

Megaloblastic anaemia in a 9-weeks old infant: A case report

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Abstract

Megaloblastic anaemia due to vitamin B12 and folic acid deficiency is uncommon in infancy and rarely reported in infants below 3 months of age. We hereby report a case of megaloblastic anaemia in a 9-weeks old infant having fever from 7th week of life. Blood picture showed pancytopenia and diagnosis was confirmed on bone marrow biopsy and serum level of vitamins. Patient positively responded to vitamin B12 and folic acid supplementation. Infants with pancytopenia even younger than 2 months, should also be investigated for vitamin B12 and folate deficiency. Mother of the baby was not antenatally investigated for anaemia. Prompt antenatal diagnosis and treatment of mothers can reduce the incidence in the infants.

Keywords: Megaloblastic anaemia, vitamin B12 deficiency, folic acid deficiency, infant

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Introduction

Megaloblastic anaemia is a condition in which there is production of large, abnormal and immature red blood cells that occurs due to inhibition of DNA synthesis during red blood cell production. Megaloblastic anaemia due to vitamin B12 and folic acid deficiency is uncommon in infancy.¹ It may occur in exclusively breast fed infants of mothers who are deficient in vitamin B12 and folic acid.² Congenital deficiency of transcobalamin can also cause megaloblastic anaemia.³ Vitamin B12 and folic acid deficiency in infants usually presents with non-specific symptoms.⁴ Laboratory investigations may reveal pancytopenia. Although signs and symptoms can appear from 2 to 12 months of age⁵ but literature review indicates that it has been rarely reported before 3 months. Nutritional deficiencies of the mothers can cause the foetus to develop megaloblastic anaemia in neonatal period.⁶ Bone marrow findings include hyper cellular marrow with all active cell stages of myelopoiesis. Hyperplastic and megaloblastic erythropoiesis is

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dominant.⁷ Prognosis is good if diagnosed timely and treatment is given for vitamin B12 and folic acid deficiency.⁸ We hereby report a case of a 9-weeks old infant with megaloblastic anaemia having fever from 7th week of life. Case has been reported after taking informed consent from the parents.

Case Report

A 9 weeks old male baby, resident of Abbottabad was presented to OPD of Federal Government Polyclinic (PGMI) on 5th October 2018 with fever for 2 weeks. Fever was high grade (102°F), intermittent and relieved by antipyretics. Fever was not associated with any other complaints. Baby was delivered by Spontaneous Vaginal Delivery (SVD) at a private hospital with good APGAR score. Baby's birth weight was 3 kg. Mother was reported anaemic in pregnancy but no detailed workup was done and mother only used iron supplements during pregnancy. Baby was also admitted to the hospital one month back for sepsis but no growth was seen on blood culture at that time and was discharged on oral medications. On examination, baby weighed 3.5 kg (<3rd percentile) with a length of 53 cm (<5th percentile) and occipitofrontal circumference of 37.5 cm (5th percentile). Baby was markedly pale with heart rate of 160/min (Normal value: 80-140/min) and respiratory rate of 40/min (Normal Value: 20-50/min). Liver, spleen and lymph nodes were not palpable. Lymph nodes were not palpable. No other specific sign apart from anaemia and failure to thrive was seen.

Laboratory investigation revealed pancytopenia in complete blood picture. Total leukocyte count was 2800/mm³ (Normal value: 4.5-11× 10³/mm³), platelet count was 10000/mm³, (Normal value: 150-350× 10³/mm³) however no active bleeding was seen and haemoglobin was 4g/dl (Normal Value: 13.5-17.5g/dl) with a haematocrit of 12% (Normal Value: 41-53%) and MCV of 92fL (Normal value: 80-100fL). Peripheral film showed few hyper-segmented polymorphs while reticulocyte count was 0.2% (Normal Value: 0.5-2.5%). C- reactive protein was within normal limits and blood culture was negative. Chest X-ray was also normal. Platelets were transfused as platelet count

was 10,000/mm³ and post transfusion blood complete picture showed platelet count of 30,000/mm³. Intravenous antibiotics were started, baby became afebrile after 48 hours however there was persistent pancytopenia in subsequent blood complete pictures. Since there was persistent pancytopenia even after the treatment of sepsis, so bone marrow biopsy was considered to rule out other causes of pancytopenia that includes aplastic anaemia, leukaemias or any other disease involving bone marrow failure. Bone marrow biopsy showed hyperplastic and megaloblastic erythropoiesis. Giant metamyelocytes were also seen. Iron stores were increased in the fragments. Findings of both blood picture and bone marrow were suggestive of megaloblastic anaemia. Vitamin B12 and folate levels of baby were determined and found to be low i.e. 90pg/mL (Normal Value: 180-350pg/mL) and 1.7ng/mL (Normal Value >3ng/mL) respectively. Since the baby was exclusively breast fed for first 6 weeks so mother's vitamin B12 and folate levels were also done were markedly low i.e. 140pg/mL and 2ng/mL respectively. Diagnosis of vitamin B12 and folic acid deficiency was made. Since the baby was neutropaenic and was not thriving well due to vitamin B12 and folate deficiency, he was more prone to infections and was admitted twice for sepsis. Baby was started on injections of vitamin B12 intramuscularly and folic acid supplements. Baby showed significant improvement over the course of few days and cell counts also started to rise. After two weeks, haemoglobin increased from 4 to 6.8 g/dl. Furthermore platelet counts also increased from 30,000/mm³ to 85000/mm³ and TLC increased from 2800/mm³ to 5000/mm³. Platelets were transfused initially during the treatment that slightly increased the platelet count only but all cell lines improved after vitamin B12 and folic acid supplementation hence the improvement in all cell lines was associated with vitamin B12 and folic acid supplementation. Mother was also treated in view of her low folic acid and vitamin B12 level. Baby was discharged on vitamin B12 and folic acid supplements. Baby came for a follow up visit after 3 months. His weight was 5.8kg (5th centile). A repeated blood picture showed haemoglobin of 10mg/dl, platelet counts of 170,000/mm³ and TLC of 7000/mm³. He was advised regular follow up on OPD basis.

Discussion

Vitamin B12 and folic acid deficiency in infants is

uncommon. It was first reported by Jadhav et al (1962) in six south Indian infants with megaloblastic anaemia.⁹ It is mostly related to maternal causes including dietary deficiencies and less commonly due to maternal pernicious anaemia.¹⁰ In the case under report, the cause of infant's vitamin B12 and folic acid deficiency was also maternal. The deficiency in infant was primarily due to low reserves transferred from mother and exclusive breast feeding from the mother with reduced levels of vitamin B12 and folic acid. This is supported by the finding of Specker et al that the vitamin B12 concentration in milk was directly correlated with vitamin B12 concentration in serum.¹⁰

Clinical presentation of megaloblastic anaemia due to vitamin B12 and folic acid deficiency can be widespread and include non-specific signs. Common sign and symptoms in infancy include anaemia, failure to thrive, lethargy, irritability, hypotonia, hyperreflexia, developmental regression and glossitis that can present from 2 to 12 months of age⁵ but literature review shows that it is rarely reported before 3 months. Pancytopenia can also be present.¹¹ In the case under report pancytopenia was present. Thus it is proposed that pancytopenia should be explored for vitamin B12 and folic acid deficiency.

Megaloblastic anaemia has been reported in infants of more than 3 months of age and very rarely below it. In the case under report, the age of presentation was rare as the symptoms were seen in the infant from 7th week of life who was breast-fed by a mother who had vitamin B12 and folic acid deficiency.

Vitamin B12 deficiency has generally been reported in infants of vegan mothers.^{12,13} In the case under report, infant was also exclusively breast fed but mother was not vegan, however the mother had poor dietary intake and was anaemic. The mother belonged to poor socioeconomic status therefore her diet lacked frequent intake of chicken, eggs, meat and fish. However her diet included locally available vegetables. In our country Jamal et al also reported a similar case of megaloblastic anaemia in a three months old child.³

Bone marrow biopsy was reported to have hyper cellular marrow with all cell stages of myelopoiesis. There was presence of hyperplastic and megaloblastic erythropoiesis.³ Our case report also had similar findings. Response to treatment is generally good provided if no

other co-existing infection or disease is present.

Conclusion

We hereby report a case of megaloblastic anaemia who presented in early infancy and was successfully treated. High suspicion for the diagnosis of vitamin B12 and folate deficiency should be kept in infants with pancytopenia. Maternal anaemia should always be investigated as prompt antenatal diagnosis and treatment of mothers with vitamin B12 and folate deficiency can reduce the incidence in the infants.

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Conflict of Interest: None to declare.

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