SYNJ1 Gene Information

Key Points

- You have one or two variants in the *SYNJ1* gene (please refer to your test report for details).
- *SYNJ1* variants have been associated with neurological diseases with different symptoms, which can include movement problems like those seen in Parkinson's disease.
- You should talk about this result with your neurologist to understand what this means for you.

SYNJ1

What is *SYNJ1*?

SYNJ1 is a gene that all people have. A gene is an instruction for your body. When there are two variants in the *SYNJ1* gene (one from each parent), it doesn't work well, almost always causing neurological symptoms. In some cases, it can cause movement problems like those seen in Parkinson's disease.

SYNJ1 variants can cause conditions such as:

- Developmental and epileptic encephalopathy: Symptoms can look different for each person, starting in the first days to first months of life. Symptoms may include low muscle tone, feeding issues, seizures, visual problems, missed milestones, and learning difficulties. Differences may be seen in brain imaging.
- Early onset parkinsonism with seizures: Symptoms can look different for each person, starting between the teenage years and third decade. Symptoms include uncontrollable muscle contractions (dystonia), muscle stiffness (spasticity), seizures, visual problems, parkinsonism, and thinking problems. People may partially respond to levodopa medication, with side effects being common.

What does this mean for me?

Depending on your results:

- **If you have one variant**, this finding is unlikely to be a cause for your symptoms, but you are a carrier (Rarely a second variant may be missed through testing).
- **If you have two variants** (one from each parent) you typically will have early-onset of the *SYNJ1* related neurological symptoms, and this finding could be an explanation for your symptoms.
- You should talk to your neurologist or a movement disorder specialist to understand how this result relates to you and your symptoms.

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What does this result mean for my family?

- *SYNJ1* variants are inherited, meaning they are passed down in families. Typically, a person must have two variants in the *SYNJ1* gene, one from each parent, to cause early-onset parkinsonism with additional neurological symptoms. This is called recessive inheritance.
- **If you have one variant**, this means each of your close family members (like your children, siblings and parents) has a 50% chance (1 in 2) of having the same *SYNJ*1 variant and being a carrier. If your partner also has a *SYNJ1* variant, your children have a chance to get a *SYNJ1* related early-onset neurological disease.
- **If you have two variants** (one from each parent), each of your children will have one variant and will at least be carriers.
- Close family members (like your children, siblings, and parents) may benefit from meeting with a genetic counselor to learn about their risks and genetic testing options.

What should I do next?

- Tell your neurologist about these results.
- Talk to your neurologist, movement disorder specialist, or a neurology genetic counselor if you have more questions or concerns.
- Consider sharing this finding with your family members.
- Consider participating in additional studies to help researchers better understand the potential link between *SYNJ1* variants and Parkinson's disease or its symptoms.

For additional information about Parkinson's disease genetics and research visit PDNexus.org

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