Hepatoerythropoietic Porphyria and Cerebral Abnormalities

Introduction

In porphyrias, hereditary enzyme deficiency can be recognized in the red blood cells. Four enzymes of heme 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 cutanea tarda (1).

Hepatoerythropoietic porphyria (HEP) is a rare homozygous variant of porphyria cutanea 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/mm³, thrombocyte 359000/mm³, AST 170 U/L, ALT 214 U/L alkaline phosphatase 349 U/L total protein 7.9gr./dl,
albumin 3.9 gr./dl, total bilirubine 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 amino-
laevulinic acid 15 mmol/L (0-40), porphobilinogen 10
mmol/L (0-16), uroporphyrin 500 nmol/L (0-40), cop-
roporphyrin 6985 nmol/L (0-370). Brain tomography
revealed hydrocephalus and subarachnoidal cyst. (Fig.2)

Hepatoerythropoietic porphyria (HEP) are either hepatic or erythroid, de-
pending on the specific enzymatic defect. He-
patoerythropoietic porphyria (HEP) is the resulting
from a deficiency of homozygous hepatic uro-
porphyrinogen decarboxylase activity, an enzyme that
is essential for hema biosynthesis. The condition is ex-
tremely rare in children. Most cases of childhood HEP
are familial, severe, and commonly associated with liv-
er 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, ser-
um transaminase levels were elevated and liver biopsy
showed nonspecific changes. All family members of the
patient were healthy.

Protoporphyrin overproduction occures in erytroid
tissue. The release of protoporphyrin from erythrocytes
is greatly increased if the erythrocytes are exposed to
light. The cutaneous symptoms are related by proto-
porphyrin-sensitized photodamage of endothelial cells.
Endothelial cells accumulated protoporphyrin from al-
bumin or lipoproteins present in the plasma. Uro-
porphyrin and coproporphyrin are hydrophilic and are
unbound in plasma. In hepatic and erythropoietic por-
phyrias, clinical symptoms are probably evoked by uro-
porphyrin and coproporphyrin present in the inter-
stitial tissue. Very little is know about the primary
targets of uroporphyrin and coroporphyrin photo-
damage 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 tomography
revealed hydrocephalus and subarachnoidal cyst.

Hepatoerythropoietic porphyria: Clinical,
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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.