HOXA13 Gene

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Homeobox A13

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1. Introduction

The *HOXA13* gene provides instructions for producing a protein that attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this role, the *HOXA13* gene is called a transcription factor. The *HOXA13* gene is part of a larger family of transcription factors called homeobox genes, which 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* gene 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 of polyalanine tracts in normal HOXA13 function is unknown.

2. Health Conditions Related to Genetic Changes

2.1. Hand-Foot-Genital Syndrome

At least 14 mutations in the *HOXA13* gene have been found to cause hand-foot-genital syndrome. More than half of these mutations affect one of the polyalanine tracts in the *HOXA13* gene. These mutations add extra alanines to these tracts, making them abnormally long and unstable. The resulting altered protein is degraded by the cell, so it is unavailable to regulate the activity of other genes during early development. These changes affect the development of the hands, feet, urinary tract, and reproductive system.

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

2.2. Cancers

Chromosomal rearrangements (translocations) involving the short (p) arm of chromosome 7 have been associated with rare cases of 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 *HOXA13* 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.

3. 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

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