A porphyria is a blood disorder in which cells fail to change chemicals (porphyrins) to the substance (heme) that gives blood its color. Porphyria Cutanea Tarda, also called PCT, is the most common type of porphyria. PCT can be genetic or acquired. The genetic form is determined by a deficiency in the liver chemical uroporphyrinogen or a liver enzyme called uroporphyrinogen decarboxylase. Both causes of the genetic form usually occur in individuals with a genetic predisposition in addition to liver disease or excess liver iron. The acquired (sporadic) form most often occurs after use of alcohol, estrogens, oral contraceptives, other drugs and certain environmental pollutants. Sometimes no cause can be found. Most people who consume alcohol and take estrogens do not develop a porphyria; therefore, it is likely that genetic factors are of higher importance.

Clinical signs and symptoms in order of frequency are:

1. Blistering on sun-exposed areas
2. Increased skin fragility
3. Hair on the face and other abnormal areas
4. Increased skin pigmentation
5. Ulcerations
6. Small cysts, called milia, that occur in previously blistered areas

Diagnosis of PCT is made by taking a sample of the patient’s urine. The urine may have a red-brown discoloration and high levels of porphyrin pigments, which can be seen under fluorescence. A special test called an assay should be ordered of plasma and a 24 hour urinary porphyrin. The assay provides a proportion of uroporphyrin to coproporphyrin and in PCT the ratio can be 4:1 or greater. The doctor may also take a biopsy of the affected area of the skin as well as check other liver enzymes and a stool sample.

Treatment of PCT includes phlebotomy, the therapeutic withdrawal of blood, to reduce blood and liver stores of iron. Common medications also used for treatment are chloroquine and hydroxychloroquine.