



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

ID#: 05D1034314

CA State License #: CLF 332383

Website: www.mammag.uci.edu/MITOMED

Laboratory Director: Taosheng Huang, M.D., Ph.D.

Patient Information

Clinical Indication: _____

Patient Name (Last, First, M.): _____

Date of Birth (M/D/YY): _____ Male Female

Gaurdian Name (for minors only): _____

Address: _____

City: _____ State: _____ Zip: _____

Phone: _____

Ethnic Background:

African American Native American Caucasian Hispanic

Mediterranean Middle Eastern Asian/Pacific Islander

Mitochondrial DNA Tests

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

1007 mtDNA point mutation screen (mt 1555, 3243, 3271, 3460, 8344, 8993, 11778, 14459, 14484)

1004 mtDNA neuromuscular panel (MELAS/MERF: mtDNA 3243, 3271, 8344)

1006 Leber's Hereditary optic neuropathy panel (mtDNA 3460, 11788, 14459, 14484)

1008 mtDNA single mutation: _____

1010 Full mitochondria genome sequencing

1013 mtDNA complex 1 subunits sequencing

1014 mtDNA complex 3 subunits sequencing

1015 mtDNA complex 4 subunits sequencing

1016 mtDNA complex 5 subunits sequencing

1009 mtDNA large deletions/duplications/rearrangements (mtDNA instability/PEO, Kearns Sayre, Pearsons - Southern blot)

Mitochondrial Nuclear Gene Tests

4006 Optic atrophy (DOA): OPA1

4008 Optic atrophy (DOA): OPA3

2003 AD Progressive External Ophthalmoplegia (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 Encephalomyopathic 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)

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

5009 SOD1 (Superoxide Dismutase 1) whole gene sequencing

Cystinosis

5001 CTNS 57kb Deletion Screen

5003 CTNS gene sequencing and 57kb deletion screen

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 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, RRM2B)

4007 Ophthalmoplegia/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 and tRNAscr

6002 Connexin 30 (GJB6) Sequencing

6003 Pendred Syndrome / DFNB4 (SLC26A4 Sequencing)

1011 Comprehensive mitochondrial hearing loss panel (12 srRNA, tRNAscr, tRNAleu, tRNAlys)

1001 Aminoglycoside related hearing loss (mtDNA 1555)

Cardiovascular Gene Tests and Panels

7001 Holt-Oram Syndrome (TBX5)

7003 Barth Syndrome / Endocardial Fibroelastosis Type 2 / Endocardial Fibroelastosis X-linked / MGA, Type II (G4.5/Tafazzin)

7004 Noncompaction of left ventricular myocardium (G4.5/Tafazzin)

7005 Noonan Syndrome (PTPN11)

7006 DiGeorge Syndrome / Velocardiofacial Syndrome (TBX1)

7002 Nonsyndromic Congenital Heart Defect

7002 a. NKx2.5

7002 b. GATA4

Other Tests

8001 Prader-Willi Syndrome (Methylation Test)

8002 Angelman Syndrome (Methylation Test)

8003 Keratonococus (VSX1 (Visual System Homeobox 1) sequencing)

8004 Glaucoma (MYOC (Myocillin) sequencing)

8005 Abetalipoproteinemia (MTTP (Microsomal Triglyceride Transfer Protein) sequencing)

8006 ALS (Sequestosome (P62) sequencing)

10001 Y Chromosome: SRY Gene detection

9003 DNA Extraction (Research)

9004 Other Research Project: _____

11001 Known familial mutation confirmation (Specify gene and mutation): _____



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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

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.

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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:** _____



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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	
<input type="checkbox"/>	<input type="checkbox"/>	Seizures or epilepsy	<input type="checkbox"/>	<input type="checkbox"/>	Chronic fatigue syndrome	<input type="checkbox"/>	<input type="checkbox"/>	ALS (Lou Gehrig's disease)
<input type="checkbox"/>	<input type="checkbox"/>	Tremor	<input type="checkbox"/>	<input type="checkbox"/>	Recurrent headaches	<input type="checkbox"/>	<input type="checkbox"/>	Alzheimer's disease
<input type="checkbox"/>	<input type="checkbox"/>	Sensory Neuropathy	<input type="checkbox"/>	<input type="checkbox"/>	Recurrent vomiting	<input type="checkbox"/>	<input type="checkbox"/>	Paget Disease
<input type="checkbox"/>	<input type="checkbox"/>	Cerebral Palsy	<input type="checkbox"/>	<input type="checkbox"/>	Muscle pain	<input type="checkbox"/>	<input type="checkbox"/>	Multiple Sclerosis
<input type="checkbox"/>	<input type="checkbox"/>	Contractures	<input type="checkbox"/>	<input type="checkbox"/>	Muscle weakness	<input type="checkbox"/>	<input type="checkbox"/>	Fibromyalgia
<input type="checkbox"/>	<input type="checkbox"/>	Ataxia	<input type="checkbox"/>	<input type="checkbox"/>	Dysphagia	<input type="checkbox"/>	<input type="checkbox"/>	Muscular Dystrophy
<input type="checkbox"/>	<input type="checkbox"/>	Stroke-like episodes	<input type="checkbox"/>	<input type="checkbox"/>	Muscle wasting	<input type="checkbox"/>	<input type="checkbox"/>	Autoimmune Disease

Developmental Histories – Is there anyone in the family with:

Patient	Family		Patient	Family		Patient	Family	
<input type="checkbox"/>	<input type="checkbox"/>	Autism	<input type="checkbox"/>	<input type="checkbox"/>	Learning Disabilities	<input type="checkbox"/>	<input type="checkbox"/>	Other
<input type="checkbox"/>	<input type="checkbox"/>	Developmental Delay	<input type="checkbox"/>	<input type="checkbox"/>	Mental Retardation			_____

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

Patient	Family		Patient	Family		Patient	Family	
<input type="checkbox"/>	<input type="checkbox"/>	Anxiety	<input type="checkbox"/>	<input type="checkbox"/>	Depression	<input type="checkbox"/>	<input type="checkbox"/>	Bipolar Disorder
<input type="checkbox"/>	<input type="checkbox"/>	OCD	<input type="checkbox"/>	<input type="checkbox"/>	Panic attacks	<input type="checkbox"/>	<input type="checkbox"/>	Schizophrenia
<input type="checkbox"/>	<input type="checkbox"/>	Other	<input type="checkbox"/>	<input type="checkbox"/>	Dementia	<input type="checkbox"/>	<input type="checkbox"/>	Memory Loss

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

Patient	Family		Patient	Family		Patient	Family	
<input type="checkbox"/>	<input type="checkbox"/>	Chronic constipation	<input type="checkbox"/>	<input type="checkbox"/>	Recurrent vomiting	<input type="checkbox"/>	<input type="checkbox"/>	Other
<input type="checkbox"/>	<input type="checkbox"/>	Irritable Bowel Syndrome	<input type="checkbox"/>	<input type="checkbox"/>	A known metabolic disorder			_____

Ophthalmologic Problems - Does anyone in the family have:

Patient	Family		Patient	Family		Patient	Family	
<input type="checkbox"/>	<input type="checkbox"/>	CPEO (Ophthalmoplegia)	<input type="checkbox"/>	<input type="checkbox"/>	Cataracts	<input type="checkbox"/>	<input type="checkbox"/>	Blindness
<input type="checkbox"/>	<input type="checkbox"/>	Ptosis (droopy eyelids)	<input type="checkbox"/>	<input type="checkbox"/>	Retinitis Pigmentosa	<input type="checkbox"/>	<input type="checkbox"/>	Color blindness
<input type="checkbox"/>	<input type="checkbox"/>	Macular degeneration	<input type="checkbox"/>	<input type="checkbox"/>	Visual field defect	<input type="checkbox"/>	<input type="checkbox"/>	Optic atrophy
<input type="checkbox"/>	<input type="checkbox"/>	Corneal deposits	<input type="checkbox"/>	<input type="checkbox"/>	Photophobia			

Auditory Problems – Is there anyone in the family with:

Patient	Family		Patient	Family		Patient	Family	
<input type="checkbox"/>	<input type="checkbox"/>	Hearing impaired or deaf (please describe): _____						

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

Patient	Family		Patient	Family		Patient	Family	
<input type="checkbox"/>	<input type="checkbox"/>	Chest pains	<input type="checkbox"/>	<input type="checkbox"/>	Shortness of breath	<input type="checkbox"/>	<input type="checkbox"/>	Other
<input type="checkbox"/>	<input type="checkbox"/>	Irregular Heart beat	<input type="checkbox"/>	<input type="checkbox"/>	Stroke			_____
<input type="checkbox"/>	<input type="checkbox"/>	Cardiomyopathy	<input type="checkbox"/>	<input type="checkbox"/>	Heart murmur			

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

Patient	Family		Patient	Family		Patient	Family	
<input type="checkbox"/>	<input type="checkbox"/>	Diabetes (adult or juvenile)	<input type="checkbox"/>	<input type="checkbox"/>	Kidney Problems	<input type="checkbox"/>	<input type="checkbox"/>	Short stature
<input type="checkbox"/>	<input type="checkbox"/>	Early childhood deaths	<input type="checkbox"/>	<input type="checkbox"/>	SIDS	<input type="checkbox"/>	<input type="checkbox"/>	Skin disorder
<input type="checkbox"/>	<input type="checkbox"/>	Chronic infections	<input type="checkbox"/>	<input type="checkbox"/>	Multiple miscarriages or infertility			
<input type="checkbox"/>	<input type="checkbox"/>	Cancer (please describe): _____						
<input type="checkbox"/>	<input type="checkbox"/>	Any other condition not listed here (please describe): _____						