PTRHD1 Gene Information

Key Points

- You have one or two variants in the *PTRHD1* gene (please refer to your test report for details).
- *PTRHD1* variants have been associated with intellectual disability with early-onset parkinsonism.
- You should talk about this result with your neurologist to understand what this means for you.

PTRHD1

What is *PTRHD1*?

PTRHD1 is a gene that all people have. A gene is an instruction for our body. When there are two variants in the *PTRHD1* gene (one from each parent), it does not work well, causing intellectual disability with early-onset parkinsonism. Additional neurological or psychiatric symptoms may also be present. People with two variants in the *PTRHD1* gene typically begin experiencing neurological symptoms in childhood, though parkinsonism may be delayed until the third or fourth decade. 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 intellectual disability or early-onset parkinsonism, 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 intellectual disability with early-onset parkinsonism, 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?

- *PTRHD1* variants are inherited, meaning they are passed down in families. Intellectual disability and early-onset parkinsonism happens when a person has two variants in the *PTRHD1* 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 *PTRHD1* variant and being a carrier. If your partner also has a *PTRHD1* variant, your children have a chance to have intellectual disability with early-onset parkinsonism.
- **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 *PTRHD1* variants and Parkinson's disease or its symptoms.

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

Authors: Verbrugge J, Cook L, Rumbaugh M, Miller A, Delgado Hodges P, Caceres V, Fiallos K, Raineri Tapies M, Williamson M

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