IDS gene

iduronate 2-sulfatase

Normal Function

The *IDS* gene provides instructions for producing an enzyme called iduronate 2-sulfatase (I2S), which is essential for the breakdown of large sugar molecules called glycosaminoglycans (GAGs). Specifically, I2S removes a chemical group known as a sulfate from a molecule called sulfated alpha-L-iduronic acid, which is present in two GAGs called heparan sulfate and dermatan sulfate. I2S is located in lysosomes, which are compartments within cells that digest and recycle different types of molecules.

Health Conditions Related to Genetic Changes

Mucopolysaccharidosis type II

Hundreds of variants (also called mutations) in the *IDS* gene have been found to cause mucopolysaccharidosis type II (MPS II). Variants that change one DNA building block (nucleotide) are the most common. All the variants that cause MPS II reduce or completely eliminate the function of the I2S enzyme in cells..pf0{}

There are two types of MPS II: the neuropathic form, which is more severe, and the non-neuropathic form, which is less severe. It is difficult to tell which variants cause each form of the disorder. However, variants that remove large pieces of the gene or rearrange the genetic material, completely eliminating I2S enzyme function, often cause the neuropathic form of the disorder.

Lack of I2S enzyme activity leads to the accumulation of heparan sulfate and dermatan sulfate within lysosomes in cells. The buildup of these GAGs increases the size of the lysosomes, which is why many tissues and organs are enlarged in people with MPS II. Researchers believe that the accumulated GAGs may also interfere with the functions of other proteins inside the lysosomes and disrupt the movement of molecules inside the cell. In addition, buildup in lysosomes may trigger the release of molecules called cytokines that stimulate inflammation and may contribute to the progression of MPS II.

Other Names for This Gene

- IDS_HUMAN
- iduronate 2-sulfatase (Hunter syndrome)

- iduronate-2-sulfatase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of IDS (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3423[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28iduronate+2-sulfatase%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- MUCOPOLYSACCHARIDOSIS, TYPE II; MPS2 (https://omim.org/entry/309900)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3423)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=IDS[gene])

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Genomic Location

The *IDS* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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