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

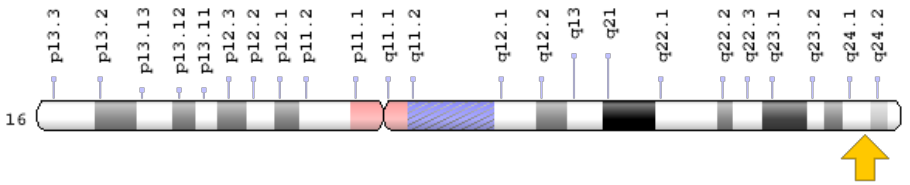
Lymphedema-distichiasis syndrome

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.

Chromosomal Location

Cytogenetic Location: 16q24.1, which is the long (q) arm of chromosome 16 at position 24.1

Molecular Location: base pairs 86,566,829 to 86,569,728 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Clinical Information from GeneReviews

- Lymphedema-Distichiasis Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1457

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/602402

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_FOXC2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FOXC2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3801
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2303
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2303
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