ATXN2 gene

ataxin 2

Normal Function

The *ATXN2* gene provides instructions for making a protein called ataxin-2. This protein is found throughout the body, but its function is unknown. Ataxin-2 is found in the fluid inside cells (cytoplasm) and seems to interact with a cell structure called the endoplasmic reticulum. The endoplasmic reticulum is involved in protein production, processing, and transport. Researchers believe that ataxin-2 may be involved in processing RNA, a chemical cousin of DNA. Ataxin-2 is also thought to play a role in the translation of genetic information to produce proteins.

One region of the *ATXN2* 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 approximately 22 times within the gene.

Health Conditions Related to Genetic Changes

Spinocerebellar ataxia type 2

Spinocerebellar ataxia type 2 (SCA2) is a condition characterized by progressive problems with movement. SCA2 results from a mutation in the *ATXN2* gene known as a trinucleotide repeat expansion. This mutation increases the length of the repeated CAG segment in the *ATXN2* gene. People with 32 or more repeats CAG repeats in the *ATXN2* gene develop SCA2.

It is unclear how the abnormally long CAG segment affects the function of the ataxin-2 protein. The abnormal protein apparently leads to cell death, as people with SCA2 show a loss of brain cells. Certain brain cells called Purkinje cells seem to be particularly sensitive to the presence of abnormal ataxin-2. Purkinje cells are located in the part of the brain that coordinates movement (cerebellum) and are involved in chemical signaling between nerve cells (neurons). It is unknown how the abnormal ataxin-2 protein leads to the death of Purkinje and other brain cells. Over time, the loss of these cells causes the movement problems characteristic of SCA2.

Amyotrophic lateral sclerosis

MedlinePlus Genetics provides information about Amyotrophic lateral sclerosis

Other Names for This Gene

- ataxin-2
- ATX2
- ATX2_HUMAN
- SCA2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ATXN2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6311[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ATXN2%5BTIAB%5D%29+OR+%28ataxin+2%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 2; ATXN2 (https://omim.org/entry/601517)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6311)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ATXN2[gene])

References

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- Velazquez Perez L, Cruz GS, Santos Falcon N, Enrique Almaguer Mederos L, Escalona Batallan K, Rodriguez Labrada R, Paneque Herrera M, Laffita Mesa JM,

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Genomic Location

The *ATXN2* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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