Case Report

Severe Vitamin B12 Deficiency in a Breast Fed Infant with Pancytopenia

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Summary

We report the case of a 7-month-old breast fed infant who presented with a nose bleed and bruises. Investigation showed severe nutritional B12 deficiency anemia with a pancytopenia. It is important to take the nutritional history of both the infant and the mother for early prevention and treatment.

Key words: megaloblastic anemia, pancytopenia, B12 deficiency, infant.

Megaloblastic anemia due to vitamin B12 deficiency is an uncommon problem in childhood that is most frequently associated with decreased ingestion or impaired absorption or utilization of B12 [1, 2]. Nutritional B12 deficiency in childhood is rare. Most cases are due to maternal insufficiency, resulting from deficient stores and intake generally among exclusively breastfed infants [3]. B12 deficiency in children often presents with nonspecific manifestations [4, 5]. We present an infant who manifested with severe megaloblastic anemia and pancytopenia.

Case Report

A 7-month-old boy presented to the emergency department with a sudden nose bleed, bruises and petechiae without any history of trauma. He had been taking iron medication irregularly for a month because of iron deficiency anemia. On physical examination, the patient was irritable and mucous membranes and conjunctivae were pale. He was slightly dyspneic. He had mild scleral icterus, a palpable spleen extending 5 cm below the left costal margin and small bruises and petechiae on the extremities. His neurologic examination revealed diminished deep tendon reflexes, hypotonia and tremors. The remainder of the physical examination was unremarkable.

The laboratory evaluation showed pancytopenia with hemoglobin of 5.8 g dl⁻¹, hematocrit 18%, red cell count 1.74 × 10¹² l⁻¹, mean corpuscular volume 103.8 fl, red cell distribution width 22.1%, a white blood cell count of 1.2 × 10⁹ l⁻¹, neutrophils of 0.6 × 10⁹ l⁻¹ and platelet count of 7 × 10⁹ l⁻¹. Peripheral blood smear examination showed anisocytosis, poikilocytosis, macrocytes, few platelets and few hypersegmented neutrophils. All bacterial, viral and fungal cultures were negative. All viral workup including TORCH, parvovirus and Ebstein–Barr virus using PCR were negative. Direct Coombs test was negative, serum B12 level was 69 pg ml⁻¹ (normal 180–914). The levels of folic acid and ferritine were 13.2 ng ml⁻¹ (range 3–20) and 86 ng ml⁻¹ (range 24–336), respectively. Methylmalonic acid spot test in urine was positive. Bone marrow examination showed erythroid precursors were increased in number and showed megaloblastic maturation. Granulocytes were decreased in number and many giant metamyelocytes and band forms were seen. The megakaryocytes were normal in number, although a rare, large multi-nucleated form was seen. No malignant cells were seen in the bone marrow.

Because our patient was exclusively breast fed, his mother was evaluated for vitamin B12 deficiency. There was no history of neurological or gastrointestinal symptoms in the mother, but the family’s socioeconomic status was very low, and their diet did not contain animal products. Complete blood count of the mother revealed hemoglobin 12.4 g dl⁻¹, red cell count 4.8 × 10¹² l⁻¹, hematocrit 38%, mean corpuscular volume 81 fl, platelets 328 × 10⁹ l⁻¹, white cell count 6.2 × 10⁹ l⁻¹ and reticulocyte count...
was 0.8%. The levels of serum iron, iron-binding capacity and B12 level in the mother were 28 mg dl\(^{-1}\) (50–120), 389 mg dl\(^{-1}\) (110–330) and 112 pg ml\(^{-1}\) (180–914), respectively. His mother was diagnosed as having a combined iron and B12 deficiency.

As a result of these findings, vitamin B12 deficiency due to nutritional inadequacy was diagnosed and our patient was treated with intramuscular vitamin B12 injections with a dose of 0.2 mcg kg\(^{-1}\) for 2 days followed by 1000 mcg day\(^{-1}\) for 2 weeks, followed by monthly 50 mcg for 6 months. Five days after vitamin B12 treatment began, the patient's reticulocyte count increased to 3.8%. During the recovery period iron therapy was initiated. Leukocyte and hemoglobin normalized in 1 month, platelets in 2 months. After the 3 months of treatment, his neurologic findings completely resolved.

Discussion

Nutritional B12 deficiency is an uncommon disorder resulting from nutritional inadequacy [5]. Exclusively breast fed infants of vegan and malnourished mothers, as well as infants of mothers with undiagnosed or untreated pernicious anemia, are at an increased risk for megaloblastic anemia [6, 7].

The amount of the vitamin stored by the fetus during pregnancy is a major determinant of B12 status during infancy. Reports from India and Guatemala show that the infants of long-term vegan mothers or those with a low intake of animal products develop deficiency symptoms within 3–8 months postpartum, at which time they present with anemia, growth failure and neurologic delay [8, 9].

Our patient was 7 months of age and the regression of his motor functions, as well as neurologic findings had become apparent in the last 2 months. His mother’s serum B12 level was low. Since our patient’s B12 stores were not sufficient and he had been exclusively breast fed, B12 deficiency was diagnosed and severe hematologic and neurologic signs and symptoms eventually became apparent. The history revealed that their socioeconomic status was low and their diet was not rich in animal products. Evaluating the B12 status of pregnant and lactating women would prevent newborns and infants from suffering the potentially severe consequences of B12 deficiency.

B12 deficiency in children often presents with nonspecific manifestations. In infancy and early childhood, manifestation of a neuro-developmental delay is common [10, 11]. The long-term prognosis of B12 deficiency is related to severity and duration of deficiency. For this reason, efforts should be directed toward preventing the deficiency.

A rare but a characteristic feature of B12 deficiency is tremors. Our patient had tremors, especially in his hands and arms. In the etiologies of tremors and abnormal movements, hyperglycinemia seems to be responsible [12]. The explanation for hyperglycinemia is uncertain but may be attributed to nonspecific interference with glycine cleavage [13].

The case reported here is a useful reminder that patients still present with advanced deficiency of vitamin B12. It also reminds us that severe megaloblastic anemia may present with suspicious findings similar to those seen in malignant disorders including pancytopenia and splenomegaly as in our patient. It is also important to take the nutritional history of both the child and the mother for early prevention and treatment. With early awareness and appropriate measures, potentially irreversible neurologic damage can be prevented in these infants.

References