

## CASE REPORT

# Congenital Erythropoietic Porphyria Without Hemolysis: A Case Report

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## ABSTRACT

**Introduction:** Porphyrrias are a group of disorders caused by deficiency of the enzymes in heme biosynthetic pathway. Congenital erythropoietic porphyria “Gunther’s disease” is an extremely rare disease with less than 200 cases documented worldwide. It is an autosomal recessive disease with mutation in the gene that codes for uroporphyrinogen III synthase. CEP is usually associated with hemolytic anemia.

**Case report:** We herein report a case of two siblings with the typical presentation of photosensitivity, hypertrichosis, mutilation of the fingers, dark-purple urine, and erythrodontia with pinkish fluorescence under a Wood’s lamp but without any feature of hemolysis.

**Conclusion:** Very rarely CEP can present without any feature of hemolytic anemia. We report this case because of rarity.

**Keywords:** Congenital erythropoietic porphyria, Gunther’s disease, Photosensitivity, Erythrodontia

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## INTRODUCTION

Congenital erythropoietic porphyria (CEP) or Günther’s disease is a very rare form of porphyria of autosomal recessive inherited deficiency of the uroporphyrinogen III synthase enzyme.<sup>1</sup> There are different mutations of the gene that code for the UROS. The CF3R mutation is most frequent.<sup>2</sup> The main site of enzyme defect is in the bone marrow. Less than 200 cases have been reported in the literature.<sup>3</sup> The disease usually starts during infancy but occasionally during adulthood. CEP is the most photosensitive and mutilating type of the porphyrias. The diagnosis is made by porphyria profile study and decreased relative enzyme activity. CEP without hemolysis is very rare. Here we report a case of CEP with classical clinical presentation but without any feature of hemolysis.

## CASE REPORT

A 5 years old male child born of a second degree consanguineous marriage presented with complaints of photosensitivity with recurrent blisters on sun exposed areas and reddish colouration of teeth and urine since the age of 4 years. Blisters used to rupture to form erosions in 1 – 2 days with crust formation, crust used to fall off spontaneously in 5-7 days leaving behind areas of hypo/hyperpigmentation. There was h/o worsening of cutaneous symptoms during summer season. No history of other systemic symptoms or acute neurological attacks was present. Milestones were appropriate for age. The patient was not on any prior medication. Similar history and examination findings were present in his 9 years old elder brother.

On examination the patient was alert and intelligent. He had mild pallor and weighed 15.3 kg. The exposed parts of the body showed generalised hyperpigmentation with a few crusted

lesions, areas of hypo/hyper-pigmentation and atrophic scarring (Fig. 1). Hypertrichosis (werewolf appearance) was also prominent over the exposed areas (Fig. 2). There was mutilation of right index finger and contractures of the fingers and toes photo induced onycholysis. Milia were present over the dorsum of hands. There was red discoloration of teeth and urine (Fig 3). Systemic examination was normal and showed no evidence of hepatosplenomegaly.

Investigations included wood's lamp examination, routine haematological and biochemical tests and the urine porphyrin tests that included PBG qualitative random urine ; quantitative analysis of total porphyrin in 24 hours urine. Wood's lamp examination showed characteristic pink red fluorescence in teeth (erythrodonia) and urine. Routine investigations : Hb - 9.2 g/dl, TLC - 8,200/cumm, Platelet count - 2.9 lac/cumm, ESR - 22 mm/hr, Peripheral Smear showed normocytic, normochromic anemia, Reticulocyte count was 1.4% of circulating erythrocytes, Liver and renal function tests was normal. Urine Porphobilinogen - absent, urinary Total porphyrin - 1087  $\mu\text{gm}$  ( $n < 150 \mu\text{gm}/\text{hour}$ ). Exact porphyrin type and isomer identification needs sophisticated equipment like HPLC unavailable in most of the laboratories in India. Hence, the total increased amount of porphyrins was considered and not the exact type of porphyrin.

So, based on the family history, clinical findings i.e. cutaneous photosensitivity , red discoloration of urine and teeth, hypertrichosis, mutilation and laboratory findings i.e. pink red fluorescence under woods lamp, raised total porphyrin in urine, diagnosis of congenital erythropoietic porphyria was made.



**Figure-1:** Photosensitivity



**Figure-2:** Hypertrichosis on photoexposed area



**Figure-3:** Red discoloration of urine

## DISCUSSION

The inheritance of two mutant alleles for the gene encoding the enzyme uroporphyrinogen III synthase leads to accumulation of predominantly type 1 porphyrin in erythrocytes, bone marrow, skin, teeth, bones and other organs that are responsible for cutaneous photosensitivity characterized by blisters, erosions, and scarring of sun-exposed skin.<sup>1</sup> Chronic damage of skin, cartilage, and bones can cause mutilation of acral tissues. Hypertrichosis, alopecia, and reddish-colored urine are often present. Erythrodonia (red fluorescence under ultraviolet light) when present is virtually pathognomonic of CEP.<sup>4</sup>

Clinical manifestations can vary from mild to severe grade, starting from non-immune hydrops foetalis as a result of severe hemolytic anemia in-utero to late onset clinical cases where the only symptom is cutaneous photosensitivity in adulthood. Hemolytic anemia can be mild or severe, with resultant splenomegaly and osseous fragility. The clinical manifestations are markedly variable due to the different mutation in

the UROIII gene.<sup>5</sup> In our case there was cutaneous photosensitivity along with cutaneous atrophic scarring, hypertrichosis, acral mutilation red-colored urine and teeth. But, strikingly there was no evidence of hemolysis.

In the past most patients died by the age of 40 years but improvement in supportive care has improved the prognosis. The only available control measure is total avoidance of sunlight. Topical sunscreen lotion, oral beta-carotene are the treatment options. Splenectomy for intractable hemolytic anemia may be required and has occasionally resulted in marked improvement both in anemia and in cutaneous photosensitivity.<sup>6</sup> Hypertransfusion with packed cells are helpful as they suppress erythropoiesis and depress the production of porphyrins. Stem cell transplantation is the only permanent curative option. In our case, we advised strict avoidance of sunlight and prescribed topical sunscreen lotion of high Sun protection factor with an oral antihistaminic, antibiotics and beta-carotene (90 mg/day). But, CEP without hemolysis is very rare. In our case there was no feature of hemolysis. This unique presentation encouraged us to report such a rare case.

## CONCLUSION

Photosensitivity reactions associated with blister formation are rare in Indian children. High index of suspicion is needed to rule out CEP. There are very few case reports of CEP in Indian literature till date. Very rarely CEP can present without any feature of hemolytic anemia.

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