

NOP56 gene

NOP56 ribonucleoprotein

Normal Function

The *NOP56* gene provides instructions for making a protein called nucleolar protein 56, which is found in the nucleus of nerve cells (neurons). This protein is mostly found in neurons within an area of the brain called the cerebellum, which is involved in coordinating movements. Nucleolar protein 56 is one part (subunit) of the ribonucleoprotein complex, which is composed of proteins and molecules of RNA, DNA's chemical cousin. The ribonucleoprotein complex is needed to make cellular structures called ribosomes, which process the cell's genetic instructions to create proteins.

Located within the *NOP56* gene, in an area known as intron 1, is a string of six DNA building blocks (nucleotides); this string, known as a hexanucleotide, is represented by the letters GGCCTG and is typically repeated 3 to 14 times within intron 1. The function of this repeated hexanucleotide is unclear.

Health Conditions Related to Genetic Changes

Spinocerebellar ataxia type 36

NOP56 gene mutations cause spinocerebellar ataxia type 36 (SCA36), which is a condition characterized by progressive movement problems that typically begin in mid-adulthood. In people with SCA36, the GGCCTG string in intron 1 is repeated at least 650 times.

To make proteins from the genetic instructions carried in genes, a molecule called messenger RNA (mRNA) is formed. This molecule acts as a genetic blueprint for protein production. However, a large increase in the number of GGCCTG repeats in the *NOP56* gene disrupts the normal structure of NOP56 mRNA. Abnormal NOP56 mRNA molecules form clumps called RNA foci within the nucleus of neurons. Other proteins become trapped in the RNA foci, where they cannot function. These proteins may be important for controlling gene activity or protein production.

Additionally, researchers believe that the large expansion of the hexanucleotide repeat in the *NOP56* gene may reduce the activity of a nearby gene called *MIR1292*. The *MIR1292* gene provides instructions for making a type of RNA that regulates the activity (expression) of genes that produce proteins called glutamate receptors. These proteins

are found on the surface of neurons and allow these cells to communicate with one another. A decrease in the production of Mir1292 RNA can lead to an increase in the production of glutamate receptors. The increased receptor activity may overexcite neurons, which disrupts normal communication between cells and can contribute to movement difficulties.

The combination of RNA foci and overly excited neurons likely leads to the death of neurons over time. Because the *NOP56* gene is especially active in neurons in the cerebellum, these cells are particularly affected by expansion of the gene, leading to cell death in the cerebellum. Deterioration in this part of the brain leads to ataxia and the other signs and symptoms of SCA36.

Other Names for This Gene

- NOL5A
- NOP56 ribonucleoprotein homolog
- nucleolar protein 56
- nucleolar protein 5A (56kDa with KKE/D repeat)

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NOP56 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10528\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10528[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28NOP56%5BTIAB%5D%29+OR+%28NOL5A%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+3600+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- NOP56 RIBONUCLEAR PROTEIN; NOP56 (<https://omim.org/entry/614154>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10528>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=NOP56\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=NOP56[gene]))

References

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Genomic Location

The *NOP56* gene is found on chromosome 20 (<https://medlineplus.gov/genetics/chromosome/20/>).

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