

Distrophinopathy: A rare cause of elevated transaminase in newborn

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ABSTRACT

Elevated transaminase levels are encountered in neonates and infancy because of several reasons. Muscular dystrophy is a rare hereditary disease compared to other disease causing elevation of transaminase. Some of them progress rapidly and result in death. Our case, who was born from a healthy non-relative marriage, delivered with NSD as term and weighed 3750g. Patient was admitted to the service with diagnosis of meconium aspiration syndrome and perinatal asphyxia due to being stained with meconium and having respiratory distress. The patient was examined because of elevated transaminase levels. There was no reason which could lead to elevation of transaminases derived from liver. We examined the patient in terms of myopathy because of the high level of creatinine kinase. Since the muscle biopsy was compatible with dystrophinopathy, it was diagnosed as dystrophinopathy. In children with prolonged transaminase levels, such clinical symptom may not be encountered. However, in these patients, it should be noted that rare myopathies may cause transaminase elevation. By reporting of this case we wanted to emphasize that determination of creatine kinase levels is important for early diagnosis.

KEY WORDS: Dystrophinopathy, Newborn, Transaminase.

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INTRODUCTION

There are many diseases that can lead to transaminase elevation.¹ The major reasons are asphyxia, infections and toxic conditions. A rare reason of the elevation is also due to myopathies. Muscular dystrophy is a group of hereditary disease affecting many systems, as well as muscular and skeletal systems. Some of them indicate symptoms during delivery and then rapidly progress. However, some of them, which progress slowly, do not show any symptom until late adulthood.² Muscular dystrophy (MD) differs from the other neuromuscular diseases due to an inherited primary myopathy, displaying a progressive degeneration and necrosed muscle fibers.²

Dystrophin gene, which is known as the largest gene in humans and X-chromosome short arm band settled to number 21, has 79 exons. Because the gene deletion, duplication or point mutation occurs in patients with dystrophinopathy, dystrophin is never produced or its production is very small.³ The absence of

dystrophy or its defective structure essentially impair muscle contractions. Increase of calcium in intracellular zone leads to the emerging of events that progress to cell necrosis.³

Male children who are affected on delivery and in infancy are infrequently symptomatic in early infancy, and may also be slightly hypotonic. Disability on keeping head erected may be the first sign of muscle weakness which is remarkable.² The patients are manifested as a characteristic waddle walking, and walk upon their toes because of bilateral weakness of hip and leg muscles.^{3,4} Ambulation failure in patients with and without treatment is around 7-12 years and converts them to the dependent to wheelchair. The expected life expectancy is approximately between 20 and 25 years.⁵ Among the cases, 75% die due to respiratory failure and 20% die due to heart failure. The rest of the patients are lost due to pneumonia, pulmonary embolism and sudden death.² In this report, we wanted to share an uncommon case of distrophinopathy which was investigated because of elevated transaminase levels in newborns and highlight the importance of creatine kinase elevation in diagnosis.

CASE REPORT

Our case, born from a healthy non-relative marriage, delivered with NSD as term and weighed 3750g. 34-year-old mother had a spontaneous abortion previously. During the labor, we had noticed the state of existing cord entanglement, and amniotic fluid had stained by meconium. Meconium had also been found in gastric lavage. Respiratory distress persisted at oxygen-assisted monitorization of the patient. So we carried the patient to the service by diagnosis of meconium aspiration syndrome and perinatal asphyxia.

Physical examination revealed a slight depression in the newborn reflexes. The nails and umbilical cord had stained by meconium. The liver was 1cm below the rib and the other system examinations were normal. Complete blood count was detected within normal limits, and twenty-fold elevation was detected in transaminases (AST: 741 ALT: 737) at laboratory analysis. During clinical follow-up, we realized hematemesis for once. Hemostasis tests of patients were within normal limits. In these findings, we thought that this might be due to perinatal asphyxia. Cranial ultrasound and chest X-ray were normal. Abdominal USG was also normal except the minimal ectasia at pelvicalyceal system of left kidney. We used dual antibiotic therapy and Ranitidine, in addition to oxygen hood therapy.

The patient was evaluated as normal at the sixth day of the physical examination. He was being fed orally by and his transaminase levels (AST 120, ALT 89) were regressed. Therefore, the patient was discharged for follow-up as outpatient. When the patient whose age was appropriate to his neurological and motor development came for follow up the transaminase levels were elevated 4-6 times. Liver size, spleen size and liver parenchymal echogenicity were normal on abdominal ultrasound.

Distrophinopathy was detected in patients on muscle biopsy which was done because of myopathy suspicion. 51 exon deletions were detected at gene analysis of the DMD/BMD gene which resulted in the levels of lactate dehydrogenase as 1381 U/L and creatine kinase as 10930 U/L. The patient was followed up in child neurology outpatient clinic by providing genetic counseling.

DISCUSSION

Transaminase elevation may be encountered in newborns and infants due to many reasons.¹ Lorio et al evaluated 425 patients who were referred to the University of Naples (Italy) because of prolonged elevation of transaminase levels between the ages of 1-18. These patients were seronegative for hepatotropic viruses and had high enzyme levels for consecutive two-month. Enzym levels decreased to normal in 259 patients at the end of the first six months. Liver disease associated with obesity was found in 75 patients, genetic diseases were found in 51 patients, autoimmune hepatitis were found in seven patients, kolelityasis were found in five patients, choledoctus cyts was found in three patients and celiac disease was found in three patients.

Genetic diseases among the patients were Wilson's disease, muscular dystrophy, alpha-1-antitrypsin deficiency, Alagille syndrome, hereditary fructose intolerance, glycogen storage disease, ornithine transcarbamylase deficiency and Shwachman's syndrome. No etiology was found in twenty two children. Genetic disease rate was around 12%.⁶ Transaminase elevation may or may not be due to reasons that are associated with the liver. Gluten enteropathy, muscle diseases, after some operations, heavy exercise, marathon, and include various endocrine diseases which may be included as non-liver related reasons.⁷

In addition, a benign idiopathic hypertransaminasemia may be observed especially in childhood during neonatal period. If the elevation of transaminase is a result of liver function tests and

liver enzymes, this might refer to serious fault because of thinking them only as the cases of liver disease by ignoring history, other clinical and laboratory findings. The determination of serum creatinine kinase the level which we wanted to point out, is important in early diagnosis, and it demonstrates the possibility of having a seldom muscle disease in children with long-term high transaminase levels, even without the existence of a clinical evidence.

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