

## FZD2 gene

frizzled class receptor 2

### Normal Function

The *FZD2* gene provides instructions for making a protein that plays a critical role in development before birth. The FZD2 protein interacts with other proteins (including those produced from the DVL genes, *DVL1*, *DVL2*, and *DVL3*) in chemical signaling pathways called Wnt signaling. These pathways control the activity of genes needed at specific times during development, and they regulate the interactions between cells when organs and tissues are forming. As a key part of Wnt signaling, the FZD2 protein is thought to be important for the normal development of the skeleton and potentially other parts of the body.

### Health Conditions Related to Genetic Changes

#### Robinow syndrome

At least four mutations in the *FZD2* gene have been found to cause autosomal dominant 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. *FZD2* gene mutations cause a form of the condition that can have a wide variety of features. These include short stature, a particular pattern of facial characteristics, shortening of the long bones in the arms and legs, and short fingers and toes (brachydactyly). This combination of features has been described as autosomal omodysplasia, but researchers now believe that it actually represents a subtype of autosomal dominant Robinow syndrome.

Three of the known mutations change a single protein building block (amino acid) at position 434 in the FZD2 protein. Another mutation leads to the production of an abnormally short version of the FZD2 protein. These mutations are thought to alter or remove a region of the protein that is needed for its interaction with DVL proteins, which 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

- frizzled 2, seven transmembrane spanning receptor

- frizzled family receptor 2
- frizzled homolog 2
- frizzled-2 precursor
- fz-2
- Fz2
- fzE2
- hFz2

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of FZD2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2535\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2535[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FZD2%5BTIAB%5D%29+OR+%28frizzled+2%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

- FRIZZLED CLASS RECEPTOR 2; FZD2 (<https://omim.org/entry/600667>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2535>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=FZD2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=FZD2[gene]))

## References

- 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-Hopkins Center for Mendelian Genomics; Brunner HG, Sutton VR, Lupski JR, Carvalho CMB. WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. *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>)

- Zhao Z, Lee CC, Baldini A, Caskey CT. A human homologue of the *Drosophila* polarity gene *frizzled* has been identified and mapped to 17q21.1. *Genomics*. 1995 May 20;27(2):370-3. doi: 10.1006/geno.1995.1060. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/7558010>)

## **Genomic Location**

The *FZD2* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

**Last updated February 1, 2018**