A New Case of Association of Megaloblastic Anemia and Pancytopenia of Infants

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Introduction

Vitamin B12, an infrequent clinical entity in pediatric age, is found almost solely in breastfed infants whose mothers are purely vegetarian, non-supplemented or with pernicious anaemia. Megaloblastic anaemia in infants presents with generalized weakness or irritability. Given this disease's rarity and potential sequelae, we present the case of an infant exclusively breastfed with severe vitamin B12 deficiency whose mother had undiagnosed pernicious anaemia.

Observation

We present the case of a nine-month-old female child, born after a normal full-term pregnancy. She presented to the Pediatric Department with complaints of irritability, reduced feeding and multiple episodes of vomiting per day for two months prior to the presentation.

Results

The developmental assessment revealed that the child had Global Developmental Delay. Laboratory examination revealed Hemoglobin=6.4g/dL, White Blood Cell count=10650/mm3, Platelet=48000/mm3, MCV=100 fL, MCH=40.8pg and Reticulocyte count=4.7%. The peripheral blood examination reported that red blood cells were macrocytic and neutrophils were hypersegmented with pancytopenia (Figure 1). Bone marrow aspiration showed megaloblasts, giant metamyelocytes and hyperlobulated megakaryocytes (Figures 2, 3). Serum vitamin B12 level was found to be 84pg/mL. The maternal Serum vitamin B12 level was found to be 195.7pg/mL. The child was treated with 5000mg of vitamin B12 and hemoglobin levels and other relevant hematological parameters rapidly improved, preceding a gradual clinical improvement.

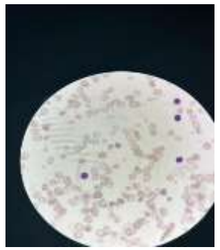

Figure 1. Depiction of macrocytosis, pancytopenia and hypersegmented neutrophils in blood film.

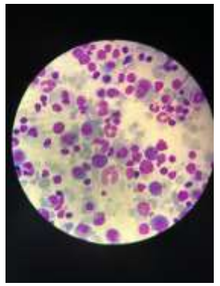

Figure 2. Medullar blood smears obtained by bone marrow puncture showed erythroblastic hyperplasia, megaloblasts and giant metamyelocytes.

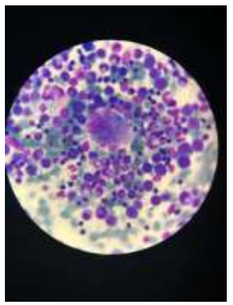

Figure 3. Medullar blood smears showed hyperlobulated megakaryocytes.

Discussion

Various factors as inborn errors of metabolism and nutritional problems, can result in deficient levels of vitamin B12 in infancy. The most frequent inborn error described is cobalamin disorder, which has been attributed to a mutation of the MMACHC gene [1]. Nutritional causes are, however, more common, and most infants found to have vitamin B12 deficiency have been noted to be born to mothers with low vitamin B12 levels and who have been exclusively breastfed. It follows, then, that the breast milk of vegan mothers is frequently poor in vitamin B12, and their newborn infants run the possibility of having low vitamin stores. Vitamin B12 deficiency has now been recognized as a fundamental cause of infant morbidity and mortality worldwide, more so in India, Mexico, Central America and certain regions of Africa [2]. Infantile vitamin B12 deficiency is essential to recognize because early recognition and treatment can prevent potentially fatal neurological sequelae. Clinical manifestations are predominantly neurologic and hematologic but are often unspecific. All blood cell lineage is affected. Red blood cells vary in size and shape and are frequently larger than usual. Anemia is macrocytic (>100 fL) and there is reticulocytopenia [2]. Vitamin B12 treatment is not well established. After therapy, it has been reported that there are clinical improvements and normalization of hematologic and neurologic parameters. In this case, after cobalamin administration and food diversification, there was a significant clinical improvement with regression of neurologic and hematologic findings.

Conclusion

This case underlines the need to consider vitamin B12 deficiency in infants with severe anaemia even if their haematological parameters do not indicate megaloblastic anaemia because the concomitant presence of substantial iron deficiency may alter the characteristics of the anemia.

References

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2.Koehler P, Cornely OA, Böttiger BW, Dusse F, Eichenauer DA, Fuchs F, et al. COVID-19 associated pulmonary aspergillosis. Mycoses. 2020;63(6):528 - 34.