Organization of the follow-up of patients with porphyrias in Switzerland – coordination within the network for rare diseases and the Swiss Reference Center for porphyrias*

I. Patients with **acute hepatic porphyrias** (acute intermittent porphyria (AIP), variegate porphyria (VP), hereditary coproporphyria (HC)) or **cutaneous porphyrias** (erythropoetic protoporphyria (EPP), X-linked protoporphyria (XLPP), porphyria cutanea tarda (PCT)) **not requiring or declining a specific therapy:**

- Referral for specialist advice at initial diagnosis to the reference center for porphyrias (pRC)*
- Thereafter:
  - in stable patients referral every 2-5 years to the pRC by non-pRC#
  - in vulnerable patients or in patients with repeat acute attacks follow-up every 6-12 months to porphyria pRC

II. Patients with **acute hepatic porphyrias** (AIP, VP, HC) or **cutaneous porphyrias** (EPP, XLPP, PCT) **with the indication for a specific therapy:**

- Referral for specialist advice at initial diagnosis to the pRC
  exception: suspicion of PCT: share laboratory and clinical data with RC– individual discussion if specialist referral is recommended (e.g. confirmation of diagnosis required, complicated co-morbidities in the patient)
- In AIP, VP, HC with symptoms of an acute porphyria attack: confirm diagnosis of the acute attack (biochemistry in spot urine), contact pRC for the evaluation of a specific therapy
- Treatment/ follow-up:
  - For PCT (phlebotomies): treatment as needed in local care facilities, follow-up in pRC upon individual discussion as needed
  - For EPP (Scenesse®): treatment every 2 months in RC (according to the specifications of the EMA and EPNET)
  - For acute hepatic porphyrias (Givosiran®):
    - first year: treatment every month in pRC
    - in *stable patients*:
      After the second year: treatment in local care facilities, follow-up in pRC at least every 6 months
      if treatment is in local care facilities: documentation of porphyria attacks/ symptoms to RC as required for insurance coverage

III. **Ultra-rare porphyrias** (ALAD-deficiency porphyria (ADP), congenital erythropoietic porphyria (CEP), hepatoerythropoietic porphyria (HEP), homozygous or compound heterozygous acute porphyrias, including harderoporphyria, EPP-2, acquired EPP, dual porphyrias)

- Referral for specialist advice and assessment at initial diagnosis to pRC
- Treatment and follow-up in close collaboration with non-pRC according to individual patient needs as recommended by the pRC

For emergencies, further information, and contact details see:
https://www.stadtzuerich.ch/triemli/de/index/kliniken_institute/swissporphyriacentre.html

*Reference Center for Porphyrias (KOSEK): Triemli Spital, Zurich. Coordinator: Dr Anna Minder, +41 44 416 32 52

#Non-porphyria Reference centers: including other RC, associate center or care facility

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