Lesch-Nyhan Syndrome

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Lesch-Nyhan Syndrome Information Page.

Lesch-Nyhan syndrome (LNS) is a rare, inherited disorder caused by a deficiency of the enzyme *hypoxanthine-guanine phosphoribosyltransferase* (HPRT). LNS is an X-linked recessive disease-- the gene is carried by the mother and passed on to her son. LNS is present at birth in baby boys. The lack of HPRT causes a build-up of uric acid in all body fluids, and leads to symptoms such as severe gout, poor muscle control, and moderate retardation, which appear in the first year of life. A striking feature of LNS is self-mutilating behaviors – characterized by lip and finger biting – that begin in the second year of life. Abnormally high uric acid levels can cause sodium urate crystals to form in the joints, kidneys, central nervous system, and other tissues of the body, leading to gout-like swelling in the joints and severe kidney problems. Neurological symptoms include facial grimacing, involuntary writhing, and repetitive movements of the arms and legs similar to those seen in Huntington's disease. Because a lack of HPRT causes the body to poorly utilize vitamin B12, some boys may develop a rare disorder called megaloblastic anemia.