Condition: Lesch-Nyhan syndrome

Inheritance:
X-linked recessive.

Genetic etiology:
Mutation in *HPRT1* gene, encoding hypoxanthine-guanine phosphoribosyltransferase.

Frequency:
1/380,000.

Clinical features:
Affected males present in infancy with hypotonia and developmental delay. There is a progressive motor disorder involving both pyramidal and extrapyramidal components. There is compulsive self-injurious behavior and deposition of uric acid in joints and uric acid calculi in the urinary tract. Milder forms may present with neurological dysfunction or hyperuricemia and renal dysfunction.

Management:
Allopurinol to block conversion of hypoxanthine and xanthine to uric acid; supportive care.

Genetic counseling:
Based on X-linked recessive inheritance. Molecular genetic testing is available.

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