
Secret Getaway


بسم الله الرحمن الرحيم

PORPHYRIAS

BY

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Porphyrias

Definition:

Porphyrias are caused by inherited (or occasionally acquired) defects in heme synthesis, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors.

Classification:

Depending on whether the enzyme deficiency occurs in red blood cells or the liver.

The porphyrias are classified into:

A. Erythropoietic porphyrias.

B. Hepatic porphyrias.

Note:

With the exception of congenital erythropoietic porphyria, which is a genetically recessive disease, all the porphyrias are inherited as autosomal dominant disorders.

CLINICAL MANIFESTATIONS

- **Abdominal pain and neuropsychiatric disturbances. Due to accumulation of ALA and porphobilinogen, such as (acute intermittent porphyria).**
- **Individuals show photosensitivity (skin itches and burns)when exposed to visible light due to accumulation of tetrapyrrole intermediates.**

Note:

- These symptoms are thought to be due to the porphyrin-mediated formation of superoxide radicals from oxygen.
- These reactive oxygen species can oxidatively damage membranes and cause the release of destructive enzymes from lysosomes.
- Destruction of cellular components leads to the photosensitivity.

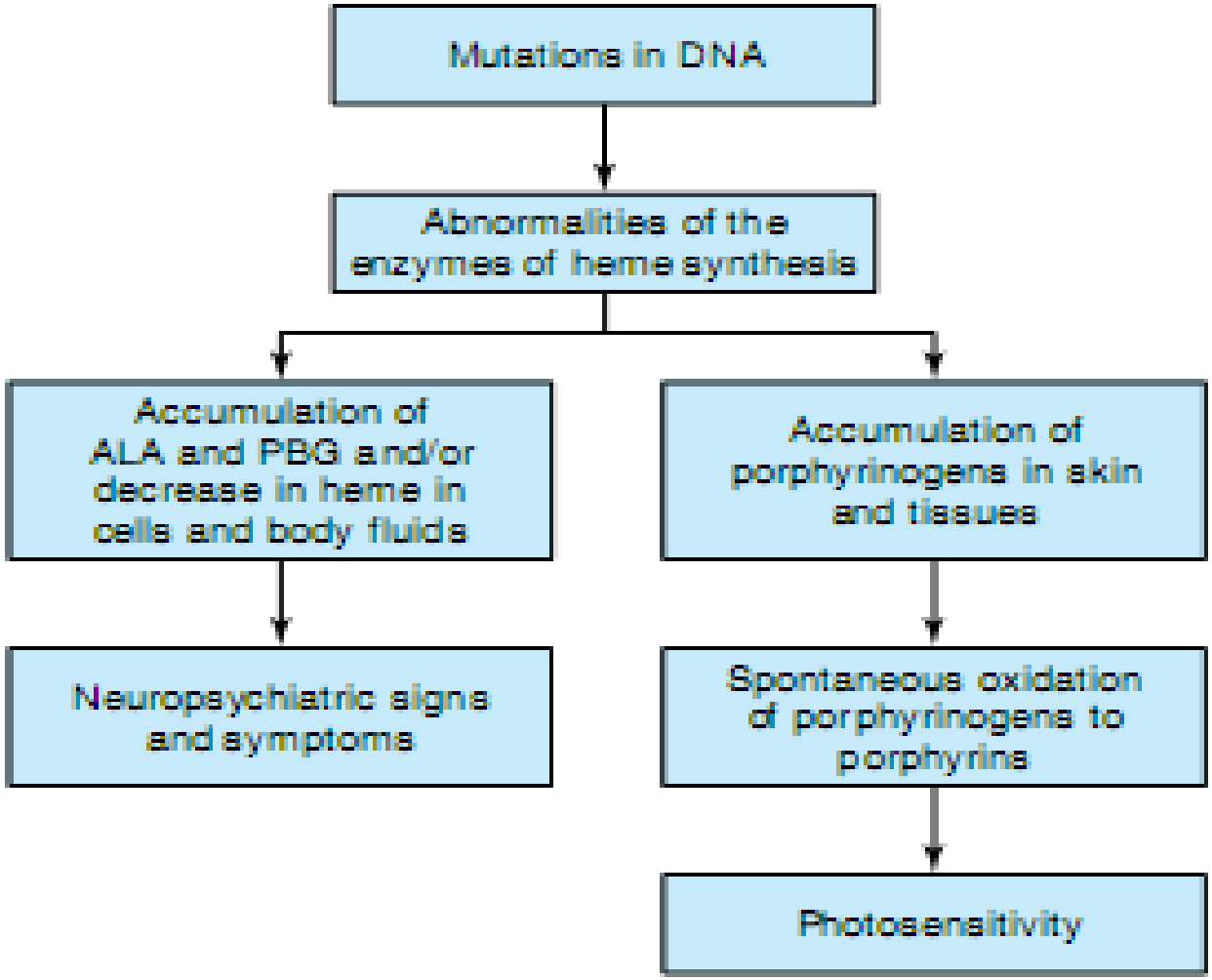


Figure 32–11. Biochemical causes of the major signs and symptoms of the porphyrias.

TYPE OF PORPHYRIA

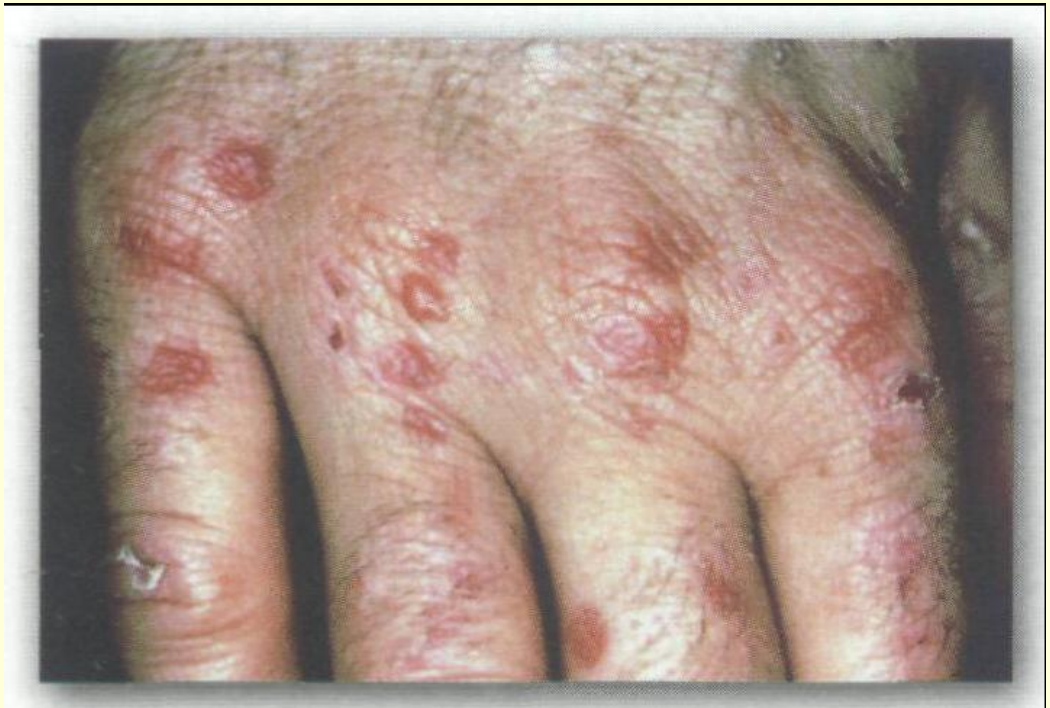
I. Chronic porphyria:(Porphyria cutanea tarda):

- **It is the most common porphyria.**
- **It is a chronic disease of the liver and erythroid tissues.**
- **It is associated with a deficiency in *uroporphyrinogen* decarboxylase.**
- Clinical appearance is influenced by various factors: as hepatic iron overload, exposure to sunlight, and presence of hepatitis B or C, or HIV infections.

Symptoms:

- Porphyrin accumulation leads to cutaneous symptoms, and urine that is red to brown in natural light, and pink to red in fluorescent light.

Skin eruptions in a patient with porphyria cutanea tarda



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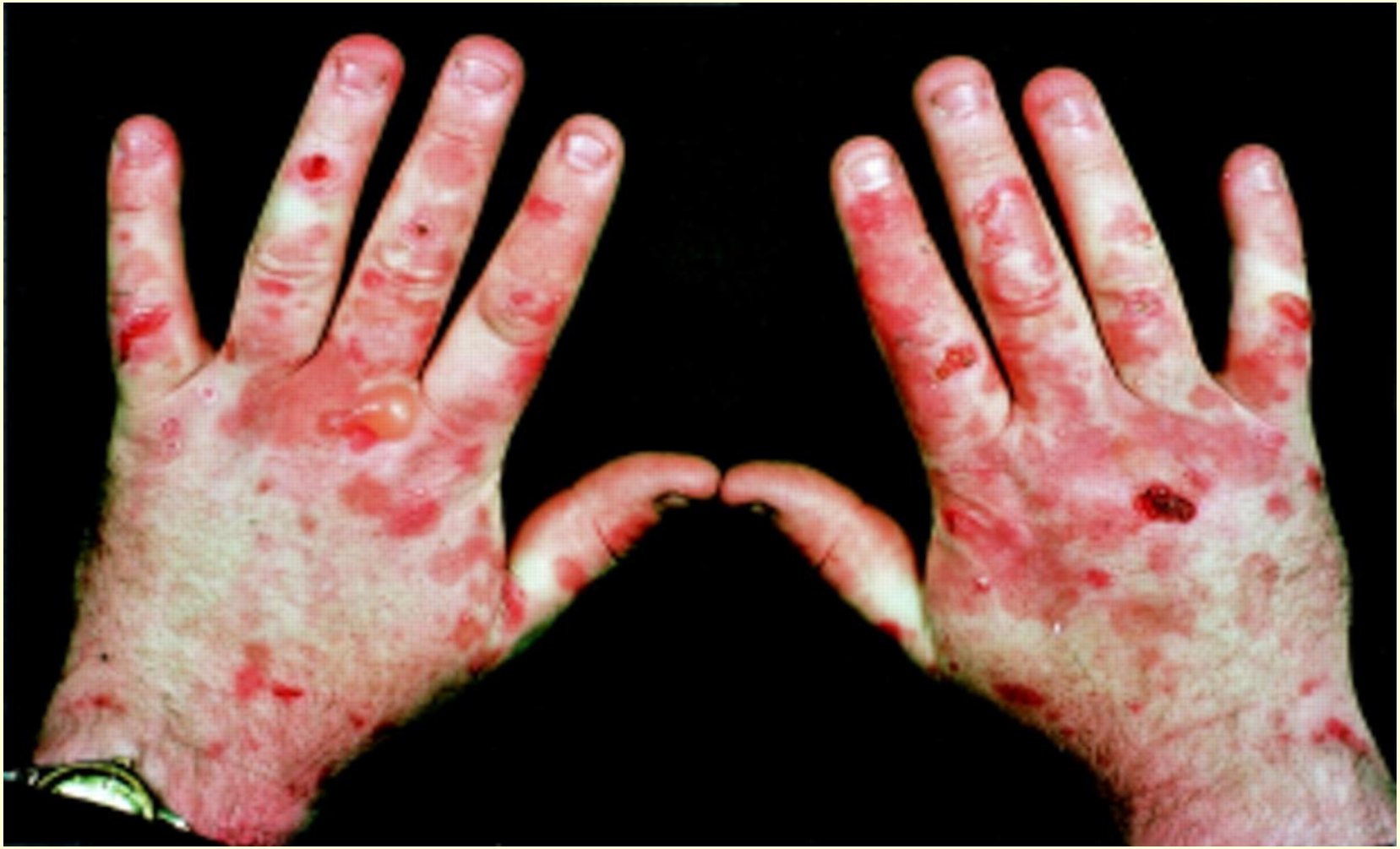

II. Acute hepatic porphyrias: (acute intermittent porphyria, hereditary coproporphyria, and varigate porphyria).

Symptoms:

- Acute attacks of gastrointestinal, neurologic/psychiatric symptoms, and cardiovascular symptoms.
- Abdominal pain and neuropsychiatric disturbances. Due to accumulation of ALA and porphobilinogen, as acute intermittent porphyria.

- Symptoms of (acute hepatic porphyrias) are precipitated by administration of drugs as barbiturates and ethanol, induce synthesis of heme-containing cytochrome P450 microsomal drug oxidation system. This further decreases the amount of available heme, which, in turn, promotes the increased synthesis of *ALA synthase.*

III. Erythropoietic porphyrias:

(congenital erythropoietic porphyria and erythropoietic protoporphyria).

Symptoms:

- **It is characterized by skin rashes and blisters appear in early childhood.**
- **The diseases are complicated by cholestatic liver cirrhosis and progressive hepatic failure.**

TREATMENT OF PORPHYRIAS

- The severity of symptoms of the porphyrias can be diminished by:

A. Intravenous injection of hemin, which acts to decrease the synthesis of ALA synthase.

B. Avoidance of sunlight.

C. Ingestion of β-carotene (a free-radical scavenger).


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