:: 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:
- Cutaneous porphyria, sporadic or familial;
- Porphyria variegata;
- Hereditary coproporphyria
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

---

**Menu**

<table>
<thead>
<tr>
<th>Pre-hospital emergency care recommendations</th>
<th>Recommendations for hospital emergency departments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Synonyms</td>
<td>Emergency situations</td>
</tr>
<tr>
<td>Aetiology</td>
<td>Drug interactions</td>
</tr>
<tr>
<td>Special risks in an emergency</td>
<td>Anaesthesia</td>
</tr>
<tr>
<td>Frequently used long term treatments</td>
<td>Additional therapeutic measures and hospitalisation</td>
</tr>
<tr>
<td>Complications</td>
<td>Organ donation</td>
</tr>
<tr>
<td>Specific medical care prior to hospitalisation</td>
<td>Documentary resources</td>
</tr>
<tr>
<td>For further information</td>
<td>Appendices</td>
</tr>
</tbody>
</table>
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
- www.porphyria-europe.com
- www.porphyrie.net
- 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 and 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-progentsatives...).
The list of authorised, uncertain and contra-indicated drugs is available on www.porphyria-europe.com and 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

- Deybach JC, Puy H. Porphyrías. Clinical Gastroenterology and Hepatology, Part 2. chapter 116 ; Fennerty, Hawkey,
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

These guidelines have been translated thanks to the financial support of Alexion.

Latest update: 10 July 2007