DVL3 gene

dishevelled segment polarity protein 3

Normal Function

The *DVL3* gene provides instructions for making a protein that plays a critical role in development before birth. It is one of three DVL genes in humans (*DVL1*, *DVL2*, and *DVL3*). The proteins produced from these genes work together in chemical signaling pathways known as Wnt signaling. These pathways control the activity of certain genes and regulate the interactions between cells during embryonic development. Signaling involving the DVL proteins appears to be important for the normal development of the brain, skeleton, and many other parts of the body.

Health Conditions Related to Genetic Changes

Robinow syndrome

At least six mutations in the *DVL3* gene have been found to cause the autosomal dominant form of Robinow syndrome, a condition that affects the development of many parts of the body, particularly the skeleton. Autosomal dominant inheritance means that one copy of the altered gene in each cell is sufficient to cause the disorder.

All of the identified *DVL3* gene mutations occur near one end of the gene and are predicted to shorten the DVL3 protein. Researchers are working to determine how these changes affect the protein's function. The changes may have a dominant-negative effect, which means that the altered protein produced from one copy of the *DVL3* gene interferes with the function of the normal protein produced from the other copy of the gene. Alternately, the changes may have a gain-of-function effect, giving the altered protein a new, as-yet-undetermined function. Either way, the abnormal DVL3 protein likely impairs Wnt signaling. Problems with Wnt signaling pathways disrupt the development of many organs and tissues before birth, leading to Robinow syndrome.

Other Names for This Gene

- dishevelled 3 (homologous to Drosophila dsh)
- dishevelled, dsh homolog 3
- DRS3
- KIAA0208

- segment polarity protein dishevelled homolog DVL-3

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DVL3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1857[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DVL3%5BTIAB%5D%29+OR+%28dishevelled+segment+polarity+protein+3%5BTIAB%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

- DISHEVELLED 3; DVL3 (https://omim.org/entry/601368)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1857)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DVL3[gene])

References

- Pizzuti A, Amati F, Calabrese G, Mari A, Colosimo A, Silani V, Giardino L,Ratti A, Penso D, Calza L, Palka G, Scarlato G, Novelli G, Dallapiccola B. cDNAcharacterization and chromosomal mapping of two human homologues of theDrosophila dishevelled polarity gene. Hum Mol Genet. 1996 Jul;5(7):953-8. doi: 10.1093/hmg/5.7.953. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/8817329)
- Semenov MV, Snyder M. Human dishevelled genes constitute a DHR-containingmultigene family. Genomics. 1997 Jun 1;42(2):302-10. doi: 10.1006/geno.1997.4713. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9192851)
- White JJ, Mazzeu JF, Coban-Akdemir Z, Bayram Y, Bahrambeigi V, Hoischen A, vanBon BWM, Gezdirici A, Gulec EY, Ramond F, Touraine R, Thevenon J, Shinawi M,Beaver E, Heeley J, Hoover-Fong J, Durmaz CD, Karabulut HG, Marzioglu-Ozdemir E,Cayir A, Duz MB, Seven M, Price S, Ferreira BM, Vianna-Morgante AM, Ellard S,Parrish A, Stals K, Flores-Daboub J, Jhangiani SN, Gibbs RA; Baylor-HopkinsCenter for Mendelian Genomics; Brunner HG, Sutton VR, Lupski JR, Carvalho CMB.WNT Signaling Perturbations Underlie the Genetic Heterogeneity of RobinowSyndrome. Am J Hum Genet. 2018 Jan 4;102(1):27-43. doi:10.1016/j.ajhg.

2017.10.002. Epub 2017 Dec 21. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/29276006)

- White JJ, Mazzeu JF, Hoischen A, Bayram Y, Withers M, Gezdirici A, Kimonis V, Steehouwer M, Jhangiani SN, Muzny DM, Gibbs RA; Baylor-Hopkins Center forMendelian Genomics; van Bon BWM, Sutton VR, Lupski JR, Brunner HG, Carvalho CMB.DVL3 Alleles Resulting in a -1 Frameshift of the Last Exon MediateAutosomal-Dominant Robinow Syndrome. Am J Hum Genet. 2016 Mar 3;98(3):553-561.doi: 10.1016/j.ajhg.2016.01.005. Epub 2016 Feb 25. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/26924530) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4800044/)

Genomic Location

The *DVL3* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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