ORIGINAL ARTICLE

Haemoglobin Electrophoresis Pattern in Normal Siblings and Beta Thalassemia Trait Siblings of Beta Thalassemia Major Patients

NASIR RASHID, MUHAMMAD JAWAD ANWAR*, RABIYA JAMIL*, *ZAHID IBRAHIM**, MUHAMMAD FAROOQ***

ABSTRACT

Aim: To determine haemoglobin electropherogram in siblings of beta thalassemia major patients. A total of 200 subjects were included in the present study and divided into two equal groups. Group IA with normal siblings of BTM patients and IB with beta thalassemia trait (BTT) siblings of BTM patients. Patients with history or diagnosis of any acute or chronic illnesses were excluded from the study. Haemoglobin electrophoresis was performed. 5 ml of blood was drawn in EDTA containing tube and was used for haemoglobin electrophoresis.

Results/Conclusions: Out of 200 siblings of beta thalassemia major (BTM) patients, 156(78%) were diagnosed as beta thalassemia trait (BTT). HbA_2 and HbF were increased in BTT siblings (IB) as compared to normal siblings of BTM patients (IA).

Keywords: Hb electrophoresis, Beta Thalassemia Trait

INTRODUCTION

The thalassemias are a group of inherited disorders of haemoglobin synthesis which result from reduced rate of production of one or more of globin chains of haemoglobin. It is the commonest single gene disorder in the world[1]. It is divided into two major categories α and β thalassaemia, depending on which globin chain is produced in reduced amounts[2]. The diagnosis of beta thalassaemia trait cases is essential to prevent births of homozygous cases by genetic counseling[3]. By this method alone the birth rate of thalassaemia major children can be reduced by as much as 90%[4]. Peripheral blood film of these cases shows microcytosis, hypochromia and anisopoikilocytosis and target cells[5]. It is based on finding microcytic red cells, target cells and basophilic stippling in peripheral blood film[6]. The red cell indices give a more reliable diagnosis. In the thalassaemia trait cases MCV and MCH are low. The red cell count is often more than $5.0x10^{12}/L$[7]. Haemoglobin electrophoresis is essential for definite diagnosis of beta thalassaemia trait. Raised HbA_2>3.5% confirms diagnosis[8]. Fetal haemoglobin is also high in β-thalassaemia minor cases as was seen in 64% of subjects with β-thalassaemia minor[5].

METHODOLOGY

A total of 200 subjects were included in the present study and divided into the two groups.

- 1A normal siblings of BTM patients i.e., HbA2<3.5%
- 1B BTT siblings of BTM patients i.e. HbA2>3.5%

They were brothers and sisters of BTM patients. They were selected from thalassemia center Sir Ganga Ram Hospital, Children Hospital, Services Hospital and institute of blood transfusion Punjab Lahore. Haemoglobin Electrophoresis was done. 5.0ml of blood was drawn in EDTA containing tube / vial (1.5mg/ml).This sample was used for haemoglobin electrophoresis. The instrument used for Hb electrophoresis was Beckman Coulter electrophoresis. It was used to determine HbA_2 concentration.At alkaline pH, haemoglobin is a negatively charged protein when subjected to electrophoresis it migrates towards the anode (+). The structural variants which have a change in the charge on the surface of the molecule at alkaline pH will separate from Hb A. The Appraise^R densitometer was used to scan the processed gel obtained by electrophoretic methods.

RESULTS

The detail of results is given in Table 1, 2 and 3

Table 1:HbA_2 lin group IA, IB

HbA_2 (%)	IA	IB
Mean ± SD	2.14 ± 0.72	4.59 ± 0.94
Ranges	0.8-3.4	2.5-7.1
Total Subjects	44	156

Statistical Analysis: IA vs IB P<0.01 (HS)
Group IA = Normal Siblings of BTM patients
Group IB = BTT Siblings of BTM patients

Assistant Professor Pathology, Amna Inayat Medical college, Sheikhupura
***Emergency Pathology Laboratory, SIMS/SHL, Lahore*
Correspondence to: Dr. Nasir Rashid, Assistant Professor Pathology, Amna Inayat Medical college, Sheikhupura

Table 2: HbA in group IA, IB

HbA(%)	IA	IB
Mean ± SD	95.5 ± 0.76	93.4 ± 7.3
Ranges	96.7-99.0	91.8-96.6
Total Subjects	44	156

Statistical Analysis: IA vs IB P>0.05 (NS)

Table 3: HbF IN GROUP IA and IB

HbF(%)	IA	IB
Mean ± SD	0.72 ± 0.12	2.1 ± 0.69
Ranges	0.3-0.8	0.4-3.4
Total Subjects	44	156

Statistical Analysis: IA vs IB P<0.05 (S)

DISCUSSION

Raised HbA_2 level is the most important finding in the confirmation of beta thalassaemia carrier state. Some studies show that HbA_2 level is affected by iron deficiency which decreases its level below the values for the diagnosis of carrier state. While others confirm that HbA_2 though decreased, in the presence of iron deficiency but never below significant values[8]. The mean ± SD values of HbA_2 of beta thalassaemia minor subjects was 4.59±0.95 (3.6--8.4%) in the present study. These values are in agreement with the findings of various studies carried out in Pakistan and outside Pakistan i.e., 3.9--10.5% by Fessas (1987)[9], 3.1--7.78% by Pootrakul et al (1973)[10]. While higher values were observed by Hashmi et al (1975)[5] i.e., 3.5—13.6% and lower values were in the range of 3.6—6.0% by Galanello et al (1990)[8] and Khattak and Saleem (1992)[11]. In the present study, the mean value of HbF was 2.1±0.69% (0.6—6.6). These results are in agreement with the values of other studies. The range of HbF by different studies was 0--7.8% by Pootrakul et al (1973)[10], while lower HbF levels were reported in which its level was <1%[11].

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