SLC6A3 Gene Information

Key Points

- You have one or two variants in the *SLC6A3* gene (please refer to your test report for details).
- *SLC6A3* variants have been associated with a dopamine transporter deficiency syndrome (DTDS), and other neurological symptoms.
- You should talk about this result with your neurologist to understand what this means for you.

SLC6A3

What is *SLC6A3*?

SLC6A3 is a gene that all people have. A gene is an instruction for our body. When there are two variants in the *SLC6A3* gene (one from each parent), it does not work well, usually causing *SLC6A3*-related dopamine transporter deficiency syndrome (DTDS). DTDS is a complex movement disorder that can have symptoms like Parkinson's disease including parkinsonism-dystonia (involuntary muscle contractions) with tremor, rigidity, and reduced facial expressions. Additional developmental and neurological symptoms may also be present. People with two variants in the *SLC6A3* gene often begin experiencing symptoms in infancy, but symptoms can be delayed until adolescence or adulthood. Age of onset and symptoms can be different for each person.

What does this mean for me?

Depending on your results:

- **If you have one variant**, you usually will not have DTDS, but you are a carrier. (Rarely a second variant may be missed through testing). Researchers are studying whether having one *SLC6A3* variant could increase the risk for Parkinson's disease later in life. Additional genetic and/or environmental factors likely contribute to causing the condition. Some individuals with one or two *SLC6A3* variants may have brain and mental health conditions including bipolar, attention-deficit-hyperactivity (ADHD), or autism spectrum, and substance use disorders, though more research is needed to understand these associations.
- **If you have two variants** (one from each parent), you typically will have DTDS, 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.
- Please refer to the website pdf for more information: https://medlineplus.gov/download/genetics/gene/slc6a3.pdf

What does this result mean for my family?

- *SLC6A3* variants are inherited, meaning they are passed down in families. DTDS usually happens when a person has two variants in the SLC6A3 gene, one from each parent. 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 *SLC6A3* variant and being a carrier. If your partner also has a *SLC6A3* variant, your children have a chance to have DTDS.
- **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 these results with your family members.
- Consider participating in additional studies to help researchers better understand the potential link between *SLC6A3* 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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