



[Authoritative facts](#) about the skin from the [New Zealand Dermatological Society Incorporated](#).

[Home](#) | [Skin signs of systemic disease](#)

Erythropoietic protoporphyria

What is erythropoietic protoporphyria?

Erythropoietic protoporphyria (EPP) is one of a group of genetic diseases called the porphyrias. EPP is due to an inherited deficiency of the enzyme ferrochelatase. Reduced activity of this enzyme causes a build-up of the chemical protoporphyrin in the skin; resulting in [photosensitivity](#) i.e. the skin is damaged by light. Abnormally high levels of protoporphyrin can rarely cause liver disease.

Who gets EPP?

People of all races may get EPP. Both males and females are equally affected. The condition usually starts in childhood.

What are the signs and symptoms?

First symptoms usually appear in infancy or early childhood and present as an uncomfortable or painful burning sensation of the skin after sun exposure. It occurs most often on the tops of the hands and feet, face and ears. In most cases visible changes to the skin are mild. The affected skin may become red and swollen and blistered. Later there are pitted scars and sometimes crusty thickened skin, particularly over the cheeks, nose and knuckles of the hands.

Erythropoietic protoporphyria



Acute sunburn
Image provided by K Searle



Scarring on cheeks and nose



Aged appearance of hands

People with EPP-induced liver disease usually have mild changes in liver blood tests. About 10% develop more severe liver disease, presenting with malaise, pain under the ribs on the right, jaundice and increasing photosensitivity.

Gallstones are common in patients with EPP.

How is EPP diagnosed?

Diagnosis is usually made by finding increased levels of protoporphyrin in the blood and reduced ferrochelatase enzyme activity.

What treatment is available?

Lifelong photosensitivity is the major problem for EPP uncomplicated by liver disease. Patients should protect their skin:

- Avoid unnecessary exposure to sunlight and wear [protective clothing](#) and wide-brimmed hats. Consider tinting windows.
- Other strong sources of light may also cause symptoms, including fluorescent and halogen lights. Protect the skin from exposure to operating lamps during a surgical procedure.
- [Sunscreens](#) may be helpful, especially formulations containing zinc oxide or titanium dioxide that reflect visible light.
- Oral beta-carotene (a food precursor of vitamin A found naturally in tomatoes and carrots) is thought to help reduce photosensitivity in some people. The dose for children is 30 to 150mg per day (1 to 5 30-mg capsules) and for adults it is 30 to 300mg (1 to 10 30-mg capsules). [Polypodium leucotomas](#) has also been used.
- Cysteine 500mg twice daily reduces photosensitivity
- Colestyramine reduces photosensitivity and hepatic protoporphyrin content.
- [Narrowband UVB](#) phototherapy increases melanin content and induces skin thickening so may reduce sun sensitivity.

Patients who also have liver disease require specialist medical treatment and possibly liver transplantation.

Congenital erythropoietic porphyria is now curable by stem cell transfusion, paving hope for the future, but there is not yet a cure available for EPP.

Related information

References:

Book: Textbook of Dermatology. Ed Rook A, Wilkinson DS, Ebling FJB, Champion RH, Burton JL. Sixth edition. Blackwell Scientific Publications.

On DermNet NZ:

- [Photosensitivity](#)
- [Porphyria cutanea tarda](#)
- [Variegated porphyria](#)

Other websites:

- [American Porphyria Foundation](#)
- [Canadian Porphyria Foundation](#)
- [New Zealand Porphyria Support Group](#) (email)
- [Erythropoietic protoporphyria](#) - e-medicine dermatology, the online textbook

Books:

See the [DermNet NZ bookstore](#)

Author: Vanessa Ngan, staff writer

DermNet does not provide an on-line consultation service.

If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

Created 2003. Last updated 13 Nov 2007. © 2008 NZDS. Disclaimer.