

## RPS10 gene

ribosomal protein S10

### Normal Function

The *RPS10* 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 *RPS10* gene is among those found in the small subunit.

The specific functions of the RPS10 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 five *RPS10* 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 *RPS10* gene mutations that cause Diamond-Blackfan anemia are believed to result in an abnormally short, nonfunctional RPS10 protein that may impair the assembly of ribosomes, but the specific effects of the mutations are not known. 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 S10
- DBA9
- MGC88819
- RS10\_HUMAN
- S10

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of RPS10 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6204\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6204[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RPS10%5BTIAB%5D%29+OR+%28ribosomal+protein+S10%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+1440+days%22%5Bdp%5D%29%29%29>)

### Catalog of Genes and Diseases from OMIM

- RIBOSOMAL PROTEIN S10; RPS10 (<https://omim.org/entry/603632>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6204>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RPS10\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RPS10[gene]))

## References

- 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>)
- 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. doi:10.1016/j.advenzreg.2011.09.008. No abstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21930148>)
- 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/>)

- Doherty L, Sheen MR, Vlachos A, Choesmel V, O’Donohue MF, Clinton C, SchneiderHE, Sieff CA, Newburger PE, Ball SE, Niewiadomska E, Matysiak M, Glader B, ArceciRJ, Farrar JE, Atsidaftos E, Lipton JM, Gleizes PE, Gazda HT. Ribosomal proteingenes RPS10 and RPS26 are commonly mutated in Diamond-Blackfan anemia. *Am J HumGenet.* 2010 Feb 12;86(2):222-8. doi: 10.1016/j.ajhg.2009.12.015. Epub 2010 Jan28. Erratum In: *Am J Hum Genet.* 2010 Apr 9;86(4):655. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20116044/>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2820177/>)
- Ellis SR, Gleizes PE. Diamond Blackfan anemia: ribosomal proteins going rogue. *Semin Hematol.* 2011 Apr;48(2):89-96. doi: 10.1053/j.seminhematol.2011.02.005. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21435505/>)
- Ellis SR. Nucleolar stress in Diamond Blackfan anemia pathophysiology. *BiochimBiophys Acta.* 2014 Jun;1842(6):765-8. doi: 10.1016/j.bbadis.2013.12.013. Epub2014 Jan 8. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24412987/>)
- Farrar JE, Dahl N. Untangling the phenotypic heterogeneity of Diamond Blackfan anemia. *Semin Hematol.* 2011 Apr;48(2):124-35. doi:10.1053/j.seminhematol.2011.02.003. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21435509/>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3078697/>)
- Mills EW, Green R. Ribosomopathies: Their strength in numbers. *Science.* 2017Nov 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: AdamMP, 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.* 2014Jun;7(3):359-72. doi: 10.1586/17474086.2014.897923. Epub 2014 Mar 26. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24665981/>)
- Yazaki M, Kamei M, Ito Y, Konno Y, Wang R, Toki T, Ito E. A novel mutation of ribosomal protein S10 gene in a Japanese patient with diamond-Blackfan anemia. *JPediatr Hematol Oncol.* 2012 May;34(4):293-5. doi: 10.1097/MPH.0b013e31824a20ab. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22510774/>)

## Genomic Location

The *RPS10* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

**Last updated September 1, 2018**