XERODERMA PIGMENTOSUM

FIRST STAGE

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MEDICAL BIOCHEMISTRY

Xeroderma Pigmentosum (XP) :

Xeroderma Pigmentosum (XP) is a rare genetic disorder characterized by

extreme sensitivity to ultraviolet (UV) rays from sunlight, leading to a high risk of skin damage and cancer.

molecular basis
Nucleotide Excision Repair (NER) pathway dysfunction is the main mechanism.

NER normally recognizes and removes bulky DNA lesions and then fills and ligates the resulting gap.

mutations in genes encoding proteins required for NER or for translesion synthesis cause XP.

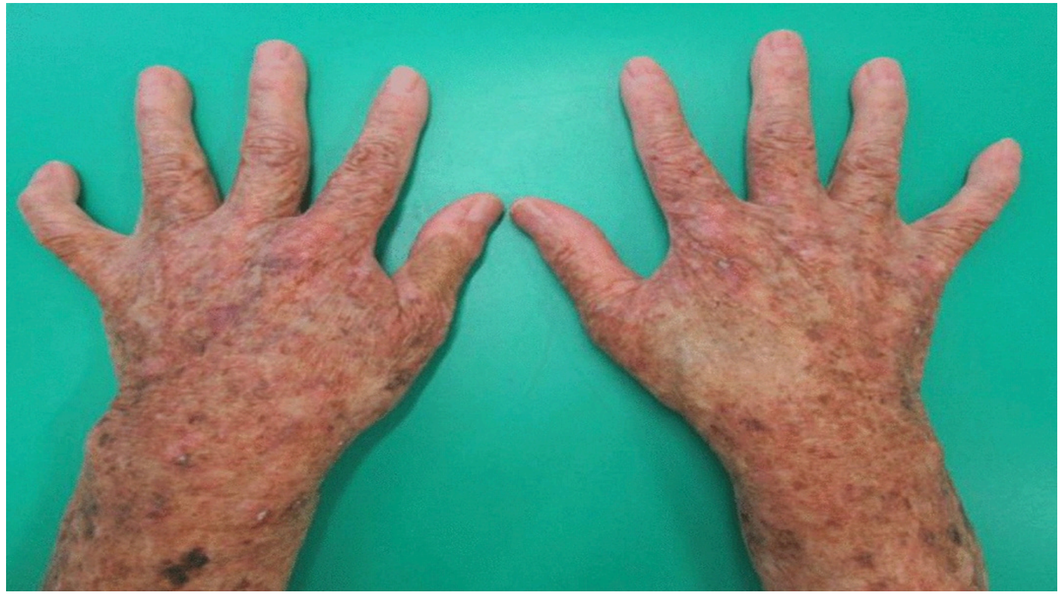
these genes form complementation groups, classically labeled XP-A through XP-G, plus a variant form (XP-V).

Cockayne Syndrome :
Cockayne Syndrome is a rare genetic disorder characterized by growth failure, premature aging, and sensitivity to sunlight.

(etymology) XERO-: from Greek "xēros" (ξηρός) meaning "dry."

DERMA: from Greek "derma" (δέρμα) meaning "skin."

PIGMENTOSUM: from Latin "pigmentum" meaning "pigment" or "coloring," with the adjectival suffix "-osum" indicating abundance—so "pigment-rich" or "marked by pigmentation."



THANK YOU FOR YOUR ATTENTION !