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CASE REPORT: HEMOGLOBIN D-PUNJAB HOMOZYGOUS IN A 23-YEAR-OLD MALE OF SINDHI COMMUNITY: A CASE REPORT IN A TERTIARY CARE CENTER

General Medicine

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ABSTRACT

Hemoglobin D-Punjab (HbD-Punjab) is a structural variant of hemoglobin resulting from a point mutation in the beta-globin gene. While heterozygous individuals are typically asymptomatic, the homozygous state is rare and can present with mild anemia or remain clinically silent. We report a case of 23 year old man with no comorbidity presented with generalised weakness, decreased appetite and lower back pain since 1 month . He was having Anemia with mild hepatosplenomegaly and pallor. Anemia due to blood loss was ruled out. His Serum iron level was normal. Serum lactate dehydrogenase level was raised and extracorpuscular causes were absent. Hemoglobin electrophoresis was performed in view of suspected hemoglobinopathy. It showed haemoglobin D Punjab heterozygous type, which is very rare.

KEYWORDS

Hemoglobin D, HbD-Punjab, Homozygous, Hemoglobinopathy, Sindhi community, HPLC

INTRODUCTION

Hemoglobin D-Punjab (HbD-Punjab), also known as HbD-Los Angeles, results from a point mutation at codon 121 (GAA → CAA) of the β-globin gene, substituting glutamine for glutamic acid. It is the most common HbD variant and is frequently found among people from the Punjab region and some ethnic groups in India, including the Sindhi community. While the heterozygous form is typically benign, homozygous cases are rare and may present with mild hemolytic features or be clinically silent [1,2].

CASE PRESENTATION :

A 23-year-old male from the Sindhi community in Gondia, Maharashtra, presented for evaluation of generalised weakness, decreased appetite and lower back pain since 1 month . There was no history of transfusion, chronic illness, or significant family history. Physical examination was normal. . He was having Anemia with mild hepatosplenomegaly and pallor. Anemia due to blood loss was ruled out. His Serum iron level was normal. Serum lactate dehydrogenase level was raised and extracorpuscular causes were absent. No jaundice or lymphadenopathy was noted.

INVESTIGATIONS

Complete Blood Count (CBC):
Hemoglobin: 9.8 g/dL
MCV: 85 fL
MCH: 28 pg
RBC Count: 4.1 million/mm³
Peripheral smear: Predominantly normocytic, Few Microcytes and mild hypochromic RBCsHigh-Performance Liquid Chromatography (HPLC):
HbD: 94.6%
HbA2: 2.8%
HbF: 2.6%

Diagnosis

Homozygous Hemoglobin D-Punjab (HbD-Punjab)

DISCUSSION

HbD-Punjab is a rare hemoglobin variant that is typically asymptomatic in heterozygous form. Homozygous HbD-Punjab, as in this case, is rarer and may present with mild anemia or no symptoms at all [3]. Diagnosis is established through HPLC and confirmed by molecular testing. The condition gains clinical importance when combined with other variants such as HbS or β-thalassemia, where more severe symptoms may occur. Genetic counseling is essential, especially in communities with higher prevalence of hemoglobinopathies.

CONCLUSION : This case highlights the importance of routine screening for hemoglobinopathies in high-risk ethnic groups. Though homozygous HbD-Punjab is rare and largely asymptomatic, early identification helps guide family planning and avoid severe compound hemoglobinopathies.

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