

# Genetic Testing for Epilepsy



## SUMMARY

- Genetic testing aims to try and accurately diagnose the underlying cause of epilepsy
- Genetic epilepsies can be caused by a change in a [single gene or on a chromosome](#)
- Not one single type of genetic test can diagnose all types of genetic epilepsies
- Depending on the test, genetic testing can be done on saliva (spit) or blood samples
- An accurate diagnosis from a genetic test may help treatment, support and family planning

## WHAT IS GENETIC TESTING FOR EPILEPSY?

Genetic testing for epilepsy requires the collection of a specimen, usually blood or spit.

DNA is then extracted from the sample, and analysed for changes in either one or a group of genes.

There are hundreds of epilepsy genes, with new ones constantly being discovered.

It is important to remember that not one single type of genetic test can examine all epilepsy genes.

This means more than one genetic test may be required for an accurate diagnosis.

For example, often, a screen of the chromosomes is done first (a chromosomal microarray), followed by a gene panel or exome test.

## WHY THINK ABOUT GENETIC TESTING FOR EPILEPSY?

There are many possible benefits to genetic testing for epilepsy. Some of these can include:

- Helping doctors select the right anti-seizure medication (ASM) and other therapies.
- Answering your questions about why epilepsy started.
- Helping you access resources such as peer support groups, financial support, disability support, information and counselling.
- Helping you plan for a family.
- Helping you come to terms with an epilepsy diagnosis

## REASONS WHY YOU MAY NOT WANT TO HAVE GENETIC TESTING FOR EPILEPSY?

A genetic test is a powerful test, which can give you information about your, or your family member's future. Some people may not feel ready for that type of information. Genetic testing does not always give an answer – about two thirds (60%) of the time an uncertain or non -diagnostic result is given. There can also be health and life insurance implications. For these reasons it is important that the [pros and cons](#) of genetic testing are carefully considered. A [genetic counsellor](#) can help you think about these issues, and your doctor must take you through the informed consent process before organising a genetic test.

## WHAT ARE THE MOST COMMON TYPES OF GENETIC TESTS?

There are many types of genetic tests for epilepsy. The types of genetic tests include:

### CHROMOSOME ARRAY

- This genetic test looks at a person's chromosomes and finds any extra (duplicated), or missing (deleted) chromosomal segments, which may contain epilepsy-associated genes. It cannot look 'inside' genes to check that they do not have a change within the gene.

## EPILEPSY GENE PANEL TEST

- This test looks at the most common types of genes associated with epilepsy.
- Depending on the gene panel itself, this test may analyse just a few genes to many hundreds of genes.
- This test can get quickly 'out of date' as genes associated with epilepsy continue to be described.

## EXOME SEQUENCING

- An exome sequencing test looks at the DNA code for all genes, including epilepsy-associated genes.
- An exome test can be 'reanalysed' over time, which means looking back to see if there are changes in new epilepsy-associated genes, which were not looked at last time.

Before any genetic test for epilepsy, you will have to provide informed consent. You have to decide if you want to undertake a genetic test.

## GENETIC TESTING RESOURCES

- [Genetic testing for epilepsy – Epilepsy Foundation USA](#)
- [Genetics and genomics testing – Syndrome Without A Name](#)
- [Genetic epilepsies – Paediatric Epilepsy Network New South Wales](#)
- [Genetic and genomic testing information sheet – Centre for Genetics Education](#)
- [Genetic and genomic testing summary – Centre of Genetics Education](#)
- [Laboratory performing genetic and genomic testing](#)
- [Easy Read guide to genes, genetic clinics and genetic conditions – GeneEQUAL](#)

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