CPNI
Cyprus Paediatric Neurology Institute
Παιδονευρολογικό Ινστιτούτο Κύπρου

LEAFLET FOR PARENTS AND PATIENTS

Neurometabolic disorders

Neurometabolic disorders are rare inherited conditions in which the body cannot properly make, break down or use certain substances. Some of these affect the brain and nerves. Several can be treated, sometimes with diet, vitamins or medicines, so early diagnosis matters.

Signs that can suggest a metabolic cause

- Developmental delay, or loss of skills
- Seizures, especially in early life or that are hard to control
- Episodes of drowsiness, vomiting, unsteadiness or confusion, often during illness or after fasting
- Problems with movement, muscle tone or walking
- Poor feeding or growth
- Problems with vision or hearing

Tests

- Blood and urine tests that measure substances made by the body's chemistry
- Sometimes a sample of spinal fluid (lumbar puncture)
- Brain MRI, which can show patterns typical of some conditions
- Genetic testing to confirm the diagnosis
- Newborn screening detects some, but not all, metabolic conditions

Treatment

- Some conditions are treated with a special diet, managed with a metabolic dietitian
- Some respond to vitamins or supplements
- Specific medicines or enzyme treatments exist for certain conditions
- An emergency plan for illness may be needed, to prevent a metabolic crisis
- Care is shared with metabolic specialists, in Cyprus or abroad

Living with a metabolic condition

- ✓ Follow the diet and medicines exactly as advised
- ✓ Carry an emergency letter, and make sure the school knows the plan
- ✓ Do not let your child go long periods without food when they are unwell
- ✓ Ask about family testing and genetic counselling

When to get urgent help

If your child has a known metabolic condition, follow their emergency plan and seek help straight away if they:

- Are vomiting and cannot keep food, drinks or medicines down
- Become unusually drowsy, floppy or confused
- Have a seizure
- Have a fever or illness and are not eating
- Are breathing fast or seem very unwell

Notes for your appointment

Write down what you notice and any questions, and bring this to your appointment.

Date	What I noticed	Questions to ask

Contact CPNI

If you have questions or would like an appointment, please call our office on +357 22 476 627 or email cpni2007@gmail.com.

More leaflets and information: www.cpni.com.cy

This leaflet gives general information. It does not replace advice from your child's doctor.

Reviewed by Dr Paola Nicolaides, Consultant Paediatric Neurologist, November 2026.

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