

RPS24 gene

ribosomal protein S24

Normal Function

The *RPS24* gene provides instructions for making one of approximately 80 different ribosomal proteins, which are components of cellular structures called ribosomes. Ribosomes process the cell's genetic instructions to create proteins.

Each ribosome is made up of two parts (subunits) called the large and small subunits. The protein produced from the *RPS24* gene is among those found in the small subunit.

The specific functions of the RPS24 protein and the other ribosomal proteins within these subunits are unclear. Some ribosomal proteins are involved in the assembly or stability of ribosomes. Others help carry out the ribosome's main function of building new proteins. Studies suggest that some ribosomal proteins may have other functions, such as participating in chemical signaling pathways within the cell, regulating cell division, and controlling the self-destruction of cells (apoptosis).

Health Conditions Related to Genetic Changes

Diamond-Blackfan anemia

At least 10 *RPS24* gene mutations have been identified in individuals with Diamond-Blackfan anemia. This disorder primarily affects the bone marrow, which produces new blood cells. People with this condition often also have physical abnormalities affecting various parts of the body.

The *RPS24* gene mutations that cause Diamond-Blackfan anemia are believed to cause problems with ribosomal function. Studies indicate that a shortage of functioning ribosomes may increase apoptosis of blood-forming cells in the bone marrow, resulting in a low number of red blood cells (anemia). Abnormal regulation of cell division or inappropriate triggering of apoptosis may contribute to the other health problems and unusual physical features that affect some people with Diamond-Blackfan anemia.

Other Names for This Gene

- 40S ribosomal protein S24
- DBA3

- eS24
- RS24_HUMAN
- S24

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RPS24 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6229\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6229[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RPS24%5BTIAB%5D%29+OR+%28ribosomal+protein+S24%5BTIAB%5D%29%29+OR+%28DBA3%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>)

Catalog of Genes and Diseases from OMIM

- RIBOSOMAL PROTEIN S24; RPS24 (<https://omim.org/entry/602412>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6229>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RPS24\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RPS24[gene]))

References

- Badhai J, Frojmark AS, J Davey E, Schuster J, Dahl N. Ribosomal protein S19 and S24 insufficiency cause distinct cell cycle defects in Diamond-Blackfan anemia. *Biochim Biophys Acta*. 2009 Oct;1792(10):1036-42. doi:10.1016/j.bbadis.2009.08.002. Epub 2009 Aug 16. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19689926>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2759502/>)
- Ball S. Diamond Blackfan anemia. *Hematology Am Soc Hematol Educ Program*. 2011;2011:487-91. doi: 10.1182/asheducation-2011.1.487. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22160079>)
- Boulton J, Pellagatti A, Wainscoat JS. Haploinsufficiency of ribosomal proteins and p53 activation in anemia: Diamond-Blackfan anemia and the 5q-syndrome. *Adv Biol Regul*. 2012 Jan;52(1):196-203. doi:10.1016/j.advenzreg.2011.09.008. No abstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21930148>)

- Choesmel V, Fribourg S, Aguisa-Toure AH, Pinaud N, Legrand P, Gazda HT, Gleizes PE. Mutation of ribosomal protein RPS24 in Diamond-Blackfan anemia results in a ribosome biogenesis disorder. *Hum Mol Genet*. 2008 May 1;17(9):1253-63. doi: 10.1093/hmg/ddn015. Epub 2008 Jan 29. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18230666>)
- Danilova N, Gazda HT. Ribosomopathies: how a common root can cause a tree of pathologies. *Dis Model Mech*. 2015 Sep;8(9):1013-26. doi: 10.1242/dmm.020529. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/26398160>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4582105/>)
- Ellis SR. Nucleolar stress in Diamond Blackfan anemia pathophysiology. *Biochim Biophys Acta*. 2014 Jun;1842(6):765-8. doi: 10.1016/j.bbadis.2013.12.013. Epub 2014 Jan 8. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24412987>)
- Gazda HT, Grabowska A, Merida-Long LB, Latawiec E, Schneider HE, Lipton JM, Vlachos A, Atsidaftos E, Ball SE, Orfali KA, Niewiadomska E, Da Costa L, Tchernia G, Niemeyer C, Meerpohl JJ, Stahl J, Schratt G, Glader B, Backer K, Wong C, Nathan DG, Beggs AH, Sieff CA. Ribosomal protein S24 gene is mutated in Diamond-Blackfan anemia. *Am J Hum Genet*. 2006 Dec;79(6):1110-8. doi:10.1086/510020. Epub 2006 Nov 2. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17186470>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1698708/>)
- Lipton JM, Ellis SR. Diamond-Blackfan anemia: diagnosis, treatment, and molecular pathogenesis. *Hematol Oncol Clin North Am*. 2009 Apr;23(2):261-82. doi:10.1016/j.hoc.2009.01.004. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19327583>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2886591/>)
- Mills EW, Green R. Ribosomopathies: There's strength in numbers. *Science*. 2017 Nov 3;358(6363):eaan2755. doi: 10.1126/science.aan2755. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/29097519>)
- Sieff C. Diamond-Blackfan Anemia. 2009 Jun 25 [updated 2023 Mar 23]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. *GeneReviews* (R) [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Available from <http://www.ncbi.nlm.nih.gov/books/NBK7047/> Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20301769>)
- Vlachos A, Blanc L, Lipton JM. Diamond Blackfan anemia: a model for the translational approach to understanding human disease. *Expert Rev Hematol*. 2014 Jun;7(3):359-72. doi: 10.1586/17474086.2014.897923. Epub 2014 Mar 26. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24665981>)

Genomic Location

The *RPS24* gene is found on chromosome 10 (<https://medlineplus.gov/genetics/chromosome/10/>).

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