Pediatric Neurology Part III: Chapter 190. Enzyme replacement therapy and substrate reduction therapy in lysosomal storage disorders with neurological expression (Handbook of Clinical Neurology)

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Lysosomal storage diseases (LSD) are inborn errors of metabolism secondary to lysosomal enzyme defects and are characterized by a progressive accumulation of nondigested macromolecules provoking cellular dysfunction and clinical manifestations. The diagnosis of these diseases can be confirmed easily in most cases by immuno-enzymatic techniques and molecular biology. Even though these enzymatic deficits result in an accumulation of pathological substrates, the underlying mechanisms responsible for the pathogenesis of the disease are not entirely known. Nevertheless, the distribution of the accumulated material determines the affected organs. More particularly in the central nervous system (CNS), neurons are often involved due to the accumulation of storage material and their incapacity of renewal. LSD can be responsible for mental retardation or for a neurodegenerative course in the central nervous system. The peripheral nervous system and the muscle can also be severely impaired. Hematopoietic stem cell transplantation was the first therapy, demonstrating efficacy especially on the neurological involvement of various LSD. Enzyme replacement therapy is now available for Gaucher disease, Fabry disease, mucopolysaccharidoses type I, type II, and type VI, and Pompe disease. Inhibition of the synthesis of the accumulated substrate by small molecules which also have the capacity to diffuse through the blood–brain barrier is another treatment option. New therapeutic strategies using the properties of molecular chaperones and of read-through molecules for nonsense mutations have been studied in vitro and hopefully will soon find clinical applications while intrathecal enzymes are currently studies in clinical trials for MPSII, MPS IIIA and MLD.

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