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Vaka Sunumu

The Infant with Vitamin B12 Deficiency

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The Infant with Vitamin B12 Deficiency

A 2-month-old male infant was admitted to the emergency clinic with complaints of loss of consciousness. The conscious and hypotonic baby had no good eye contact. In the laboratory tests of the infants who had no symptoms other than hypotonia on physical examination, WBC: 7130/mm^3, RBC: 3.17 10e6/µL, hgb: 10.6 g/dL, hct: 29.9, MCV: 94.3, MCH: 33.4, PLT: 480.103/ml, blood sugar: 108 mg/dl, Ca: 10,2 mg/dl, Mg: 2,1 mg/dl Na: 136 mEq/l, K: 3,7 mEq/l, AST: 28 IU, ALT: 32 IU, ammonia: 55.8 µg/dL were measured and tandem mass was normal. On the first day of the hospitalization, tonic contraction was observed for a few seconds. The patient's cranial USG and EEG were normal. While VSD and PFO were detected in the echocardiography of the patient, any abnormality was found in the holter ECG. Vitamin B12 level was 219.9 pg/dl, and homocysteine was 9.2 µmol/ml. Her mother's B12 level was measured at 186 pg/dl and the patient started vitamin B12 IM at 100 mcg/kg/day. The baby, whose attacks had disappeared since the treatment was started, was discharged on the 7th day of hospitalization. It was found that the baby did not have a seizure in the follow-up performed one week after discharge and the hypotonicity decreased in the control after 1 month.

Keywords: *Anemia, vitamin B12, convulsion*

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B12 Vitamini Eksikliği Olan Bir Sütçoocuğu

İki aylık erkek bebek şuur kaybı nedeniyle acil polikliniğimize getirildi. Fizik muayenesinde şuuru açık ve soluk olan bebeğin göz kontağının az olduğu saptandı. Sistem bulgularında hipotoni dışında özellik olmayan bebeğin laboratuvar tetkiklerinde WBC: 7130/mm^3, RBC: 3.17 10e6/µL, hgb: 10.6 g/dL, hct: 29.9, MCV: 94.3, MCH: 33.4, PLT: 480.103/ml, kan şekeri: 108 mg/dl, Ca: 10,2 mg/dl, Mg: 2,1 mg/dl Na: 136 mEq/l, K: 3,7 mEq/l, amonyak: 55.8 µgr/dl ve tandem mass testi normal saptandı. Yatışının ilk gününde tüm vücüdunda kasılma olan hastanın çekilen kranial USG ve EEG normal bulundu. Kardiyolojik açıdan değerlendirilen hastanın ekokardiografisinde VSD ve PFO saptanırken, holter EKG özellik saptanmadı. B12 düzeyi 219.9 pg/dl, homosistein: 9.2 µmol/ml (N: 3.3-8.3) saptanan hastann annesinin B12 düzeyi de 186 pg/dl bulunması üzerine 100 mcg/kg/gün B12 İM başlandı. Tedavi başlandıktan sora atakları kaybolan bebek, tedavisi düzenlerek yatışının 7. gününde taburcu edildi. Bir hafta sonra yapılan kontrolünde nöbet geçirmediği, 1 ay sonraki kontrolünde ise hipotonisitesinin azaldığı saptandı.

Anahtar kelimeler: *Anemi, B12 vitamini, konvülziyon*

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INTRODUCTION

Vitamin B12 is not synthesized in the human body; it is mostly found in foods of animal origin. Along with folic acid, vitamin B12 participates in DNA synthesis and plays a role in cell division and proliferation. The most common cause of vitamin B12 insufficiency is insufficient intake of the vitamin in the diet. However, lack of intrinsic factor, Imerslund-Grasbeck syndrome, ileal and gastric surgical interventions and infections with Diphyllobothrium latum may also lead to vitamin B12 deficiency [(1)]. In babies exclusively fed with breast milk, the most common cause of vitamin B12 deficiency is insufficient intake from vegetarian mothers or mothers with pernicious anaemia. Deficiency of vitamin B12 due to genetic defects is rare [(2)]. As the resources of babies born with low vitamin B12 stores are fully consumed in the first 3-18 months and when vitamin B12 intake via breast milk is insufficient, a gradual onset of clinical findings is usually observed. The severity of clinical symptoms is directly related to the severity of vitamin B12 deficiency in the mother [(3,4)]. As the clinical findings of vitamin B12 deficiency are nonspecific, they may easily be overlooked by family physicians or paediatricians, which in turn leads to a delay in the diagnosis and more severe clinical manifestations. The most common clinical signs were irritability, apathy, hypotonia, motor retardation, decrease in deep tendon reflexes, encephalopathy, coma and microcephaly [(1,2)].

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CASE

A 2-month-old male infant was admitted to the emergency clinic with complaints of loss of consciousness. Pregnancy and birth history of infant were unremarkable. She was the only child of non-consanguineous parents. Physical examination revealed no dysmorphic features, hepatosplenomegaly or neurocutaneous findings. Head circumference was 38 cm (75-90 p). The conscious and hypotonic baby had no good eye contact and smiled very rarely. In the laboratory tests of the infants whose characteristics were not found in the system findings, WBC: 7130, RBC: 3.17 10 6, HGB: 10.6 gr, HCT: 29.9, MCV: 94.3, MCH: 33.4, PLT: 480.103 / ml, blood sugar: 108 mg/dl, Ca: 10,2 mg /dl, Mg: 2,1 mg /dl Na: 136 mEq / l, K: 3,7 mEq /l, AST:28 IU, ALT:32 IU, ammonia:55.8 µg/dL and Tandem mass was normal. On the first day of the hospitalization, tonic contraction was observed for a few seconds. The patient's cranial USG and EEG were normal. While VSD and PFO were detected in the echocardiography of the patient, any abnormality was found in the holter ECG. Vitamin B12 level was 219.9 pg /dl, and homocysteine was 9.2 µmol / ml (N: 3.3-8.3). Her mother's B12 level was measured at 186 pg/dl and the patient started vitamin B12 IM at 100 mcg/kg/day. The baby, whose attacks had disappeared since the treatment was started, was discharged on the 7th day of hospitalization. It was found that the baby did not have a seizure in the follow-up performed one week after discharge and the hypotonicity decreased in the control after 1 month.

DISCUSSION

Vitamin B12 deficiency is preventable, potentially reversible disorder, but may result in serious neurological problems if not treated properly. Vitamin B12 deficiency can present with various clinical manifestations, especially during infancy. Although it can present through slowly progressive and insidious neurologic impairment such as motor delay, developmental regression, and apathy, it might also present with acute neurologic findings such as seizures or involuntary movements [6].

B12 deficiency and seizures have been described in the literature. İncecik et al. reported 7 infants with vitamin B12 deficiency, 4 of whom had generalized tonic-clonic seizures, 1 had generalized tonic and 2 had focal seizures [7]. Taşkesen et al. reported 12 children with seizures of 42 with nutritional vitamin B12 deficiency. B12 deficiency may also cause West syndrome [8]. In some of the cases after the treatment the convulsions are dissapear. Resistant seizures continue in some cases after treatment and learning difficulties may occur in these patients [9,10]. We have found that atipic movements have disappeared since B12 therapy started.

The impact of vitamin B12 deficiency on such different neurologic findings has not yet been fully elucidated. Delayed myelination or demyelination, impaired ratio of S-adenosylmethionine to S-adenosylhomocysteine, an imbalance between tumor necrosis factor-a and epidermal growth factor and lactate accumulation are the mechanisms that could plausibly play a role [11].

The most common neuroradiological findings are cortical atrophy, hypoplasia of the corpus callosum, retardation in myelinisation and moderate enlargement in the ventricle. While the haematological findings rapidly improve following treatment, the neurological findings may persist. Whether neurological damage will lead to a sequel or not depends on the severity and duration of vitamin B12 deficiency [1]. Cranial USG was normal in our patient.

B12 deficiency should be started and continue on maintenance-level doses of cyanocobalamin. Options include parenteral (intramuscular [IM] or subcutaneous [SC]), oral, sublingual, or intranasal routes. Parenteral therapy using the IM or SC route is by far the most reliable and most familiar treatment for vitamin B12 deficiency [12].

While the haematological findings rapidly improve following treatment, the neurological findings may persist. Whether neurological damage will lead to a sequel or not depends on the severity and duration of vitamin B12 deficiency. Von Schenck et alstated that the mental states of patients diagnosed before the age of 10 months were found to be normal compared with cases with delayed diagnosis. In another trial, the long-term follow-up (5-10 years) of two cases with convulsions at the time of diagnosis revealed neurological damage [13].

Treatment with high doses in severe vitamin b12 deficiencies may lead to hypokalemia, congestive heart failure and tremors in the hands [14].

It is of great importance to prevent, diagnose, and treat vitamin B12 deficiency promptly to prevent the long-term neurologic problems. The World Health Organization concluded that vitamin B12 has a clear impact on child development and directly affects the cognitive scores of school-aged children [15]. Because vitamin B12 deficiency may present with various neurologic symptoms prior to systemic symptoms, every child presenting with neurologic problems should have their levels checked, and if deficient, treated immediately. The prevention of B12 deficiency is a more effective approach than treatment. In countries where vitamin B12 deficiency is common, levels should be checked in all pregnant women and infants within the framework of a health policy, and additional focus should also be given to infants presenting with neurologic symptoms.

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