



RERE gene

arginine-glutamic acid dipeptide repeats

Normal Function

The *RERE* gene provides instructions for making a protein that is critical for normal development before birth. This protein interacts with other proteins called transcription factors, which control the activity (transcription) of particular genes. The RERE protein helps these transcription factors turn on (activate) and turn off (repress) a number of genes important for early development, ensuring that the genes are activated (expressed) at the right time and place for proper tissue formation. Research indicates that the RERE protein plays a role in the development of the brain, eyes, inner ear, heart, and kidneys.

Health Conditions Related to Genetic Changes

Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart

More than 15 mutations in the *RERE* gene have been found to cause a condition called neurodevelopmental disorder with or without anomalies of the brain, eye, or heart (NEDBEH). This condition primarily affects brain development, causing intellectual disability, delayed development, or autism spectrum disorder, which is a condition that affects communication and social interaction. Abnormalities of other body systems, including the eyes, heart, inner ear, or kidneys, can also occur.

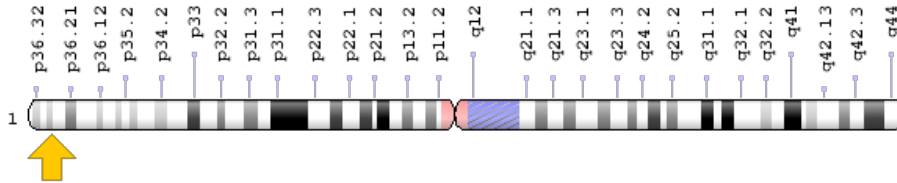
Many of the *RERE* gene mutations that cause NEDBEH change single DNA building blocks (nucleotides). Other mutations add or remove nucleotides. Researchers suspect that these genetic changes reduce or eliminate the function of the RERE protein. A shortage of RERE protein function likely alters the activity of many genes involved in development before birth. These changes prevent the normal development of tissues in the brain, eyes, heart, and other organs. Researchers are working to identify which genes are affected and how changes in their activity lead to the signs and symptoms of NEDBEH.

It is unknown why some people with NEDBEH have only neurological problems and others also have structural abnormalities. Researchers suspect that the severity of the condition may be related to the location and type of mutation in the *RERE* gene. Additional genetic factors that have not been identified, including variations in other genes, may also help determine which body systems are affected.

Chromosomal Location

Cytogenetic Location: 1p36.23, which is the short (p) arm of chromosome 1 at position 36.23

Molecular Location: base pairs 8,352,404 to 8,817,640 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARG
- arginine-glutamic acid dipeptide (RE) repeats
- arginine-glutamic acid repeats-encoding gene
- ARP
- ATN1L
- atrophin-1 like protein
- atrophin-1 related protein
- atrophin 2
- atrophn-related protein
- deleted in neuroblastoma-1
- DNB1
- KIAA0458

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Transcription Factors
https://www.ncbi.nlm.nih.gov/books/NBK10023/#_A763_

Clinical Information from GeneReviews

- RERE-Related Disorders
<https://www.ncbi.nlm.nih.gov/books/NBK538938>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RERE%5BTIAB%5D%29+OR+%28arginine-glutamic+acid+dipeptide+repeats%5BTIAB%5D%29%29+OR+%28%28NEDBEH%5BTIAB%5D%29+OR+%28atrophin+2%5BTIAB%5D%29+OR+%28atrophin-1+like+protein%5BTIAB%5D%29+OR+%28atrophin-1+related+protein%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

- RE REPEATS-ENCODING GENE
<http://omim.org/entry/605226>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_RERE.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RERE%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:9965
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:473>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/473>
- UniProt
<https://www.uniprot.org/uniprot/Q9P2R6>

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