**GAUCHER DISEASE**


**POMPE DISEASE**

Electrical changes in resting, exercise, and Holter electrocardiography in Fabry cardiomyopathy. In: http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4656035/

**NPS I DISEASE**

ABSTRACT: Sphingolipids represent a major class of lipids that are essential for several cellular processes. Sphingolipids are involved in the development and progression of several human diseases, including a series of lysosomal storage diseases. The most prevalent is Gaucher disease, an autosomal recessive disorder due to a defect in lysosomal glucocerebrosidase catalysis. In Gaucher disease, glucocerebroside and its deacylated product glucosylsphingosine accumulate in macrophages of the reticuloendothelial system and are responsible for the clinical manifestations of the disease. In addition to the well-known clinical manifestations, Gaucher disease is characterized by a set of hematological complications, such as anemia, thrombocytopenia, and platelet dysfunction. The mechanism underlying these hematological complications is not fully understood, but it is known that glucocerebroside and its deacylated product glucosylsphingosine are able to disrupt platelet physiology and function.

**FABRY DISEASE**

The latest American Academy of Pediatrics guideline on Fabry disease includes recommendations for diagnosis, treatment and management.