MitoMed Diagnostic Laboratory

2501E Hewitt Hall, University of California Irvine, Irvine, CA 92697-3940

Telephone: (949) 824 1886 Fax: (949) 824 3007 Email: mdl.lab@uci.edu

CLIA #: 05D1034314 CA State License #: CLF 332383 Website: www.mammag.uci.edu/MITOMED

Laboratory Director: Elizabeth Chao, M.D., FACMG.

Patient Information

Clinical Indication:

☐ Diagnostic ☐ Carrier Test
☐ Know familial study ☐ Positive Newborn Screening
☐ Family History

Clinical Findings: _____

Family History: _____

Patient Name (Last, First, M.): _____
Date of Birth (M/D/YY): _____ ☐Male ☐Female
Guardian Name (for minors only): _____
Address: _____
City: _____ State: _____ Zip: _____
Phone: _____

Ethnic Background:

☐African American ☐Native American ☐Caucasian ☐Hispanic
☐Mediterranean ☐Middle Eastern ☐Asian/Pacific Islander

Referring Physician Information

Name (Last, First, M.): _____
Address: _____
City: _____ State: _____ Zip: _____
Phone: _____ Fax: _____
Email: _____
MD's Signature: _____
Genetic Counselor: _____
Preferred Method of reporting: ☐Email ☐Fax ☐Mail ☐Phone

MITOCHONDRIAL DNA TESTS

(Tissue specific - Please contact laboratory regarding highest detection rate)

1004 ☐ mtDNA Neuromuscular panel (MELAS/MERRF: mtDNA 583, 3243, 3256, 3271, 3291, 3697, 4332, 8344, 8356, 8363, 12147, 13513, 13514)
1006 ☐ Leber's Hereditary Optic Neuropathy panel (mtDNA 3460, 3635, 3697, 3700, 3733, 4171, 10197, 10663, 11778, 13513, 14459, 14482, 14484, 14495, 14568)
1007 ☐ mtDNA Point Mutation Screen (57 mtDNA point mutations)

1001 ☐ Aminoglycoside related hearing loss (mtDNA 14494, 1555)
1010 ☐ Full mitochondria genome sequencing
1011 ☐ Comprehensive mitochondrial hearing loss panel (12 srRNA, tRNAser, tRNAleu, tRNAlys, MTTS)
1013 ☐ mtDNA complex 1 subunits sequencing
1014 ☐ mtDNA complex 3 subunits sequencing
1015 ☐ mtDNA complex 4 subunits sequencing
1016 ☐ mtDNA complex 5 subunits sequencing

MITOCHONDRIAL NUCLEAR GENE TESTS

2003 ☐ AD Progressive External Opthalmoplegia (PEO), Cardiomyopathy (ANT1 (SLC25A4) sequencing)
2005 ☐ POLG-related disorders: AD-PEO, SANDO, MIRAS (POLG1 sequencing)
2006 ☐ Infantile-Onset Spinocerebellar ataxia (IOSCA), AD-PEO Twinkle (PEO1) sequencing
2007 ☐ Myopathic mtDNA Depletion (TK-2 sequencing)
2008 ☐ AD PEO (POLG2 sequencing)
2009 ☐ Mitochondrial myopathy and methylmalonic aciduria (SUCLA2 sequencing)
2010 ☐ Hepatocerebral mitochondrial depletion syndrome (DGUOK sequencing)
2011 ☐ Encephelomyopathic mitochondrial depletion syndrome, AD-PEO (RRM2B sequencing)
2012 ☐ Leigh Syndrome, Complex 4 deficiency (SURF1 sequencing)
2014 ☐ Hepatocerebral mitochondria depletion syndrome (MPV17 Sequencing)
2015 ☐ MINGIE (TYMP Sequencing)
4006 ☐ Optic atrophy (DOA): OPA1
4008 ☐ Optic atrophy (DOA): OPA3

IBMPFD/ALS

5006 ☐ IBMPFD VCP Exon 5 Sequencing
5007 ☐ IBMPFD VCP Exon 3, 5, 6, 7 & 10 sequencing
5008 ☐ VCP (Valosin Containing Protein) whole gene sequencing

CYSTINOSIS

5001 ☐ CTNS 57kb Deletion Screen
5002 ☐ CTNS gene sequencing
5003 ☐ CTNS gene sequencing and 57kb deletion screen

MITOCHONDRIAL NUCLEAR GENE PANELS

4001 ☐ Leigh Syndrome panel (Full mtDNA sequencing, POLG1, SURF1)
4002 ☐ mtDNA Depletion and Multiple Deletion panel (POLG1, SUCLA2, DGUOK, TK2)
4003 ☐ Myopathic mtDNA instability panel (POLG1, RRM2B, TK2, Twinkle)
4004 ☐ Hepatocerebral mtDNA instability panel (POLG1, DGUOK, MPV17, Twinkle)
4005 ☐ Encephalomyopathic mtDNA instability panel (POLG1, SUCLA2, RR2MB)
4007 ☐ Opthalmoplegia/PEO sequencing panel (POLG1, POLG2, Twinkle, ANT1)

HEARING LOSS GENE TESTS AND PANELS

6001 ☐ Comprehensive Sensory Neural Hearing Loss Panel (6001a-6001d)
6001 a. ☐ GJB2 (Connexin 26) Sequencing
6001 b. ☐ GJB6 (Connexin 30) Deletion
6001 c. ☐ Pendred (p.Leu236Pro, p.Thr416Pro, c1001+1G>A)
6001 d. ☐ Mitochondrial genes 12SrRNA, tRNAser
6002 ☐ Connexin 30 (GJB6) Sequencing
6003 ☐ Pendred Syndrome / DFNB4 (SLC26A4 Sequencing)
1011 ☐ Comprehensive mitochondrial hearing loss panel (12 srRNA, tRNAser, tRNAleu, tRNAlys, MTTS)
1001 ☐ Aminoglycoside related hearing loss (mtDNA 14494, 1555)

CARDIOVASCULAR GENE TESTS

7006 ☐ DiGeorge Syndrome / Velocardiofacial Syndrome (TBX1)

OCCULAR DISEASES GENE TESTS

4006 ☐ Optic atrophy (DOA): OPA1
4008 ☐ Optic atrophy (DOA): OPA3
8003 ☐ Keratoconus (VSX1 (Visual System Homeobox 1) sequencing)

OTHER TESTS

8005 ☐ Abetalipoprotenemia (MTTP (Microsomal Triglyceride Transfer Protein) sequencing)
9003 ☐ DNA Extraction (Clinical/Research)
9004 ☐ Other Research Project: _____
11001 ☐ Known familial mutation confirmation (Specify gene and mutation): _____

MitoMed Diagnostic Laboratory

2501E Hewitt Hall, University of California Irvine, Irvine, CA 92697-3940

Telephone: (949) 824 1886 Fax: (949) 824 3007 Email: mdl.lab@uci.edu

CLIA #: 05D1034314 CA State License #: CLF 332383 Website: www.mammag.uci.edu/MITOMED

Laboratory Director: Elizabeth Chao, M.D., FACMG.

Billing Information

MitoMed can only accept institutional billing or cash/check payments.

Party Responsible for Payment:

☐ UC Sendouts / UC campus

UC Campus: _____
Department: _____
Recharge Account number: _____
Account Manager: _____
Phone Number: _____

☐ Institutional Billing

Referring Physician: _____
Institution: _____
Billing Address: _____
City:_____ State: _____ Zip: _____
Phone: _____
Email: _____

☐ Patient Self-Pay

Patient Name (Last, First, M.):_____
Date of Birth: _____ SSN: _____
Billing Address: _____
City:_____ State: _____ Zip: _____
Phone: _____
Email: _____

Cash Payment Options:

Check or money order must be sent at the time of sample submission to the address below. If a sample cannot be processed, payment will be refunded. Please see rejection criteria for details).

Make checks payable to: Regents of the University of California
Send checks to: Union Bank of California
PO Box 515297
Los Angeles, CA 90047

***Important: Please have patient or guardian sign our DNA consent on Page 3, and complete the Health History Checklist on Page 4**

Specimen Information

Collection Date: _____

Sample Type:

Muscle and other tissue have the highest detection rate for mtDNA mutations. Please refer to test information for appropriate tissues.

☐ **Whole Blood**
- 1-3 cc for children
- 6-9 cc for adults
- EDTA/Lavender top tubes
- (Please send at Room Temperature)

☐ **Urine**
- For MELAS testing if muscle is not available
- 30-50 mL morning urine
- Send at 4 °C or in urine cup overnight at room temperature

☐ **Buccal swabs**
- 2-4 swabs
- Assay dependant – send at room temperature

☐ **Tissue**
- Specify: _____
- *For mtDNA studies, minimum tissue: 100 mg
- No paraffin: please send frozen on dry ice.

☐ **DNA**
- Specify tissue of origin: _____
- Specify concentration: _____
- Minimum amount varies by test. Average amount is 5 micrograms. Please send at room temperature by overnight shipping. Previously extracted DNA is not recommended for full mitochondria genome sequencing.

Shipment Information

Ship all samples to:

MitoMed Diagnostic Laboratory
University of California, Irvine
2501E Hewitt Hall
Irvine, CA 92697-3940
USA

Ship all samples by overnight mail. Do not ship on Fridays. Samples may be received Monday-Friday, 8am-5pm. Call to alert laboratory of pending shipment, or email the tracking number to us.

Phone: (949) 824 1886
Email: mdl.lab@uci.edu

MitoMed Diagnostic Laboratory

2501E Hewitt Hall, University of California Irvine, Irvine, CA 92697-3940

Telephone: (949) 824 1886 Fax: (949) 824 3007 Email: mdl.lab@uci.edu

CLIA #: 05D1034314 CA State License #: CLF 332383 Website: www.mammag.uci.edu/MITOMED

Laboratory Director: Elizabeth Chao, M.D., FACMG.

Informed Consent for DNA Testing

I, _____ (Patient or legal guardian name), request and authorize MitoMed Diagnostic Laboratory to perform the requested test(s) for the person(s) list below. I acknowledge the benefits, risks, and limitations outlined below.

Patient's Name	Date of Birth	Sex	Date of Sample Collection
_____	_____	_____	_____
_____	_____	_____	_____
_____	_____	_____	_____
_____	_____	_____	_____

I understand that:

A. An inaccurate diagnosis may result if I report incorrect medical or family history information. Other sources of error may be possible, including but not limited to sample handling, contamination, or misidentification.
B. Interpreting DNA results can be complex. Therefore my results will be made available through a Genetic Counselor or my referring physician.
C. I understand that the molecular tests need to be interpreted within the context of additional clinical and laboratory findings.
D. This test is known to be a highly complex test and the performance characteristics have been validated by the MitoMed Diagnostics Laboratory but have not been approved by the FDA (Food and Drug Administration). Any results from this testing are not intended to be used as the sole means for clinical diagnosis or to treat any disease.
E. The absence of a known mutation does not guarantee the absence of the disease. For example, more than one gene may cause the disease, I may have a mutation not detectable with the technique applied, or I may have a mutation which has not previously been described and cannot be interpreted at this time. I can speak with my physician or genetic counselor to clarify the results of this testing if I have questions.
F. The MitoMed Laboratory does not return patient samples. Sometimes there is enough sample stored to request additional tests or at my request to send out samples to other institutions. Once my test result has been released, some DNA or cells that remain may be de-identified to be used for laboratory quality control or research. I can withdraw my consent at any time by calling the MitoMed Laboratory at (949) 824 1886.
G. Any additional testing must be requested by my referring physician and will incur additional charges to my insurance or to me.
H. All results and patient information are confidential but may be made available to insurance providers if needed for reimbursements.
I. My signature below indicates that I have read the above information. All my questions have been answered and my inquiries regarding the purpose of this test have been discussed and fully understood by me.

Patient or Guardian Signature: _____ **Date: _____**

I confirm that the patient understood the limitations, risks, and benefits of the DNA testing, and that the inquiries, questions, and concerns of the patient have been answered.

Physician or Genetic Counselor's name: _____ **Phone: _____**

Physician or Genetic Counselor's Signature: _____ **Date: _____**

MitoMed Diagnostic Laboratory

2501E Hewitt Hall, University of California Irvine, Irvine, CA 92697-3940

Telephone: (949) 824 1886 Fax: (949) 824 3007 Email: mdl.lab@uci.edu

CLIA #: 05D1034314 CA State License #: CLF 332383 Website: www.mammag.uci.edu/MITOMED

Laboratory Director: Elizabeth Chao, M.D., FACMG.

Patient and Family History

Patient's Name: _____ Date of Birth: _____

Maternal Ethnicity: _____ Paternal Ethnicity: _____

Consanguinity: Are the parents of the patient related to each other by blood (e.g., second cousins)? ☐ Yes ☐ No

If so, how are they related?:_____

Please describe the patient's symptoms and family history using the checklist below as a guideline. Please attach a pedigree if available:_____

_____

_____

Please describe previously abnormal tests – e.g. Metabolic tests, MRI, Echo, Muscle histology, and functional studies:

_____

_____

_____

Neurological/Muscular Symptoms - Does anyone in the family have:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	Seizures or epilepsy	☐	☐	Chronic fatigue syndrome	☐	☐	ALS (Lou Gehrig's disease)
☐	☐	Tremor	☐	☐	Recurrent headaches	☐	☐	Alzheimer's disease
☐	☐	Sensory Neuropathy	☐	☐	Recurrent vomiting	☐	☐	Paget Disease
☐	☐	Cerebral Palsy	☐	☐	Muscle pain	☐	☐	Multiple Sclerosis
☐	☐	Contractures	☐	☐	Muscle weakness	☐	☐	Fibromyalgia
☐	☐	Ataxia	☐	☐	Dysphagia	☐	☐	Muscular Dystrophy
☐	☐	Stroke-like episodes	☐	☐	Muscle wasting	☐	☐	Autoimmune Disease

Developmental Histories – Is there anyone in the family with:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	Autism	☐	☐	Learning Disabilities	☐	☐	Other
☐	☐	Developmental Delay	☐	☐	Mental Retardation			_____

Psychiatric Issues - Does anyone in the family have a psychiatric disorder, such as:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	Anxiety	☐	☐	Depression	☐	☐	Bipolar Disorder
☐	☐	OCD	☐	☐	Panic attacks	☐	☐	Schizophrenia
☐	☐	Other	☐	☐	Dementia	☐	☐	Memory Loss

Gastrointestinal & Metabolic disease – Is there anyone in the family with:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	Chronic constipation	☐	☐	Recurrent vomiting	☐	☐	Other
☐	☐	Irritable Bowel Syndrome	☐	☐	A known metabolic disorder			_____

Ophthalmologic Problems - Does anyone in the family have:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	CPEO (Opthalmoplegia)	☐	☐	Cataracts	☐	☐	Blindness
☐	☐	Ptosis (droopy eyelids)	☐	☐	Retinitis Pigmentosa	☐	☐	Color blindess
☐	☐	Macular degeneration	☐	☐	Visual field defect	☐	☐	Optic atrophy
☐	☐	Corneal deposits	☐	☐	Photophobia			

Auditory Problems – Is there anyone in the family with:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	Hearing impaired or deaf (please describe): _____						

Cardiac disease/symptoms - Does anyone in the family have:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	Chest pains	☐	☐	Shortness of breath	☐	☐	Other
☐	☐	Irregular Heart beat	☐	☐	Stroke			_____
☐	☐	Cardiomyopathy	☐	☐	Heart murmur			

Other health concerns – Is there anyone in the family with:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	Diabetes (adult or juvenile)	☐	☐	Kidney Problems	☐	☐	Short stature
☐	☐	Early childhood deaths	☐	☐	SIDS	☐	☐	Skin disorder
☐	☐	Chronic infections	☐	☐	Multiple miscarriages or infertility			
☐	☐	Cancer (please describe):_ _____						
☐	☐	Any other condition not listed here (please describe):_ _____						