ATP1A3 Gene Information

Key Points

- You have a variant in the *ATP1A3* gene.
- *ATP1A3* variants have been associated with a range of neurologic conditions that can affect movement and development, some of which can look like Parkinson's disease.
- You should talk about this result with your neurologist to understand what this means for you.

ATP1A3

What is *ATP1A3*?

ATP1A3 is a gene that all people have. A gene is an instruction for your body. You have a variant in your *ATP1A3* gene that makes it not work right. When there is a variant in the *ATP1A3* gene, it doesn't work well. This can cause different neurologic conditions that affect movement and development. In some cases, it can cause symptoms similar to those seen in Parkinson's disease. However, people with changes in the *ATP1A3* gene can have other symptoms or conditions such as:

- Rapid-onset dystonia parkinsonism (RDP)- abnormally clenched muscles (dystonia) and parkinsonism (slow movements and balance problems) that develop quickly, usually after a stressful event. Symptoms can start anywhere from childhood to mid-adulthood.
- Alternating hemiplegia of childhood (AHC)- paroxysmal (sudden) episodes of weakness or clenched muscles (dystonia) on one side of the body that last from minutes to weeks. The episodes usually start before the age of 2.
- Cerebellar ataxia (coordination problems), areflexia (absent reflexes), pes cavus (high arch of foot), optic atrophy (loss/reduced function of optic nerve), sensorineural hearing loss syndrome (CAPOS)

A person with a variant in *ATP1A3* may have symptoms that fit with one or more of these conditions. Neuropsychiatric and cognitive problems have also been reported.

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What does this mean for me?

- This finding may 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. They may refer you to other specialists for further evaluation of *ATP1A3*-related symptoms.
- Please refer to this link for more information: https://medlineplus.gov/genetics/gene/atp1a3/

What does this result mean for my family?

- *ATP1A3* variants are inherited, meaning they are passed down in families. *ATP1A3*-related conditions happen when a person has one variant in the *ATP1A3* gene, from one parent. This is called dominant inheritance.
- Your close family members (like your children, siblings and, parents) have a 50% (1 in 2) chance of having the same *ATP1A3* variant. However, not everyone with an *ATP1A3* variant will have symptoms.
- 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 *ATP1A3* 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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