Mitochondrial Disease

What are mitochondria?

- Mitochondria are tiny organelles found in almost every cell in the body.
- They are known as the "powerhouse of the cell."
- They are responsible for creating more than 90% of cellular energy.
- They are necessary in the body to sustain life and support growth.
- They are composed of tiny packages of enzymes that turn nutrients into cellular energy
- Mitochondrial failure causes cell injury that leads to cell death. When multiple organ cells die there is organ failure.

What is Mitochondrial Disease?

- Mitochondrial disease is a chronic, (progressive), genetic disorder that occurs when the mitochondrial of the cell fail.
- The incidence about 1:3000-4000 individuals in the US. This is similar to the incidence of cystic fibrosis of caucasian births in the U.S.
- There are more than 40 different types of mitochondrial disease.
- Mitochondrial disease is inherited in a number of different ways
- Mitochondrial disease presents very differently from individual to individual.
- There may be one individual in a family or many individuals affected over a number of generations.

What are symptoms of mitochondrial disease?

- Loss of muscle coordination, muscle weakness
- Neurological problems, seizures
- Visual and/or hearing problems
- Developmental delays, learning disabilities
- Heart, liver or kidney disease
- Gastrointestinal disorders, severe constipation
- Diabetes
- Increased risk of infection
- Thyroid and/or adrenal dysfunction
- Autonomic dysfunction
- Neuropsychological changes characterized by confusion, disorientation and memory loss.

How is mitochondrial disease diagnosed?

- There is no reliable and consistent means of diagnosis.
- Diagnosis can be made by one of the few physicians that specializes in mitochondrial disease.
- Diagnosis can be made by blood DNA testing and/or muscle biopsy but neither of these tests are completely reliable.

How is mitochondrial disease treated?

- Treatment consists of vitamin therapy and conserving energy
- The goal is to improve symptoms and slow progression of the disease.
- Conserve energy
- Pace activities
- Maintain an ambient environmental temperature
- Avoid exposure to illness
- Ensure adequate nutrition and hydration.

What are the challenges of living with mitochondrial disease?

- Affects multiple organs, affects multiple family members, affects multiple generations.
- Lack of awareness and understanding of the disease
- Families are continuously forces to expend their very limited energy to
- explain their disease, advocate for themselves and fight for services.
- Mitochondrial disease is often an " invisible disease."
    - Good day – patients look fine and healthy. They have more energy and appear rested.
    - Bad day - - patients appear tired to significantly ill. They are obviously fatigued and/or have significant illness. Repeated "bad days"often lead to decompensation and patients have difficulty returning to baseline.
- Mitochondrial disease is unpredictable. Day to day, hour to hour patients can develop symptoms and their stability can be threatened.
- Difficulties establishing a diagnosis interfere with a patient's ability to obtain adequate recognition, medical care, adequate insurance coverage, healthcare supports and disability services.
- Lack of understanding of the disease and misinterpretation of symptoms can lead to misdiagnosis. Further progression of symptoms can occur if the symptoms are missed and opportunities for treatment and support are not recognized.
- An individual can become symptomatic at any time in life despite the fact that it is inherited.
- It is difficult to diagnose.

What is the prognosis?

- The prognosis is variable. Some people live a normal life and are minimally affected, others can be severely compromised with the disease.
- It is completely individualized
- The prognosis is unpredictable.

Future Goals

- To develop a better understanding of the biology of mitochondria.
- To learn more about mitochondrial disease in human beings.
- To refine diagnostic methods.
- To improve and expand treatment options.
- To educate the general public and medical arena.
- To improve the day to day life of individuals living with mitochondrial disease.