POLG Gene Information

Key Points

- You have a variant(s) in the *POLG* gene.
- *POLG* variants are associated with a group of disorders involving the mitochondria.
- You should talk about this result with your neurologist to understand what this means for you.

POLG

What is *POLG*?

POLG is a gene that all people have. A gene is an instruction for your body. When there is a variant(s) in the *POLG* gene, it may not function well, causing a condition that affects the function of mitochondria. The mitochondria are parts inside our cells that act like batteries, giving energy to the whole body. People with a *POLG*-related mitochondrial disease can have many different symptoms involving organs with high energy needs. These symptoms can include walking difficulties (ataxia), nerve problems leading to weakness, numbness, and pain (peripheral neuropathy), muscle weakness, and eye muscle weakness (ptosis or progressive external ophthalmoplegia). Symptoms in children and young adults can include seizures, developmental delay or disabilities, liver problems, and stroke-like episodes. The symptoms can be different for each person.

What does this mean for me?

- Depending on the results, most will not have *POLG*-associated mitochondrial disease or symptoms.
- Researchers are studying if carrying one *POLG* variant causes mild expression of *POLG*-associated conditions or if it could be a small risk factor for typical Parkinson's disease later in life.
- You should talk to your neurologist or a movement disorder specialist to understand how this result relates to you and your symptoms. There may be treatment for *POLG*-associated symptoms.
- Please refer to the United Mitochondrial Disease Foundation for more information: https://umdf.org/polg/

What does this result mean for my family?

- *POLG* variants are inherited, meaning they are passed down in families. *POLG* - associated conditions usually happen when a person has two variants in the *POLG* 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 *POLG* variant as you and being a carrier. If your partner also has a *POLG* variant, your children have a chance to get *POLG*-associated mitochondrial 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.**
- **Talk to your doctor about your *POLG* results if you are prescribed valproic acid (or other anti-seizure medications) since there are reports of liver toxicity associated with *POLG* variants.**
- 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 *POLG* 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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Updated: August 21, 2025