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.

Other Names for This Gene

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RERE (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=473[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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

- ARGININE-GLUTAMIC ACID DIPEPTIDE REPEATS; RERE (https://omim.org/entry/605226)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/473)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=RERE[gene])

References

- Fregeau B, Kim BJ, Hernandez-Garcia A, Jordan VK, Cho MT, Schnur RE, MonaghanKG, Juusola J, Rosenfeld JA, Bhoj E, Zackai EH, Sacharow S, Baranano K, BoschDGM, de Vries BBA, Lindstrom K, Schroeder A, James P, Kulch P, Lalani SR, vanHaelst MM, van Gassen KLI, van Binsbergen E, Barkovich AJ, Scott DA, Sherr EH. DeNovo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap ThoseAssociated with Proximal 1p36 Deletions. Am J Hum Genet. 2016 May5;98(5):963-970. doi: 10.1016/j.ajhg.2016.03.002. Epub 2016 Apr 14. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/27087320) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4863473/)
- Jordan VK, Fregeau B, Ge X, Giordano J, Wapner RJ, Balci TB, Carter MT, BernatJA, Moccia AN, Srivastava A, Martin DM, Bielas SL, Pappas J, Svoboda MD, Rio M,Boddaert N, Cantagrel V, Lewis AM, Scaglia F; Undiagnosed Diseases Network;Kohler JN, Bernstein JA, Dries AM, Rosenfeld JA, DeFilippo C, Thorson W, Yang Y,Sherr EH, Bi W, Scott DA. Genotype-phenotype correlations in individuals withpathogenic RERE variants. Hum Mutat. 2018 May;39(5):666-675. doi:10.1002/humu.23400. Epub 2018 Jan 25. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/29330883) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5903952/)
- Kim BJ, Scott DA. Mouse model reveals the role of RERE in cerebellar foliationand the migration and maturation of Purkinje cells. PLoS One. 2014 Jan23;9(1):e87518. doi: 10.1371/journal.pone.0087518. eCollection 2014. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24466353) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3900724/)
- Kim BJ, Zaveri HP, Jordan VK, Hernandez-Garcia A, Jacob DJ, Zamora DL, Yu W, Schwartz RJ, Scott DA. RERE deficiency leads to decreased expression of GATA4 andthe development of ventricular septal defects. Dis Model Mech. 2018 Aug28;11(9):dmm031534. doi: 10.1242/dmm.031534. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/30061196)
- Kim BJ, Zaveri HP, Shchelochkov OA, Yu Z, Hernandez-Garcia A, Seymour ML, Oghalai JS, Pereira FA, Stockton DW, Justice MJ, Lee B, Scott DA. An allelicseries of mice reveals a role for RERE in the development of multiple organsaffected in chromosome 1p36 deletions. PLoS One. 2013;8(2):e57460. doi:10.1371/journal.pone.0057460. Epub 2013 Feb 25. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23451234) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3581587/)
- Vilhais-Neto GC, Maruhashi M, Smith KT, Vasseur-Cognet M, Peterson AS, WorkmanJL, Pourquie O. Rere controls retinoic acid signalling and somite bilateralsymmetry. Nature. 2010 Feb 18;463(7283):953-7. doi: 10.1038/nature08763. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20164929)
- Zoltewicz JS, Stewart NJ, Leung R, Peterson AS. Atrophin 2 recruits histonedeacetylase and is required for the function of multiple signaling centers duringmouse embryogenesis. Development. 2004 Jan;131(1):3-14. doi: 10.1242/dev.00908.Epub 2003 Nov 26. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14645126)

Genomic Location

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

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