

## RPL5 gene

ribosomal protein L5

### Normal Function

The *RPL5* 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 *RPL5* gene is among those found in the large subunit.

The specific functions of the RPL5 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).

Research suggests that the protein produced from the *RPL5* gene also normally has tumor suppressor function, which means that it helps keep cells from growing and dividing too rapidly or in an uncontrolled way.

### Health Conditions Related to Genetic Changes

#### Diamond-Blackfan anemia

More than 70 *RPL5* 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 *RPL5* gene mutations that cause Diamond-Blackfan anemia are either inherited or occur early in development before birth, and occur in every cell in the body. The mutations 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.

## Cancers

Mutations and deletions affecting the *RPL5* gene have been identified in cancerous tumors of several types, including breast cancer, a type of brain tumor called glioblastoma, and a skin cancer called melanoma. These genetic changes are somatic, which means that they are not inherited and occur only in the cancer cells. Mutations or deletions in the *RPL5* gene that interfere with the RPL5 protein's tumor suppressor function can result in the uncontrolled cell growth and division that leads to cancer.

## **Other Names for This Gene**

- 60S ribosomal protein L5
- DBA6
- L5
- MGC117339
- MSTP030
- PPP1R135
- RL5\_HUMAN
- uL18

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of RPL5 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6125\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6125[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RPL5%5BTIAB%5D%29+OR+%28ribosomal+protein+L5%5BTIAB%5D%29%29+OR+%2860S+ribosomal+protein+L5%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%29%29>)

### Catalog of Genes and Diseases from OMIM

- RIBOSOMAL PROTEIN L5; RPL5 (<https://omim.org/entry/603634>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6125>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RPL5\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RPL5[gene]))

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## Genomic Location

The *RPL5* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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