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Case Series

Three rare and accidental findings of hemoglobinopathies encountered in high-performance liquid chromatography: case series

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ABSTRACT

Hemoglobinopathies are the leading cause of some major genetic and social health problem in India. Among all hemoglobinopathies, sickle cell disorder and thalassemia are commonly found in Gujarat state. Double heterozygous state of hemoglobin S and D, hemoglobin E trait, hemoglobin D disease are very uncommon. In present instance, one case of 25-year-old male was diagnosed with sickle cell hemoglobin D disease. The case was confirmed through slide-based sickle test and high-performance liquid chromatography (HPLC). The peripheral smear findings showed presence of microcytic hypochromic red blood cells (RBCs) and many sickled RBCs. Ultrasonography (USG) findings showed hepatomegaly. Second case of 30-year-old female was diagnosed with hemoglobin E trait. The case was confirmed by HPLC. The peripheral findings showed normocytic normochromic RBCs and occasional target cells. Presence of gross hepatomegaly on palpation. Third case of 20-year-old female was diagnosed with hemoglobin D trait. The case was confirmed by HPLC. The peripheral findings showed normocytic normochromic RBCs.

Keywords: Sickle cell disorder, Hemoglobin D trait, Hemoglobin E trait, HPLC

INTRODUCTION

Hemoglobinopathies are one of the most commonly observed blood disorder widely prevalent in India. It is one of the major public major health problems. There are regional variants for these variations in structure of haemoglobin. The sickle cell hemoglobin (Hb S) is structurally abnormal variant with presence of valine at 6th position of β globin polypeptide chain instead of glutamic acid. HbE disease caused by a mutation leading to presence of lysine instead of glutamic acid in the 26th position of the β chain of hemoglobin. Substitution of glutamine at 121 positions of β chain instead of glutamic acid gives rise hemoglobin D. Tribal communities constitute a major part of the Indian population and are particularly vulnerable to hemoglobinopathies. Reports have designated it as the second most common hemoglobinopathy variant globally with observations that this mutation has evolved to provide resistance against malaria.[1]

In India, a considerable size of population spanning across Central India, from Odisha in the east to Maharashtra and Gujarat in the west is affected by the sickle cell disorder (SCD).[2,3] Three major variants of this disease are widely reported in India –sickle hemoglobin (Hb S), hemoglobin E and hemoglobin D (Hb D Punjab).[4,5]

According to the World Health Organization (WHO) report, hemoglobin disorders are originally endemic to 61% of the 229 countries around the world and 5.2% of the world population carries a significant hemoglobin variant. Again approximately 1.1% of couples are at risk for having children with a hemoglobin disorder and 2.7 per 1000 conceptions are affected worldwide.[6]

CASE SERIES

Case 1

A 25-year-old male presented with complains of fever, chest pain, abdominal pain and constipation since one week.

Complete blood count findings show hemoglobin 8.7 g/dl, mean corpuscular volume (MCV) 83 µm^3, mean corpuscular hemoglobin (MCH) 27.5 pg, and mean corpuscular hemoglobin concentration (MCHC) 33.2 g/dl. The peripheral smear findings showed presence of microcytic hypochromic red blood cells (RBCs) and many sickled RBCs.

Ultrasonography (USG) findings showed hepatomegaly. Slide test for sickle cell disease showed presence of many sickled RBCs. The diagnosis of sickle cell hemoglobin D disease was confirmed by HPLC. The graph showed peak of Hb S with value 38.0%, Hb D 53.4% and Hb F 5.8 %. The value adult haemoglobin HbA0 was 2.8%.

Case 2

A 30-year-old 9 months pregnant female presented with complains of weakness and fatigue since one month. History of repeated of blood transfusion since last 4 months.

Complete blood count findings show hemoglobin 10.2 g/dl, MCV 78 µm^3, MCH 25.2 pg, and MCHC 33.2 g/dl. The peripheral findings showed normocytic normochromic RBCs and occasional target cells. Presence of gross hepatomegaly on palpation. The diagnosis of hemoglobin E trait was made by HPLC. The graph of HPLC showed peak of Hb E at 102 seconds and value of peak was 12.3%. Hb F value was 5.3% and adult haemoglobin HbA0 value was 74.8%.

Case 3

A 20-year-old female presented with complain of menstrual irregularities since 2 month.

Complete blood count was normal with normal peripheral smear findings. HPLC was done to rule out hemoglobinopathies and it showed graph plating at 140 seconds of Hb D with value of 43.0%. The value of adult haemoglobin HbA0 was 48.4%.

In our first case, three major peaks were plotted at 11, 140 and 161 seconds respectively. The interpretation of the graph showed presence of Hb F eluted at 11 second with value being 5.8%. Hb D eluted at 140 seconds and value being 53.4%. Hb S eluted at 161 seconds with value being 38.0%. Thus, the diagnosis of sickle cell hemoglobin D disease was made (Figure 1).

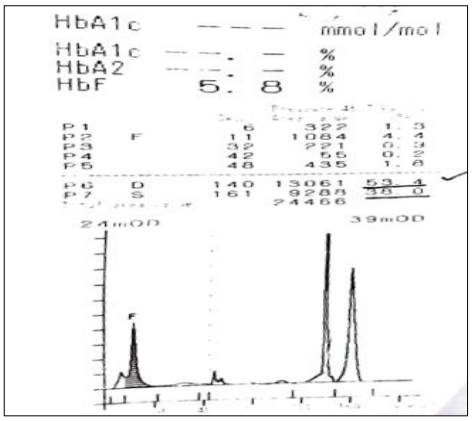

Figure 1: HPLC graph of sickle cell hemoglobin D disease.

Our second case showed elution of Hb E peak at 102 seconds with value 12.3%. HbA0 was eluted at 79 seconds with value 74.8%. Value of Hb F was noted as 5.3%. Thus, diagnosis of hemoglobin E trait was made (Figure 2).

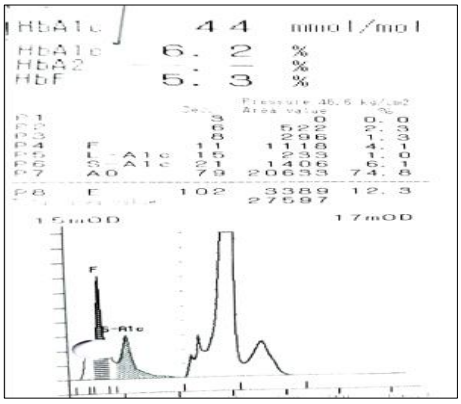

Figure 2: HPLC graph of hemoglobin E trait.

In our third case, elution of Hb D was present at 140 seconds with peak value 43.0%. Value of adult hemoglobin level was 48.4% with normal Hb F value. Thus, the diagnosis of Hb D trait was made (Figure 3).

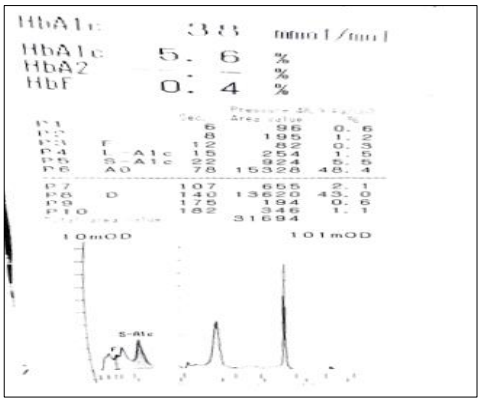

Figure 3: HPLC graph of hemoglobin E trait.

DISCUSSION

Sickle haemoglobin is known to interact hemoglobin D and hemoglobin E. The first description of hemoglobin D Punjab was made by Itano in year 1951. It is predominantly seen in India, Pakistan and China. One such case was reported in Maharashtra.[8] The elution time of hemoglobin D according to Arkray machine of HPLC is 140 seconds and elution time of Hb S is between 160–163 seconds. The peak of Hb D appears immediately after Hb A2.

The HbE disorder in India is commonly seen in north-eastern states and West Bengal and gene frequency has been reported to be about 10.9% in north eastern regions.[9,10] Rural population contribute a major part of the Indian population and are particularly prone to hemoglobinopathies. In a rural population, 1.4% prevalence of HbE disorders was recorded.[11,12] The disease pattern of Hb E is similar to that of β-thalassemia.[13] HbE trait is reported to have a content of approximately 30% of HbA2 along with HbE whereas homozygous HbE patients have approximately 90% HbE+A2 with minor elevation of HbF.[12]

HbD Punjab also known as HbD Los Angeles, is one of the commoner hemoglobinopathies in India, Pakistan, England, Ireland, Holland, China, Turkey and Brazil. Its prevalence is 1-3% of population in north-west India especially in Punjab (Sikhs) and in Gujarat.

CONCLUSION

Present trend of migration of population leads to demographic variation in hemoglobinopathies. Detailed geographic history along with family history should be elicited before stamping the diagnosis. There is a need to establish thorough practices of testing and evaluation in almost all regions of the state.

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