Case Report

A Case of HbQ India Heterozygous in a Patient of Bengali Origin

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Abstract

Background : HbQ India (HbA1:c. 193 G>C), is relatively an uncommon alpha 1 -chain structural Hemoglobin variant, due to mutation at codon 64 of the alpha1-globin gene causing an amino acid substitution of histidine for aspartic acid. With an overall incidence of 0.4% HbQ India is predominantly found amongst the Sindhi population, mostly in individuals from western and Northern India. We reported a case of HbQ India Heterozygous by HPLC in a 33-year-old female of Bengali origin in Eastern part of India.

Key words : HbQ India, Alpha Chain Variant, Mutation.

India is a land of plenty of Hemoglobin variants. HbQ India has a prevalence of 0.4% in the Indian subcontinent. HbQ India (HbA1:c. 193 G>C), is relatively an uncommon alpha 1 -chain structural hemoglobin variant, due to mutation at codon 64 of the alpha1-globin gene causing an amino acid substitution of histidine for aspartic acid[1]. HbQ was first described by Vella, *et al*, in association with alpha Thalassaemia in a Chinese patient[2]. There are three variations that have been described namely India (alpha 64 Asp to His), Thailand (alpha 74 Asp to His) and Iran (alpha 75 Asp to His)[3]. This rare disorder has been detected in the Homozygous and heterozygous states mostly in association with alpha and beta Thalassemia. HbQ-India is clinically silent normally. The replacement of aspartic acid with histidine, which is on the surface of the protein structure, does not affect the protein interchain contacts and electrical charges of the molecule; therefore, it does not cause any changes in hematologic parameters and indices[4]. It becomes symptomatic when it is present in association with other conditions like beta-thalassemia, alpha-thalassemia, HbE, HbH and nutritional Anemia. HbQ India is pre -dominantly found amongst the Sindhi population, mostly in individuals from Western and Northern India[5]. However, our case is of Bengali origin from West Bengal, Eastern India (Fig 1 & Table 1).

We recently encountered a case of HbQ India Heterozygous in a 33-year-old female patient who came to us for Hemoglobin typing by HPLC for pre-marital screening for Thalassemia trait.

Her HPLC (using D10 Hemoglobin Testing System (Bio-Rad) revealed normal HbF <0.8%, HbA0 76.6%, HbA2 1.6% and an unknown peak in the retention window of 4.48 mintutes, the percentage of area was 10.35%. HbQ India is an alpha chain variant and does not cause any clinical manifestation which is obvious from the hematological parameters. There is no anemia or reticulocytosis, MCV & MCH are normal. Adult Hemoglobin though reduced yet HbA2 being <3% rules out association of beta Thalassemia trait. The unknown peak at 4.48 minutes show the area percentage to be in the range of

Editor's Comment :

- HbQ India is a rare, clinically silent alpha globin chain variant which may be an incidental finding for routine hemoglobinopathy screening by HPLC. Although heterozygous cases typically show normal hematological parameters, accurate identification and differentiation from other hemoglobin variants are essential, especially in premarital or antenatal screening settings.
- Confirmation can be done by molecular testing and familial studies to ensure appropriate genetic counseling and ruling out co-inheritance with clinically significant hemoglobinopathies.

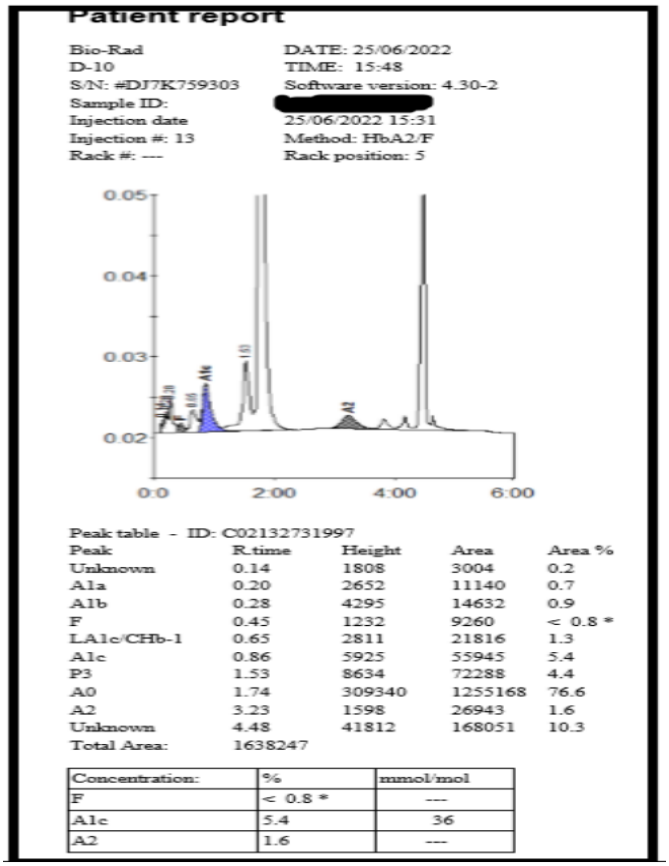

Fig 1 — Showing Patient Report

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Received on : 17/08/2022
Accepted on : 27/07/2024

How to cite this article : A Case of HbQ India Heterozygous in a Patient of Bengali Origin. Ghosh A, Chowdhury N, Chakrabarti S. *J Indian Med Assoc* 2026; **124(3):** 71-2.

Table 1 — *Hematological Parameters of the Patient*

Parameters	Values	Ref int
Hemoglobin	14.0 gm/dl	13-17 gm/dl
RBC	5.06*10^6/µl	4.5-5.5*10^6/µl
Hematocrit	43.9%	40-50%
MCV	86.9 fl	83-101 fl
MCH	27.6 pg	27-32pg
RDW	14.7%	11.6-14%

10-20% which differentiates it from Homozygous condition where the presence of HbQ is >35%[6]. The patient's family members (parents and siblings) were requested to undergo Hemoglobin typing (HPLC) to find out the inheritance history, however it was denied by the party. In presence of abnormal Hemoglobin the use of a single test to establish presumptive identification is inappropriate and second or even third line testing procedures should be in place. The patient was suggested to undergo a parental screening and DNA analysis for the same

Conclusion

India Is a land of huge prevalence of different Hemoglobinopathies which are yet to be identified. Now-a-days various procedures like HPLC, IEF, ARMS–PCR, DNA sequencing are available for diagnosis of the abnormal Hemoglobin.

Moreover, consanguineous marriage being common in India , screening and genetic counseling are essential to prevent the occurrence of Homozygous Hemoglobinopathies .

Funding : This is a case study from the retrospective data from Department of Clinical Chemistry and Special Immunology, Suraksha Diagnostics Pvt Ltd. No additional funding required.

Conflict of Interest : The authors declare no conflict of interest.

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