Defective HGSNAT does not acetylate Heparan chain(1)

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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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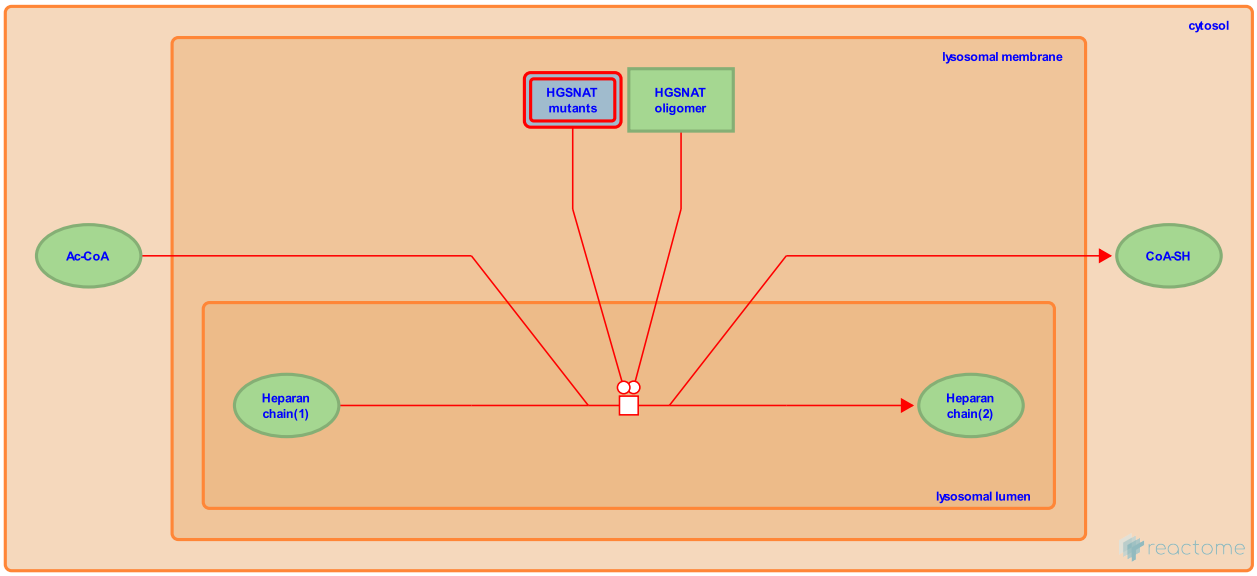
Stable identifier: R-HSA-2263492

Type: transition

Compartments: lysosomal lumen, cytosol, lysosomal membrane

Diseases: mucopolysaccharidosis III



Enzyme misfolding due to missense mutations results in incorrect glycosylation therefore HGSNAT is not targeted to the lysosome and stays in the ER (Feldhammer et al. 2009). This, together with mutations giving rise to nonsense-mediated mRNA decay (Fedele & Hopwood 2010), appear to be the major molecular mechanisms underlying MPSIIIC. More than 50 mutations are known in the HGSNAT gene. Some of them drastically reduce enzyme activity; W403C/A615T double mutant (Fedele & Hopwood 2010), R344C, S518F and R384X (Fedele et al. 2007, Ruijter et al.2008).

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Editions

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