# PURA gene

purine rich element binding protein A

## Normal Function

The *PURA* gene provides instructions for making a protein called Pur-alpha (Purα), which is able to attach (bind) to DNA and RNA (a molecular cousin of DNA). This protein has multiple roles in cells, including controlling the activity of genes (gene transcription) and aiding in the copying (replication) of DNA.

The Purα protein is important for normal brain development. The protein helps direct the growth and division of nerve cells (neurons). It may also be involved in the formation or maturation of myelin, the protective substance that covers nerves and promotes the efficient transmission of nerve impulses.

## Health Conditions Related to Genetic Changes

### 5q31.3 microdeletion syndrome

5q31.3 microdeletion syndrome is caused by a chromosomal change in which a small piece of chromosome 5 is deleted in each cell. This rare condition is characterized by severely delayed or impaired development of speech and walking, weak muscle tone (hypotonia), breathing problems, recurrent seizures (epilepsy) or seizure-like episodes, and distinctive facial features. The deletion that causes this condition occurs on the long (q) arm of the chromosome at a position designated q31.3. The size of the deletion can range from several thousand to several million DNA building blocks (base pairs). The deleted region typically contains at least three genes, one of which is *PURA*.

A loss of one copy of the *PURA* gene is thought to alter normal brain development and impair the function of neurons, leading to developmental delay, hypotonia, and other neurological problems in people with 5q31.3 microdeletion syndrome. Some studies suggest that loss of another nearby gene called *NRG2* increases the severity of the signs and symptoms. It is unclear how the loss of other genes in the deleted region contributes to development of 5q31.3 microdeletion syndrome.

### PURA syndrome

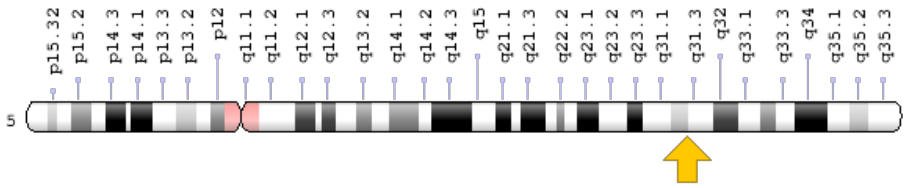
At least 22 *PURA* gene mutations have been found to cause *PURA* syndrome, a condition characterized by intellectual disability, delayed development of speech and walking, and epilepsy. Some of these genetic changes remove small segments of DNA from the *PURA* gene. Others change single building blocks (amino acids) in the Purα protein or lead to production of an abnormally short protein. These

mutations are thought to reduce the amount of functional Purα protein. Although it is not understood how a partial loss of Purα function leads to the signs and symptoms of *PURA* syndrome, researchers suspect that it may alter normal brain development and impair the function of neurons, leading to developmental problems and seizures in people with the condition.

## Chromosomal Location

Cytogenetic Location: 5q31.3, which is the long (q) arm of chromosome 5 at position 31.3

Molecular Location: base pairs 140,114,123 to 140,119,416 on chromosome 5 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- MRD31
- PUR-ALPHA
- PUR1
- PURALPHA
- purine-rich single-stranded DNA-binding protein alpha
- transcriptional activator protein Pur-alpha

## Additional Information & Resources

Clinical Information from GeneReviews

- PURA-Related Neurodevelopmental Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK426063

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PURA%5BTIAB%5D%29+OR+%28purine+rich+element+binding+protein+A%5BTIAB%5D%29%29+OR+%28%28PUR-ALPHA%5BTIAB%5D%29+OR+%28PUR1%5BTIAB%5D%29+OR+%28PURALPHA%5BTIAB%5D%29+OR+%28purine-rich+single-stranded+DNA-binding+protein+alpha%5BTIAB%5D%29+OR+%28transcriptional+activator+protein+Pur-alpha%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+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PURINE-RICH ELEMENT-BINDING PROTEIN A
  http://omim.org/entry/600473

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PURA.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PURA%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:9701
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5813
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5813
- UniProt
  https://www.uniprot.org/uniprot/Q00577

## Sources for This Summary


- Brown N, Burgess T, Forbes R, McGillivray G, Kornberg A, Mandelstam S, Stark Z. 5q31.3 Microdeletion syndrome: clinical and molecular characterization of two further cases. Am J Med Genet A. 2013 Oct;161A(10):2604-8. doi: 10.1002/ajmg.a.36108. Epub 2013 Aug 15.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/23950017
- Hokkanen S, Feldmann HM, Ding H, Jung CK, Bojarski L, Renner-Müller I, Schüller U, Kretzschmar H, Wolf E, Herms J. Lack of Pur-alpha alters postnatal brain development and causes megalencephaly. Hum Mol Genet. 2012 Feb 1;21(3):473-84. doi: 10.1093/hmg/ddr476. Epub 2011 Oct 18.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/22010047

- Hosoki K, Ohta T, Natsume J, Imai S, Okumura A, Matsui T, Harada N, Bacino CA, Scaglia F, Jones JY, Niikawa N, Saitoh S. Clinical phenotype and candidate genes for the 5q31.3 microdeletion syndrome. Am J Med Genet A. 2012 Aug;158A(8):1891-6. doi: 10.1002/ajmg.a.35439. Epub 2012 Jun 18.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/22711443
- Lalani SR, Zhang J, Schaaf CP, Brown CW, Magoulas P, Tsai AC, El-Gharbawy A, Wierenga KJ, Bartholomew D, Fong CT, Barbaro-Dieber T, Kukolich MK, Burrage LC, Austin E, Keller K, Pastore M, Fernandez F, Lotze T, Wilfong A, Purcarin G, Zhu W, Craigen WJ, McGuire M, Jain M, Cooney E, Azamian M, Bainbridge MN, Muzny DM, Boerwinkle E, Person RE, Niu Z, Eng CM, Lupski JR, Gibbs RA, Beaudet AL, Yang Y, Wang MC, Xia F. Mutations in PURA cause profound neonatal hypotonia, seizures, and encephalopathy in 5q31.3 microdeletion syndrome. Am J Hum Genet. 2014 Nov 6;95(5):579-83. doi: 10.1016/j.ajhg.2014.09.014. Epub 2014 Oct 16.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/25439098
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4225583/
- OMIM: PURINE-RICH ELEMENT-BINDING PROTEIN A
  http://omim.org/entry/600473
- Shimojima K, Isidor B, Le Caignec C, Kondo A, Sakata S, Ohno K, Yamamoto T. A new microdeletion syndrome of 5q31.3 characterized by severe developmental delays, distinctive facial features, and delayed myelination. Am J Med Genet A. 2011 Apr;155A(4):732-6. doi: 10.1002/ajmg.a.33891. Epub 2011 Mar 15. Erratum in: Am J Med Genet A. 2011 Nov;155A(11):2903.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/21594995
- Tanaka AJ, Bai R, Cho MT, Anyane-Yeboa K, Ahimaz P, Wilson AL, Kendall F, Hay B, Moss T, Nardini M, Bauer M, Retterer K, Juusola J, Chung WK. De novo mutations in PURA are associated with hypotonia and developmental delay. Cold Spring Harb Mol Case Stud. 2015 Oct;1(1):a000356. doi: 10.1101/mcs.a000356.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/27148565
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4850890/
- Weber J, Bao H, Hartlmüller C, Wang Z, Windhager A, Janowski R, Madl T, Jin P, Niessing D. Structural basis of nucleic-acid recognition and double-strand unwinding by the essential neuronal protein Pur-alpha. Elife. 2016 Jan 8;5. pii: e11297. doi: 10.7554/eLife.11297.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/26744780
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4764581/
- White MK, Johnson EM, Khalili K. Multiple roles for Puralpha in cellular and viral regulation. Cell Cycle. 2009 Feb 1;8(3):1-7. Epub 2009 Feb 10. Review.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/19182532
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2683411/


---


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

Reviewed: August 2017
Published: May 14, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services