Defective GUSB does not hydrolyse (HA)2

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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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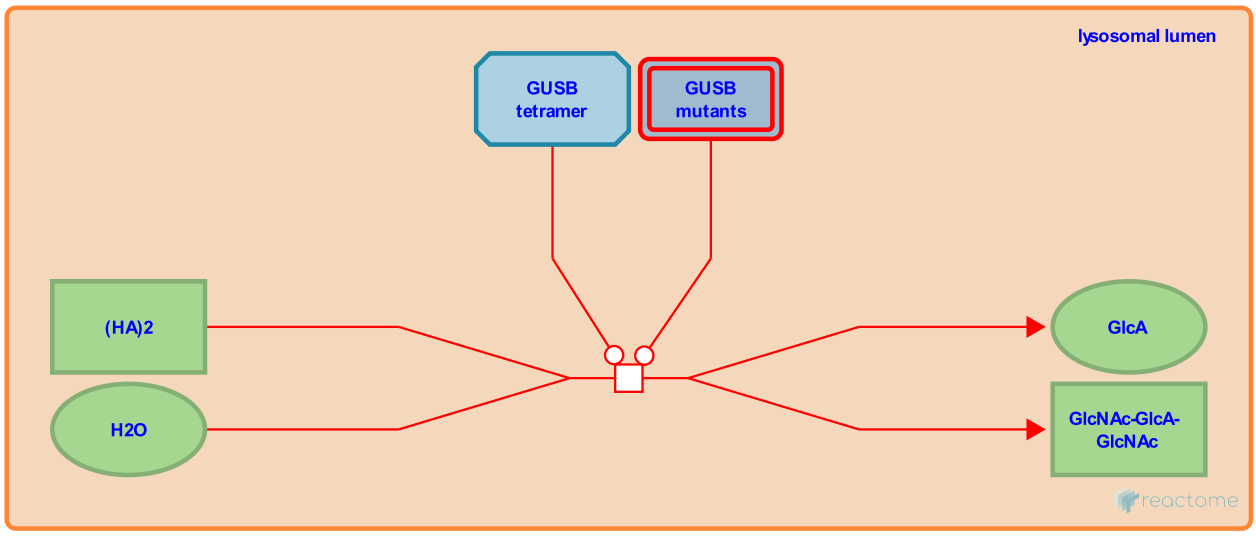
Stable identifier: R-HSA-2318373

Type: transition

Compartments: lysosomal lumen

Diseases: mucopolysaccharidosis VII



Tetrameric lysosomal enzyme beta-glucuronidase (GUSB tetramer) hydrolyses glucuronate from the HA tetrasaccharide (HA(2)) resulting in the single sugars glucuronic acid and N-acetylglucosamine. Defects in beta-glucuronidase (GUSB; MIM:611499) cause mucopolysaccharidosis type VII (MPS VII, Sly syndrome, beta-glucuronidase deficiency; MIM:253220), an autosomal recessive lysosomal storage disease. Mutations causing severe forms of the disease are R356* (Shipley et al. 1993), A354V and R611W (Wu & Sly 1993), S52F (Vervoot et al. 1997) and R216W (Vervoort et al. 1996).

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Editions

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