

EARLY ACUTE PRESENTATION IN THE NEONATAL PERIOD OR IN INFANTS (BEFORE LANGUAGE ACQUISITION)

After phototherapy for neonatal jaundice:

Generalised erythema
Oedema

After sun exposure: on sun-exposed skin

Erythema
Oedema of the hands and face
Crying
Hand rubbing

ACUTE PRESENTATION AFTER LANGUAGE ACQUISITION

From a few minutes to several dozen minutes after sun exposure

Reversible prodromal signs on sun-exposed skin
Tingling
Itching
Sensation of heat or burning

If sun exposure is continued: phototoxic crisis (lasting 2 to 7 days)

Unbearable burning pain affecting the hands, nose, mouth, ears and feet

Inconsistent objective lesions:

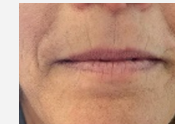
- Erythema
- Oedema
- Ulcerated and crusted lesions on the hands, ears, and nose

CHRONIC SYMPTOMS

Licheniform thickening of the skin on the backs of the interphalangeal and metacarpophalangeal joints



Perioral rhagades



Significant psychological impact
Signs of vitamin D deficiency

Additional tests

Standard blood workup

Standard workup may be normal or show:

Moderate elevation of AST/ALT
Rare cases of cholestatic hepatitis progressing to liver failure
Iron deficiency
Microcytic anaemia
Vitamin D deficiency

Erythropoietic protoporphyria?

Specialist assessment in collaboration with an expert centre

Measurement of erythrocyte and plasma protoporphyrin IX (abnormal even outside acute episodes)

Specialist confirmatory workup (biochemical, enzymatic and molecular porphyria typing) and follow-up in conjunction with an Expert Centre: Reference Centre for Rare Diseases:
<https://www.porphyrrie.net/>

Specific treatment for phototoxic crisis:

- Short course of corticosteroids if oedema is present
- Even step 3 analgesics provide no relief
- Patient should remain in darkness
- Gentle airflow at room temperature
- Use of a mist spray
- Allodynia: avoid touching the patient

Preventive treatment:

- UVB therapy, opaque topical creams
- Anti-UV cream ineffective

Genetic counselling, family screening in a specialised centre

