AMERICAN PORPHYRIA FOUNDATION

2020 MEDIA KIT

porphyriafoundation.org

PORPHYRIA

Porphyria is not a single disease but a group of eight inherited genetic disorders that differ considerably from each other. A common feature in all Porphyrias is the accumulation in the body of porphyrins or porphyrin precursors. Although these are normal body chemicals, they normally do not accumulate. Precisely which of these chemicals builds up depends on the type of Porphyria.

The terms porphyrin and porphyria are derived from the Greek word porphyrus, meaning purple. Urine from some Porphyria patients may be reddish-purple in color due to the presence of excess porphyrins and related substances in the urine, and the urine may darken after exposure to light.

CAUSE

> Porphyria arises as a result of a malfunction in one of the eight steps in the body's synthesis of a complex molecule called heme. Heme is essential for the transport of oxygen to cells in the body. If any step in the synthesis of heme is blocked, an intermediate chemical accumulates in the cell, resulting in oxygen depletion. Those intermediate chemicals, known as porphyrins or porphyrin precursors, are the substances of which heme is composed. Each type of Porphyria represents a deficiency or a specific enzyme needed for the synthesis of heme.

TYPES OF PORPHYRIA

ACUTE

Acute Intermittent Porphyria
Hereditary Coproporphyria
Variegate Porphyria
ALA-D Porphyria

Acute porphyria attacks generally evolve and become more severe over several days, especially the abdominal pain; two of these, Variegate Porphyria and Hereditary Coproporphyria, may also have skin symptoms of blistering after sun exposure.

CUTANEOUS

Porphyria Cutanea Tarda
Erythropoietic Protoporphyria
X-Linked Protoporphyria
Congenital Erythropoietic Porphyria

Cutaneous Porphyrias present with blistering and scarring of the skin, pain, and/or redness and swelling in sun-exposed areas.

ABOUT THE APF

Supporting the Porphyria community since 1983, the APF's mission is to improve the health and well-being of all individuals and families impacted by Porphyria. The APF maintains a focus on education, advocacy, support services and research for the prevention, treatment and cure of the Porphyrias. Additionally, the APF serves patient advocates to public, private, and government agencies interested in funding research and educational programs. Its Scientific Advisory Board is made up of the world's foremost experts in Porphyria management, diagnosis, and research.

SCIENTIFIC ADVISORY BOARD

The Scientific Advisory Board is made up of the world's foremost experts in Porphyria management, diagnosis, and research. They have written or approved the medical information on the APF's website. Many of the APF's SAB members have over 40 years experience working on Porphyria, from conducting cutting-edge research, and writing peer-reviewed article for major medical journals to authoring the chapters on Porphyria in medical school textbooks. Doctors worldwide consult with these Porphyria specialists for help with diagnosis and treatment of their patients.

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PROTECT THE FUTURE

The Protect the Future campaign was established to attract and train the next generation of doctors and specialists in the field of Porphyria. Over the next decade, ninety percent of our valued Porphyria experts will retire. These men and women have led Porphyria research, testing and treatment for the past 30 years. Without financial support, there is a real risk of losing knowledge of the disease, quality testing, diagnosis, and treatment, and ultimately a cure.

Protect the Future supports young doctors who work and study with long-time experts, seeing patients and doing research, in order to gain the expertise they will need to care for the U.S. Porphyria patient population for decades to come.

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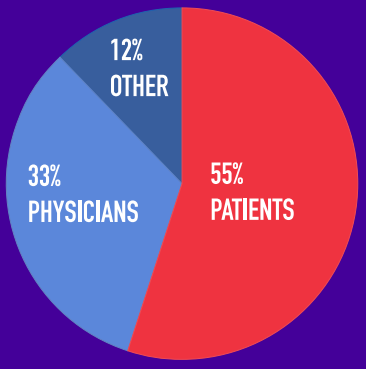
AMERICAN PORPHYRIA FOUNDATION

MEMBERSHIP

Membership Database over 12,000



MISSION

The APF is dedicated to improving the health and well-being of individuals and families affected by Porphyria.

ABOUT PORPHYRIA

Porphyria is a group of eight rare inherited genetic disorders that differ considerably from each other, A common feature in all Porphyrias is the accumulation in the body of porphyrins or porphyrin precursors. Although these are normal body chemicals, they normally do not accumulate. Precisely which of these chemicals builds up depends on the type of Porphyria.The terms porphyrin and porphyria are derived from the Greek word porphyrus, meaning purple.

HOW CAN THE APF HELP?

- SUPPORT & ASSISTANCE PROGRAMS
- PHYSICIAN EDUCATION PROGRAMS
- PATIENT EDUCATION PROGRAMS
- RESEARCH EFFORTS
- INTERNATIONAL EFFORTS
- ADVOCACY EFFORTS

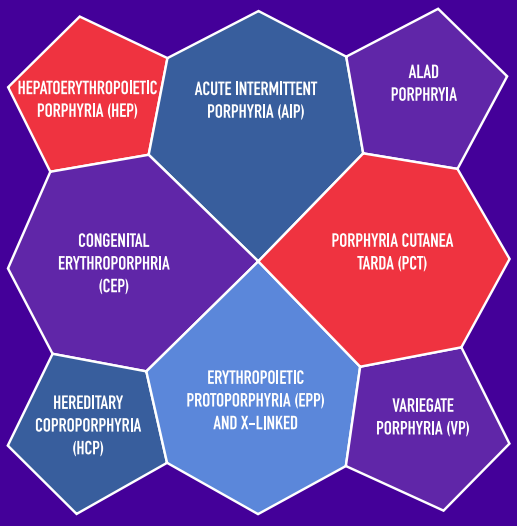
REMEMBER ... RESEARCH IS THE KEY TO YOUR CURE!

People with porphyria experience:

PAIN, ISOLATION, AVOIDANCE OF TRIGGERS, LACK OF TREATMENT

PATIENT & DOCTOR DATABASE

Make sure your contact information is up to date in the APF database to receive the latest news and educational materials.



PORPHYRIA CENTERS AND SATELLITE LOCATIONS

FACTS

7,000 known rare diseases

93% have no treatment

80% are genetic

#AskMeAboutPorphyria

PORPHYRIA AWARENESS WEEK

18-25 April, 2020

DONATE TODAY

WWW.PORPHYRIAFOUNDATION.ORG | 866-APF-3635 | 4915 St. Elmo Avenue, Suite 200 Bethesda, Maryland 20814

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). Skin photosensitivity results in severe blistering and scarring, often with mutilation and loss of facial features and fingers. Increased hair growth (hypertrichosis) on sun-exposed skin, brownish-colored teeth (erythrodontia), and reddish-colored urine are common.

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.

CUTANEOUS PORPHYRIAS

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 through phototoxic damage & infection
- Anemia

Hepatoerythropoietic Porphyria (HEP)

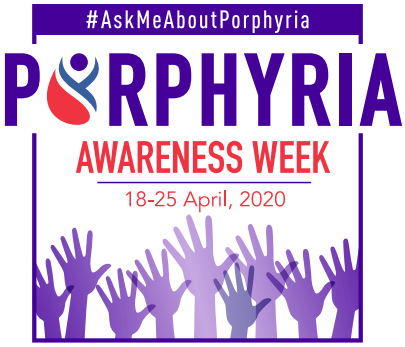
Hepatoerythropoietic Porphyria (HEP) is a deficiency of the enzyme uroporphyrinogen decarboxylase; it is the autosomal recessive form of familial Porphyria Cutanea Tarda (f-PCT).

Signs and Symptoms

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

LEARN MORE:

WWW.PORPHYRIAFOUNDATION.ORG | 866-APF-3635



FOR IMMEDIATE RELEASE: 04/10/2020
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"ASK ME ABOUT PORPHYRIA" CAMPAIGN BRINGS ATTENTION TO RARE, LIFE-THREATENING DISEASE

American Porphyria Foundation to Host Porphyria Awareness Week April 18-25, 2020

(BETHESDA, MD) – Porphyria affects fewer than 200,000 Americans. Due to the wide array of symptoms and the rarity of the condition, porphyria is often misdiagnosed and misunderstood by the medical community. The American Porphyria Foundation is working to change that.

The non-profit porphyria research and advocacy organization is hosting Porphyria Awareness Week April 18-25 to spread the word about Porphyria.

"Porphyria Awareness Week has grown substantially in previous years and now represents a push for global awareness of porphyria research efforts and treatment," says APF Executive Director, Kristen Wheeden.

Porphyria is not a single disease but a group of eight inherited genetic disorders that differ considerably from each other. A common feature in all the Porphyrias is the accumulation in the body of porphyrins or porphyrin precursors. These are normal body chemicals, but they normally do not accumulate. Precisely which of these chemicals builds up depends on the type of Porphyria.

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Throughout Porphyria Awareness Week, the APF will continue to educate others on porphyria by collecting and distributing awareness information from patients and the APF on Twitter, Instagram and Facebook. Visit the Porphyria Awareness Week 2020 landing page for more information, listing of special events, and a media kit.

For more information on Porphyria, Porphyria Awareness Week and the American Porphyria Foundation, visit https://porphyriafoundation.org.

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For the all the latest in porphyria, visit porphyriafoundation.org/news

porphyriafoundation.org