CPNI
Cyprus Paediatric Neurology Institute
Παιδονευρολογικό Ινστιτούτο Κύπρου

LEAFLET FOR PARENTS AND PATIENTS

Genetic and rare neurological conditions

Many neurological and developmental conditions in children have a genetic cause. Each individual condition may be rare, but together rare conditions affect many families. Finding the cause can explain your child's difficulties, guide treatment and help with family planning.

When might a genetic cause be considered?

- Developmental delay or learning difficulties, especially with no clear cause
- Autism, especially together with developmental delay, epilepsy or unusual physical features
- Epilepsy that starts early or is hard to control, including developmental and epileptic encephalopathies (epilepsy that affects a child's development), for example Dravet syndrome or GEFS+ (genetic epilepsy with febrile seizures plus), often linked to the SCN1A gene
- Unusual physical features or growth
- Problems with movement, muscles or coordination
- Epilepsy or a similar condition in a first-degree relative (a parent, brother or sister)
- Loss of skills a child had before

Genetic tests

- Chromosomal microarray, which looks for missing or extra pieces of chromosomes
- Gene panels, which test a group of genes linked to your child's condition
- Exome or genome sequencing, which looks at most or all genes at once
- Testing parents can help us understand whether a change is new in your child or inherited
- Most tests use a blood sample; results can take weeks to months

What results can mean

- ✓ A clear answer: a genetic change that explains your child's condition
- ✓ A variant of uncertain significance: a change we cannot yet interpret, which may be reviewed again later
- ✓ No cause found: this does not rule out a genetic condition, as knowledge grows every year

How a diagnosis can help

- ✓ Understanding what to expect, and planning care and surveillance
- ✓ Sometimes a specific treatment, diet or medicine to avoid
- ✓ Information about the chance of the condition happening again in the family
- ✓ Connecting with other families and patient organisations
- ✓ Access to research studies

Our approach

We work closely with colleagues at the Genetics Clinic at Makarios Hospital, the Karaiskakio Foundation and the Cyprus Institute of Neurology and Genetics (CING), and collaborate on research into rare neurogenetic conditions with The Neurogenetics Lab, UCL Queen Square Institute of Neurology, London. We will explain each test, ask for your consent, and go through the results with you in person.

Questions you may want to ask

Families often find it helpful to ask:

- Which test is being done, and why?
- How long will the results take?
- Could the result affect other family members?
- Is there a patient group for this condition?
- Are there research studies we could join?

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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