FOXF1 gene

forkhead box F1

Normal Function

The *FOXF1* gene provides instructions for making the forkhead box F1 (FOXF1) protein. 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. The FOXF1 protein is important in the development of pulmonary mesenchyme, the embryonic tissue from which blood vessels of the lung arise. It is also involved in the development of the gastrointestinal tract.

Health Conditions Related to Genetic Changes

Alveolar capillary dysplasia with misalignment of pulmonary veins

At least four mutations in the *FOXF1* gene have been identified in infants with alveolar capillary dysplasia with misalignment of pulmonary veins (ACD/MPV). Some mutations change single protein building blocks (amino acids) used to make the FOXF1 protein. Other mutations insert or delete genetic material in the *FOXF1* gene. These mutations result in an inactive protein that cannot regulate development, leading to abnormal formation of the pulmonary blood vessels. Affected infants with *FOXF1* gene mutations usually also have gastrointestinal abnormalities.

Other Names for This Gene

- ACDMPV
- FKHL5
- Forkhead, drosophila, homolog-like 5
- forkhead-related activator 1
- FOXF1_HUMAN
- FREAC1
- MGC105125

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FOXF1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2294[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FOXF1%5BTIAB%5D%29+OR+%28forkhead+box+F1%5BTIAB%5D%29%29+OR+%28FKHL5%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- FORKHEAD BOX F1; FOXF1 (https://omim.org/entry/601089)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2294)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=FOXF1[gene])

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Genomic Location

The *FOXF1* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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