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 a 6-year-old girl who was detected to have severe anemia 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 hemoglobin of 1.9 gm% with reticulocyte count of 0.3%. Other investigations were done to establish the cause of anemia. The sickling test was negative, peripheral blood smear revealed macrocytic anemia, Hb electrophoresis revealed fetal hemoglobin 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 hemoglobin 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
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