Mitomed Diagnostic Laboratory

MITOMED
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ITOWIKI

University of California Irvine ▪ 2014 Hewitt Hall, Irvine CA 92697-3940
Telephone (949) 824-1886 Fax (949) 824-3007
email: mdl.lab@uci.edu http://mitomed.bio.uci.edu
CLIA # 05D1034314 CA State License: CLF 332383

Patient Information

Clinical Indication: _____
Patient Name: (Last, First, Middle): _____
DOB (M/D/Y): _____ Sex: ☐M ☐F
Guardian Name (for minor patients only): _____
Address:_____
City: _____ State: ____ ZIP: _____
Phone: _____

Ethnic Background

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

Mitochondrial DNA Testing

(Tissue Specific- please call laboratory if you need input regarding sending samples for highest detection rate)

- ☑ **1007** mtDNA mutation screen of positions: 1555, 3243, 3271, 3460, 8344, 8993, 11778, 14459, 14484
- ☑ **1009** mtDNA Large Deletions/Duplications/Rearrangements (Kearns Sayre, Pearson's-Southern Blot)
- ☐ **1012** mt DNA Instability/PEO, multisystemic disorder screen (muscle samples only-Southern Blot/LX-PCR)
- ☐ **1010** Full mtDNA 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
- ☐ **1001** Aminoglycoside related hearing loss
- ☐ **1011** Comprehensive Maternally inherited hearing loss panel
- ☐ **1006** Maternally inherited optic neuropathy panel
- ☐ **1004** mtDNA neuromuscular Panel (MELAS/MERRF/NARP) (send Urine unless tissue available)
- ☐ **1008** mtDNA single Mutation : Specify Mutation_____

Mitochondrial Nuclear Gene Test Panels

- ☐ **4001** Leigh Syndrome Panel: MT-DNA 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
- ☐ **4006** Hereditary Optic Neuropathy / Optic Atrophy Panel OPA1, MT-DNA Sequencing
- ☐ **4007** Opthalmoplegia / PEO sequencing Panel POLG1, POLG2, Twinkle, ANT1

Mitochondrial Functional Tests

- ☐ **4008** Electron-transport-chain-studies (frozen muscle homogenate)

Research Support

- ☐ **3001** Generation of Cybrids (research purposes only-no CPT codes)
- ☐ **3003** mtDNA Haplotyping (research purposes only-no CPT codes)

Referring Physician Information

Name: (Last, First, Middle):
_____
Address: _____ City_____
State: ____ ZIP: _____ Phone:_____
Fax #: _____ E-mail Address:_____
Physician's signature:_____
Genetic Counselor Contact Info:_____

Preferred Method of reporting: email☐ Fax☐ Mail☐ phone☐

Name of Institution to be Billed:_____

PLEASE HAVE PATIENT OR GUARDIAN SIGN DNA Consent! Attach previous laboratory findings / Pedigree and family history for test interpretation purposes.

Shipment Information

Ship all samples to:
Mitomed Diagnostic Laboratory, University of California Irvine, 2014 Hewitt Hall, Irvine CA 92697-3940, USA
by overnight mail. (Do not ship on Fridays) Samples may be received Monday-Friday from 8am to 5pm. Call to alert laboratory of pending shipment, or email the tracking number to us
Phone (949) 824-1886, Fax (949) 824-3007 mdl.lab@uci.edu

Specimen Information

Sample type: (muscle and other tissue has the highest detection rate for mtDNA mutations, please refer to test information for appropriate tissue)

☐ **Whole blood**
For DNA: 1 -3cc children, 6-9cc adults (EDTA /lavender Top tube) send at Room Temperature (RT) or 4°C ,
For Cybrids: 8cc in dark green top (Sodium Heparin) tube at Room Temperature, mail immediately by overnight shipping

☐ **Urine (for MELAS testing if muscle not available)**
30-50ml morning urine, send at 4°C or RT in urine cup overnight

☐ **Buccal Swabs: 2-4 swabs- assay dependent (Room temp.)**

☐ **Tissue: specify: _____ (for mtDNA studies: minimum 30mg, for Electron-transport-chain studies: minimum 100mg; No paraffin; please send frozen on dry ice)**

☐ **DNA: specify tissue of origin: _____ (Minimum amount varies by test, average 5 micrograms. Please send at Room Temperature by overnight shipping. Previously extracted DNA is not recommended for full mt DNA genome sequencing)**

Date of sample collection (M/D/Y) _____/___/____ Time_____
Collected by: _____

For Laboratory use only:
Date sample received (M/D/Y) _____/___/____ Time_____
Received by: _____

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Single Gene Tests

Test Code	Name of Disorder	Gene
☐ 2001	Autosomal Dominant (AD) Optic Atrophy	**OPA1** sequencing
☐ 2003	AD Progressive External Opthalmoplegia (PEO), cardiomyopathy	**ANT-1 (SLC25A4)** sequencing
☐ 2005	POLG related Disorders: Alpers, 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,	**SURF-1** sequencing
☐ 2014	Hepatocerebral Mitochondrial Depletion Syndrome	**MPV17** sequencing
☐ 5001	Cystinosis	**CTNS** 57kb deletion screen
☐ 5003	Cystinosis	**CTNS** gene Sequencing
☐ 5006	Inclusion Body Myopathy with Paget's disease +/- Frontotemporal Dementia (IBMPFD)	**VCP** (Valosin containing Protein) whole gene sequencing
☐ 5007	IBMPFD	**VCP** exon 5 sequencing
☐ 5008	IBMPFD	**VCP** exon 3, 5, 6, 10 sequencing

Custom DNA Testing

☐ 9001a	Known Familial Mutation sequencing, dominant genes (for genes on the Mitomed test menu)
☐ 9001b	Known Familial Mutation sequencing, recessive genes (for genes on the Mitomed test menu)
☐ 9002a	Validation of research results sequencing, dominant genes (for genes not on the Mitomed test menu)
☐ 9002b	Validation of research results sequencing, recessive genes (for genes on the Mitomed test menu)

Mitomed can also design other custom molecular assays tailored to our clients' needs. Please contact us for details.

IMPORTANT: Please have Patient or Guardian sign our DNA Consent on page 3, complete the Institutional Billing Form as well as the health history checklist on pages 4 and 5.

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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) listed below. I acknowledge the benefits, risks and limitations outlined below.

Patient's name	Date of Birth	Sex	Date of 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 results 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 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 reimbursement.

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, concerns and questions of the patient have been answered.

Physician or Genetic Counselor's Name _____
Signature _____ Date _____ Phone _____

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BILLING INFORMATION:

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

Party Responsible for payment:

- ☐ **UCI Sendouts/Other UC Campus**
- ☐ **Institutional Billing**
- ☐ **Patient Self Pay**

Patient Information

Patient Name (Last, First, Middle Initial): _____
Date of Birth: _____ **SSN or ID:** _____
Address: _____
City, State, Zip: _____
Phone:_____ **E-mail:** _____

Institution/Referring Physician Information

Referring Physician or Institution: _____

Billing address: _____
City, State, Zip: _____
Contact person: _____
Phone/E-mail: _____
Clinical Diagnosis/ICD-9 Code: _____

Cash Payment Options: Check or money order must be sent at the time of sample submission, to the address below. If a sample can not be processed payment will be refunded (please see rejection criteria for details).

Please make check payable to: **Regents of the University of California**
Send checks to: **Union Bank of California**
PO Box 515297
Los Angeles 515297

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Patient's name _____ Birthdate:_____ Ethnicity (maternal):_____(paternal):_____

Sperm or Egg donor?: ☐ No ☐ Yes
Consanguinity: Are the parents of the patient related to each other by blood (e.g. second cousins) ? ☐ No ☐ Yes
If so, how are they related?_____

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

_____

_____

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

_____

_____

Neurologic/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 or migraines	☐	☐	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 _____
☐	☐	Obsessive-compulsive disorder	☐	☐	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 (ophthalmoplegia- weak eye muscles)	☐	☐	Cataracts	☐	☐	Blindness
☐	☐	Ptosis (droopy eyelids)	☐	☐	Retinitis pigmentosa	☐	☐	Color blindness
☐	☐	Macular degeneration	☐	☐	Visual Field Defect	☐	☐	Optic Atrophy
☐	☐	Corneal Deposits	☐	☐	Photophobia			

Auditory Problems: Is there anyone in the family who is:

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

Cardiac Disease/Symptoms:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	Chest Pains	☐	☐	Shortness of breath	☐	☐	Other:
☐	☐	Irregular Heart Beat	☐	☐	Stroke			
☐	☐	Cardiomyopathy	☐	☐	Heart Murmur			

Other health concerns: Is there any family history of:

Patient	Family		Patient	Family		Patient	Family	
☐	☐	Diabetes (Adult or Juvenile type)	☐	☐	Kidney problems	☐	☐	Short stature
☐	☐	Early childhood deaths	☐	☐	SIDS	☐	☐	Skin disorder
☐	☐	Chronic infections (bacterial or viral	☐	☐	Multiple miscarriages or infertility			

☐ ☐ Cancer: ☐ Hematological _____ Type _____ Age of onset_____ Current status_____
☐ Solid _____ Type _____ Age of onset _____ Current status_____

☐ ☐ Any other condition not listed above:_____