Condition: Ataxia-telangiectasia

Inheritance:

Autosomal recessive.

Genetic etiology:

Mutation in *ATM* gene, which encodes a protein involved in the detection of DNA damage.

Frequency:

1/40,000 – 1/100,000.

Clinical features:

Progressive ataxia begins in childhood, along with slurred speech and oculomotor apraxia (difficult following movement with eyes), and choreoathetosis. Telangiectasia occur on skin and conjunctivae. There is increased susceptibility to infection due to immunodeficiency, and a markedly increased frequency of leukemia and lymphoma.

Management:

Supportive care.

Genetic counseling:

Parents of affected child have 25% risk of recurrence. Genetic testing is available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.