

CASE REPORTS

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Nomimmune Hydrops Fetalis due to Diamond-Blackfan Anemia

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We describe case report of a baby with Diamond Blackfan anemia, who presented as non-immune hydrops fetalis. The diagnosis was confirmed by measurement of red cell adenosine deaminase activity which is increased in Diamond-Blackfan anemia. At 2 years of age he is dependent on small dose of alternate day steroid to maintain his hemoglobin.

Key words: *Congenital pure red cell aplasia, Diamond-Blackfan anemia, Hydrops fetalis.*

A male baby was born at 39 weeks gestation by emergency caesarean section for fetal distress. Pregnancy was eventful till then. Parents were unrelated and the previous 3 children were healthy.

The baby was pale and floppy at birth,

weighed 3.4 kg and required immediate institution of ventilatory support. He was also noted to have generalized edema, hydrocoele and ascitis but no pleural effusion. In the initial few hours the baby was stabilized by correction of severe metabolic acidosis (base excess of -18 mmol/L), anemia (Hemoglobin 5.4 g/dL) and institution of ionotropic support. On the first day baby also developed convulsions, which necessitated phenobarbitone, phenytoin and clonazepam. CT scan showed hematoma in the temporal lobe and EEG showed burst suppression. He received prophylactic antibiotics and intravenous Aciclovir for possible herpes infection. However, maternal and baby's herpes serology was negative subsequently. On day 3 he developed persistent pulmonary

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hypertension, which was treated with tolazoline. Other metabolic problems like hypocalcemia, hypoglycemia and hematological problems like disseminated intravascular coagulation were treated appropriately. The screening for hydrops fetalis showed negative Coomb's test, TORCH serology, normal karyotype, echo cardiogram and no abnormality of urinary amino and organic acids. The baby had a total of 4 packed cell transfusions during the neonatal period and was discharged home on 25th day of life.

He was readmitted to the children's ward at 10 weeks of age with hemoglobin of 3.2 g/dL and reticulocyte count of <1% with normal leucocyte and platelet count. Bone marrow aspiration showed absence of erythropoiesis and increased red cell adenosine deaminase activity, confirming the diagnosis of Diamond-Blackfan anemia. The anemia responded to oral prednisolone but attempts to wean it off were unsuccessful. At 2 years of age baby was still steroid dependent, weight remained between 1 and 9 centile. His development is mildly delayed for the age.

Discussion

Hematological disorders are implicated in approximately 10% to 27% of cases of nonimmune hydrops fetalis(1). Diamond-Blackfan anemia is an unusual cause of nonimmune hydrops fetalis(2). Anemia, short stature, developmental delay, normocellular

bone marrow with erythroid hypoplasia, characterizes the disease. Red cell adenosine deaminase activity is increased in Diamond-Blackfan anemia and its measurement is useful in diagnosing this condition(3). A majority of patients respond to prednisolone, and often erythropoiesis can be maintained with low doses of the drug. Both remissions and increased resistance to steroid treatment can occur. Patients who do not respond to treatment are usually transfusion dependent. We are at present attempting to taper and stop steroid. However, this may incur patient becoming transfusion dependant. One of the siblings of this child is HLA matched for bone marrow donation. Bone marrow transplantation in Diamond-Blackfan anemia can be curative(4).

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