Summary

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The Porphyrrias

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The porphyrias are a group of uncommon diseases due to enzymatic deficiencies in the heme pathway. This produces accumulation of intermediary metabolites (the porphyrins) which are responsible for the diverse clinical and biochemical manifestations [1].

The porphyrias have been classified into two groups in relation to the main organ where the abnormal porphyrin synthesis occurs (hepatic and erythropoietic porphyrias). Other classifications are based in the genetics of these diseases (recessive, dominant, and non-hereditary porphyrias). However, based on their clinical expression, the porphyrias could be divided into three groups:

A. Acute porphyrias

Acute intermittent porphyria (AIP)

Porphyria due to uroporphobilinogen-synthase deficiency (PBG-S)

B. Cutaneous porphyrias

Porphyria cutanea tarda (PCT)

Erythropoietic protoporphyria (EPP)

Congenital erythropoietic porphyria (CEP)

Hepatoerythropoietic porphyria (HEP)

C. Mixed porphyrias

Porphyria variegata (PV)

Hereditary coproporphyria (HCP)

In the period 1969-1993 we have been able to study 750 cases of porphyria in the Department of Dermatology of the Hospital Clinic of Barcelona. Most patients were affected with PCT (688 cases). But we have also seen other types of porphyria (23 cases of EPP, 23 of PV, 5 of CEP, 5 of HEP, 5 of AIP, 1 of HCP).

Dermatologic manifestations of porphyrias include a subacute syndrome of hyperfragility of the skin (with post-

traumatic blisters that develop on the dorsa of the hands after minimal trauma) and an acute syndrome of cutaneous photosensitivity (with erythema, oedema and purpuric lesions appearing after a short exposure to sunlight). The syndrome of hyperfragility, which is associated to hypertrichosis, hyperpigmentation, premature ageing of the skin and scleroderma-like lesions, develops in porphyrias where hydrophilic porphyrins accumulate (PCT, CEP, HCP, HEP, PV). The porphyrias with increased hydrophilic porphyrins may also present cutaneous photosensitivity if the abnormal porphyrins are present in very high amounts.

Cutaneous porphyrias may also present with non-cutaneous manifestations. Liver alteration is almost constant in PCT, even in patients who have no clinical or laboratory evidence of hepatic alteration. Toxic, infectious and hormonal factors (as alcohol or hepatitis C virus infection) play a role in triggering liver involvement in PCT [2]. In EPP, the liver may also be severely involved. Other non-cutaneous manifestations include ocular alterations (conjunctivitis, keratitis, loss of eyelashes, scleromalacia) and, in CEP, hemolytic anemia.

Laboratory determination of urinary, fecal and blood porphyrin patterns permit to identify the different types of porphyria. More sophisticated studies may precise the nature and the degree of enzyme deficiency.

The management of porphyrias must not only include the conventional therapy (as phlebotomies, oral antimalarials, iron chelators in PCT, or betacarotene in EPP) but also photoprotection and diverse prophylactic measures to decrease the risk of mutilating tendency of cutaneous lesions in the severe forms and the progression of associated liver disease.

References

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