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A CASE REPORT OF DIAMOND-BLACKFAN ANEMIA IN A CHILD

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Abstract: Diamond–Blackfan anemia (DBA) is a rare congenital bone marrow failure syndrome characterized by normochromic macrocytic or normocytic anemia with markedly reduced or absent erythroid precursors in the bone marrow. It typically presents in early infancy. Once diagnosed, corticosteroids and red blood cell transfusions constitute the mainstays of treatment, although some patients ultimately require hematopoietic stem cell transplantation. To describe the clinical and hematological characteristics of a one-year-old female child diagnosed with Diamond–Blackfan anemia. We report the case of a one-year-old female child who presented with severe anemia and reticulocytopenia, requiring regular red blood cell transfusions from the fourth day of life. Bone marrow examination revealed selective suppression of the erythroid lineage, confirming the diagnosis of Diamond–Blackfan anemia. It should be strongly suspected in infants, with or without associated congenital anomalies, who present early in life with anemia and reticulocytopenia. Early recognition is essential for appropriate management and long-term follow-up.

Keywords: Diamond–Blackfan anemia; congenital pure red cell aplasia; macrocytic anemia; reticulocytopenia; bone marrow failure; erythroid hypoplasia

Introduction

Diamond–Blackfan anemia (DBA) is a rare congenital bone marrow failure syndrome characterized by selective impairment of erythropoiesis. It presents with severe normochromic macrocytic or normocytic anemia, reticulocytopenia, and a marked reduction or absence of erythroid precursors in the bone marrow, while leukocyte and platelet counts are usually preserved. Approximately 90% of patients are diagnosed within the first year of life, often during early infancy[1].

In addition to hematologic abnormalities, 40–50% of patients with DBA exhibit congenital anomalies, most commonly involving the craniofacial region, upper limbs, cardiovascular system, and genitourinary tract[2]. At the molecular level, DBA is most frequently caused by mutations in genes encoding ribosomal proteins, such as RPS19, leading to defective ribosome biogenesis and increased apoptosis of erythroid progenitor cells through activation of the p53 pathway[3],[4]. These findings classify DBA as a prototype ribosomopathy.

Management of DBA includes corticosteroids as first-line therapy, with chronic red blood cell transfusions required in steroid-refractory or steroid-dependent patients. Hematopoietic stem cell transplantation remains the only curative treatment for severe cases[2],[5]. Early diagnosis is essential to optimize therapy, monitor complications such as iron overload, and provide appropriate long-term follow-up and genetic counseling.

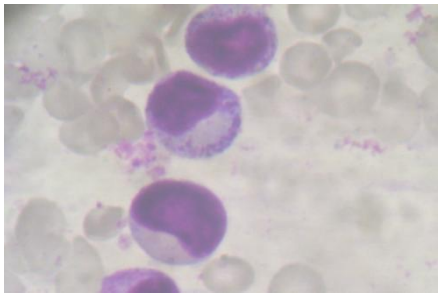
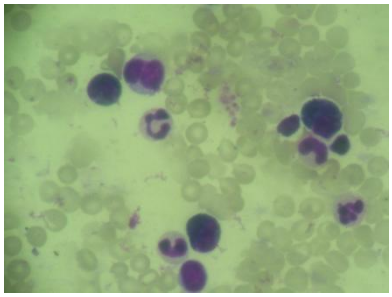
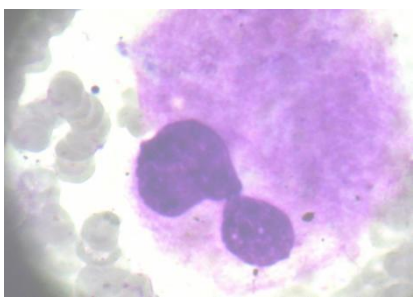
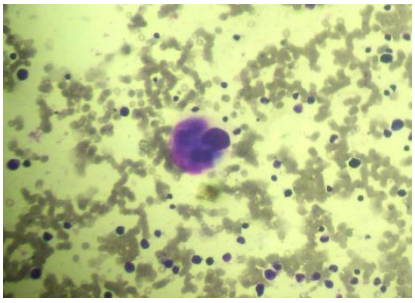
Case Report

One year old female child was brought to the emergency with the complaint of progressive pallor for one month. She was a full term hospital delivery with appropriate weight for the gestation without any antenatal and perinatal complications. On the fourth day of life, she was admitted in

the hospital for blood transfusion because of pallor. After that she had been admitted five times in the hospital for workup and blood transfusion. There was no history of consanguinity amongst parents and she had no affected siblings.

On examination, she was alert and had weight of 7 kg (71.4% of expected), length of 72 cm (96% of expected) and head circumference of 43 cm (below 5th centile). She had severe pallor and mild hepatomegaly. Spleen was not palpable. She didn't have thumb anomaly. No other congenital anomalies were observed during physical examination. Other systems were normal. Investigations revealed hemoglobin of 1.9 gm% with a reticulocyte count of 0.2%. Total leucocyte count was 9,400/cu mm and platelet count was 140,000/cu mm. RBC count was 2,550,000/cu mm and peripheral smear revealed normochromic and normocytic red cells without any immature cells. The MCV was 100.7 fL. Fetal hemoglobin (HbF) was 1.4%. Skeletal survey was normal.

The diagnosis was confirmed on bone marrow aspiration, which showed marked erythroid hypoplasia with normal megakaryopoiesis and myelopoiesis. She was given one transfusion of whole blood at 20ml/kg body weight, following which she was given oral prednisolone in the dose of 2 mg/kg body weight/day in three divided doses. She came to follow up only after one month of prednisolone and at that time her Hemoglobin was 15.1 gm% and RBC count was 4,920,000/cu mm. The total leucocyte count was 10,200/cu mm and Reticulocyte was 0.3%. Prednisolone was planned to taper slowly till the effective Hemoglobin level is stabilized then will be continued in the lowest effective dose.

These bone marrow slides of the patient show present of all other bone marrow precursors except red cells precursors. There is severe erythroid hypoplasia.

Discussion

Congenital pure red cell aplasia was first described by Josephs in 1936, and subsequently by Diamond and Blackfan in 1938, leading to recognition of DBA as a distinct clinical entity[1]. The disorder typically presents in infancy, although cases diagnosed later in childhood have been reported. A slight male predominance has been observed, with a male-to-female ratio of approximately 1.2:1[2].

Congenital anomalies are present in 25–33% of patients and commonly involve craniofacial structures, upper limbs, cardiovascular system, and genitourinary tract[2],[4]. The coexistence of malformations and erythroid failure suggests a shared pathogenic mechanism affecting both embryogenesis and hematopoiesis, though the precise link remains unclear[4]. Most cases are sporadic, while approximately 10% exhibit familial inheritance, usually autosomal dominant. Mutations in the ribosomal protein S19 gene on chromosome 19q13.2 account for nearly one-quarter of cases, with additional loci, including chromosome 8p, implicated[4],[5].

The primary defect in DBA is a reduction in number or function of erythroid progenitors, specifically BFU-E and CFU-E[4]. Elevated serum erythropoietin suggests insensitivity of progenitors to erythropoietin stimulation[6]. Although immune-mediated mechanisms were previously proposed, subsequent studies have yielded inconsistent results, and immunologic suppression is no longer considered central to pathogenesis[7],[8].

The main differential diagnosis is transient erythroblastopenia of childhood (TEC), which presents later, lacks congenital anomalies, has normal mean corpuscular volume and fetal hemoglobin, and typically resolves spontaneously within 1–2 months[2].

Diagnosis relies on established criteria combining clinical, hematologic, and supportive features[11]. Therapeutically, corticosteroids remain first-line treatment, with response rates of 60–70%[2]. Red blood cell transfusions remain essential for steroid-refractory patients, while allogeneic hematopoietic stem cell transplantation offers a definitive cure in selected cases[9],[10]. Early recognition and long-term multidisciplinary follow-up are critical to improve outcomes and minimize morbidity.

Conclusion

Diamond–Blackfan Anemia (DBA) is a rare congenital red cell disorder presenting in infancy, often with associated congenital anomalies. Most cases are sporadic, though some are inherited, frequently involving ribosomal protein gene mutations. The primary defect is impaired erythroid progenitor function, leading to anemia and elevated erythropoietin. Diagnosis is clinical and hematologic, with corticosteroids as first-line therapy, transfusions for refractory cases, and stem cell transplantation as a definitive cure. Early recognition and multidisciplinary management are essential for improving outcomes.

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