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Case report **Biochemistry**

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A CASE OF SYMPTOMATIC SICKLE CELL TRAIT-A VERY RARE CASE REPORT AND DIAGNOSTIC APPROACH.

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ABSTRACT

Among the various hereditary diseases known, there are a few rare diseases which differ from the others in manifestations. One such example is the Sickle cell trait which is usually asymptomatic. Here, we discuss about a symptomatic case which will lead to a difficulty in diagnosis.

KEYWORDS : HbS, Electrophoresis, IVP

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INTRODUCTION

The haemoglobin present in the red blood cells is a tetramere which is mainly responsible for oxygen transport and its delivery to the tissues . Various structural and functional mal- and dysfunctions of the hemoglobin will result in various clinical syndromes and diseases, which may be hereditary or acquired, one of which is the sickle cell disease. Sickle cell disease is an important hereditary structural haemoglobinopathy, a type of disease characterized by the production of defective hemoglobin. It is inherited by autosomal co-dominant traits. The adult hemoglobin HbA has 2 alpha and 2 beta globin chains. In human alpha chain genes are located on chromosome no:16 and beta type genes clustered on chromosome no:11. Any mutation in the gene leading to an altered amino-acid sequence in the beta globin chain will result in the structural deformity. In a sickle cell, the 6^{th} amino-acid Glutamic acid is replaced by Valine. This results in the sickling of the hemoglobin, hence the name. This sickling is responsible even for functional disabilities of the erythrocytes such as stiffening of their membranes, increased viscosity and decreased elasticity , resulting in various clinical manifestations like vaso-occlusive crises with spleen, brain, marrow, kidney infarction; aseptic bony necrosis, gall stones, priapism, etc… this disorder is common in areas among the African population in which malaria is endemic assuming to reflect a selective survival advantage. There is a heterozygous condition known as the sickle cell trait where only about 40% of the total hemoglobin are affected and the rest being normal. This condition is usually asymptomatic. Anaemia and painful crises are exceedingly rare. But here we are going to discuss about a case of sickle cell trait among the Arabs and also being symptomatic , which is a rare case indeed.

CASE REPORT

A 32 year old man of Arabn origin presented with complaints of painless hematuria for the past 1 year. He had no other related clinical symptoms or comorbid diseases.He had no history of trauma or nephrolithiasis. His physical examination was completely normal. He was admitted under the urologist and certain investigations were carried out including the routine blood and urine tests and cystoscopy. Every investigation ,including the coagulation profile and the renal parameters were normal , except for some, which are as follows.

1. Urine routine examination : blood 3+, protein 3+. No RBC casts or white blood cells seen.
2. Peripheral blood smear : presence of sickled and normal RBCs.

Therefore hematologist's opinion was sought . During the further history collection we came to know that his father died due to jaundice and the reason was unknown. This created a suspicion about some sort of hemoglobinopathy and Electrophoresis was thought of. It showed both HbS and HbA.

DIAGNOSIS

Based on the above clinical features, familial history, blood and urine investigations and the electrophoresis, the case was diagnosed to be suffering from sickle cell trait.

DISCUSSION

Sickle cell trait is a heterozygous condition where only 40% of the hemoglobin is abnormal unlike the homozygous sickle cell disease where 100% is defective. Therefore the sickle cell trait as such does not produce any clinical symptoms and such persons generally have a normal life span except under certain extreme conditions such as higher altitudes, hypoxia and chronic lung diseases. An uncommon, but highly distinctive, symptom is painless hematuria often occurring in adolescent males, probably due to papillary necrosis. The case discussed here has painless hematuria and is also probably due to papillary necrosis as

mentioned in various textbooks, which is a rare presentation.The electrophoresis report, being the gold standard investigation, with both HbS and HbA bands directed us to a conclusive diagnosis of sickle cell trait. In the studies conducted by *Gino Zadei et al*[1] and *Richard I. Chrone et al*[2], the case presented with clinical symptoms of painless gross hematuria and was diagnosed to have sickle cell trait. Further , in the study conducted by *Gino Zadei et al*[1], renal papillary necrosis was also diagnosed during IVP(Intravenous pyelogram), correlating and deeply resembling our study. Unlike the studies of *Brian F Birnham et al* [3] ,*April Edwards et al*[4]and*Vanessa K Wong et al*[5], our's is not a known case of sickle cell disease and moreover doesn't have any of the classical symptoms or complications like priapism, spinal artery infarction or pyomyositis. Usually the disease is common among the African and Asian population. And also sickle cell traits are usually asymptomatic and lead a very usual and healthy life. The case here is an Arab and presents with a very unusual symptom, making it a very rare case report. The only drawback of the case is that, he wasn't willing for an IVP and the difficulty in follow-up.

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