Diamond-Blackfan Anemia

Diamond-Blackfan Anemia is a type of pure red cell aplasia that presents as macrocytic anemia in infancy. On examination, the patient may display short stature, craniofacial anomalies, and triphalangeal thumbs. Labs will show increased fetal hemoglobin. Patients can be managed with red blood cell transfusion and steroids. Allogenic stem cell transplantation is curative.

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Characteristics

Red Blood Cell Aplasia
RBC Plate
Red Blood Cell Aplasia is the absence of erythroid precursors in the bone marrow leading to decreased reticulocytes. It is thought to be due to IgG antibodies against erythroblasts.

Macrocytic Anemia
Macaroni Anemone
Macrocytic anemia is defined as anemia with a red blood cell mean corpuscular volume (MCV) of greater than 100. It is mainly due to DNA maturation defects. In Diamond-Blackfan anemia, this is a consequence of “stress erythropoiesis” and skipped erythroid cell divisions.

Clinical Features

Short Stature
Shorts Statue
A child who is less than two standard deviations below the mean in height for children of the same age and sex is said to have short stature, which is characteristic of this disease.

Craniofacial Anomalies
Crane Building Face and Anomaly-Animal
Craniofacial abnormalities like microcephaly, micrognathia, hypertelorism, flat nasal bridge, and cleft palate may be evident in these patients.

Triphalangeal Thumbs
Triphalangeal Thumbs-Up
Normally the thumb has two phalanges (proximal and distal). If it has three phalanges, it is called triphalangeal thumb. Patients with DBA can often demonstrate this clinical feature.

Diagnosis

Increased HbF (Fetal Hemoglobin)
Up-arrow He-man-globe with Fetus
Fetal circulation has hemoglobin made up of two alpha chains and two gamma chains. It has a higher affinity for oxygen than adult hemoglobin. In Diamond Blackfan anemia, HbF is elevated but total hemoglobin levels are low.

Management

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RBC Transfusion

Blood Transfusion

Patients receiving chronic red cell transfusion therapy generally require transfusions of 10 to 15 mL/kg every 3 to 5 weeks to maintain a hemoglobin level of more than 8 g/dL.

Steroids

Steroid-stairs

In Diamond-Blackfan anemia, abnormal T cell function leads to IgG antibodies against erythroid progenitor cells. Immunosuppression is therefore important.

Hematopoietic Stem Cell Transplantation

Stems with Blood Cells in Transplant Train-plant

Allogeneic hematopoietic stem cell transplantation is the only curative option for patients with Diamond-Blackfan anemia. Pluripotent stem cells are transplanted from a donor with healthy RBCs.

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