Letter to Dermatology

Dermatology 1994;188:329-330

Plasmasorbent Therapy with Activated Charcoal Column for Congenital Erythropoietic Porphyria

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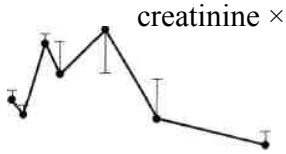
Congenital erythropoietic porphyria (CEP), classically known as ‘Günther’s disease’, is an extremely rare autosomal reces-sively inherited deficiency characterized by mutilating cutaneous photosensitivity and abnormal porphyrin heme synthesis in the bone marrow [1]. Although several therapeutic trials for CEP have been conducted previously, they have been generally only partially successful [2-5]. We report a trial of plasmasorbent therapy with an activated charcoal column in a CEP patient which considerably reduced the plasma concentration of porphyrin shortly after treatment, without side effects.

The patient, a 21-year-old Japanese woman with consanguineous parents, was admitted to our hospital for further evaluation and treatment of CEP. She was first noted to have marked red urine from birth and was subsequently diagnosed with CEP. Her past medical history revealed cholecysto-lithiasis and acute hepatitis. Physical examination and laboratory investigations showed the classic stigma of CEP. After gaining written informed consent we performed plasmasorbent therapy with a 100-g activated charcoal column with the Plasorba system (Asahi Medical Co.. Tokyo, Japan). The membrane plasma separator utilized was Plasmaflo Hi-05, and the absorbent column of uncoated activated charcoal was a Hcmo-sorba N-180, consisting of a closed loop in which plasma porphyrins were circulated from the inlet through the charcoal column and back into the outlet. During plasmasorbent therapy, plasma porphyrin levels were measured by high-performance liquid chro-
matography (HPLC) analysis. Ninety percent of porphyrins were reduced through the column (fig. 1). Sequential measurement of plasma and urinary porphyrins before and after 72 h of a treatment was also taken (fig. 2). As shown in figure 2, plasma porphyrin levels rapidly and transiently decreased shortly after treatment, whereas urinary excretion of porphyrin increased 24 h after a treatment and gradually fell to the basic levels.

100
Inlet
Outlet
Fig. 1. Plasma porphyrin levels (mean < ± SEM) of the inlet and the outlet were measured by HPLC during plasmasorbent therapy. More than 90% of porphyrins were reduced through the activated charcoal column. *p < 0.01.

CEP results from the deficient activity of uroporphyrinogen III synthase (UROS-syn-thase). which plays an important role from hydroxymethylbilane to URO III in heme synthesis. Highly advanced molecular biological techniques revealed genetic point mutations of the URO III gene in CEP [6-9]. Several therapeutic treatments which have been conducted were generally only partially successful, and some side effects that have led to or not led to death have been reported [3, 5].

Plasma
µg/dl 200-|
100
µg/g
Urine
creatinine × 104
⅛,
15
o 10



72
0 12 24 48 Time, h

Fig. 2. Sequential measurement of plasma and urinary porphyrins before and after 72 h of therapy.

1018-8665/94/1884-0329 $ 5.00/0

In our case, plasmasorbent therapy with an activated charcoal column considerably reduced the plasma concentration of porphy-rin shortly after treatment without side effects. On the other hand, urinary excretion of porphyrins increased 24 h after a treatment and gradually fell to the basic level. These
results suggest that not only circulating plasma porphyrins but also porphyrins accumulated in the peripheral tissue such as skin might be removed and excreted through urine.

The prognosis, which is under the influence of the hemolytic process due to the pres-
ence of circulating crythroblasts, is extremely poor. Therefore, at the present time, plasmasorbent therapy is one of the practical treatments for CEP and is recommended. Moreover, more extended studies regarding repetitive therapy and the long-term prognosis are needed.

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