What is epilepsy?

Epilepsy is a group of neurological disorders in which individuals are at risk for recurrent/repeating seizures. The diagnosis of epilepsy is made when someone has two or more unprovoked seizures during their lifetime. In the general population, about 1 in 10 (10%) people will have one seizure during their lifetime, and about 3% will be diagnosed with epilepsy.

What causes epilepsy?

There are a number of different causes for epilepsy. The majority of epilepsy is caused by a combination of genetic and non-genetic factors, with most related to infection, stroke, head trauma, brain tumor, or a brain malformation. Factors that impact whether an individual has an underlying genetic cause to their epilepsy include:

- the type of seizure (partial, generalized, or other)
- age of onset
- family history
- other health issues, such as intellectual disability, autism spectrum disorder, or birth defects.

An underlying genetic cause can be identified in 30-50% of those with epilepsy. The genetic causes may include chromosomal abnormalities or single gene disorders. The different causes may be de novo (new) or inherited in an autosomal dominant, autosomal recessive, X-linked, or mitochondrial pattern.

What type of genetic testing may be recommended?

For those with epilepsy, particularly with those who have symptoms in childhood, there are multiple types of genetic tests that may be considered. Recommendations may differ between patients depending on whether they have other health issues. Testing may include biochemical screening, chromosome microarray, single gene tests, multigene panels, or whole exome sequencing.

For those with other health conditions such as intellectual disability, autism spectrum disorder, or birth defects, the chromosome microarray may be considered as an initial test. The chromosome microarray is used to identify small missing or extra pieces of chromosome material, and can identify or rule out several hundred known genetic syndromes. This testing has a detection rate (the chance for the testing to come back positive) of about

20-30% in general. For all individuals with epilepsy the detection rate is less than 5%.

Further testing that may be considered includes single gene testing, multigene panels, or whole exome sequencing. About 15-40% of individuals with epilepsy are found to have a single gene disorder on one of these tests.

Genetic testing should be considered for those with epilepsy because in some cases finding the underlying genetic cause of someone's epilepsy may change what is recommended for treatment. For instance, for some genetic forms of epilepsy there may be specific medications that should or should not be taken, or other treatments like dietary management that can be helpful.

Click here to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

References:

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