

APT_X gene

aprataxin

Normal Function

The *APT_X* gene provides instructions for making a protein called aprataxin that is involved in the repair of DNA damage in cells. Aprataxin is located in the nucleus of cells and is produced in various tissues, including the brain, spinal cord, and muscles. Different parts of the aprataxin protein allow the protein to interact with other DNA repair proteins to make repairs. At the site of the damage, aprataxin modifies the broken ends of the DNA strands so they can be joined back together correctly.

Health Conditions Related to Genetic Changes

Ataxia with oculomotor apraxia

More than 30 mutations in the *APT_X* gene have been found to cause ataxia with oculomotor apraxia type 1. This condition is characterized by difficulty coordinating movements (ataxia) and problems with side-to-side movements of the eyes (oculomotor apraxia). Most mutations change single protein building blocks (amino acids) in aprataxin, resulting in an unstable aprataxin protein that is quickly broken down in the cell. A lack of functional aprataxin disrupts DNA repair and can lead to an accumulation of damage in cells, particularly affecting cells in the part of the brain involved in coordinating movements (the cerebellum). This accumulation can lead to cell death in the cerebellum, causing the characteristic movement problems of ataxia with oculomotor apraxia type 1.

Other Names for This Gene

- AOA
- AOA1
- APT_X_HUMAN
- ataxia 1, early onset with hypoalbuminemia
- AXA1
- EAOH
- EOAHA
- FHA-HIT

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of APTX ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=54840\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=54840[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28APTX%5BTIAB%5D%29+OR+%28aprataxin%5BTIAB%5D%29+OR+%28AOA1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- APRATAXIN; APTX (<https://omim.org/entry/606350>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/54840>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=APTX\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=APTX[gene]))

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Genomic Location

The *APTX* gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

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