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PREGNANCY OUTCOME IN WOMAN WITH BETA THALASSEMIA INTERMEDIA: A RARE CASE REPORT

Obstetrics & Gynaecology

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ABSTRACT

Introduction: Thalassemia is one among hemoglobinopathies, it is inherited as autosomal recessive disorder. Beta thalassemia is consequence of impaired beta globin chain production or alpha chain instability where beta chain production is decreased, with excess alpha chains precipitate to cause red cell membrane damage resulting in anemia. Thalassemia during pregnancy could be associated with significant complications to mother and fetus. **Objective:** Reporting rare case of beta thalassemia intermedia in pregnancy with severe anemia and its maternal and fetal outcome. **Case Report:** A 20 years old Gravida 2, abortion 1 presented to OBG outpatient department with 20 weeks of gestation with known case of hypothyroidism. In first antenatal visit complete blood count showed severe anemia with hemoglobin 6.3g/dl with decreased blood indices. Hemoglobin electrophoresis with reflex PCR was done, which revealed beta thalassemia intermedia. Patient had regular antenatal visits. In antenatal period, patient was admitted 6 times, total 9 pints of PRBC transfusions were done at regular intervals to maintain her hemoglobin concentration at 10g/dl. At 38 weeks of gestation patient came with labor pain and leak per vagina was admitted. **Management:** After complete investigations, emergency LSCS was done in view of fetal distress. Approximate blood loss was ~500-600ml. Intraoperative period was uneventful. One pint of PRBC was transfused post operatively. She had uneventful post operative period with healthy mother and baby, was discharged in satisfactory condition on postoperative day 7. At time of discharge hemoglobin level was 10.4g/dl. **Conclusion:** Inherited autosomal recessive disorder like thalassemia is a major global health concern. Nowadays with aggressive use of iron chelation therapy, the rate of pregnancy in women with thalassemia major is fast increasing. To reduce associated maternal ,neonatal mortality and morbidity, preconceptional councelling, monitoring during antenatal, intrapartum and postpartum period is important.

KEYWORDS

Beta thalassemia intermedia; hemoglobin electrophoresis; PRBC;

INTRODUCTION

Thalassemia is one among hemoglobinopathies and it is inherited as an autosomal recessive disorder. Annual incidence of beta thalassemia is 1/100,000 throughtout world. Asian countries like India, Pakistan, and Bangladesh account for 79% thalassemia births. Beta thalassemia intermedia is the most common cause of chronic hemolytic anemia and accounts for one-fourth of beta thalassemia cases. Beta thalassemia are consequences of impaired beta globin chain production or alpha chain instability and beta chain production is decreased, and excess alpha chains precipitate to cause red cell membrane damage. Thalassemia during pregnancy could be associated with significant complications to mother as well as fetus. Therefore, universal antenatal screening for thalassemia carriers should be implemented in populations having a high prevalence of this condition.

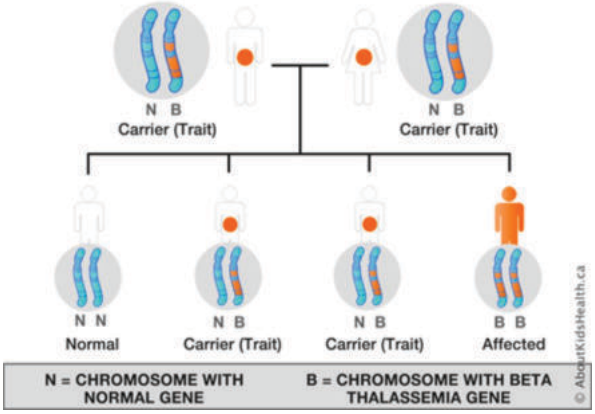

Figure 1: Genetics in Beta Thalassemia

CASE REPORT

A 20 years old Gravida 2, abortion 1 presented to outpatient department with 20 weeks of gestation with known case of hypothyroidism. She had history of moderate anemia since childhood where HB level ranged between 8-9g/dl. In her first antenatal visit complete blood count showed severe anemia with hemoglobin 6.3g/dl with decreased blood indices MCH-71.2fl, MCH-23.0Pg.Patient was refered to hematologist,hemoglobin electrophoresis with reflex PCR was done at 20 weeks of gestation in view of severe anemia and severe anisopoikilocytosis, which revealed beta thalassemia intermedia with elevated HbA2.Patient had regular antenatal visits ,HB levels with blood indices were assessed every 3 weeks , serum ferritin levels every 4 weeks ,ultrasonographic evaluation of fetal growth every 4 weeks starting at 24-26 weeks.In antenatal period, patient was admitted 6 times with total 9 pints of PRBC's transfusions were done with regular intervals to maintain her hemoglobin concentration at 10mg/dl.Iron profile was done to rule out iron over load due to more number of blood transfusions.At 38 weeks of gestation patient came with pain abdomen and per vaginal leak and was admitted .

Investigations:

Hemoglobin electrophoresis with reflex PCR at 20 weeks revealed beta thalassemiaHbA-88.3% (normal range->1year-96.8%-97.8%)HbF-7.6% (normal range->1year-0.0%-2%)HbA2-4.1%(normal range->1year-2.0%-3.3%)Peripheral smear- dimorphic anemia with anisopoikilocytosis Osmotic fragility test/ eosin 5 malemidefor hereditary spherocytosis was negative.Iron profile done on 24 weeks showed decreased total iron binding capacity 224.3mcg/dl and increased serum ferritin 536.7ng/ml transferrin saturation was 62.12%. Iron chelation was not recommended as it was contraindicated in pregnancy. Oral iron supplementation was stopped. At 36 weeks of gestation USG was normalAt term hemoglobin level was 7.5g/dl

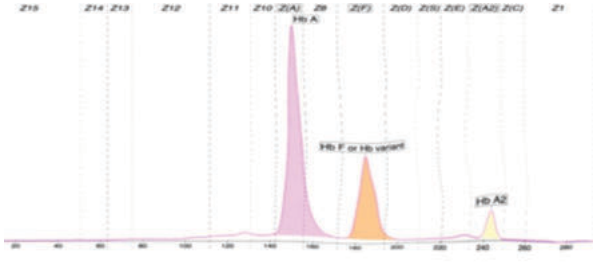

Figure 2: Haemoglobin Electrophoresis

On examination patient is moderately built and nourished, afebrile, no edema.Pallor was presentBP-110/70mmHg, PR- 72bpm. **Systemic Examination:** CVS/CNS/RS- Normal PER **Abdominal Examination:** Uterine height was term size, irritableGrips showed cephalic presentation and head was engaged.Fetal heart sound was present on left spino-umbilical line. CTG- Non reassuring.

Management: After complete investigations emergency lower segment caesarean section was done in view of fetal distress. Intra operative findings: Face to pubis presentation , short cord was noted, liquor was clear and adequate. A live male baby weighing 2.97kg was extracted and its APGAR score was 7/10 in one minute and 9/10 in 5 minutes. Approximate blood loss was nearly 500-600ml. One pint of packed red blood cells was transfused post operatively.

Figure 3: Image Showing Healthy Mother and Baby Before and After Delivery

DISCUSSION

In thalassemia intermedia, mutations can sometimes occur in one or both alleles causing moderate impairment of beta chains production. Woman's clinical condition is intermediate between major and minor forms of beta thalassemia. Thalassemia intermedia is defined as a group of patients with beta thalassemia whose disease severity varies. At the end of the spectrum, there are patients who are completely asymptomatic until adulthood. Further definitive diagnosis can be done by globin chain synthesis studies and DNA analysis.In these patients during pregnancy, if there is worsening maternal anemia or evidence of FGR, regular transfusions to be considered until target hemoglobin is managed.

CONCLUSION

Inherited autosomal recessive disorder like thalassemia is a major global health concern. Thalassemia syndromes should be suspected in pregnancy in patients with constant anemia.Beta thalassemia intermedia during pregnancy should be considered high risk and can present unique management challenges and requires close maternal and fetal surveillance.Multidisciplinary team incorporating genetic counceller, fetal medicine specialist, hematologist, approach is required for management of pregnancy with thalassemia. Time and mode of delivery should be individualized.Proper management of pregnant thalassemia patient by appropriate fetal and maternal monitoring during the preconceptional, antenatal, intrapartum, and postpartum periods can considerably help reduce the associated maternal and neonatal mortality and morbidity.

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