

Congenital Erythropoietic Porphyria

Congenital erythropoietic porphyria (CEP) is a rare cutaneous porphyria which presents with photosensitivity, complicated by presence of hemolytic anemia. There is no permanent cure and the treatment is supportive. A 3 day-old male baby was admitted with neonatal jaundice for phototherapy. The baby developed bullous lesions over both the feet within minutes of starting phototherapy (**Fig. 1**). These lesions were prominently present over the toes and were filled with clear fluid. We observed the color of the urine to be pink. The baby had no hepatosplenomegaly or any evidence of hemolysis. A urine analysis showed the presence of uroporphyrin and coproporphyrin which are not usually detectable suggesting the diagnosis of CEP. A quantitative assay could not be done because of financial reasons. The lesions healed in 2 weeks time following which the patient was discharged with advice regarding photoprotection and regular follow up. The patient is now 3 months old and has not had any further episodes of blister formation.

CEP, also known as Gunther's disease, is an autosomal recessive inherited deficiency of the uroporphyrinogen III cosynthetase enzyme leading to accumulation of type I porphyrins. Less than a hundred cases are reported worldwide(1). The earliest sign of CEP could be brownish discoloration of amniotic fluid or pink to brown staining of the nappies. Severe photosensitivity often begins in the neonatal period itself with blisters developing on



FIG. 1 Bullae over feet due to photosensitivity following phototherapy.

exposure to light. There have been earlier case reports of CEP in which the newborns developed photosensitivity following phototherapy but in most of these newborns hemolytic anemia was the main presenting feature(2,3). There are reports of older children and adults with CEP from India also(4,5). Genetic counseling is important for the parents of an affected offspring. Antenatal diagnosis can be made by measuring the uroporphyrin I concentration in the amniotic fluid which is increased as early as 16 weeks *in utero*.

Diagnosis is made by demonstrating the presence of uroporphyrin and coproporphyrin in the urine and blood. A plasma spectrofluorimetry is seen at 615-620 nm. A positive test with a characteristic history is highly suggestive, although quantitative screening using spectrophotometric or fluorimetric techniques is ideally the best. As this facility was not available to us and the patient was unable to afford it we were not able to do conduct a definitive diagnostic test. We were also not able to do a Woods lamp examination of the eyes or urine because of lack of the facility.

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Acute Hemorrhagic Edema of Infancy

Acute hemorrhagic edema of infancy (AHEI), a dramatic but benign and self-limiting small vessel vasculitis, was first described by Snow in the US in 1913(1).

A 7 month-old male was admitted with sudden onset edema of dorsum of hands and feet, multiple spontaneous ecchymotic spots over face and limbs and low-grade fever. Before he presented to us, the child had undergone incision and drainage of the dorsum of the foot on 2 occasions, the fluid drained was sero-sanguinous. Examination revealed edema of hands and feet (**Fig.1**) and swelling of interphalangeal joints. Over the next 2-3 days edema and ecchymosis almost disappeared without any specific therapy. Another 4 days later, he again developed



FIG.1 Edematous lower limbs with scars of surgery. Note also the swelling of the interphalangeal joints of the great toes.

edema of the whole right lower limb and dorsum of left foot, restricted extension of the right knee joint and circinate skin lesions on the face. A clinical diagnosis of AHEI was considered at this stage.

Laboratory investigations showed progressive anemia (hemoglobin 6.3g/dL), rising ESR (up to 60mm/h) with thrombocytosis ($13 \times 10^3/\mu\text{L}$). Other investigations to rule out mimicking conditions were normal. Skin biopsy from lesion on face showed luminal narrowing and destruction of the elastic lamina of the small and medium sized arteries. Parents were reassured and sent home without specific therapy. After a waxing and waning course over 4-6 weeks, the child improved.

AHEI is a distinctive cutaneous, small vessel leucocytoclastic vasculitis. It is common in males without racial predilection, usually benign and without sequel and with spontaneous recovery within 1-3 weeks(2,3). It presents at 4-24 months and occurs during winter(4,5). It is usually preceded by viral infections, drug intake or vaccination(2). Clinical findings develop rapidly over 24-48 hrs. The two primary features include (i) large cockade (rosette or knot of ribbons), annular or targetoid purpuric lesion found primarily on face, ears and extremities, and (ii) non-tender edema of the limbs and face. It may be asymmetric. Low-grade fever is common(2). Recurrent episodes may occur. Arthritis, nephritis, abdominal pain, GI bleeding and lethal intestinal complications are rare(2). The entity should be differentiated from erythema multiforme, HSP, and (iii) meningococemia(2).

A slightly prolonged course of the disease with anemia, thrombocytosis and elevated ESR were unusual features in our patient.

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