin biopsy or fresh blood \$1,450

:Storage of cell line (even if no testing performed) \$200

2. Mutation analysis for Mitochondrial DNA, *POLG* and *SURF1*

These tests are performed by the VCGS Pathology Molecular Genetics Laboratory (Lab Head: Dr Desiree DuSart, ph 03 8341 6333). Request cards should specify what mutation testing is desired. If the request is not explicit, we will interpret requests in the following way:

“mtDNA mutations” will be tested for the four common point mutations listed below;

“MELAS” will be tested for the common A3243G mutation;

“MERRF” will be tested for the common A8344G mutation;

“Kearns Sayre” or “CPEO” or “Pearson syndrome” will be tested for mitochondrial deletions;

“POLG” will be tested for the three common *POLG* mutations listed below;

“SURF1” will be tested for the common *SURF1* mutation listed below.

N.B. Tissue biopsies and cell lines referred to the Mitochondrial Laboratory for enzyme analysis will not have mtDNA mutation analysis performed unless it is specifically requested.

VCGS Laboratories	Prepared by: David Thorburn	Doc No.: BG- E-133
Laboratory: Biochemical Genetics	Authorised by: David Thorburn	Version: 8
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	<u>Fee</u>
Common mtDNA point mutations include :	
mt.3243A>G, 8344A>G, 8993T>G & 8993T>C	\$250
mt.3243A>G (causes ~80% of MELAS; also diabetes, deafness, Leigh disease)	
mt.8344A>G (causes ~80% of MERRF; also Leigh disease)	
mt.8993T>G & mt.8993T>C (NARP, Leigh disease)	
Rare mtDNA point mutations (only tested when specifically requested) include :	
mt.3271T>C, 13513G>A, 8356T>C, 14459G>A, 9176T>C & 1555A>G	\$250
mt.3271T>C (MELAS)	
mt. 13513G>A (MELAS, Leigh disease)	
mt.8356T>C (MERRF, MELAS)	
mt.14459G>A (Leigh disease, dystonia)	
mt.9176T>C (Leigh disease)	
mt.1555A>G (deafness)	
mtDNA deletions tested by quantitative PCR and Long Range PCR (CPEO, Kearns Sayre, Pearson syndromes)	\$400
<i>POLG</i> mutations (3 common mutations, all apparently of ancient European origin) p.A467T, p.W748S, p.G848S	\$220
<i>POLG</i> sequencing	\$850
<i>SURF1</i> mutation (common mutation in exon 4, apparently of British origin) c.312delTCTGCCAGCC,insAT	\$275

Preferred sample: Tissue biopsy, EDTA blood sample or urine DNA, as appropriate.

3. Other Enzyme Tests

Preferred sample

Pyruvate Dehydrogenase Complex (PDHC)	skin fibroblasts
Fumarase	skin fibroblasts
Holocarboxylase Synthetase (indirect assay of multiple carboxylases)	skin fibroblasts

Fees for each enzyme: Cultured Cells: \$690

4. Prenatal Diagnosis

Enzymatic Prenatal Diagnosis

Fee for enzyme assays on CVS samples dissected free of maternal tissue \$2,500.

N.B. Enzymatic prenatal diagnosis for these disorders can only be attempted in a minority of diagnosed cases. In respiratory chain disorders, residual enzyme activity is usually substantial, and the mode of inheritance and tissue expression are often unclear. Extra control samples and long term cultures of multiple CVS and fibroblast cell lines (including fibroblasts from parents and the proband) are therefore required, increasing costs substantially. Each specific case must be discussed in advance with our laboratory to confirm that the test can be offered, and to clarify any ambiguities about test outcomes and result interpretation.

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