ATP7B Gene Information

Key Points

- You have one or two variants in the *ATP7B* gene (please refer to your test report for details).
- *ATP7B* variants have been associated with a condition called Wilson disease, 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.

ATP7B

What is *ATP7B*?

ATP7B is a gene that all people have. A gene is an instruction for our body. When there are two variants in the *ATP7B* gene (one from each parent), it doesn't work well, causing Wilson disease. This causes too much copper to build up in the body. People with Wilson disease can have liver, neurological, and mental health problems. Some people with Wilson disease also have movement problems that look like Parkinson's disease. The 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 Wilson disease, but you are a carrier. (Rarely a second variant may be missed through testing and will require further evaluation.)
- **If you have two variants** (one from each parent) you typically will have Wilson disease. The good news is that there is treatment for Wilson disease to help lower the amount of copper in the body.
- You should talk to your neurologist or a movement disorder specialist to understand how this result relates to you and your symptoms. This is especially urgent if you have two variants.
- Please visit the Wilson Disease Association website for more information: https://wilsondisease.org/

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What does this result mean for my family?

- *ATP7B* variants are inherited, meaning they are passed down in families. Wilson disease happens when a person has two variants in the *ATP7B* 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 *ATP7B* variant and being a carrier. If your partner also has an *ATP7B* variant, your children could have Wilson 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**. You may need additional tests and evaluations to determine if you have Wilson disease.
- 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 *ATP7B* 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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