Niemann-Pick Disease

Niemann-Pick disease is a lysosomal storage disease caused by a defect in the enzyme sphingomyelinase commonly seen in the Eastern European Ashkenazi Jewish population. Defect of this enzyme causes accumulation of sphingomyelin in various organs including the retina, the central nervous system, and liver and spleen. On histology, Niemann-Pick disease is characterized by foam cells, which are numerous small vacuoles filled with lipids. The most common type of Niemann-Pick disease has an extremely poor prognosis with the majority of cases being diagnosed by 18 months of age.



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Pathophysiology

Autosomal Recessive
Recessive-chocolate
Niemann-Pick is inherited in an autosomal recessive modality. This means two copies of the abnormal gene must be present in order for the disease to develop.

Sphingomyelinase Deficiency
Broken Sphinx-on-my-leg
Sphingomyelinase, is deficient in this disorder, leading to an accumulation of sphingomyelin. This build up of sphingomyelin is attributed to the physiological symptoms seen in Niemann-Pick.

More Common in Ashkenazi Jewish Population
Yarmulke (Yamaka)
There is an increased disease prevalence in the Eastern European Ashkenazi Jewish population.

Signs and Symptoms

Cherry Red Spot on Macula
Cherry-eyes
On fundoscopic examination, a small red spot is seen in the center of the macula of the retina. It is caused by accumulation of sphingomyelin in the retina and the relative transparency of the macula. Cherry red macula can be seen in several different lipid storage diseases.

Hepatosplenomegaly
Liver-and-spleen-balloons
An accumulation of sphingomyelin in the liver and spleen causes enlargement of these organs at an early age.

Neurodegeneration
Degenerating-nerve-arm
Neurodegeneration involves progressive deterioration of the central nervous system due to accumulation of sphingomyelin.

Diagnosis

Foam Cells
Foam
Foam cells are phagocytes that have accumulated large amounts of oxidized low-density lipoproteins. Numerous small vacuoles filled with lipids are created causing the cytoplasm to appear foamy.

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