FOXN1 gene

forkhead box N1

Normal Function

The *FOXN1* gene provides instructions for making a protein that attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this action, the FOXN1 protein is called a transcription factor.

The FOXN1 protein is important for development of the skin, hair, nails, and immune system. Studies suggest that this protein helps guide the formation of hair follicles and the growth of fingernails and toenails. The FOXN1 protein also plays a critical role in the formation of the thymus, which is a gland located behind the breastbone where immune system cells called T cells mature and become functional. T cells recognize and attack foreign invaders, such as viruses and bacteria, to help prevent infection.

Researchers suspect that the FOXN1 protein is also involved in the development of the brain and spinal cord (central nervous system), although its role is unclear.

Health Conditions Related to Genetic Changes

T-cell immunodeficiency, congenital alopecia, and nail dystrophy

At least one mutation in the *FOXN1* gene has been found to cause T-cell immunodeficiency, congenital alopecia, and nail dystrophy, a condition that affects the immune system and growth of the hair and nails. The known mutation, which is written as Arg255Ter or R255X, replaces the protein building block (amino acid) arginine with a signal to stop protein production. No functional FOXN1 protein is produced from the mutated gene.

A lack of FOXN1 protein prevents the formation of the thymus. When this gland is not present, the immune system cannot produce mature, functional T cells to fight infections. As a result, people with T-cell immunodeficiency, congenital alopecia, and nail dystrophy develop recurrent serious infections starting early in life. Loss of the FOXN1 protein also prevents the formation of hair follicles, leading to an absence of hair (alopecia). In addition, a shortage of this protein causes malformations of the fingernails and toenails (nail dystrophy).

Researchers have described abnormalities of the central nervous system in at least two cases of T-cell immunodeficiency, congenital alopecia, and nail dystrophy. However, it

is not yet known whether central nervous system abnormalities are a common feature of this condition. It is unclear how a shortage of the FOXN1 protein might contribute to these abnormalities.

Other Names for This Gene

- FKHL20
- forkhead box protein N1
- RONU
- Rowett nude
- WHN
- winged helix nude
- winged-helix nude
- winged-helix transcription factor nude

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FOXN1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8456[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FOXN1%5BTIAB%5D%29+OR+%28forkhead+box+N1%5BTIAB%5D%29%29+OR+%28winged+helix+nude%5BTIAB%5D%29+OR+%28%28WHN%5BTIAB%5D%29+AND+%28winged+helix%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

- FORKHEAD BOX N1; FOXN1 (https://omim.org/entry/600838)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/8456)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=FOXN1[gene])

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Genomic Location

The *FOXN1* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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