

YY1AP1 gene

YY1 associated protein 1

Normal Function

The *YY1AP1* gene provides instructions for making part of a group of associated proteins known as the INO80 chromatin remodeling complex. In the cell nucleus, this complex attaches (binds) to chromatin, which is the network of DNA and proteins that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling by the INO80 chromatin remodeling complex allows cells to control the activity (expression) of certain genes. This complex also helps regulate several other critical cell functions, including repair of damaged DNA, cell specialization (differentiation), and cell growth and division (proliferation). Activity of the INO80 chromatin remodeling complex appears to be particularly important in smooth muscle cells, which line the walls of blood vessels.

Health Conditions Related to Genetic Changes

Grange syndrome

At least five mutations in the *YY1AP1* gene have been identified in people with Grange syndrome. This rare condition causes narrowing (stenosis) and blockage (occlusion) of blood vessels that supply blood to many organs and tissues, including the kidneys, brain, and heart. Grange syndrome can also be associated with short fingers and toes (brachydactyly), fusion of some of the fingers or toes (syndactyly), fragile bones that are prone to breakage, learning disabilities, and heart defects.

Mutations in the *YY1AP1* gene are described as "loss-of-function" because they prevent the production of any functional YY1AP1 protein. A loss of this protein disrupts the function of the INO80 chromatin remodeling complex, which alters the activity of multiple genes in smooth muscle cells. Researchers suspect that these changes in gene expression lead to reduced cell proliferation and differentiation. However, it is unclear how these changes cause narrowing and blockage of arteries. It is also unknown how *YY1AP1* gene mutations are related to other features of Grange syndrome, such as bone abnormalities and learning disabilities.

Other Names for This Gene

- GRNG

- HCCA1
- HCCA2
- hepatocellular carcinoma susceptibility protein
- hepatocellular carcinoma-associated protein 2
- YAP
- YY1AP

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of YY1AP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55249\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55249[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28YY1AP1%5BTIAB%5D%29+OR+%28YY1+associated+protein+1%5BTIAB%5D%29+OR+%28hepatocellular+carcinoma+susceptibility+protein%5BTIAB%5D%29+OR+%28YY1+associated+protein%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

Catalog of Genes and Diseases from OMIM

- YY1 ASSOCIATED PROTEIN 1; YY1AP1 (<https://omim.org/entry/607860>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/55249>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=YY1AP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=YY1AP1[gene]))

References

- Guo DC, Duan XY, Regalado ES, Mellor-Crummey L, Kwartler CS, Kim D, Lieberman K, de Vries BBA, Pfundt R, Schinzel A, Kotzot D, Shen X, Yang ML; University of Washington Center for Mendelian Genomics; Bamshad MJ, Nickerson DA, Gornik HL, Ganesh SK, Braverman AC, Grange DK, Milewicz DM. Loss-of-Function Mutations in YY1AP1 Lead to Grange Syndrome and a Fibromuscular Dysplasia-Like Vascular Disease. *Am J Hum Genet.* 2017 Jan 5;100(1):21-30. doi:10.1016/j.ajhg.2016.11.008. Epub 2016 Dec 8. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/27939641>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5223026/>)
- Morrison AJ, Shen X. Chromatin remodelling beyond transcription: the INO80 and SWR1 complexes. *Nat Rev Mol Cell Biol.* 2009 Jun;10(6):373-84. doi:10.1038/

nrm2693. Epub 2009 May 8. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19424290>)

- Wang ZX, Wang HY, Wu MC. Identification and characterization of a novel humanhepatocellular carcinoma-associated gene. Br J Cancer. 2001 Oct 19;85(8): 1162-7.doi: 10.1054/bjoc.2001.2059. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11710830>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2375167/>)

Genomic Location

The *YY1AP1* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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