CCN6 gene

cellular communication network factor 6

Normal Function

The *CCN6* gene provides instructions for making a protein that appears to be involved in bone growth and the maintenance of cartilage, which covers and protects the ends of bones. The function of the CCN6 protein is not well understood. It is part of a family of proteins that are involved in the growth and maintenance of connective tissues, such as bone, cartilage, and blood vessels. The CCN6 protein is made in cells called chondrocytes, which produce and maintain cartilage, and is associated with the production of certain proteins that make up cartilage, but its role in their production is unclear. CCN6 may also help control signaling pathways involved in the development of cartilage and bone and may help regulate the breakdown of cartilage components.

Health Conditions Related to Genetic Changes

Progressive pseudorheumatoid dysplasia

Mutations in the *CCN6* gene cause progressive pseudorheumatoid dysplasia (PPRD), which is a condition that causes stiffness and pain in the joints of the hands, hips, knees, and spine. The joint problems worsen over time, and movement in the joints becomes limited. Most of the mutations involved in this condition lead to production of an abnormally short CCN6 protein that is probably nonfunctional. Other mutations change single protein building blocks (amino acids) in the protein. Loss of CCN6 protein function likely disrupts normal cartilage maintenance and bone growth, leading to the joint problems in PPRD.

Juvenile idiopathic arthritis

MedlinePlus Genetics provides information about Juvenile idiopathic arthritis

Other Names for This Gene

- CCN family member 6
- LIBC
- PPAC
- PPD

- WISP-3
- WISP3
- WISP3_HUMAN
- WNT1 inducible signaling pathway protein 3
- WNT1-inducible-signaling pathway protein 3

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CCN6 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8838[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28WISP3%5BTIAB%5D%29+OR+%28WNT1+inducible+signaling+pathway+protein+3%5BTIAB%5D%29+OR+%28CCN6%29%29+OR+%28%28WNT1-inducible-signaling+pathway+protein+3%5BTIAB%5D%29+OR+%28WISP-3%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CELLULAR COMMUNICATION NETWORK FACTOR 6; CCN6 (https://omim.org/entry/603400)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/8838)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CCN6[gene])

References

- Baker N, Sharpe P, Culley K, Otero M, Bevan D, Newham P, Barker W, ClementsKM, Langham CJ, Goldring MB, Gavrilovic J. Dual regulation of metalloproteinaseexpression in chondrocytes by Wnt-1-inducible signaling pathway protein 3/CCN6.Arthritis Rheum. 2012 Jul;64(7):2289-99. doi: 10.1002/art.34411. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22294415) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3366172/)
- Davis L, Chen Y, Sen M. WISP-3 functions as a ligand and promotes superoxidedismutase activity. Biochem Biophys Res Commun. 2006 Mar 31;342(1): 259-65. doi:10.1016/j.bbrc.2006.01.132. Epub 2006 Feb 3. Citation on PubMed (http

s://pubmed.ncbi.nlm.nih.gov/16480948)

- Nakamura Y, Weidinger G, Liang JO, Aquilina-Beck A, Tamai K, Moon RT, WarmanML. The CCN family member Wisp3, mutant in progressive pseudorheumatoiddysplasia, modulates BMP and Wnt signaling. J Clin Invest. 2007Oct;117(10):3075-86. doi: 10.1172/JCI32001. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17823661) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1964511/)
- Sen M, Cheng YH, Goldring MB, Lotz MK, Carson DA. WISP3-dependent regulationof type II collagen and aggrecan production in chondrocytes. Arthritis Rheum.2004 Feb;50(2):488-97. doi: 10.1002/art.20005. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14872491)

Genomic Location

The *CCN6* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

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