



Empowering Children. Supporting Families. Building Brighter Futures.

Resource 074 | Metabolic and hormonal conditions

Mitochondrial disorders

What it means

Mitochondrial disorders are a varied group of conditions affecting the cell structures that help produce usable energy. They can involve several organs, including the brain and muscles. Developmental and medical effects differ substantially. A precise diagnosis is more useful than assuming one course or treatment for the whole group. [1]

Causes and risk factors

Disease-causing changes can occur in mitochondrial DNA or genes in the cell nucleus. Inheritance therefore varies and is not always through the mother. Energy-intensive organs may be particularly affected. Genetic counseling helps explain the specific disorder, while some cases remain difficult to confirm. [1,2]

General family education. A concern deserves assessment, but this guide does not diagnose a child or prescribe treatment.



Signs and everyday impact

Possible signs include muscle weakness, unusual fatigue, developmental delay or regression, seizures, hearing or vision differences, growth or feeding problems, and involvement of the heart or other organs. These symptoms have many possible causes. Changes during illness should be assessed in light of the child's diagnosis and usual functioning. [1,2]

Assessment and diagnosis

Specialist assessment integrates the clinical pattern with genetic and selected metabolic investigations. Blood tests, imaging, and occasionally other procedures are chosen individually; no single screening test detects every mitochondrial disorder. Assessment also identifies which organs need monitoring and which developmental supports would improve participation. [1,2]

Treatment and therapy

Care treats the specific disorder and affected organs where possible, alongside nutrition, rehabilitation, communication, and educational support. Some conditions have targeted treatments; many require supportive management. Supplements or an 'energy cocktail' are not a universal solution and require specialist review. The team should provide an individualized plan for illness, feeding interruption, and procedures. [1,2,3]

Support at home

Follow the agreed activity, rest, nutrition, and illness plans. Balance participation with fatigue rather than pushing through marked exhaustion. Make communication and mobility supports accessible. Keep a medical summary available, and ask the team how to handle reduced intake; fasting restrictions and emergency needs depend on the specific condition.

When to seek help

Follow the emergency illness plan for vomiting, reduced intake, or unusual lethargy. New weakness, regression, seizures, breathing difficulty, or reduced consciousness needs urgent assessment.

Questions for your care team

- What specific disorder or genetic finding is established?
- Which organs and developmental skills need monitoring?
- What should we do during illness or interrupted feeding?



Podcast preparation

Use this outline to develop a later podcast script.

Suggested opening

Welcome to Mariel Early Intervention. Today we discuss mitochondrial disorders. Mitochondrial disorders are a varied group of conditions affecting the cell structures that help produce usable energy. This is general education; each child needs individualized care.

Opening: Define the topic and its developmental context.

Understanding: Explain causes and signs without inviting self-diagnosis.

Assessment: Describe evaluation and distinguish screening from diagnosis.

Support: Discuss treatment, therapy, and practical family supports.

Closing: Explain urgent signs and one clear next step.

Misconception to address: Myth: all mitochondrial disorders are inherited from the mother.
Nuclear-gene disorders have other inheritance patterns.

Before recording: update clinical references and local service details, arrange clinical review, and use fully consented or fictional examples.

Terms in plain language

- Mitochondria: cell structures involved in producing usable energy.
- Mitochondrial DNA: genetic material within mitochondria.
- Metabolic specialist: a clinician with expertise in disorders of body chemistry.

Sources and further reading

Sources accessed 3 October 2026. Numbered references support the clinical overview. Treatment availability and referral routes vary by country.

[1] Mitochondrial Diseases: MedlinePlus

<https://medlineplus.gov/mitochondrialdiseases.html>

[2] Metabolic Disorders: MedlinePlus

<https://medlineplus.gov/metabolicdisorders.html>

[3] Condition Information Newborn Screening

<https://newbornscreening.hrsa.gov/conditions>