WNT10A gene

Wnt family member 10A

Normal Function

The *WNT10A* 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 *WNT10A* gene plays a role in the development of many parts of the body. It appears to be essential for the formation of tissues that arise from an embryonic cell layer called the ectoderm. These tissues include the skin, hair, nails, teeth, and sweat glands. Researchers believe that the WNT10A protein is particularly important for the formation and shaping of both baby (primary) teeth and adult (permanent) teeth.

Health Conditions Related to Genetic Changes

Hypohidrotic ectodermal dysplasia

Several mutations in the *WNT10A* gene have been found to cause hypohidrotic ectodermal dysplasia, the most common form of ectodermal dysplasia. Starting before birth, ectodermal dysplasias result in the abnormal development of the skin, hair, nails, teeth, and sweat glands. Hypohidrotic ectodermal dysplasia is characterized by a reduced ability to sweat (hypohidrosis), sparse scalp and body hair (hypotrichosis), and several missing teeth (hypodontia) or teeth that are malformed. *WNT10A* gene mutations account for about 5 percent of all cases of hypohidrotic ectodermal dysplasia.

Most of the *WNT10A* gene mutations associated with hypohidrotic ectodermal dysplasia change single protein building blocks (amino acids) in the WNT10A protein, which impairs its function. The resulting shortage of functional WNT10A protein disrupts Wnt signaling during the development of ectodermal tissues, particularly the teeth.

Hypohidrotic ectodermal dysplasia can result from mutations in several genes. When the condition is caused by *WNT10A* gene mutations, its features are more variable than when the condition is caused by mutations in any other gene. Signs and symptoms range from mild to severe, and mutations in this gene are more likely to cause all of the

permanent teeth to be missing.

Keratoconus

MedlinePlus Genetics provides information about Keratoconus

Other disorders

Mutations in the *WNT10A* gene have been reported to cause several other, rare forms of ectodermal dysplasia, including odonto-onycho-dermal dysplasia (OODD) and Schopf-Schulz-Passarge syndrome (SSPS). OODD is characterized by dry hair, missing teeth, a smooth tongue, fingernail and toenail abnormalities, thickened skin on the palms of the hands and soles of the feet (palmoplantar keratoderma), and increased sweating (hyperhidrosis) of the palms and soles. The major features of SSPS include missing teeth, hypotrichosis, palmoplantar keratoderma, and fluid-filled sacs (cysts) on the edges of the eyelids.

WNT10A gene mutations are also frequently associated with a condition called nonsyndromic tooth agenesis. This condition causes one or more teeth not to form. Although missing teeth is a common feature of ectodermal dysplasias, "nonsyndromic" suggests that in these cases tooth agenesis occurs without the other signs and symptoms of those conditions.

More than 70 *WNT10A* gene mutations have been identified in people with various forms of ectodermal dysplasia or nonsyndromic tooth agenesis. Most of these mutations change single amino acids in the WNT10A protein, which impairs its function. The resulting shortage of functional WNT10A protein disrupts Wnt signaling during the development of ectodermal tissues, including the skin, hair, nails, teeth, and sweat glands. Researchers are working to determine why mutations in this gene can cause several different disorders.

Other Names for This Gene

- OODD
- protein Wnt-10a precursor
- STHAG4
- wingless-type MMTV integration site family, member 10A

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of WNT10A (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80326[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28WNT10A%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- TOOTH AGENESIS, SELECTIVE, 4; STHAG4 (https://omim.org/entry/150400)
- SCHOPF-SCHULZ-PASSARGE SYNDROME; SSPS (https://omim.org/entry/224750)
- ODONTOONYCHODERMAL DYSPLASIA; OODD (https://omim.org/entry/257980)
- WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 10A; WNT10A (https://omim.org/entry/606268)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/80326)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=WNT10A[gene])

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Genomic Location

The *WNT10A* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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