ATXN1 gene

ataxin 1

Normal Function

The *ATXN1* gene provides instructions for making a protein called ataxin-1. This protein is found throughout the body, but its function is unknown. Within cells, ataxin-1 is located in the nucleus. Researchers believe that ataxin-1 may be involved in regulating various aspects of producing proteins, including the first stage of protein production (transcription) and processing RNA, a chemical cousin of DNA.

One region of the *ATXN1* gene contains a DNA segment known as a CAG trinucleotide repeat. This segment is made up of a series of three DNA building blocks (cytosine, adenine, and guanine) that appear multiple times in a row. Normally, the CAG segment is repeated 4 to 39 times within the gene.

Health Conditions Related to Genetic Changes

Spinocerebellar ataxia type 1

Spinocerebellar ataxia type 1 (SCA1) is a condition characterized by progressive problems with movement. SCA1 results from a mutation in the *ATXN1* gene known as a trinucleotide repeat expansion. This mutation increases the length of the repeated CAG segment in the *ATXN1* gene. People with SCA1 have from 40 to more than 80 CAG repeats in most cells.

The expanded CAG segment leads to the production of an abnormally long version of the ataxin-1 protein that folds into the wrong 3-dimensional shape. This abnormal protein clusters with other proteins to form clumps (aggregates) within the nucleus of cells. These aggregates prevent the ataxin-1 protein from functioning normally, which damages cells. For reasons that are unclear, aggregates of ataxin-1 are found only in the brain and spinal cord (central nervous system). Certain brain cells called Purkinje cells seem to be particularly sensitive to the accumulation of these aggregates. Purkinje cells are located in the part of the brain that coordinates movement (cerebellum) and are involved in chemical signaling between nerve cells (neurons). The accumulation of protein aggregates in the cells of the central nervous system, particularly in Purkinje cells, leads to cell death. Over time, the loss of these cells causes the movement problems characteristic of SCA1.

Other Names for This Gene

- ataxin-1
- ATX1
- ATX1_HUMAN
- SCA1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ATXN1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6310[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ATXN1%5BTIAB%5D%29+OR+%28ataxin+1%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ATAXIN 1; ATXN1 (https://omim.org/entry/601556)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6310)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ATXN1[gene])

References

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- Lim J, Crespo-Barreto J, Jafar-Nejad P, Bowman AB, Richman R, Hill DE, Orr HT, Zoghbi HY. Opposing effects of polyglutamine expansion on native proteincomplexes contribute to SCA1. Nature. 2008 Apr 10;452(7188):713-8. doi:10.1038/nature06731. Epub 2008 Mar 12. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18337722) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2377396/)
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Genomic Location

The *ATXN1* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

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