2017

www.jmscr.igmpublication.org Impact Factor 5.84 Index Copernicus Value: 83.27 ISSN (e)-2347-176x ISSN (p) 2455-0450 crossref DOI: _https://dx.doi.org/10.18535/jmscr/v5i5.56

Jo IGM Publication

Journal Of Medical Science And Clinical Research An Official Publication Of IGM Publication

Infantile Spasm-Rare Presentation of "Vitamin B12 Encephalopathy"

Authors

Ali Nasreen, Agarwalla Kumar Sunil, Muhamed Shabeer, Pandia Kumar Hemant MKCG Medical College

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, disease, H.Pyloric infection, celiac 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

JMSCR Vol||05||Issue||05||Page 21583-21586||May

2017

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 it seized completely. The patient was 8 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

JMSCR Vol||05||Issue||05||Page 21583-21586||May

2017



Figure 4 showing CBC (low Hb and high MCV)

181	Contraction of the local division of the loc			
12		2/A		
12	pthe	2. 1	-	
E		2 1	Vational Reference La	
and the second of the second s	15 million	1	these between the second	11 - 3010-3204
EARS	ETROL PUMP	Brie Dr. Arrian	Wighter B.	D sqisticity www.idea more
		Padam Clini		tendary la
		A Statement		Dr. Vandana Lal M.D. Datro, D.Car Chad of Port
Baby DEEPIK	BANAYA	And a state of the		trappent charge and
: 236425904	Ago: 2 Years	and the Part of		the second second
Status : P	Ref By: Dr en	iender: Female	Received	: 5/4/2017 4:00.00PM
	SA AGARY	VALLA	Reported	: 6/4/2017 4:33:27PM
Tost Namo			suport Status	: Final
VITAMIN B12; CYANOCOBALAM	IIN, SEDUM	Results	11-11	
Note: To differentiate)	97.00	Pg/mL	Bio. Ref. Interval
Homocusteles I	12 & folate detail			211.00 - 911.00
Comments Vitamin B12 performs many co-enzyme for reducing ribon dietary intake is not the	important functions in ucleotides to deoxyribol commonest cause for	easurement of Metr the body, but th nucleotides, a ste	nyl malonic acid in e most significa p in the format	unine & serum nt function is to act as on of genés, inadequate
Comments Comments Vitamin B12 performs many co-enzyme for reducing ribon distary intake is not the matabaseption either due to a leads to Megaloblastic anemia sufficient to last for 3-6 years suggett	d important functions in ucleotides to deoxribo commonest cause for itrophy of gastic muce and demyelination of a Sources of Vitamin	the body, but th nucleotides, a ste cobalamine det osa or diseases large nerve fibres B12 are liver, she	e most significa p in the format ficiency. The z of terminal lieur of spinal cord.	Urino & serum nt function is to act as on of genés, inadequate sost common cause is a. Cobatamino déficiency Normal body stores are k. eggs, mik, chese & 6
Comments Comments Vitamin B12 performs many co-enzyme for reducing ribon distary intake is not the malabsorption either due to a leads to Megaloblastic anemia sufficient to last for 3-6 years yogur.	important functions in ucleotides to deoxyribo commonest cause for throphy of gastric muce and demyelination of Sources of Vitamin i	the body, but the nucleotides, a ste cobalamine del sa or diseases large nerve fibres B12 are liver, she	nyi malonic acid in e most significa p in the format iciency. The r of terminal lieur of spinal cord. allfish, fish, mea	urine & serum nt function is to act as on of gends, Inadequate osst common cause is n. Cobatamine deficiency Normal body stores are Å. eggs, milk, cheese &
Comments Comments Vitamin B12 performs many oenzyme for reducing niton dietary intel 2 performs to another ideatary intel 2 performs autocinent to land for 3-6 years yogur. December 2 performance for the form - lack of bitming for the form of the - lack of bitming for the form of the - lack of bitming for the form	important functions in ucleotides to deoxyribo commonest cause for throphy of gastric muce and demyelination of . Sources of Vitamin I	the body, but the nucleotides, a ste cobalamine det as or diseases large nerve fibres B12 are liver, she	nyi malonic acid ir e most significa p in the format ficiency. The r of terminal ileur of spinal cord, alfish, fish, mea	urine & serum nt function is to act as on of genés, inudequate nost common cause is A Cobatamise deficiency Normat body stores are & eggs, milk, cheese &
Comments Comments Vitamin B12 performs many oterazyme for reducing ribon dietary intake is not the matabsorption either due to a kedda to Megaboblastic anemaia sufficient to last for 3-6 years yogur. Decreased Levels - Lack of Intrinsic factor: T - Matabsorption: Regional	important functions in ucleotides to deoxyrbo trophy of gastric muse and demyelination of . Sources of Vitamin i otal or partial gastrecton lelis, resected hower Tr	the body, but the nucleotides, a ste cobalamine del sa or diseases large nerve fibres B12 are liver, she ny, Atrophic gastritt	nyi matonic acid ir e most significa p in the format liciency. The c of terminal ileur of spinal cord. allfish, fish, mea	urine & serum nt function is to act as on of gends, inadequate not common cause is n. Cobatamine deficiency Normal body screes are k. eggs, milk, cheese & antiBockies
Comments Comments Varianti B2 performs many co-enzyme for reducing ribon giatary intake is not the malabsorption either due to a sufficient to last for 3-6 years vogur. Decreased Levels • Lack of Intrinsic factors T • Malabsorption Regional insufficiency, bacterial over	d mportant functions in ucleotides to decrytebo commonest cause for itrophy of gastic muc and demyelination of . Sources of Vitamin I otal or partial gastrector leitis, resected bowl, Tr growth & achtorhydria	the body, but in nucleotides, a ste cobalamine del sa or diseases B12 are liver, she ny, Atrophic gastrit opical Sprue, Cella	tyl matonic acid ir e most significa p in the format liciency. The c of terminal ileur of spinal cord, alifish, fish, mea ss, Intrinsic factor ac disease, pancr	unhe & serum If function is to act as on of gends, inadequate nost common cause is Codatamie difficiency Normal body stores are k. eggs, mik, cheese & antibodies eatic
Comments Comments Warmin B12 performs many co-enzyme for reducing ribon distay intake is not the malabsorption either due to a badds to Megaloblastic anema autorition either due to a badds to Megaloblastic anema sogur Decreased Levels • Lack of Intrinsic factor: T • Malabsorption: Regional • Malabsorption: Regional	d important functions in urdeotides to decrytelo commonest cuest trophy of gastic muc- and demyelination of . Sources of Vitamin I otal or partial gastrector letils, resected bower, Tr growth & achlorhydia growth & achlorhydia	the body, but th nucleotides, a ste cobalarnine de osa or diseases B12 are liver, she ny, Atrophic gastrit opical Sprue, Cella	nyl matonic acid ir e most significa p in the format liciency. The c of terminal ileur of spinal cord, illfish, fish, mea is, Intrinsic factor ac disease, pancr	urine & serum nt function is to act as on of gends, inadequate out common cause is Normal body stores are k. eggs, milk, cheese & antibodies astic
Analogianan kevel is suggest Analogian and a suggest Marini B2 partorns many observations for reducing rition observations for reducing rition observations for reducing rition observations for reducing rition observations for reducing analogiant and analogian and analogiant and analogiant and for reducing ritions and analogiant and analogiant and for reducing ritions and analogiant analogiant and for reducing ritions and analogiant anal	d mportant functions in nucleotides to decoxyteo commonest cause for and demyelination of sources of Vitamin I folal or partial gastrector lettis, resected boxel, Tr growth & achtorhydia B21: fish tapeworm rians	the body, but th nucleotides, a ste cobalamine del soa or diseases large nerve fibres B12 are liver, she hy, Atrophic gastrit opical Sprue, Cella	yf malionic acid ir e most significi p in the format licency. The z of terminal lear of spinal cord. Illifah, fah, mea s, Intrinsic factor s, clisesse, pancr	urine & serum int function is to act as on of gends, inadequate nost common cause is nost common cause is
Comments Comments Vitamin 182 performs many ocerazyme for reducing niton ocerazyme for reducing niton ocerazyme for reducing niton ocerazyme for reducing niton insufficient to last for 3-6 years yogur. Decreased Levels 1. Loss of ingested vitamin 1. Loss of ingested vitamin 1. Loss of ingested vitamin 1. Dietary dieficiency: Vegeti 2. Congenital disorders: Otto	d mportant functions in ucleotides to decoypte commonest cause for furphy of gastric mass and demyeliantion of Sources: of Vitamin otal or partial gastrector letils, resected bowel, Tr growth & achtorhydria B12: finit papevorm rins bis activitie & transcobal	Ihe body, but th nucleotides, a the coolamine del sia or diseases any nerve fibres 112 are liver, she ny. Altophic gastrit ny. Altophic gastrit amine deficiency	yf matonic acid ir 9 most significa 9 in the format Gioney. The z of terminal ileur of terminal iceu diffish, fish, mea 8, Intrinsic factor s, Intrinsic factor	ume & serum In function is to act as on of gends, inadequate note common dause is n. Cobatamon deficiency Normal body stores are & eggs, mill, cheese & antibodies. estic
Annexysterie fevel is suggest Annexysterie fevel is suggest Annexysterie fevel is suggest between the suggest and association either due for and association either due for association either due for associat	d mportant functions in incolocidies to decoyribio commonest cause for introphy of gastic muco and demyelination of . Sources of Vitamin I colal or partial gastrecton testis, resected bowel, T growth & achtorhydria B12: fielt tapeworm ritans fie andkuria & transcobal incy specially last trimesa	the body, but the nucleotides, a site coolarmine del sia or diseases a large nerve fibres 112 are liver, site ny, Atrophic gastrit, ny, Atrophic gastrit, ny, Atrophic gastrit, amine deficiency ter	y matonic acid in e most significa p in the format licitory. The r of spinal cord. efficiency. Signification s, Intrinsic factors ac disease, panor	urine & serum Inf. function is to act as on of gends, inadequate nost common cause is Codatamie deficiency Normal body stores are k. eggs, mik, cheese & antibodies eatic
Comental Alternation of the suggest Comment Maning Alt performs many co-enzyme for reducing ribon indiabsorption either due to autificiant to last for 3-6 years upper Decreased Levels Alternational disorders: Ore Distant deficiency: Vegets Distant deficiency: Distant def	d durant functions in turbefoldes to decrypte commonest cause for trophy of gastic mass and demyelination of . Sources of Vitamin I celal or partial gastrector lettis, resected bows, I could or partial gastrector lettis, resected bows, I growth & achtorhydria B12: fish tapeworm rians tic aciduria & transcobal necy specially last trimes	the body, but in muleicaldes, a tel coblamme de datases, a targe nerve faces B12 are liver, abr months and the second second second second pical Sprue, Cells amline deficiency ter	9 matonic acid in 9 most significa p in the format licitney. The a of terminal licent of spinal cord, allfah, fish, maxi- si, Intrinsic factor is, Intrinsic factor ac disease, panor	urine & serum Inf function is to act as on of genés, inadequate not common cause is A codatamine deficiency Normal body stores are & eggs, milk, cheese & antibodes estic
Analogical and the second of the second	d mportant functions in ucleotides to deoxytebo commonest cause for introphy of gastic mus- and demyeliation of Sources: of Vitamin i obial or partial gastrection tellis, resected bowel, Tr growth & achiorhydris B12: fint hapevorm rins tic aciduria & transcobal incry specially last trimes a heart failure, Acu er disease, Drug induce	Ine body, but in me body, but in cooldamine det as or diseases as or diseases are news firms 12 are inver, she hy, Atrophic gastrit opical Sprue, Celli amine deficiency ter le & Chronic di cholestasis & F	nyf matoric acid ir e most significa p in the format licency. The r of terminal licency of spinal cord. illich, fish, mea s, Intrinsic factor ac disease, panor Myeloid Leuke Protein malnutrili	ume & serum In function is to act as on of gends, inadequate not common datas is n. Cobatamon datas n. Cobatamon datas n. Cobatamon datas n. Cobatamon datas n. Cobatamon datas n. Cobatamon datas n. Cobatamon datas setto: mia, Pobycythemia vera, m

Figure 5 showing serum B12 level



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

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

JMSCR Vol||05||Issue||05||Page 21583-21586||May

2017

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.

REFERENCE

- Gardner, William. & Osler, William. (1877). Case of progressive pernicious anaemia (idiopathic of Addison)
- Singh NN, Thomas FP, Diamond AL, Diamond R. Vitamin B-12 associated neurological diseases. E Medicine Neurology. 2010:1-22.
- Rasmussen SA, Fernhoff PM, Scanlon KS. Vitamin B12 deficiency in children and adolescents. The Journal of pediatrics. 2001 Jan 1;138(1):10-7.
- Black MM. Effects of vitamin B12 and folate deficiency on brain development in children. Food and nutrition bulletin. 2008 Jun;29(2_suppl1):S126-31.
- Wolansky LJ, Goldstein G, Gozo A, Lee HJ, Sills I, Chatkupt S. Subacute combined degeneration of the spinal cord: MRI detection of preferential involvement of the posterior columns in a child. Pediatric radiology. 1995 Mar 1;25(2): 140-1.