

A 4-month-old male infant is brought to the office by his parents due to progressive lethargy, poor feeding, fatigue and increasing pallor for the past four weeks. His antenatal and birth histories are unremarkable. His diet consists mainly of breast milk. His immunizations are up-to-date. His mother's blood type is O+. Physical examination reveals a webbed neck, cleft lip, shielded chest, triphalangeal thumbs, and pale mucous membranes and conjunctivae. Cardiac auscultation reveals mild tachycardia and a systolic ejection murmur over the left upper sternal border. The initial investigations reveal the following:

Hb	8 g/dL
Ht	26 %
WBCs	7,000/cmm
Platelets	300,000 /cmm
Reticulocytes	0.4%
MCV	104 fL
Blood type	A -
Bilirubin direct	0.1 mg/dL
Bilirubin total	1.0 mg/dL

What is the most likely diagnosis?

- ☐ A. Wiskott-Aldrich syndrome
- ☐ B. Transient erythroblastopenia of childhood
- ☐ C. Idiopathic aplastic anemia
- ☐ D. Fanconi's anemia
- ☐ E. Diamond-Blackfan anemia
- ☐ F. Rhesus incompatibility

Submit



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What is the most likely diagnosis?

- ☐ A. Wiskott-Aldrich syndrome [8%]
- ☐ B. Transient erythroblastopenia of childhood [7%]
- ☐ C. Idiopathic aplastic anemia [3%]
- ☐ D. Fanconi's anemia [27%]
- ☒ E. **Diamond-Blackfan anemia** [50%]
- ☐ F. Rhesus incompatibility [5%]

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### Explanation:

User Id: [REDACTED]

Suspect Diamond-Blackfan syndrome (DBS), also called congenital hypoplastic anemia, in a child with macrocytic anemia, low reticulocyte count, and congenital anomalies. The majority of cases are sporadic, although dominant and recessive inheritance is found in 15 percent of cases. The primary pathology is an intrinsic defect of erythroid progenitor cells which results in increased apoptosis (programmed cell death). The condition often presents with pallor in the neonatal period. Over 90 percent of cases are diagnosed within the first year of life, with the average age of diagnosis being 2 months. Congenital



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Suspect Diamond-Blackfan syndrome (DBS), also called congenital hypoplastic anemia, in a child with macrocytic anemia, low reticulocyte count, and congenital anomalies. The majority of cases are sporadic, although dominant and recessive inheritance is found in 15 percent of cases. The primary pathology is an intrinsic defect of erythroid progenitor cells which results in increased apoptosis (programmed cell death). The condition often presents with pallor in the neonatal period. Over 90 percent of cases are diagnosed within the first year of life, with the average age of diagnosis being 3 months. Congenital anomalies (as described in this case) are present in over 50 percent of cases.

The macrocytic anemia of DBS is distinct from that of megaloblastic anemia because there is no hypersegmentation of the nucleus in neutrophils and other blood cells in the former. Electrophoresis reveals elevated fetal Hb levels. Chromosomal studies are normal. Therapy is mainly corticosteroids. For unresponsive patients, transfusion therapy is indicated.

**(Choice A)** Wiskott-Aldrich syndrome is an X-linked disorder characterized by eczema, thrombocytopenia and hypogammaglobulinemia.

**(Choice B)** Transient erythroblastopenia of childhood is a pure red cell aplasia without macrocytosis. Most cases are diagnosed after 1 year of age and there are no associated congenital anomalies.

**(Choice C)** Idiopathic aplastic anemia is an acquired disease that results in pancytopenia. It may be due to chemicals (e.g., benzene, phenylbutazone), drugs (e.g., chloramphenicol, sulfonamides), infectious agents (e.g., viral hepatitis) or ionizing radiation.

**(Choice D)** Fanconi's anemia is an autosomal recessive disorder characterized by progressive pancytopenia and macrocytosis. The average age at diagnosis is 8 years. Associated deformities include cafe-au-lait spots, microcephaly, microphthalmia, short stature, horseshoe kidneys and absent thumbs (not triphalangeal thumbs).

**(Choice F)** The mother in this vignette is rhesus-positive; therefore, there is no risk of rhesus incompatibility for the baby.

**Educational Objective:**

DBS is a macrocytic pure red aplasia associated with several congenital anomalies such as short stature, webbed neck, cleft lip, shielded chest and triphalangeal thumbs.

Time Spent: 2 seconds

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