Acid Lipase Disease

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Acid Lipase Disease Information Page.

Acid lipase disease or deficiency occurs when the enzyme needed to break down certain fats that are normally digested by the body is lacking or missing, resulting in the toxic buildup of these fats in the body's cells and tissues. These fatty substances, called lipids, include fatty acids, oils, and cholesterol. Two rare lipid storage diseases are caused by the deficiency of the enzyme lysosomal acid lipase, both of which are interited and affect males and females

- Wolman's disease (also known as acid lipase deficiency) is marked by the buildup of cholesteryl esters (normally a tranport form of cholesterol that brings nutrients into the cells and carries out waste) and triglycerides (a chemical form in which fats exist in the body). Infants with the disorder appear normal at birth but quickly develop progressive mental deterioration, low muscle tone, enlarged liver and grossly enlarged spleen, gastrointestinal problems, jaundice, anemia, vomiting, and calcium deposits in the adrenal glands, which causes them to harden.

- Cholesteryl ester storage disease (CESD) is an extremely rare disorder that results from storage of cholesteryl esters and triglycerides in cells in the blood and lymph and lymphoid tissue. Children develop an enlarged liver, leading to cirrhosis and chronic liver failure before adulthood. Children may also develop calcium deposits in the adrenal glands and jaundice. Onset varies, and the disorder may not be diagnosed until adulthood.