HOXA13 gene

homeobox A13

Normal Function

The *HOXA13* gene provides instructions for producing a type of protein called a transcription factor. These proteins attach (bind) to specific regions of DNA and helps control the activity of other genes. The *HOXA13* gene is part of a larger family genes called homeobox genes, which provide instructions for making transcription factors that act during early embryonic development to control the formation of many body structures. Specifically, the HOXA13 protein appears to be critical for the formation and development of the limbs (particularly the hands and feet), urinary tract, and reproductive system.

The HOXA13 protein contains three areas where a protein building block (amino acid) called alanine is repeated multiple times. These stretches of alanines are known as polyalanine tracts or poly(A) tracts. The role these polyalanine tracts play in the normal function of this protein is unknown.

Health Conditions Related to Genetic Changes

Hand-foot-genital syndrome

Variants (also called mutations) in the *HOXA13* gene have been found to cause hand-foot-genital syndrome. As its name suggests, this condition affects the development of the hands and feet, the urinary tract, and the reproductive system. More than half of the variants affect one of the polyalanine tracts in the HOXA13 protein. These variants add extra alanines to these tracts, making them abnormally long and unstable. The resulting altered protein is degraded by the cell, so it cannot regulate the activity of other genes during early development.

Other *HOXA13* gene variants result in the production of an abnormally short, nonfunctional version of the HOXA13 protein or change single amino acids in the protein. Variants that substitute one amino acid for another amino acid may change the way the HOXA13 protein folds. The altered protein may or may not function or bind to DNA normally. Variants that result in an altered but functional HOXA13 protein may cause more severe signs and symptoms of hand-foot-genital syndrome than variants that lead to a nonfunctional version of this protein.

Cancers

Chromosomal rearrangements (translocations) involving the short (p) arm of chromosome 7 have been associated with leukemia, a cancer of blood-forming cells. These translocations disrupt the region of chromosome 7 that contains several similar homeobox genes, including *HOXA13*.

Within cancer cells, researchers have found translocations between chromosome 7 and chromosome 11 in several people with leukemia. These rearrangements abnormally fuse part of the *HOXA13* gene or a similar gene on chromosome 7 to part of the *NUP98* gene on chromosome 11. The protein produced from the fused gene probably signals abnormal cells to continue dividing without control or order, which likely contributes to the development of cancer.

Other Names for This Gene

- homeo box 1J
- homeo box A13
- Homeobox protein Hox-A13
- homeobox protein HOXA13
- Hox-1J
- HOX1
- HOX1J
- HXA13_HUMAN
- transcription factor HOXA13

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of HOXA13 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3209[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28HOXA13%5BTIAB%5D%29+OR+%28HOX1J%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%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

- HOMEOBOX A13; HOXA13 (https://omim.org/entry/142959)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3209)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=HOXA13[gene])

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Genomic Location

The *HOXA13* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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