SUCCESSFUL PREGNANCY IN HOMOZYGOUS BETA THALASSAEMIA

(Report of a case)

by

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Introduction

In heterozygous form of beta thalassaemia the defect is minor and this is commonly known as "thalassaemia minor". On the other hand, homozygous beta thalassaemia or "thalassaemia major" is a crippling disorder with a severe anaemia leading to death in childhood. Thalassaemia major patients rarely achieve parenthood. The following is the case of homogyous beta thalassaemia with pregnancy who presented at AIIMS.

Case Report

Mrs. K.L. aged 29 years attended the Medical O.P.D. in January 1982 at 2 months gestation with anaemia, bad obstetric history and jaundice in previous pregnancies. She was admitted in medical ward at 18 weeks of gestation as a case of pregnancy with severe anaemia, jaundice and hepatosplenomegaly. She was transfused three units of blood during her stay of one month is the hospital. The patient was re-admitted in the maternity ward at 30 weeks gestation with history of pale coloured stool and itching.

This was her fifth pregnancy and had no living issues. During her first pregnancy (1974) she developed nausea vomiting and jaundice in the third month of pregnancy. She delivered a live born baby who died 2 hours after birth. The jaundice disappeared one month after delivery. In her second pregnancy she developed jaundice in the third month. The pregnancy was terminated prematurely in a still born baby. The jaundice again disappeared after one month of delivery. During her 3rd pregnancy she developed jaundice and a lump in left hypochondrium. She attended All India Institute of Medical Sciences and was diagnosed as beta thalassaemia with cholestatic jaundice of pregnancy. She had an intrauterine death at 31 weeks, and delivered a macerated still born baby following induction with oxytocin. Her 4th pregnancy was also complicated by jaundice and terminated prematurally at home in a fresh still born baby.

On examination, she was a pale, icteric woman of 4' 9" height weighing 87 lbs with thalassaemic facies. An ejection systolic murmur was heard in mitral area with widely fixed split 2nd sound. There was a diastolic murmur in tricuspid area. Liver was enlarged to 6 cm below costal margin. Spleen was 9 cm in spleno-umbilical line. Uterus was 28 to 30 weeks by gestation with cephalic presentation. Fetal heart rate was regular.

Management

She was investigated for bad obstetric history. Her haematologic profile and liver tests were carried out regularly. (Table I and II).

Fetal monitoring was done by weight gain, rise in fundal height, weekly urinary estriol, daily fetal movement score and twice weekly

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TABLE I

Hematologic Profile

Date	Hb.	Reticulocytes	Peripheral smear	Hb F	Hb A_2	Others
March 1982	4.7	2	Microcytic hypochronic target cells +	21.7	3.2	Coomb's neg.
June 1982	6.3	3.2	same	23.1	3.4	SFC 172 mg% TIBC 297 mg% % saturation 58%
18-6-82	6.7	3	same			Platelets 120 x 10^3/CC
10-7-82	7.5					
28-7-82	8.4					Electrophoresis done no other bands detected
9-8-82 (post op.)	10.4					

TABLE II

Liver Function Tests

Date	S. bilirubin Total	S. bilirubin Conj.	S. alk. Phos.	SGOT/PT		S. proteins Total	S. proteins Albumin	HBs AG
March 1982	3.8	1.0	34	60	24	6.7	3.4	Neg.
June 1982	7.4	4.8	10	15	18	8.2	5.1	Neg.
18-6-82	5.6		29	30	20	6.0	3.0	
10-7-82	7.2	5.0	32	60	42			
28-7-82	4.2	2.0	30	40	20	6.2	3.5	
9-8-82 (post op.)	2.2	1.5		25	40	6.6	3.8	

HB AG — Australia antigen.

non-stress test. She was transfused 4 units of blood. At 37 weeks gestation when she noticed sudden fall in her fetal movement score and had a non-reactive non-stress test, induction was decided upon. Artificial rupture of membranes done and syntocinon infusion started. After 18 hours of labour she was taken up for emergency lower segment caesarean section in view of the non-progress of labour. She was delivered of a 2.4 kg female baby with an Apgar score of 9/10. She was given 3 units of blood during operation. Her postoperative period was uneventful and was discharged on 10th post operative day.

Discussion

In Homozygous beta thalassaemia or thalassaemia major the disorder leads to

severe anaemia and very rarely patients attain puberty. Even if they attain adulthood, the disorder is usually associated with infertility. The literature was reviewed and only 1 case of homozygous beta thalassaemia who had two children has been reported in 1971 (Perkin).

The above discussed case of beta thalassaemia major has the unique feature of reaching parenthood without childhood transfusion and succeeding in having a live born baby.

Reference

1. Perkins, R. P.: Obstet. Gynec. 111: 120, 1971.