Lesch-Nyhan Syndrome

National Cancer Institute

Source

National Cancer Institute. Lesch-Nyhan Syndrome. NCI Thesaurus. Code C61255.

An X-linked inherited syndrome caused by mutations in the gene that encodes the enzyme hypoxanthine-guanine phosphoribosyltransferase, resulting in accumulation of uric acid in the body. It affects males and is characterized by neurologic defects, moderate mental retardation, muscle hypotonia, and a tendency for self-mutilation (self-biting of lips, tongue, and fingertips).