Defective SGSH does not hydrolyse Heparan sulfate chain(2)

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30/04/2024

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.

The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

Literature references

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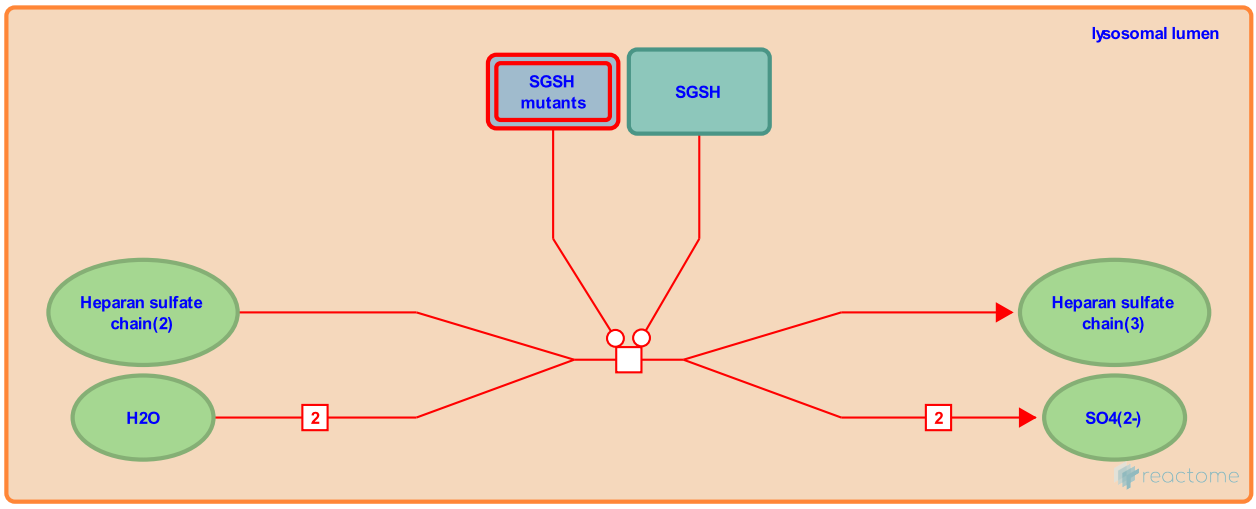
Stable identifier: R-HSA-9036050

Type: transition

Compartments: lysosomal lumen

Diseases: mucopolysaccharidosis III



MPS IIIA (Sanfilippo syndrome A, mucopolysaccharidosis IIIA, MIM:252900) is a rare, autosomal recessive lysosomal storage disease. A deficiency of the enzyme N-sulphoglucosamine sulphohydrolase (SGSH, MIM:605270), which normally hydrolyses the sulfate group from the terminal N-sulphoglucosamine residue of heparan sulfate (HS) leads to the build up of HS in cells and tissues, characterised by severe CNS degeneration in early childhood leading to death between 10 and 20 years of age.
Four mutations (R74C, R245H, S66W, and 1091delC) are known to be prevalent in Polish (Bunge et al. 1997), Dutch (Weber et al. 1997), Italian (Di Natale et al. 1998), and Spanish (Montfort et al. 1998) populations, respectively. These mutations abolish the activity of SGSH being associated with the classic severe phenotype.

Literature references

Zaremba, J., Hopwood, JJ., Weber, B., Ince, H., Van Diggelen, OP., Steglich, C. et al. (1997). Identification of 16 sulfamidase gene mutations including the common R74C in patients with mucopolysaccharidosis type IIIA (Sanfilippo A). *Hum Mutat, 10*, 479-85. ↗

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Editions

2012-05-21	Authored, Edited	Jassal, B.
2012-08-27	Reviewed	Coutinho, MF., Matos, L., Alves, S.