



Canadian Association for Porphyria



CANADIAN ASSOCIATION FOR PORPHYRIA INC.

***Our Mission: "To Improve the Quality of
Life for People Affected by Porphyria"***





What Is Porphyria?

- Not a single disease
- A group of at least 8 disorders
- Condition in which excessive amounts of porphyrins form in the body
- Porphyrins are normal body chemicals
- Accumulation causes porphyria





History Of Organization

- CAP was established in 1988
- Cofounders were Lois Aitken and Dr. David Chapman
- The Charity's opening balance was ~\$350
- Budget increased to \$50,000 after 14 years
- Current budget is \$114,000





Inheritance

- Many forms of Porphyria are inherited
- Passed on from parents to children
- Can be DOMINANT or RECESSIVE
- Parent may or may not show signs and symptoms of the condition
- Your doctor should know about Porphyria in family history

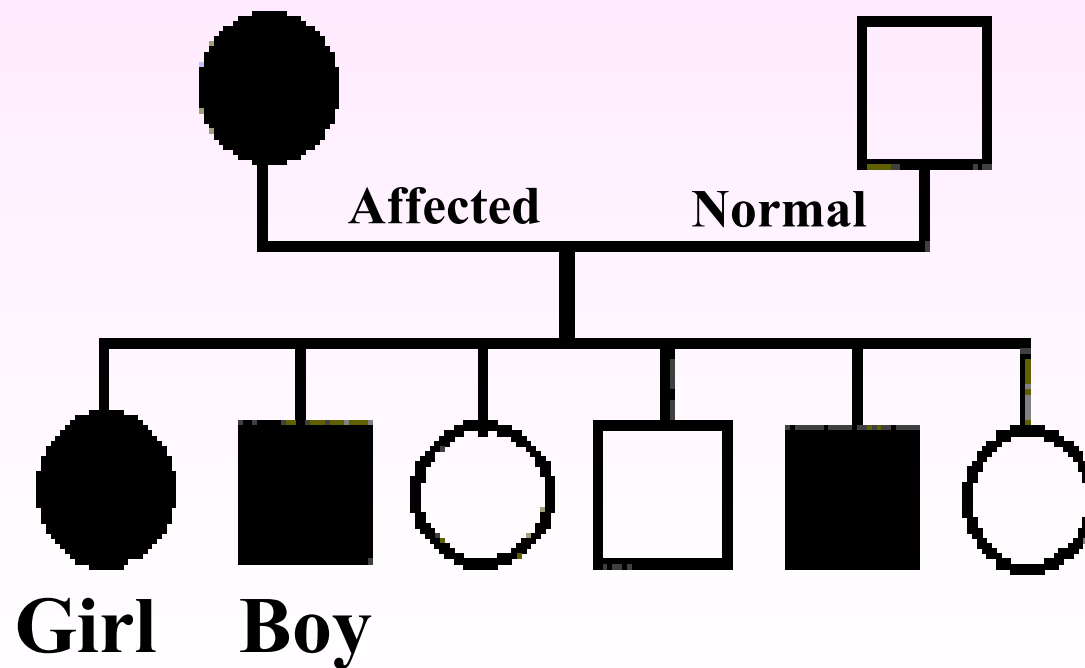




Dominant

Mother

Father



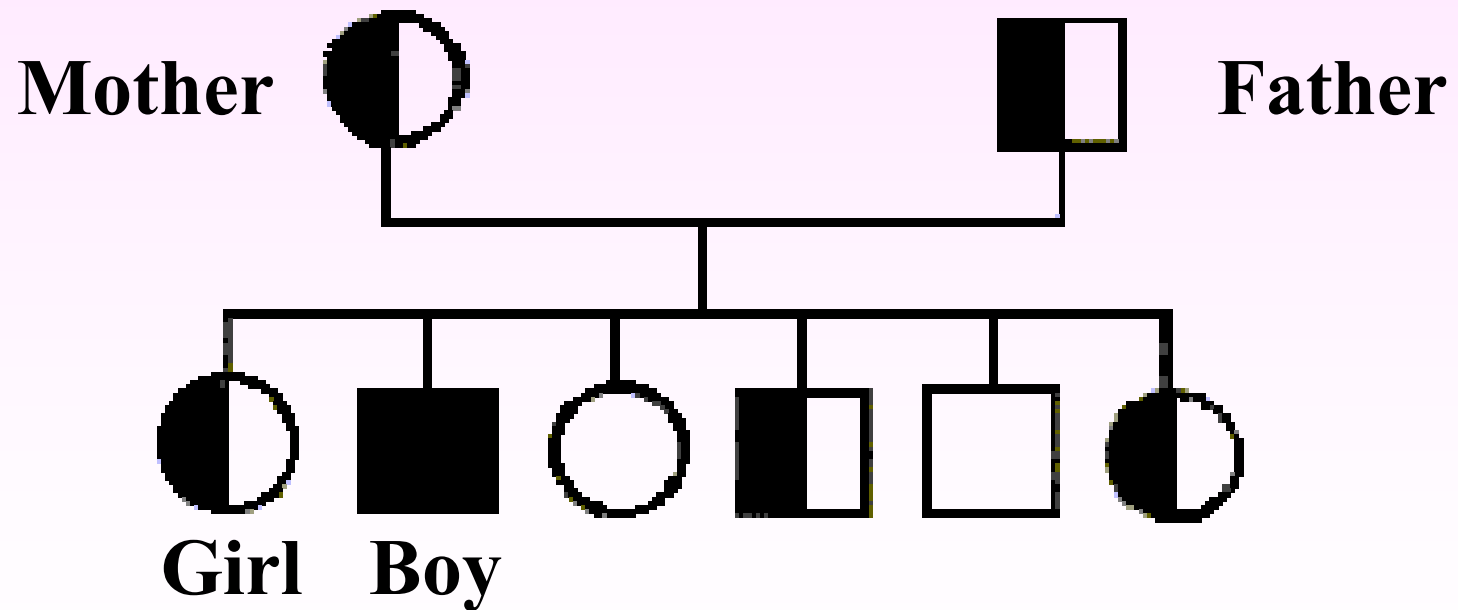
Affected (1:2 Chance)





Recessive

Both Carriers But Appear Normal



Affected (1:4 Chance)





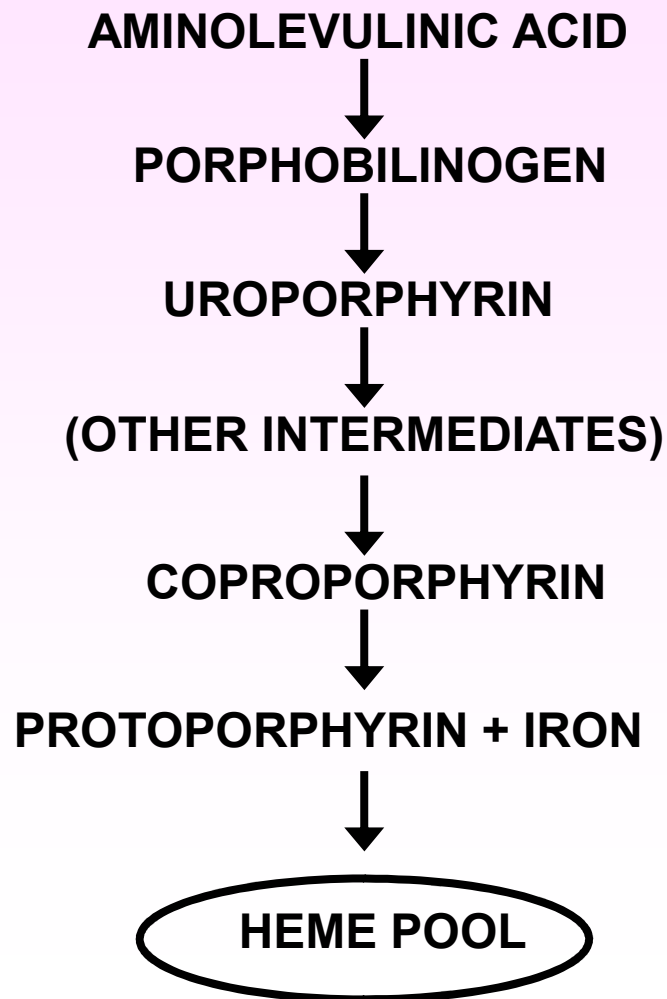
Explanation Terminology

- **PORPHYRINS** — light-sensitive compounds necessary for and involved in production of...
- **HEME** — mixture of iron and porphyrins to make...
- **HEMOGLOBIN** — in red blood cells; holds onto oxygen; gives cells and blood red colour





The Heme Pathway



This is a brief model of the pathway that *porphyrins* must pass through to create *heme* and *hemoglobin*.

Certain irritants can cause porphyrins to build up in stages of this path.

If one part of the pathway experiences problems, then the whole system will be affected.





Heme Pathway Analogy

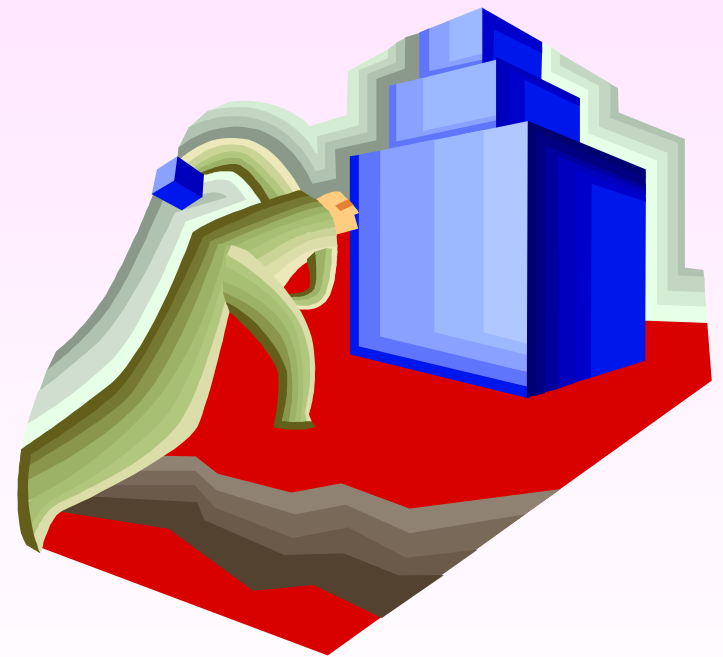
- It's as if there is a gatekeeper at each stage of the heme pathway
- This gatekeeper is the only one who can perform the job of letting porphyrins through to the next stage





Analogy Continued

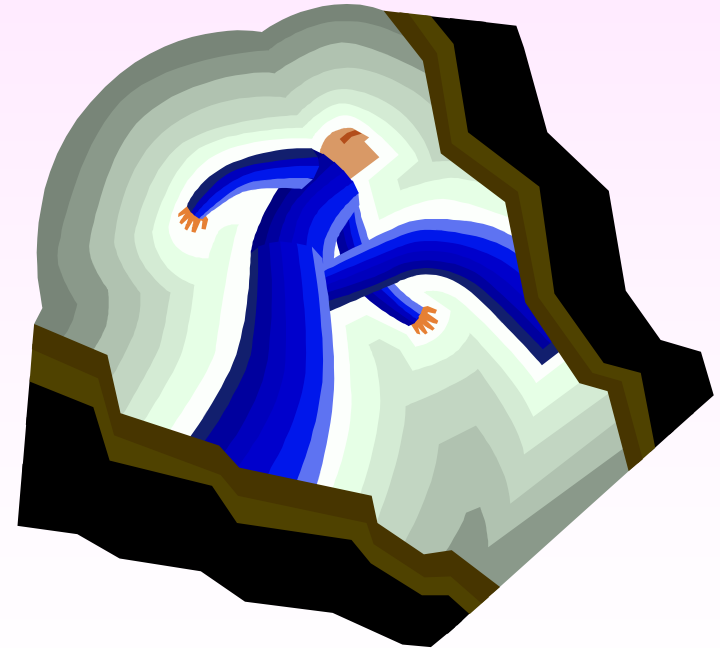
- When exposed to irritants, the gatekeeper will not function properly
- Porphyrins will build up at certain stages because the gate isn't opening normally
- The heme pool will not be getting enough porphyrins





Analogy Continued

- The heme pool then calls upon the first stage to produce more porphyrins
- Once this happens, the problem stage will experience a flood





Porphyria

- The cause of Porphyria is very intricate
- The Heme Pathway has many stages
- Porphyria can occur at any stage
- This is why there are many

TYPES

- Each different type has different

SYMPTOMS





Main Types

- Acute Intermittent Porphyrria (AIP)
- Hereditary Coproporphyrria (HCP)
- Variegate Porhyria (VP)
- Porphyrria Cutanea Tarda (PCT)
- Erythropoietic Protoporphyrria (EPP)
- Congenital Erythropoietic Porphyrria(CEP)
- ALAD Porphyrria (ADP)





Symptoms

- Easily damaged, sensitive skin
- Blistering, scarring, and darkening of skin
- Abdominal pain, cramps
- Constipation
- Rapid heart beat
- Nausea or vomiting
- Anxiety or disturbed behaviour
- Weakness or paralysis (extreme cases)
- Seizures
- Death (possible but fortunately VERY rare)





Prevention

Attacks may be caused by...

- Sunlight
- Numerous drugs (including birth control pill)
- Exposure to some chemicals
- Alcohol
- Exhaustion
- Severe Dieting





Prevention Continued

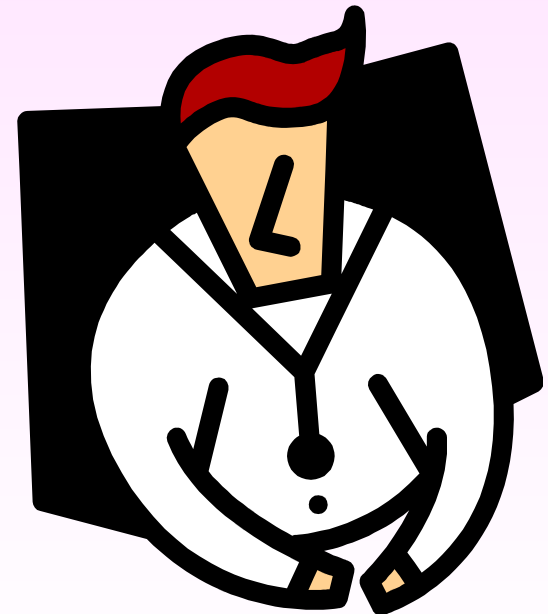
- Porphyrics must avoid these triggers
- High carbohydrate diet is helpful
- Knowledge of safe and unsafe drugs
- Contact CAP to meet someone with similar symptoms
- Keep informed to learn new discoveries





Treatment

- Treatment depends on type
- Can be quite successful
- Safe drugs can be effective
- Supportive therapy is a key
- Drugs to treat other conditions can affect Porphyria
- If your family has Porphyria, your doctor must know!





How To Donate



- In memory of someone who has passed away
- Celebrate an event
- Designate your United Way
- Your employer might match it!





Canadian Association for Porphyria



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Types of Porphyrria

The following information provides a brief description of the 8 types of porphyrria and should not be considered as all inclusive.

For complete information, ask your doctor or contact the Canadian Association for Porphyrria to determine where further information may be obtained.





ACUTE INTERMITTENT PORPHYRIA (AIP)

- a rare disease that is usually inherited from one parent
- deficiency in the enzyme porphobilinogen deaminase (PBGD)
- a deficiency of PBGD is not sufficient by itself to produce AIP, other activating factors must also be present and include hormones, drugs, exposure to chemicals and dietary changes
- symptoms usually occur as attacks that develop over several hours or days. When the disorder is active, it can cause intermittent attacks of abdominal pain as well as a variety of symptoms, which range from mild to life-threatening
- hospitalization is often necessary for acute attacks
- a high intake of glucose or other carbohydrates can help suppress disease activity





CONGENITAL ERYTHROPOIETIC PORPHYRIA (CEP)

- extremely rare and autosomal recessive
- the deficient enzyme is uroporphyrinogen III cosynthase (or uroporphyrinogen III synthase)
- skin photosensitivity may be extreme and lead to blistering, severe scarring and increased hair growth. Bacteria may infect the damaged skin. Facial features and fingers may be lost through phototoxic damage as well as infection. Red blood cells have a shortened life-span, and anemia often results
- blood transfusions and perhaps removing the spleen may reduce porphyrin production by the bone marrow. Activated charcoal given by mouth is sometimes effective. Bone Marrow Transplantation has been very effective in some patients





PORPHYRIA CUTANEA TARDA (PCT)

- deficiency of the enzyme, uroporphyrinogen decarboxylase (UROD)
- PCT is essentially an acquired disease, but some individuals have a genetic (autosomal dominant) deficiency of UROD that contributes to its development
- most individuals with the inherited enzyme deficiency remain latent and never have symptoms
- blisters develop on sun-exposed areas of the skin, such as the hands and face. The skin in these areas may blister or peel after minor trauma
- liver function abnormalities are common but are usually mild. These sometimes progress to cirrhosis and even liver cancer
- most widely recommended treatment is a schedule of repeated phlebotomies (removal of blood), with the aim of reducing iron in the liver
- alcohol, iron, estrogens and Hepatitis C Virus are factors that can activate PCT





ALAD PORPHYRIA (ADP)

- inherited as an autosomal recessive trait and seems to be extremely rare
- there is a deficiency of the enzyme delta-aminolevulinic acid dehydratase (ALAD) and increased excretion of delta-aminolevulinic acid (ALA) in the urine
- the symptoms are very similar to acute intermittent porphyria





HEPATOERYTHROPOIETIC PORPHYRIA (HEP)

- a very rare type of porphyria due to a deficiency of uroporphyrinogen decarboxylase
- the enzyme deficiency is inherited as an autosomal recessive trait
- the manifestations of HEP resemble CEP, with symptoms of skin blistering usually beginning in infancy
- porphyrins are increased in bone marrow and red blood cells, as well as liver, plasma, urine and feces





HEREDITARY COPROPORPHYRIA (HCP)

- an autosomal dominant form of hepatic porphyria that is similar to AIP, except that some patients develop skin photosensitivity
- deficient enzyme is coproporphyrinogen oxidase
- precautions and treatment for acute attacks are as described for AIP





VARIEGATE PORPHYRIA (VP)

- an autosomal dominant disorder and may produce acute attacks (as in AIP) as well as skin photosensitivity
- the deficient enzyme is protoporphyrinogen oxidase
- in patients with skin manifestations, it is important to distinguish VP or HCP from PCT, because treatment by phlebotomy or low-dose chloroquine is not successful in VP and HCP
- acute attacks are managed and may be prevented as in AIP





ERYTHROPOIETIC PROTOPORPHYRIA (EPP) or PROTOPORPHYRIA

- due to a deficiency of ferrochelatase
- inherited as an autosomal dominant trait
- swelling, burning, itching, and redness of the skin may appear during or immediately after exposure to sunlight, including sunlight that passes through window glass
- other manifestations may include gallstones containing protoporphyrin, and occasionally, severe liver complications
- treatment with beta-carotene improves sunlight tolerance but does not lower porphyrin levels
- a few patients with EPP and advanced liver damage have developed neurological symptoms similar to those in the "acute" porphyrias

