



Empowering Children. Supporting Families. Building Brighter Futures.

**Resource 075 | Metabolic and hormonal conditions**

# Inherited metabolic disorders

## What it means

Inherited metabolic disorders are conditions in which genetic changes disrupt the body's handling of nutrients or other substances. They include many distinct diagnoses with very different treatments. Some affect development gradually; others can cause sudden illness. Early recognition and diagnosis can be crucial for conditions with effective treatment. [1]

## Causes and risk factors

Changes in genes may affect enzymes, transport proteins, or other processes involved in metabolism. Harmful substances may accumulate, or the body may lack an essential product or energy supply. Inheritance varies by disorder. A broad label does not identify the correct diet, medicine, or emergency plan. [1,2]

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



## Signs and everyday impact

An infant may initially appear well, then develop poor feeding, vomiting, unusual sleepiness, seizures, or loss of skills. Other conditions present later with growth, movement, learning, or organ problems. These signs are nonspecific. A newborn screening result can identify risk before symptoms, but screening does not cover every disorder. [1,2]

## Assessment and diagnosis

An abnormal screen or concerning clinical pattern requires appropriate confirmatory assessment. A metabolic team may use blood and urine studies, genetic testing, and other targeted investigations. A normal newborn screen does not exclude all inherited metabolic disorders. Diagnosis should clarify the condition before a specialized long-term treatment plan is chosen. [1,2]

## Treatment and therapy

Depending on the diagnosis, treatment may involve carefully prescribed nutrition, medical formula, medicines, enzyme-related treatment, or other specialist interventions. Some disorders need rapid treatment during illness. Developmental therapies and educational supports address functional needs. Restrictions used for one disorder may harm a child with another; families should follow only the child's individualized plan. [1,2,3]

## Support at home

Keep emergency instructions and the metabolic team's contact details accessible. Follow the prescribed meal, medicine, formula, and monitoring schedule. Ask childcare staff to understand the plan. Report reduced intake promptly when the disorder requires it, and avoid trying fasting, detox products, or diet changes to test a suspected condition.

## When to seek help

Repeated vomiting, poor feeding, unusual lethargy, seizures, or altered breathing can signal serious illness. Follow the emergency metabolic plan and obtain urgent care; do not wait for a routine developmental visit.

## Questions for your care team

- What condition has been confirmed and how?
- What nutrition and treatment plan is specific to it?
- When should we use the emergency illness instructions?



## Podcast preparation

Use this outline to develop a later podcast script.

### Suggested opening

Welcome to Mariel Early Intervention. Today we discuss inherited metabolic disorders. Inherited metabolic disorders are conditions in which genetic changes disrupt the body's handling of nutrients or other substances. 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: a normal newborn screen excludes every metabolic disorder. Coverage and detection limits vary.

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

### Terms in plain language

- Metabolism: the body's chemical processes for using and changing substances.
- Enzyme: a protein that helps a chemical reaction occur.
- Metabolic crisis: acute illness caused by failure of a metabolic process.

### Sources and further reading

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

**[1] Metabolic Disorders: MedlinePlus**

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

**[2] Condition Information Newborn Screening**

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

**[3] How do health care providers diagnose phenylketonuria (PKU)?**

<https://www.nichd.nih.gov/health/topics/pku/conditioninfo/diagnosed>