Defective IDS does not hydrolyse dermatan sulfate (Chebi:63517 chain)

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Introduction

Reactome is open-source, open access, manually curated and peer-reviewed pathway database. Pathway annotations are authored by expert biologists, in collaboration with Reactome editorial staff and cross-referenced to many bioinformatics databases. A system of evidence tracking ensures that all assertions are backed up by the primary literature. Reactome is used by clinicians, geneticists, genomics researchers, and molecular biologists to interpret the results of high-throughput experimental studies, by bioinformaticians seeking to develop novel algorithms for mining knowledge from genomic studies, and by systems biologists building predictive models of normal and disease variant pathways.

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Literature references

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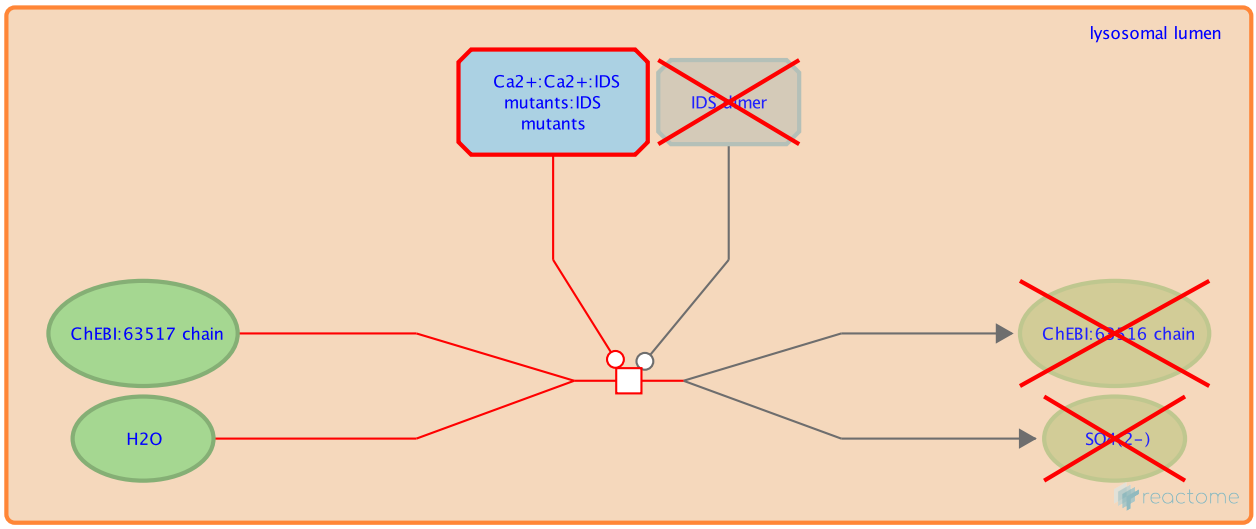
Stable identifier: R-HSA-2262743

Type: transition

Compartments: lysosomal lumen

Diseases: mucopolysaccharidosis II



Mucopolysaccharidosis II (MPS II, Hunter syndrome, MIM:309900) is an X-linked genetic disorder caused by defects in the gene encoding the enzyme iduronate 2-sulfatase (IDS, MIM:300823). This causes an accumulation of the GAGs dermatan sulfate and heparan sulfate and their excessive excretion in urine. MPS II has a broad range of severity with variable mental retardation and life expectancy. This disease has a prevelence of approximately 1 in 170,000 male births (Muenzer et al. 2009). The R468 codon may be a mutational hot-spot, as it has been noted in patients with diverse ethnic origins: R468W (Crotty et al. 1992), R468L and R468Q (Isogai et al. 1998). R443X is also a frequent mutation (Froissart et al. 1998).

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Editions

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