Genetics Home Reference

Your Guide to Understanding Genetic Conditions

FGD1 gene

FYVE, RhoGEF and PH domain containing 1

Normal Function

The *FGD1* gene provides instructions for making a protein known as a guanine nucleotide exchange factor (GEF). GEFs turn on (activate) proteins called GTPases, which play an important role in chemical signaling within cells. GTPases are turned off (inactivated) when they are attached (bound) to a molecule called GDP and are activated when they are bound to another molecule called GTP. The FGD1 protein activates the GTPase known as Cdc42 by stimulating the exchange of GDP for GTP. Once Cdc42 is active, it transmits signals that are critical for various aspects of embryonic development, particularly formation of the skeleton.

Health Conditions Related to Genetic Changes

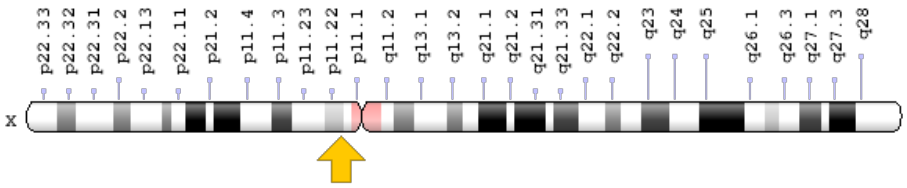
Aarskog-Scott syndrome

Approximately 20 mutations in the *FGD1* gene have been found to cause Aarskog-Scott syndrome. These mutations lead to the production of an abnormally functioning protein, which disrupts Cdc42 signaling. Altering the transmission of Cdc42 signals results in the wide variety of developmental abnormalities seen in Aarskog-Scott syndrome.

Chromosomal Location

Cytogenetic Location: Xp11.22, which is the short (p) arm of the X chromosome at position 11.22

Molecular Location: base pairs 54,445,454 to 54,496,166 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AAS
- faciogenital dysplasia protein
- FGD1_HUMAN
- FGDY
- ZFYVE3

Additional Information & Resources

Educational Resources

- Eurekah Bioscience Collection: The Rho GDP/GTP Cycle Is Regulated by GEFs and GAPs
  https://www.ncbi.nlm.nih.gov/books/NBK6594/#A39190
- Molecular Biology of the Cell (fourth edition, 2002): Regulatory Proteins Control the Activity of GTP-Binding Proteins by Determining Whether GTP or GDP Is Bound
  https://www.ncbi.nlm.nih.gov/books/NBK26911/#A515

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28FGD1%5BTIAB%5D%29+OR+%28%28faciogenital+dysplasia+protein%5BTIAB%5D%29+OR+%28FGDY%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28%28glucocorticoid%29+OR+%28FGD4%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- FYVE, RhoGEF, AND PH DOMAIN-CONTAINING PROTEIN 1
  http://omim.org/entry/300546

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_FGD1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FGD1%5Bgene%5D
- HGNC Gene Family: Pleckstrin homology domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/682
- HGNC Gene Family: Rho guanine nucleotide exchange factors
  http://www.genenames.org/cgi-bin/genefamilies/set/722

- HGNC Gene Family: Zinc fingers FYVE-type
  http://www.genenames.org/cgi-bin/genefamilies/set/81
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3663
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2245
- UniProt
  http://www.uniprot.org/uniprot/P98174

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11181572
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  http://omim.org/entry/300546
- Gorski JL, Estrada L, Hu C, Liu Z. Skeletal-specific expression of Fgd1 during bone formation and skeletal defects in faciogenital dysplasia (FGDY; Aarskog syndrome). Dev Dyn. 2000 Aug;218(4):573-86.
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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14560308

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