NIEMANN-PICK DISEASE
TYPES A & B

WHAT IS NIEMANN-PICK DISEASE?

Niemann-Pick disease is classified as a lysosomal storage disorder (LSD). Lysosomes are cell organelles that digest and recycle various molecules. Disorders that result in the accumulation of molecules within the lysosomes, including Niemann-Pick disease, are identified as LSDs.

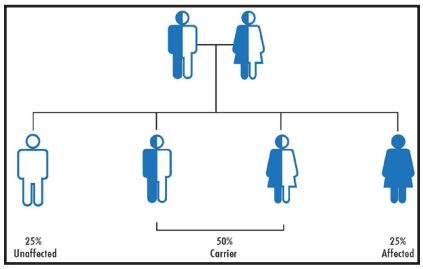
Niemann-Pick disease is divided into three types: type A, type B, and type C. These types are organized according to the genetic cause and symptoms.

THE CAUSE

Niemann-Pick disease types A and B are caused by mutations in the SMPD1 gene. The purpose of this gene is to code for the production of an enzyme called acid sphingomyelinase. This enzyme is found in the lysosomes and is responsible for converting a lipid called sphingomyelin into another lipid called ceramide. Mutations in the gene result in reduced quantity or complete absence of the enzyme, causing lipid accumulation in the cells and eventual loss of function in tissues and organs. Organs most affected include the brain, lungs, spleen, and liver.

INHERITANCE PATTERN

Niemann-Pick disease is inherited as an autosomal recessive (AR) condition. In order to be affected, the individual would have had to inherit one mutation in the gene from each parent. Each parent is a carrier and does not show symptoms of the condition. When two carrier parents of an AR condition have children, there is a 25% chance of having an affected child.



Niemann-Pick disease types A and B affects 1 in 250,000 infants. There is a higher incidence of the disease among individuals of Ashkenazi Jewish descent (1 in 40,000).

PATIENT CARE

DIAGNOSIS

1. Eye exam for detection of cherry-red spot
2. Enzymatic testing to measure acid sphingomyelinase activity levels in white blood cells
3. DNA mutation analysis of the SMPD1 gene

MANAGEMENT and TREATMENT

There is no specific treatment for Niemann-Pick disease types A and B. Supportive care should be provided to manage symptoms and includes pulmonology, genetics, cardiology, physical therapy, and a developmental specialist.

For Niemann-Pick type B, bone marrow transplantation has been performed in a few individuals. Researchers are also exploring enzyme replacement as a treatment option.

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WHAT ARE THE SYMPTOMS?

Niemann-Pick type A

Symptoms appear in infancy. Babies develop hepatosplenomegaly, fail to gain weight and grow, and have a progressive loss of movement and neurodevelopmental growth. Children with this disease generally do not survive beyond early childhood.

- Hepatosplenomegaly
- Failure to thrive
- Interstitial lung disease with recurrent respiratory infections
- Cherry-red spot on the macula
- Eye movement difficulties

Niemann-Pick type B

Symptoms present in mid-childhood to pre-teen years. Individuals with this disorder usually survive into adulthood.

- Hepatosplenomegaly
- Frequent lung infections
- Thrombocytopenia
- Short stature
- 30% will have the cherry-red spot on the macula
- Delayed bone age and puberty

RESOURCES

National Niemann-Pick Disease Foundation
npdf.org

National Institute of Neurological Disorders and Stroke
ninds.nih.gov/Disorders

Genetics Home Reference
ghr.nlm.nih.gov/condition/niemann-pick-disease

Genetic and Rare Diseases
rarediseases.info.nih.gov

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