POST-SAN DIEGO 2023
Novità dal Meeting della Società Americana di Ematologia

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Verona
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Malattia di Gaucher

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Disclosures of Adriano Venditti

Company name	Research support	Employee	Consultant	Stockholder	Speakers bureau	Advisory board	Other
Abbvie			X			X	
Astellas			X			X	
Servier			X			X	
Menarini			X			X	
BMS			X				X
Pfizer			X		X	X	
Medac			X				X
Janssen			X			X	X
AstraZeneca			X			X	
Otzuka			X			X	
Beigene			X			X	
GSK			X			X	
Jazz Pharmaceuticals			X			X	X
Novartis			X			X	
Delbert Laboratoires						X	

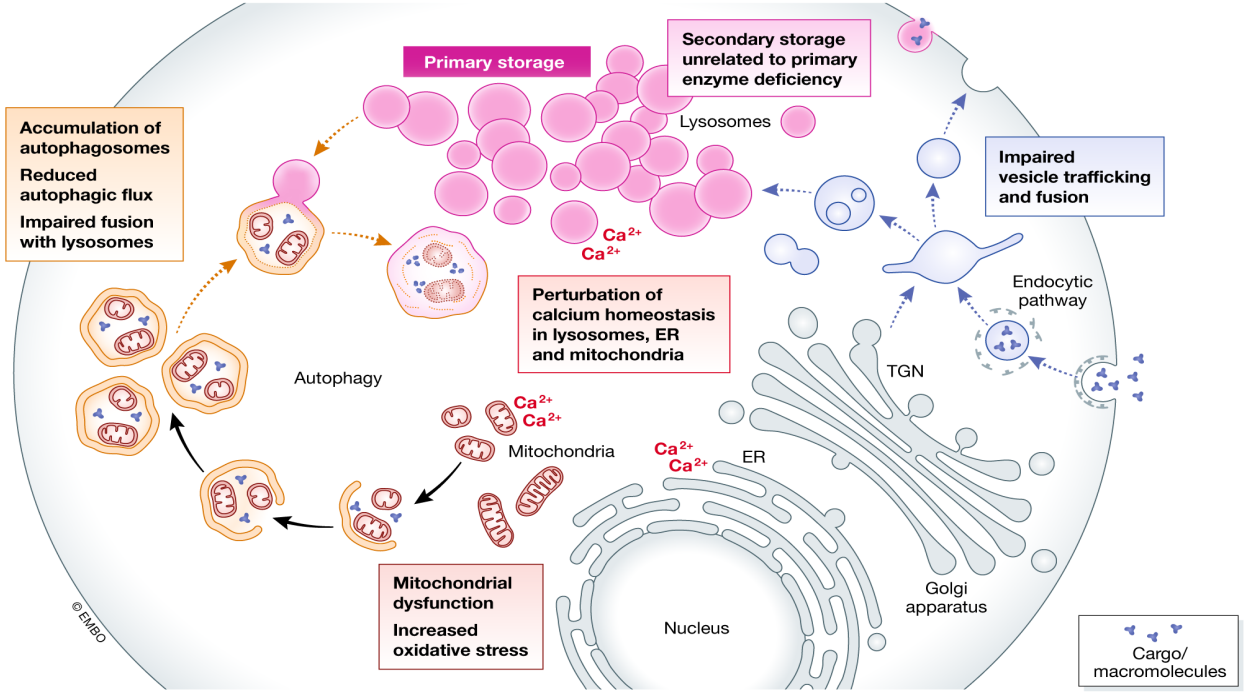
- Lysosomal storage disorders (LSDs) are inherited metabolic diseases due to deficiency of several components of lysosomal function
- LSDs are characterized by the accumulation of substrates in excess in several organs such as brain, spleen, liver, heart, bones, and muscles[1]
- Most LSDs are autosomal recessive disorders
  - Tree exceptions that are X-linked : *Fabry Disease, Hunter Syndrome and Danon Disease*
- Incidence of LSDs as a group was calculated to be 1 in 5000-8000 births

1. Demczko M. Merck Manual Professional. 2018. [Epub]..

The family of lysosomal storage disorders

- Glycogen storage disease type II
- Mucopolysaccharidoses
- Mucolipidoses
- Oligosaccharidoses
- Lipidoses
- **Sphingolipidoses (Gaucher Disease)**
- Lysosomal transport diseases

The view of lysosomal storage disorders



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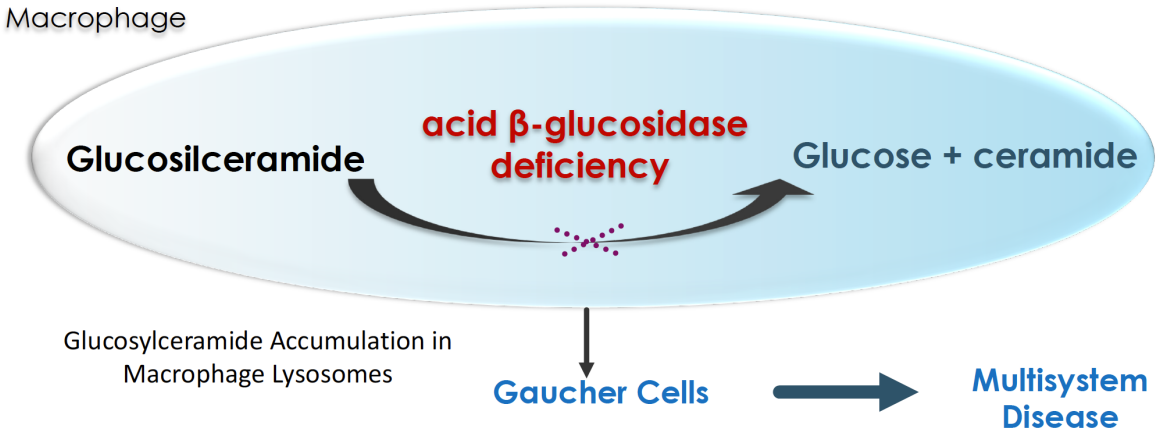
Gaucher Disease is the most frequent lysosomal storage disorder

The pathogenesis of Gaucher disease depends on mutations in the GBA1 gene that lead to deficiency of acid β-glucosidase enzyme activity[1–4]

This enzyme deficiency causes the accumulation of glucosylceramide within macrophage lysosomes → Gaucher cells



1. Stirnemann *et al. Int J Mol Sci* 2017;18:441 2. Deegan PB & Cox TM. *Drug Des Devel Ther* 2012;6:81 3. Cerezyme® (imiglucerase) summary of product characteristics. Genzyme Europe B.V. (September 2017) 4. Nagral A. *J Clin Exp Hepatol* 2014;4:37

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