

IDUA gene

alpha-L-iduronidase

Normal Function

The *IDUA* gene provides instructions for producing an enzyme called alpha-L-iduronidase, which is essential for the breakdown of large sugar molecules called glycosaminoglycans (GAGs). Through a process called hydrolysis, alpha-L-iduronidase uses water molecules to break down a molecule known as unsulfated alpha-L-iduronic acid, which is present in two GAGs called heparan sulfate and dermatan sulfate. Alpha-L-iduronidase is located in lysosomes, compartments within cells that digest and recycle different types of molecules.

Health Conditions Related to Genetic Changes

Mucopolysaccharidosis type I

More than 100 mutations in the *IDUA* gene have been found to cause mucopolysaccharidosis type I (MPS I). Mutations that change one DNA building block (nucleotide) are the most common. All mutations that cause MPS I reduce or completely eliminate the function of alpha-L-iduronidase. It usually cannot be determined whether a certain mutation will cause severe or attenuated MPS I; however, people who do not produce any alpha-L-iduronidase have the severe form of this disorder.

The lack of alpha-L-iduronidase enzyme activity leads to the accumulation of heparan sulfate and dermatan sulfate within the lysosomes. The buildup of these GAGs increases the size of the lysosomes, which is why many tissues and organs are enlarged in MPS I. 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.

Other Names for This Gene

- alpha-L-iduronidase
- IDUA_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of IDUA ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3425\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3425[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28IDUA%5BTIAB%5D%29+OR+%28alpha-L-iduronidase%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+1440+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- ALPHA-L-IDURONIDASE; IDUA (<https://omim.org/entry/252800>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3425>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=IDUA\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=IDUA[gene]))

References

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Genomic Location

The *IDUA* gene is found on chromosome 4 (<https://medlineplus.gov/genetics/chromosome/4/>).

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