Journal of Pediatric Sciences

Diamond Blackfan Syndrome

Rahul Sinha, Daljit Singh, Kirandeep Sodhi, YK Kiran, Biju John

Journal of Pediatric Sciences; 2010; 2; 1e

How to cite this article:

Rahul S., Daljit S., Kirandeep S., Kiran Y.K., John B. Diamond Blackfan Syndrome.
Journal of Pediatric Sciences 2010;2:1e

CASE REPORT

Diamond Blackfan Syndrome

Rahul Sinha[1], Daljit Singh[2], Kirandeep Sodhi[1], YK Kiran[1], Biju John[1]

Abstract: *We report a case of Diamond Blackfan syndrome in 6yr old girl who was detected to have severe anaemia on D4 of life. The baby was detected to have polydactyly right hand (preaxial) and weak radial pulse on right side. On examination there was severe pallor without hepatosplenomegaly. The investigations revealed haemoglobin of 1.9 gm% with reticulocyte count of 0.3%. Other investigations were done to establish the cause of anaemia. The sickling test was negative, Peripheral blood smearrevealed macrocytic anaemia, Hb electrophoresis revealed fetal haemoglobin of 2.7 %. Bone marrow examination revealed markedly reduced erythroid series, stress cytogenetics study done later was negative for any chromosomal breakage. Based on the clinical profile and investigation reports the diagnosis of Diamond Blackfan Syndrome was made. The child was put on corticosteroids which were gradually tapered. Subsequently any attempt at withdrawl of steroids resulted in fall inhaemoglobin levels. Hence the child has been maintained on low dose steroids and has remained symptom free.*

Key words*: Diamond Blackfan, anaemia*
Received: 30/12/2009; Accepted: 08/01/2010

Introduction

Diamond-Blackfan anemia (DBA) is a congenital erythroid aplasia that usually presents in infancy [1]. DBA patients have low red blood cell counts (anemia). The rest of their blood cells (the platelets and the white blood cells) are normal. A variety of other congenital abnormalities may also occur.

Case Report

We report a case of Diamond Blackfan syndrome in 6yr old girl who is under follow up in our hospital. This child was born to non consanguineous parents at term by lower segment caesarean section with birth weight of 3 kg. The baby was detected to have polydactyly right hand (preaxial) [Fig.1] and weak radial pulse on right side. There was no hyperpigmentation or dysmorphic features. The immediate post natal period was uneventful. The child was asymptomatic till 72 days of life when she was first brought to medical attention with complaints of poor feeding and lethargy. On examination she was found to be normal for height and weight with severe pallor without hepatosplenomegaly. The investigations revealed haemoglobin of 1.9 gm% with reticulocyte count of 0.3%. The child was initially managed with packed cell transfusion. Other investigations were done to establish the cause of anaemia. The sickling test was negative, Peripheral blood smear revealed macrocytic anaemia, Hb electrophoresis revealed fetal haemoglobin of 2.7 %. Bone marrow examination revealed markedly reduced erythroid series; stress cytogenetics study done later was negative for any chromosomal breakage. Based on the clinical profile and investigation reports the diagnosis of Diamond Blackfan Syndrome was made. The child was put on corticosteroids which were gradually tapered. Subsequently any attempt at withdrawal of steroids resulted in fall in haemoglobin levels. Hence the child has been maintained on low dose steroids and has remained symptom free.

Discussion

Diamond and Blackfan described congenital hypoplastic anemia in 1938. In 1997 a region on chromosome 19 was determined to carry a gene mutated in DBA [2]. In 1999, mutations in the ribosomal protein S19 gene (RPS19) were found to be

Rahul S.[1] MD, Daljit S.[2], MD and Kirandeep S.[3] MD
Command Hospital Bangalore, [1]Department of Pediatrics, [2]Department of Neonatalogy, , India

Corresponding Author: ***Rahul Sinha, MD***
Command Hospital Bangalore, Department of Pediatrics, India
e-mail: drrahul_2000@yahoo.com

Conflict of interest: *non identified*

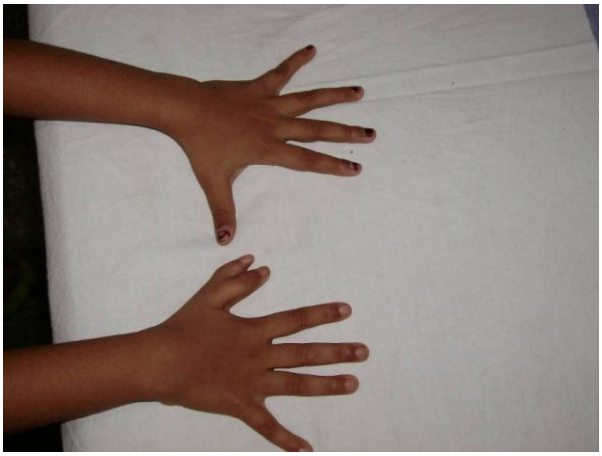

Figure.1 Showing photograph of right hand showing polydactyl (preaxial)

associated with disease in 42 of 172 DBA patients [3]. Diamond-Blackfan anaemia is a congenital erythroid aplasia that usually presents in infancy as severely hypoplastic macrocytic anaemia. Approximately 30 to 40% of patients have other congenital anomalies, particularly of the upper limb and craniofacial regions. Although the majority of cases are sporadic, approximately 10 to 25% are familial, with most showing autosomal dominant inheritance [4]. Some children may not develop anaemia until later on in childhood. Hand deformities include a triphalangeal thumb and thenar muscle hypoplasia. There may be weak radial pulse. Many affected children are very short for their age, and may have delayed puberty. Development is otherwise normal and it is unusual for affected children to have learning difficulties. Leukocyte and platelet counts are normal or slightly reduced. DBA patients have a modest risk of developing leukemia and other malignancies. A diagnosis of DBA is made on the basis of anemia, low reticulocyte (immature red blood cells) counts, and diminished erythroid precursors in bone marrow. Features that support a diagnosis of DBA include the presence of congenital abnormalities, macrocytosis, elevated fetal hemoglobin, and elevated adenosine deaminase levels in red blood cells. Most patients are diagnosed in the first two years of life. However, some mildly affected individuals only receive attention after a more severely affected family member is identified. About 20-25% of DBA patients may be identified with a genetic test for mutations in the RPS19 gene. Corticosteroids can be used to treat anemia in DBA. In a large study of 225 patients, 82% initially responded to this therapy, although many side effects were noted [5]. Some patients remained responsive to steroids, while efficacy waned in others. Blood transfusions can also be used to treat severe anemia in DBA. Periods of remission may occur, during which transfusions and steroid treatments are not required. Bone marrow transplantation (BMT) can cure hematological aspects of DBA. This option may be considered when patients become transfusion-dependent because frequent transfusions can lead to iron overloading and organ damage. However, data from a large DBA patient registry indicated that adverse events in transfusion-dependent patients were more frequently caused by BMTs than iron overloading [6]. A recent study in Japan found an 85% success rate with haematopoietic stem cell transplantation [7]. There is an increased risk of leukemia in these children [8]. In our case child had polydactyly, weak radial pulse, with marked reduced erythroid series in bone marrow and macrocytic anaemia and good response to low dose steroids. Being steroid responsive, she is not a candidate for bone marrow transplantation.

We report this case to highlight the possibility of this syndrome especially in anaemia of infancy with polydactyly and anatomical variation of blood vessels.

REFERENCES

1. Diamond LK, Allen DW, Magill FB. Congenital (erythroid) hypoplastic anemia: a 25 year study. 1961;102: 403-15.

2. Gustavsson P, Willing TN, van Haeringen A, Tchernia G, Dianzani I, Donner M, Elinder G, Henter JI, Nilsson PG, Gordon L, Skeppner G, van't Veer-Korthof L, Kreuger A, Dahl N. Diamond-Blackfan anaemia: genetic homogeneity for a gene on chromosome 19q13 restricted to 1.8 Mb. 1997; 16: 368-71

3. Draptchinskaia N, Gustavsson P, Andersson B, Pettersson M, Willig TN, Dianzani I, Ball S, Tchernia G, Klar J, Matsson H, Tentler D, Mohandas N, Carlsson B, Dahl N. The gene encoding ribosomal protein S19 is mutated in Diamond-Blackfan anaemia. 1999; 21: 168-75.

4. Orfali KA, Ohene-Abuakwa Y, Ball SE. Diamond Blackfan anaemia in the UK: clinical and genetic heterogeneity. Br J Haematol. 2004;125:243-52.

5. Draptchinskaia N, Gustavsson P, Andersson B, Pettersson M, Willig TN, Dianzani I, Ball S, Tchernia G, Klar J, Matsson H, Tentler D, Mohandas N, Carlsson B, Dahl N. The gene encoding ribosomal protein S19 is mutated in Diamond-Blackfan anaemia. 1999; 21: 168-75.

6. Roy V, Perez WS, Eapen M, et al. Bone marrow transplantation for diamond-blackfan anemia. Biol Blood Marrow Transplant. 2005;11:600-8.

7. Ohga S, Mugishima H, Ohara A, et al. Diamond-Blackfan anemia in Japan: clinical outcomes of prednisolone therapy and hematopoietic stem cell transplantation. Int J Hematol. 2004;79:22-30

8. Krishnan EU, Wegner K, Garg SK. Congenital hypoplastic anemia terminating in acute promyelocytic leukemia.1978;61:898-901.