Mitrochondrial Disease

- Information for patients, parents and families

Neuromuscular and Neurometabolic Clinic
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What is mitochondria?

- Mitochondria are tiny structures inside nearly every cell in the body.
- Their main role is to make energy for daily life and growth.

What is mitochondrial disease?

- Mitochondrial disease refers to a group of inherited disorders.
- A gene mutation affects the mitochondria and they fail to make enough energy for cells to function.
- When mitochondria fail, cells can become stressed, damaged or die.
- Depending on the number and location of the affected cells, many symptoms are possible and range from mild to severe.
- Each person with Mitochondrial disease may have different symptoms, even within the same family.
- Mitochondrial disease may affect one person or many people in a family.

How do you get it?

- Most people with Mitochondrial disease were born with it. This is called primary Mitochondrial disease. Symptoms may appear shortly after birth or later in life.
- Mitochondrial disease is not infectious. You can't "catch it" from another person.
- It is not caused by something you did. There is no way to prevent it.

What are the symptoms?

- Most people with Mitochondrial disease have several of these symptoms:
  - developmental delay
  - dementia
  - movement disorder (dystonia)
  - seizures
  - migraines or strokes
  - muscle weakness or pain
  - poor muscle tone
  - poor balance
  - unable to tolerate exercise
  - fatigue (usually with activity)
  - vomiting
  - abdominal pain
  - constipation
  - problems with vision
    (low vision, droopy eyes, impaired eye movement)
  - hearing loss
  - heart enlargement
  - liver or kidney disease (usually only in children)

How is the diagnosis made?

- It takes many tests and procedures to confirm the diagnosis.
- Diagnostic testing may include:
  - testing blood and urine samples
  - muscle biopsy
  - genetic testing
  - magnetic resonance imaging (MRI) and magnetic resonance spectroscopy (MRS)
  - exercise and strength testing

How is it treated?

- The goal of treatment is to ease symptoms, improve function and slow down the progression of the disease.
- Treatment is not the same for everyone. It is designed for each person.
- Treatment consists of a combination of therapies, including diet, exercise, medications and vitamin supplements.

What is the prognosis?

- The outcomes vary from person to person.
- Many people with Mitochondrial disease live productive and normal lives.
- Some people have few symptoms or learn to adapt. Others are severely affected and Mitochondrial disease shortens their lives.
- Our team and others around the world are continually working to improve the quality and duration of life for people with Mitochondrial disease.
- Through grants and donations, we have made strides towards improved treatments. This is the main goal of the clinic and research program at the Neuromuscular and Neurometabolic Clinic.
- Research is underway to find better treatments and ultimately a cure.

How can I help?

- Learn more about Mitochondrial disease.
- Talk about Mitochondrial disease with your family, friends and in your community.
- Offer to have your name and contact number added to our parent support group.
- Support research to better understand the disease, find better treatments and search for a cure.

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