



RPL11 gene

ribosomal protein L11

Normal Function

The *RPL11* 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 *RPL11* gene is among those found in the large subunit.

The specific functions of the RPL11 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

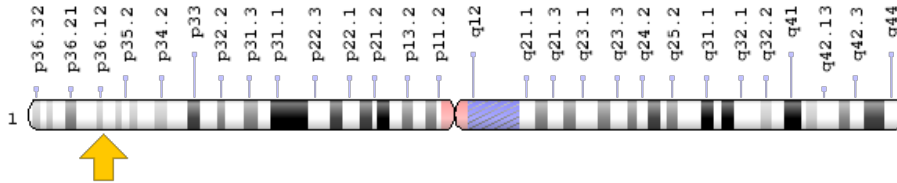
At least 44 *RPL11* gene mutations have been identified in individuals with Diamond-Blackfan anemia. This disorder primarily affects the bone marrow, which produces new blood cells. People with this condition often also have physical abnormalities affecting various parts of the body.

The *RPL11* 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: 1p36.11, which is the short (p) arm of chromosome 1 at position 36.11

Molecular Location: base pairs 23,691,779 to 23,696,835 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 60S ribosomal protein L11
- cell growth-inhibiting protein 34
- CLL-associated antigen KW-12
- DBA7
- GIG34
- L11
- RL11_HUMAN
- uL5

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%28RPL11%5BTIAB%5D%29+OR+%28ribosomal+protein+L11%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 L11
<http://omim.org/entry/604175>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_RPL11.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RPL11%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10301
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:6135>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6135>
- UniProt
<https://www.uniprot.org/uniprot/P62913>

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<https://ghr.nlm.nih.gov/gene/RPL11>

Reviewed: September 2018
Published: September 10, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services