Pediatric Neurology Part III: Chapter 175. Gaucher disease (Handbook of Clinical Neurology)

Cyril Mignot, Antoinette Gelot, Thierry Billette De Villemeur

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Gaucher disease is an autosomal recessive condition due to glucocerebrosidase deficiency responsible for the lysosomal accumulation of glucosylceramide, a complex lipid derived from cell membranes, mainly in macrophages. It is due to mutations mostly in the GBA gene, although saposine C deficiency is due to mutations in the PSAP gene. It encompasses an extremely heterogeneous spectrum of clinical involvement from the fetus to adulthood. Splenomegaly, blood cytopenia, and bone involvement are the main manifestations of Gaucher disease, but nervous system degeneration is observed in about 5–10% of patients. The accumulation in neurons of glucosylceramide and its derivative, psychosine, are thought to underlie neuronal dysfunction and death, although Gaucher cells that mostly accumulate such substances are mainly macrophages. Enzyme replacement therapy dramatically improves the outcome of patients because of its extreme efficacy in the treatment of the systemic involvement. However, it has only limited effects on most neurological signs.

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