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Beta thalassemia trait with priapism: A unique presentation

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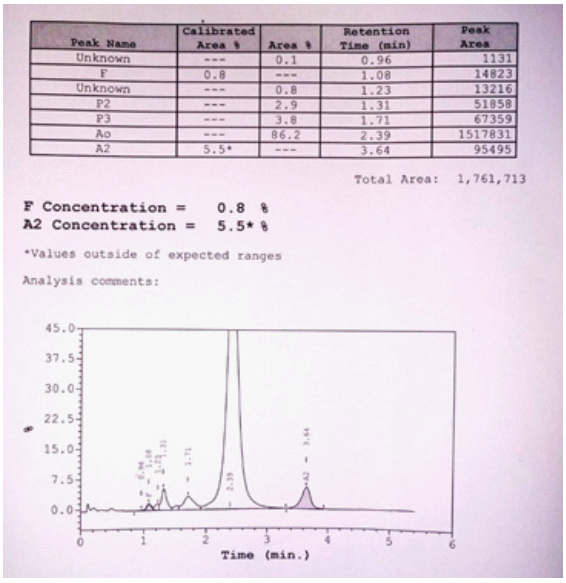
Introduction

Beta thalassemias are characterized by decreased to absent beta globin chain synthesis. These are autosomal recessive disorders caused commonly due to point mutations in the beta globin gene on chromosome 11. Rarely there maybe deletions in the beta globin genes [1]. Reduced beta chains lead to excess of unbound alpha chains which form homotetramers. Alpha tetramers are highly unstable and precipitate as inclusion bodies which damages RBCs causing ineffective erythropoiesis [2]. Beta thalassemias can be broadly categorized as homozygous beta thalassemia major and heterozygous beta thalassemia trait. Children with beta thalassemia major become symptomatic within 2 years of life and require regular blood transfusions while thalassemia trait results in silent carrier state [1]. In India, prevalence of beta thalassemia trait is 2.78% while 10000-12000 children of beta thalassemia major are born yearly [3]. The major complications of beta thalassemia include cardiac arrythmias and failure, liver cirrhosis, and endocrinopathies like growth hormone deficiency and adrenal insufficiency. All of these complications arise due to iron overload resulting from regular blood transfusions [1]. Priapism has been very rarely associated in patients of beta thalassemia. We report a case of beta thalassemia trait patient presenting with priapism.

Case presentation

A 43 year old man presented to urologic emergency with priapism for 8 hours. He had no previous episodes of priapism. There was no history of trauma. No lower urinary tract symptoms/hematuria/fever. There was no association with sexual activity. He had no history of erectile dysfunction or intake of pro erectile drugs. He had taken homoeopathic medications for bilateral upper and lower limbs pain 7 days back. However, he denied intake of any other drugs or alcohol. He had no other significant medical history. On examination, he had tender rigid penile erection. No evidence of pallor or icterus. No cervical/axillary/inguinal lymph nodes were palpable. There was no hepatosplenomegaly. Penile doppler showed bilateral engorged cavernosa with absent blood flow in bilateral cavernosal artery. A diagnosis of low flow priapism was made. Cavernosal aspiration and irrigation yielded dark red blood followed by phenylephrine injection in cavernosa. The patient was then referred to hematology for workup of any underlying hematological cause. He had no history of chest pain/dyspnea/dactylitis. No history of previous blood transfusion. No family history suggestive of hemolytic anemia. On examination, he had no pallor or icterus. No evidence of inflamed small joints. There was no hepatosplenomegaly on per abdomen examination. CBC revealed Hb of 10.3 g/dl, TLC of 10700/dl and platelet count of 1.6 L/dl. PS showed microcytic hypochromic RBCs with occasional target cells. No

evidence of sickle cells. Hb HPLC revealed HbA2 of 5.5%, HbA of 86.2%% and HbF of 0.8%. There was no elution in HbS window. Hence, patient was diagnosed as beta thalassemia trait. He had no further episodes of priapism and is doing well on follow up.



Peak Name	Calibrated Area %	Area %	Retention Time (min)	Peak Area
Unknown	---	0.1	0.96	1131
F	0.8	---	1.08	14823
Unknown	---	0.8	1.23	13216
P2	---	2.9	1.31	51858
P3	---	3.8	1.71	67359
Ao	---	86.2	2.39	1517831
A2	5.5*	---	3.64	95495

Total Area: 1,761,713

F Concentration = 0.8 %
A2 Concentration = 5.5* %

*Values outside of expected ranges

Analysis comments:

Figure 1: HPLC of the patient showing beta thalassemia trait

Discussion

Priapism is characterized by painful penile erection persisting for more than 4 hours. Priapism can be of 3 types: ischaemic, non ischaemic and stuttering. Ischaemic or low flow priapism results due to retention of venous blood in corpora cavernosa with no arterial inflow causing markedly rigid corpora. The tissue ischaemia causes painful priapism. This is a penile compartment syndrome and requires emergency intervention. Non ischaemic or high flow priapism results due to high arterial blood flow into non rigid corpora cavernosa. There is no venous obstruction. Clinically, it leads to painless persistent penile erection. Stuttering priapism, commonly seen in sickle cell disease, causes recurrent episodes of priapism but these episodes are self limiting. Penile arterial blood gas assessment can also be used to differentiate between the types of priapism [4].

Ischaemic priapism is the commonest type with a wide variety of causes, most notably iatrogenic, due to intracavernosal injections for treatment of erectile dysfunction. It can also be caused by an array of hematological conditions like sickle cell disease, G6PD deficiency, thrombophilia, hyperviscosity and anticoagulants. Hence, a thorough hematologic workup is essential in all cases of ischaemic priapism. Even stuttering priapism is characteristic of sickle cell disease. Non ischaemic priapism on the other hand is caused due to trauma affecting the caversonal artery [5].

Among hematological conditions, sickle cell disease is the foremost causing priapism. Upto 35% of men with sickle cell disease are affected by ischaemic priapism [6]. Thalassemia has been rarely associated with priapism. There are as few as nine case reports, which includes thalassemia major as well as intermedia, presenting with priapism, both ischaemic and stuttering type. Recurrent episodes of priapism in thalassemia patients may lead to penile fibrosis and erectile dysfunction [7].

There are two proposed theories delineating the pathophysiology of priapism in thalassemia. A cellular mechanism states that increased blood viscosity and thrombosis may obstruct the venous system of corpora cavernosa causing rigid penile erection. The functional mechanism states that increased NO consumption causing vasoconstriction and decreased phosphodiesterase-5 activity leads to altered vascular homoeostasis and priapism in thalassemia [8]. It has also been observed that thalassemia patients post splenectomy are more predisposed to developing priapism. The underlying mechanism is that post splenectomy thrombocytosis and increased nRBC leads to thromboembolic obstruction of the venules of corpora cavernosa [9].

A literature review of thalassemia with priapism revealed a mean Hb of 9.14 ± 1.32 g/dL. Thalassemia intermedia was slightly more common, comprising of 55% of cases [7]. There has been only one reported case of beta thalassemia trait with priapism [10]. Our patient also a Hb of 10.3 g/dl. However, he has never required any blood transfusion. Priapism is a medical emergency since it may cause tissue ischaemia and necrosis. So the corpora has to be decompressed and arterial blood flow has to be restored to limit tissue injury. The first line management of priapism involves aspiration of inspissated blood followed by irrigation of corpora and intracavernous alpha agonist injection [5]. Refractory cases require surgical shunt procedures. Patients of sickle cell disease maybe additionally managed with hydration, oxygentation, alkalanization and exchange transfusion. Our patient was also managed along similar lines with cavernous phenylephrine injection. His priapism subsided with the procedure. He did not develop any further such episodes.

Conclusion

Priapism has been uncommonly observed in association with thalassemia, especially thalassemia intermedia and splenectomized patients. However, there has been only one reported case of beta thalassemia trait with priapism. We report only second such rare case of beta thalassemia trait associated with priapism as first presentation. Hence, all patients of priapism should be evaluated for underlying hematological conditions.

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