Mitochondrial Disorders

What are mitochondrial disorders?

- Mitochondrial disorders are a diverse group of diseases caused by damage to small structures found in human cells that are essential in converting food to energy. The result is decreased energy production and associated symptoms.
- Mitochondria are the "furnace" of the body, producing energy for all the body's functions. There are many different types of mitochondrial disorders. Each disorder affects the hundreds of chemical reactions required to convert food and oxygen into energy. Some specific disorders include
  - **KSS (Kearns-Sayre syndrome):** Onset of KSS usually occurs before the age of 20 years. Symptoms include eye-movement problems, droopy eyelids, muscle weakness, short stature, hearing loss, loss of coordination, heart problems, cognitive delays, and diabetes.
  - **MERRF (myoclonus epilepsy with ragged-red fibers):** Symptoms include seizures, loss of coordination, short stature, buildup of lactic acid in the blood, difficulty speaking, dementia, and muscle weakness.
  - **MELAS (mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes):** Symptoms include muscle weakness, stroke-like events, eye-muscle paralysis, and cognitive impairment.
  - **LHON (Leber hereditary optic neuropathy):** Leber hereditary optic neuropathy causes progressive loss of vision that results in various degrees of blindness.
  - **Leigh disease:** This degenerative brain disorder is usually diagnosed at a young age (eg, before age 2 years [ie, 24 months]). Deterioration is often rapid, with symptoms such as seizures, dementia, feeding and speech difficulties, respiratory dysfunction, heart problems, and muscle weakness. Prognosis is poor, with death typically occurring within a few years of diagnosis.
  - **Pearson syndrome:** Difficulty gaining weight, diarrhea, and enlarged liver are signs of Pearson syndrome.

How common are they?

Approximately 1,000 to 4,000 children are born with a mitochondrial disease in the United States each year. Typically, by the age of 10 years, approximately 1 in 4,000 children in the United States is diagnosed as having a mitochondrial disease.

What are some common characteristics of children with mitochondrial disorders?

- Some of the more common symptoms of mitochondrial diseases by organ system include
  - **Brain:** Confusion, memory loss, headaches, seizures, developmental delays, and stroke-like episodes.
  - **Nerves:** Pain caused by nerve abnormalities (neuropathic pain), gastrointestinal problems linked to nerve abnormalities, abnormal sweating, and fainting.
  - **Skeletal muscles:** Muscle weakness, muscle cramping, muscle pain, loss of coordination, exercise intolerance, and poor growth.
  - **Liver:** Liver failure.
  - **Heart:** Heart muscle weakness and disturbed electrical signals in the heart (called *heart block*).
  - **Kidneys:** Abnormalities that cause difficulty with absorbing nutrients and electrolytes back into the body.
  - **Ears:** Hearing loss.
  - **Eyes:** Eye-muscle paralysis and progressive loss of vision.
  - **Pancreas:** Diabetes and pancreatic failure.
- Other symptoms include failure to thrive in infants, poor growth, short stature, fatigue, respiratory disorders, swallowing difficulties, and increased risk of infection.

Who might be on the treatment team?

- Children with mitochondrial disorders usually receive a diagnosis from a neurologist or a geneticist (or both). They may also see pediatric doctors who specialize in cardiology, gastroenterology, ophthalmology, urology, and surgery. Pediatric palliative care doctors often help children and families with pain and the stress of a severe chronic illness.
- Physical, occupational, and speech-language therapies may be needed to help these children achieve normal milestones.
- Sometimes, surgery is necessary for these children because of the condition. Nurses who work with surgeons can be very helpful if a child with a mitochondrial disorder needs a gastrostomy tube, a vesicostomy (bladder) tube, or a central venous catheter (for intravenous nutrition).
- Children who are younger than 3 years (ie, 36 months) may receive therapies through *early intervention* services.
- For children 3 years and older, *special education and related services* are available through public schools to provide the accommodations necessary for school achievement and adaptation.

Mitochondrial Disorders (*continued*)

What adaptations may be needed?

Care Plans may include

- Adaptive equipment to support the gross-motor, fine-motor, and speech development of these children when they are younger. Many of these children tire easily, so an adaptive wheelchair or stroller is helpful.
- Many children will appear younger than their same-age peers because of their difficulties with growth. Be sure to take their ages and normal intelligences into account as you interact with them.
- Many children are at risk for teasing because of their physical appearances. Work to foster self-confidence with the child as well as understanding among the child's classmates.

What should be considered an emergency?

Notify parents/guardians immediately for

- Unexplained fatigue or lethargy
- Vomiting
- Fever

What are some resources?

- Centers for Disease Control and Prevention: "Mitochondrial Disease - Frequently Asked Questions" (Web page), www.cdc.gov/ncbddd/autism/mitochondrial-faq.html
- Encyclopedia of Children's Health: "Mitochondrial Disorders" (Web page), www.healthofchildren.com/M/Mitochondrial-Disorders.html#ixzz3cfhAtWpx
- National Institute of Neurological Disorders and Stroke: "Mitochondrial Myopathies Information Page" (Web page), www.ninds.nih.gov/Disorders/All-Disorders/Mitochondrial-Myopathy-Information-Page
- United Mitochondrial Disease Foundation: www.umdf.org, 1-888-317-UMDF (1-888-317-8633)

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