Niemann-Pick syndrome

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Niemann-Pick syndrome is a hereditary autosomal recessive disorder associated with lysosomal storage dysfunction. It is characterized by a deficiency of one or more lysosomal enzymes or components involved in the hydrolysis or transport of lipids and their derivatives. In most cases, there is a deficiency of acid sphingomyelinase, an enzyme that catalyzes the hydrolysis of sphingomyelin into ceramide and phosphocholine. As a result, sphingomyelin and its precursor lipids accumulate within lysosomes. In some variants, abnormal cholesterol accumulation is observed due to impaired function of cholesterol transporter proteins.

Niemann-Pick disease is classified as a rare disorder, with a global incidence estimated at approximately 0.5 cases per 100,000 live births. In Georgia, no official statistics have yet been published by healthcare providers or public health authorities regarding the prevalence of lysosomal storage diseases, including Niemann-Pick disease. The condition is more frequently observed among individuals of Ashkenazi Jewish descent.

Genetic screening for Niemann-Pick disease is available through certain laboratories, such as Synevo; however, testing is performed abroad, primarily in European facilities, indicating a likely low number of diagnosed cases in Georgia.

Due to its rarity, research into Niemann-Pick disease is challenging. It typically manifests in early childhood, which further complicates clinical and experimental studies. Nonetheless, mouse models of the disease have been developed to facilitate investigation. Studies have demonstrated the disease's multisystemic impact, varying in severity depending on the subtype and underlying pathophysiological mechanisms. Modern medicine continues to search for effective treatments. Nevertheless, advancements in genetic research and enzyme replacement therapies provide promising future prospects.

The objective of this paper is to raise awareness about Niemann-Pick disease among the public. It will cover the clinical manifestations, subtypes, genetic and biochemical basis, differential diagnoses, and current hypotheses regarding the disease.