

DLL3 gene

delta like canonical Notch ligand 3

Normal Function

The *DLL3* gene provides instructions for making a protein that helps control (regulate) the Notch pathway, an important pathway in embryonic development. The Notch pathway plays a critical role in the development of vertebrae. Specifically, the DLL3 protein and the Notch pathway are involved in separating future vertebrae from one another during early development, in a complex process called somite segmentation. Although the exact mechanism of somite segmentation is unclear, it appears to require the activity of several proteins in the Notch pathway, including the NOTCH1 protein, to be turned on and off (oscillate) in a specific pattern.

The DLL3 protein regulates the activity of the NOTCH1 protein. The DLL3 protein attaches (binds) to the inactive NOTCH1 protein and isolates (sequesters) it or marks it to be broken down so that it cannot be activated.

Health Conditions Related to Genetic Changes

Spondylocostal dysostosis

At least 20 mutations in the *DLL3* gene have been found to cause spondylocostal dysostosis type 1, the most common type of spondylocostal dysostosis. This condition is characterized by the abnormal development of bones in the spine and ribs. The known mutations in the *DLL3* gene prevent the production of any protein or lead to the production of an abnormally short, nonfunctional protein. When the DLL3 protein is nonfunctional or absent, the NOTCH1 protein is abnormally active and does not oscillate, so somite segmentation does not occur properly. This results in the malformation and fusion of the bones of the spine and ribs seen in spondylocostal dysostosis type 1.

Other Names for This Gene

- delta-like 3 (Drosophila)
- delta-like protein 3
- delta-like protein 3 isoform 1 precursor
- delta-like protein 3 isoform 2 precursor

- delta3
- DLL3_HUMAN
- drosophila Delta homolog 3
- SCDO1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DLL3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10683\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10683[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DLL3%5BTIAB%5D%29+OR+%28delta-like+3%5BTIAB%5D%29%29+OR+%28SCDO1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- DELTA-LIKE CANONICAL NOTCH LIGAND 3; DLL3 (<https://omim.org/entry/602768>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10683>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=DLL3\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=DLL3[gene]))

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Genomic Location

The *DLL3* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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