FGD1 gene

FYVE, RhoGEF and PH domain containing 1

Normal Function

The *FGD1* gene provides instructions for making a protein that functions 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 turned on (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 development before and after birth, particularly the development of bones. The FGD1 protein may also be involved in maintenance (remodeling) of the extracellular matrix, which is the intricate lattice of proteins and other molecules that forms in the spaces between cells. Through this process, the protein appears to play a role in cell movement (migration) and the remodeling of blood vessels.

Health Conditions Related to Genetic Changes

Aarskog-Scott syndrome

More than 40 variants (also known as mutations) in the *FGD1* gene have been found to cause Aarskog-Scott syndrome, a rare condition that occurs primarily in males. Affected boys typically have distinctive facial features, genital abnormalities, childhood short stature, and other skeletal abnormalities. The *FGD1* gene variants lead to the production of an abnormally functioning FGD1 protein, which disrupts Cdc42 signaling. Altering the transmission of Cdc42 signals likely impairs normal development of bones and other tissues, resulting in the wide variety of abnormalities that occur in people with Aarskog-Scott syndrome.

Other Names for This Gene

- AAS
- faciogenital dysplasia protein
- FGD1_HUMAN
- FGDY

- ZFYVE3

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FGD1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2245[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28FGD1%5BTIAB%5D%29+OR+%28%28faciogenital+dysplasia+protein%5BTIAB%5D%29+OR+%28FGDY%5BTIAB%5D%29%29+NOT+%28%28glucocorticoid%29+OR+%28FGD4%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- FYVE, RhoGEF, AND PH DOMAIN-CONTAINING PROTEIN 1; FGD1 (https://omim.org/entry/300546)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2245)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=FGD1[gene])

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Genomic Location

The *FGD1* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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