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Case Report
Olgu Sunumu

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A Rare Case Report of A Newborn That May Be Confused with Rickets and other Metabolic Bone Disorders: I-Cell Disease

Hiccup as a Rare Complication Following Lomber Dorsal Root Ganglion Pulsed Radiofrequency and Transforaminal Steroid Injection

Hüseyin Şimşek, Mustafa Akçalı, Mehtap Durukan Tosun, Mustafa Yıldırım,

Mersin City Training and Research Hospital Neonatal Intensive Care Unit, Mersin, Türkiye.

ORCID ID of the authors
HŞ. 0000-0002-3453-6802
MA. 0000-0002-0496-542X
MDT. 0000-0002-4041-2777
MY. 0000-0002-2196-9617

Correspondence / Sorumlu yazar:
Hüseyin ŞİMŞEK

Mersin City Training and Research Hospital Neonatal Intensive Care Unit, Mersin, Türkiye.

e-mail: drhuseyinsmsk84@hotmail.com

Abstract: I-cell disease is an autosomal recessive disorder caused by a transfer defect of several lysosomal hydrolase enzymes. The disease is characterized by rapidly progressive dysostosis multiplex, herniations, gingival hyperplasia and cardiac involvement. In this article, we would like to present a case with a fracture of the tibia bone, laboratory findings mimicking rickets, an alkaline phosphatase value of 4325 U/L and a genetic diagnosis of I cell disease.
Keywords: Bone deformity, Mucolipidosis, Neonatal

Özet: İ cell hastalığı (Mukolipidoz II), ciddi iskelet anormallikleri, intrauterin gelişme geriliği, kaba yüz görünümü, ilerleyici psikomotor gerilik, cillte kalınlaşma, dişeti hiperplazisi, fıtıklar ve erken dönemde ölüm ile karakterize otozomal resesif geçişli nadir görülen lizozomal depo hastalığıdır.Normalde endoplazmik retikulumda veya Golgi aygıtında bulunan N-asetilglukozamin1-fosfotransferaz eksikliğinden kaynaklanır. Hastalığın klinik, biyokimyasal ve laboratuvar bulguları raşitizmi taklit ettiği için tanı başlangıçta zor olabilmektedir
Anahtar Kelimeler: Kemik Deformiteleri, Mukolipoz, Yenidoğan

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1. Introduction

I cell disease (Mucolipidosis II) is a rare autosomal recessive lysosomal storage disease characterised by severe skeletal abnormalities, intrauterine growth retardation, coarse facial appearance, progressive psychomotor retardation, thickening of the skin, gingival hyperplasia, hernias and early death[1]. It is caused by a deficiency of N-acetylglucosamine 1-phosphotransferase which is normally found in the endoplasmic reticulum or Golgi apparatus[2]. Since the clinical, biochemical and laboratory findings of the disease mimic rickets, the diagnosis may be difficult at the beginning[3].

2. Case Report

The case who was born at 37 weeks of gestation, 2120 grams, Apgar 6/8 at 37 weeks of gestation as the 2nd living baby from the 4th pregnancy of a 35-year-old mother was admitted to the neonatal intensive care unit with the diagnoses of preterm birth, respiratory distress, intrauterine growth retardation and skeletal dysplasia. There was a 1st degree consanguinity between the parents, and 2 siblings were also exited due to epilepsy and syndromic infant. Neither of his siblings had a genetically confirmed diagnosis. Physical examination revealed rough facial appearance, high and narrow forehead, macrostomy, antevert nostrils, curvature of the left arm and right leg and mild joint restriction. Demineralization and osteopenic changes in the patient's costae are shown in Figure 1. On direct radiography, diffuse subperiosteal demineralisation in the long bones and costae, marked osteopenia, these skeletal changes were consistent with severe rachitic findings. Serum biochemistry revealed serum calcium 9.3 mg/dL, phosphorus 3.26 mg/dL, ALP 4325 U/L, PTH 613 pg/ml, 25(OH)D 14.7 ng/ml and tubular phosphate reabsorption 97.2%. Serum calcium, phosphorus, ALP, PTH and 25(OH)D levels were normal in maternal investigations. Echocardiography revealed hypertrophy in the left ventricular wall and interventricular septum. On the 20th day of hospitalisation, a distal fracture of the right tibia was found on radiograph taken because of swelling in the right tibia and splint treatment was applied. Mucolipidos 2 (I cell disease) was diagnosed in the patient who was found to be GNTAB homozygous as a result of whole exon sequencing performed for diagnosis. He died of sepsis and multiorgan failure on the 147th day of hospitalisation.

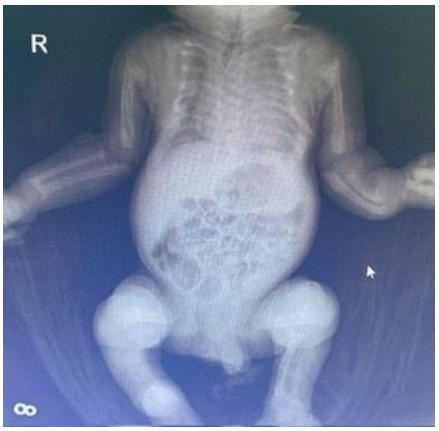

Figure 1. Significant osteopenia in the humerus and ribs

3. Discussion

Neonatal bone disorders are a process that begins in the fetal period and are caused by structural defects or mineralisation defects. Despite extensive genetic investigations, the cause of the bone disorder is rarely identified. As advances are made in the field of genetics, diagnosis will become easier. Further targeted molecular genetic tests, prenatal diagnosis in families at risk, prenatal counselling should be provided thanks to quality antenatal imaging[4]. In bone deformities and fractures in the neonatal and early infancy period, skeletal dysplasias such as rickets and osteogenesis imperfecta should be considered, as well as I-Cell disease. Although there is no definite treatment for mucolipidosis II, bone marrow transplantation has been reported in a limited number of patients[5]. It should be aimed to protect infections, provide nutrition and manage general health care of these patients by

multidisciplinary health teams. In our case, only supportive treatment was given and he died of infection and multiorgan failure.

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