

A Rare Case Of Co-Existent Hb Q India-Beta Thalassemia Trait

D Desai, C Parmar, H Dhanani, R Patel, D Master

Citation

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Abstract

Hb Q- India is a rare alpha chain variant and usually it presents in the heterozygous state. It's presence along with beta thalassemia trait again adds to its rarity. Such a rare entity can be diagnosed by careful screening in routine practice with the use of the techniques like Hb electrophoresis, Solubility test. We report a case of Hb Q in a concomitant presence of Beta thalassemia trait.

CASE REPORT

A 19 years old boy presented to us for the screening of the beta thalassemia minor and was found to have variant hemoglobin migrating in the position of the Hemoglobin S (HbS) on cellulose acetate electrophoresis at alkaline pH. The patient was asymptomatic without any significant past or family history and clinical examination.

Automated cell counter used to determine the cell counts and indices showed the presence of thalassemic indices, whereas solubility test for sickling was negative (Table-1).

Suspecting the presence of beta thalassemia trait, Hb electrophoresis at pH 8.4 was performed which showed presence of an abnormal band in the position of Hb S and duplicated bands of Hb A₂ apart from normal band of Hb A, suggesting the presence of alpha chain variant in heterozygous state (Figure-1).

Figure 1

Figure 1: Photograph showing the electrophoretic mobility of hemoglobin at pH 8.4 in propositus, father, mother, sister, and control of a known case of sickle cell trait.

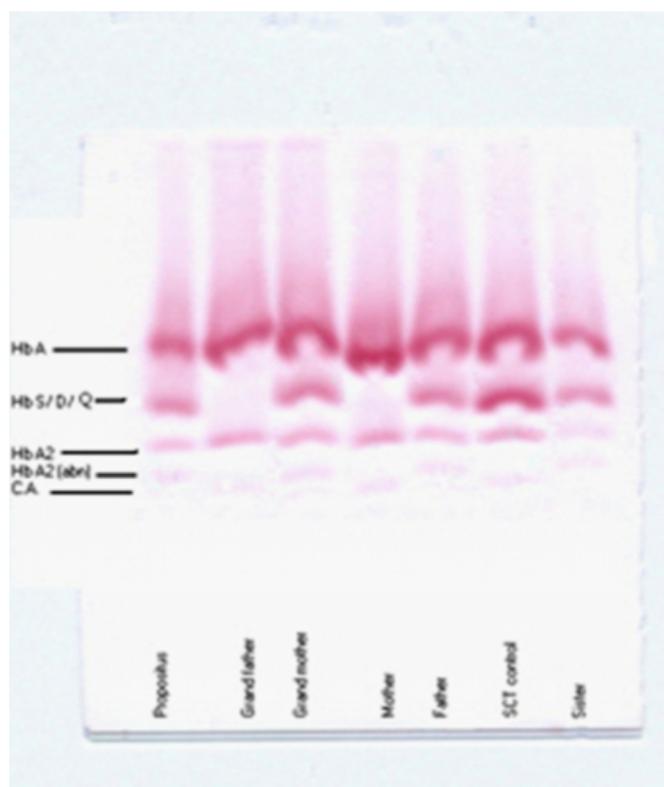


Figure 2

Table 1

	Hb g/dl	RBC 10 ¹² /L	Hct %	MCV fL	MCH Pg	MCHC g/dL	RDW fL	Solubility Test
Propositus	10.0	6.20	34.3	55	16.1	29.2	39.1	Negative
Father	11.3	6.72	38.8	58	16.8	29.1	38.3	Negative
Mother	10.3	6.15	35.0	57	16.7	29.3	38.3	Negative
Sister	9.2	5.68	33.0	58	16.2	27.9	46.9	Negative

Microcolumn chromatography using glycine-potassium cyanide developers showed the presence of high levels of Hb A₂ in the thalassemic range (Table-2).

Figure 3

Table 2

	Hb A ₂ %	Hb A %	Hb X %	Hb F % (Singer's)
Propositus	5.0	81.2	12.8	1.0
Father	5.3	94.1	---	0.6
Mother	4.7	81.3	12.9	1.1
Sister	5.1	94.5	---	0.4

Fetal hemoglobin (HbF) was determined using the alkali denaturation technique. Later on, HPLC was done to confirm and detect the type of alpha chain variant and it showed abnormal peak with a retention time of 4.73 minute along with Hb A₂ level of 5.2 % (Figure-2).

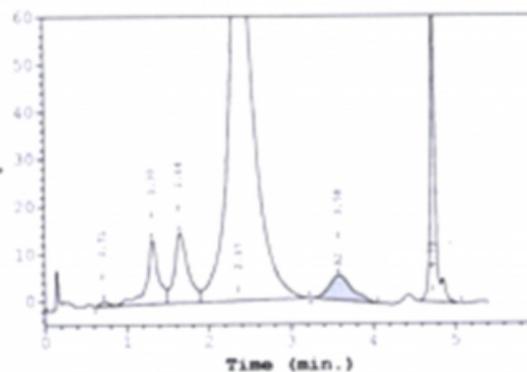
Figure 4

Figure 2: High performance liquid chromatography report.

Peak Name	Calibrated Area %	Area %	Retention Time (min)	Peak Area
P1	---	0.3	0.71	5323
P2	---	5.6	1.30	115858
P3	---	1.0	1.64	144033
A ₂	---	12.6	2.37	1501043
A ₁	5.2*	---	3.58	84733
Unknown	---	10.5	4.73	237219

Total Area: 2068169

F Concentration = %
 A₂ Concentration = 5.2*%
 *Values outside of expected ranges
 Analysis comments:



It confirmed the concomitant presence of Hb Q-India with beta thalassemia trait. Subsequently, family studies showed the similar abnormality in the mother along with the beta thalassemia trait in father and sister.

DISCUSSION

Hb Q variants are the alpha globin chain variants due to structural mutations. Three molecular variant types have been documented, namely HbQ-India, HbQ-Thailand and HbQ-Iran.

Normally HbQ is clinically silent. Even its presence along with beta thalassemia trait does not seem to produce any clinical abnormality. But, HbQ-H disease can give clinical manifestations though it is very rare. ¹ Quantities of HbQ variant is usually determined by the ratio of alpha A, alpha Q and beta A globin chains. Presence of alpha thalassemia favors the formation of HbQ, whereas beta thalassemia reduces the formation of HbQ. ^{2,3} This is explained to be due to post translational control mechanism. Our study also showed the decreased levels of HbQ due to concomitant presence of beta thalassemia trait as against the predictive value of 25 % in case of a stable structural alpha variant in heterozygous form. ⁴

In our study, primary screening by cellulose acetate electrophoresis showed abnormal Hb moving in the position of HbS (with sickling negative) and duplicated Hb A₂ suggesting the presence of alpha chain structural variant in heterozygous form. Hb A₂ levels and indices suggested the concomitant presence of beta thalassemia trait. HPLC confirmed the abnormality. Still definitive diagnosis can be done using the techniques like IEF, ARMS-PCR, DNA sequencing.

We stress on the point that careful screening of the samples using routine techniques like Hb Electrophoresis, Chromatography can be the basis of identification of abnormal hemoglobin variants like HbQ.

ANNOUNCEMENT

This case report was presented in the 29th Annual conference of IAPM, Gujarat Chapter at Surendranagar on 6th February, 2005.

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Author Information

Devenkumar Desai, MBBS, DCP

Division of Human molecular and Cytogenetics, Medical College & SSG Hospital

Chirag Parmar, MBBS, DCP

Division of Human molecular and Cytogenetics, Medical College & SSG Hospital

Hiren Dhanani, MBBS, MD

Division of Human molecular and Cytogenetics, Medical College & SSG Hospital

R. Z. Patel, MBBS, MD, DCP

Division of Human molecular and Cytogenetics, Medical College & SSG Hospital

D. C. Master, PhD

Division of Human molecular and Cytogenetics, Medical College & SSG Hospital