Defective HYAL1 does not hydrolyse Chondroitin chains

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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

Fabregat, A., Sidiropoulos, K., Viteri, G., Forner, O., Marin-Garcia, P., Arnau, V. et al. (2017). Reactome pathway analysis: a high-performance in-memory approach. *BMC bioinformatics, 18*, 142. ↗

Sidiropoulos, K., Viteri, G., Sevilla, C., Jupe, S., Webber, M., Orlic-Milacic, M. et al. (2017). Reactome enhanced pathway visualization. *Bioinformatics, 33*, 3461-3467. ↗

Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

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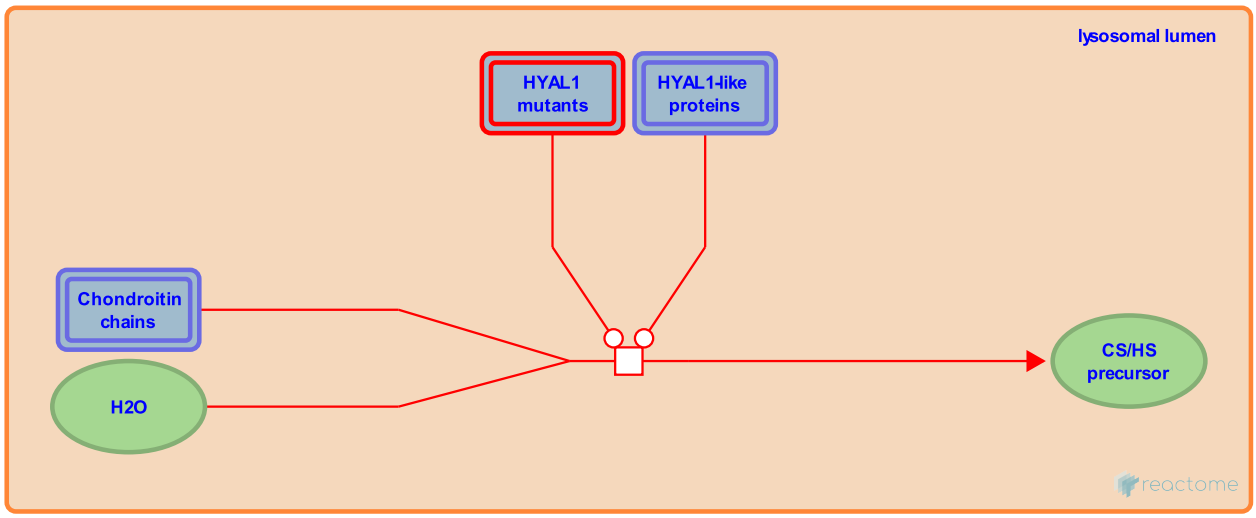
Stable identifier: R-HSA-2318585

Type: transition

Compartments: lysosomal lumen

Diseases: mucopolysaccharidosis



Hyaluronidase 1 (HYAL1) hydrolyses 1-4 linkages between GalNAc and D-glucuronate residues in chondroitin (or dermatan). It also hydrolyses this linkage in hyaluronate, another glycosaminoglycan (GAG) composed of repeating disaccharide units but the only one which is non-sulfated. Defects in HYAL1 (MIM:607071) cause mucopolysaccharidosis type IX (MPS IX, Natowicz syndrome, Hyaluronidase deficiency, MIM:601492), a rare lysosomal storage disease. Triggs-Raine et al. identified a patient with two mutations in HYAL1 alleles, a nonconservative amino acid substitution (Glu268Lys) and a complex intragenic rearrangement (1361del37ins14) that results in a premature termination codon (Triggs-Raine et al. 1999).

Literature references

Salo, TJ., Natowicz, MR., Triggs-Raine, B., Wicklow, BA., Zhang, H. (1999). Mutations in HYAL1, a member of a tandemly distributed multigene family encoding disparate hyaluronidase activities, cause a newly described lysosomal disorder, mucopolysaccharidosis IX. *Proc. Natl. Acad. Sci. U.S.A., 96*, 6296-300.

Editions

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