

Case Report

Unusual presentation of vitamin B12 deficiency in a 3 month old infant: presentation with hemolysis and thrombocytopenia

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ABSTRACT

A 3 month old infant exclusively breast fed presented with vomiting and poor weight gain with purpuric and echymotic patches all over the body. The child also had hyperpigmentation over knuckles and icterus. Laboratory investigations revealed severe dimorphic anemia with thrombocytopenia, elevated bilirubin and LDH levels and severe vitamin B12 deficiency. Following vitamin B12 supplementation there was improvement in well-being including feed tolerance, icterus resolved and in follow up lab studies there was improvement in hemoglobin and platelet counts along with reduced bilirubin levels. Through this case report we want to emphasize the possibility of vitamin B12 deficiency presenting as hemolytic anemia and psoedothrombotic microangiopathy.

Keywords: Hemolytic anemia, Icterus, Purpura, Thrombocytopenia, Vitamin B12 deficiency

INTRODUCTION

Vitamin B12 deficiency is considered rare in infants these may be seen in breast feeding mothers who themselves are deficient.^{1,2} Inadequate dietary intake of animal foods by mother and pernicious anemia are the important causes of vitamin B12 deficiency in infants and children.^{3,4}

A study has put incidence of vitamin B12 deficiency as 32%, folate deficiency as 6.8% and combined deficiency as 20% in north Indian mothers.⁵ In approximately 10% cases they may present with life threatening conditions like haemolytic anemia, pancytopenia, bleeding.⁶

The deficiency of vitamin B12 causes megaloblastic anemia. The clinical picture reveals hyperpigmentation of skin on knuckles, enlargement of liver and spleen (30-40%). Neurological signs may be present including developmental delay and regression of milestones. Purpura and bleeding manifestations are noticed in 25%

cases. Pancytopenia and hepatomegaly can make it difficult from other cause like leukemia.⁷

CASE REPORT

A 3-month-old male infant born to a non consanguinously married couple, presented with vomiting and not gaining weight noted by mother for past 1 month. The mother also noticed bluish black rashes all over the body for 1 month. The baby was born full term with birth weight of 2.8kg and there were no postnatal complications. The developmental milestones were appropriate for age. The child was immunised appropriate for age and was exclusively breast fed. The parents were pure vegetarians.

Physical examination revealed the baby was pale, sclera was icteric and bounding pulse with tachycardia was present. There was hyperpigmentation over knuckles (Figure 1), pupuric and echymotic patches (Figure 2) were present all over the body. Systemic examination revealed palpable liver with span of 5cm and also tip of

spleen was palpable. CVS examination revealed grade 3 systolic murmur.



Figure 1: Knuckle hyperpigmentation.



Figure 2: Multiple purpuric lesions.

Investigations

Thyroid profile

- T3 - 201 ng/dl(80-265)
- T4 - 13.34 mcg/dl(5.6-16)
- TSH - 3.38 mIU/ml(0.73-8.35)

Iron profile

- IRON 196.4 mcg/dl(37-158)
- TIBC - 234(228-428)
- UIBC - 320(180-370)
- FERRITIN - 268(30-400)

Urine routine

Normal

Peripheral smear

RBC: Reduced in number with dimorphic picture having microcytic hypochromic with few macrocytes with anisopoikilocytosis, elliptocytes, tear drops, fragmented

RBC and occasional polychromatophilic cells. (Mimicking microangiopathic hemolytic anemia).

WBC: normal in number with increased lymphocytes with few toxic granules, no blasts cells.

Platelet: reduced in number.

Differential diagnosis

G6PD, autoimmune hemolytic anemia, leukemia, infectious causes, vitamin B12 deficiency, aplastic anemia were considered. Clinical and laboratory investigation ruled out other causes and vitamin B12 levels were largely reduced.

Treatment

The child was started on 1000mcg of vitamin B12 in alternate day for 1 week and then weekly injection for 2 weeks was advised. Iron, folic acid supplementation was given.

DISCUSSION

Vitamin B12 is required for DNA synthesis, haematopoiesis and myelination as cofactor for methionine synthase and for methylmelanoyl coenzyme a mutase.^{3,4,8}

Humans are unable to synthesize vitamin B12 and should be ingested in diet. The most common cause of vitamin B12 deficiency in infant exclusively breast fed is maternal deficiency. Other causes like malabsorption syndromes, pernicious anemia should be kept in mind. The average requirement of vitamin B12 in infant is 0.5 - 0.6 mcg/day. In this case the parents were pure vegetarians. In a study on vitamin B12 deficiency 10% of cases had severe complications like hemolytic anemia, pancytopenia, bleeding.⁶ Our patient too had the features of hemolysis shown by peripheral smear and direct bilirubinemia there were purpuric and echymotic patches suggestive of thrombocytopenia. Concurrent hemolysis in a patient with vitamin B12 deficiency has been attributed to intramedullary destruction of erythrocytes.⁹

Cobalamine is essential for DNA synthesis and haematopoiesis and if deficient not only causes anemia but can also cause other cell deficiencies like thrombocytopenia in this case.¹⁰⁻¹² Hepatosplenomegaly can also be seen in severe vitamin B12 deficiency as it can be due to extramedullary erythropoiesis.¹⁰

There is a study conducted by Noël et al on patient with pseudothrombotic microangiopathy showed low retic count and elevated LDH levels. Low vitamin B12 as possible underlying cause.

In vitamin B12 deficiency cases cause should be identified. In case of pernicious anemia lifelong B12

supplementation is required. The parents were counselled regarding dietary habits to improve vitamin B12 sources and they were advised to continue with vitamin B12 supplementation and further follow up was advised.

The followup investigation showed improvement in Vitamin B12 levels and haemoglobin along with reduction in bilirubin and LDH levels (Table 1).

Table 1: Comparison of various laboratory parameters after starting with vitamin B12 supplementation.

Investigation names	Initial reports	Follow up after 1 month
Haemoglobin	6.9 gm%	10.2 gm%
Total count	10650 cells/cumm	8760 cells/cumm
Platelet	0.37 lakh/cumm	1.33 lakh/cumm
PCV	20.6%	31%
MCV	107.5 fl	90.5 fl
MCH	32.4 pg	33pg
MCHC	33.2%	34%
RDW	21.6%	
Retic count	1.8%	
DCT	Negative	
CRP	0.07 mg/dl (Negative)	
LDH	1350 (270-340) IU/l	241 IU/l
Total bilirubin	5.2 mg/dl	2.8 mg/dl
Indirect bilirubin	4.6 mg/dl	2.4 mg/dl
Vitamin B12	68(140-320) pmol/l	540 pmol/l

They were also told about further workup for vitamin B12 deficiency causes if the child again presents with similar complaints, including for causes like pernicious anemia, atrophic gastritis, transcobalamine deficiency.

CONCLUSION

The proper clinical suspicion is crucial for diagnosis of vitamin B12 deficiency in infant, as it is rare. Unusual presentations like hemolytic picture and bleeding manifestation to be kept in mind. Early diagnosis helps in prevention of serious neurological deficits especially in this developmental age.

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