Case Report

Lichtheim disease: A rare manifestation of pernicious anemia

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ABSTRACT

Subacute combined degeneration of the spinal cord (SACD) is an important neurological complication of vitamin B12 deficiency. It is an important treatable cause of progressive neurological disability. We report a case of a 12-year-old girl, which presented with features of SACD of the spinal cord with low vitamin B12 levels and positive anti-parietal cell antibodies, suggesting vitamin B12 deficiency due to autoimmune etiology. The girl was started on a regimen of intramuscular vitamin B12 treatment. After which she had complete neurological recovery. She was advised of lifelong supplementation of parenteral vitamin B12 due to the autoimmune etiology.

Key words: Methylcobalamin, Pernicious anemia, Subacute combined degeneration, Vitamin B12

Vitamin B12 deficiency is prevalent in the Indian population with a prevalence of 34.4% among adolescents [1]. Subacute combined degeneration (SACD) of the spinal cord, also known as Lichtheim disease, is a rare yet important complication of vitamin B12 deficiency. It refers to the degeneration of the posterior and lateral columns of the spinal cord. The incidence is more common in the elderly population with a peak incidence in the 6th and 7th decades, but cases have been reported in children and adolescents [2]. Pernicious anemia is an important cause of vitamin B12 deficiency. The prevalence of pernicious anemia is 0.1% in the general population and contributes to 25–50% of cobalamin deficiency cases [3].

CASE REPORT

An 8-year-old girl, born of third-degree consanguinity, developmentally appropriate for age presented with complaints of difficulty in walking and multiple falls for the past 20 days. This was associated with difficulty in wearing slippers and holding objects tightly with hands, and tingling numbness of both hands and feet. The girl was vegetarian by diet. Perinatal history was insignificant and family history was not contributory. On general examination, pallor and hyperpigmentation of knuckles (Fig. 1) were noted along with multiple scars over both knees due to repeated falls. On CNS examination, the higher mental functions and cranial nerves were intact. Power was normal (5/5 of Medical Research Council muscle power grading [MRC]) in a proximal group of muscles in both upper and lower limbs, whereas it was affected (3/5) in a distal group of muscles in both upper and lower limbs. The sensation of touch, pain, and the temperature was preserved in all the dermatomes. Vibration sense was affected in bilateral lower limbs in dermatomes L4, L5, and S1. However, stereognosis was intact. Among superficial reflexes, corneal, conjunctival, and abdominal reflexes were intact bilaterally, whereas anal and plantar reflexes were not elicitable on both sides. Among the deep tendon reflexes, biceps and triceps reflexes were normal (2+ of MRC grading of reflexes), whereas knee and ankle jerks were absent bilaterally. A clinical diagnosis of SACD was made. Complete hemogram revealed a hemoglobin of 9.9 g/dl (11–14 g/dl), mean corpuscular volume of 108 fl (76-96 fl), WBC counts – 7400 cells/cu mm (4000–11,000 cu mm), and platelet count – 2 lakhs/cu mm (1.5–4 lakh/cu mm). Blood vitamin B12 levels were 152 pg/ml (200–1000 pg/ml). Serum homocysteine levels were 146 µmol/l (13–18 µmol/l). Serum anti-parietal cell antibodies were positive, which was 50 units (Normal: <20 Units) pointing toward pernicious etiology (Table 1)

Nerve conduction velocity (NCV) was suggestive of axonal sensory polyneuropathy. MRI of the spinal cord was planned but was not feasible due to financial constraints. The patient was started on intramuscular Inj. Methylcobalamin 1000 µg/day and was continued for 2 weeks. In view of autoimmune pernicious etiology, the girl was advised monthly intramuscular injections of Inj. Methylcobalamin lifelong. On follow-up after 3 months, no gait problems and posterior column signs were seen and the knuckle hyperpigmentation had subsided. Repeat hemogram on follow-up showed an increase in hemoglobin to 11.2 g/dl and vitamin B12 levels to 320 pg/ml.

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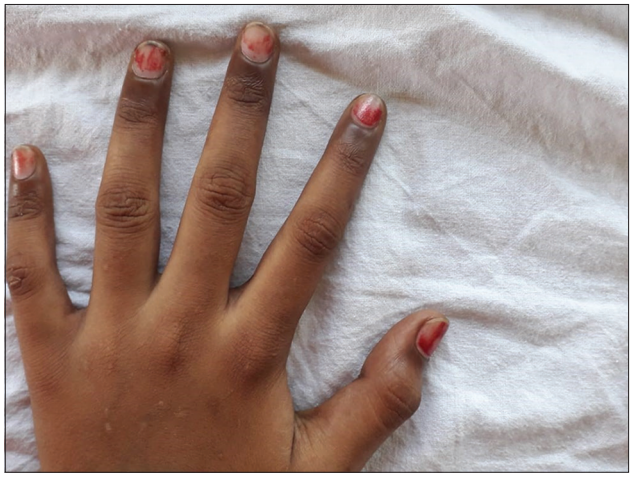

Figure 1: Hyperpigmentation of knuckles of hand

Table 1: Laboratory parameters before and after treatment

S. No.	Parameters	Pre-treatment	Post-treatment (after 3 months)
1.	Hemoglobin	9.9 g/dl	13.1 g/dl
2.	MCV	108 fl	90 fl
3.	WBC	7800 cu/mm^3	5500 cu/mm^3
4.	Vitamin B12 levels	152 pg/ml	400pg/ml
5.	Serum homocysteine levels	146 µmol/l	42 µmol/l
6.	Serum anti-parietal cell antibodies	50 Units	

DISCUSSION

Vitamin B12 deficiency can occur due to various causes. Animal food is the major source of vitamin B12; hence, deficiency can occur in a strict vegetarian or vegan. Our patient was a vegetarian. Another important cause for development of pernicious anemia is where antibodies to intrinsic factors or parietal cells interfere with the absorption of vitamin B12 (like in our case).

The neurological abnormality in SACD is due to decreased methionine synthase activity [4]. There is degeneration of posterior and lateral columns of the spinal cord along with peripheral nerve lesions. Symptom onset is often with a vague sensation of weakness and paresthesia. The "pins and needles" usually involve hands more than the feet and tends to be progressive. Ataxic paraplegia may occur as the illness progresses [5]. Additional affective/behavioral and cognitive symptoms may occur and in some patients, these symptoms can precede any cord symptoms [6]. Diagnosis is established by a hemogram showing macrocytic anemia which, in some cases, is accompanied by neutropenia and thrombocytopenia, low blood vitamin B12 levels, and elevated homocysteine levels. MRI spinal cord shows T2 hyperintense signal alterations usually confined to posterior columns, which may involve the lateral columns and rarely brainstem [7]. NCV can be normal or may suggest axonal sensory neuropathy. There are many treatment regimens for vitamin B12 deficiency. For SACD, Inj. vitamin B12 1 mg should be given for initial 2 weeks [8] and later put on maintenance therapy. The early replenishment of vitamin B12 body stores is associated with a better prognosis and reduces the likelihood of long-term residual disability [6]. In cases with etiology being pernicious anemia (as in our case), parenteral vitamin B12 should be administered regularly even after the resolution of neurological features [2]. The association with other autoimmune diseases such as type 1 diabetes, autoimmune thyroiditis, and Vitiligo is common. The association between pernicious anemia and autoimmune thyroiditis is most frequent [9]. Unexplained poor linear growth is a classical initial finding [10]. Parents were counseled regarding the importance of follow-up.

CONCLUSION

Although dietary factors of patients contributing to vitamin B12 deficiency are very common, a thorough workup should be done to rule out pernicious anemia, because oral vitamin B12 is not helpful in the treatment of such cases. The treatment with parenteral vitamin B12 in these patients improves the symptoms and prevents further complications of SACD.

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