

SEVERE VITAMIN B12 DEFICIENCY LEADING TO CEREBRAL ATROPHY

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Introduction -

Vitamin B deficiency is rare disorder in infants and is generally due to vitamin B12 (Cobalamin) deficiency in mother which is usually secondary to pernicious anemia, strict vegetarian diet and malnourishment. Neurological symptoms of vitamin B12 deficiency in infants include irritability, failure to thrive, hypotonia and developmental regression /delay. We report a 9-month – old infant with insidious developmental regression hypotonia, macrocytic anemia and generalized cerebral atrophy on CT scan brain which is markedly improved after vitamin B12 administration

Key Words : Vitamin B12 deficiency, Cerebral atrophy.

Case Report -

A -9 Month – old child born of non consanguineous marriage was admitted to our hospital with high grade fever cough and dyspnea for period of 4days. The patient was exclusively breast fed until the age 6 months after that complementary feeding was started. He developed normally during his first seven months; from seven months onward he gradually becomes less active and lost the ability to hold neck, roll over and at nine months of age he was unable to sit without support and developed dysphagia to animal based nutrients such as milk and egg.

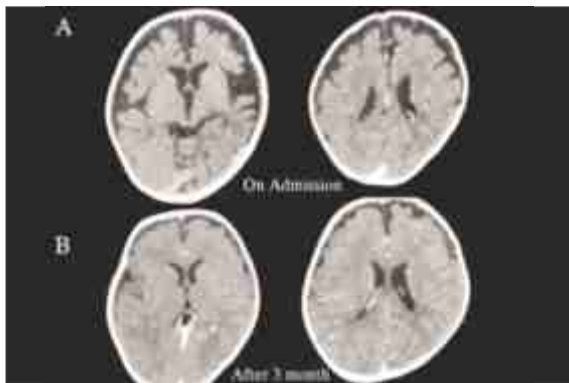
Family history revealed that his mother was vegetarian and had been treated for iron deficiency anemia during the pregnancy. On examination there was pallor, knuckle pigmentation, respiratory

distress and hepatomegaly. He did not establish eye contacts, unable to smile, lacked head control and was not able to sit without support.

Investigation - Hemoglobin 6 gm/dl, white blood cell count 5000 mm³, platelet count 2.5 lacks mm³, mean corpuscular volume 100fL, red cell distribution width (RDW) 20% peripheral smear showed anisocytosis, macrocytosis, poikilocytosis, hyper segmented neutrophils and adequate platelets. Biochemical profile, thyroid profile, serum ferritin, iron level were normal. Serum vitamin B12 level of baby was 140nf/L and of mother was 160ng/L. CT scan brain shows generalized atrophy of brain mainly involving bilateral fronto–parietal lobe and ventriculomegaly (fig. 1A).

Treatment- Initially vitamin B 12 was given intramuscular daily for seven days, and then alternate day for 7 days, followed by weekly for four to five weeks and then started orally. Within 1 to 2 weeks of treatment patient became active began to smile and swallow and show interest in surrounding, muscles tone improvement and achieved head control. After three months treatment baby's anemia improved and was able to waves, and walk with support. Repeat CT scan brain done shows marked improvement (Fig 1B)

Discussion - Infantile Vitamin B12 deficiency is rare but treatable cause of development delay and deterioration affecting exclusively breast fed infants born to Vitamin B12 deficient mother which is usually secondary to pernicious anaemia or a strict vegetarian diet^[1] Vitamin B12 is necessary for the production of methylenetetrahydrofolate which is essential for DNA synthesis in the rapidly growing hematopoietic cells and this can result in macrocytic anemia, hyper segmentation of neutrophils, leucopenia, thrombocytopenia and pancytopenia. Accompanying neurological abnormalities include paresthesia, sensory deficits, and loss of deep tendon. Reflexes movement disorder, developmental regression, dementia nad neuropsychiatric changes. Brain atrophy and delayed myelination can be observed in neuroImaging studies.^[2,3]



In infants vitamin B12 deficiency causes fatigue, restlessness, vomiting, difficulty of swallowing solid food and delay or regression of growth and development. Vitamin B 12 deficiency can also cause irreversible neurological damage and this effect is more prominent in infants who have been exposed to deficiency during the intrauterine period. Brain atrophy, microcephaly and myoclonic seizure are among the most commonly neurological sequelae in vitamin B12 deficiency^[4,5].

Developmental regression, megaloblastic anemia and cerebral atrophy on neuroimaging as seen in our patient is not surprising and have been reported before. Jadhav et al first described nutritional vitamin B12 deficiency in 6 Indian infants aged 6-12 months that presented with megaloblastic anemia, psychomotor regression and hyper pigmentation of the skin^[6]. The pathogenic mechanism by which vitamin B12 deficiency causes neurological symptoms is not clear, it is believed that congenital anomalies in homocysteine remethylation or an abnormality in the methionine.

Synthetase reactions lead to neurological symptoms. Although vitamin B12 supplements were reported to result in rapid improvement, concern has been raised regarding long term development. Mongale et al reported that two out of six vitamin B12 deficient patients developed long term neurological sequelae even after treatment^[2]. Von Schenck et al reported that early diagnosis done before 10 months age was associated with normal outcome, while those in which diagnosis was delayed made after one year of age had permanent neurological abnormality^[7].

Conclusion -

In case of patient presenting with hypotonia and development delay physician should obtain antenatal history and dietary history in detail instead of thinking extreme diagnosis and doing unnecessary laboratory work up. It is also important to emphasize that vitamin B12 supplementation during pregnancy and lactation should be provided for strict vegetarians and individuals with pernicious anemia to avoid irreversible neurological damage in exclusively breast fed babies.

References -

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