



Multicystic Renal Dysplasia in a Child with Split Hand/Split Foot Malformation

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Abstract

Split hand/split foot malformation is a human developmental disorder characterized by missing central digits and other distal limb malformations. Multicystic renal dysplasia is the most common cause of an abdominal mass in the new born period and is the most common cystic malformation of the kidney in infancy. Here, we report a case of split hand/split foot malformation with a submucosed cleft palate and multicystic renal dysplasia in one-year old boy.

Key words: Ectrodactyly, multicystic, renal, dysplasia

Split El/Ayak Malformasyonlu Çocukta Multikistik Böbrek Displazisi

Özet

Split el/ayak malformasyonu santral parmakların kaybı ve diğer distal ekstremitel malformasyonları ile karakterize bir insan gelişimsel bozukluktur. Multikistik renal displazi yeni doğan dönemde abdominal kitlenin en sık nedenidir ve bebeklik döneminde böbreğin en sık görülen kistik bir anomalisidir. Burada, bir yaşındaki oğlan çocuğunda bir mukozaaltı yarık damak ve multikistik renal displazi ile split el/ayak malformasyonu sunulmaktadır.

Anahtar kelimeler: Ektrodaktili, multikistik, böbrek, displazi

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INTRODUCTION

Split-hand/split-foot malformation (SHFM), also known as ectrodactyly, is a congenital limb malformation, characterized by a deep median cleft of the hand and/or foot due to the absence of the central rays (1). SHFM may occur both as an isolated malformation and as part of several syndromes including the Ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome and acrorenal syndrome (2,3). Associated renal lesions described so far comprise agenesis, bilateral hypoplasia (originally diagnosed as oligomeganephronia), and duplication abnormalities (3). In this report, we present a case of split-hand/split-foot malformation with cleft palate, multicystic renal dysplasia, rickets and severe iron deficiency anemia that did not take any medical help until 12 months of age because of the curious religious reasons.

CASE

A one-year-old boy was admitted to our clinic with the complaints of failure in suction, pallor and vomiting. He was the second child of the third degree consanguineous parents. Family history was unremarkable and the other sibling was healthy. Physical examination revealed a submucosed cleft palate, epiphyseal enlargement at the wrists, palpable enlargement of the costochondral junctions (the rachitic rozary), pallor, tachycardia, and 2-3/6 degree systolic murmur at cardiac auscultation. Extremity examinations showed deep median cleft in his hands and feet and fusion of the remaining digits (Figure 1 and 2). Laboratory investigation showed hemoglobin 3,3 g/dl, mean corpuscular volume 46 fl, red blood cell distribution width 22%, serum ferritin level 2,8 ng/ml, serum iron level 12 ug/dl, iron binding capacity of the serum 356 ug/dl, calcium 8,1 mg/dl, phosphor 2,1mg/dl, alkaline phosphatase 1300 U/L, 25-OH-Vitamin D level 5,16 ug/L, parathormon level 441 pg/ml. Bone survey investigation showed cupping and fraying of the distal ends of the radius and ulna, rudimented proximal phalanx in the right hand middle phalanx, absence of the third phalanx and transverse situated bone from third metacarpal to fourth phalanks in the left hand, absence of the 1-3 metatarsus in the right foot and absence of the 1-4 metatarses in the left foot. Abdominal ultrasonography showed multicystic renal dysplasia in the left kidney. The shape and size of the right kidney was in normal ranges for age.



Figure 1. Split Hand

Figure 2. Split Foot

Erythrocyte suspension was given in a dose of 10 ml/kg for severe anemia, oral iron replacement therapy was begun, oral stoss Vitamin D treatment was administered in a dose of 300.000 IU (three times 100.000 IU in one hour intervals) and oral calcium lactat and 400 IU vitamin D treatment was given for continue therapy.

DISCUSSION

Split hand/foot malformation (SHFM) results from failure of formation of parts of hands, feet or both due to a variable deficiency of central rays of the autopod. (4) SHFM, is a rare congenital malformation and the incidence of SHFM approximates 1/90.000 live birth.(5,6) Inherited as an autosomal dominant trait, both sporadic and familial cases have been described. (7,8) but there was no similar family history in our patient. Associated renal lesions described so far comprise agenesis, bilateral hypoplasia (originally diagnosed as oligomeganephronia), and duplication abnormalities(3). In our case abdominal ultrasonography showed multicystic renal dysplasia in the left kidney.

EEC (ectrodactyly, ectodermal dysplasia, cleft lip/palate) syndrome must be considered in differential diagnosis, in which besides limb malformations, there is cleft lip and palate, lacrimal duct atresia and ectodermal defects involving skin, hair, teeth and nails (6). In our patient, there were no atypical anhydrotic ectodermal dysplasia and lacrimal duct atresia. A similar overlap has been observed in Adams-Oliver syndrome with associated defects of calvarium and scalp(5). There were no calvarium or scalp defects in our case.

SHFM, is not a big problem for functional activities but unfortunately is an important problem for psychosocial situation. In appropriate ages with appropriate surgical treatment the best result can be provided.

In conclusion, a case of a split-hand/split foot malformation with multicystic renal dysplasia is reported and to the best of our knowledge no other SHFM and multicystic renal dysplasia together case report has been published to date.

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