Bloom Syndrome

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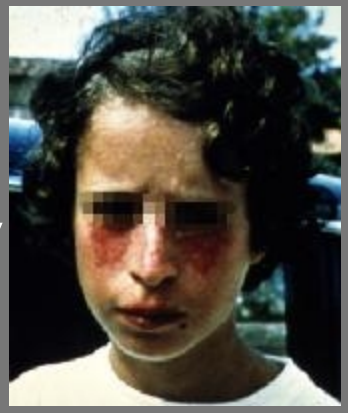
Clinical Features:

- Short stature; small cranium
- Sparseness of subcutaneous fat tissue in infancy
- *Facial rash, often butterfly-shaped on cheek*
- High-pitched voice
- Long, narrow face



more characteristics:

- Men are sterile; women have reduced fertility
- Most have normal intellectual ability
- Longest survival: 49 years old [mean: 24 years old]

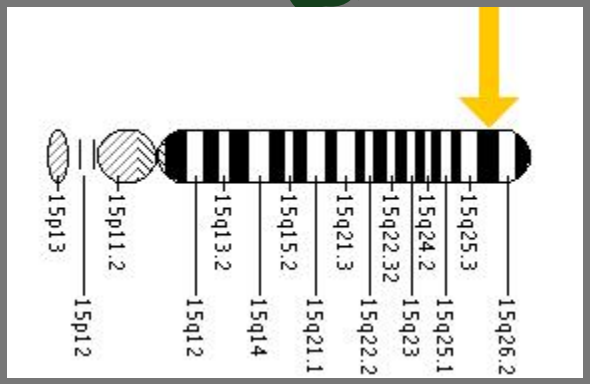
Susceptible to:

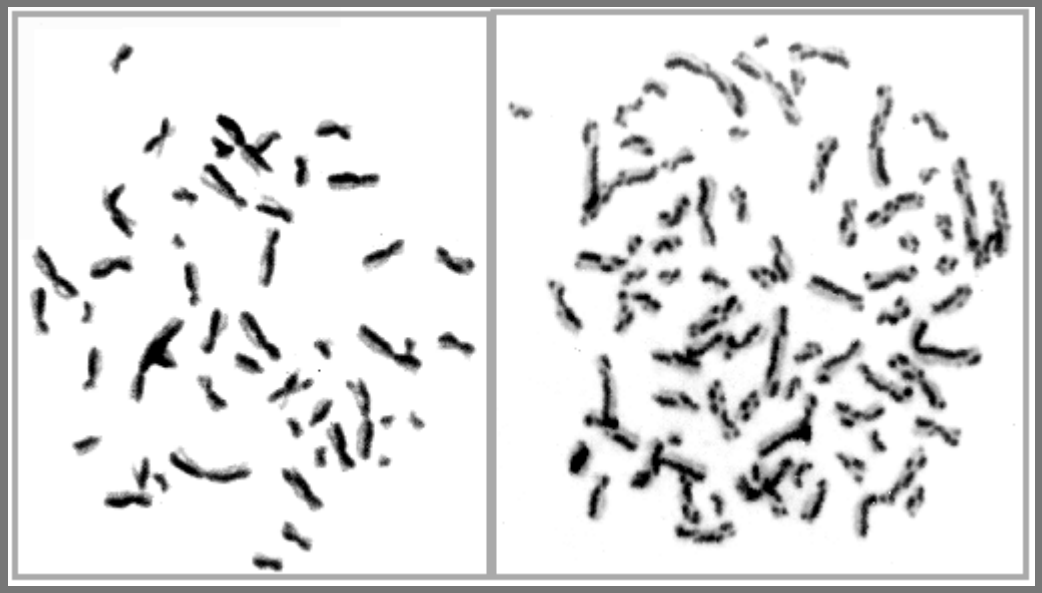
- Lower urinary tract obstruction in men
- Chronic obstructive pulminary disease
- Diabetes Mellitus
- Myelodisplacia
- ***Cancer***
  - including Leukemia
- Infections
  - including Otitus media
  - including Pneumonia

What's happening?

BLM Gene *(loss of function)*

- located on Chromosome 15 (15q26.1)
- DNA helicase RecQ protein-like-3
  - Opens DNA for replication
  - Maintains "genetic integrity"





Normal SCE

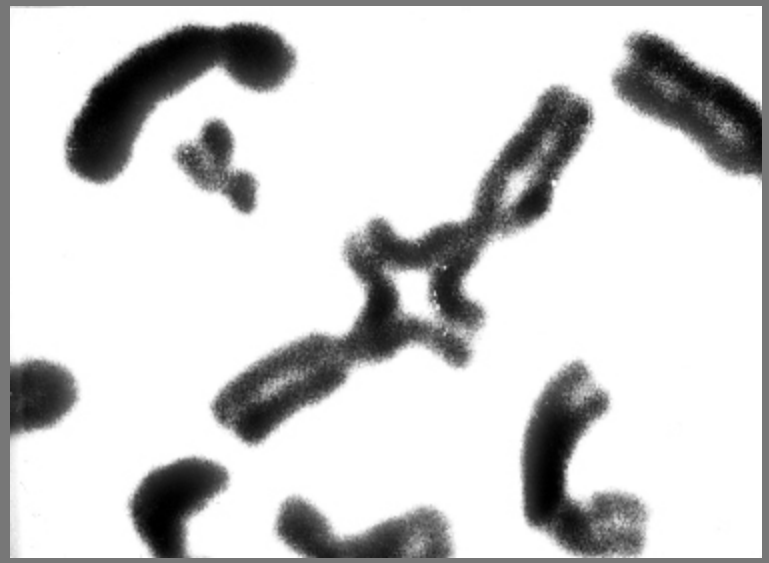
SCE in Bloom syndrome patient

Increase in Sister Chromatid exchange (SCE)

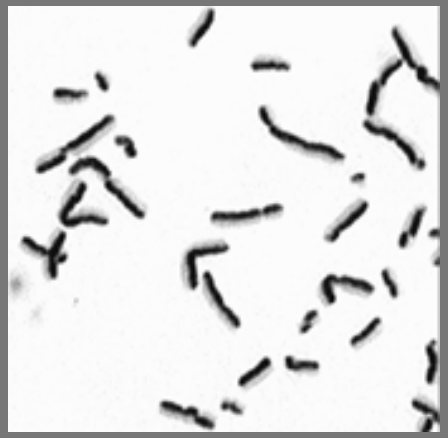
Diagnostic Methods:

Phenotype in Cytogenetics (chromosomes)

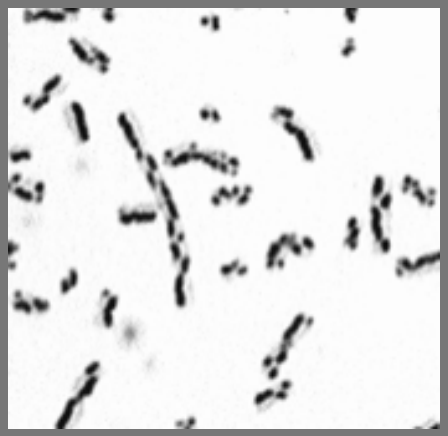
1. Quadriradial configurations: symmetrical, four-armed chromatid interchange configuration



2. Increased Frequency of SCEs



Few (<10) SCEs/cell



90+ SCEs/cell

Diagnostic Methods:

Genotype with knowledge of the BLM gene

- Molecular genetic analysis of BLM gene
- Prenatal diagnostics

Treatment:

- Can't treat disease directly
- Treat the manifestations:
  - **Infections**: routine antibiotic treatment
  - **Diabetes metillus**: standard treatment
  - **Cancer**: *modification* of standard treatment, reducing dosage and duration

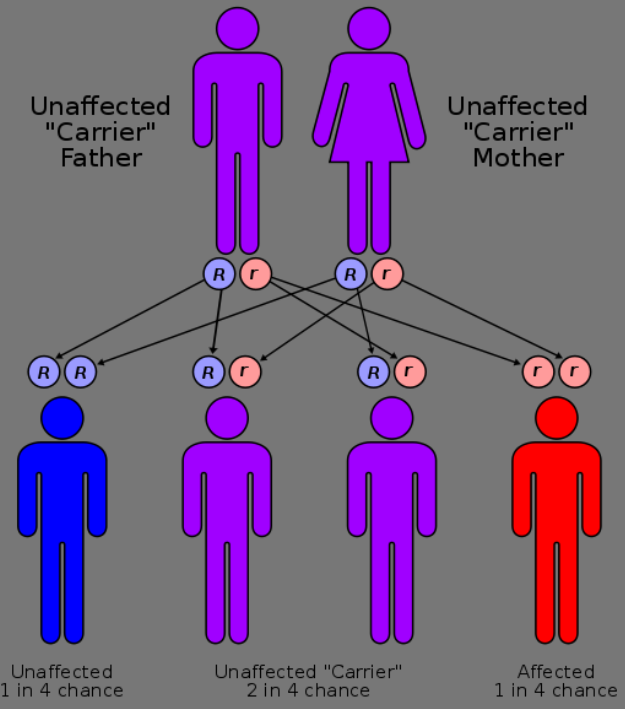
Management:

- Avoid sun exposure to face
- Frequent breast + colon cancer screening

Ashkenazi Jews

- 72 of the 265 infected (2009) were of Ashkenazi Jewish ancestry
- High % of Ashkenazi Jewish carriers
  - New York: 1/100
  - Israel: 1/37

•**Autosomal recessive disorder**; two carriers have 25% chance of having affected child

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Sources

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- http://ghr.nlm.nih.gov/condition/bloom-syndrome
- http://www.ncbi.nlm.nih.gov/books/NBK1398/
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