



RPS17 gene

ribosomal protein S17

Normal Function

The *RPS17* 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 *RPS17* gene is among those found in the small subunit.

The specific functions of the RPS17 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

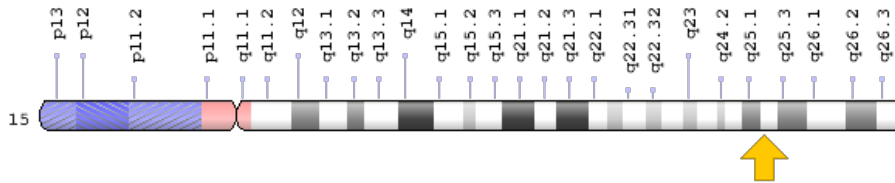
At least 18 *RPS17* gene mutations have been identified in individuals with Diamond-Blackfan anemia. This disorder primarily affects the bone marrow, which produces new blood cells. People with this condition often also have physical abnormalities affecting various parts of the body.

The *RPS17* 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.

Chromosomal Location

Cytogenetic Location: 15q25.2, which is the long (q) arm of chromosome 15 at position 25.2

Molecular Location: base pairs 82,536,750 to 82,540,564 on chromosome 15 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 40S ribosomal protein S17
- DBA4
- MGC72007
- RPS17L1
- RPS17L2
- RS17_HUMAN
- S17

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The RNA Message is Decoded on Ribosomes
<https://www.ncbi.nlm.nih.gov/books/NBK26829/#A1071>

Clinical Information from GeneReviews

- Diamond-Blackfan Anemia
<https://www.ncbi.nlm.nih.gov/books/NBK7047>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RPS17%5BTIAB%5D%29+OR+%28ribosomal+protein+S17%5BTIAB%5D%29%29+OR+%28%28DBA4%5BTIAB%5D%29+OR+%2840S+ribosomal+protein+S17%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>

Catalog of Genes and Diseases from OMIM

- RIBOSOMAL PROTEIN S17
<http://omim.org/entry/180472>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RPS17%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10397
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:6218>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6218>
- UniProt
<https://www.uniprot.org/uniprot/P08708>

Sources for This Summary

- Ball S. Diamond Blackfan anemia. Hematology Am Soc Hematol Educ Program. 2011;2011:487-91. doi: 10.1182/asheducation-2011.1.487. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22160079>
- Boulwood 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. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21930148>
- Cmejla R, Cmejlova J, Handrkova H, Petrak J, Pospisilova D. Ribosomal protein S17 gene (RPS17) is mutated in Diamond-Blackfan anemia. Hum Mutat. 2007 Dec;28(12):1178-82.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17647292>
- 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. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/26398160>
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. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24412987>

- Gazda HT, Sheen MR, Vlachos A, Choemmel V, O'Donohue MF, Schneider H, Darras N, Hasman C, Sieff CA, Newburger PE, Ball SE, Niewiadomska E, Matysiak M, Zaucha JM, Glader B, Niemeyer C, Meerpohl JJ, Atsidaftos E, Lipton JM, Gleizes PE, Beggs AH. Ribosomal protein L5 and L11 mutations are associated with cleft palate and abnormal thumbs in Diamond-Blackfan anemia patients. *Am J Hum Genet.* 2008 Dec;83(6):769-80. doi: 10.1016/j.ajhg.2008.11.004.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19061985>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2668101/>
- Kenney SP, Meng XJ. Identification and fine mapping of nuclear and nucleolar localization signals within the human ribosomal protein S17. *PLoS One.* 2015 Apr 8;10(4):e0124396. doi: 10.1371/journal.pone.0124396. eCollection 2015.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/25853866>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4390217/>
- Mills EW, Green R. Ribosomopathies: There's strength in numbers. *Science.* 2017 Nov 3; 358(6363). pii: eaan2755. doi: 10.1126/science.aan2755. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/29097519>
- OMIM: RIBOSOMAL PROTEIN S17
<http://omim.org/entry/180472>
- Song MJ, Yoo EH, Lee KO, Kim GN, Kim HJ, Kim SY, Kim SH. A novel initiation codon mutation in the ribosomal protein S17 gene (RPS17) in a patient with Diamond-Blackfan anemia. *Pediatr Blood Cancer.* 2010 Apr;54(4):629-31. doi: 10.1002/pbc.22316.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19953637>
- 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. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24665981>

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