

MITOCHONDRIAL DISEASES

What is it?

Mitochondrial diseases is a group of progressive metabolic, often neurological, disorders that result from defects in the mitochondria. The mitochondria are commonly referred to as the “power house” of cells in the body containing genetic material. They are responsible for creating more than 90% of the energy needed to support growth and sustain life. Energy deprivation results in mitochondrial failure and inhibits the body’s ability to function. If this process is repeated throughout the body, cell injury and death results in organ dysfunction and organ system failure.

Mitochondrial diseases can affect almost any part of the body, making it challenging to diagnose. If three or more organ systems are involved, mitochondrial disease should be suspected. Not everyone with a mitochondrial disease will show symptoms. However, when discussing the group of mitochondrial diseases that tend to affect children, symptoms usually appear in the toddler and preschool years.

What are the symptoms or characteristics?

Mitochondrial diseases do not usually follow a set pattern. Symptoms will depend on which body cells are affected. Such symptoms will vary in severity and age of onset. The heart, brain, lungs, muscles, and gastrointestinal tract are most affected because they require the most energy. Mitochondrial diseases can also affect cells of the nerves, kidneys, liver, eyes, ears, or pancreas.

Complications like cardiac and respiratory issues, liver failure, seizures, and diabetes are possible. Fatigue and weakness are common. Other common signs and symptoms include:

- Poor growth
- Muscle weakness, low tone, movement disorders
- Pain
- Neurological issues like seizures, headaches, stroke
- Increased risk of infection
- GI issues like reflux, constipation, diarrhea
- Temperature instability
- Vision and hearing problems
- Autism and autism-like features
- Thyroid problems

What is the treatment?

There is no cure or effective treatment for mitochondrial disease. It is important to identify and treat the different medical problems that can occur with the disorder. Treatments and interventions must be based on the unique needs of each individual. This could mean regular monitoring for potential medical problems like cardiac or respiratory conditions, nutritional management, vitamin/amino acid supplement, or specialized services to treat things like low muscle tone.



Kennedy Krieger Institute

The Specialized Health Needs Interagency Collaboration (SHNIC) program is a collaborative partnership between the Kennedy Krieger Institute and the Maryland State Department of Education.

Suggested school accommodations

Students with mitochondrial disorders often need accommodations to help conserve energy while learning in the school setting. Supporting students with these conditions in the school require educators and parents/guardian to work as a team. Some accommodations to consider for a 504/IEP could include:

- PT/OT/SLP evaluation
- Modified or flexible school day
- Plan for absences and make-up work
- Limiting stressors
- Provide support
- Energy conservation strategies
- Frequent breaks
- Monitor for memory loss
- Limit physical activities when needed
- Climate control environment in all educational settings
- Hydration, snacks
- Rest area
- Hand hygiene
- Staff education/training as appropriate
- Emergency Evacuation Plan (EEP)

Specific health issues for Individualized Healthcare Plan

- Name of mitochondrial disorder, severity
- Body parts and functions affected
- Current medications including PRN for pain or spasms
- Last hospitalization
- Fever protocol
- Dietary orders, snacks, fluid goals
- Feeding tube orders for use and replacement, if applicable
- Orders for temperature control for cold stress/hot stress
- Orders for rest breaks or nap times
- Repositioning for muscle cramping
- Communicate with school staff, parents/guardian, and provider any changes or concerns about the disease
- Emergency Care Plan(s) (ECP) related to medical needs in the school setting and staff education/training as appropriate for each

Resources & Manuals

Kennedy Krieger Institute: The Neurology and Neurogenetics Clinic

<https://www.kennedykrieger.org/patient-care/centers-and-programs/neurology-and-neurogenetics-clinic>

United Mitochondrial Disease Foundation

<https://www.umdf.org/>

United Mitochondrial Disease Foundation– Teacher/Educator Resources

<https://www.umdf.org/teacher-educator-resources/>

Mito Action

<https://www.mitoaction.org/>