



Severe Vit B12 Deficiency Presented As Encephalopathy in a 5 Yr Child

Authors

**Dr Subhranshu Sekhar Dhal¹, Dr Sunil Kumar Agarwalla², Dr Tapan Kumar Biswas³,
Dr Shantanu Kumar Meher⁴**

¹Junior Resident, Dept. Of Paediatrics, M.K.C.G Medical College

²Associate Professor, Dept. Of Paediatrics, M.K.C.G Medical College

Email: sunil_9910@yahoo.com, Mobile no-9861070101

³Junior Resident, Dept. Of Paediatrics, M.K.C.G Medical College

Email- biswas.tapan433@gmail.com, Mobile no.-8908050325

⁴Junior Resident, Dept. of Paediatrics, M.K.C.G Medical College

Email-drshantanukrmeher@gmail.com, Mobile no.-9853454594

Corresponding Author

Dr Subhranshu Sekhar Dhal

Address- P.G Hostel 2, M.K.C.G Medical College, Berhampur, Ganjam, Odisha, 760004, India

Email: subhransumkcg@gmail.com, Mobile No.-9861726493

Abstract

Vit B12 plays important role in different organ in our body, most importantly haematological system and central as well as peripheral nervous system. So its deficiency in children can cause poor weight gain, developmental regression, mental changes, abnormal movements, encephalopathy or may leads to long-term neurological sequelae. Early diagnosis and prompt treatment can alter the disease process. Here we present a case of severe vit B12 deficiency, who admitted with encephalopathy and successfully treated with a good neurological outcome almost towards normal.

Keywords- *vit B12 deficiency, encephalopathy, developmental regression.*

Introduction

Vit B12 is also known as cyanocobalamin. It is essential for functioning of brain, spinal cord, peripheral or cranial nerves, and blood cell production. Deficiency produces dementia, peripheral neuropathy, subacute combined degeneration of the spinal cord, nutritional amblyopia (visual loss) and cognitive dysfunction^[1]. Vitamin B₁₂ deficiency can lead to serious sequelae in developing children. Honzik et al^[2]. Describe failure to thrive, hypotonia, irritability,

lethargy, and developmental delay/regression, epilepsy and movement disorders associated with vitamin B₁₂ deficiency. They also report brain atrophy, delayed myelination, poly-neuropathy and abnormal evoked responses. One series of 27 patients with vitamin B₁₂ deficiency found failure to thrive in 63%, hypotonia in 66.7%, refusal to wean in 81.3% and absence of eye contact/social retardation in 51.9%^[3]. A study in the year 2000 indicates that B₁₂ deficiency is far more widespread than formerly believed. The study

found that 39 percent of studied group of 3,000 had low values ^[4]. This Tufts University study used the B₁₂ concentration 258 pmol/l (= 350 pg/mL) as a criterion of "low level". However, recent research has found that B₁₂ deficiency may occur at a much higher B₁₂ concentration (500–600 pg/mL) ^[5]. Against this background, there are reasons to believe that B₁₂ deficiency is present in a far greater proportion of the population than 39% as reported by Tufts University

Case Presentation

A 5 yrs. old male child, product of non-consanguineous marriage presented with unable to walk, stand, sit, speak with abnormal body movements and altered sensorium for last 15 days. (fig.1) There was no history of fever, convulsion, loose stool, and vomiting, respiratory difficulty. There was also no history of birth asphyxia and child is neuro developmentally normal. No similar episodes in past or no sibling in the family having similar problem. On examination child was in altered sensorium, vitals was stable, anthropometric measurement was normal for the age. On head to toe examination child having knuckle and periungual hyperpigmentation of both limbs (fig.2, 3), angular stomatitis with sparse hypo pigmented brittle hair, (fig.4, 5) some pallor, nocyanosis, clubbing, icterus, edema or lymphadenopathy. On CNS examination no cranial nerve deficit, hypotonia of both upper and lower limbs, power of both limbs was diminished (3/5). All superficial reflexes are normal except planter extensor, B/L Knee jerk was exaggerated and ankle jerk was diminished. Other systemic examination was within normal limits. On investigation complete blood count suggestive of megaloblastic anemia. Hb-6.7 gm%, MCV-110.8 fl. Serum electrolytes, urea, creatinine, liver function test was normal. Serum vitB12 was estimated and it was very low (73 pg/ml). CECT brain was also normal.



Fig 01



Fig 02



Fig 03

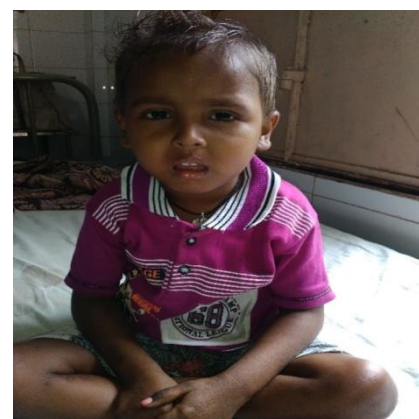


Fig 04



Fig 05



Fig 06

Discussion

Vitamin B₁₂ deficiency is though rare in the paediatric age group but in can happen due to a strict vegan diet, malabsorptive disorders, pernicious anemia, previous gastric surgery and terminal ileum resection [6]. Inborn errors of metabolism or transport defects are uncommon causes. There have been very rare cases reported as pernicious anemia with autoimmune disorders such as polyglandular syndrome [7]. The main syndrome of vitamin B₁₂ deficiency is characterized by a triad of symptoms:

- 1) Megaloblastic anaemia. This is due to the inhibition of DNA synthesis (specifically purines and thymidine)
- 2) Gastrointestinal symptoms: alteration in bowel motility, such as mild diarrhoea or constipation, and loss of bladder or bowel control [8]. There is an association with GAVE syndrome (commonly called watermelon stomach) and pernicious anemia [9].
- 3) Neurological symptoms: Sensory or motor deficiencies (absent reflexes, diminished

vibration or soft touch sensation), subacute combined degeneration of spinal cord, seizures, [10,11,12,13] or even symptoms of dementia [14] and or other psychiatric symptoms may be present. Methyl malonic acid, if not properly handled by B₁₂, remains in the myelin sheath, causing fragility. Dementia and depression have been associated with this deficiency as well, possibly from the under-production of methionine because of the inability to convert homocysteine into this product. Methionine is a necessary cofactor in the production of several neurotransmitters.

Each of those symptoms can occur either alone or along with others. The neurological complex, defined as myelosis funicularis, consists of the following symptoms:

- 1) Impaired perception of deep touch, pressure and vibration, loss of sense of touch, very annoying and persistent paresthesias
- 2) Ataxia of dorsal chord type
- 3) Decrease or loss of deep muscle-tendon reflexes
- 4) Pathological reflexes — Babinski, Rossolimo and others, also severe paresis

Vitamin B₁₂ deficiency can cause severe and irreversible damage, especially to the brain and nervous system. These symptoms of neuronal damage may not reverse after correction of hematological abnormalities, and the chance of complete reversal decreases with the length of time the neurological symptoms have been present.

Tinnitus may be associated with vitamin B₁₂ deficiency [15].

This child having very low serum vit b12 and presented to us with encephalopathy characterised by altered sensorium with abnormal deep tendon reflexes with typical knuckle hyperpigmentation. Treatment is usually supportive with supplementation of vit b12 with folic acid. It will take long time to recover but early treatment can lead to better outcome. After giving 2 wks. of

daily vit B12(1mg IM) and folic acid, neurological and general well-being improved(fig 6).Repeat vit B12 level was 512pg/ml. Child discharged with im vit B12 wkly for 8 wk. then monthly once for 6 month along with folate therapy .The child was advised for monthly check up.

Conclusion

Encephalopathy due to vit B12 deficiency is very rare in children but any child presented with encephalopathy with knuckle hyperpigmentation without any prior history fever, loose stool, vomiting one has to do CBC, PS and serum B12 level to rule out B12 deficiency. Management with vit B12 supplementation and folic acid is mainstay of therapy. Long term neurological sequele depends on time of diagnosis and initiation of treatment.

References

1. S. P. Stabler, R. H. Allen, D. G. Savage, and J. Lindenbaum, "Clinical spectrum and diagnosis of cobalamin deficiency," *Blood*, vol 76, no. 5, pp. 871–881, 1990.
2. Honzik T, Adamovicova M, Smolka V, Magner M, Hrubá E, Zeman J. Clinical presentation and metabolic consequences in 40 breastfed infants with nutritional vitamin B₁₂ deficiency – what have we learned? *Eur J Paediatr Neuro*. 2010; 14:488–95. [PubMed]
3. Zengin E, Sarper N, Caki Kiliç S. Clinical manifestations of infants with nutritional vitamin B₁₂ deficiency due to maternal dietary deficiency. *Acta Paediatr*. 2009; 98:98–102. [PubMed]
4. Jump up to:^{a b c} McBride, Judy (2 August 2000). "B12 Deficiency May Be More Widespread Than Thought". Agricultural Research Service. United States Dept of Agriculture. Retrieved 2 July 2012.
5. Jump up^ Thomas, Donald (2011-06-22). *New Jump Swing Healthy Aging and Athletic Nutrition Program*. Xlibris. p. 41. ISBN 978-1-4628-8427-8.
6. Kamei M, Ito Y, Ando N, Awaya T, Yamada T, Nakagawa M. Brain atrophy caused by vitamin B₁₂-deficient anemia in an infant. *J Pediatr Hematol Oncol*. 2011; 33:556–8. [PubMed]
7. van Loon M, Postels DG, Heikens GT, Molyneux E. Severe pernicious anaemia in an 8-year-old African girl. *Ann Trop Paediatr*. 2009; 29:231–4. [PubMed]
8. Jump up^ Schwartz, William (2012). *The 5-minute pediatric consult (6th ed.)*. Philadelphia: Wolters Kluwer Health/Lippincott Williams & Wilkins. p. 535. ISBN 9781451116564.
9. Jump up^ Reynolds, E.H. (2014). "The neurology of folic acid deficiency". *Handb Clin Neurol*. 120: 927–43. doi:10.1016/B978-0-7020-4087-0.00061-9. PMID 24365361.
10. Jump up^ Briani C, Dalla Torre C, Citton V, Manara R, Pompanin S, Binotto G, Adami F (2013)."Cobalamin Deficiency: Clinical Picture and Radiological Findings". *Nutrients*. 5 (11): 4521–4539. doi:10.3390/nu5114521. ISSN 2072-6643. PMC 3847746. PMID 24248213
11. Jump up^ DN Amarapurka MD, ND Patel MD (Sept 2004). "Gastric Antral Vascular Ectasia Syndrome". *Journal of The Association of Physicians of India*. 52: 757.
12. Jump up^ Matsumoto A, Shiga Y, Shimizu H, Kimura I, Hisanaga K (Apr 2009). "[Encephalomyelopathy due to vitamin B12 deficiency with seizures as a predominant symptom]". *Rinshō Shinkeigaku = Clinical Neurology*. 49 (4): 179–85. doi:10.5692/clinicalneuro.49.179. PMID 19462816.
13. Jump up^ Kumar S (Mar 2004). "Recurrent seizures: an unusual manifestation of vitamin B12 deficiency". *Neurology India*. *Neurologyindia.com*. 52 (1): 122–123. PMID 15069260.
14. Jump up^ Mustafa TAŞKESEN; Ahmet YARAMIŞ; Selahattin KATAR; Ayfer

GÖZÜ PİRİNÇÇİOĞLU; Murat SÖKER. "Neurological presentations of nutritional vitamin B12 deficiency in 42 breastfed infants in Southeast Turkey" (PDF). Turk J Med Sci. 41 (6): 1091–1096. Retrieved 2013-12-29.

15. Jump up^ Yavuz H (Sep 2008). "'Vitamin B12 deficiency and seizures'". *Developmental Medicine and Child Neurology* (Open access). 50 (9): 720. doi:10.1111/j.1469-8749.2008.03083.x.PMID 18754925