

Porphyria

Porphyria is not a single disease. It is a group of at least eight disorders, all of which can cause severe pain and other serious health effects.

All eight forms of porphyria have a common cause. They are caused when *porphyrins* or *porphyrin precursors* – building blocks in chemicals that are part of metabolism – keep building up inside the body, rather than being used and discharged. Porphyrins tend to be purple in color – the word "porphyria" is derived from the Greek word for "purple" – and for this reason, the urine of people with porphyria may be, or may become after exposure to light, reddish in color, which can be one clue to the presence of the condition.

The form of porphyria a person develops depends on which of the various porphyrins builds up in the body. Some forms of porphyria chiefly affect the skin, causing extreme sensitivity to sunlight that may make it necessary to avoid porphyria altogether. Others affect the nervous system. Symptoms of these forms can include excruciating abdominal pain – pain that seems literally unbearable – faintness, loss of feeling or control in the limbs, and, in some cases, symptoms as extreme as hallucinations.

Porphyria is rare and tends to have a genetic cause. Tests have to be performed to determine if an individual has the ailment. When it is diagnosed, genetic testing can help determine whether other family members may be at risk. The number of laboratories able to perform the tests in reliable fashion is limited, and proper supervision of tests is key.

For persons who are diagnosed with porphyria, help is available. There are medications that can reduce the risks of an onset of symptoms. There are also things that individuals may need to avoid to reduce their risk of attacks. For example, some medicines are believed frequently to be responsible for inducing attacks. Alcohol can also cause attacks. So can failure to consume enough carbohydrates. Of course, exposure to sunlight can trigger the painful skin conditions associated with some porphyrias.

As with any painful physical ailment, only a doctor can diagnose porphyria or prescribe a course of treatment for it. Some doctors have special expertise in dealing with porphyria. The American Porphyria Foundation is an excellent source of further information and support. Its website is at <http://porphyriafoundation.com>.