E9.THE PORPHYRIAS.

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Traditionally, the porphyrias have been classified as either hepatic or erytropoiethic, depending on the primary site of the overproduction and accumulation of porphyrin and porphyrin precursors, although some porphyrias have overlapping features. For simplicity, we have classified the eight major porphyrias into three groups: (1) the four acute hepatic porphyrias, (2) the single hepatic cutaneous porphyria (PCT) and (3) the three erythropoietic cutaneous porphyrias.

The acute hepatic porphyrias includes AIP,HCP,VP (AIP is the most common). The major manifestations of these disorders are acute neurological attacks, abdominal pain, cramps, constipation, abdominal distension, increased bowel sounds, nausea, vomitting, tachicardia, hypertension, menthal symptoms, chest pain, headache, muscle weakness, tremors,disuria, bladder distension. Once a biochemical diagnosis is established , mutation analysis of the genes for AIP (HMBS), HCP (CPOX),VP (PPOX) and then ADP (ALAD) should be undertaken.

CEP is an AR porphyria that results from deficient activity of URO-synthase and results in the accumulation of Uroporphyrin I and Coproporphyrin I isomers. The skin areas overexposed to sunlight are friable and blisters and vesicles are prone to rupture and infection. Skin thickening, focal hypo- and hyperpigmentation and hyperthricosis of the face and extremities are characteristic. The diagnosis is confirmed by the demonstration of significantly deficient URO-synthase activity or the identification of specific mutations of the UROS-gene.

The knowledge of these advances is relevant for hematologists because they administer the hematin infusions to treat the acute attacks in patients with acute hepatic porphiryas, perform the chronic phlebotomies to reduce iron overload, clear the dermatologic lessions in PCT, diagnose and treat the erithropoietic porphiryas, including chronic erythrocyte transfusions, B.M. or SCT transplants and experimental pharmacologic chaperone and stem cell gene therapies for CEP. These developments are relevant to update hematologists on the latest advances in these diverse disorders.