Porphyria Overview
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Background

Porphyria is named from the ancient Greek word porphura, meaning purple.¹ Porphyrins are precursors of heme, a part of the hemoglobin molecule. Heme is manufactured in a multistep process. Defects of enzymes needed at various steps of heme synthesis result in distinct clinical syndromes known as porphyrias. These syndromes can be clinically classified into those predominantly involving the skin, those manifesting as disorders of the liver/nervous system, and a combination involving all 3 entities (see Image 1, or below).

Porphyrias can be inherited or (rarely) acquired.² With the exception of congenital erythropoietic porphyria (CEP), which is autosomal recessive, all other porphyrias are inherited as autosomal dominant disorders. They invariably result in accumulation and increased excretion of porphyrins and their precursors. Some porphyrias have acute presentations (acute intermittent, variegate, hereditary coproporphyria), whereas others have a chronic, relatively stable presentation (congenital, erythropoietic).³

King George III of England had symptoms of abdominal pain, rashes, reddish urine, and psychotic episodes that are
consistent with porphyria, although the account is disputed by many.\(^4\)
During the period 1955-1959, approximately 4000 people in southeast Anatolia (Turkey) developed porphyria due to the ingestion of hexachlorobenzene (HCB), a fungicide that was added to wheat seedlings.\(^5\)

**Pathophysiology**

Urine and stool studies in various types of porphyria are summarized in Image 2.

<table>
<thead>
<tr>
<th>Porphyria Type</th>
<th>Urine</th>
<th>Stool</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acute intermittent</td>
<td>ALA/PBG/ ?</td>
<td>ALA PBG/ ?</td>
</tr>
<tr>
<td>Variegate</td>
<td>ALA/PBG/Copro/ ?</td>
<td>Copro/ ?</td>
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<tr>
<td>Coproporphyrin</td>
<td>ALA/PBG/Copro/ ?</td>
<td>Copro/ ?</td>
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<tr>
<td>Doss porphyria</td>
<td>ALA</td>
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<tr>
<td>Cutanea Tarda</td>
<td>Uro</td>
<td>Iso Copro</td>
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<td>Congenital Enzymopathic</td>
<td>Uro/Copro</td>
<td>Copro</td>
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<tr>
<td>Enzymopathic</td>
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<td>Proto</td>
</tr>
</tbody>
</table>

**Porphyrins in stool and urine in different porphyrias.**

Heme synthesis is summarized in Image 3. In some porphyria patients and families, however, these diagnostic tools can reveal simultaneous findings that are compatible with 2 different forms of porphyria, a phenomenon referred to as dual porphyria.\(^6\)
Heme biosynthesis and the porphyrias.

Porphyrias present in 2 distinct syndromes, acute and chronic.

**Acute porphyrias**

The acute porphyrias are characterized by periodic acute attacks of neurovisceral symptoms and may stay occult for a long time. Four major disorders in this group are the Doss porphyria, acute intermittent porphyria, hereditary coproporphyria, and variegate porphyria. These porphyria syndromes are characterized by abdominal pain, neurologic deficits, psychiatric symptoms, and colored (red) urine.

**Chronic porphyrias**

The chronic porphyrias are dermatologic diseases that may or may not involve the liver and nervous system and do not present with acute attacks as described for the acute porphyrias above. These syndromes include congenital
erythropoietic porphyria, erythropoietic porphyria, and porphyria cutanea tarda.

**Clinical manifestations**

Clinical manifestations depend on the step in which the enzymatic defect occurs. If the enzymatic defects are in the initial steps of the metabolic cascade, early metabolic intermediates will accumulate (ie, aminolevulinic acid [ALA] and porphobilinogen [PBG]), which are responsible for attacks of neurologic dysfunction. If the enzymatic defects are in the final steps, sunlight-induced cutaneous lesions (photosensitivity) due to porphyrin accumulation in the skin will develop.7

**Doss porphyria / plumboporphyria**

Doss porphyria, also known as plumboporphyria (ALA dehydratase deficiency), is extremely rare.8 Abdominal pain and polyneuropathy are typical of this syndrome. Urinary ALA and coproporphyrin are markedly increased. Molecular genetic studies of the ALA dehydratase gene reveal the mutated nucleotides. In some patients, the development of the acute porphyria syndrome while the patient received pharmacologic doses of erythropoietin, which resolved when the drug was stopped, suggests that by stimulating heme synthesis, erythropoietin may unmask an enzyme deficiency resulting in the clinical expression of ALA dehydratase deficiency porphyria.9. Sometimes, exposure to lead may unmask occult plumboporphyria.10

**Acute intermittent porphyria**

Acute porphyria attacks are brought about by uncontrolled upregulation of the ALA synthase enzyme. This can be precipitated by certain lipophilic drugs (see the Drugs to Avoid section), hypoglycemia ("the glucose effect"),11 and a deficiency of heme, the end-product of the heme pathway (see Image 3) that acts as a negative feedback mechanism in normal circumstances.12

**Hereditary coproporphyria**

Hereditary coproporphyria results in most cases from half-normal activity (50%) of coproporphyrin oxidase.13 The disease is an acute hepatic porphyria that is characterized by abdominal pain, neuropsychiatric symptoms, and cutaneous photosensitivity14. In rare homozygous cases, enzyme activity decreases to <10% and the term harderoporphyria is used.15

**Variegate porphyria**

Variegate porphyria is an autosomal dominant inherited trait that results in decreased activity of protoporphyrinogen oxidase. It is characterized clinically by photosensitive skin disease and a propensity to acute neurovisceral crises. Variegate porphyria is found worldwide but has an exceptionally high frequency in South Africa.16

**Erythropoietic porphyria**

In erythropoietic porphyria, the protoporphyrin molecule accumulates and can be excited by absorbing light energy. This causes the generation of free radicals and, thereby, photosensitivity of all tissues exposed to light. In the dark, several other toxic mechanisms have been described: deposition of protoporphyrin crystals in hepatocytes and bile canaliculi, interference with redox systems and, recently, formation of cytotoxic bile. Clinical manifestations of erythropoietic porphyria are photosensitivity, insignificant hematologic abnormalities, and liver disease.17 The hepatic manifestations of the disease are diverse: mildly disturbed liver enzymes in 20% to fatal hepatic failure in less than 5%.18
Porphyria cutanea tarda

Porphyria cutanea tarda (PCT) is characterized by the defective uroporphyrinogen III decarboxylase enzyme. There are 3 types of porphyria cutanea tarda with typical skin manifestations; patients present with skin fragility, erosions, vesicles, bullae, and milia in sun-exposed areas of the skin. Sometimes, there is the presence of periorbital mottled hyperpigmentation and hypertrichosis, sclerodermoid changes, and ulceration.

Hemochromatosis gene (HFE) mutations and the hepatitis C virus (HCV) are both risk factors for PCT. In a French cohort of PCT patients with both C282Y and H63D were more frequent in PCT+ patients than in controls, but there was no difference in HFE genotype according to HCV seropositivity. Up to two thirds of Saxon patients with PCT carry the classic HFE mutations (C282Y and/or H63D). PCT is associated with antibodies to HCV. PCT is an important extrahepatic manifestation of HCV-infection. In a Swedish cohort, 38% of male patients with PCT had a history of alcohol abuse.

Congenital erythropoietic porphyria (Gunther’s disease)

Congenital erythropoietic porphyria, or Gunther’s disease, is one of the least common porphyrias. It results from a deficient activity of uroporphyrinogen III synthase (URO-synthase). Hemolysis may be a feature in homozygous cases. Prolonged exposure to sunlight may precipitate a blistering rash, red urine, and even blindness.

Prevalence

- The combined prevalence of the acute porphyrrias is approximately 5 cases per 100,000 persons.
- Porphyria cutanea tarda is the most common porphyria.
- The prevalence of erythropoietic porphyria in a study was estimated at 1.75 per 100,000 population.
- Congenital erythropoietic porphyria is extremely rare. Less than 200 cases have been reported in the literature.

History

Abdominal pain

The most common presenting symptom (90%) of an acute porphyria is abdominal pain. This is usually colicky in nature, located in the left lower abdomen, and lasts hours to days. The abdominal pain is rarely accompanied by fever, leukocytosis, or peritoneal signs. Nausea and vomiting appear common. There is a very characteristic discrepancy between the serious complaints and the actual clinical findings.

In a minority of patients, paresis may be the only presentation without abdominal pain. When a patient has repetitive visits to the emergency department because of severe abdominal pain without reasonable causes and needs narcotics for pain control, acute porphyria should be taken into consideration.

Muscle weakness and neurologic deficits

Muscle weakness and focal neurologic defictssuch as tetraparesis may be the presenting feature, especially in women of reproductive age. Limb pain is common.

The lifetime prevalence of acute intermittent porphryia-associated seizures has been reported as 2.2% of all those with known acute intermittent porphryia and 5.1% of all those with manifest acute intermittent porphryia. Epileptic seizures
among persons with acute intermittent porphyria are less common than has been previously described.\textsuperscript{36}

**Psychiatric symptoms**

Some patients develop psychiatric symptoms such as psychosis similar to schizophrenia. Diagnostic difficulty may lead to underdiagnosis of patients who present with strictly psychiatric symptoms. This assumption is supported by a high prevalence of acute intermittent porphyria in psychiatric hospitals.\textsuperscript{37}

Anxiety is raised in patients with acute intermittent porphyria and with variegate porphyria, in males and females, compared with the normal population.\textsuperscript{38}

**Other**

Acute intermittent porphyria should be suspected in individuals presenting with unexplained acute abdominal pain following international air travel.\textsuperscript{39}

The relative risk of an acute attack in acute intermittent porphyria compared with that in variegate porphyria was 14 in a series of 112 patients from South Africa with porphyria attack.\textsuperscript{40}

A thorough family history for porphyria and the patient's occupational history must be obtained.

**Physical Examination**

- In acute porphyrias presenting as abdominal pain, attention should be paid to peritoneal signs. The presence of localized tenderness, rebound tenderness, vaginal discharge, cervical motion tenderness, and/or genitourinary bleeding should raise "red flags" even in patients known to carry a diagnosis of porphyria, and alternative diagnoses should be sought.
- Jaundice may or may not be present.\textsuperscript{41}
- A focused neurologic exam should be performed to identify motor and sensory deficits and peripheral neuropathy.
- In some but not all acute porphyrias, a skin rash may be seen. Variegate porphyria and, much less commonly, hereditary coproporphyria can also cause chronic, blistering lesions on sun-exposed skin that are identical to those in porphyria cutanea tarda, a much more common condition.
- Gross, biochemical, and microscopic examination of the patient's urine is paramount if porphyria is on the differential diagnosis. The urine of patients with porphyria cutanea tarda is red to brown in natural light and pink to red in fluorescent light.\textsuperscript{42}

**Laboratory**

- **Urine**
  - Urine porphyrin studies are the mainstay in the diagnosis of acute porphyria attacks. Establish the diagnosis promptly by testing for increased porphobilinogen in a single-void urine. An expert guidelines panel recommended the Trace PBG Kit [Thermo Trace/DMA, Arlington, Tex].\textsuperscript{29}
  - Patients with acute exacerbations of porphyria have logarithmic increases (5-100 times) in metabolic precursors (ALA, PBG, etc). Minor elevations of these precursors are nondiagnostic and nonspecific.\textsuperscript{43}
Significantly increased ALA and PBG in urine have 100% specificity (i.e., rules in) for acute intermittent (hepatic) porphyria, variegate porphyria, and coproporphyria. A normal urine PBG result has a sensitivity of almost 100% (i.e., rules out) in the diagnosis of porphyria in acutely symptomatic patients.\textsuperscript{44}

- **Stool**
  - Stool coproporphyrin and protoporphyrin are the most commonly measured porphyrins in feces. The ratio of fecal coproporphyrin to fecal protoporphyrin varies among the porphyrias. For example, fecal protoporphyrin always exceeds coproporphyrin (P > C = V) in variegate porphyria, whereas the reverse is true in hereditary coproporphyria.\textsuperscript{45}

- **Erythrocyte uroporphyrinogen decarboxylase**
  - Erythrocyte uroporphyrinogen decarboxylase activity is a specific and intrinsic defect in porphyria cutanea tarda; measurement of this enzyme is a reliable diagnostic test for this disease.\textsuperscript{46}

- **Electrolytes**
  - Hyponatremia is typical.\textsuperscript{47} In 1966, lesions of the median eminence of the hypothalamus and both hypothalamic–hypophyseal tracts were described in a patient with acute intermittent porphyria and syndrome of inappropriate antidiuretic hormone (SIADH). It was suggested that SIADH occurred because of damage to these areas of the brain from excessive exposure to porphyrins.\textsuperscript{48}

- **C73R mutation**
  - Prenatal diagnosis is possible in some types of porphyria. For example, in congenital erythropoietic porphyria, pink fluorescence of the amniotic fluid examined fortuitously in sunlight is suggestive. DNA analysis may show the mutation C73R in the gene for URO-synthase.\textsuperscript{49}
  - A mutation screening for family members should be conducted to identify symptom-free carriers, especially in cases where there is a positive family history.\textsuperscript{34}

### Imaging

**Computed tomography (CT) scanning of the abdomen and pelvis**

CT scanning helps clinicians to rule out other diagnoses of excruciating abdominal pain, such as a ruptured viscus or vessel, and may help to pick up concomitant pathology, such as intussusception or infarction.\textsuperscript{50} Focal, fatty nodularity of the liver may be noted in some patients.\textsuperscript{51}

**Magnetic resonance imaging (MRI)**

MRI of acute intermittent porphyria demonstrates multiple large, contrast-enhancing, subcortical white matter lesions, which regress with glucose and hematin infusions. Diffusion-weighted MRI is normal, and MR spectroscopy excludes acute demyelination or tissue necrosis. MRI findings of acute intermittent porphyria can differ from those in posterior reversible encephalopathy syndrome by virtue of intense contrast enhancement. Because diffusion-weighted MRI and MR spectroscopy are normal, the lesions are likely caused by reversible vasogenic edema and transient breakdown of the blood-brain barrier.\textsuperscript{52}

T2-weighted MRI sequences demonstrated multiple white-matter, high-signal lesions in 4 of 16 acute intermittent porphyria gene carriers (25%).\textsuperscript{53} Kupferschmidt and colleagues\textsuperscript{54} described 2 patients with acute intermittent porphyria who presented with cortical blindness. MRI showed bilateral occipital lesions, and the investigators speculated that these lesions were caused by vasospasm-induced ischemia due to unopposed cerebral vasoconstriction that resulted from a deficiency of nitrous oxide synthase, a major vascular dilator.
The striking feature of the MRI findings in these cases and in those of acute intermittent porphyria described in the literature is that the lesions are bioccipital and partially or totally reversible. These characteristics are typical of MRI findings seen in patients with hypertensive encephalopathy, and many patients presenting with acute intermittent porphyria attacks have high blood pressure on presentation.\(^\text{55}\)

In cases of porphyria cutanea tarda, MRI of the liver shows poorly defined areas, which, on T2-weighted sequences, exhibit a hypersignal with fat saturation. Treatment of porphyria cutanea tarda may lead to clinical remission and resolution of radiologic abnormalities.\(^\text{56}\)

### Treatment

- Withdrawal of any culprit medications, alcohol, drugs, toxins, and chemicals is the key to therapy.
- Supportive care such as fluid, electrolytes, and nutrition is paramount.
- Monitor for hyponatremia or hypomagnesemia, and treat vigorously if found.
- Aggressively treat respiratory failure, which may ensue once muscle weakness involves the diaphragm, and ventilate in an intensive care unit as appropriate.
- Monitor patients on telemetry for prompt identification and treatment of arrhythmias, which are a common occurrence.
- Treat pain with parenteral meperidine or morphine. Complicated and debilitating chronic cases may require celiac plexus injection.\(^\text{57}\)
- Administer phenothiazines for nausea, vomiting, agitation, etc.
- Treat tachycardia and/or hypertension with propranolol or nadolol, which can be safely used for beta blockade.
- Promptly start glucose infusion in the form of 10% dextrose. At least 300-400 g should be given in 24 hours. The infusion is a time-buying measure to bridge the patient to more definitive treatment with hemin; by itself, glucose infusion is only effective for mild symptoms.\(^\text{58}\)
- Treat acute porphyria attacks with hemin (intravenous heme); 3-4 mg/kg/d for 3-5 days is the definitive treatment and mainstay of management. Thrombophlebitis is the major adverse effect.
  - At least two thirds of the patients have a good response, with resolution of pain and neurologic deficits.\(^\text{59,60,61}\) Tin protoporphyrin appears to have a synergistic effect with hemin.\(^\text{52}\)
  - Recombinant PBG deaminase has been tested in phase I studies of acute hepatic porphyria and found to be effective and promising.\(^\text{63}\) Further data are awaited.
  - Hemodialysis has been used in dire circumstances with some benefit when hemin was not available.\(^\text{64}\)
  - Termination of pregnancy may have to be considered in acute fulminant attacks presenting during pregnancy.\(^\text{65}\)
  - Gonadotropin-releasing hormone (GdRH) analogues have been reportedly effective in some cases of acute intermittent porphyria, but these agents are not widely used.\(^\text{66}\)
- Avoidance of sunlight is the key in treating cutaneous porphyrias.\(^\text{67}\)
- Other
  - Magnesium sulfate has been used to control seizures, as many regular anti-seizure medications are contraindicated.\(^\text{68}\)
  - Neither red blood cell exchange transfusion nor plasmapheresis prevented progressive hepatic deterioration in 2 cases of advanced hepatic erythropoietic protoporphyria despite a significant decrease in protoporphyrin levels.\(^\text{69}\)
  - Hematopoietic stem cell transplantation (HSCT) has been applied with success in severe cases of congenital erythropoietic porphyria.\(^\text{70,71}\)
  - Liver transplantation is an option in cases in which cirrhosis complicates hepatic porphyria.
The use of beta-carotene has shown some benefit for cutaneous porphyrias.

Porphyria cutanea tarda can be effectively treated by phlebotomy. High-dose chloroquine therapy for porphyria cutanea tarda is rarely used now because of its hepatic side-effects.

**Prognosis**

Among 206 adult Finnish patients with acute intermittent porphyria or variegate porphyria, 47 patients had a total of 117 acute attacks during the period of 1967-1989. Six of these patients died during an attack, and 21 attacks were associated with paresis; the frequency of severe attacks was significantly smaller than before 1967. Most pareses and deaths occurred because of a delay in diagnosis and inappropriate treatment of porphyria. Milder symptoms of porphyria were more common among those who had had previous attacks than among those who had not.

In cases of acute intermittent porphyria, the risk of attacks correlated with the excretion of PBG in the urine during remission among adults; a low rate of excretion predicted freedom from acute attacks. Two percent of the surgical operations and 4% of the pregnancies were associated with acute attacks. Nearly one third of the women had symptoms of porphyria associated with the menstrual cycle, but these seldom proceeded to an acute attack. Forty-six percent of the women had used sex-hormone preparations regularly; 2 (4.5%) of the women experienced associated acute attacks. Patients with acute intermittent porphyria or variegate porphyria showed increased incidences of hepatocellular carcinoma.

Erythropoietic porphyria is a persistent, severely painful, socially disabling disease with a marked impact on quality of life.

**Prevention**

- Educate patients about their porphyria disease, inheritance, precipitating drugs and events, and the importance of seeking treatment in early stages of an attack.
- Encourage patients to wear medical alert bracelets.
- Perioperative management includes the use of filters on operative lights to prevent skin burns and intestinal perforation. To prevent burn injuries, astral lamps in the operating room are covered with yellow film filters.

**Expertise, Testing, and Centers of Excellence**

A listing of experts, testing, and centers of excellence in the porphyrias is available at the American Porphyria Foundation Website (www.porphyriafoundation.com).

The European Porphyria Initiative (EPI) (www.porphyria-europe.org) network was formed in 2001 to compare the experience among countries in an attempt to develop a common approach to the management of the porphyrias, particularly concerning the recommendations of safe medications and warnings against unsafe drugs, and to facilitate international collaborative clinical and biologic research. The main achievements of the EPI during this period have been: the drafting of and agreement to consensus protocols for the diagnosis and management of acute hepatic porphyrias, as well as the creation of a multilingual Website, particularly focusing on guidelines for common prescribing problems in acute porphyrias and providing information to patients that is now available in more than a dozen languages.
Diet and Nutrition

Smoking, which increases hepatic cytochrome P450 enzymes and presumably heme synthesis, is associated with more frequent porphyria attacks.79

Drugs to Avoid

The list of drugs to avoid continues to grow. Major culprits include barbiturates, anticonvulsants, progestins, and rifampin.

Individual medications can be checked against a safe and unsafe drug database that is maintained by the American Porphyria Foundation.

Acute intermittent porphyrias are rare complications of ovulation induction with clomiphene citrate, but these syndromes should be considered in patients who develop unexplained hyponatremia or neurovisceral symptoms.80

Different drugs in the same class may have different effects in the porphyrias. For example, although lidocaine should be avoided, dental treatment using bupivacaine or levobupivacaine as local analgesic agents was successfully and safely provided for 5 children with a diagnosis of latent acute intermittent porphyria or who had a family history of acute intermittent porphyria.81

Differential Diagnosis

- Acute abdomen
- Hereditary tyrosinemia type I82
- Lead poisoning
- Pseudoporphyria is a bullous photosensitivity, the commonest etiology being secondary to various ingested medications, such as voriconazole (a relatively new second-generation triazole antifungal agent)83
- Psychosis

References


Keywords

porphyria overview, porphyria, hepatic porphyria, erythropoietic porphyria, acute porphyria, acute intermittent porphyria, cutaneous porphyria, porphyria cutanea tarda, hepatoerythropoietic porphyria, variegate porphyria, Doss porphyria, plumboporphyria, hereditary coproporphyria, congenital erythropoietic porphyria, Gunther’s disease, Gunther disease, EPP, PCT, AIP, CEP

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Further Reading

Related eMedicine Topics

- Congenital Erythropoietic Porphyria [in the Dermatology section]
- Diseases of Tetrapyrole Metabolism - Refsum Disease and the Hepatic Porphyrias [in the Neurology section]
- Erythropoietic Protoporphyria [in the Dermatology section]
- Porphyria, Acute [in the Pediatrics: General Medicine section]
- Porphyria, Acute Intermittent [in the Hematology section]
- Porphyria, Chester
- Porphyria Cutanea Tarda [in the Dermatology section]
- Porphyria, Cutaneous [in the Pediatrics: General Medicine section]
- Porphyria, Hereditary Coproporphyria
- Pseudoporphyria [in the Dermatology section]
- Variegate Porphyria [in the Dermatology section]

Clinical Trials

- Does Exercise and Heat Increase the Lightsensitivity in Patients With Erythropoietic Protoporphyria
- Phase III Confirmatory Study in Erythropoietic Protoporphyria (EPP)
- Pilot Trial of Deferasirox in the Treatment of Porphyria Cutanea Tarda
- Studies in Porphyria I: Characterization of Enzyme Defects

Additional Resources


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