Condition: Bloom syndrome

Inheritance:
Autosomal recessive.

Genetic etiology:
Mutation in *BLM* gene that encodes a DNA helicase protein.

Frequency:
Rare (more common in Ashkenazi Jewish population).

Clinical features:
Individuals with Bloom syndrome have low birth weight and proportionate short stature as they grow. There are distinctive facies, with underdeveloped malar bones and a sun-sensitive rash appears on the face, hands, and forearms. There may be several café-au-lait spots. There is an immune deficiency with low immunoglobulins and increased susceptibility to infection. Susceptibility to multiple forms of cancer usually is the cause of death.

Management:
Monitoring for major complications; supportive care.

Genetic counseling:
Parents of affected child have 25% recurrence risk; molecular genetic testing available.

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