

A 7-year-old boy is brought to the physician by his mother because of bleeding gums for the past 3 months. The mother reports that he regularly brushes his teeth and visits a dentist twice per year. Examination at his most recent dentist visit 6 months ago was normal. The patient also complains of easy fatigability and a pounding sensation in his ears. He has no other medical problems and takes no medication. Physical examination shows that he is at the 5th percentile for height and 25th percentile for weight for his age. His thumbs are slightly bent, and several areas of hypopigmentation are noted on his skin. Laboratory findings reveal:

WBC count	3000/mm ³
Hemoglobin	7.8 g/dL
RBC count	3 million/mm ³
MCV	112 fL
Platelet count	40,000/mm ³

Which of the following is the most likely cause of this patient's condition?

- ☐ A. Chromosomal breaks
- ☐ B. RBC enzyme deficiency
- ☐ C. Congenital infection
- ☐ D. Benzene exposure
- ☐ E. Thymic tumor
- ☐ F. Cobalamin deficiency

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Which of the following is the most likely cause of this patient's condition?

- ☒ A. Chromosomal breaks [48%]
- ☐ B. RBC enzyme deficiency [8%]
- ☐ C. Congenital infection [5%]
- ☐ D. Benzene exposure [13%]
- ☐ E. Thymic tumor [4%]
- ☐ F. Cobalamin deficiency [21%]

Proceed to Next Item

Explanation:

User Id: [REDACTED]

Acquired Causes of Aplastic Anemia

Drugs (e.g., NSAIDs, sulfonamides, etc.)

Toxic chemicals (e.g., benzene, glue, etc.)

Explanation:

User Id: **Acquired Causes of Aplastic Anemia**

Drugs (e.g., NSAIDs, sulfonamides, etc.)

Toxic chemicals (e.g., benzene, glue, etc.)

Idiopathic

Viral infections (e.g., HIV, EBV, etc.)

Immune disorders

Thymoma

This patient presents with symptoms and laboratory values consistent with aplastic anemia, which can be acquired or congenital as shown above. Congenital causes are more common in children, and Fanconi anemia (FA) is the most common congenital cause. It is an autosomal recessive or X-linked disorder associated with the clinical manifestations summarized below. Most patients with FA are diagnosed by the age of 16 years and have a predisposition for developing cancer. Numerous genes, all believed to involve DNA repair, have been implicated.

Location	Clinical Manifestations of Fanconi Anemia
Bone marrow	Aplastic anemia and progressive bone marrow failure
Appearance	Short stature, microcephaly, abnormal thumbs, and hypogonadism
Skin	Hypopigmented/hyperpigmented areas, café au lait spots, and large freckles
Eyes/ears	Strabismus, low-set ears, and middle ear abnormalities (e.g., hemorrhage, incomplete development, chronic infections, deafness, etc.)

Diagnosis of FA is made by chromosomal breaks on genetic analysis combined with the clinical findings. This patient likely has bleeding secondary to thrombocytopenia, fatigue from macrocytic anemia, and pounding in his ears from possible conduction defects or

SKIN	and large freckles
Eyes/ears	Strabismus, low-set ears, and middle ear abnormalities (e.g., hemorrhage, incomplete development, chronic infections, deafness, etc.)

Diagnosis of FA is made by chromosomal breaks on genetic analysis combined with the clinical findings. This patient likely has bleeding secondary to thrombocytopenia, fatigue from macrocytic anemia, and pounding in his ears from possible conduction defects or chronic hemorrhage. The definitive treatment for aplastic anemia is hematopoietic stem cell transplantation.

(Choice B) The most common RBC enzyme deficiency causing anemia is glucose-6-phosphate dehydrogenase (G6PD) deficiency. In affected patients, oxidant drugs (e.g., antimalarials and sulfas) and infection can cause episodic hemolysis.

(Choice C) The TORCH (toxoplasmosis, other infections [e.g., syphilis], rubella, cytomegalovirus, and herpes simplex) infections are known to cause significant neonatal and perinatal morbidity and mortality. They commonly cause growth restriction but not chronic pancytopenia, which is seen in this patient.

(Choice D) Benzene is an industrial chemical known to cause aplastic anemia. Based on this patient's history, there is no reason to suspect that he has been exposed to benzene. Furthermore, benzene exposure does not explain his skin changes.

(Choice E) Approximately 5%-15% of the patients with thymic tumors have pure red cell aplasia. This finding is most common in older women. This patient does not have an isolated red cell aplasia because he is also thrombocytopenic.

(Choice F) Autoimmune pernicious anemia due to anti-intrinsic factor autoantibodies is the leading cause of cobalamin deficiency. However, this patient is somewhat young to have pernicious anemia.

Educational objective:

Fanconi anemia is an autosomal recessive disorder that causes congenital marrow failure, poor growth, morphologic abnormalities, and usually macrocytic anemia.

References:

1. [Fanconi anemia and its diagnosis](#)
2. [X-linked inheritance of Fanconi anemia complementation group B](#)