CONGENITAL ERYTHROPOIETIC PORPHYRIA (GUNTHERS DISEASE)

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Summary

Two cases of congenital erythropoietic porphyria occurring in two muslim siblings and seen for the first time in Kashmir are reported.

Congenital erythropoietic porphyria is a rare disease[1]. Two siblings suffering from this disease, seen in Dermatology department of Medical College, Srinagar are being reported.

Case Reports

Case No. 1

7 years old muslim male child developed vesiculobullous eruption on the face, dorsae of hands, feet, forearms and legs during first fortnight of life. Remissions and recurrences were frequent, since then. The lesions were more severe during summer months. Some lesions became purulent and healed with scars and hyperpigmentation. Mother had also noticed reddish brown urine and pinkish teeth in the child from very young age. He suffered from frequent respiratory tract infections.

One of patient's two sisters had similar disease (reported below). Father had a history of pulmonary tuberculosis (treated). Parents were first cousins. Past three generations did not record any similar disease.

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Received for publication on 16—9—1975

Examination revealed scattered bullous lesions and a few purulent and crusted lesions on the exposed areas of body and the knees. Areas of hyperpigmentation and scars of healed lesions were also present on exposed areas of body. Patient had slight hypertrichosis of eyebrows and eyelashes. The deciduous and some permanent teeth were reddish brown in colour. Liver was just palpable, soft and non-tender. Spleen was enlarged to 1 cm below the costal margin, soft and non-tender.

Investigations

Hemoglobin of 10 gms%, white blood cell count, 14800/cmm, N 47%, L 32%, E 19%, M 1% and B 1%. Platelet count 300,000/cmm, Red blood cells count 3.9 million/cmm, Packed cell volume 28%, Reticulocyte count 9%, ESR 18mm first hour (Wintrobe's method). Urine showed increased Urobilinogen. Liver function tests revealed total proteins of 6.4 gm% (Albumin 3.5 gm%, Globulin 2.9 mg%) Thymol turbidity 4 units serum alkaline phosphatase 8 KA units, serum bilirubin .4 mg%. Bone marrow showed a normoblastic reaction. The myelocytic series showed increased eosinophilic precursors. No megaloblasts were seen. X-ray chest showed old healed calcified spots in hilar region.

Case No. 2

4 years old muslim female, younger sister of case 1 also developed eruptions similar to that of her brother, during first month of life. Remissions and relapses were frequent with eruptions more severe and widespread in summer. Examination showed bullous eruptions and areas of hyperpigmentation and scars on exposed parts of body, slight hypertrichosis of eyebrows and eyelashes and erythrodontia. Systemic examination revealed liver just palpable and spleen 3 centimeters below costal margin, soft and non-tender.

Investigations

Blood Hb.9 gm% WBC count 10,800/cmm, P 48%, L .36%, E 1 % M 2%, Platelet count: 360000/cmm, RBC count 3.3 million/cmm, PCV 26%, Reticulocyte count 10% and ESR 62 mm first hour. (Wintrobe's method). Urine showed increased urobilinogen. Stools–Ova of ascaris lumbricoides, Liver function tests total proteins 8.0 gm% (Alb. 3.6 gm%, Glob. 4.4 gm%), Thymol turbidity 8 units, serum alkaline phosphatase 7 KA unit, serum bilirubin 0.8 mg%. Bone marrow showed a normoblastic reaction. No other abnormality was seen. Porphyrin studies of both patients are as in Table 1.

TABLE 1

	Urinary Uroporphyrins micrograms per litre	Urinary Coproporphyrins micrograms per litre
Case I	5059.7	8680.0
Case II	4816 9	14614.7

Table indicating Urinary Uro and Copro porphyrins.

Discussion

Congenital erythropoietic porphyria, an exceedingly rare hereditary disease was first reported in 1874[2,3]. A total of 86 cases of this disorder including 21 from India have been reported in the World Literature uptill 1972[4], with an additional case by Seervai et al. In India first report of this disorder was in 2 siblings reported by Taneja et al[5]. An analysis of available case reports has revealed no significant racial or ethnic group being prone to this disease. However in India cases have been reported mostly from Bengali Brahmins and Sikhs[6] with striking absence among muslims where consanguinity is frequent. Both sexes are equally affected[4,7]. The age of onset was the first year of life in about 67 percent. In 30 percent cases it was within first month of life[4]. One of the patients however experienced the first symptom at the age of 54 years[8]. Parental consanguinity has been reported and[5,9-13] so the occurence of the disease among siblings suggested an autosomal[5,11,14-19] recessive mode of transmission. Among 86 cases reported till 1972 Anaemia, Splenomegaly, erythrodontia and consanguinity was reported to be present in 76, 81, 76 and 14 percent cases and absent in 13, 8, 1 and 44 percent respectively[4].

In congenital erythropoietic porphyria about 50% of developing erythroblasts do not convert porphobilinogen to urogen III. Instead, Urogen and Coprogen I are formed in excess which are excreted in urine and stool. Only prophobilinogen (PBG) deaminase is required for conversion of PBG to Urogen I whereas Urogen Isomerase is required in addition to deaminase for its conversion to Urogen III. Thus the defect probably lies in an excessive amount or activity of PBG deaminase or a deficiency or defect in Urogen Isomerase[7].

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