SETX gene

senataxin

Normal Function

The *SETX* gene provides instructions for making a protein called senataxin. Senataxin is produced in a wide range of tissues, including the brain, spinal cord, and muscles. Based on the structure of senataxin, researchers believe that it is one of a class of proteins called helicases, which attach to particular regions of DNA or RNA (a chemical cousin of DNA) and temporarily unwind the strands of the molecule. By unwinding the strands, helicases allow other proteins to reach the strands to perform their function. Although senataxin's role in cells is not completely understood, it appears to be involved in the production of proteins from genes (transcription), the processing of RNA molecules, and the repair of damaged DNA.

Health Conditions Related to Genetic Changes

Amyotrophic lateral sclerosis

MedlinePlus Genetics provides information about Amyotrophic lateral sclerosis

Ataxia with oculomotor apraxia

At least 125 mutations in the *SETX* gene have been found to cause ataxia with oculomotor apraxia type 2. This condition is characterized by difficulty coordinating movements (ataxia) and problems with side-to-side movements of the eyes (oculomotor apraxia). Most mutations replace single protein building blocks (amino acids) in senataxin. The mutations associated with ataxia with oculomotor apraxia type 2 are thought to disrupt the helicase function of senataxin. Although it is unclear how impaired senataxin function leads to the signs and symptoms of ataxia with oculomotor apraxia type 2, some researchers suggest that it disrupts DNA repair and can lead to an accumulation of DNA damage in cells. This accumulation can lead to cell death and seems particularly harmful to cells in the part of the brain involved in coordinating movements (the cerebellum), causing the characteristic movement problems of ataxia with oculomotor apraxia type 2.

Charcot-Marie-Tooth disease

MedlinePlus Genetics provides information about Charcot-Marie-Tooth disease

Other Names for This Gene

- ALS4
- AOA2
- KIAA0625
- SCAR1
- Sen1
- SETX_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SETX (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23064[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28senataxin%5BTIAB%5D%29+OR+%28SETX%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SENATAXIN; SETX (https://omim.org/entry/608465)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/23064)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SETX[gene])

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Genomic Location

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

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