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**Case Report**

# Pregnancy in porphyria and its complications: a case report

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## ABSTRACT

As with many rare diseases, little is known about the porphyrias and reproduction. Knowledge pertaining to complications in pregnancy in a patient with porphyria is even more scant. The information which we have is from attending on individual cases. The following case report demonstrates a rare case of porphyria cutanea tarda in a 30 year old female patient, diagnosed around pubertal age, who presented to the Emergency department as a case of severe anemia who had delivered a still born baby the same day. Still births along with spontaneous abortions, preeclampsia and low birth weights are few of the known complications of pregnancy in patients with porphyria.

**Keywords:** Porphyria cutanea tarda, Acute intermittent porphyria, Stillbirth, Preeclampsia, Pregnancy

## INTRODUCTION

The Porphyrias are a group of rare hereditary metabolic disorders, resulting from defective porphyrins, which are enzymes involved in heme synthesis. Depending on the specific enzymes, they can manifest as acute or chronic, with symptoms being predominantly cutaneous, neurologic, psychiatric or combination of these. One of the common subtype of porphyria is porphyria cutanea tarda (PCT) with a prevalence of approximately 1 in 10,000, caused by deficiency of uroporphyrinogen decarboxylase (UROD) which primarily presents with skin manifestations later in life. Other types include acute porphyrias like acute intermittent porphyria, hereditary erythropoetic porphyria, variegate porphyria, have been implied to be risk factors for low birth weight, growth retardation, premature delivery, spontaneous abortion, and perinatal death[1,2,3,4].

Our case report discusses about one such complication in pregnancy, perinatal death, in a patient with PCT, who delivered a preterm still birth.

## CASE REPORT

A 30 year old female patient, known case of porphyria, presented in casualty with severe anemia who had delivered a still born few hours before in a private hospital on the same day. Patient had edema feet since 15 days. Patient had no complaints of generalized weakness or breathlessness. Patient also had no history of pregnancy induced hypertension in the recent pregnancy. However, patient was a known case of hypothyroidism for which she was on treatment. Patient conceived spontaneously after 2.5 years of marriage, with no history of any abortions.

Patient was diagnosed as a case of porphyria cutanea tarda since the age of 11 years, around her pubertal age and had been on treatment for the same, with immunomodulators and steroids. Patient received three doses of injection iron sucrose during her antenatal care period as she was anemic.

During her pregnancy, patient gave history of increased blister formation over extensor regions of bilateral lower limbs and upper limbs, which subsided soon after

delivery. Patient's elder sister also suffered from porphyria but of less severe form, and she had two healthy children with uneventful pregnancies.

On examination, patient was conscious, oriented but irritable. Patient's blood pressure was 120/80 mm of Hg and pulse 90 per minute. Pallor was present. Facial hair was present. Hyperpigmented skin patches were present all over the body including face, with scarring seen.

Respiratory system examination was normal. Cardiovascular examination revealed a pansystolic murmur of grade III in the tricuspid area. Abdomen was soft and uterus was well retracted with minimal per vaginum bleeding.

Patient was further evaluated and a dermatology opinion was taken, where the diagnosis of porphyria cutanea tarda was confirmed. Patient had diffuse thickened hypersegmented skin over extensor aspect of bilateral upper and lower limbs with scar with milia formation. The scalp, oral cavity, palms and soles had no abnormality. There was evidence of blister formation followed by scarring over sun exposed parts. Patient was advised strict sun protection with application of ultraviolet sunscreen. Various investigations were advised like urine and teeth porphyrin under woodlamp's, serum porphyrin levels, ophthalmology opinion and hand X-ray. 2d echocardiography was done which was suggestive of congenital heart disease with 12 mm and 14 mm os atrial septal defect (primum to secondum) with moderate TR and moderate PAH with intact interventricular septum and normal pericardium. Intensive cardiology rehabilitation was advised.

Renal scan and renal Doppler were within normal limits while ultrasonography of Abdomen was suggestive of moderate splenomegaly. Laboratory studies included hemoglobin 5.2 gm% which increased upto 7.8 gm% after two packed red blood transfusions. WBC and platelet counts were within normal limits; total bilirubin was raised to 1.3 mg/dL; alkaline phosphatase, aspartate transaminase, alanine transaminase, serum urea, serum creatinine and serum electrolytes were within normal limits. Old reports showed raised serum porphyrin levels to 220 nmol/L (<15 nmol/L) and raised porphyrins in urine, mainly uroporphyrins at 360 nmol/L (<140 nmol/L).

Patient received two pint of whole blood transfusions to build up her hemoglobin and was started on multivitamin injections daily, tablet vitamin C 500 mg OD, tablet ferrous sulphate 200 mg BD, tablet calcium 500 mg TDS and was advised strict sun protection with local application of UV protection sunscreen. Patient was advised to follow up in dermatology OPD and cardiology OPD for further line of treatment and was counseled regarding the complications that would occur with subsequent pregnancies in future.

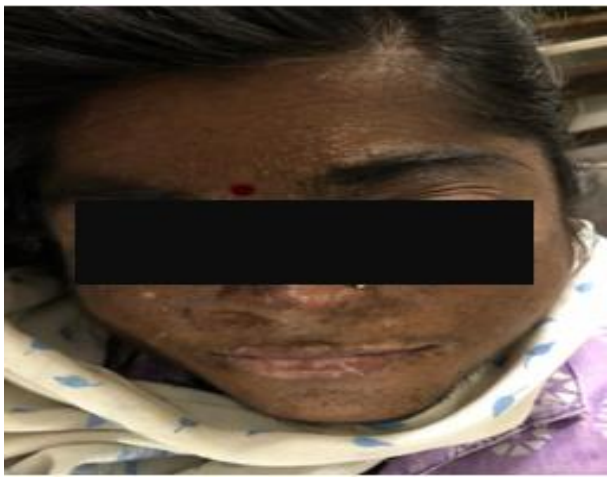

**Figure 1: Hyperpigmented patches with milia and hypertrichiosis on face.**

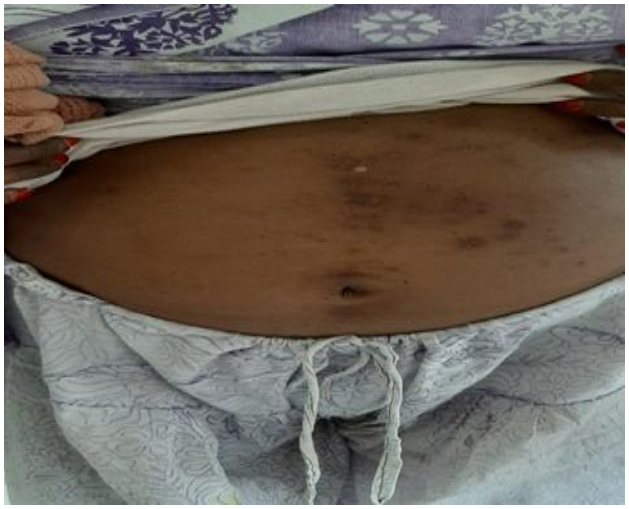

**Figure 2: Scarring seen over abdomen.**

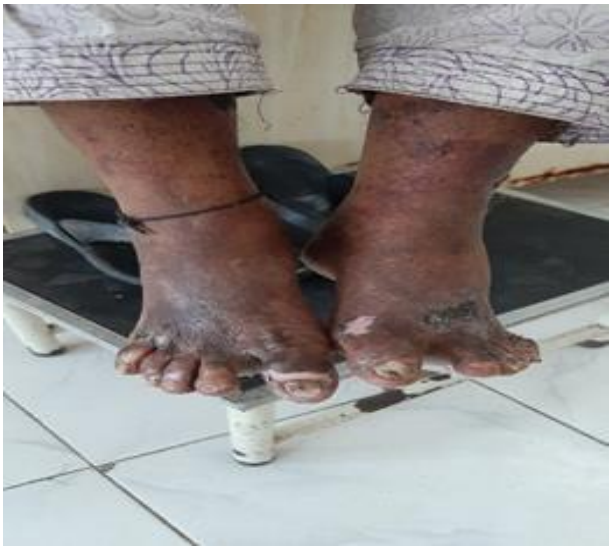

**Figure 3: Scleroderma like plaques seen over bilateral feet.**

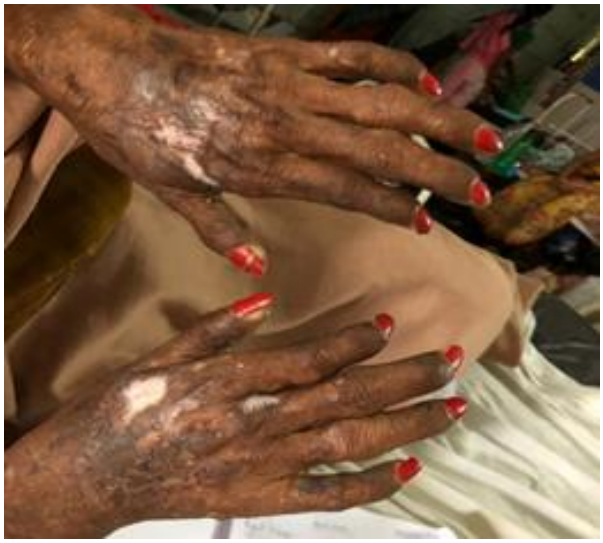

**Figure 4: Diffuse thickened patchy skin over extensor surface of hands.**

## DISCUSSION

This case shows one of the few complications of porphyria cutanea tarda in pregnancy, which resulted in still birth. Just as pregnancy can be complicated by porphyria, similarly the increased levels of progesterone during pregnancy can itself precipitate porphyria.

Information about pregnancy in porphyria is scarce. Information pertaining to complications in pregnancy in a patient with porphyria is even more scant. The information we have is from attending on individual cases. One such case report was of a female patient suffering from acute intermittent pophyria (AIP), who had presented with missed abortion of 7 weeks, depicting one of the complications of pregnancy in porphyria.[5] Other published case reports on four pregnancies by three women with active AIP reported that all four infants were born small for gestational age (SGA) and one died shortly after.[2,6] It was also reported that of four pregnancies by a woman with variegate porphyria, two infants were SGA and two died perinatally.[4] Brodie et al reported that babies born after an acute attack had occurred during pregnancy had lower birth weight than if no acute attack had occurred, but is not apparent whether this difference was due to in utero growth retardation or premature delivery.[1] A publication bias toward pregnancies with adverse outcomes is to be expected in case reports. Clinical features develop due to build up of porphyrins in our body, negatively affecting the skin or the nervous system. The most common photo cutaneous manifestations of porphyria, especially observed in PCT are caused by a buildup of porphyrin compounds (specifically uroporphyrinogen) under skin oxidized by free radicals or sunlight leading to type IV hypersensitivity reaction. These cause erosions and blisters in exposed areas of the skin which form painful indolent sores that heal with milia, dyspigmentation, and scarring. Other features include hypertrichosis, scleroderma like plaques that may develop dystrophic calcification and excretion of discoloured urine.

Diagnosis of porphyria is confirmed based on porphyrin studies with biochemical analysis of blood, urine and stool; urine estimation of porphobilinogen (PBG) in acute cases. During pregnancy in female patients, prenatal diagnosis can be done, by measuring porphyrins in amniotic fluid and amniotic cell culture.[6,8] Cord blood can be tested to determine the inheritance. Treatment of porphyria is symptomatic and oriented to improving skin conditions and clinical manifestations. It is important to avoid precipitating factors like estrogen, valproic acid, barbiturates, sulfonamides and hydantoins. Other factors involved in acute crisis are alcohol, hypocaloric diets, and infections.[6,7] Acute attacks of porphyria respond well to treatment with heme arginate, given in a dose of 2-3 mg/kg/day during four consecutive days administered in slow infusion. But effects on fetus are unknown hence this treatment is not recommended during pregnancy.[6,7,9,10]

Use of contraceptives is controversial, as its progesterone component is known to provoke acute attacks. The most recommended is barrier method of contraception, especially in our case as oestrogen based preparations can make active PCT symptoms worse.

## CONCLUSION

To sum up, pregnancy in porphyria is a high risk pregnancy, and it is important to have frequent ANC checkups for maternal well being with regular ANC Doppler to monitor fetal growth, and if need be early induction to prevent complications like spontaneous abortions, preeclampsia and perinatal loss.

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