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THE WONDER HAS A NAME

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#MADEEASY

XERODERMA PIGMENTOSUM

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- ***Xeroderma Pigmentosum (XP) is a rare, inherited disorder characterized by extreme sensitivity to ultraviolet (UV) rays from sunlight.***
- ***It is a result of defects in the nucleotide excision repair pathway, which is responsible for repairing UV-induced DNA damage.***
- ***Think of individuals with XP as having paper-thin skin when it comes to sunlight, where even minimal exposure can cause significant damage.***

Pathophysiology

- ***Autosomal recessive disorder caused by mutations in any one of the several genes involved in the NER pathway (e.g., XPA, XPC, ERCC2, ERCC3, DDB2).***
- ***This pathway is like the body's "repair shop" for fixing DNA damaged by UV light.***
- ***Defective DNA repair leads to accumulation of UV-induced DNA damage, resulting in skin changes, premature aging, and a high risk of skin cancers.***

Clinical Features

- ***Cutaneous Manifestations:***
  - ***Photosensitivity causing severe sunburns after minimal sun exposure***
    - ***Imagine someone who gets sunburned just by sitting near a sunny window***
  - ***Early onset of extensive freckling in sun-exposed areas***
  - ***Abnormally dry, scaly skin (Xerosis).***
  - ***Irregular pigmentation, hypopigmented macules***
  - ***Thinning of the skin, loss of elasticity.***
  - ***Premature aging***
  - ***High incidence of basal cell carcinoma, squamous cell carcinoma, and melanoma at a young age***

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- ***Ocular Manifestations:***
  Photophobia (Sensitivity to light)
  Chronic inflammation of the conjunctiva.
  Keratitis (Inflammation of the cornea)
  Corneal Opacities like clouding of the cornea, leading to vision impairment.
- ***Neurological Manifestations (in some cases):***
  Microcephaly
  Intellectual Disability
  Progressive hearing loss

Diagnosis

- ***Clinical Evaluation based on characteristic skin and eye findings, especially in the context of early-onset and severe photosensitivity.***
- ***Genetic Testing confirms the diagnosis by identifying mutations in NER pathway genes.***
- ***Skin Biopsy shows histopathological changes typical of sun damage and skin cancer.***

Management

- ***Strict avoidance of UV exposure through the use of high-SPF sunscreens, protective clothing, wide-brimmed hats, and UV-blocking sunglasses.***
- ***Topical Treatments: Use of topical retinoids and 5-fluorouracil for premalignant lesions.***
- ***Oral Retinoids: May help reduce the risk of skin cancers in some patients.***
- ***Regular skin checks and prompt removal of any suspicious lesions.***
- ***Regular eye check-ups to monitor and treat ocular complications.***
- ***Assessment and support for any neurological symptoms.***
- ***Quality of life can be significantly impacted by the need for constant UV protection and the risk of skin cancers and neurological complications.***