DONE BY:

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WITH CARE OF:

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INTRODUCTION

- Xeroderma Pigmentosum is an AUTOSOMAL RECISIVE HERIDITARY DISEASE of DNA repair.

- Passed down through families in which the skin and tissue covering the eye are extremely sensitive to ultraviolet light.
First discovered in late 1800s by Moritz Kaposi.

Severe lesions, tumors and skin deformations result from sun exposure (UV).
CAUSES

Exposure to unfiltered sunlight cause the formation of pyrimidine dimers (usually thymines). In normal condition, the body repairs this damage. But in persons with xeroderma pigmentosa, the body does not correct the damage, because of there are hereditary defects in the genes responsible for producing enzymes that correct dimers mainly (UV specific endonuclease or UV rABC exonuclease).

This leading to accumulation of mutations and after that leading to cancer.

Skin cancer occurs before the child is 5 years old.
XERODERMA PIGMENTOSUM
DNA DAMAGES

Exposure to UV light

Thymine dimer
(this dimers prevent DNA polymerase from replicating the DNA strand).

No replicating DNA strand
autosomal recessive genetic disorder of DNA repair
SYMPTOMS

- Sunburn that does not heal after just a little bit of sun exposure.
- Blistering after just a little bit of sun exposure.
- Spider-like blood vessels under the skin.
- Patches of discolored skin that get worse.
- Crusting & Scaling of the skin.
- Oozing raw skin surface.
- Discomfort when being in bright light (photophobia).
TREATMENT

- Children with this condition need total protection from sunlight. Even the light coming through windows and fluorescent bulbs are dangerous.
- When these children must go out in the sun, they should wear protective clothing or mask.
- Wear high protection sunscreen and very dark, UV-protected glasses. Your doctor may prescribe medicine to help prevent certain skin cancers.
Protective mask
Many patients die at an early age from skin cancers. but, if a person is diagnosed early, does not have severe neurological symptoms and takes all the precautionary measures to avoid exposure to UV light, they may survive beyond middle age.
< 40% of individuals with the disease survive beyond age 20. Some with less severe cases manage to live well into their 40s.
Experts recommend genetic counseling for persons with a family history of xeroderma pigmentosa who wish to have children.
WHEN TO CONTACT A DOCTOR

Call for an appointment with your health care provider if you or your child has symptoms of xeroderma pigmentosa.
THANK YOU