Lesch-Nyhan Syndrome

Lesch-Nyhan syndrome (LNS) is a rare, genetic disorder caused by a deficiency of the enzyme hypoxanthine-guanine phosphoribosyltransferase or HPRT. It is an X-linked recessive disease, and the gene has been localized to the long arm of the X chromosome. LNS is characterized by self-mutilating behaviors such as lip and finger biting and/or head banging. The symptoms (problems) of LNS usually appear between the ages of 3 and 6 months. Frequently the first symptom is the presence of orange-colored crystal-like deposits (orange sand) in the diapers of affected infants. The deposits, which are called urate crystal formation, are caused by increased levels of uric acid in the urine. Uric acid levels, which are abnormally high in individuals with LNS, may also cause sodium urate crystals to form in the joints, kidneys, central nervous system, and other tissues of the body. Other symptoms of LNS may include kidney stones, blood in the urine, pain and swelling of the joints, difficulty swallowing (dysphagia) and eating, and vomiting, impaired kidney function, irritability, uncontrolled aggressive and/or compulsive actions, muscle weakness (hypotonia), uncontrolled spastic muscle movements, and neurological problems such as involuntary writhing movements of the arms and legs (athetosis) and purposeless repetitive movements (chorea) such as shoulder raising and lowering and/or facial grimacing. Moderate mental retardation is also common. Some individuals may develop a rare disorder called megaloblastic anemia (low red blood cells).

TREATMENT

Treatment for LNS is symptomatic. The drug allopurinol may be used to control excessive amounts of uric acid. Kidney stones may be treated with lithotripsy. There is no standard treatment for the neurological symptoms of LNS. Some symptoms may be relieved with the drugs carbidopa/levodopa, diazepam, phenobarbital, or haloperidol.

WHAT WILL HAPPEN

The prognosis (likely outcome) for individuals with LNS is poor as there is no treatment for the neurological defects. The build-up of uric acid within the body causes agonizing episodes of self-mutilation and may result in severe retardation and, ultimately, death.

RESEARCH BEING DONE

The NINDS supports and conducts research on genetic disorders such as LNS in an effort to find ways to prevent, treat, and, ultimately, cure these disorders.

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

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