Mitochondrial Disease

Background

Mitochondrial disease occurs when the mitochondria, or "powerhouse" of the cell responsible for turning sugar and oxygen into energy, malfunctions. When mitochondria fail to produce enough energy, cells of the body do not function properly and can lead to cell injury, cell death, and organ system failure. The brain, heart and muscles are the most common organs to experience damage as they require the most energy. Three or more malfunctioning body systems and recurrent flare-ups associated with acute infections are considered red flags for diagnosis. Mitochondrial disease is progressive and can cause debilitating physical, developmental and cognitive disabilities.

There are many types of mitochondrial diseases, and a spectrum of symptoms can arise in any organ, at any age. The variation in symptoms, ranging from mild to debilitating, are dependent upon the number and type of cells affected. General symptoms of mitochondrial disease include:

- Fatigue
- Muscle weakness
- Pain
- Headache
- Poor growth
- Vision and hearing problems

Cardiac and respiratory issues (e.g., cardiomyopathy, irregular heartbeat, unstable blood pressure, shortness of breath) may require special intervention and short or long-term care.

Gastrointestinal problems can include nausea, vomiting, dysmotility and dehydration. Diets including anti-inflammatory foods to decrease stress on the body have been suggested.

Neurological symptoms can cause seizures, stroke, hearing loss, vision problems, developmental delays, learning disabilities, and problems with attention and memory.

A child diagnosed with a mitochondrial disease may also be at increased risk of infection and temperature intolerance. A thyroid, adrenal or diabetic diagnosis may also occur.

Top Takeaways for School

Mitochondrial disease is a genetic disorder in which the mitochondria do not function properly and deprive the body's cells of energy.

School plans should focus on energy management (both physical and mental). Prioritizing, pacing, and planning will support energy demands for student success.

A large majority of children with mitochondrial disorders have a developmental delay.

Any metabolic stress on the body (e.g., illness, dehydration, fasting) can cause progression of disease symptoms. Cells may be unable to cope with the increased demand.

Reducing stress by optimizing nutrition (e.g., small frequent meals, vitamins, supplements) and maintaining muscle strength are all therapeutic approaches to care.

Considerations for the Individualized Healthcare Plan (IHP)

- Nursing diagnoses: Fatigue, risk for injury and risk for infection
- Student-specific triggers and prevention tips
- Respiratory interventions and equipment (consider tracheostomy brand/size and downsize, suctioning brand/size, frequency of suctioning, ventilator brand and settings); note location of suctioning, use of private duty nursing if applicable
- Nutrition interventions and equipment (consider brand/size of feeding tube, tube replacement, water flushes, fluid intake goal and supplements); note school district policy on tube replacement and consider keeping backup feeding tube kit at school if applicable
- Use of specialized equipment, adaptive equipment and orthotics
- Activity, positioning, transferring (consider precautions and/or restrictions)
- Equipment troubleshooting (consider equipment/device user manual, battery, charger)
- Consider emergency action plans (EAPs) and emergency evacuation plans (EEPs) related to special health care needs, including staff education/training

Discussion Starters for the Educational Team

1. Would the student benefit from evaluations or assessments in any of the following areas: physical therapy, occupational therapy, speech and language therapy, assistive technology, adapted physical education, functional behavior, psychology, hearing and vision?
2. Would the student benefit from additional academic support and/or modified education (e.g., copies of notes, extra time, reduced workload, simplified instructions, alternative formats for presentation of material, 504/IEP)?
3. Can strategies be implemented to assist the student with executive function (e.g., plan, prompts, organizers, agendas)?
4. Would schedule flexibility support the student?
5. Can rest breaks, safe spaces or reduced stimulation times be built into the student's schedule?
6. Will staff receive education/training to implement the student-specific emergency plan?

Resources

Kennedy Krieger Institute: Neurology and Neurogenetics Clinics
kennedykrieger.org/patient-care/centers-and-programs/neurology-and-neurogenetics-clinics

United Mitochondrial Disease Foundation
umdf.org/

Mito Foundation: Mito for Kids
mito.org.au/school-education/mito-4-kids/

Mito Action
mitoaction.org/

For more information, please scan the QR code or visit: KennedyKrieger.org/SHNIC

Kennedy Krieger