Optic Atrophy Due to Vitamin B₁₂ Deficiency Without Anemia

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Vitamin B₁₂ deficiency can cause neurologic findings without anemia. Optic neuropathy due to vitamin B₁₂ deficiency occurs occasionally in adult patients but it is rarely seen in children. A 16 year-old boy was followed up in the outpatient clinic with blurred vision. On physical examination, he had optic atrophy bilaterally without other neurologic findings. He had low levels of serum vitamin B₁₂ and had no anemia. After cyanocobalamin treatment, visual acuity was improved but ophthalmologic findings were little changed despite serum vitamin B₁₂ levels returned to be a normal. In conclusion, we want to draw attention that unexplained visual loss in childhood, especially in adolescence period, might be due to vitamin B₁₂ deficiency and improvement of the visual symptoms is very well with vitamin B₁₂ treatment. Optic atrophy without anemia may be the first sign of vitamin B₁₂ deficiency.

Key words: Optic neuropathy, vitamin B₁₂, child

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INTRODUCTION

The main systems affected due to vitamin B12 deficiency are the hematological and the nervous system (Aaron et al., 2005). Neurologic symptoms of vitamin B12 deficiency are variable in all age groups (Healton et al., 1991). In adults, myeloneuropathy, cognitive dysfunction, peripheral neuropathy, neurophysiologic manifestations and dementia are the main clinical findings (Aaron et al., 2005). In children, it is often presents with nonspecific manifestations, such as developmental delay, irritability, weakness, failure to thrive and involuntary movement especially in infancy (Rasmussen et al., 2001; Kalyaci et al., 1996). It is well recognized that patients with vitamin B12 deficiency can develop neurologic findings without anemia in adults but relatively few data exists in children (Aaron et al., 2005; Healton et al., 1991; Lindenbaum et al., 1988). Optic neuropathy due to vitamin B12 deficiency occurs occasionally in adult patients but it is rarely seen in children and can be evaluated postmortem examination (Moschos and Droutsas, 1998; Areekul et al., 1992; Moschos et al., 1999; Lessel, 1998). Herein, we presented 16 year-old-boy with optic atrophy and vitamin B12 deficiency without anemia.

CASE REPORT

Sixteen-year-old boy admitted to our clinic with blurred vision for one year. He also complained episodes of diarrhea and abdominal pain monthly past two years and he had no history of the tobacco and alcohol exposure and family history and that he occasionally preferred to eat meat and meat products. But he was not strict vegetarian.

Physical examination revealed that his weight and height were both under third percentile and systemic findings were normal. On neurologic examination cranial nerves were normal except bilateral complete optic atrophy. His muscle tone was normal. His deep tendon reflexes were normoactive and his Babinski’s response was negative bilaterally. There were no motor abnormalities, no cerebellar signs and sensory deficit. Ophthalmologic examination revealed his best-corrected visual acuity was 1/10 in right eye and 2/10 in left eye. Direct and indirect light reactions in both pupils were diminished and no relative afferent pupillary deficit. Slit-lamp biomicroscopy and intraocular pressure was normal. There was no ptosis and proptosis. Examination of the extraocular motility revealed normal in all directions of gaze. Visual testing by Humphery Visual Field Analyzer showed visual field of patient who has very advanced generalized field loss in both eyes. Disc appearances were white, flat disc with clearly delineated margin, reduction in number of small blood vessels on the disc, attenuation of peripapillary nerve fiber layer on dilated fundus examination.

Complete blood count were as follows: Hemoglobin 14 g dL⁻¹, white blood cell count 8000 mm³ as 65% polymorphonuclear leukocyte and 35% lymphocyte, platelet count 283,000 mm³, hematocrit 43,1%, MCV 80.7 fl, MCH 28.3 pg. Peripheral blood smear showed that normochrom normocytic erythrocytes. Bone marrow aspiration showed nucleocytoplasmic dissociation and flow cytometry evaluation was normal. Serum iron level was 92 mg dL⁻¹, iron binding capacity was 316 mg dL⁻¹, vitamin B12 level was 87 µg mL⁻¹ (140-700), folic acid level was 7.77 ng mL⁻¹ (6-18,6). Gastric pH was 5 and antitibodies were negative. The visual evoked responses were significantly affected bilaterally. Thyroid functions, blood and urine amino acids levels, somatosensory evoked potential, electromyography and cranial magnetic resonance imaging were normal.

He was given intramuscular injections of 100 µg of cyanocobalamin daily during first week and tapered in two weeks and was continued monthly. Three months later, serum vitamin B12 level and visual performance returned to be normal and his visual acuity was 5/10 in right eye and 6/10 in left. But fundoscopic findings were little changed in follow up examination. His visual acuity was gradually increased after one year follow-up visit.

DISCUSSION

Although hematological finding such as macrocytic anemia was the main finding of vitamin B12 deficiency, neurologic disorders might be seen in the absence of the anemia (Rasmussen et al., 2001). Lindenbaum et al. (1988) reported 28% of cases with B12 deficiency had neurological findings such as sensory loss, ataxia, psychiatric disorders but had no macrocytic anemia. Parasympathetic, peripheral neuropathy and ataxia, even without anemias, may be the first sign of the cobalamin deficiency and also decreased vibration and position sense can be seen (Aaron et al., 2005; Healton et al., 1991; Rasmussen et al., 2001).

Optic neuropathy due to vitamin B12 deficiency is extremely rare in children (Moschos and Droutsas, 1998; Areekul et al., 1992; Moschos et al., 1999; Lessel, 1998). Lesions in optic nerve have been demonstrated in
postmortem examination of adult patient with pernicious anemia. Abnormal visual evoked responses was reported with pernicious anemia who have no visual symptoms, suggesting that there may also be subclinical damage to the visual pathway. Optic nerve pathology in vitamin B₁₂ deficiency is similar to the tobacco-alcohol amblyopia such as symmetrical, painless and progressive visual loss. Central and centrocecal scotomas are the main ophthalmologic findings and the optic disc appears normal in the early stages of the condition (Lessel, 1998).

Our patients had vitamin B₁₂ deficiency but had no anemia. In our patient, the only neurologic sign was blurred vision due to optic atrophy. Optic neuropathy due to alcohol or tobacco exposure may have similar symptoms but our patient had no history of these substances abuse. His blood and urine aminoacid levels and cranial magnetic resonance imaging were normal. Leber's Hereditary Optic Neuropathy (LHON) may be seen during adolescence period, but our patient ophthalmologic findings were not compatible with LHON (Lessel, 1998). Clinical and radiological findings were not compatible with demyelinating disease or tumour and we suggested that optic atrophy due to low serum vitamin B₁₂ level.

Recovery of the symptoms and signs of vitamin B₁₂ deficiency is contradictory (Healton et al., 1991; Moretti et al., 2004). In nonanemic patients in whom diagnosis was delayed, neurologic progression frequently occurred although the hematocrit remains normal (Healton et al., 1991). In our patient the diagnosis of vitamin B₁₂ deficiency was delayed because of lack of anemia symptoms. After treatment of cobalamin for three months, serum vitamin B₁₂ levels returned to be normal. His visual acuity was improved but ophthalmologic findings were little changed. Clinical response can be seen during the first three months of treatment (Healton et al., 1991). During childhood, treatment may resolve the complications such as hematological, failure to thrive, but permanent neurologic damage may have already occurred. It has been several studies reported that recovery of the visual acuity with vitamin B₁₂ therapy while ophthalmologic findings persisting (Moschos and Droitsas, 1998; Arekuk et al., 1992; Moschos et al., 1999). The percent improvement over baseline neurologic status after treatment was inversely related to duration of symptoms and hematocrit (Healton et al., 1991).

In conclusion, we want to draw attention that unexplained visual loss in childhood, especially in adolescence period, might be due to vitamin B₁₂ deficiency and improvement of the visual symptoms is very well with vitamin B₁₂ treatment.

REFERENCES


