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 pulmonary disease
- Diabetes Mellitus
- Myelodisplasia
- **Cancer**
  - including Leukemia
- Infections
  - including Otitis 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”

**Increase in Sister Chromatid exchange (SCE)**

Normal SCE  |  SCE in Bloom syndrome patient
Diagnostic Methods:

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

Phenotype in Cytogenetics (chromosomes)

2. Increased Frequency of SCEs

- Few (<10) SCEs/cell
- 90+ SCEs/cell
Diagnostic Methods:

- Molecular genetic analysis of BLM gene
- Prenatal diagnostics

Genotype with knowledge of the BLM gene
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

- http://atlasgeneticsoncology.org/Kprones/BLO10002.html
- http://www.med.cornell.edu/bsr/lab_diagnosis/