DCTN1 Gene Information

Key Points

- You have a variant in the *DCTN1* gene.
- *DCTN1* variants have been associated with a spectrum of neurological diseases including Perry syndrome.
- You should talk about this result with your neurologist to understand what this means for you.

DCTN1

What is *DCTN1*?

DCTN1 is a gene that all people have. A gene is an instruction for your body. When there is a variant in the *DCTN1* gene, it doesn't work well and can lead to problems referred to as *DCTN1*-related neurodegeneration. People with *DCTN1* variants are most likely to have Perry syndrome with symptoms like parkinsonism, mood and personality changes, weight loss, and breathing problems.

There can be other symptoms or conditions associated with *DCTN1* variants that include:

- Distal hereditary motor neuropathy type 7B (dHMN7B) - progressive muscle weakness and muscle loss, mostly in the hands and feet
- Frontotemporal dementia (FTD) - problems with behavior, language, and thinking
- Amyotrophic lateral sclerosis (ALS) also called Lou Gehrig's disease – muscles get weaker over time, causing an inability to move
- Progressive supranuclear palsy (PSP) - problems with movement, balance, vision, speech, swallowing, and mood/behavior

What does this mean for me?

- Within the spectrum of *DCTN1*-related neurodegeneration, individual's symptoms may look like Parkinson's disease or other conditions.
- 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 website for more information: https://medlineplus.gov/genetics/gene/dctn1/

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What does this result mean for my family?

- *DCTN1* variants are inherited, meaning they are passed down in families. *DCTN1*-related conditions happen when a person has one variant in the *DCTN1* 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 *DCTN1* variant. Currently, there is limited information about the risk of developing symptoms in people who have the variant but are not affected. However, the risk appears to increase with age.
- 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.**
- People with *DCTN1*-related neurodegeneration can have breathing problems, so it is important to avoid or limit substances that slow down breathing, like benzodiazepines, alcohol, or narcotics.
- 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 *DCTN1* 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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