Homozygous Beta Thalassemia Presenting As Neonatal Jaundice

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In hemolytic anaemia there is shorter than normal erythrocytic survival resulting in increased destruction of red blood cells (RBCs) which leads to raised bilirubin levels. Appearance of jaundice, however, depends not only Upon increased biirubin production but also on clearance mechanisms in the liver. These clearance mechanisms are not well developed in the neonates resulting in greater incidence of hemolysis related jaundice as compared with adults and older children[1]. Anemias caused by the production of an abnormal type of haemoglobin or those that are a result of abnormal .haemoglobin synthesis, such as beta thalassaemia, rarely manifest themselves in the first week of life[2]. At birth fetal hemoglobin (HbF) accounts for 50 to 60% of haemoglobin production. This declines to about 5% by the age of jhree months when most of it is replaced by adult type of hemoglobin containing two alpha and two beta globin chains. Gamma globin production is usually unimpaired in utero in individuals with severe beta thalassemia, thus, only when such newborns need to replace their fetal red cells with cells that contain predominantly haemoglobin A. does the defect in beta globin synthesis become apparent[3]. We present a case of homozygous beta thalassemia which developed hyper bilirubinemia soon after birth and presented at the age of 3 months with persistent jaundice since birth.

Case Report

A 3 months old male child presented to the Children's Hospitaiwithahistory ofyellowishdiscolourationofskinand sciera sincebirth. His stools were yellowish in colour and there was a histor of highcoloured urine which was persisting. The child was exclusively breast fed and. there were no problems with either his feeding or growth. Pregnancy and delivery were uneventful and he was of normal birth weight. He had an older male sibling who was normal. On examination, he was well-nourished and his weight was at the 75th percentile. He was an active alert baby with jaundice. His liver was 4 cms and spleen 3 cms palpable below the costal margin. All other systems were normal. Investigations revealed a serum bilirubin of 3.2 rng/dl with un conjugated bilimbin being 2.5 mg/dl. Liver enzymes were within normal limits, haemoglobin was 4.4 G/dl with reticulocyte Count of 18%. Peripheral smear showed microcytosis (Mean Corpuscular Volume: 75.6) and hypochromia (Mean corpuscular haernoglobin: 22.3). Baby's glucose 6 phosphate dehydrogenase screening revealed a normal value of 9.1 u/g. Hb. Both direct and indirect coornbs test were negative on two occasions. Haemoglobin electrophoresis at pH 8.6 showed Hb F of 67%. Haemoglobin electrophoresis of both the parents showed both to be positive for beta thalassemia trait. The child was t.ransfused with packed cells, twice in one week. Haemoglobin at the time of discharge was 8.9 G/dl. Jaundice had clear ed on discharge one week later, except for the yellow discolouration of sciera. Repeat liver function tests showed a fall in bilirubin to normal. On follow-up two weeks later, the haemoglobin had reduced to 8 Gm/dl with 7% reticulocyte count and there was no evidence of jaundice.

Discussion

The usual presentation of beta thalassernia is between 3 to 6 months of age with increasing pallor and hepatosplenomegaly. Before this age, the diagnosis of beta thalassemia is unusual due to lack of

clinical symptomatology in this age group. However, any disorder that causes the fetus to destroy his existing red blood cell population will result ma replacement with red cells having a markedly different HbF:HbA ratio at the time of delivery. Fetomaternal blood group incompatibility and intrauterine blood loss may, for example, unmask a beta chain defect earlier than might otherwise occur. In the absence of such causes, it is rare for beta thalassemia to present in the newborn period[4] Erlandson and Hilgarter[5] diagnosed four infants as having thalassemia major between 1 to 2 months of life. All four infants had abnormally low hemoglobin concentrations. In one infant erythrocytic morphological abnormalities were present as early as three days of age. None of these infants had hype thilimbinemia. Association between neonatal hyper bilirubinemia and gamma beta thalassenlia has been documented[6]. Oort et al[7], described an extensive dutch pedigree in which nine newborn infants had a syndrome of severe hemolytic anemia caused by beta-gamma thalassemia. Many of these infants had severe hyperbilirubinemia and five of the six survivors needed exchange transfusion. In this case, it is difficult to draw any definitive conclusions regarding the association of neonatal jaundice and beta thalassemia due to the long delay in seeking medical help. The baby presented to us at three months of age when the opportunity to examine and investigate him at birth had already been lost. Although, the blood group of both the mother and baby was B positive and coombs test was negative on two occasions the possibility of minor blood group incompatibility as the cause of hypethilirubinemia present at birth, cannot be completely ruled out. Likewise the possibility of intrauterine blood loss cannot be ruled out on the basis of history alone, specially in the absence of any antenatal and delivery records. On the other hand, persistence of un conjugated hyper bilirubinemia with a very high reticulocyte count in the absence of any other cause of haemolysis at the time of jresentation and its disappearance after blood transfusion strongly supports our impression that jaundice at birth was due to beta thalassemia, which may or may not have been complicated by some hernolytic episode during intrauterine life or at the time of birth.

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