ATN1 gene
atrophin 1

Normal Function

The ATN1 gene provides instructions for making a protein called atrophin 1. Although the exact function of this protein is unknown, it appears to play an important role in nerve cells (neurons) in many areas of the brain. Based on studies in other animals, researchers speculate that atrophin 1 may act as a transcriptional co-repressor. A transcriptional co-repressor is a protein that interacts with other DNA-binding proteins to suppress the activity of certain genes, although it cannot attach (bind) to DNA by itself.

One region of the ATN1 gene contains a particular 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. In most people, the number of CAG repeats in the ATN1 gene ranges from 6 to 35.

Health Conditions Related to Genetic Changes

Dentatorubral-pallidoluysian atrophy

Dentatorubral-pallidoluysian atrophy (DRPLA) results from an increased number of copies (expansion) of the CAG trinucleotide repeat in the ATN1 gene. In people with this condition, the CAG segment is abnormally repeated at least 48 times, and the repeat region may be two or three times its usual length. Although the extended CAG region changes the structure of atrophin 1, it is unclear how the altered protein damages brain cells. Researchers believe that abnormal atrophin 1 accumulates in neurons and interferes with normal cell functions. The dysfunction and eventual death of neurons in many parts of the brain lead to involuntary movements, intellectual decline, and the other characteristic features of DRPLA.
Chromosomal Location

Cytogenetic Location: 12p13.31, which is the short (p) arm of chromosome 12 at position 13.31

Molecular Location: base pairs 6,924,459 to 6,942,321 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene
- ATN1_HUMAN
- atrophin-1
- B37
- D12S755E
- dentatorubral-pallidoluysian atrophy protein
- DRPLA
- NOD

Additional Information & Resources

Educational Resources
- Biochemistry (fifth edition, 2002): Some genetic diseases are caused by the expansion of repeats of three nucleotides
  https://www.ncbi.nlm.nih.gov/books/NBK22525/#A3843

Clinical Information from GeneReviews
- DRPLA
  https://www.ncbi.nlm.nih.gov/books/NBK1491
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ATN1%5BTIAB%5D%29+OR+%28atrophin+1%5BTIAB%5D%29+OR+%28atrophin-1%5BTIAB%5D%29+OR+%28DRPLA%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+AND+Genetic+Phenomena%5BMH%5D%29+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ATROPHIN 1
  http://omim.org/entry/607462

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ATN1%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1822

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P54259

Sources for This Summary


  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2175251/


Reprinted from Genetics Home Reference:
  https://ghr.nlm.nih.gov/gene/ATN1

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