

Lipid Storage

including sphingomyelin and cholesterol accumulation



SMPD1 gene mutation cause acid sphingomyelinase activity deficiency



Niemann-Pick disease type A and B

Glycolipid Storage

Glucocerebrosidase deficiency



Gaucher's disease

Glycosphingolipid Storage

α-galactosidase A deficiency



Fabry diseases

Mutations of NPC1 and NPC2 genes cause impairment in cellular processing and transport of LDL-cholesterol



Niemann-Pick disease type C

Glycogen Storage

Acid alpha-glucosidase or acid maltase deficiency

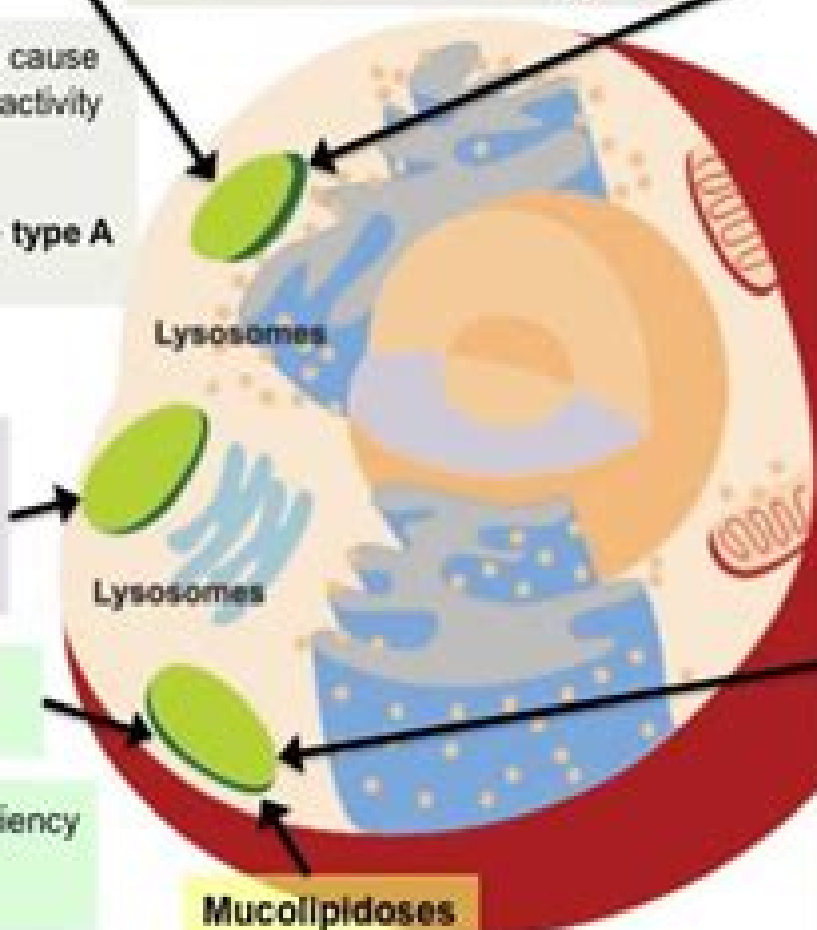


Pompe disease

Glycosaminoglycans (mucopolysaccharides) Storage

- α-L-iduronidase deficiency → mucopolysaccharidosis I
- Iduronate sulfatase deficiency → mucopolysaccharidosis II
- Heparan sulfamidase deficiency → mucopolysaccharidosis IIIA
- N-acetylglucosaminidase deficiency → mucopolysaccharidosis IIIB
- Heparan-α-glucosaminide N-acetyltransferase deficiency → mucopolysaccharidosis IIIC
- N-acetylglucosamine 6-sulfatase deficiency → mucopolysaccharidosis IIID
- Galactose-6-sulfate sulfatase deficiency → mucopolysaccharidosis IVA
- β-galactosidase deficiency → mucopolysaccharidosis IVB
- N-acetylgalactosamine-4-sulfatase deficiency → mucopolysaccharidosis VI
- β-glucuronidase deficiency → mucopolysaccharidosis VII
- Hyaluronidase deficiency → mucopolysaccharidosis IX

- α-N-acetyl neuraminidase (sialidase) → mucopolipidosis type I
- N-acetylglucosamine-1-phosphotransferase deficiency → mucopolipidosis type II and type III
- MCONLN1 gene mutation encoding mucopolipin1 → mucopolipidosis type IV



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