

Key Points

- You have one or two variants in the *TH* gene (please refer to your test report for details).
- *TH* variants have been associated with neurological diseases that often affect movement. This 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.

TH

What is *TH*?

TH is a gene that all people have. A gene is an instruction for your body. When there are two variants in the *TH* gene (one from each parent), it doesn't work well, causing tyrosine hydroxylase deficiency (*TH* deficiency). People with *TH* deficiency most often have dopa-responsive dystonia (uncontrolled muscle contractions which can be treated with levodopa) starting in early childhood. In severe cases, symptoms start in infancy and also include parkinsonism, low muscle tone (hypotonia) and delayed motor milestones or intellectual disability.

What does this mean for me?

Depending on your results:

- **If you have one variant**, you usually will not have *TH* deficiency, 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 tyrosine hydroxylase deficiency, and this finding could be an explanation for your symptoms, especially if you have dystonia.
- You should talk to your neurologist or a movement disorder specialist to understand how this result relates to you and your symptoms.
- Please visit this website for more information:
<https://medlineplus.gov/genetics/condition/tyrosine-hydroxylase-deficiency/>

What does this result mean for my family?

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