HAEMOGLOBIN ELECTROPHORESIS IN DIAGNOSING A CASE OF SICKLE CELL ANAEMIA ASSOCIATED WITH β -THALASSEMIA

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ABSTRACT

Alkaline haemoglobin electrophoresis is a useful tool in diagnosing β -thalassemia and sickle-cell anaemia. In this report, using this simple technique, β -thalassemia associated with sickle-cell anaemia is diagnosed. This is the first case we have diagnosed in our laboratory using agarose gel electrophoresis.

KEY WORDS: Haemoglobin electrophoresis, β -thalassemia, sickle-cell anaemia.

INTRODUCTION

Haemoglobin electrophoresis at alkaline pH of 8.4 is a simple and informative screening test for abnormal haemoglobins. This procedure detects haemoglobin variants that migrate differentially from normally occurring haemoglobins. Using this method, HbS and HbC, the most clinically significant abnormal variants which have distinctly different electrophoretic mobilities can be differentiated from each other and from HbA. Haemoglobin electrophoresis in acid conditions is useful in differentiating HbS from HbD and HbG, HbC from HbE, HbO and HbC Harlem.

In our laboratory, we use alkaline haemoglobin electrophoresis as the preliminary test in diagnosing haemoglobinopathies such as sickle cell anaemia and thalassemia.

Thalassemias are hereditary disorders characterized by a reduction in the rate of synthesis of one or another type of globin chains. Reduction in α -chain synthesis results in α -thalassemia and is highly prevalent in people of Southeast Asian origin (1). Reduction or absence in the synthesis of β -chain results in β -thalassemia. This disorder is historically associated with Mediterranean people. However its prevalence appears to be high in Asians of which 4-

6% are Indians (2). The β -thalassemias are classified as β^0 -thalassemia, β^+ -thalassemia and β^{++} -thalassemia depending on the degree or deficiency of β -chain synthesis. Thalassemias can also be associated with other haemoglobinopathies, one of them being β -thalassemia associated with sickle cell disease where HbA levels are less than HbS (3).

In this case report, the patient was referred to us with a diagnosis of sickle cell disease from a local hospital. Alkaline haemoglobin electrophoresis (Beckman Paragon) was done and we arrived at a diagnosis of sickle cell disease associated with β -thalassemia and this modified the treatment protocol of the patient.

CASE REPORT

A 18 years old male was referred to our hospital with history of fever associated with chills and rigors since one month. He also complained of colicky type of pain in the left hypochondrium. Family history revealed that he belonged to a tribal race called "Yadigalu" originating from Gujarat.

On clinical examination, pallor and icterus were present. Systemic examination showed hepatomegaly and splenomegaly. Considering the history and the outside report of sickle cell in peripheral smear, a differential diagnosis of sickle cell crisis and malaria was made. To confirm the same, the following

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preliminary investigations were carried out. (Tables 1 and 2).

Table 1. Haematological investigations.

Investigation	Results	Normal Range
Haemoglobin	5.1 g/dl	Male: 13-18 g/dl
WBC-TC	13,000/ul	4000-11000/ul
DC-Neutrophils	72%	Adults: 40 – 75%
Lymphocytes	21%	20 – 45%
Eosinophils	2%	1 – 6%
Monocytes	9%	2 – 10%
Basophils	2%	< 1%
M P smear	Negative	

Table 2. Liver function tests.

Investigation	Results	Normal range
Total proteins	8.5 g/dl	6.0 – 8.0 g/dl
Albumin	4.2 g/dl	3.2 – 5.5 g/dl
A/G ratio	0.9	1.0 – 1.5
Bilirubin: total	4.3 mg/dl	0.1 – 1.0 mg/dl
conjugated	0.4 mg/dl	0.0 – 0.4 mg/dl
Aspartate transaminase	64 U/L	Upto 37 U/L
Alanine transaminase	35 U/L	Upto 65 U/L
γ -Glutamyl transferase	23 U/L	Male: 11 – 50 U/L
Lactate dehydrogenase	495 U/L	240 – 480 U/L

Thus from the preliminary investigations, malaria was ruled out. A complete haemoglobinopathy work up was carried out and the results were as follows.

1. Peripheral smear = sickle cell positive. 2. Alkali denaturation of HbF = 9%. 3. Hb Electrophoresis = Pattern suggestive of thalassemia associated with sickle haemoglobin. Since HbA is present, most probable diagnosis will be type I HbS- β^+ -thalassemia (Fig 1).

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Fig. 1. Illustrates alkaline haemoglobin electrophoresis of the patient with increase in HbS and HbF.

Thus the above findings clearly pointed towards the diagnosis of sickle cell anaemia associated with β -thalassemia. By proper interpretation of the haemoglobin electrophoresis report, the patient was also diagnosed as type I HbS- β^+ -thalassemia due to the presence of HbA. The recommended further investigations for confirmation could be by more advanced techniques like polymerase chain reaction.

Conclusion

Advancements in technology have an advantage in diagnosing haemoglobinopathies at the molecular level. But these technologies are present only in few laboratories. Alkaline haemoglobin electrophoresis is a simple technology which can be set up even at a primary level and thus aids in diagnosis of a syndrome like β -thalassemia associated with sickle cell anaemia even at a grass root level.