WNT3 gene

Wnt family member 3

Normal Function

The *WNT3* gene is part of a large family of WNT genes, which play critical roles in development before birth. WNT genes provide instructions for making proteins that participate in chemical signaling pathways in the body. These pathways control the activity of certain genes and regulate the interactions between cells during embryonic development.

Research in animals indicates that the protein produced from the *WNT3* gene is critical for outgrowth of the limbs in the developing embryo. The WNT3 protein also appears to play an important role in determining the anterior-posterior axis (the imaginary line that runs from head to tail in animals) during the earliest stages of embryonic development. Additionally, the effects of mutations in the human *WNT3* gene suggest that the protein may be involved in the normal formation of the facial features, head, heart, lungs, nervous system, skeleton, and genitalia.

Health Conditions Related to Genetic Changes

Tetra-amelia syndrome

A mutation in the *WNT3* gene has been shown to cause tetra-amelia syndrome. This condition is very rare and characterized by the absence of all four limbs. The *WNT3* gene variant causes a form of this condition known as tetra-amelia syndrome type 1. Because children with this condition have such serious medical problems, most are stillborn or die shortly after birth.

The *WNT3* gene variant that causes tetra-amelia syndrome type 1, which occurs in both copies of the *WNT3* gene in each cell, replaces one protein building block (amino acid) with a premature stop signal in the instructions for making the WNT3 protein. This variant is written as Gln83Ter or Q83X.

Researchers believe that the Gln83Ter variant results in the production of an abnormally short, nonfunctional version of the WNT3 protein. Loss of WNT3 protein activity disrupts normal limb formation before birth and leads to the other serious birth defects associated with tetra-amelia syndrome type 1.

Other Names for This Gene

- INT4
- Oncogene INT4
- Proto-oncogene protein Wnt-3
- wingless-type MMTV integration site family member 3
- wingless-type MMTV integration site family, member 3
- WNT-3 proto-oncogene protein
- WNT3_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of WNT3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7473[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=(WNT3%5BTIAB%5D)+AND+((Genes%5BMH%5D)+OR+(Genetic+Phenomena%5BMH%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 3; WNT3 (https://omim.org/entry/165330)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7473)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=WNT3[gene])

References

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Genomic Location

The *WNT3* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

Last updated February 17, 2023