

DLG4 gene

discs large MAGUK scaffold protein 4

Normal Function

The *DLG4* gene provides instructions for making a protein that plays a role in nerve cells (neurons) in the brain. The DLG4 protein is found at synapses, which are the connections between neurons where cell-to-cell communication occurs. Connected nerve cells act as the "wiring" in the circuitry, carrying signals between the brain and the rest of the body. At synapses, the DLG4 protein interacts with other proteins to regulate a process called synaptic plasticity, which allows synapses to change and adapt over time in response to experiences. Synaptic plasticity is critical for learning and memory.

Health Conditions Related to Genetic Changes

DLG4-related synaptopathy

Variants (also called mutations) in the *DLG4* gene cause *DLG4*-related synaptopathy. This condition affects neurological development and often causes intellectual disabilities, delayed development, movement disorders, and seizures. Most of the variants impair the normal function of the DLG4 protein by decreasing its ability to interact with the other proteins involved in synaptic plasticity or decreasing its ability to help synapses send signals. Other variants prevent the production of any normal DLG4 protein. As a result, synaptic plasticity and other synaptic functions are impaired. The signs and symptoms of *DLG4*-related synaptopathy are likely caused by the reduced ability of synapses to change and adapt during important periods of brain development.

Other Names for This Gene

- discs large, drosophila, homolog of, 4
- postsynaptic density 95
- PSD95
- SAP90
- synapse-associated protein 90

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DLG4 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1742\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1742[geneid]))

Scientific Articles on PubMed

- PubMed ([https://pubmed.ncbi.nlm.nih.gov/?term=\(DLG4%5BTtitle%5D\)+OR+\(PSD-95%5BTtitle%5D\)&filter=hum_ani.humans&filter=lang.english&sort=date\)](https://pubmed.ncbi.nlm.nih.gov/?term=(DLG4%5BTtitle%5D)+OR+(PSD-95%5BTtitle%5D)&filter=hum_ani.humans&filter=lang.english&sort=date)))

Catalog of Genes and Diseases from OMIM

- DISCS LARGE MAGUK SCAFFOLD PROTEIN 4; DLG4 (<https://omim.org/entry/602887>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1742>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=DLG4\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=DLG4[gene]))

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