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Ellis-van creveld syndrome accompanied by cleft glans penis-epispadias: a case report

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ÖZET

Cleft glans penis-epispadias'ın eşlik ettiği ellıs-van creveld sendromu: olgu sunumu

Fetal taşikardi en iyi bilinen non-immun hidrops fetalis nedenidir. Fetal taşikardi tedavi edilmezse hidrops fetalise ve fetal kayıba neden olabilmektedir. Hidrops fetaliste prognoz antiaritmik tedaviye hızlı yanıta, gebelik haftasına, fetusun iyilik haline ve yüksek kardiyovasküler profil skora (KVPS) bağlıdır. Biz burada SVT' ye sekonder gelişen 28 haftalık hidropslu fetusu olgu olarak sunduk. Biz KVPS'u tartışmak amacıyla bu olgu sunumunu sunduk . Olgumuz da Digoksin ve sotalol den oluşan kombine maternal antiaritmik tedavinin 1. günde fetal taşikardinin başarılı şekilde sinüs ritmine dönüştüğü izlendi fakat fetusun iyilik hali gittikçe bozulup hidrops fetalis ağırlaştı ve tedavinin 21. günü fetal kayıp gerçekleşti. Fetusun kardiyovasküler profil skoru (KVPS) 7/10 (cilt ödemi 2 puan, duktus venozusta revers atriyal A dalgası 1 puan kayıp) idi. Ve bu yaşamla bağdaşır kabul edilebilir bir skordu. Fakat olgumuzda hayatla bağdaşır (7/10) KVPS olmasına rağmen fetal kayıp oluşmuştur. Kardiyovasküler profil skor SVT'ye sekonder gelişen hidrops fetalisin prognozunu belirlenmede yanıltıcı olabilir

Anahtar Kelimeler: *Hidrops fetalis; Fetal Taşiaritmi, Kardiyovasküler Profil Skor*

SUMMARY

Ellis-van Creveld (EVC) syndrome is a rare autosomal recessive skeletal dysplasia which presents with chondrodysplasia, ectodermal dysplasia, polydactyly, and congenital heart disease. Urinary tract anomalies such as renal agenesis, congenital megaureter, hypospadias and hypoplastic external genitalia are some of the rare associations of the EVC syndrome. EVC syndrome and cleft glans penis-epispadias association was not reported before.

Key words: *Ellis-van Creveld syndrome, cleft glans penis-epispadias, child*

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Introduction

Ellis-van Creveld (EVC) syndrome also called 'chondroectodermal dysplasia' and 'mesodermal dysplasia' is an autosomal recessive disorder characterized by short-limb dwarfism, polydactyly, thoracic defects, congenital heart disease, ectodermal defects, and other rare but wide range spectrum of clinical manifestations [1]. This syndrome is included within short-rib dysplasias syndromes together with asphyxiating thoracic dystrophy (Jeune syndrome-ATD), and short-rib-polydactyly (SRP) syndromes [2]. Urinary tract anomalies are rare in EVC syndrome [3]. In the literature, there are only two cases of cleft glans penis associated with epispadias (CGPE). We present an EVC syndrome and CGPE association, which was not reported previously.

Case Report:

A 4-year-old boy referred our department for circumcision. On physical examination the patient has low-set shoulder, a narrow thorax, lumbar lordosis, genu valgus, broad hands and feet, sausage shaped fingers and hypoplastic nails. When the intact prepuce retracted, clefted glans penis and midshaft epispadias was dedected (Fig.1). The external urethral mea was found on the dorsum penis at the midshaft position. Cardiovascular system examination was normal. In his history there was not person with similar features in his family. Blood and urine analysis were within normal limits.

On x-radiographic examinations, the skull and spine were unremarkable. The pelvis was small and iliac blades were squared-shaped and flattened in addition to horizontal acetabuli. Thickening and shortening of long bones, widening of proximal tibial shaft with delayed development of tibial plateau (dome-shaped metapysis) and dislocation of radial head due to shortening of ulna were also observed (Fig 2A). Beside, hypoplasia of terminal phalanges with cone-shaped epiphyses, bilateral polysyndactyly, and fusion of the right carpal bones were present (Fig.2B). Cardiothoracic ratio increase was observed as well as high location of clavicules on chest x-ray. The chest was bell-shaped (Fig.2C). All above radiographic findings were suggesting EVC syndrome.

Patient was operated for cleft glans penis and epispadias. Postoperative period was uneventfull. Patient was discharged 21 days after the operation. The other anomalies were managed by related clinics.

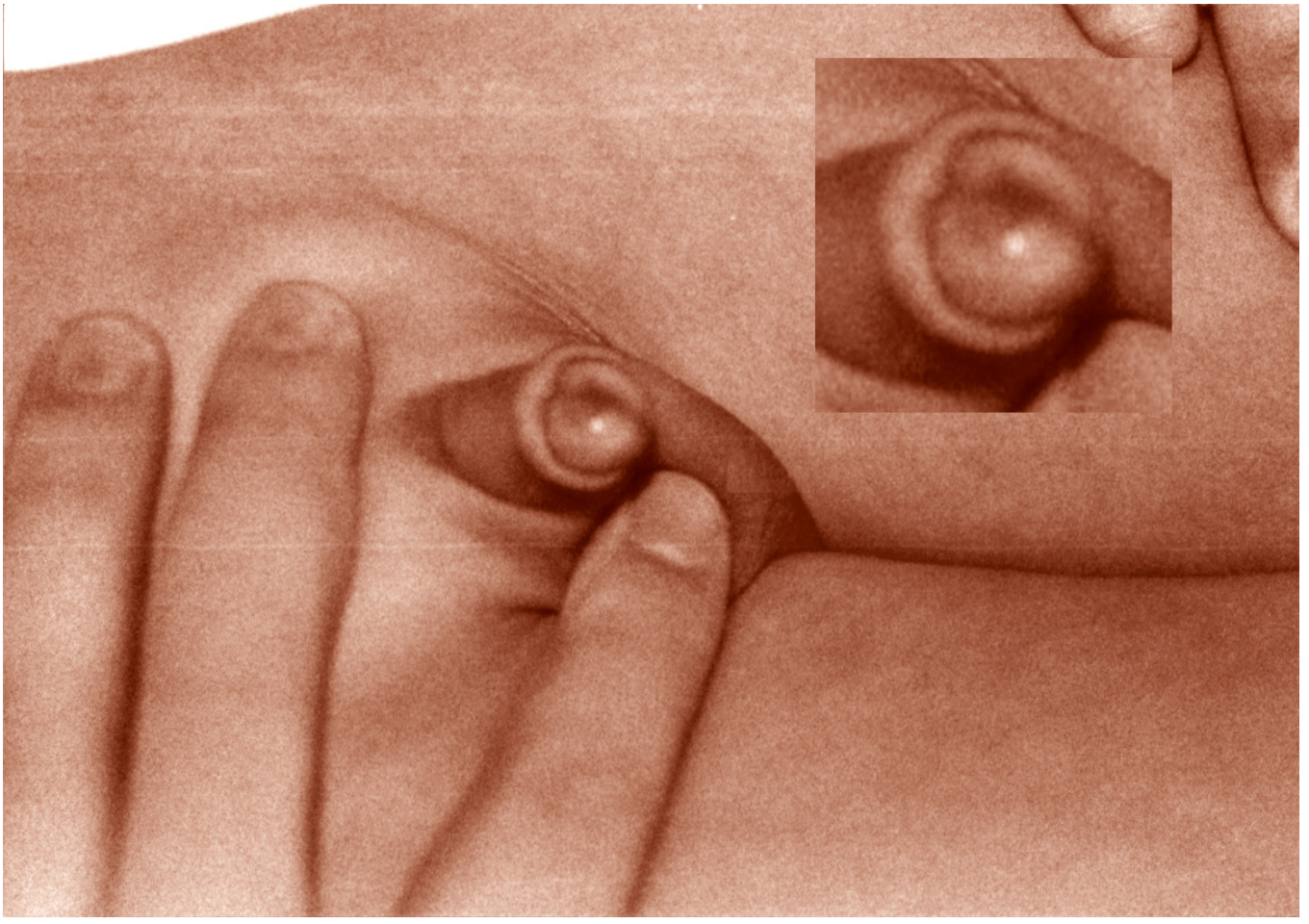

Fig. 1. Clinical photograph shows cleft glans penis-epispadias.

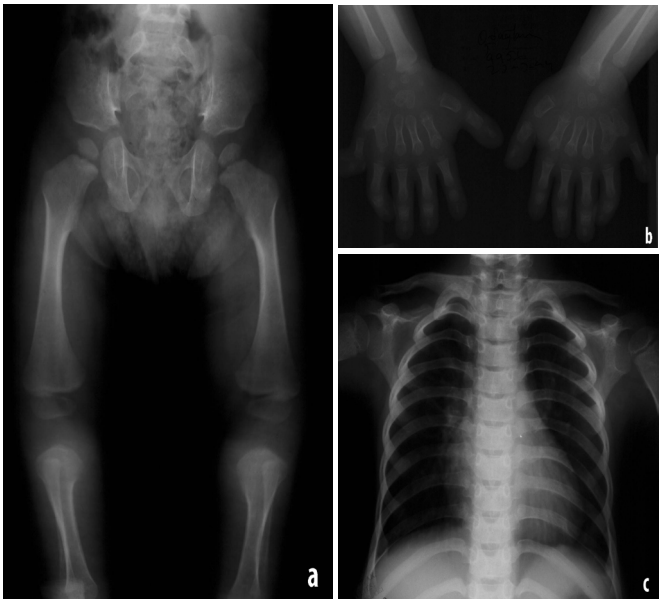

Fig. 2. Pelvis and lower extremity x-radiography **(A)** demonstrates small pelvis, squared shaped iliac blades and horizontal acetabuli. Long bones are undertubulated and proximal tibial plateaus are dome-shaped. On x-radiography of hands **(B)** bilateral polysyndactyly, right carpal bones fusion and distal phalangeal hypoplasia are noted. Chest x-radiography **(C)** shows highly located clavicles, bell-shaped thorax and increased cardiothoracic ratio.

Discussion:

Ellis-van Creveld (EVC) syndrome is a skeletal dysplasia which have common features of short rib-polydactyly (SRP) syndromes and asphyxiating thoracic dystrophy (ATD) [1]. Renal agenesis, congenital megaureter, hypospadias and hypoplastic external genitalia are some of the rare associations of the EVC syndrome [3]. Our case had CGPE association.

The first case of cleft glans penis was reported by Hofmokl in 1897 [4]. Since then other single cases were reported which urethra was normal ventral position or hypospadiac. Till today only two cleft glans penis and epispadias (CGPE) cases were reported according to our knowledge [4,5]. In the literature, we could not find EVC syndrome and CGPE association.

Polydactyly, another frequent manifestation of EVC syndrome, is usually seen as postaxial hexadactyly of the hands, although it may be entirely absent. Moreover, hypoplasia of terminal phalanges and cone-shaped epiphyses may present. Rarely polydactyly of the feet may also be seen. Our case has bilateral upper and lower limb polydactyly.

Manifestations of main ectodermal dysplasia including sparse hair, nail dystrophy and malformed teeth present in 70% of patients [1]. Congenital heart defects was reported at least in 60% of patients and include persistent, common, atrioventricular canal, ventricular septal defect, or atrial septal defect [6]. On routine cardiovascular examination of

the patient, we did not find any pathology so we did not apply other advanced examination methods.

The essential radiologic findings of EVC syndrome are distalward shortening of limps and generalized thickness, and coarseness of bones [1]. Radiologic features of EVC syndrome are curvature of the humerus, enlargement of distal ends of the radii and ulnae, supernumerary carpal bone centers with fusion, synmetacarpalism, synostosis, wedge-shaped tibial epiphyses, genu valgus, small pelvis and iliac bones, trident shape of acetabuli, highly located clavicles and bell-shaped thorax vary depending on severity of the disease, and age [1,2].

ATD and SRP syndromes are also autosomal recessive skeletal dysplasias with associated multiorgan involvement. While the prognosis of ATD is poor either due to respiratory insufficiency in infancy or to renal failure in older ages, SRP syndromes are subdivided four groups based on clinical, radiologic and cartilage histomorphology.

Because of the radiographic and clinical similarities, these disorders were suggested as a part of continuous spectrum ranging from pre-natally lethal SRP to mild cases of EVC and ATD. In addition to these phenotypic resemblances, it was also speculated that EVC, ATD and SRP syndromes were allelic.

Patients with EVC syndrome should be investigated for possible genital and urinary system anomalies. Both in the patient and the family members genetic counseling should be recommended.

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