Bloom’s syndrome

Genetics

-BLM (15q26.1)

-AR

Clinical findings/Dysmorphic features

-Severe pre- and postnatal growth deficiency; short stature throughout postnatal life

-Sparseness of subcutaneous fat tissue throughout infancy and early childhood

-Erythematous and sun-sensitive skin lesion of the face (Butterfly rash)

-Women may be fertile, but menopause occurs unusually early; men are infertile

-Immunodeficiency; increased risk of cancer (wide distribution of type and site (colon most common), often multiple primary tumors)

Etiology

-1/100 carrier frequency in AJ

Pathogenesis

-Abnormal DNA replication and repair leading to genomic instability --> chromosome breakage

Genetic testing/diagnosis

-Identification of biallelic pathogenic variants in BLM

-c.2207\_2212delinsTAGTTC in AJ (97%), no del/dup reported

-If genetic testing is inconclusive --> increased frequency of sister-chromatid exchanges on specialized cytogenetic studies

Others

-Normal intelligence

-Chromatid/chromosome breaks; triradial and quadriradial figures

-Harlequin Chromosomes