MEOX1 gene

mesenchyme homeobox 1

Normal Function

The *MEOX1* gene provides instructions for making a protein called homeobox protein MOX-1, which is a member of the homeobox protein family. Homeobox proteins direct the formation of body structures during early embryonic development. Homeobox protein MOX-1 regulates the process that begins separating vertebrae from one another, a process called somite segmentation. The protein functions as a transcription factor, which means it attaches to DNA and controls the activity (expression) of other genes. Homeobox protein MOX-1 likely controls the expression of genes that regulate somite segmentation. Homeobox protein MOX-1 also ensures that the developing vertebral bone is maintained in its correct position for proper formation. Additionally, the homeobox protein MOX-1 plays a role in the formation of the joints that connect the base of the skull and the top of spine (cranio-cervical joints).

Health Conditions Related to Genetic Changes

Klippel-Feil syndrome

At least three mutations in the *MEOX1* gene have been found to cause Klippel-Feil syndrome. This condition is characterized by the abnormal joining (fusion) of two or more spinal bones in the neck (cervical vertebrae) and a variety of other features affecting many parts of the body. The *MEOX1* gene mutations result in a lack of functional homeobox protein MOX-1. While the effect of the loss of this protein on vertebral development is unclear, it is likely that absence of this protein leads to unregulated somite segmentation and incorrect vertebral positioning. As a result, the cervical vertebrae do not separate during development but instead are fused together. It is unclear why this condition affects the cervical vertebrae more severely than other bones.

Other Names for This Gene

- homeobox protein MOX-1
- MEOX1_HUMAN
- MOX1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MEOX1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4222[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MEOX1%5BTIAB%5D%29+OR+%28mesenchyme+homeobox+1%5BTIAB%5D%29+OR+%28MOX1%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)

Catalog of Genes and Diseases from OMIM

- MESENCHYME HOMEOBOX 1; MEOX1 (https://omim.org/entry/600147)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4222)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MEOX1[gene])

References

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- Douville JM, Cheung DY, Herbert KL, Moffatt T, Wigle JT. Mechanisms of MEOX1and MEOX2 regulation of the cyclin dependent kinase inhibitors p21 and p16 invascular endothelial cells. PLoS One. 2011;6(12):e29099. doi:10.1371/journal.pone.0029099. Epub 2011 Dec 20. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22206000) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3243699/)
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Genomic Location

The *MEOX1* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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