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Short Report

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Hepatoerythropoietic Porphyria and Cerebral Abnormalities

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Introduction

In porphyrias, hereditary enzyme deficiency can be recognized in the red blood cells. Four enzymes of hema biosynthesis can be detected: porphobilinogen synthase (delta-aminolevulinic acid dehydrase), uroporphyrinogen synthase, cosynthas and decarboxylase. A decrease of porphobilinogen synthase observes in lead intoxication. Uroporphyrinogen cosynthase decreases in congenital erythropoietic porphyria. Acute intermittent porphyria is characterized by deficient uroporphyrinogen synthase. Uroporphyrinogen decarboxylase decreases in the genetic type of porphyria cutunea tarda (1)

Hepatoerythropoietic porphyria (HEP) is a rare homozygous variant of porphyria cutunea tarda, autosomal recessive disorder due to deficient hepatic uroporphyrinogen decarboxylase enzyme activity (2). We reported a 5-year old boy who had the clinical and biochemical findings of HEP. Brain tomography revealed hydrocephalus and subarachnoidal cyst. Central nervous system abnormalities are a rare feature in association with HEP.

A 5-year old boy, was hospitalized with photosensitivity, red colour urine, hypertrichosis and hyperpigmentation particularly on his face and all body surfaces; The brownish wounds on his face, and hands healed with white scars (fig 1). These features were first appeared three years ago. He was delivered after an uneventful pregnancy, and normal labor. All the family member were healthy and there was no history of illness. The parents were not relatives. On physical examination, the head circumference was 50 cm (90 p), the weight 19.2 kg (50-75 p) and the length 106 cm (25 p). Blood pressure ws 100/60 mmHg, the breath 26/minute, and the pulse 132/minute. The patient was considered as mentally retarded because of the apperance and behaviours, but the age of the patient was too small for performing the intelligence test. Liver was 2 cm palpable and liver biopsy showed nonspecific changes. Spleen was nonpalpable. Laboratory findings: Hb 9.3 gr./dl, WBC 6400/mm^3, thrombocyt 359000/mm^3, AST 170 U/L, ALT 214 U/L alkaline phosphatase 349 U/L total protein 7.9gr./dl,

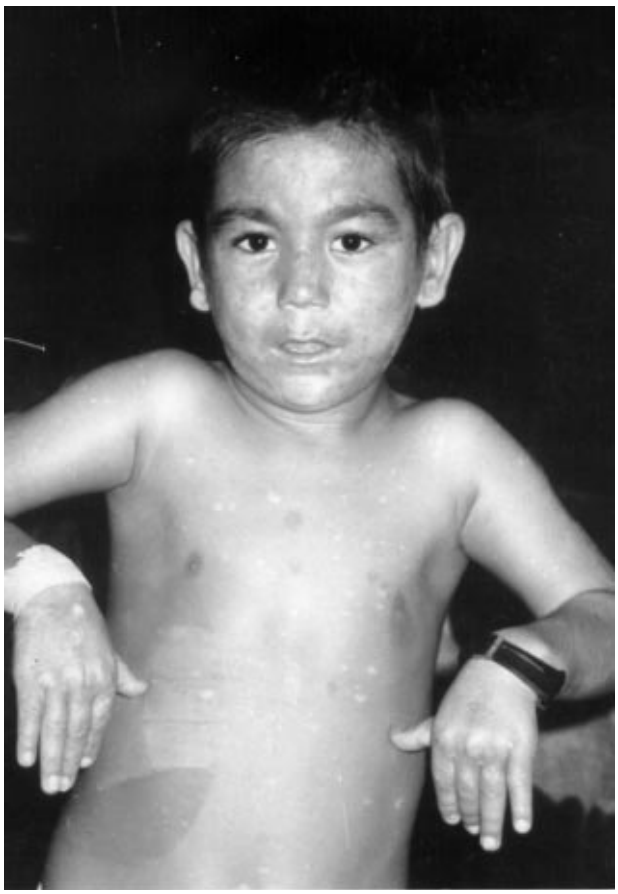

Figure 1. Brownish wounds of on his face and hand healing with white scars and hypertrichosis on his face are seen.

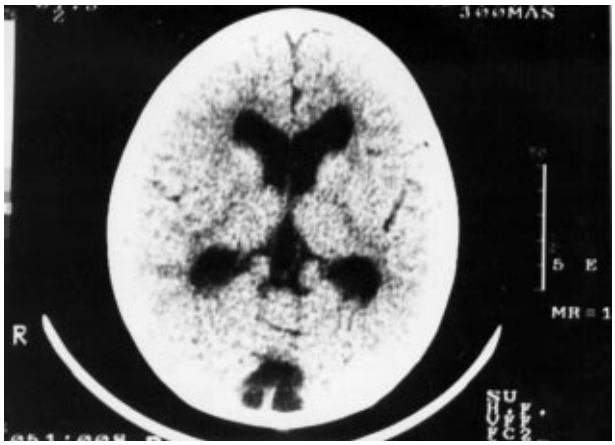

Figure 2. Brain tomography reveals hydrocephalus and subarachnoidal cyst.

albumin 3.9 gr./dl, total biluribine 0.35 mg/dl, direct bilirubine 0.20 mg/dl the makers of the hepatitis A, B, C and D were negatives, serum iron 33 µg/dl aminolaevulinic acid 15 mmol/L (0-40), porphobilinogen 10 mmol/L (0-16), uroporphyrin 500 nmol/L (0-40), coproporphyrin 6985 nmol/L (0-370). Brain tomography revealed hydrocephalus and subarchnoidal cyst. (Fig.2)

Porphyrias are either hepatic or erythroid, depending on the specific enzymatic defect. Hepatoerythropoietic porphyria (HEP) is the resulting from a deficiency of homozygous hepatic uroporphyrinogen decarboxylase activity, an enzyme that is essential for hema biosynthesis. The condition is extremely rare in children. Most cases of childhood HEP are familial, severe, and commonly associated with liver disease and hepatic iron overload (3). In our case, biochemically elevated levels of uroporphyrines in urine and coproporphyrins in feaces are markers of this form of porphyria. Serum iron level was normal, serum transaminase levels were elevated and liver biopsy showed nonspecific changes. All family members of the patient were healthy.

HEP has typical cutaneous manifestations: photosensitiity with blistering and mild scarring and hypertrichosis (4). Our patient had hyperpigmentation particularly on his face and all of body surface brownish wounds on his face and hand healing with white scars and hypertrichosis on his face.

Protoporphyrin overproduction occures in erytroid tissue. The release of protoporphyrin from erytrocytes is greatly increased if the erythrocytes are exposed to light. The cutaneous symptoms are related by protoporphyrin-sensitized photodamage of endothelial cellls. Endothelial cells accumulated protoporphyrin from albumin or lipoproteins present in the plasma. Uroporphyrin and coproporphyrin are hydrophilic and are unbound in plasma. In hepatic and erythropoietic porphyrias, clinical symptoms are probably evoked by uroporphyrin and coproporphyrin present in the interstitial tissue. Very little is know about the primary targets of uroporphyrin and coroporphyrin photodamage in these disorders (5).

Central nervous system abnormalities have been previously reported in association with HEP (2) but there are rare features. In our case, brain totography revealed hydrocephalus and subarachnoidal cyst.

HEP is controlled by removal of iron by phlebotomies or with lowdose chloroquine. Skin symptoms in porphyria can be reduced with beta caroten there is no effective treatment in congenital porphyria but suggested that bone marrow transppantation (6,7). Caudry performed gene transferred in Congenital erythropoetic porphiria (8). Serum iron levels was normal and liver biopsy showed nonspecific changes in our patient. We didn't use betacaroten for skin symptoms.

Eleven patients with HEP have been reported until 1984 (9). This case was found interesting because of the rare occurrence of HEP and very rare presentation of it with central nervous system abnormalities.

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