

Glycogen Storage

Acid alpha-glucosidase or acid maltase deficiency

Pompe disease

Glycosaminoglycans (mucopolysaccharides) Storage

- α-L-iduronidase deficiency → mucopolyssacharidosis I
- Iduronate sulfatase deficiency → mucopolyssacharidosis II
- Heparan sulfamidase deficiency → mucopolyssacharidosis IIIA
- N-acetylglucosaminidase deficiency → mucopolyssacharidosis IIIB
 - Heparan-α-glucosaminide Nacetyltransferase deficiency →
- mucopolyssacharidosis IIIC
 N-acetylglucosamine 6-sulfatase
- Galactose-6-sulfate sulfatase deficiency->mucopolyssacharidosis IVA

deficiency→mucopolyssacharidosis IIID

- β-galactosidase deficiency → mucopolyssacharidosis IVB
- N-acetylgalactosamine-4-sulfatase deficiency →mucopolyssacharidosis VI
 - β-glucoronidase deficiency → mucopolyssacharidosis VII
 - Hyaluronidase deficiency -> mucopolyssacharidosis IX

Lysosomal Storage Disease

Ilkka Mononen, Nathan N. Aronson

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