BLOOM SYNDROME

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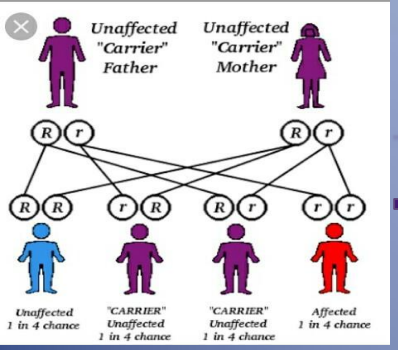


INTRODUCTION

- Bloom syndrome, also called Bloom-Torre-Machacek syndrome or congenital telangiectatic erythema (MIM 210900), is a rare autosomal recessive inherited disorder characterized by genomic instability and predisposition to the development all types of cancer.

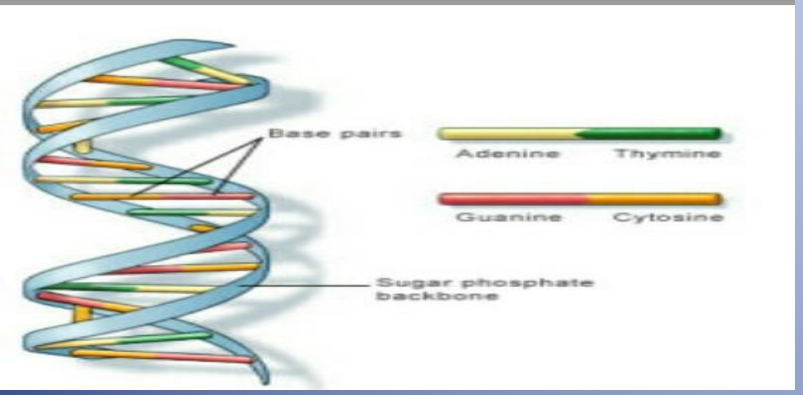
- Bloom syndrome is due to mutations in the *BLM* gene inducing the formation of an abnormal DNA helicase protein.

- The most prominent features include growth deficiency of prenatal onset, mild immunodeficiency, excessive photosensitivity with facial lupus-like skin lesions, type 2 diabetes mellitus and hypogonadism.

DEFINITION

- Bloom syndrome is an inherited disorder that is characterized by short stature, sun sensitive skin changes, &increased chances of cancer and other serious health problems.

- Cells from a person with Bloom Syndrome exhibit a striking genomic instability that includes excessive homologous recombination.



CAUSES

- The genetic abnormality in Bloom Syndrome causes problems with DNA repair, resulting in a high number of chromosome breaks and rearrangements.

- The abnormal DNA repair is responsible for cancer.

- Bloom Syndrome is inherited as an autosomal recessive genetic trait.

SYMPTOMS

- *The most prominent features include short stature, and arash on the face that develops early in life when exposed to the sun.*

- *The skin rash is erythematous or red;telangiectatic, i.e. characterized by dilated or spider veins; infiltrated and scaly, it can appear across the nose, on the cheeks and around the lips.*

DIFFERENT THERAPY

- Bloom syndrome (congenital telangiectatic erythema) has no specific treatment; however, avoiding sun exposure and using sunscreens can help prevent some of the cutaneous changes associated with photosensitivity.

- Efforts to minimize exposure to other known environmental mutagens are also advisable.

- Identification of café au lait macules by dermatologists would help to identify the risk of neoplasms in these patients.

PREVENTION

Since Bloom syndrome is a genetic disorder, no preventative measures can be taken against the disorder. However, prenatal diagnosis is possible with amniocentesis. Also, DNA analysis is expected to be available in the future.

BIBLIOGRAPHY & REFERENCE

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Thank
You