

## Myths and Facts About Mitochondrial Diseases

### MYTH

All mitochondrial diseases are known by acronym abbreviations (eg, MELAS, MERRF, NARP, LHON)

### FACT

Acronyms were commonly used when these disorders were first described. Today, the naming of mitochondrial disorders is evolving. Mitochondrial disorders are currently named by any of the following methods:

- By acronym descriptions (still in use although most people with a mitochondrial disorder do not have an "acronymic-named" disorder)
- By a name based on a person who described the disease
- By a name based on a specific genetic mutation
- By a name based on a microscopic description of tissue, or
- By a name based on the deficient enzyme

It is important to note that the labels given this disorder do not, in and of themselves, predict the long-term outcome or alter treatment.

### MYTH

Mitochondrial diseases are inherited only from your mother

### FACT

The current thinking is that most mitochondrial diseases are the result of one or more complex inheritance patterns. Most mitochondrial diseases are the result of mutations (changes) in DNA located in the nucleus of the cell. Only mitochondrial disorders caused by mutations in the mitochondrial DNA (a specific structure in living cells, located outside the nucleus) are exclusively inherited from mothers. Another source of mitochondrial disorders, affecting a large percentage of patients, are poorly functioning mitochondria that become that way:

- as a consequence of another disease process (including other chromosomal disorders)
- as a result of exposure to toxins or viruses
- as a result of other inherited genetic mutations that are not disease-causing until "triggered" by some other genetic factor

## **MYTH**

**Mitochondrial disease is a childhood disease**

### **FACT**

Although mitochondrial disorders are commonly seen in infants and children, they can occur at any age.

## **MYTH**

**An individual with mitochondrial disease has mental retardation, growth problems, and/or seizures**

### **FACT**

Only some individuals have these developmental problems. Patients' symptoms can range from extremely mild to severe, involve one or more body systems, and can emerge at any age. The brain, muscles, heart, liver, nerves, eyes, ears and kidneys are the organs and tissues most affected. Most patients' symptoms fluctuate over the course of their illness -- at some times experiencing no or few symptoms while at other times experiencing many and/or severe symptoms. Even family members with the same disorder can experience vastly different symptoms.

## **MYTH**

**Since mitochondrial diseases are incurable, no treatments can be given to these patients**

### **FACT**

Even though these disorders are long term and incurable, treatments are available. Early treatment of symptoms can reduce their impact and limit further disability. Avoiding certain medications and stressful situations that worsen symptoms is also helpful. Certain medications and supplements may improve mitochondrial disease-related symptoms - just as they do for other incurable diseases -- such as diabetes and emphysema.

## **MYTH**

**Patients with mitochondrial disease all have elevated lactic acid levels in their blood**

### **FACT**

An elevated lactic acid level, along with other symptoms, typically does indicate a mitochondrial problem and requires further investigation. However, elevated lactic acid levels are not seen in all types of mitochondrial diseases. In making the diagnosis, your doctor will look for other signs of mitochondrial disorders in blood, urine, and spinal fluid samples.

## **MYTH**

**A muscle biopsy is the "gold standard" for diagnosis of mitochondrial disease**

### **FACT**

Although the muscle biopsy is a powerful diagnostic tool, it should not be considered a "gold standard." Examination of a biopsy includes microscopic evaluation, enzyme testing, and genetic testing. Although all

U.S. labs that offer muscle biopsy meet strict laboratory guidelines, there is no agreed-upon standard approach for enzyme testing. Furthermore, a muscle biopsy with full analysis costs well over \$10,000 and poses both surgical and anesthetic risks. In some patients, the diagnosis can be made based on clinical symptoms and a positive blood test (identifying a genetic mutation) or a combination of clinical findings and other non-invasive testing - in either case, a muscle biopsy is not necessary. Finally, since biopsy results usually do not alter the long-term outcome or treatment considerations, some specialists and patients choose to treat without the need for a muscle biopsy.

## MYTH

**A muscle biopsy is a muscle biopsy no matter where and how it is done**

## FACT

Muscle removed for biopsy can be tested in many ways. For example, enzyme testing can be done on either ground-up muscle or on mitochondria extracted from muscle. Testing on extracted mitochondria is performed in only a few medical center laboratories and must be performed immediately. This procedure is known as a "fresh biopsy." In an alternative procedure, called a "frozen biopsy," the muscle is quickly cooled and stored at -80 degrees Celsius for testing at an outside facility. The scientific community is currently debating the advantages of testing "fresh vs frozen" mitochondria. Some evidence indicates that the "fresh biopsy" may be the superior method. Other types of mitochondrial testing of the muscle biopsy may need to be conducted; a limited number of laboratories offer such testing.

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