

GALNS gene

galactosamine (N-acetyl)-6-sulfatase

Normal Function

The *GALNS* gene provides instructions for producing an enzyme called N-acetylgalactosamine 6-sulfatase. This enzyme is located in lysosomes, which are compartments within cells that break down and recycle different types of molecules. N-acetylgalactosamine 6-sulfatase is involved in the breakdown of large sugar molecules called glycosaminoglycans (GAGs) or mucopolysaccharides. Specifically, this enzyme removes a chemical group known as a sulfate from a GAG called keratan sulfate. Keratan sulfate is particularly abundant in cartilage and the clear covering of the eye (cornea).

Health Conditions Related to Genetic Changes

Mucopolysaccharidosis type IV

More than 148 mutations in the *GALNS* gene have been found to cause mucopolysaccharidosis type IV (MPS IV). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS IV reduce or eliminate the function of N-acetylgalactosamine 6-sulfatase.

The lack of N-acetylgalactosamine 6-sulfatase activity leads to the accumulation of keratan sulfate within lysosomes. Because keratan sulfate is predominantly found in cartilage and the cornea, the buildup of this substance causes skeletal abnormalities and cloudy corneas. Researchers believe that a buildup of GAGs may also cause the features of MPS IV by interfering with the functions of other proteins inside lysosomes and disrupting the movement of molecules inside the cell.

Other Names for This Gene

- chondroitinase
- chondroitinsulfatase
- FLJ17434
- FLJ42844
- FLJ98217
- galactosamine (N-acetyl)-6-sulfate sulfatase

- galactose-6-sulfate sulfatase
- GALNAC6S
- galNAc6S sulfatase
- GALNS_HUMAN
- GAS
- MPS4A
- N-acetylgalactosamine-6-sulfatase
- N-acetylgalactosamine-6-sulfatase precursor
- N-acetylgalactosamine-6-sulfate sulfatase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GALNS ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2588\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2588[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GALNS%5BTIAB%5D%29+OR+%28galactosamine+-6-sulfate+sulfatase%5BTIAB%5D%29%29+OR+%28%28MPS4A%5BTIAB%5D%29+OR+%28N-acetylgalactosamine-6-sulfatase%5BTIAB%5D%29+OR+%28chondroitinsulfatase%5BTIAB%5D%29+OR+%28galactose-6-sulfate+sulfatase%5BTIAB%5D%29+OR+%28N-acetylgalactosamine-6-sulfate+sulfatase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- GALACTOSAMINE-6-SULFATE SULFATASE; GALNS (<https://omim.org/entry/612222>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2588>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GALNS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GALNS[gene]))

References

- Montano AM, Sukegawa K, Kato Z, Carrozzo R, Di Natale P, Christensen E, Orii KO, Orii T, Kondo N, Tomatsu S. Effect of γ -attenuated γ -mutations

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- Tomatsu S, Montano AM, Lopez P, Trandafirescu G, Gutierrez MA, Oikawa H, Nishioka T, Vieira MB, Orii T, Noguchi A. Determinant factors of spectrum of missense variants in mucopolysaccharidosis IVA gene. *Mol Genet Metab*. 2006 Sep-Oct;89(1-2):139-49. doi: 10.1016/j.ymgme.2006.05.012. Epub 2006 Jul 11. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16837223>)
- Tomatsu S, Montano AM, Nishioka T, Gutierrez MA, Pena OM, Tranda Firescu GG, Lopez P, Yamaguchi S, Noguchi A, Orii T. Mutation and polymorphism spectrum of the GALNS gene in mucopolysaccharidosis IVA (Morquio A). *Hum Mutat*. 2005 Dec;26(6):500-12. doi: 10.1002/humu.20257. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16287098>)

Genomic Location

The *GALNS* gene is found on chromosome 16 (<https://medlineplus.gov/genetics/chromosome/16/>).

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