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# PORPHYRIA: A CASE STUDY

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**A B S T R A C T**

Porphyria is a group of liver disorders in which substances called porphyrins build up in the body, negatively affecting the skin or nervous system. The types that affect the nervous system are also known as acute porphyria, as symptoms are rapid in onset and short in duration. Symptoms of an attack include abdominal pain, chest pain, vomiting, confusion, constipation, fever, high blood pressure, and high heart rate. The attacks usually last for days to weeks. Complications may include paralysis, low blood sodium levels, and seizures. Attacks may be triggered by alcohol, smoking, hormonal changes, fasting, stress, or certain medications.If the skin is affected, blisters or itching may occur with sunlight exposure.Most types of porphyria are inherited from one or both of a person's parents and are due to a mutation in one of the genes that make heme. They may be inherited in an autosomal dominant, autosomal recessive, or X-linked dominant manner. One type, porphyria cutanea tarda, may also be due to hemochromatosis (increased iron in the liver), hepatitis C, alcohol, or HIV/AIDS. The underlying mechanism results in a decrease in the amount of heme produced and a build-up of substances involved in making heme. Porphyrias may also be classified by whether the liver or bone marrow is affected

***Keywords****: Porphyria, Delta-aminolevulinic acid (ALA) and porphobilinogen (PBG)*

## 1. INTRODUCTION

Porphyria refers to a group of disorders that result from a buildup of natural chemicals that produce porphyrin in your body. Porphyrins are essential for the function of hemoglobin — a protein in your red blood cells that links to porphyrin, binds iron, and carries oxygen to your organs and tissues. High levels of porphyrins can cause significant problems[2]. There are two general categories of porphyria: acute, which mainly affects the nervous system, and cutaneous, which mainly affects the skin. Some types of porphyria have both nervous system symptoms and skin symptoms.

Porphyrias have been subdivided into hepatic and erythropoietic forms, according to the site of expression of the dysfunctional enzyme. In a clinical approach, it is much more convenient to classify them according to their clinical manifestations—as acute (neurovisceral) versus non-acute (cutaneous) porphyrias. In some of the acute porphyria types both neurovisceral and cutaneous symptoms may be present[2].

The acute neurovisceral forms are characterized by overproduction of delta-aminolevulinic acid (ALA) and porphobilinogen (PBG), which are porphyrin precursors, at the initial steps of heme synthesis (while the cutaneous ones are characterized by accumulation of porphyrins, which are the precursors at the final steps of the synthesis. This fundamental difference is the basis of the different clinical symptoms.[3].

## 2. CASE PRESENTATION

A 18 years old female student visited IGMC, Shimla medicine department with the complaints of abdomen pain, vomiting( multiple episode ), she was experiencing these conditions from last 2 days. After hospitalization few days later abnormal body movement and altered sensorium present.

**Past Medical History:**

There was not significant history of any past medical illness.

**General Examination**

Weight: 49 Kg

Height: 160 cm

BMI: 19.14kg/m2

**Special investigations:**

Blood culture, urine culture, PFT/LFT, MRI Brain.

## 3. TREATMENT

Inj. Doxycycline 200mg Iv stat, Inj. Pantop 40 mg Iv OD, Inj. Phenytoin 1gm Iv, Inj ceftriaxone 2 gm Iv BD, Inj. Perinorm 10mg Iv OD.

## 4. INTERVENTION

**Supportive care:**

Patient put on mechanical ventilator(SIMV mode, tidal volume-320, PEEP- 6, Fio2-60%).

IV fluids was given ; NS-RL-NS

1unit PRBC administered to patient.

## 5. CARE PLAN

- Physical activity/Passive exercise-
- Educate family members regarding passive exercise, change the position every 2nd hourly and back care for increasing circulation of body.
- Eat high protein diet – skim or low- fat milk, yogurt, fat free or low- fat cheese and eggs.
- Increased salt intake

## 6. OUTCOME

- After supportive care pateint was on oxygen support .
- Educate family member to give timely medication to the patient.
- Educate family member that proper care has to be given like back care.
- Family member was advised to visit hospital after one month for follwup.

## 7. DISCUSSION

Porphyria a rare hereditary disease in which there is abnormal metabolism of the blood pigment haemoglobin. Porphyrins are excreted in the urine, which becomes dark; other symptoms include mental disturbances and extreme sensitivity of the skin to light. Prevalence of porphyria approximately 1 in 20,000 and the prevalence of the most common erythropoietic porphyria, erythropoietic protoporphyria, is estimated at 1 in 50,000 to 75,000. clinical features include- Pain areas: in the abdomen, Gastrointestinal: constipation, nausea, or vomiting, Skin: rashes, blister, or darkening of the skin, Muscular: cramping or muscle weakness, Sensory: pins and needles or sensitivity to light, Whole body: nervous system dysfunction or water-electrolyte imbalance, Also common: anxiety, blood in urine, fast heart rate, itching, mental confusion, or seizures. If we talk about pathophysiology when enzyme of the heme synthesis is deficient, its substrate and any other heme precursors normally modified by that enzyme may accumulate in bone marrow, liver, skin or other tissues and have toxic effect. Diagnostic test for porphyria is genetic testing of a blood sample. This type of test is very accurate. Treatment of porphyria blood transfusion, dietary, supplementary and antibiotic therapy. This case shows that better outcome as per the treatment protocols followed.

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