PORPHYRIA CUTANEA TARDA, HEPATITIS C, AND HEPATIC HEMOSIDEROSIS

C. Clementi[1], G. Gimignani[2]

[1]Casa di Cura "Santo Volto" Santa Marinella-Roma; [2]UOC Medicina Interna Osp.San Paolo Civitavecchia, Roma.

Porphyrias are a group of disorders of heme biosynthesis, each characterized by an enzymatic defect in the heme biosynthetic pathway. The pathogenesis of Porphyria Cutanea Tarda involves a hepatic deficiency of uroporphyrinogen decarboxylase (UROD). Under normal conditions, UROD converts uroporphyrinogen to coproporphyrinogen through a series of decarboxylations. In the presence of iron and free radicals, uroporphyrinogen is partially oxidized, leading to the formation of a uroporphyrinogen inhibitor of UROD. Subsequent accumulation and oxidation of the corresponding porphyrins, predominantly uroporphyrins in plasma, are responsible for the cutaneous manifestations of PCT. Most patients with PCT show signs of siderosis during liver biopsy and resolve with depletion. PCT manifests as skin fragility, blisters and skin lesions on sun-exposed areas, dark urine, and elevated porphyrins in plasma and urine. One of the most frequent risk factors is the presence of HCV-related hepatitis, obviously associated with a genetic predisposition. In our case, a patient over 80 years of age was an asymptomatic HCV carrier with no signs of cytolysis. Based on the appearance of a beard, blisters, and sores on the backs of the hands and subsequent investigations, a diagnosis of Porphyria Cutanea Tarda was made, also associated with elevated ferritin levels and a positive HCV RNA. The biopsy confirmed the presence of liver iron. Treatment with direct-acting antiviral drugs (DAAs) led to HCV eradication and clinical and biochemical remission of the porphyria cutanea tarda. Porphyria cutanea tarda is therefore an acquired condition with susceptibility factors associated with the pathogenesis of the disease.

Abstracts printing tool powered by pagepress.org