

Disease

X-linked erythropoietic protoporphyria^{1, 2}

Gene

ALAS2

Inheritance	XL
Current Treatments	Afamelanotide, Bitoperton, avoidance of sunlight and UV light
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	free erythrocyte protoporphyrin and zinc-chelated erythrocyte protoporphyrin ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=Protoporphyria&city=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK121284/>

² https://www.discmedicine.com/wp-content/uploads/2024/12/Results_from_the_BEACON_Trial.pdf