Niemann Pick Disease: Type C1 (NPC1)

Niemann-Pick Disease Type C1 is caused by mutations in the NPC1 gene. This gene is involved in producing a protein involved in the transportation of lipids, including cholesterol. Mutations in the NPC2 gene cause similar effects (Type C2). Symptoms of Niemann Pick C1 typically develop in early childhood, but can first appear in adults. Muscle weakness and ataxia (poor control of movements, etc.) become gradually apparent, along with liver disease, seizures (in some cases), and progressive mental deterioration. Patients are often unable to move their eyes vertically (vertical supranuclear gaze palsy). Swallowing and speech deteriorate progressively, leading to a complete inability to ingest food. Respiratory failure may also occur. Most patients die in their 20's or 30's. When the disease first appears in adulthood, psychiatric symptoms tend to predominate.

The total incidence of Niemann-Pick disease Type C is around 1 in 120,000 births. About 90% are due to the NPC1 gene. The disease is more common in some ethnic groups, such as Hispanics whose ancestors lived in the upper Rio Grande valley. The defective genes are inherited in an autosomal recessive manner, typically requiring both parents to be asymptomatic carriers.

Sources

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