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Case Report

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Optic Atrophy Due to Vitamin B_{12} Deficiency Without Anemia

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Vitamin B_{12} deficiency can cause neurologic findings without anemia. Optic neuropathy due to vitamin B_{12} 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 B_{12} and had no anemia. After cyanocobalamine treatment, visual acuity was improved but ophthalmologic findings were little changed despite serum vitamin B_{12} 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 B_{12} deficiency and improvement of the visual symptoms is very well with vitamin B_{12} treatment. Optic atrophy without anemia may be the first sign of vitamin B_{12} deficiency.

Key words: Optic neuropathy, vitamin B_{12}, child

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INTRODUCTION

The main systems affected due to vitamin B_{12} deficiency are the hematological and the nervous system (Aaron *et al*., 2005). Neurologic symptoms of vitamin B_{12} deficiency are variable in all age groups (Healton *et al*., 1991). In adults, myeloneuropathy, cognitive dysfunction, peripheral neuropathy, neurophysciatric 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; Kalayci *et al*., 1996). It is well recognized that patients with vitamin B_{12} 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 B_{12} 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 B_{12} 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 mm^3 as 65% polymorphonuclear leukocyte and 35% lymphocyte, platelet count 283.000 mm^3, 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 B_{12} level was 87 pg mL^{-1} (140-700), folic acid level was 7.77 ng mL^{-1} (6-18.6). Gastric pH was 5 and antigliadin antibodies were negative. The visual evoked responses were significantly affected bilaterally. Thyroid functions, blood and urine aminoacids levels, somatosensorial evoked potential, electromyography and cranial magnetic resonance imaging were normal.

He was given intramuscular injections of 100 μg of cyanocobalamine daily during first week and tapered in two weeks and was continued monthly. Three months later, serum vitamin B_{12} 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 B_{12} 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 B_{12} deficiency had neurological findings such as sensory loss, ataxia, psychiatric disorders but had no macrocytic anemia. Paraesthesia, peripheral neuropathy and ataxia, even without anemias, may be the first sign of the cobalamine 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 B_{12} 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 B_{12} 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 B_{12} 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 B_{12} level.

Recovery of the symptoms and signs of vitamin B_{12} 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 B_{12} deficiency was delayed because of lack of anemia symptoms. After treatment of cobalamine for three months, serum vitamin B_{12} levels returned to be normal. His visual acuity was improved but opthalmologic findings were little changed. Clinical response can be seen during the firs 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 B_{12} therapy while ophthalmologic findings persisting (Moschos and Droutsas, 1998; Areekul *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 B_{12} deficiency and improvement of the visual symptoms is very well with vitamin B_{12} treatment.

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