GUSB gene

glucuronidase beta

Normal Function

The *GUSB* gene provides instructions for producing an enzyme called beta-glucuronidase (β-glucuronidase). This enzyme is located in lysosomes, compartments within cells that digest and recycle different types of molecules. β-glucuronidase is involved in the breakdown of large molecules called glycosaminoglycans (GAGs). GAGs are composed of sugar molecules that are linked together to form a long string. To break down these large molecules, individual sugars are removed one at a time from one end of the molecule. β-glucuronidase is involved in the break down of three types of GAGs: dermatan sulfate, heparan sulfate, and chondroitin sulfate. This enzyme removes a sugar called glucuronic acid when it is at the end of the GAG chain.

Health Conditions Related to Genetic Changes

Mucopolysaccharidosis type VII

At least 55 mutations in the *GUSB* gene have been found to cause mucopolysaccharidosis type VII (MPS VII). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS VII reduce or eliminate the function of β-glucuronidase.

The shortage (deficiency) of β-glucuronidase leads to the accumulation of dermatan sulfate, heparan sulfate, and chondroitin sulfate within lysosomes in virtually all tissues and organs. The buildup of these GAGs increases the size of the lysosomes, which is why many tissues and organs are enlarged in MPS VII. Researchers believe that the accumulated GAGs may also interfere with the functions of other proteins inside the lysosomes and disrupt many normal functions of cells.

Other Names for This Gene

- beta-D-glucuronidase
- beta-G1
- beta-glucuronidase
- beta-glucuronidase precursor
- BG

- BGLR_HUMAN
- FLJ39445
- glucuronidase, beta
- MPS7

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GUSB (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2990[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GUSB%5BTI%5D%29+OR+%28beta+glucuronidase%5BTI%5D%29%29+OR+%28MPS+VII%5BTIAB%5D%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

- BETA-GLUCURONIDASE; GUSB (https://omim.org/entry/611499)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2990)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GUSB[gene])

References

- Storch S, Wittenstein B, Islam R, Ullrich K, Sly WS, Braulke T. Mutationalanalysis in longest known survivor of mucopolysaccharidosis type VII. Hum Genet.2003 Feb; 112(2):190-4. doi: 10.1007/s00439-002-0849-5. Epub 2002 Nov 5. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12522561)
- Tomatsu S, Montano AM, Dung VC, Grubb JH, Sly WS. Mutations and polymorphismsin GUSB gene in mucopolysaccharidosis VII (Sly Syndrome). Hum Mutat. 2009Apr;30(4):511-9. doi: 10.1002/humu.20828. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19224584) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3048808/)
- Vervoort R, Islam MR, Sly WS, Zabot MT, Kleijer WJ, Chabas A, Fensom A, YoungEP, Liebaers I, Lissens W. Molecular analysis of patients with beta-glucuronidasedeficiency presenting as hydrops fetalis or as early

mucopolysaccharidosis VII.Am J Hum Genet. 1996 Mar;58(3):457-71. Citation on PubMed (https:
d.ncbi.nlm.nih.gov/8644704) or Free article on PubMed Central (https://www.ncbi.nl
m.nih.gov/pmc/articles/PMC1914559/)

Genomic Location

The *GUSB* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromoso
me/7/).

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