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Case Report

Gunther's Disease

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Abstract

60 years female patient unmarried Resident of remote mountainous region of northern Pakistan is admitted via OPD as a case of leprosy with nasal septum deformities and bilateral upper limbs digits deformities.

So This patient was admitted for workup of Anaemia , persistent hematuria , Acute kidney injury and shortness of breath and abdominal distention .

General physical examination shows thin pale ill looking female with red dry eyes and increased facial hair along with prominent and elongated canine teeth with nasal septum deformity and digits auto amputation bilaterally, with gross ascites, Bilateral pitting pedal oedema, reddish urine and low Blood pressure with Anaemia. Systemic examination showed splenomegaly, with ascites and bilateral decreased air entry at bases of the lungs. Echocardiography show cor pulmonale with RVSP of 54 and abdominal ultrasound showed splenomegaly. Creatinine was 3.4 with Hb of 8.9 and ALT of 212 with SBR of 3.2.

Dermatology department was consulted for opinion and they advised to consider a very rare disease CEP as the primary diagnosis since the features were all matching with this entity. Finally diagnosis was confirmed to be CEP by clinical signs and raised uroporphyrin level 1 and coproporphyrin 1. Unfortunately we lost the patient secondary to septic shock in spite of good antibiotic coverage and inotropic support as patient was having persistent Hypotension with worsening AKI.

The unique presentation of this case in the form of splenomegaly, Ascites, Anaemia, Nasal septum and digits amputation along with positive childhood history for blisters according to the family and presumptive diagnosis of leprosy made it a challenging case well deserving to be in the category of rarest medical case in our region. Similarly it also highlights the positive role of thinking in internal medicine diagnosis as well as the value of interdepartmental consultation that could well save the resources and improve patient care at tertiary level. The lesson learned in this case is to promote awareness about negative aspects of consanguinity which is practiced at much higher frequencies in our community in-order to avoid congenital diseases.

Keywords: Gunther's Disease, coproporphyrin, Erythrodontia.

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Introduction

Congenital erythropoietic porphyria (CEP or Gunther's disease) is an extremely rare subtype of the non acute group of porphyria ,with an estimation of <0.9 in ten million people.[1] Similarly all organs are vulnerable to the porphyrin accumulation effects, but the most affected ones are the hematopoietic system, teeth, skin, bones and sclera. CEP affects all ethnic groups and so far only 200 cases have been reported worldwide. The age of onset and clinical severity of CEP are highly variable, In most cases, severe photosensitivity develops soon after birth. Pink or red brown staining of diapers due to large amounts of urinary porphyrins may be the first clue to the disease in the infant.[2] The major clinical features are cutaneous photosensitivity, anemia. Exposure to sunlight causes a vesicular eruption that can lead to erosions, infections and scarring. Mutilation of light exposed areas such as nose, Digits and ear may also occur. Anemia due to hemolysis can also be severe if the bone marrow doesn't compensate and some individuals may become transfusion dependent. Splenomegaly is secondary to hemolysis, and the liver can be enlarged secondary to iron overload. Erythrodontia, a pinkish brown discolouration of teeth due to porphyrin deposition is almost path-gnomic sign and that's why it is also called vampire disease due to typical appearance

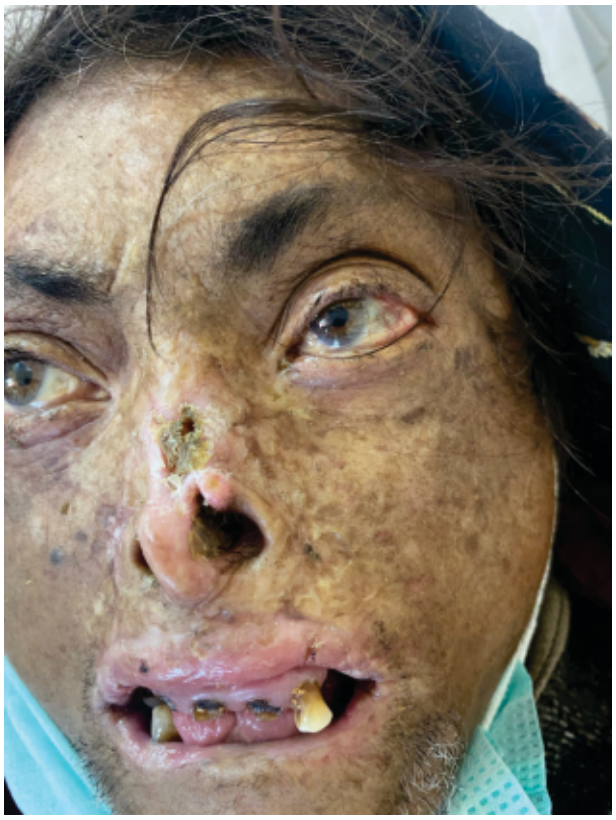
of the teeth on red fluorescence under ultraviolet light.[3]

Diagnosis is usually confirmed through clinical examination, age of onset of the disease and raised level of uroporphyrin 1 and coproporphyrin1.[4] Treatment of CEP includes avoidance of sunlight, protective clothing, barrier skin creams ,along with red blood cell transfusion and hematin administration. Allogeneic bone marrow transplantation have proved curative in some limited cases.[5] Our case is unique in the sense that a false diagnosis of leprosy due to nasal septum mutilation and worsening clinical parameters of the patient made us to re-evaluate the initial diagnosis in a limited span of time.

Case Description

We had this 60 years old female patient unmarried Resident of remote mountainous region of northern Pakistan who was admitted via OPD as a case of leprosy with nasal septum deformities and bilateral upper limbs digits deformities.

The very first look of the patient would led you to the



diagnosis of Leprosy but it never was the final diagnosis because of very unusual presentation not present in leprosy disease.

So This patient was admitted for workup of Anaemia, persistent hematuria, Acute kidney injury and shortness of breath and abdominal distention.

General physical examination shows thin pale ill looking

female with red dry eyes and increased facial hair along with prominent and elongated canine teeth with nasal septum deformity and digits auto amputation bilaterally, with gross ascites, Bilateral pitting pedal oedema, reddish urine and low Blood pressure with Anaemia. Systemic examination showed splenomegaly, with ascites And bilateral decreased air entry at bases of the lungs. Echocardiography show cor pulmonale with RVSP of 54 and abdominal ultrasound showed splenomegaly. creatinine was 3.4 with Hb of 8.9 and ALT of 212 with SBR of 3.2.

The presence of splenomegaly, jaundice, ascites, hematuria that was never picked in labs by urine analysis and shock due to low BP led to the suspicion of reconsidering the diagnosis as leprosy clinical presentation was not matching in this case .Also the urinary catheter was repeatedly checked but the reddish coloration never vanished and urine analysis was always negative for Red blood cells.so finally dermatology department was consulted for opinion and they advised to consider a very rare disease CEP as the primary diagnosis since the features were all matching with this entity. Finally diagnosis was confirmed to be CEP by clinical signs and raised uroporphyrin level 1 and coproporphyrin 1. unfortunately we lost the patient secondary to septic shock in spite of good antibiotic coverage and inotropic support as patient was having persistent Hypotension with worsening AKI.

Labs

WBC	18.5*10
HB.	8.9
Platelets.	160*10
Creatinine.	3.4
Urea.	105
ALT.	212
SBR.	3.2
Uroporphyrin1.	5388
Coproporphyrin1	3211

Discussion

This case as described above in detail highlights the importance of early interventions in the form of promotion, prevention, treatment and avoiding complications while dealing a congenital rare disease. The misleading diagnosis of Leprosy complicated this case at first as we were treating the patient in the context of leprosy but the continuous red colour discolouration of urine with no proven urinary tract trauma or infections along with splenomegaly compelled us to search for alternate diagnosis which we eventually succeeded with help from dermatology department and a well proven final diagnosis of CEP in this patient explained all our queries of the underlying cause of splenomegaly, vampire teeth, Hemolytic anaemia and urine discolouration in this particular case. Later on the previous medical history of the patient was confirmed through a well educated family elder of that patient giving positive clues of her continuous symptoms since childhood thus making this case a well investigated case of CEP.

Lessons learned

Timely interdepartmental consultation is a key to success while diagnosing rare cases.

Community awareness regarding medical drawbacks of consanguinity in our region must be improved.

Patient and their family awareness about all congenital diseases must be ensured so that patient can be protected from developing complications from the said disease.

A definitive cure is still awaited for this rare disease and stem cell transplantation can be a breakthrough if well researched for CEP.

Conflict of Interest: *None*

Funding Source: *None*

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