
TEACHING FILE

Anemia in a 6 month old

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Case Report: - A 6 month old male child presented with progressive pallor noticed by parents for the past two months and dark coloured stools for past 1 month. For the above complaints, child was shown to a doctor and was investigated and found to have hemoglobin of 3 gm% for which blood transfusion was given and some medications were given. However, the pallor has again increased. Birth history and immunization, milestones are normal. The child is on breast feeds only. On examination, the child is pale, has jaundice and hepatosplenomegaly. Other systems are normal. Investigations show:

- Hemoglobin = 5 gm%, WBC = 10,000/cumm (48% polymorphs, 52% lymphocytes)
- Platelet count = 2,70,000/cumm
- Reticulocyte count = 0.9%
- Peripheral smear = Hypochromia, microcytosis, target cells
- Bilirubin = 4 mg% (Indirect = 3 mg/dl, Direct = 1 mg/dl)
- Liver transaminases and albumin = Normal
- Ultrasound abdomen = Hepatosplenomegaly. No portal hypertension
- Stool occult blood = Negative

What is the diagnosis? Why did the child have dark stools?

Expert's opinion: - This child has presented with progressive anemia inspite of blood transfusion suggestive of transfusion dependent anemia. Since the indirect bilirubin is high, it suggests increased hemolysis

of RBCs. The hemolysis of RBCs could be in the spleen (in which case reticulocyte count would be high) or there is ineffective erythropoiesis in the bone marrow (in which case reticulocyte count will not be elevated). Of all the hemolytic anemias, thalassemia is associated with ineffective erythropoiesis and normal reticulocyte count. Also peripheral smear may show presence of hypochromic, microcytic anemia. Thus, most likely this child has a thalassemia major. Now regarding the dark stools, it may be due to loss of RBCs in the stools as seen with worm infestation or the child could be bleeding from the intestines. Worm infestation in a 6 month old child on breast feeds is unlikely. Also malena due to GI losses causing severe anemia without frank blood in stools is a rare etiology especially since the child also has hepatosplenomegaly. Hence dark stools could be due to iron supplements. Since the dark stools have started later and the child has been on some medicines following the blood transfusion, one would have to go more in detail about the medicine history. It is very likely that the doctor who treated the child initially would have given iron supplements for the anemia. In this child, on further detailed history, the medicine was iron supplements and dark stools stopped when iron supplements were stopped. The child's electrophoresis showed presence of HbF (60%) and thus child was diagnosed to be thalassemia major and both parents were thalassemia minor.

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