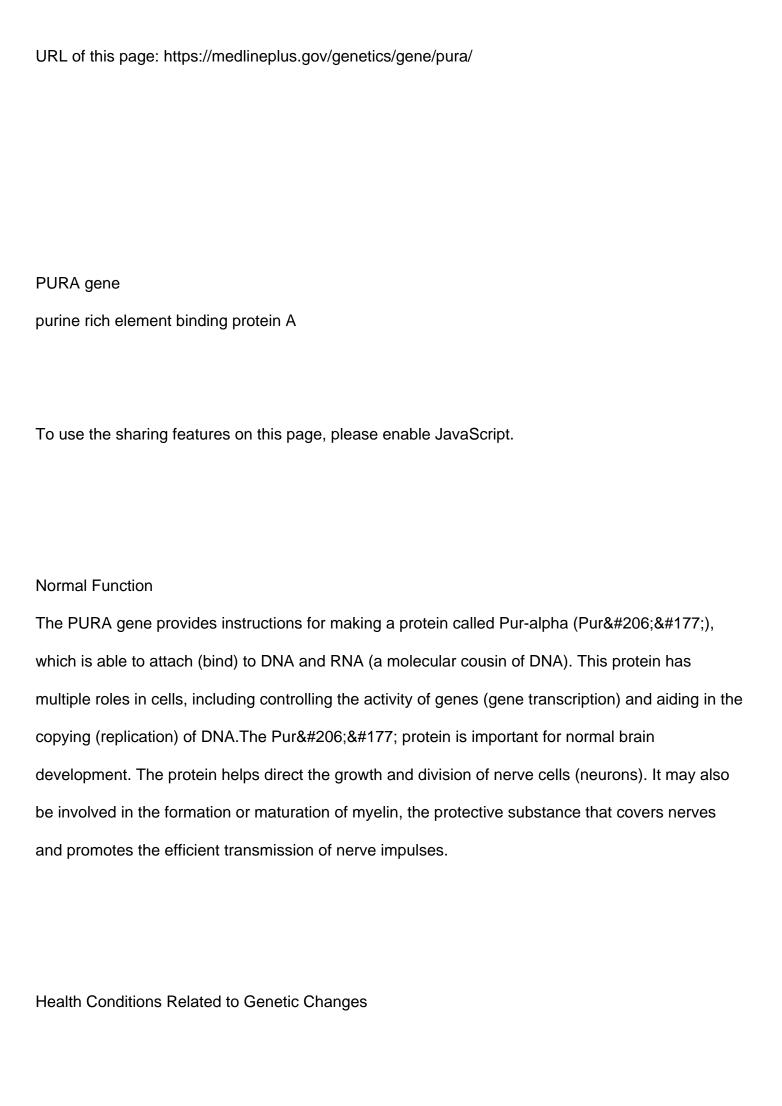
PURA And 5q31

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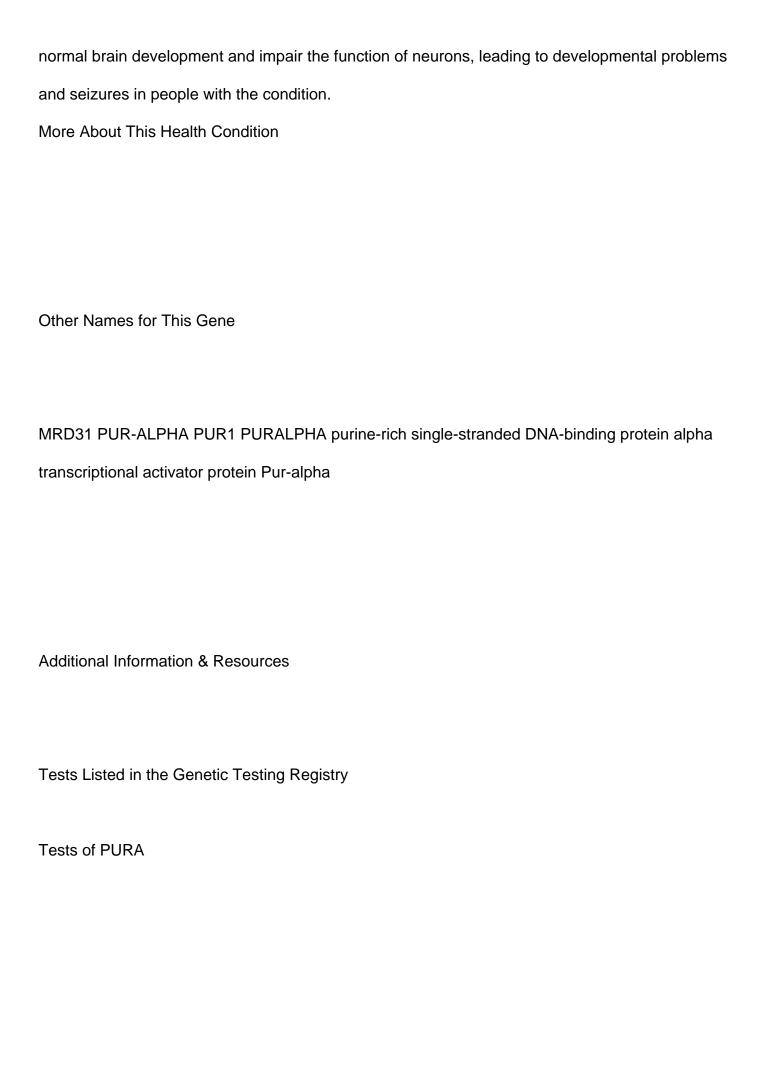


5q31.3 microdeletion syndrome

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



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References

Brown N, Burgess T, Forbes R, McGillivray G, Kornberg A, Mandelstam S, Stark Z. 5g31.3 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 Hokkanen S, Feldmann HM, Ding H, Jung CK, Bojarski L, Renner-Muller I, Schuller 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 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 5g31.3 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 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 5g31.3 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 or Free article on PubMed Central

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

Tanaka AJ, Bai R, Cho MT, Anyane-Yeboa K, Ahimaz P, Wilson AL, Kendall F, Hay

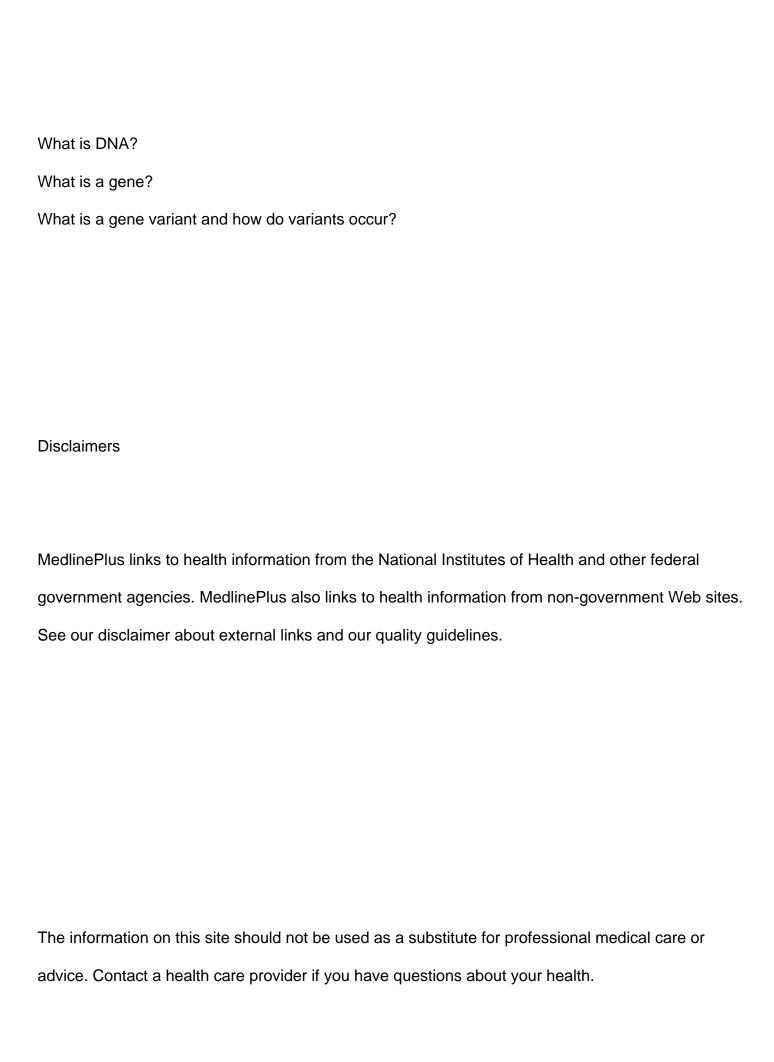
B, Moss T, Nardini M, Bauer M, Retterer K, Juusola J, Chung WK. De novo mutations

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 or Free article on PubMed Central

Weber J, Bao H, Hartlmuller 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:e11297. doi: 10.7554/eLife.11297. Citation on PubMed or Free article on PubMed Central White MK, Johnson EM, Khalili K. Multiple roles for Puralpha in cellular and viral regulation. Cell Cycle. 2009 Feb 1;8(3):1-7. doi: 10.4161/cc.8.3.7585. Epub 2009 Feb 10. Citation on PubMed or Free article on PubMed Central

Genomic LocationThe PURA gene is found on chromosome 5.
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