FOXC2 gene

forkhead box C2

Normal Function

The *FOXC2* gene provides instructions for making a protein that plays a critical role in the formation of many organs and tissues before birth. This protein is a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of many other genes. Researchers believe that the FOXC2 protein has a role in a variety of developmental processes, such as the formation of veins and the development of the lungs, eyes, kidneys and urinary tract, cardiovascular system, and the transport system for immune cells (lymphatic vessels).

Health Conditions Related to Genetic Changes

Lymphedema-distichiasis syndrome

More than 50 mutations in the *FOXC2* gene can cause lymphedema-distichiasis syndrome. Most of these mutations insert or delete a few DNA building blocks (nucleotides), which results in a premature stop signal in the instructions for making the FOXC2 protein. These mutations lead to the production of a FOXC2 protein that is abnormally small and cannot effectively attach (bind) to DNA. As a result, the altered protein cannot regulate the activity of other genes. Other mutations change one protein building block (amino acid) in the area of the FOXC2 protein that binds to DNA, preventing the protein from regulating gene activity. It is not clear why mutations in the *FOXC2* gene affect the development of the eye area and lymphatic vessels, the primary regions of the body affected by lymphedema-distichiasis syndrome.

Other Names for This Gene

- FKHL14
- forkhead (Drosophila)-like 14
- forkhead, Drosophila, homolog-like 14
- FOXC2_HUMAN
- LD
- MFH-1
- MFH-1,mesenchyme forkhead 1

- MFH1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FOXC2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2303[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FOXC2%5BTIAB%5D%29+OR+%28forkhead+box+C2%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- FORKHEAD BOX C2; FOXC2 (https://omim.org/entry/602402)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2303)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=FOXC2[gene])

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Genomic Location

The *FOXC2* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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