Understanding the disease of porphyria

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The **Canadian Porphyria Foundation** (CPF) had its beginning in the mind of a person who had experienced the lack of knowledge and support when searching for porphyria information in the mid-1970s. In January 1988, a group of six volunteers armed with $350 set out on a mission "to improve the quality of life for people affected by porphyria". The Canadian Porphyria Foundation was formed by September 1988 with the national office located in Neepawa, Manitoba.

The CPF is the only organization of its kind in Canada and is dedicated to providing quality service and support to porphyria sufferers not only in Canada, but globally as well. Since its inception 17 years ago, the CPF has come a long way. It has progressed from being a small rural organization into an international resource of information for porphyric patients. Recently, the CPF had its educational guides printed bilingually and will soon have its "Porphyria Safe and Unsafe Drug Guide" updated and also printed bilingually. This year on June 1, National Porphyria Day was celebrated for the first time in Canadian medical history.

Many people, even those in the medical field, have not heard of porphyria or don't know that it is a disease. Porphyria is not a single disease, but is a group of disorders of at least eight different types. It is the result of an accumulation of porphyrins in the body caused by an enzyme deficiency that occurs due to a genetic defect. Porphyrins are normal body chemicals that can be found in plants and animals. They are produced in the liver and bone marrow and are necessary in the production of heme, which goes on to make hemoglobin. Hemoglobin is the red blood cells that transport oxygen from the lungs to the tissues of the body.

Porphyria is a hereditary disease, but also can be acquired after exposure to certain chemicals. Symptoms of porphyria may include extreme sun sensitivity, abdominal pain, nausea and vomiting, muscle pain and weakness, psychosis, hallucinations, seizures and limb and respiratory paralysis. In rare cases, it can result in death.

Porphyrias are generally divided into two groups: the **acute porphyrias** and **non-acute porphyrias**. Non-acute porphyria is better known as the sun-sensitive or photosensitive kind. However, this does not mean that you can have only one kind of porphyria. Some people suffer from acute porphyria with symptoms of sun sensitivity.

The acute porphyrias are acute intermittent porphyria (AIP), variegate porphyria (VP), hereditary coproporphyria (HCP), and ALAD deficiency porphyria. AIP is perhaps the most severe of all the porphyric syndromes in terms of its symptomatology. It is inherited and is a deficiency in the enzyme porphobilinogen deaminase. There are several triggers that bring about a porphyric attack. Some of these include starvation or unusual diets, street drugs, prescribed medications and environmental stimuli. Endogenous stimuli are also involved, including stress, intercurrent illness and normal menstrual cycles. High concentrations of glucose and other carbohydrates are helpful. The treatment of hematin is used in acute attacks. The prognosis of the acute attacks is good, and most symptoms settle quickly although, at times, severe nerve damage and its associated signs of weakness and sensory disturbance may take several months to improve.

VP is a type of porphyria that may be similar in its acute attacks to AIP, but is also associated with a classic photosensitive skin disorder. It is inherited and is relatively common in the white population of South Africa. The disease rarely appears before puberty, is most common in the young adult, but may suddenly occur at any age including the elderly. It is caused by a deficiency of the enzyme protoporphyrinogen oxidase. These changes include skin fragility, erosions and blisters during the acute attack, and abnormal pigmentation, skin thickening and hirsuitism due to chronic exposure. The precipitating factors and treatment are similar to those of AIP. Patients should be advised to avoid sun exposure and to use sunscreens containing zinc oxide or titanium oxide. If both parents carry the abnormal gene, the disease will be present in early childhood and will be rather severe.

HCP is inherited as an uncommon type of porphyria. It is caused by a deficiency of the enzyme coproporphyrinogen oxidase. The clinical symptoms are similar to those of AIP, but it can be associated with the type of photosensitive dermatitis seen in PCT. Fatigue and muscle weakness are symptoms and sometimes the patient may be jaundiced. The treatment is essentially the same as AIP, with hematin usually being effective.

ALAD is a very rare form of porphyria that is inherited in an autosomal recessive fashion and has been diagnosed in a very small number of patients whose ages range from infancy to adulthood. There is almost a complete lack of enzyme activity.

The non-acute porphyrias are porphyria cutanea tarda (PCT), erythropoietic protoporphyria (EPP), congenital erythropoietic porphyria (CEP), and hepatoerythropoietic porphyria (HEP). PCT is the most common of all the porphyrias. It is caused by the deficiency of the enzyme uroporphyrinogen decarboxylase. Excessive alcohol ingestion has long been recognized as an important cause, possibly related to the development of chronic liver disease. Estrogen therapy may also be a factor in this disease. Infections have been implicated. Iron overload states may cause or magnify the disease.

EPP is due to a deficiency of ferrochelatase. The symptoms are precipitated primarily by sunlight and cause burning, itching, swelling and redness of the skin. Occasionally, liver disease may develop and gall bladder disease is a common problem because the high concentration of protoporphyrin in the bile will lead to gall stone formation. Treatment with beta-carotene improves sunlight tolerance.

CEP is extremely rare. The enzyme that is deficient is uroporphyrin III cosynthase. It occurs at a very young age and has a marked degree of photosensitivity. Total avoidance of sunlight is usually essential to prevent disfiguration. This is the only type of porphyria that can be diagnosed prenatally.

HEP is a very rare type of porphyria due to a deficiency of uroporphyrinogen decarboxylase. Marked phototoxic skin lesions develop early in childhood along with a variety of neurological abnormalities.

If you are interested in learning more about the Canadian Porphyria Foundation, porphyria or about National Porphyria Day, you can contact the Canadian Porphyria Foundation at 1 (866) 476-2801, or visit our website at www.cpf-inc.ca, or e-mail us at porphyria@cpf-inc.ca.

Much of the information in this article is taken from the Canadian Porphyria Foundation's **A Guide to Porphyria booklet by Dr. Barry Tobe.

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Tapeworms

By Ted Sellers, RN, BHScN, ENC(C) - Ontario

Some facts about tapeworms

- They are intestinal parasites of vertebrate animals (including humans)
- They absorb partially digested food through body surfaces as they have no mouths or digestive canals
- Most infections occur in Africa, Yugoslavia, Middle East, Southeast Asia, Mexico, parts of South America and the former U.S.S.R.
- In the U.S., some forms can be contracted from infected dogs or cats when children eat infected fleas
- Some worms have been known to live up to 20 years and exceed 10 metres in length

Tapeworm characteristics

- Flattened worms range from 0.5 inches to 30 feet long
- The head (scolex) has a crown of hooklets for attachment to intestinal lining
- They have a narrow neck and then body segments (proglottids) that are budded off asexually
- They may have as few as three or as many as several thousand proglottids
- The proglottids contain organs of sexual reproduction – both testes and ovaries
- They are ribbon-shaped, segmented worms
- Usually fertilization is between worms, but some species self-fertilize
- Some species shed eggs continuously in the feces, others store the eggs and shed them in the proglottid

Transmission

- The proglottids furthest away from the head mature most rapidly. When they mature, they break off and pass out of the host in the feces
- This newly detached proglottid contains several eggs with embryonic tapeworms
- This living proglottid is ingested by another primary host, regenerates a new scolex that attaches itself to the intestinal wall and resumes growth
- When eggs are ingested, they hatch into larvae, then burrow into the tissue of the host and form cysts. These are known as bladder worms, cycticeri, hydatids and measles
- These larvae attack certain selected tissues (e.g., liver in humans and dogs, brain in sheep)
- When the larvae are ingested, their growth into tapeworms is stimulated by gastric juices

Classes of tapeworms

- Together, they form the class called CESTODA
- Dwarf tapeworm – hymenolepsis nana – is transmitted through fecal contamination
- Fish tapeworm – diphyllobothrium latum – is seen in fish, especially in pike
- Liver tapeworm – taenia coenurus – are also known as hydatid cysts
- Sheep brain tapeworm – taenia coenurus – causes the disease in sheep known as "gid" or "staggers"
- Pork tapeworm – taenia solium
- Beef tapeworm – taenia saginata – can occur often in people who eat raw or lightly cooked beef

Signs and symptoms of tapeworms

- Often patients are asymptomatic with tapeworms
- If symptoms are present, they may include unexplained weight loss, symptoms of pernicious anemia, presence of white eggs or ribbon-like segments of worm in stool, abdominal discomfort, diarrhea, constipation

Treatment

- To eradicate the worm, the scolex must be dislodged from the bowel. If this is not done, the worm will regrow
- Quinacrine hydrochloride (atabrine) medication kills the worm

Resources

http://encarta.msn.com/encnet/refpages/refarticle.aspx?refid=761566007
http://www.1upinfo.com/encyclopedia/T/tapeworm.html
http://www.medhelp.org/glossary/new/gls_4070.htm
http://www.infoplease.com/ce6/sci/A0861444.html
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