

## NPRL3 gene

NPR3 like, GATOR1 complex subunit

### Normal Function

The *NPRL3* gene provides instructions for making a protein that is one piece of a group of proteins (complex) called GATOR1. This complex is found in cells throughout the body, where it regulates a signaling pathway called the mTOR pathway. The mTOR pathway is involved in cell growth and division (proliferation), the survival of cells, and the creation (synthesis) of new proteins. The role of the GATOR1 complex is to block this pathway by inhibiting (stopping) the activity of a complex called mTOR complex 1 (mTORC1) that is integral to the mTOR pathway.

In the brain, the mTOR pathway regulates many processes, including the growth and development of nerve cells and their ability to change and adapt over time (plasticity).

### Health Conditions Related to Genetic Changes

#### Familial focal epilepsy with variable foci

At least ten *NPRL3* gene mutations have been found to cause familial focal epilepsy with variable foci (FFEVF), which is an uncommon form of recurrent seizures (epilepsy) that runs in families. Most of these mutations lead to the production of an abnormally short, nonfunctional protein. As a result, formation of normal GATOR1 complex is reduced, leading to overactivity of mTORC1 and excessive signaling of the mTOR pathway. It is not clear how an abnormally active mTOR pathway leads to the seizures of FFEVF. Research suggests that increased mTOR pathway signaling in the brain leads to changes in the connections between nerve cells (synapses) and increased activation (excitation) of nerve cells, which can cause seizures.

### Other Names for This Gene

- alpha-globin regulatory element-containing gene protein
- C16orf35
- CGTHBA
- conserved gene telomeric to alpha globin cluster
- HS-40
- MARE

- NPR3
- RMD11

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of NPRL3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8131\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8131[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28NPRL3%5BTIAB%5D%29+OR+%28NPR3+like,+GATOR1+complex+subunit%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+3600+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- NITROGEN PERMEASE REGULATOR-LIKE 3; NPRL3 (<https://omim.org/entry/600928>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8131>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=NPRL3\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=NPRL3[gene]))

## **References**

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## Genomic Location

The *NPRL3* gene is found on chromosome 16 (<https://medlineplus.gov/genetics/chromosome/16/>).

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