JPH3 gene

junctophilin 3

Normal Function

The *JPH3* gene provides instructions for making a protein called junctophilin-3, which is found primarily in the brain. Although the exact function of this protein is unclear, researchers believe that it plays a role in the formation of a structure called the junctional membrane complex. This complex connects certain channels inside cells with other channels at the cell surface. The junctional membrane complex appears to be involved in the release of charged calcium atoms (calcium ions), which are critical for transmitting signals within cells. As part of the junctional membrane complex, junctophilin-3 is probably involved in signaling within and between nerve cells (neurons) in the brain.

One region of the *JPH3* gene contains a particular DNA segment known as a CAG/CTG trinucleotide repeat. This segment is made up of a series of three DNA building blocks (nucleotides) that appear multiple times in a row. Normally, the CAG/CTG segment is repeated 6 to 28 times within the gene.

Health Conditions Related to Genetic Changes

Huntington's disease-like syndrome

A particular type of variant (also called a mutation) in the *JPH3* gene has been found to cause signs and symptoms that resemble those of Huntington's disease, including uncontrolled movements, emotional problems, and loss of thinking ability. Researchers have named this condition Huntington's disease-like 2 (HDL2).

The variant associated with HDL2 increases the size of the CAG/CTG trinucleotide repeat in the *JPH3* gene. People with this condition have 44 to 59 CAG/CTG repeats. People with 29 to about 43 CAG/CTG repeats may or may not develop the signs and symptoms of HDL2.

Researchers are working to determine the effects of the abnormally large CAG/CTG segment. They believe that the altered *JPH3* gene produces an altered version of messenger RNA, which is a molecular blueprint of the gene that is normally used for protein production. The abnormal messenger RNA forms clumps inside neurons that interfere with the normal functions of these cells. The dysfunction and eventual death of

neurons in certain areas of the brain underlie the signs and symptoms of HDL2.

Other Names for This Gene

- CAGL237
- FLJ44707
- HDL2
- JP-3
- JP3
- JPH3_HUMAN
- junctophilin type 3
- junctophilin-3
- TNRC22
- trinucleotide repeat containing 22

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of JPH3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57338[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28JPH3%5BTIAB%5D%29+OR+%28junctophilin+3%5BTIAB%5D%29%29+OR+%28%28HDL-2%5BTIAB%5D%29+OR+%28JP-3%5BTIAB%5D%29+OR+%28JP3%5BTIAB%5D%29+OR+%28junctophilin+type+3%5BTIAB%5D%29+OR+%28trinucleotide+repeat+containing+22%5BTIAB%5D%29+OR+%28junctophilin-3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- JUNCTOPHILIN 3; JPH3 (https://omim.org/entry/605268)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/57338)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=JPH3[gene])

References

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Genomic Location

The *JPH3* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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