Disability Awareness

Lesch-Nyhan Syndrome

What is Lesch-Nyhan syndrome?
Lesch-Nyhan syndrome is a condition that occurs almost exclusively in males. It is characterized by neurological and behavioral abnormalities and the overproduction of uric acid. Uric acid is a waste product of normal chemical processes and is found in blood and urine. Excess uric acid can be released from the blood and build up under the skin and cause gouty arthritis (arthritis caused by an accumulation of uric acid in the joints). Uric acid accumulation can also cause kidney and bladder stones.

The nervous system and behavioral disturbances experienced by people with Lesch-Nyhan syndrome include abnormal involuntary muscle movements, such as tensing of various muscles (dystonia), jerking movements (chorea), and flailing of the limbs (ballismus). People with Lesch-Nyhan syndrome usually cannot walk, require assistance sitting, and generally use a wheelchair. Self-injury (including biting and head banging) is the most common and distinctive behavioral problem in individuals with Lesch-Nyhan syndrome.

How common is Lesch-Nyhan syndrome?
The prevalence of Lesch-Nyhan syndrome is approximately 1 in 380,000 individuals. This condition occurs with a similar frequency in all populations.

What genes are related to Lesch-Nyhan syndrome?
Mutations in the HPRT1 gene cause Lesch-Nyhan syndrome. The HPRT1 gene provides instructions for making an enzyme called hypoxanthine phosphoribosyltransferase 1. This enzyme is responsible for recycling purines, a type of building block of DNA and its chemical cousin RNA. Recycling purines ensures that cells have a plentiful supply of building blocks for the production of DNA and RNA.

HPRT1 gene mutations that cause Lesch-Nyhan syndrome result in a severe shortage (deficiency) or complete absence of hypoxanthine phosphoribosyltransferase 1. When this enzyme is lacking, purines are broken down but not recycled, producing abnormally high levels of uric acid. For unknown reasons, a deficiency of hypoxanthine phosphoribosyltransferase 1 is associated with low levels of a chemical messenger in the brain called dopamine. Dopamine transmits messages that help the brain control physical movement and emotional behavior, and its shortage may play a role in the movement problems and other features of this disorder. However, it is unclear how a shortage of hypoxanthine phosphoribosyltransferase 1 causes the neurological and behavioral problems characteristic of Lesch-Nyhan syndrome.

Some people with HPRT1 gene mutations produce some functional enzyme. These individuals are said to have Lesch-Nyhan variant. The signs and symptoms of Lesch-Nyhan variant are often milder than those of Lesch-Nyhan syndrome and do not include self-injury.

How do people inherit Lesch-Nyhan syndrome?
This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males

(cont. on page 29)

GLADD Dances. 3rd Friday of each month, 5:30-7:30 pm, September-June. So. Park Comm. Ctr., 4851 So. Tacoma Way. $4 suggested donation per person. Music & snacks provided. For individuals 18 & over. Call Geri for more info. 253-223-4800. If you need special care or supervision, please bring support.

FrienDS Down Syndrome Play Group
Meets on Saturdays. Contact Susan Jackson at PAVE/Parent to Parent Program for more information. 253-565-2266 x107.

Community Inclusion Program. Activities are held at various locations around the county. Three Pierce County areas monthly. For more information or a schedule of events, please contact PAVE at 253-565-2266 or visit www.communityinclusionprogram.org

Companionship Program. Please contact Companionship Program at 253-798-2997 or email PCCompanionship@co.pierce.wa.us for program schedule.

(cont. from page 28)

(who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

What other names do people use for Lesch-Nyhan syndrome?
choreoathetosis self-mutilation syndrome
complete HPRT deficiency
complete hypoxanthine-guanine phosphoribosyltransferase deficiency
deficiency of guanine phosphoribosyltransferase
deficiency of hypoxanthine phosphoribosyltransferase
HGPRT deficiency
hypoxanthine guanine phosphoribosyltransferase deficiency
hypoxanthine phosphoribosyltransferase deficiency
juvenile gout, choreoathetosis, mental retardation syndrome
juvenile hyperuricemia syndrome
Lesch-Nyhan disease
LND
LNS
primary hyperuricemia syndrome
total HPRT deficiency
total hypoxanthine-guanine phosphoribosyl transferase deficiency
X-linked hyperuricemia
X-linked primary hyperuricemia
X-linked uric aciduria enzyme defect

This resource information should not be used as a substitute for professional medical care or advice. Users seeking information about a personal genetic disease, syndrome, or condition should consult with a qualified healthcare professional.

Edited from: http://ghr.nlm.nih.gov/condition/lesch-nyhan-syndrome