

## **WNT5A gene**

Wnt family member 5A

### **Normal Function**

The *WNT5A* gene is part of a large family of WNT genes, which play critical roles in development starting before birth. These genes provide instructions for making proteins that participate in chemical signaling pathways in the body. Wnt signaling controls the activity of certain genes and regulates the interactions between cells during embryonic development.

The protein produced from the *WNT5A* gene is part of chemical signaling pathways that control the movement of cells (cell migration) and attachment of cells to one another (cell adhesion) during early development. Studies suggest that the *WNT5A* protein plays important roles in the normal development of many parts of the body, including the brain, skeleton, blood cells, and fatty (adipose) tissue.

### **Health Conditions Related to Genetic Changes**

#### Robinow syndrome

At least six mutations in the *WNT5A* 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 bones. Autosomal dominant inheritance means that one copy of the altered gene in each cell is sufficient to cause the disorder. Most of the known mutations change a single protein building block (amino acid) in the *WNT5A* protein. These mutations alter the structure of the protein, which likely affects its interactions with other proteins involved in Wnt signaling. The resulting impairment of Wnt signaling pathways disrupts the development of many organs and tissues, leading to the features of Robinow syndrome.

### **Other Names for This Gene**

- hWNT5A
- wingless-type MMTV integration site family member 5A
- wingless-type MMTV integration site family, member 5A
- WNT-5A protein

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of WNT5A ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7474\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7474[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28WNT5A%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 5A; WNT5A (<https://omim.org/entry/164975>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7474>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=WNT5A\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=WNT5A[gene]))

## **References**

- Kikuchi A, Yamamoto H, Sato A, Matsumoto S. Wnt5a: its signalling, functions and implication in diseases. *Acta Physiol (Oxf)*. 2012 Jan;204(1):17-33. doi:10.1111/j.1748-1716.2011.02294.x. Epub 2011 Apr 22. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21518267>)
- Mikels AJ, Nusse R. Purified Wnt5a protein activates or inhibits beta-catenin-TCF signaling depending on receptor context. *PLoS Biol*. 2006 Apr;4(4):e115. doi: 10.1371/journal.pbio.0040115. Epub 2006 Apr 4. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16602827>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1420652/>)
- Nishita M, Enomoto M, Yamagata K, Minami Y. Cell/tissue-tropic functions of Wnt5a signaling in normal and cancer cells. *Trends Cell Biol*. 2010 Jun;20(6):346-54. doi: 10.1016/j.tcb.2010.03.001. Epub 2010 Mar 30. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20359892>)
- Person AD, Beiraghi S, Sieben CM, Hermanson S, Neumann AN, Robu ME, Schleiffarth JR, Billington CJ Jr, van Bokhoven H, Hoogeboom JM, Mazzeu JF, Petryk A, Schimmenti LA, Brunner HG, Ekker SC, Lohr JL. WNT5A mutations in patients with autosomal dominant Robinow syndrome. *Dev Dyn*. 2010 Jan;239(1):327-37. doi: 10.1002/dvdy.22156. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19918918>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4059519/>)

- Roifman M, Marcelis CL, Paton T, Marshall C, Silver R, Lohr JL, Yntema HG, Venselaar H, Kayserili H, van Bon B, Seaward G; FORGE Canada Consortium; BrunnerHG, Chitayat D. De novo WNT5A-associated autosomal dominant Robinow syndromesuggests specificity of genotype and phenotype. *Clin Genet.* 2015;87(1):34-41.doi: 10.1111/cge.12401. Epub 2014 May 24. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24716670/>)
- 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/>)

## Genomic Location

The *WNT5A* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

**Last updated February 1, 2018**