



## :: Cutaneous porphyrias



- This document is a translation of the French recommendations drafted by Prof. Jean-Charles Deybach and Dr. Hervé Puy, reviewed and published by Orphanet in 2007.
- Some of the procedures mentioned, particularly drug treatments, may not be validated in the country where you practice.



- See also the emergency guidelines for [acute hepatic porphyria neuro-visceral crisis](#)

### Synonyms:

Bullous porphyria:

- 1- Cutaneous porphyria, sporadic or familial;
- 2- Porphyria variegata;
- 3- Hereditary coproporphyrin

Günther disease or congenital erythropoietic porphyria

Photosensitive porphyria: erythropoietic porphyria

### Definition:

Porphyrias are monogenic and autosomal genetic conditions and each is linked to a deficiency of haem metabolising enzymes.

Cutaneous porphyrias are characterised by specific cutaneous lesions in zones exposed to sunlight (photodermatosis). There are two groups of cutaneous porphyrias: **bullous porphyrias** and **erythropoietic protoporphyrias** that are mainly photosensitive reactions.

### Further information:

[See the Orphanet abstract](#)

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# Pre-hospital emergency care recommendations

## Call for a patient suffering from cutaneous porphyria

### Synonyms

- ▶ Porphyric photodermatoses: bullous porphyria, photosensitive porphyria, Günther's disease

### Aetiology

- ▶ Deficiency of an enzyme involved in haem synthesis

### Special risks in an emergency

- ▶ hepatic colic, hepatic insufficiency
- ▶ haemolysis (Günther's disease)
- ▶ vesicular eruptions, skin pain, sun photosensitivity, cutaneous secondary infections
- ▶ spontaneous fractures

### Frequently used long term treatments

- ▶ beta-carotene
- ▶ cutaneous antiseptics
- ▶ transfusions
- ▶ immunosuppressors (transplant patient: bone marrow, liver)

### Complications

- be careful to exposure to the sun (contra-indicated) and trauma, even if minor
- triggering factors and contra-indicated medicaments: alcohol, oestrogens, porphyrinogenic drugs
- be careful: depending on the sources (official websites) the list of authorised, uncertain or contra-indicated medicaments are not identical

### Specific medical care prior to hospitalisation

- ▶ no vital risk other than haematological and hepatic risks
- ▶ contra-indication: barbiturates, etomidate, ketamine, sulfamides, photosensitisers...
- ▶ authorised drugs: morphine, fentanyl, midazolam, succinylcholine, vecuronium, atracurium...
- ▶ sun protection is essential, minimise micro-traumas, careful asepsis, avoid vascular approach in a photo-exposed zone
- ▶ orientation: intensive care if hepato-cellular involvement is suspected
- ▶ no specific treatment prior to hospitalisation

### For further information

- ▶ [www.orpha.net](http://www.orpha.net)
- ▶ [www.porphyria-europe.com](http://www.porphyria-europe.com)
- ▶ [www.porphyrie.net](http://www.porphyrie.net)
- ▶ [www.drugs-porphyria.org](http://www.drugs-porphyria.org)

# Recommendations for hospital emergency departments

## Emergency situations

### 1. Emergency diagnostic measures

#### ▶ Evaluate the gravity

- dermatological criteria (local secondary infection)
- hepatic complications (cutaneous porphyria, erythropoietic protoporphyria)
- haemolytic complications (Günther's disease)

#### ▶ Emergency investigations

- clinical dermatological examinations
- porphyrin blood, urine and faecal concentrations
- complete blood screen
- complete hepatic screen (cholestasis, cytolysis)
- iron screen
- Eliminate trigger factors: alcohol, oestrogens, porphyrinogenic medicaments (list on [www.porphyria-europe.com](http://www.porphyria-europe.com) and [www.drugs-porphyria.org](http://www.drugs-porphyria.org)), hepatitis (HBV, HCV), HIV, excess iron

### 2. Emergency therapeutic measures

**Porphyric photodermatoses do not always require hospitalisation when there are no hepatic and/or haemolytic complications.**

#### ▶ Cutaneous porphyrias (**Appendices: fig.1**):

- Avoid minor trauma and prolonged exposure to the sun
- Treat concurrent infections
- Check that triggers have been eliminated (alcohol, drugs...)

#### ▶ Günther's disease (**Appendices: fig.2**):

- Avoid minor traumas and prolonged exposure to the sun
- Rigorous cutaneous asepsis
- Transfusions of red blood cells and/or
- Splenectomy if there are haemolytic episodes.

#### ▶ Erythropoietic protoporphyria:

- Avoid prolonged exposure to the sun
- If there are signs of hepatic complications, transfer to intensive care
- Liver transplant is a last resource and is used in the rare cases of irreversible hepatic lesions.

## Drug interactions

- ▶ **Hepatic cutaneous porphyrias (cutaneous porphyria, hereditary coproporphyria, porphyria variegata)** are often triggered by alcohol or use of drugs that require hepatic induction of some P450 cytochromes (barbiturates, sulfamides, oestro-progestatives...).

- ▶ The **list of authorised, uncertain and contra-indicated drugs is available on** [www.porphyria-europe.com](http://www.porphyria-europe.com) and [www.drugs-porphyria.org](http://www.drugs-porphyria.org)
- ▶ **Günther's disease and erythropoietic protoporphyria** (erythropoietic and non-hepatic porphyria)
  - The list of porphyrinogenic drugs is not useful
  - Avoid photosensitising compounds

## Anaesthesia

- ▶ **Cutaneous hepatic vesicular porphyria (cutaneous porphyria, hereditary coproporphyria, porphyria variegata)**
  - **In a life threatening situation: propofol** should be used in combination **with an opiate**
  - **General anaesthesia:** the anaesthetist should **contact an specialised centre in all cases**
  - **Epidural / spinal nerve block: Bupivacain** is authorised
  - **Cutaneous surface anaesthesia: Bupivacain and local anaesthetic cream** are authorised
  - **Dental anaesthesia: Articain + epinephrine** is authorised **for healthy carriers or patients in long remission.** A urine sample should be taken from the first micturition the following day and sent to a specialised centre. However, all chronic cases (recurring acute crises) should be discussed with a doctor of a specialised centre.
- ▶ **Haemolysis (Günther's disease)**
  - No specific contra-indication
- ▶ **Erythropoietic protoporphyria**
  - It may be necessary to use **filters that block light emissions** of certain wavelengths using operating room lights to avoid serious lesions developing

## Additional therapeutic measures and hospitalisation

- ▶ Use 'total block' creams (factor 50+) to avoid aggravating or causing complications
- ▶ Increased hygiene of hands and exposed parts of the body (antisepsis)
- ▶ Management of infectious and/or inflammatory episodes
- ▶ Elimination of triggers (alcohol, medicaments...)
- ▶ The family must be advised and made aware of the situation during screening and given the contact details for an expert centre that the family should consult at least once

## Organ donation



- Despite there being little published data, **it is not advisable to propose organ donation** with these conditions because they involve the liver and erythropoietic tissue.

## Documentary resources

- ▶ Badminton MN, Elder GH. Management of acute and cutaneous porphyrias. *Int J Clin Pract.* 2002 May;54(4):272-8.
- ▶ [www.porphyria-europe.com](http://www.porphyria-europe.com) : site européen dédié aux malades porphyriques et aux médecins les prenant en charge. *Orphanet Letter* 2004.
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- ▶ Deybach JC, Puy H. Les porphyries héréditaires chez l'enfant. *Enc. Med. Chir.* Elsevier eds. 2007.
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- ▶ Sassa S. Modern diagnosis and management of the porphyrias. *Br J Haematol.* 2006 Nov;135(3):281-92.

## Appendices

Figure 1: Cutaneous porphyria Porphyrie



Figure 2: Günther's disease



*These guidelines have been prepared in collaboration with Professor Jean-Charles DEYBACH - Doctor Hervé PUY Centre de référence pour les porphyries - Centre français des porphyries - Service de Biochimie et Génétique moléculaire - Hôpital Louis Mourier – Colombes, and Doctor Gilles Bagou - Doctor Gaële Comte - SAMU-69 Lyon*

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