

PROTOPORPHYRIA PATIENT CARE

Erythropoietic Protoporphyria (EPP) & X-Linked Prophyria (XLP)

OVERVIEW

EPP and XLP affect multiple body systems, causing severe phototoxic reactions and potential liver complications. This tool is designed to help patients with EPP and XLP communicate with their healthcare providers about the recommended checks and management strategies for their condition. It is based on consensus guidelines developed by experts from the Porphyrrias Consortium and the United Porphyrrias Association (UPA). *This tool is not intended to be medical advice. Please refer to the full article, consult your physician, or contact the UPA for referral to a porphyria specialist.*

- **Reference:** Dickey, Amy K et al. "Evidence-based consensus guidelines for the diagnosis and management of erythropoietic protoporphyria and X-linked protoporphyria." *Journal of the American Academy of Dermatology* vol. 89,6 (2023): 1227-1237. doi:10.1016/j.jaad.2022.08.036
- **Patient friendly summary:** porphyria.org/experts-in-action-page/sumup8

DIAGNOSIS

Labs/Test	Frequency	Purpose/Notes
Blood Test for Total Erythrocyte Protoporphyrin	Once for diagnosis, then as below in section of screening and monitoring	To diagnose EPP and XLP. Ensure the test measures metal-free and zinc-bound protoporphyrin and is completed at a recommended lab (UMTB Galveston Porphyria Center or Mayo Clinic- contact UPA for more information).
Genetic Testing	Once, after biochemical diagnosis	To distinguish between EPP (FECH gene mutations) and XLP (ALAS2 gene mutations).

RECOMMENDED SCREENING AND MONITORING

Labs/Test	Frequency	Purpose/Notes
Complete Blood Count (CBC)	Annually	Screen for anemia, common in EPP and XLP. Iron supplementation considerations differ between EPP and XLP (see section on treatment).
Total Erythrocyte Protoporphyrin	Annually	Monitor for changes from baseline. Increases in PPIX in a patient may result in increased photosensitivity and can indicate liver damage.
Liver Function Tests	Annually	Screen for liver disease. The recommended monitoring intervals will increase to every 3 or 6 months if you have abnormal liver function. Please see the separate EPP/XLP liver disease guidelines for more information.
Blood Test for Vitamin D Levels	Annually	Vitamin D deficiency is common due to lifelong sunlight avoidance, treat deficiencies according to local guidelines.
Psychological Screening	As recommended for general population	Screen for anxiety and depression and provide appropriate care.

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TREATMENT

Condition/ Symptom	Treatment	Frequency
Photosensitivity	Afamelanotide (Scenesse) may be prescribed to adults to increase pain-free sun exposure and quality of life. Dermatology check-ups recommended every 6 months for those on afamelanotide	Implant every 2 months
Phototoxic Reactions	Cooling devices and cold compresses may provide minor relief.	As needed
Anemia	EPP: Iron supplementation only if anemia symptoms are present along with low hemoglobin/ferritin. Supplementation may result in increased protoporphyrin levels and photosensitivity. XLP: Iron supplementation recommended and may reduce protoporphyrin levels and photosensitivity	As needed

MANAGING AND LIVING WITH PORPHYRIA

	Recommendations
Preventing Phototoxic Reactions	<ul style="list-style-type: none"> • Sunlight avoidance, including reflections off snow and water; may need to avoid fluorescent and other artificial lights in 340-650nm range • Opaque clothing • Broad-spectrum sunscreens (zinc oxide or titanium dioxide) • Window tinting
Protect Liver and Bile System Health and Reduce Risk of Liver Disease	<ul style="list-style-type: none"> • Hepatitis A and B vaccination • Avoid contraceptives with high doses of estrogen • Minimizing alcohol intake • Healthy lifestyle- maintain healthy body weight, exercise, healthy diet
Build a Support Network	Engage with United Porphyrins Association to connect with healthcare professionals, research, and patient support activities and groups.
Accommodations	Seek accommodations at work or school and educate peers about the condition.
Self-Advocacy	Learn to explain this disease and advocate for your needs.

SPECIAL CONSIDERATIONS

Consideration	Test/Procedure	Purpose/Notes
Surgery	Light acrylate yellow filters for wavelengths 340-470nm for prolonged surgeries	Ensures safety during long surgeries. No anesthetics are contraindicated.
Pregnancy	Standard prenatal care	No increased monitoring needed. Porphyrin symptoms may improve, but this is inconsistent.
Light and Laser Therapies	Use lights outside 340-650nm range	Ensure safety with a spot test before treatment.

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ANNUAL EPP MONITORING AND TESTING TRACKER (2024-2030)

Use this table to keep track of recommended testing.

Year	Complete Blood Count (CBC)	Liver Function Tests	Vitamin D Levels	Psychological Screening (as recommended)	Afamelanotide (Scenesse) (every 2 months, if applicable)	Dermatology Check-up (every 6 months)
Sample	Jan 15, 2023	Jan 15, 2023	Jan 15, 2023		Jan 15, Mar 12, May 7, July 11, Sept 6, Nov 8	Feb 3, 2023 Aug 7, 2023
2024						
2025						
2026						
2027						
2028						
2029						
2030						

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