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Infantile Spasm-Rare Presentation of "Vitamin B12 Encephalopathy"

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ABSTRACT
Vitamin B12 deficiency manifests as triad of anaemia, gastrointestinal abnormalities and neurological abnormalities. The children with vitamin B12 deficiency are often misdiagnosed as it mimics autism spectrum disorders, colics and gastroenteritis. The existence of vitamin B12 deficiency neuropathy was recognised in 1958[1]. Seizures are rare but are seen especially in infants and there are only a few reports regarding the relationship between infantile spasm and vitamin B12 deficiency. Here we report a case of 19 month old female baby who presented to MKCG medical college, paediatric casualty with complains of hyperpigmentation of skin for 4 months, inability to stand with support for 3 months and sudden flexion of neck, arms and thighs multiple times throught the day for 2 months.
Keywords- *Vitamin B12 deficiency, Autism spectrum disease, Seizure, Infantile spasm.*

INTRODUCTION

In 1877, Gardner and osler coined the term pernicious anaemia to describe a patient with progressive arm numbness with difficulty in buttoning and using tools.[1] The prevalence of vitamin B12 is difficult to asertain because of its etiology and different assays[2]. It is an easily treated disorder that often goes undiagnosed in infant and children, placing them at high risk for permanent brain injury. Total body stores are 2-5mg of which half, stored in liver. The daily recommended dose is 0.7mcg/day in children and in adolescence 2mcg/day[3]. Vitamin B12 deficiency occurs in infants born to mother's with vitamin B12 deficiency due to any cause like vegetarianism, autoimmune pernicious anaemia, celiac disease, H.Pyloric infection, Crohn's disease, gastric bypass, partial ilectomy,eating disorders, use of PPI etc. Infants who are exclusively breast fed may have significantly less stored vitamin B12 and can develop a deficiency within first year of life. Even in infants who are formula fed have suboptimal B12 levels as the amount of B12 in formula is not enough to correct their deficiency. Compared to adults, infants with B12 deficiency have much more rapid onset of symptoms. Children presents with non specific manifestation such as developmental delay, irritability, weakness, failure to thrive, abmormal pigmentation, hypotonia and hepatosplenomegaly. Vitamin B12 has a role in DNA synthesis, delayed DNA synthesis in rapidly growing hematopoietic cells may result in macrocytic anaemia. The neurological manifestation of cobalamin deficiency is may be due to homocysteine toxicity deposits in brain and infants may be predisposed due to incompletely formed blood brain barrier[3]. Restoration of development skills after therapy is

variable with some children remaining moderately or severely retarded.

CASE REPORT

A 19 month old female baby admitted to paediatric department of MKCG medical college with complains of hyperpigmentation of skin for 4 months, unable to stand with support for 3 months and sudden flexion of neck, arms and thighs multiple times for 2 months. The child was born out of non-consanguinous marriage by normal vaginal delivery, the child has been continuing breast feed till now along with mixed diet from family pot. The family being vegetarian. The child was apparently normal till 15 months of age and had attained all milestones appropriate for age till 15 months of life, following which she gradually lost the ability to stand with support and sit by herself. There was no associated fever, headache and vomiting. On examination, the child was irritable. There was intermittent flexor spasm (infantile spasm) multiple times a day and there was hyperpigmentation of skin over toungue, knuckles, knee and thighs (Figure 1,2 and 3). There were no signs of meningitis, reflexes were brisk and B/L plantar was flexor.CSF study was done to rule out meningitis, which came out to be normal.CBC showed severe anaemia and MCV was 94 fL (Figure 4). Because of macrocytic anaemia, knuckle pigmentation and neurological signs with a history of vegetarian diet,a provisional diagnosis of vitamin B12 deficiency was made. It was confirmed by doing serum B12 level, which came to be very low (<100pg/ml) (Figure 5).EEG came out to be normal. The patient was treated with IM neurobion injections daily for 7 days followed by weekly dose for 7 weeks. To control infantile spasm IV valproate started and after 72 hours oral clonazepam was added as seizure persisted. The patients cognition improved by day 3.The infantile spasm came under control from 6th day onwards and from day 8 it seized completely. The patient was successfully discharged after regaining all the developmental milestones appropriate for age (figure 6).

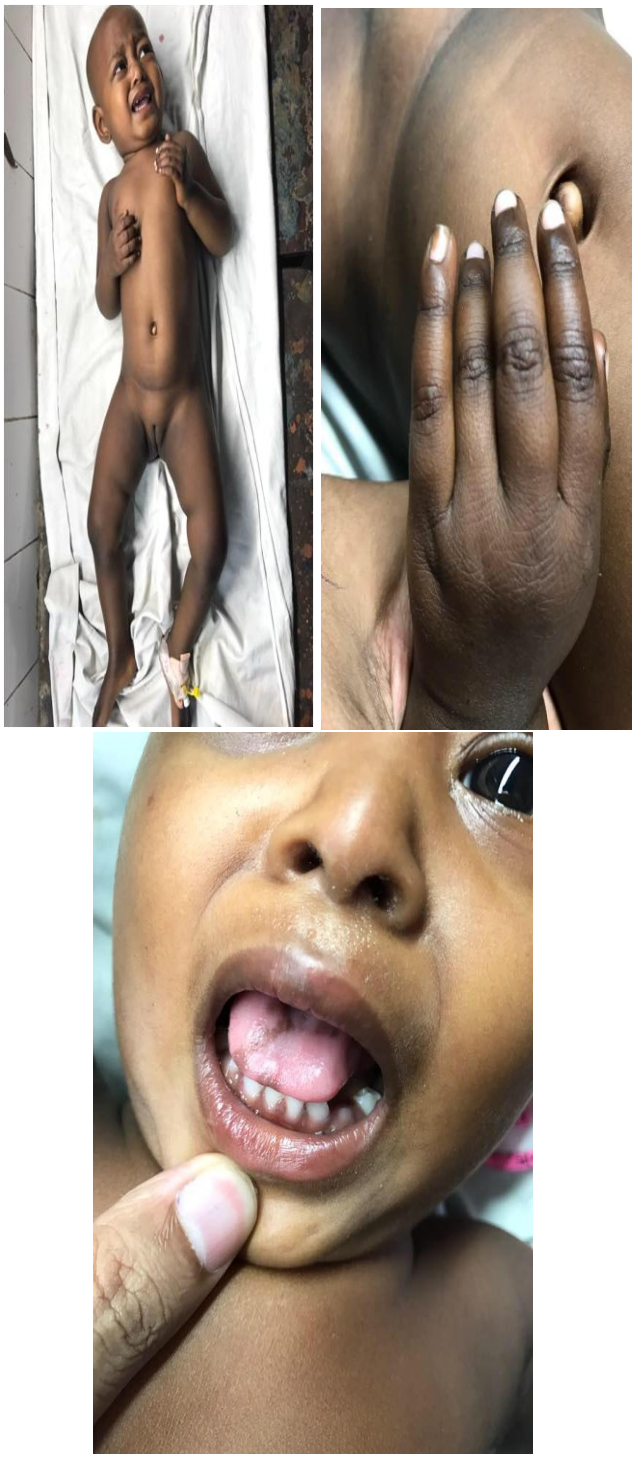

Figure 1 ,2 and 3 showing pigmentation of lower limbs, hand and tongue

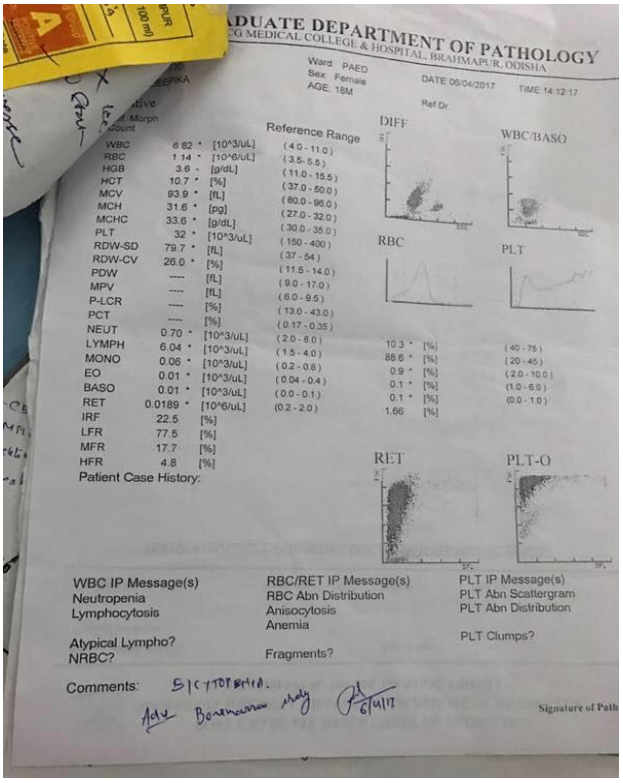

Figure 4 showing CBC (low Hb and high MCV)

Figure 6 showing happy child with ability to sit on its own

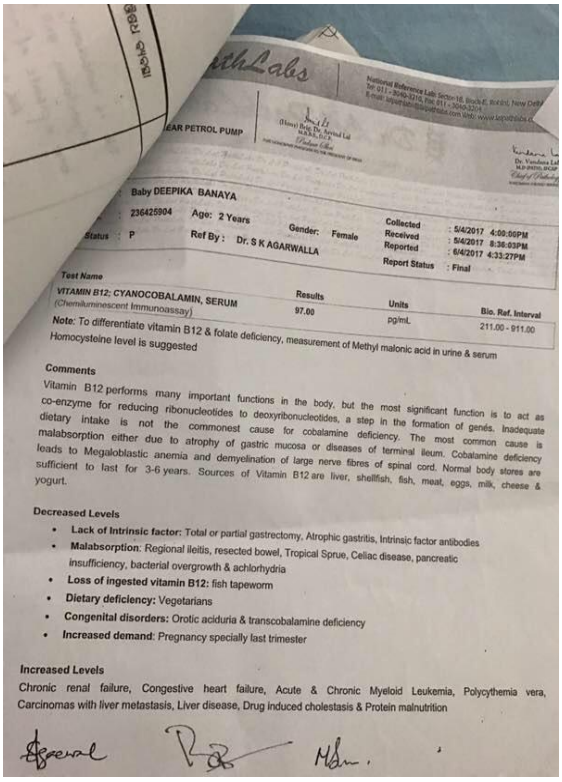

Figure 5 showing serum B12 level

DISCUSSION

Most of the initial data regarding vitamin B12 deficiency in infancy are from case studies of infants exclusively breast fed by mothers on vegetarian diet. This case reiterates the association between infantile spasm and vitamin B12 deficiency. Infantile spasm are a unique form of seizure disorder as their occurrenceis mosltly limited to infancy and they are refractory to conventional anticonvulsant drugs. In India, a hospital population radioassay study with a cut off of 200 pg/ml found a vitamin B12 deficiency in 0.88% of patients with boder line values in 3.8%[3].Infants born to vitamin B12 replete mothers have stores of vitamin B12 that are adequate to sustain them for first several months post partum hence vitamin B12 rarely occurs before 4 months of age[4].

CONCLUSION

Vitamin B12 deficiency may be a treatable cause of infantile spasms and should be considered in the associated cause of infantile spasm especially if there is nutritional inadequacy in strict vegan

children.90% patients have improvement in symptoms of >50% and rest 10% have residual moderate to severe disability following early treatment[5]. Hence early diagnosis and treatment is required.

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