MEDomics

MitoDx™

The FIRST Next Generation sequencing test of the mitochondrial genome for the diagnosis of mitochondrial disease

MitoDx offers unparalleled sensitivity. Using Next Generation Sequencing (SOLiD platform), heteroplasmy levels down to 1% can be detected. Pathogenic mutations found in very low levels in non-invasive tissues, like blood and saliva, provide a definitive result for the physician and the patient, guiding you to the most effective treatments.

What is mitochondrial disease?

Mitochondrial diseases are disorders of energy metabolism caused in many cases by mutations in the mitochondrial DNA (mtDNA).

How does mitochondrial disease manifest itself?

Organ System	*Clinical Presentation*	
Brain/CNS	• Seizures • Developmental delay • Stroke and stroke like episodes • Migraine • Mental retardation	• Dementia • Atypical cerebral palsy • Psychiatric episodes • Atypical autism
GI	• Severe dysmotility • Pseudo obstruction • Constipation	• Cyclic vomiting • GERD
Muscle	• Myopathy • Weakness • Exercise intolerance	• Hypotonia • Cramping • Muscle pain
Heart/Cardiac	• Conduction block	• Cardiomyopathy
Nerves	• Neuropathy • Weakness • Absent reflexes	• Sweating • Poor thermo-regulation • Fainting spells
Eye/Optic	• Visual loss and blindness • Ophthalmoplegia (Chronic/Progressive)	• Retinitis Pigmentosa • Nystagmus • Ptosis
Hearing	• Deafness	• SensoriNeural Hearing loss
Pancreas	• Diabetes	• Exocrine pancreatic failure (inability to make enzymes)
Liver	• Liver failure	• Hypoglycemia
Systemic	• Short stature • Failure to thrive • Lactic acidosis	• Air hunger • Respiratory distress

When to suspect mitochondrial disease:

1) Suspected maternal inheritance
2) Phenotypically different siblings (one with migraines, one with myopathy, etc.)
3) Three or more organ systems involved
4) Chronic disease “flare up” after stress or infection
5) Autism spectrum disorder plus (Autism and epilepsy, etc.)
6) Dysautonomic or “functional” symptoms not otherwise explained

Myths about mitochondrial disease:

1) It is a rare disorder. In fact, mitochondrial disease affects more people than cystic fibrosis and muscular dystrophy combined. (1 in 2,000-3,000 individuals).
2) It is a childhood disease. Mitochondrial disease affects many adults and may be a contributing factor in Parkinson’s Disease, Progressive External Ophthalmoplegia, Alzheimer's and some forms of cancer.
3) To diagnose mitochondrial disease, a muscle biopsy is required. Actually, due to advances in genetic testing, MEDomics can often detect mitochondrial disease with a blood or cheek swab.

Why order the MitoDx test from MEDomics?

1) **Unparalleled sensitivity of testing:** Using Next Generation Sequencing (SOLiD platform), MEDomics is able to detect heteroplasmy levels down to 1%.
2) **Unsurpassed bioinformatics:** The MEDomics team, led by Dr. Steve Sommer MD, PhD, has been at the forefront of genetic testing and data interpretation for two decades.
3) **Genetic counseling:** Each result is followed up by a consult from a specialized mitochondrial genetic counselor.
4) **The least invasive testing available:** MEDomics can use blood, buccal swab or saliva samples. Doing a MitoDx test before a muscle biopsy is less traumatic and much less costly to the patient and institution.

For more information:

MEDomics, LLC
426 North San Gabriel Avenue
Azusa, CA 91702 USA

www.MEDomics.com
info@MEDomics.com

(626) 804-3645 voice
(626) 529-0907 fax

MEDomics is proud to be a partner with MitoAction and supports their educational mission.