

RPSA gene

ribosomal protein SA

Normal Function

The *RPSA* gene provides instructions for making a protein called ribosomal protein SA, which is one of approximately 80 different ribosomal proteins. These proteins come together to form 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 subunit and the small subunit. Ribosomal protein SA is part of the small subunit.

The specific roles of each of the ribosomal proteins within the ribosome are not entirely understood. 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. Research suggests that ribosomal protein SA helps the ribosome control the production of certain proteins, many of which are likely important for development before birth.

Health Conditions Related to Genetic Changes

Isolated congenital asplenia

At least 20 *RPSA* gene mutations have been identified in individuals with isolated congenital asplenia. People with this condition do not have a spleen but have no other developmental abnormalities. The spleen plays an important role in the immune system. Without this organ, affected individuals are highly susceptible to bacterial infections, which can be life-threatening.

RPSA gene mutations are thought to reduce the amount of functional ribosomal protein SA. A shortage of the normal protein likely impairs the assembly of ribosomes, but the specific effects of the mutations are not known. It is unclear why *RPSA* gene mutations solely affect the development of the spleen.

Other Names for This Gene

- 40S Ribosomal Protein SA
- LAMBR
- Laminin Receptor

- LAMININ RECEPTOR 1
- laminin receptor 1, human
- LAMININ RECEPTOR, 67-KD
- Laminin Receptor-1
- LAMR1
- Lamr1 protein, human
- Ribosomal Protein SA Gene
- RPSA Gene

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RPSA ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3921\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3921[geneid]))

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- RIBOSOMAL PROTEIN SA; RPSA (<https://omim.org/entry/150370>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3921>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RPSA\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RPSA[gene]))

References

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Genomic Location

The *RPSA* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

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