PORPHYRIAS
AT A GLANCE

Porphyria is not a single disease but a group of eight inherited genetic disorders that differ considerably from each other. Most commonly they are divided into the "acute "and "cutaneous" porphyrias, depending on the primary symptoms. A common feature in all Porphyrias is the accumulation in the body of porphyrins or porphyrin precursors. These body chemicals do not normally accumulate. Precisely which of these chemicals builds up depends on the type of Porphyria.

ACUTE PORPHYRIAS

There are four types of acute porphyria: Acute Intermittent Porphyria (AIP), Hereditary Coproporphyria (HCP), Variegate Porphyria (VP), and δ-Aminolevulinic Acid Dehydratase (ALAD) Deficiency Porphyria (ADP), which are characterized by episodes of debilitating attacks.

Signs and Symptoms

- » Severe Abdominal pain
- » Vomiting, nausea, constipation
- » Pain in the back, arms and legs
- » Muscle weakness
- » Urinary retention
- » Tachycardia
- » Confusion, hallucinations, seizures
- » Insomnia

Acute Intermittent Porphyria (AIP)

Acute Intermittent Porphyria (AIP) is characterized by deficiency of the enzyme hydroxymethylbilane synthase (HMBS), also known as porphobilinogen deaminase (PBGD). Additional factors such as endocrine influences (e.g. hormonal changes), the use of certain drugs, excess alcohol consumption, infections, and fasting or dietary changes are required to trigger the appearance of symptoms.

Hereditary Coproporphyria (HCP)

Hereditary Coproporphyria (HCP) is a rare metabolic disorder characterized by deficiency of the enzyme coproporphyrinogen oxidase (CPOX). This enzyme deficiency results in the accumulation of toxic porphyrin precursors in the body. Some affected individuals experience acute attacks or episodes that generally develop and become more severe over a period of days. In some cases, affected individuals may develop skin (cutaneous) lesions affecting the sun-exposed areas of skin such as the hands and face.

Variegate Porphyria (VP)

Variegate Porphyria is a rare genetic metabolic disorder characterized by deficient function of the enzyme protoporphyrinogen oxidase (PPO or PPOX). Some affected individuals present with skin symptoms, some with neurological symptoms and some with both. Blistering and fragility of sun-exposed skin are the most common skin (cutaneous) symptoms.

ALAD-Deficiency Porphyria (ADP)

ALAD Deficiency Porphyria (ADP) is rare and is characterized by almost complete deficiency of the enzyme delta-aminolevulinic acid (ALA) dehydratase. ADP is more severe than the other acute Porphyrias and can present in childhood. Only ~10 cases have been reported worldwide.

LEARN MORE:

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CUTANEOUS PORPHYRIAS

There are four types of cutaneous porphyria: Porphyria Cutanea Tarda (PCT), Erythropoietic Protoporphyria (EPP), and X-Linked Protoporphyria (XLP), Congenital Erythropoietic Porphyria (CEP), and Hepatoerythropoietic Porphyria (HEP), which are characterized by blistering and scarring of the skin, pain, and/or redness and swelling on sun-exposed areas.

Porphyria Cutanea Tarda (PCT)

Porphyria Cutanea Tarda (PCT) is the most common of the Porphyrias and results from a deficiency of the enzyme uroporphyrinogen decarboxylase (UROD).

Subdivisions of Porphyria Cutanea Tarda

- » Sporadic or acquired Porphyria Cutanea Tarda (80-90%)
- » Familial Porphyria Cutanea Tarda or f-PCT (10-20%)

Signs and Symptoms

- » Skin abnormalities characterize this disorder
- » Susceptibility to damage of the skin from photosensitivity
- » Extremely fragile skin
- » Abnormal, excessive hair growth
- » Liver abnormalities

PCT and Hepatitis C Virus

PCT is frequently associated with Hepatitis C Virus (HCV) infection.

Erythropoietic Protoporphyria (EPP) and X-Linked Protoporphyria (XLP)

Erythropoietic Protoporphyria (EPP) is a rare inherited metabolic disorder characterized by a deficiency of the enzyme ferrochelatase (FECH). When EPP is due to an ALAS2 mutation it is termed X-linked protoporphyria (XLP), because that gene is found on the X chromosome.

Signs and Symptoms

- » Photosensitivity, swelling, burning, itching, redness of the skin after exposure to sunlight

Congenital Erythropoietic Porphyria (CEP)

Congenital Erythropoietic Porphyria (CEP) is a very rare inherited metabolic disorder resulting from the deficient function of the enzyme uroporphyrinogen III cosynthase (UROS), the fourth enzyme in the heme biosynthetic pathway.

Signs and Symptoms

- » Reddish color of the urine
- » Skin photosensitivity
- » Increased hair growth
- » Loss of facial features and fingers by phototoxic damage & infection
- » Anemia

Hepatoerythropoietic Porphyria (HEP)

Hepatoerythropoietic Porphyria (HEP) is a deficiency of the enzyme uroporphyrinogen decarboxylase; it is the autosomal recessive form of familial Porphyria Cutanea Tarda (f-PCT).

Signs and Symptoms

- » Severe cutaneous Photosensitivity
- » Erythema
- » Hypertrichosis
- » Anemia
- » Hepatosplenomegaly