

RED-FLAG SYMPTOMS

Neurologic

- Cerebral stroke-like lesions in non-vascular pattern
- Basal ganglia disease
- Encephalopathy
- Neurodegeneration
- Epilepsia partialis continua
- Myoclonus
- Ataxia
- MRI consistent w/Leigh's disease
- Characteristic MRS peaks
 - Lactate peak at 1.3 ppm
 - Succinate peak at 2.4 ppm

Cardiovascular

- Hypertrophic cardiomyopathy with rhythm disturbance
- Unexplained heart block
- Cardiomyopathy with lactic acidosis
- Dilated cardiomyopathy with muscle weakness
- Wolff-Parkinson-White arrhythmia

Ophthalmologic

- Retinal degeneration
- Ophthalmoplegia/paresis
- Fluctuating, dysconjugate eye movements
- Ptosis
- Sudden or insidious onset optic neuropathy/atrophy

Gastroenterologic

- Unexplained or valproate-induced liver failure
- Severe dysmotility
- Pseudo-obstructive episodes

Other

- Unexplained hypotonia, weakness, failure to thrive and a metabolic acidosis in infant or young child.
- Exercise intolerance disproportionate to weakness
- Hypersensitivity to general anesthesia
- Episodes of acute rhabdomyolysis

Information courtesy of Mitochondrial Medicine Society (www.mitosoc.org)

Think mitochondrial disease when three or more organ systems are involved.

INITIAL EVALUATION

Metabolic Screening in Blood and Urine (all patients)

- Basic chemistries
- Complete blood count
- Blood lactate, pyruvate, L:P ratio
- Quantitative plasma amino acids
- Liver enzymes & ammonia
- Creatinine kinase (CPK)
- Plasma acylcarnitine analysis
- Quantitative urine organic acids

Characterize Systemic Involvement (all patients)

- Echocardiogram
- Ophthalmologic exam
- Brain MRI
- Electrocardiogram (EKG)
- Audiology testing

Metabolic Screening in Spinal Fluid

(patients with neurological symptoms)

- Lactate & pyruvate
- Routine studies including cell count, glucose and protein measurement
- Quantitative amino acids

Clinical Neurogenetics Evaluation

(patients with developmental delays)

- Karyotype
- Child neurology consultation
- Fragile X test
- Genetics consultation

COULD IT BE MITOCHONDRIAL DISEASE?

**A QUICK-REFERENCE GUIDE
FOR HEALTHCARE PROFESSIONALS**

- ***Mitochondrial disease is the body's inability to turn food into the energy needed to sustain life. There is no cure...yet.***
- ***Mitochondrial disease is under-diagnosed, and it's rarity is questionable. Every 30 minutes, a child is born who will develop a mitochondrial disease by age 10.***
- ***Diagnosing mitochondrial disease can be a nightmare. Many experts refer to it as a "notorious masquerader" because it wears the "mask" of many different illnesses.***
- ***Adult onset of mitochondrial disease can result in drastic changes from active lifestyle to debilitating illness in a short amount of time.***
- ***The United Mitochondrial Disease Foundation promotes research and education for the diagnosis, treatment and cure of mitochondrial disorders and provides support to affected individuals and families.***

**FOR MORE INFORMATION
ON MITOCHONDRIAL DISEASE CONTACT:**



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