

SFRP4 gene

secreted frizzled related protein 4

Normal Function

The *SFRP4* gene provides instructions for making a protein called secreted frizzled-related protein 4 (SFRP4). This protein blocks (inhibits) a process called Wnt signaling. Wnt signaling plays an important role in the development of several tissues and organs throughout the body. In particular, regulation of this signaling process by SFRP4 is critical for normal bone development and remodeling. Bone remodeling is a normal process in which old bone is broken down and new bone is created to replace it. The SFRP4 protein also plays a role in the development of fatty (adipose) tissue.

Health Conditions Related to Genetic Changes

Pyle disease

At least four *SFRP4* gene mutations have been found in individuals with a bone disorder called Pyle disease. This condition is characterized by a bone abnormality in which the ends (metaphyses) of the long bones in the arms and legs are abnormally wide, resembling a boat oar or paddle. Other bones may also be abnormal in Pyle disease, including the collar bones (clavicles), ribs, and bones in the fingers and hands. The *SFRP4* gene mutations are thought to lead to production of an abnormally short SFRP4 protein with impaired function, or they result in no SFRP4 protein production at all. Studies suggest that loss of functional SFRP4 dysregulates Wnt signaling, which disrupts normal bone development and remodeling. Abnormal bone formation leads to the characteristics of Pyle disease.

Dupuytren contracture

MedlinePlus Genetics provides information about Dupuytren contracture

Other Names for This Gene

- frizzled protein, human endometrium
- FRP-4
- FRPHE
- PYL

- secreted frizzled-related protein 4 precursor
- secreted frizzled-related protein 4; secreted frizzled-related protein 4
- SFRP-4

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SFRP4 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6424\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6424[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SFRP4%5BTIAB%5D%29+OR+%28secreted+frizzled+related+protein+4%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+1440+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- SECRETED FRIZZLED-RELATED PROTEIN 4; SFRP4 (<https://omim.org/entry/606570>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6424>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SFRP4\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SFRP4[gene]))

References

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Genomic Location

The *SFRP4* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

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