ATP13A2 Gene Information

Key Points

- You have one or two variants in the *ATP13A2* gene (please refer to your test report for details).
- *ATP13A2* variants have been associated with juvenile-onset atypical Parkinson's disease.
- You should talk about this result with your neurologist to understand what this means for you.

ATP13A2

What is *ATP13A2*?

ATP13A2 is a gene that all people have. A gene is an instruction for our body. When there are two variants in the *ATP13A2* (one from each parent), it doesn't work well, causing juvenile-onset atypical Parkinson's disease. This condition can also be called Kufor-Rakeb syndrome, with symptoms of Parkinsonism, eye problems, and thinking problems. People with two variants in the *ATP13A2* gene typically begin experiencing disease symptoms as a child (12-22 years old), though symptoms can start earlier or later in life. Some people with *ATP13A2* variants will have other symptoms, which can include abnormal reflexes and tight muscles, or have muscle weakness, intellectual/developmental disabilities, and issues with their behavior. These 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 juvenile-onset atypical Parkinson's disease or Kufor-Rakeb Syndrome, but you are a carrier. (Rarely a second variant may be missed through testing). Researchers are studying whether having one *ATP13A2* variant could increase the risk for Parkinson's disease later in life. Additional genetic and/or environmental factors likely contribute to causing the condition.
- **If you have two variants** (one from each parent), you typically will have juvenile-onset Parkinson's disease (Kufor-Rakeb syndrome), 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?

- *ATP13A2* variants are inherited, meaning they are passed down in families. This form of juvenile-onset Parkinson's disease happens when a person has two variants in the *ATP13A2* 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 *ATP13A2* variant and being a carrier. If your partner also has a *ATP13A2* variant, your children have a chance to have juvenile-onset Parkinson's disease and other serious symptoms.
- **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 *ATP13A2* 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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