

RPS14 gene

ribosomal protein S14

Normal Function

The *RPS14* 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 *RPS14* gene is among those found in the small subunit.

The specific functions of the RPS14 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

5q minus syndrome

The *RPS14* gene is involved in a condition called 5q minus (5q-) syndrome. This condition is a type of bone marrow disorder called myelodysplastic syndrome (MDS), in which immature blood cells fail to develop normally. Individuals with 5q- syndrome often have a shortage of red blood cells (anemia) and abnormalities in blood cells called megakaryocytes, which produce platelets, the cells involved in blood clotting. Affected individuals also have an increased risk of developing a fast-growing blood cancer known as acute myeloid leukemia (AML).

5q- syndrome is caused by deletion of a region of DNA from the long (q) arm of chromosome 5. This deletion occurs in immature blood cells during a person's lifetime and affects one copy of chromosome 5 in each cell. Most people with 5q- syndrome are missing a sequence of about 1.5 million DNA building blocks (base pairs), also written as 1.5 megabases (Mb). This region of DNA contains 40 genes, including *RPS14*. Loss of one copy of the *RPS14* gene reduces the amount of RPS14 protein that is made. Studies indicate that a shortage of functioning ribosomal proteins increases the self-destruction of blood-forming cells in the bone marrow, resulting in anemia. Research

suggests that the other features of the condition are associated with other genes in the deleted segment of DNA.

Other Names for This Gene

- 40S ribosomal protein S14
- emetine resistance
- EMTB
- S14

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RPS14 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6208\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6208[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RPS14%5BTIAB%5D%29+OR+%28ribosomal+protein+S14%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%29%29%29>)

Catalog of Genes and Diseases from OMIM

- RIBOSOMAL PROTEIN S14; RPS14 (<https://omim.org/entry/130620>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6208>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RPS14\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RPS14[gene]))

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Genomic Location

The *RPS14* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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