



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

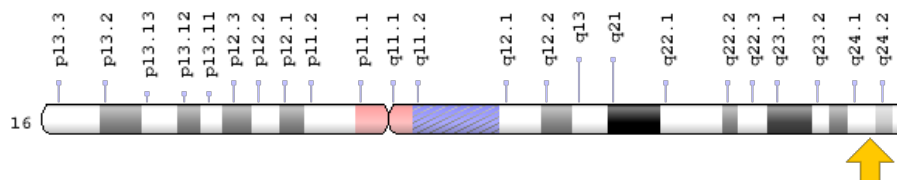
Alveolar capillary dysplasia with misalignment of pulmonary veins

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.

Chromosomal Location

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

Molecular Location: base pairs 86,510,527 to 86,515,422 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

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

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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
<http://omim.org/entry/601089>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/FOXF1ID40628ch16q24.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=FOXF1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3809
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:2294>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2294>
- UniProt
<https://www.uniprot.org/uniprot/Q12946>

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