CASE REPORTS

CONGENITAL ERYTHROPOIETIC PORPHYRIA ASSOCIATED WITH VENTRICULAR SEPTAL DEFECT

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A case of congenital erythropoietic porphyria associated with ventricular septal defect in a 6-year-old girl is reported.

Key Words: Porphyria, Congenital erythropoietic porphyria, Gunther's disease, Ventricular septal defect

Introduction

Congenital erythropoietic porphyria (CEP) or Gunther's disease is an extremely rare autosomal recessive inborn error of metabolism, also due to gene mutation. CEP is characterised by mutilating cutaneous photosensitivity and abnormal haem-synthesis in bone marrow. Reduced activity of uroporphyrinogen III synthase causes this disease. The clinical manifestations are red coloured urine, photosensitivity, mutilation, hypertrichosis, reddish or pinkish discolouration of the teeth, finger nails, sclera[1] and stunted growth. Of the internal manifestations, hepatosplenomegaly, hemolytic anaemia, nephrotic syndrome, renal siderosis [2] and osteodystrophy [3] have so far been reported. Adult onset CEP can present with thrombocytopaenia.[4] We report a case of Gunther's disease in a girl who had associated VSD.

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Case Report

A 6-year-old girl born of consanguinous parents was seen for blisters occurring over the sun exposed areas and dyspnoea since early childhood. The mother gave a

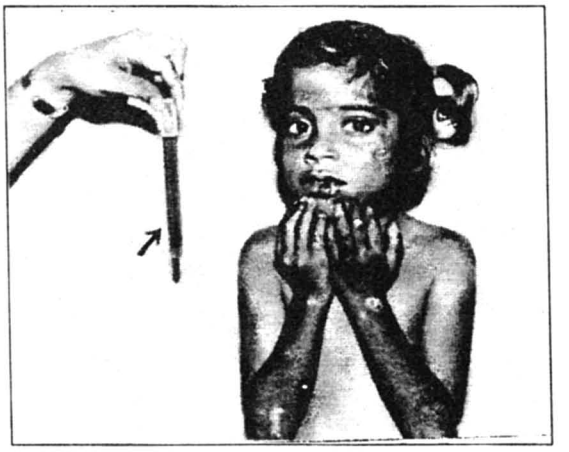

Fig.1. Congenital erythropoietic porphyria associated with VSD

history that the child has been passing red coloured urine since infancy. Bullae occurred over the sun exposed areas which healed with scarring and pigmentary changes. Dyspnoea was exertional and there was no palpitation, syncope or cyanosis. There was no history suggestive of

other systemic involvement.

On examination, the child was moderately built showing hypertrichosis over the face and extremities (sun exposed sites). Lower lip showed ulcers covered by haemorrhagic crust (Fig.1). There were vesicles, ulcers, milia, scarring and post inflammatory pigmentary changes over the sun exposed parts of the body. There was nail dystrophy and mild absorption of the left fifth terminal phalanx. Skin over the covered sites were apparently normal. Systemic examination revealed ventricular septal defect (VSD). With this picture a clinical diagnosis of CEP with VSD was made. She was treated with antibiotics, vitamins, topical sun screen and was advised to avoid exposure to sunlight.

Investigations revealed poikilocytosis, reddish fluorescence of the urine for porphyrins. Biochemical tests were positive for urinary porphyrins. X-ray chest revealed cardiomegaly and ECHO revealed perimembranous VSD. Ultrasound abdomen and liver function tests were normal. Skin biopsy showed histopathological features compatible with porphyria.

Discussion

Congenital erythropoietic porphyria is a rare photosensitive genodermatosis affecting childhood. History of passing red coloured urine since infancy is very characteristic. An early diagnosis can be confirmed by simple biochemical test and Wood's lamp examination. Therapy is very vital in preventing mutilation which is irreversible. In such families genetic counselling and prenatal diagnosis will help in detecting the possibility of porphyria in utero itself. This case is reported for its rare occurrence. Its association with VSD has so far not been reported in literature to our knowledge. Though this association could be a chance occurrence, possibility of mutation affecting the gene responsible for both disorders cannot be excluded. In addition, as the phenotypic expression of an offending gene is variable from ' forme fruste' to 'full-fledged' form such association with ventricular septal defect must be looked for in cases of congenital erythropoietic porphyria in future for its significance.

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