

# Defective GLB1 does not hydrolyse linker chain(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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Reactome database release: 88

This document contains 1 reaction ([see Table of Contents](#))

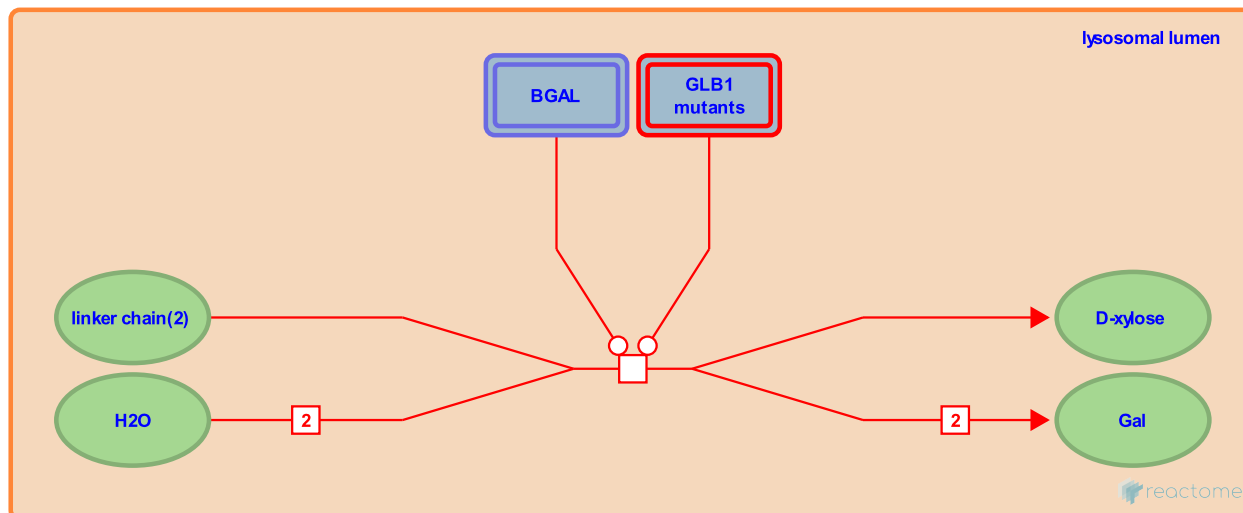
## Defective GLB1 does not hydrolyse linker chain(2) [↗](#)

**Stable identifier:** R-HSA-9036061

**Type:** transition

**Compartments:** lysosomal lumen

**Diseases:** mucopolysaccharidosis



Defects in beta-galactosidase (GLB1, MIM:611458) result in galactose moieties not being hydrolysed from keratan sulfate (KS) or the GAG linker chain, a tetrasaccharide sequence required for some GAG biosyntheses to take place. Mucopolysaccharidosis IV B (MPSIVB, Morquio's syndrome B; MIM:253010) is the result of GLB1 deficiency. GLB1 mutations causing severe phenotypes are R482C (Ishii et al. 1995), W509C (Oshima et al. 1991), Y83C (Santamaria et al. 2006) and W273L Paschke et al. 2001. Mild phenotypes where a partial loss of enzyme activity occurs can involve the mutants G438E, N484K, T500A (Bagshaw et al. 2002) and Y83H (Ishii et al. 1995). These mild phenotype mutants are not detailed here.

### Literature references

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### Editions

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