

Disease

ERCC2 associated xeroderma pigmentosum^{1, 2, 3}

Gene

ERCC2

Inheritance	AR
Current Treatments	avoid exposure to UVA and UVB (found in sunlight) and UVC (found in some artificial light sources). Oral isotretinoin, oral niacinamide, topical imiquimod and topical fluorouracil
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	no
Disease Frequency (per 100,000)	0.2 ⁵
Genes Included in Disease Frequency	DDB2, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Xeroderma+Pigmentosum&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1397/>

² <https://omim.org/entry/278730>

³ <https://pubmed.ncbi.nlm.nih.gov/35520754/>

⁴ <https://pubmed.ncbi.nlm.nih.gov/28771907/>

⁵ <https://pubmed.ncbi.nlm.nih.gov/35246173/>