



PPP2R5D gene

protein phosphatase 2 regulatory subunit B'delta

Normal Function

The *PPP2R5D* gene provides instructions for making a protein called B56-delta (B56 δ). This protein is one piece (the B subunit) of an enzyme called protein phosphatase 2A (PP2A). (B56 δ is one of several possible B subunits.) The PP2A enzyme removes phosphate groups, consisting of three oxygen atoms and one phosphorus atom, from certain proteins. This process, called dephosphorylation, helps control whether the protein is turned on or off. The B subunit determines which proteins are dephosphorylated by PP2A and regulates the activity of the enzyme.

PP2A removes phosphate groups from proteins that are part of signaling pathways involved in cell growth and turning genes on and off. PP2A enzymes containing the B56 δ protein are found mainly in the brain, where they are thought to be primarily involved in controlling the activity of signaling pathways that play roles in the normal development and function of nerve cells (neurons).

Health Conditions Related to Genetic Changes

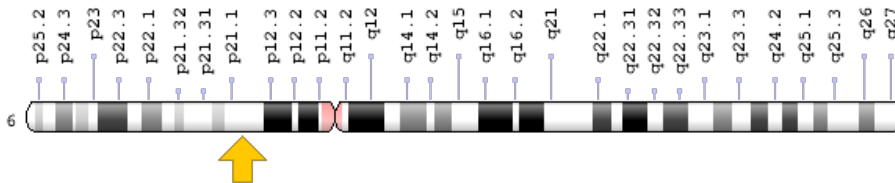
PPP2R5D-related intellectual disability

At least seven *PPP2R5D* gene mutations have been found to cause moderate to severe intellectual disability. Individuals with *PPP2R5D*-related intellectual disability typically also have weak muscle tone (hypotonia) and delayed development of speech and walking. These mutations affect one copy of the gene. They change single protein building blocks (amino acids) in the B56 δ protein. Although the effects of these changes are unclear, researchers suspect that they alter or impair the activity of the PP2A enzyme. Abnormal or reduced PP2A enzyme activity is thought to disrupt signaling pathways in neurons, impairing their normal development and functioning, which may underlie intellectual disability and other features of *PPP2R5D*-related intellectual disability.

Chromosomal Location

Cytogenetic Location: 6p21.1, which is the short (p) arm of chromosome 6 at position 21.1

Molecular Location: base pairs 42,984,499 to 43,012,345 on chromosome 6 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- B56D
- B56delta
- MRD35
- PP2A, B subunit, B' delta isoform
- PP2A, B subunit, B56 delta isoform
- PP2A, B subunit, PR61 delta isoform
- PP2A, B subunit, R5 delta isoform
- protein phosphatase 2, regulatory subunit B (B56), delta isoform
- Serine/threonine protein phosphatase 2A, 56 kDa regulatory subunit, delta isoform

Additional Information & Resources

Educational Resources

- Basic Neurochemistry: Molecular, Cellular and Medical Aspects (sixth edition, 1999): Protein Serine-Threonine Phosphatases
<https://www.ncbi.nlm.nih.gov/books/NBK28069/>

Clinical Information from GeneReviews

- PPP2R5D-Related Neurodevelopmental Disorder
<https://www.ncbi.nlm.nih.gov/books/NBK536360>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PPP2R5D%5BTIAB%5D%29+OR+%28protein+phosphatase+2+regulatory+subunit+B'delta%5BTIAB%5D%29%29+OR+%28%28B56delta%5BTIAB%5D%29+OR+%28serine/threonine-protein+phosphatase+2A+56+kDa+regulatory+subunit+delta+isoform+isoform+1%5BTIAB%5D%29+OR+%28serine/threonine-protein+phosphatase+2A+56+kDa+regulatory+subunit+delta+isoform+isoform+2%5BTIAB%5D%29+OR+%28serine/threonine-protein+phosphatase+2A+56+kDa+regulatory+subunit+delta+isoform+isoform+3%5BTIAB%5D%29+OR+%28serine/threonine-protein+phosphatase+2A+56+kDa+regulatory+subunit+delta+isoform+isoform+4%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

- PROTEIN PHOSPHATASE 2, REGULATORY SUBUNIT B (B56), DELTA
<http://omim.org/entry/601646>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PPP2R5D.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PPP2R5D%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:9312
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:5528>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/5528>
- UniProt
<https://www.uniprot.org/uniprot/Q14738>

Sources for This Summary

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<https://ghr.nlm.nih.gov/gene/PPP2R5D>

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