RPS19 gene

ribosomal protein S19

Normal Function

The *RPS19* 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 *RPS19* gene is among those found in the small subunit.

The specific functions of the RPS19 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

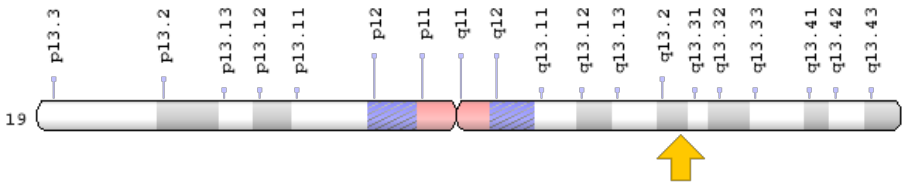
More than 170 *RPS19* gene mutations have been identified in individuals with Diamond-Blackfan anemia. This disorder primarily affects the bone marrow, which produces new blood cells. People with this condition often also have physical abnormalities affecting various parts of the body.

The *RPS19* 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: 19q13.2, which is the long (q) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 41,859,918 to 41,871,416 on chromosome 19 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 40S ribosomal protein S19
- DBA
- eS19
- RS19_HUMAN
- S19

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%28RPS19%5BTIAB%5D%29+OR+%28ribosomal+protein+S19%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

- RIBOSOMAL PROTEIN S19
  http://omim.org/entry/603474

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_RPS19.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RPS19%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10402
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:6223
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6223
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