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Case Report

Hemoglobin D-Punjab heterozygous encountered in Assam: a case report

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ABSTRACT

Hemoglobin (Hb) D-Punjab is a haemoglobin variant with a point mutation in beta globin gene. A case of Hb D was encountered during screening of Hb variants among undergraduate students. With a prevalence rate of 2% in India, Hb D is generally rare in the Northeast region of India. Among the 200 screened cases, one Hb D heterozygous was detected by analysing the blood sample of the subject with an automated haematology analyser for complete blood count (CBC) and by using high performance liquid chromatography (HPLC) based Hb typing method. The haematological parameters were done in cell counter with Hg % 15.1 g/dl, mean corpuscular volume (MCV) 95 fl and mean corpuscular hemoglobin (MCH) 32.2 pg. The platelet value was high and high mean corpuscular hemoglobin concentration (MCHC) was obtained and the HPLC based haemoglobin typing was done with A2-2.9, A0-53.3, F- <0.8 area percentage and unknown window with 38.2 area percentage (retention time 3.86). Hb D Punjab heterozygous case has been reported in this report as prevalence of Hb D Punjab is very rare in the state of Assam.

Keywords: Haemoglobinopathies, Hb D, Genetic haematological disease, HPLC

INTRODUCTION

Hemoglobin (Hb) D-Punjab is a variant derived from a point mutation in the beta-globin gene in the first base of the 121 codon (GAA→CAA) where glutamic acid is substituted by glutamine (Glu>Gln) in the beta globin chain.[1] In India, Hb D Punjab is found with the greatest prevalence (2%) in Punjab as well as in nearby Gujarat (1%).[2] Hb D is predominantly seen in other northwestern states like Uttar Pradesh, and Jammu and Kashmir.[3] The prevalence of Hb D in northeastern states like Assam are rare. Several studies conducted have shown a prevalence of 0.02-0.09%. Also, the heterozygous form of Hb D with Hb S or β-thalassemia in the north east regions are extremely rare.[4,5] The Hb D-Punjab can be inherited in heterozygous form with normal Hb A, characterizing the heterozygous trait. This condition presents no clinical or haematological alterations. The homozygous component Hb DD is the rarest form of inheritance, not commonly related to symptomatic cases, but occasionally individuals with this profile can develop mild to moderate haemolytic anaemia. The association of this variant with other abnormal haemoglobins, such as Hb S or thalassemia can also occur. It is most prevalent in Punjabi, Sikh, and migrant Sindhi populations.[2]

CASE REPORT

During a screening study of haemoglobinopathies and thalassaemia, done among the undergraduate students in a tertiary care hospital of Assam, a case of Hb D heterozygous was encountered. The subject was a 20 years old male. When we enquired about the details, it was found that the subject is a native of Rajasthan. 3 ml blood sample

was collected from the subject in sterile vacutainers (EDTA) after filling up the proforma and taking written informed consent.

After sample was collected, complete blood count (CBC) was done within 1 hour using an automated haematology analyzer machine (Horiba ABX Micros ES 60) (Table 1) and then sample was further processed for Hb typing by BIORAD D-10 (dual mode) HPLC machine using the standard kits and reagents. An HbA2/F calibrator and two level controls were analysed at the beginning of each run for quality check. The result obtained was interpreted and recorded. The Hb D heterozygous Hb variant was identified (Figure 1).

Table 1: Haematological indices of the subject (complete blood count).

Parameters	Results
Hb% (g/dl)	15.1
WBC (10^3/mm^3)	9.3
RBC (10^6/mm^3)	4.68
Hct (%)	44.6
MCV (fl)	95
MCH (pg)	32.2
MCHC (g/dl)	33.8
Platelet count (10^3/mm^3)	439

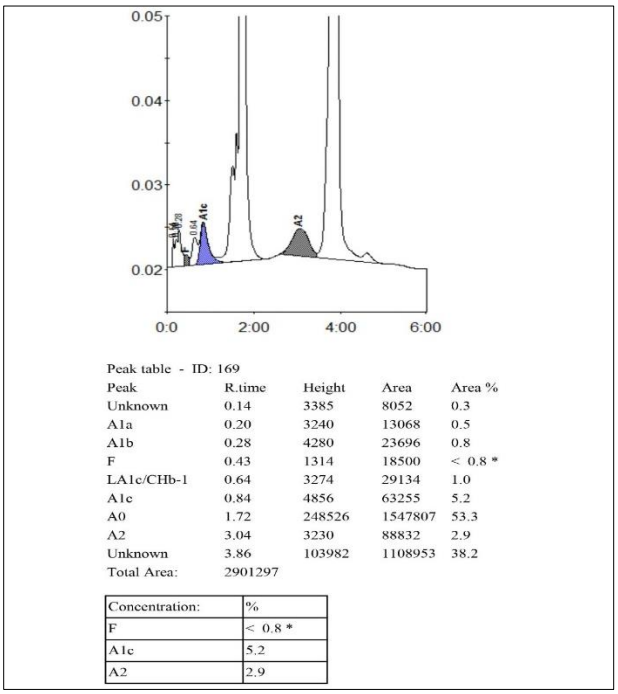

Figure 1: Chromatogram of the subject with Hb D Punjab heterozygous.

DISCUSSION

For Hb D Punjab, the chromatogram shows an unknown window with a retention time of approximately 3.8±0.1 minutes in HPLC based haemoglobin typing machine. The levels of Hb F are normal whereas the levels of Hb A2 are towards the lower side.[6] As per the standard BIORAD D-10 protocol for Hb D Punjab, the values for Hb D elutes in an unknown window of retention time 3.8±0.1 minutes with an area percentage of 22–40% for Hb D heterozygous.[7] The values correlate with our findings for this Hb D case.

Generally, the Hb D Punjab carriers are phenotypically asymptomatic and the prevalence rate in the general north Indian population is 3%.[7,8] In patients with Hb D disease, the size and number of RBCs decreases causing mild anaemia. However, it does not cause any serious health issues.[9] Although, in India, Hb D is found in north-western states like Punjab, Gujarat and in few other states like Uttar Pradesh and Jammu and Kashmir also, but a very few cases has been reported in literature till now.[10]

A rare case report of Hb D in a 24-year-old female was reported in Chennai by Anuradha et al, the values of abnormal haemoglobin were similar to our findings.[11] A compound heterozygous case of Hb D/Hb Q was reported in a 48-year-old Punjabi female by Dhanani et al, which was detected accidentally while analyzing HbA1c by HPLC from Gujarat.[12] This shows that Hb D generally remains as silent in carrier state and does not show much clinical manifestations. Also, Desai et al reported another case of Hb D disease in a 13-year-old girl from Gujarat.[13] A compound heterozygous Hb SD Punjab case in a 10-year-old female child was reported from Nagpur, Maharashtra by Mukherjee et al.[14] Hb D is very rare in North-East India and not much cases have been reported till date. As per Sharma et al, a case of Hb D heterozygous and double heterozygous state with Hb E (Hb ED) was reported in an Ahom family affiliated to Tai-Kadai linguistic group of Assam.[15] The HPLC findings of the present reported case resemble the findings of the cited literatures which were reported from different regions of India.

Though the heterozygous cases are asymptomatic and does not show much phenotypic symptoms, but if they marry other heterozygous cases then the chances of having offspring with homozygous and compound homozygous cases increases which lead to a socio-economic burden to the family and society. Most of the heterozygous Hb variant cases remain unaware of their disease status. Screening among young adults is very important and necessary. Premarital screening, screening of young adults, and also screening the couples at risk are important to reduce the occurrence of these genetic haematological diseases in the near future generation. In this case also, the subject was unaware of his carrier status. With this finding, we suggested screening for Hb typing for his family members.

CONCLUSION

As most of the genetic haematological diseases in the heterozygous state do not show clinical manifestations, the

subjects remain unaware of their carrier state. Also, as there are no definite neonatal screening programs or community-based screening programs, this shows that majority of the population remains unaware of the carrier state. So, awareness programs, screening programs and genetic counselling should be adopted to reduce the occurrence of these genetic haematological diseases.

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