SCN2A
AUSTRALIA

INFORMATION SHEET

Genetic testing: what to ask

A question prompt for your first genetics appointment

A genetic test looks at your child's **DNA** (the instructions inside their cells) to find a change that may explain their epilepsy or development. A clear answer can guide treatment, connect you to the right supports and research, and inform questions about future pregnancies. This sheet won't replace your appointment; it will help you get the most from it. Take it with you and make notes.

BRING WITH YOU

- A short timeline: pregnancy and birth, when seizures started, development so far.
- Previous results you have (EEG, MRI, blood tests, any earlier genetic tests).
- A list of relatives with epilepsy, developmental or learning differences.
- Your top three questions.

A About the test itself

? Which test are you recommending, and why?

Common options: a **gene panel** (a set of epilepsy genes); **whole exome sequencing** (WES, the protein-coding parts of all genes); **whole genome sequencing** (WGS, almost all the DNA); or a **chromosomal microarray** (CMA, looks for missing or extra pieces of DNA).

? Will you test our child only, or us as parents too?

Testing the child and both parents together (a “trio”) can speed up clear answers.

? How long will the results take?

? Is the test funded through Medicare or the hospital, or is there a cost to us?

B About the results

? What are the possible outcomes?

A clear cause is found; nothing is found; or a **variant of uncertain significance** (VUS), a change that can't yet be classified as harmful or harmless.

? If you find a cause, will it change my child's treatment?

For some genes, including **SCN2A**, the type of change can guide which seizure medicines to use or avoid.[3]

? If the result is uncertain or negative, can the data be re-checked later as knowledge grows?

This is called **reanalysis**, and it finds new answers over time.

C What it means for our family

- ? Is the change **de novo** (new in my child) or inherited from a parent?
- ? What is the chance of having another child with the same condition?
  Even when a change is new in the child, the chance isn't quite zero. A parent can carry the change in some egg or sperm cells only, called **gonadal mosaicism**. Your genetics team can give a figure for your situation.
- ? What are our options for future pregnancies?
  For example, testing during a pregnancy, or testing embryos before pregnancy.
- ? Should other relatives be tested?

D Support and next steps

- ? Who will explain the result to us, and can we have it in writing?
- ? Will you connect us with a condition-specific group?
  For SCN2A, that is **SCN2A Australia**.
- ? Are there registries, natural history studies or clinical trials we could join?
- ? Who is our main contact if we have questions later?

GOOD TO KNOW

- Genetic testing finds a cause in roughly **a quarter to a third** of children tested for epilepsy, and more often when seizures start in the first months of life.[1,2]
- A “no result” today does not mean never. Reanalysis later can find answers as more genes are discovered.[1]
- A diagnosis can do more than name the condition: it can refine treatment, end years of searching, and open doors to research.

General information, not medical advice. Always discuss your child's diagnosis, testing and treatment with your treating team. Genetics knowledge changes quickly; this reflects current understanding at the date shown.

SOURCES

1. Sheidley BR, et al. Genetic testing for the epilepsies: a systematic review. *Epilepsia*. 2022;63(2):375–387.
2. Sánchez Fernández I, et al. Diagnostic yield of genetic tests in epilepsy: a meta-analysis and cost-effectiveness study. *Neurology*. 2019;92(5):e418–e428.
3. Zuberi SM, et al. ILAE classification of epilepsy syndromes with onset in neonates and infants. *Epilepsia*. 2022;63(6):1349–1397.