

## PPP2R5D gene

protein phosphatase 2 regulatory subunit B&#x27;delta

### Normal Function

The *PPP2R5D* gene provides instructions for making a protein called B56-delta (B56δ). This protein is one piece (the B subunit) of an enzyme called protein phosphatase 2A (PP2A). (B56δ is one of several possible B subunits.) The PP2A enzyme removes phosphate groups, consisting of clusters of oxygen and phosphorus atoms, 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 B56δ 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 B56δ 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.

### Other Names for This Gene

- B56D
- B56delta

- MRD35
- PP2A, B subunit, B&#x27; 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**

### Tests Listed in the Genetic Testing Registry

- Tests of PPP2R5D ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5528\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5528[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PPP2R5D%5BTIAB%5D%29+OR+%28protein+phosphatase+2+regulatory+subunit+B&#x27;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; PPP2R5D (<https://omim.org/entry/601646>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5528>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PPP2R5D\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PPP2R5D[gene]))

## References

- Janssens V, Goris J. Protein phosphatase 2A: a highly regulated family of serine/threonine phosphatases implicated in cell growth and signalling. *Biochem J*. 2001 Feb 1;353(Pt 3):417-39. doi: 10.1042/0264-6021:3530417. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11171037>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1221586/>)
- Loveday C, Tatton-Brown K, Clarke M, Westwood I, Renwick A, Ramsay E, Nemeth A, Campbell J, Joss S, Gardner M, Zachariou A, Elliott A, Ruark E, van Montfort R; Childhood Overgrowth Collaboration; Rahman N. Mutations in the PP2A regulatory subunit B family genes PPP2R5B, PPP2R5C and PPP2R5D cause human overgrowth. *Hum Mol Genet*. 2015 Sep 1;24(17):4775-9. doi: 10.1093/hmg/ddv182. Epub 2015 May 13. Erratum In: *Hum Mol Genet*. 2019 May 1;28(9):1578. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25972378>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4527483/>)
- Shang L, Henderson LB, Cho MT, Petrey DS, Fong CT, Haude KM, Shur N, Lundberg J, Hauser N, Carmichael J, Innis J, Schuette J, Wu YW, Asaika S, Pearson M, Folk L, Retterer K, Monaghan KG, Chung WK. De novo missense variants in PPP2R5D are associated with intellectual disability, macrocephaly, hypotonia, and autism. *Neurogenetics*. 2016 Jan;17(1):43-9. doi: 10.1007/s10048-015-0466-9. Epub 2015 Nov 17. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/26576547>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4765493/>)
- Yu UY, Yoo BC, Ahn JH. Regulatory B Subunits of Protein Phosphatase 2A Are Involved in Site-specific Regulation of Tau Protein Phosphorylation. *Korean J Physiol Pharmacol*. 2014 Apr;18(2):155-61. doi: 10.4196/kjpp.2014.18.2.155. Epub 2014 Apr 3. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24757378>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3994303/>)

## Genomic Location

The *PPP2R5D* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

**Last updated August 1, 2017**