PLA2G6 Gene Information

Key Points

- You have one or two variants in the *PLA2G6* gene (please refer to your test report for details).
- *PLA2G6* variants have been associated with neurological diseases with different symptoms, 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.

PLA2G6

What is *PLA2G6*?

PLA2G6 is a gene that all people have. A gene is an instruction for your body. When there are two variants in the *PLA2G6* gene (one from each parent), it doesn't work well, causing a range of neurological disorders called *PLA2G6*-associated neurodegeneration (PLAN). People with PLAN can have various symptoms in childhood or early adulthood. They may include walking difficulty (ataxia), uncontrollable muscle contractions (dystonia), muscle stiffness (spasticity), thinking and mood problems, and movement problems like those seen in Parkinson's disease.

What does this mean for me?

Depending on your results:

- **If you have one variant**, you usually will not have PLAN, but you are a carrier. (Rarely a second variant may be missed through testing). Researchers are studying whether having one *PLA2G6* 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 PLAN, 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 visit NBIA disorders association for more information: https://www.nbiadisorders.org/about-nbia/plan

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What does this result mean for my family?

- *PLA2G6* variants are inherited, meaning they are passed down in families. PLAN happens when a person has two variants in the *PLA2G6* 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 *PLA2G6* variant and being a carrier. If your partner also has a *PLA2G6* variant, your children have a chance to get PLAN.
- **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 *PLA2G6* 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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