Condition: Xeroderma pigmentosum

Inheritance:

Autosomal recessive.

Genetic etiology:

Genetically heterogeneous; mutation in various genes involved in DNA repair.

Frequency:

Approximately 1/1,000,000.

Clinical features:

Skin changes, including pigmentation and dryness, occur with sun exposure; approximately 50% have severe sunburn on UV exposure. Abnormalities of anterior portion of eye, including inflammation, carcinoma, and melanoma, are common. Approximately 30% have neurological abnormalities, including microcephaly and cognitive impairment.

Management:

Avoidance of sun exposure; supportive care.

Genetic counseling:

Parents of affected child have 25% recurrence risk; molecular genetic testing for some of the genes responsible for XP is available.

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