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Letter to Editor

Letter to the Editor regarding the article "Porphyria cutanea tarda: A novel mutation" by Patil R et al. in doi:10.1016/j.phoj.2016.04.001

To the Editor

Dr (Surg Cdr) Gaurav Narula

I suggest another diagnosis for this child; I believe it may be a case of hepatoerythropoietic porphyria (HEP). Hepatoerythropoietic porphyria (HEP) is a very rare type of autosomal recessive porphyria caused by severe deficiency of uroporphyrinogen decarboxylase (UROD) [1]. Clinically it resembles congenital erythropoietic porphyria (CEP), but biochemical findings are similar to those found in porphyria cutanea tarda (PCT).

Deficiency in UROD activity was firstly described by Elder et al. [3] in 1981. In most cases, the catalytic activity of UROD is below 10% of normal level. HEP is caused by biallelic (i.e., homozygous or compound heterozygous) *UROD* mutations [2]. Several mutations were identified in UROD gene of patients with HEP, indicating the molecular heterogeneity of this disease [4–9]. Five to ten mutations identified in HEP were also found in family PCT (type II), which confirms the idea that HEP is the homozygous form of PCT [10]. Mutations of UROD found in PCT apparently are more critical to the enzymatic activity of UROD than the mutations found in HEP [11].

Clinical manifestations begin in childhood and are characterized by severe photosensitivity, pruritus and bullous lesions that progress to mutilating scars on sun-exposed areas. These patients have shortened distal phalanges, thickened skin, and severe hypertrichosis [12] (which worsens with age). Photosensitivity tends to decrease with age. The sclerodermiform skin changes are similar to those found in CEP. Erythrodontia and eye disorders can occur in these patients. Some patients may develop hemolytic anemia and hepatesplenomegalia [13,14].

Biochemical findings show increase in urinary porphyrins, predominantly URO and to a lesser extent heptacarboxyl porphyrin. The ISOCOPRO has a concentration greater than or equal to COPRO and can be found in urine and feces. Zn-PROTO in erythrocytes is also increased. The increase in ISOCOPRO (urine and feces) and Zn-PROTO (erythrocytes) are diagnostic criteria [14]. Some patients may experience changes in liver enzymes, but serum iron levels are normal. The pattern of urinary porphyrins in HEP is similar to that found in PCT with a ratio URO/COPRO greater than 5: 1 and again it is the increase in erythrocyte Zn-PROTO that allows the differentiation [13].

Unlike PCT, as iron concentrations are normal, phlebotomy is not effective in this disease. We treat our patients in Brazil with low dose regimen of hydroxychloroquine (400 mg, 2 times per week) and the photosensitivity and blisters improve. As the child grows the number of blisters tend to decrease. Avoidance of sun exposure is very important.

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