

RARS2 gene

arginyl-tRNA synthetase 2, mitochondrial

Normal Function

The *RARS2* gene provides instructions for making an enzyme called mitochondrial arginyl-tRNA synthetase. This enzyme is active in cell structures called mitochondria. Each cell contains hundreds or thousands of mitochondria, which convert the energy from food into a form that cells can use.

Mitochondrial arginyl-tRNA synthetase interacts with a molecule called transfer RNA (tRNA). This molecule, which is a chemical cousin of DNA, helps assemble protein building blocks called amino acids into functioning proteins. To build new proteins, tRNA must collect different amino acids and then attach them to one another in the correct order. Mitochondrial arginyl-tRNA synthetase is one of several enzymes that link amino acids to tRNA. Specifically, this enzyme links the amino acid arginine to the tRNA molecule, which then incorporates it into new proteins in mitochondria.

Health Conditions Related to Genetic Changes

Pontocerebellar hypoplasia

At least 13 mutations in the *RARS2* gene have been identified in people with a disorder of brain development called pontocerebellar hypoplasia. The major features of this condition include delayed development, problems with movement, and intellectual disability. Most of the known *RARS2* gene mutations cause a form of the disorder designated pontocerebellar hypoplasia type 6 (PCH6). One mutation has been found in an individual with the characteristic features of another form of the condition, pontocerebellar hypoplasia type 1 (PCH1).

The *RARS2* gene mutations that cause pontocerebellar hypoplasia significantly reduce or eliminate the function of mitochondrial arginyl-tRNA synthetase. A loss of this enzyme's function likely disrupts the production of new proteins in mitochondria. However, it is unknown how these changes lead to abnormal brain development in people with pontocerebellar hypoplasia.

Other Names for This Gene

- arginine-tRNA ligase

- arginyl-tRNA synthetase 2, mitochondrial precursor
- arginyl-tRNA synthetase-like
- ArgRS
- DALRD2
- dJ382I10.6
- MGC14993
- MGC23778
- PRO1992
- RARSL
- SYRM_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RARS2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57038\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57038[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RARS2%5BTIAB%5D%29+OR+%28RARSL%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

- ARGINYL-tRNA SYNTHETASE 2; RARS2 (<https://omim.org/entry/611524>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/57038>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RARS2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RARS2[gene]))

References

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Genomic Location

The *RARS2* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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