

Disease

Erythropoietic protoporphyria-1^{1, 2}

Gene

FECH

Inheritance	AR
Current Treatments	Afamelanotide, Bitopertin, avoidance of sunlight and UV light
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	free protoporphyrin ¹
Disease Frequency (per 100,000)	0.35 ^{3, 4}
Genes Included in Disease Frequency	FECH
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Protoporphyria&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK100826/>
- ² https://www.discmedicine.com/wp-content/uploads/2024/12/Results_from_the_BEACON_Trial.pdf
- ³ <https://www.ncbi.nlm.nih.gov/books/NBK100826/#epp-ar.Summary>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/32873934/>