Farber's Disease

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Farber's Disease Information Page.

Farber's disease, also known as Farber's lipogranulomatosis, describes a group of inherited metabolic disorders called lipid storage diseases, in which excess amounts of lipids (oils, fatty acids, and related compounds) build up to harmful levels in the joints, tissues, and central nervous system. The liver, heart, and kidneys may also be affected. Disease onset typically begins in early infancy but may occur later in life. Symptoms of the classic form may have moderately impaired mental ability and difficulty with swallowing. Other symptoms may include chronic shortening of muscles or tendons around joints. arthritis, swollen lymph nodes and joints, hoarseness, nodules under the skin (and sometimes in the lungs and other parts of the body), and vomiting. Some people may need a breathing tube. In severe cases, the liver and spleen are enlarged. Farber's disease is caused by a deficiency of the enzyme ceramidase. The disease occurs when both parents carry and pass on the defective gene that regulates the protein sphingomyelin. Children born to these parents have a 25 percent chance of inheriting the disorder and a 50 percent chance of carrying the faulty gene. The disorder affects both males and females.