

Disease

**X-linked erythropoietic protoporphyria<sup>1, 2</sup>**

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 <sup>1</sup>                                                     |
| Experimental / Investigational Therapies | <a href="https://clinicaltrials.gov/search?cond=Protoporphyria&amp;city=">https://clinicaltrials.gov/search?cond=Protoporphyria&amp;city=</a> |

References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK121284/>
- <sup>2</sup> [https://www.discmedicine.com/wp-content/uploads/2024/12/Results\\_from\\_the\\_BEACON\\_Trial.pdf](https://www.discmedicine.com/wp-content/uploads/2024/12/Results_from_the_BEACON_Trial.pdf)