Niemann-Pick Disease

Niemann-Pick disease (NP) is an inherited metabolic disorder. With it, harmful amounts of a fatty substance build up in the spleen, liver, lungs, bone marrow, and, in some people, the brain. The disease is subdivided into 4 related types.

- In **TYPES A AND B,** there is not enough activity of an enzyme called sphingomyelinase. This causes the build up of toxic amounts of sphingomyelin. That is a fatty substance present in every cell of the body.
- **TYPE A, INFANTILE NP** is the most common type. It happens in infants and it causes jaundice, enlargement of the liver and profound brain damage. Children with this type rarely live beyond 18 months.
- **TYPE B, JUVENILE NON-NEURONOPATHIC NP,** involves enlargement of the liver and spleen. It often occurs in the pre-teen years; the brain is not affected.
- **TYPES C** and **D** cause a defect that disrupts the transport of cholesterol between brain cells. They may appear early in life, or they may be delayed into the teen years. Children with these types have only moderate enlargement of the spleen and liver. But brain damage may be extensive. It may cause:
  - Inability to look up and down.
  - Difficulty in walking and swallowing.
  - Progressive loss of vision and hearing.

TYPE D only occurs in people with an ancestral background in Nova Scotia.

TREATMENT

There is currently no effective treatment for patients with type A. Bone marrow transplantation has been attempted in a few patients with type B. Encouraging results have been reported. The development of enzyme replacement and gene therapies might also be helpful for those with type B. Children and young teens with types C and D are often placed on a low cholesterol diet; however, its clinical benefit is not convincing.

WHAT WILL HAPPEN

Patients with type A die in infancy. Type B patients may live a comparatively long time but many require supplemental oxygen due to lung impairment. The life expectancies of patients with types C and D vary; some patients die in childhood; others, who appear to be less drastically affected, live into adulthood.

RESEARCH BEING DONE

Investigators at NINDS have identified two different genes that, when defective, contribute to Niemann-Pick disease, types C and D. Research supported by the NINDS also includes studies to understand how genetic defects can cause neurological disorders. These studies open promising new avenues for the development of successful treatments.

Research is continually being done to learn more about this problem.

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