RAB39B Gene Information

Key Points

- You have a variant in the *RAB39B* gene.
- *RAB39B* variants have been associated with early onset Parkinson's disease and intellectual disability.
- You should talk about this result with your neurologist to understand what this means for you.

RAB39B

What is *RAB39B*?

RAB39B is a gene that all people have. A gene is an instruction for your body. When there is a variant in the *RAB39B* gene, it doesn't work well, causing a rare form of early-onset Parkinson's disease (onset before age 50) and/or variable degrees of intellectual disability, and other neurological problems. This condition most often affects people who are assigned male at birth. The *RAB39B* gene is on the X chromosome, which is a sex chromosome. In humans, typically, people assigned female at birth have two X chromosomes and people assigned male at birth have an X and a Y chromosome.

What does this mean for me?

- **If you have an X and a Y chromosome (generally assigned male at birth)**, this gene variant likely contributed to your Parkinson's disease and any other related symptoms.
- **If you have two X chromosomes (generally assigned female at birth)**, this gene variant may have contributed to your Parkinson's disease.
- You should talk to your neurologist or a movement disorder specialist to understand how this result relates to you and your symptoms.

What does this result mean for my family?

- *RAB39B* variants are inherited, meaning they are passed down in families. *RAB39B* conditions happen typically in males, who have only one X chromosome, and have a *RAB39B* variant. Less frequently, females with the variant may have mild features. This is called X-linked inheritance.

- **If you have an X and a Y chromosome (generally assigned male at birth)**, and one *RAB39B* variant, all of your children with two X chromosomes (typically daughters) will inherit the *RAB39B* variant and could have an increased risk for Parkinson's disease, possibly at a young age. None of your children with an X and Y chromosome (typically sons) will inherit the *RAB39B* variant from you. Your siblings have a 50% chance (1 in 2) of having the *RAB39B* variant regardless of their sex chromosomes.
- **If you have two X chromosomes (generally assigned female at birth)** and one *RAB39B* variant, each of your children will have a 50% chance (1 in 2) of having the same *RAB39B* variant. Your children with an X and a Y chromosome (typically sons) who inherit the variant would most likely have early-onset Parkinson's, intellectual disability, or both. Your children with two X chromosomes (typically daughters) who inherit the *RAB39B* variant may have a higher risk for Parkinson's disease, which could start before age 50. They likely would not have intellectual disability. Each of your siblings has up to a 50% chance of having the same *RAB39B* variant.
- 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 *RAB39B* 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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