GCH1 Gene Information

Key Points

- You have a variant in the *GCH1* gene.
- *GCH1* variants are associated with dystonia (uncontrolled muscle contractions), which responds well to dopamine medicines. This condition is called dopa-responsive dystonia (DRD). Less often, these variants can be associated with parkinsonism or Parkinson's disease.
- You should talk about this result with your neurologist to understand what this means for you.

GCH1

What is *GCH1*?

GCH1 is a gene that all people have. A gene is an instruction for your body. When there is a variant in the *GCH1* gene, it doesn't work well. This causes a condition called dopa-responsive dystonia (DRD) that usually starts in childhood. People with DRD may experience dystonia, increased reflexes, and other problems with walking and movement. Often, they feel better in the beginning of the day and experience more symptoms as the day advances. Treatment with dopamine medicines is usually very helpful for people with DRD, and compared with Parkinson's, the condition is not necessarily progressive (degenerative). *GCH1* variants less commonly cause parkinsonism (slow movement, muscle stiffness, tremor, balance instability) presenting in adult years. These symptoms can be different for each person.

What does this mean for me?

- This finding may 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 refer to this link for more information: https://medlineplus.gov/genetics/gene/gch1/

What does this result mean for my family?

- *GCH1* variants are inherited, meaning they are passed down in families. *GCH1*-related conditions happen when a person has one variant in the *GCH1* gene, from one parent. This is called dominant inheritance.
- Your close family members (like your children, siblings and, parents) have a 50% (1 in 2) chance of having the same *GCH1* variant. However, not everyone with a *GCH1* variant will have symptoms. Whether you develop symptoms may depend on your sex (assigned at birth), with females more likely to have symptoms.

- If both parents have *GCH1* variants, they can have a child with a more severe condition called tetrahydrobiopterin deficiency. Visit this website for more information about this condition: https://medlineplus.gov/genetics/condition/tetrahydrobiopterin-deficiency/
- 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 *GCH1* 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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