Türk Göğüs Kalp Damar Cerrahisi Dergisi 2012;20(3):619-621
doi: 10.5606/tgkdc.dergisi.2012.119

*Case report / Olgu Sunumu*

# Polydactyly and ostium primum type atrial septal defect: Ellis-van Creveld syndrome

## Polidaktili ve ostium primum tip atriyal septal defekt: Ellis-van Creveld sendromu

**Orhan Veli Doğan,[1] Murat Günday,[1] Ergün Haliloğlu,[1] Ali Rıza Akyüz[2]**

Departments of [1]Cardiovascular Surgery, [2]Cardiology,
Ahi Evren Thoracic and Cardiovascular Surgery Hospital, Trabzon, Turkey

Comorbid polydactyly and atrial septal defect (ASD) is a very rarely seen anomaly. Basic clinical findings of Ellis-van Creveld syndrome include muscular skeletal disorders in the hands in 100% of cases, additional symmetrical postaxial polydactyly in the feet in 10-25% of cases, disproportionate dwarfism, a narrow thorax leading to respiratory problems, ectodermic dysplasia in the skin, nails, and teeth, a high palate, malocclusion, micrognathia, and congenital heart diseases, especially ASDs, in almost half of the patients (50-60%). In this article, we report a case with polydactyly and primum type ASD along with the surgical treatment and outcomes.

*Key words:* Ellis-van Creveld syndrome; polydactyly; primum atrial septal defect.

Polidaktili ve atrial septal defekt (ASD) birlikteliği çok nadir rastlanan bir anomalidir. Ellis van Creveld sendromu (EVC) ile ilgili temel klinik bulgular %100 olguda ellerde, %10-25 olguda buna ilaveten ayaklarda simetrik postaksiyel polidaktili içeren kas iskelet sistemi bozuklukları, orantısız cücelik, solunum sorunlarına neden olan dar göğüs kafesi, cilt, tırnaklar ve dişlerde ektodermal displazi, yüksek damak, maloklüzyon, mikrognati ve hastaların hemen hemen yarısında (%50-60) özellikle ASD'ler olmak üzere konjenital kalp hastalıklarıdır Bu yazıda, cerrahi tedavi ve sonuçları ile birlikte polidaktili ve primum tip ASD saptanan bir olgu sunuldu.

*Anahtar sözcükler:* Ellis-van Creveld sendromu; Polidaktili; primum atriyal septal defekt.

Polydactyly and atrial septal defect (ASD) occurring concurrently is a rare anomaly. The basic clinical findings for Ellis-van Creveld syndrome include muscular skeletal disorders in the hands in 100% of cases, additional symmetrical postaxial polydactyly in the feet in 10-25% of cases, disproportionate dwarfism, a narrow thorax leading to respiratory problems, ectodermic dysplasia in the skin, nails, and teeth, a high palate, malocclusion, micrognathia, and congenital heart diseases, especially ASDs, in almost half of the patients (50-60%).[1]

We report a case with polydactyly and ostium primum type ASD with appropriate surgical treatment.

## CASE REPORT

A 40-year-old female patient was admitted to our cardiology clinic with palpitation. The patient was 1.45 cm with normal intelligence, distinctively short extremities, fingernails on the short hand, and six fingers and toes on each hand and foot (Figures 1 and 2). Physical examination revealed accessory labiogingival frenulum in the upper lip (Figure 3), and patient had prosthetic teeth. The patient also had a high palate (Figure 4). During cardiac examination, 3/6 systolic murmur was detected on pulmonary focus, and cardiomegaly was observed on the chest X-ray. Echocardiography revealed ostium primum type ASD, a mitral cleft with third degree mitral valve

Available online at
www.tgkdc.dergisi.org
doi: 10.5606/tgkdc.dergisi.2012.119
QR (Quick Response) Code

*Received:* February 19, 2010 *Accepted:* April 5, 2010

Correspondence: Murat Günday, M.D. Başkent Üniversitesi, Konya Uygulama ve Araştırma Merkezi, Kalp ve Damar Cerrahisi Kliniği, 42080 Selçuklu, Konya, Turkey.
Tel: +90 332 - 257 06 06 e-mail: gundaymurat@yahoo.com

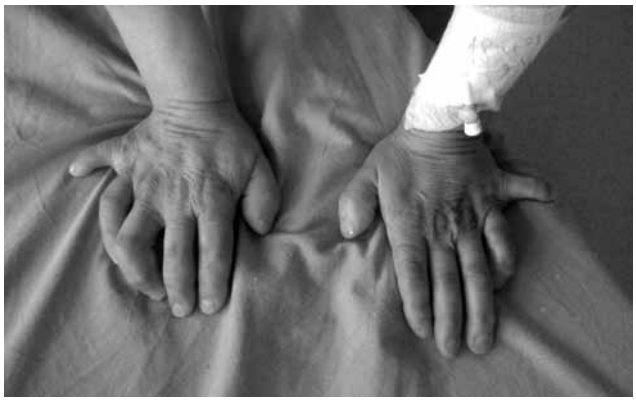

**Figure 1.** Polydactyly and fingernails on the short hand.

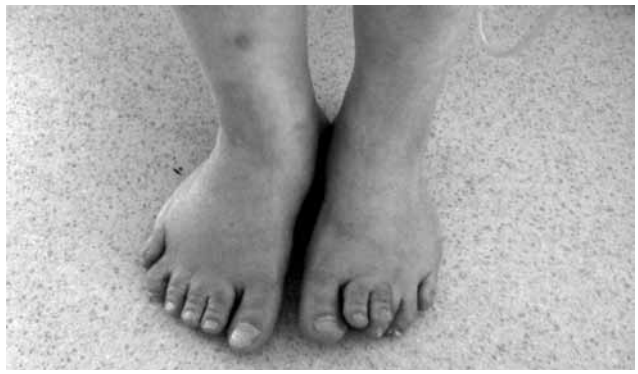

**Figure 2.** 10-25% of cases in feet symmetrical postaxial polydactyly.

insufficiency, and pulmonary hypertension (45 mmHg). The pulmonary artery pressure was 46/17 (29 mmHg on average), the mixed venous oxygen saturation was 73.9%, and the shunt ratio was Qp/Qs= 3.2 at right heart catheterization. A right atriotomy was then performed to correct the ostium primum defect using a midsternal incision, extra-corporeal circulation, and selective caval cannulation. The atrial septum was reconstituted with the autologous pericardium, leaving the coronary sinus in the right atrium. The mitral cleft was primarily mended and flushed to obtain adequate coaptation. The patient was extubated the first day in the intensive care unit (ICU). Control echocardiography revealed minimal mitral insufficiency and no interatrial shunt, and the patient was discharged from the hospital on the postoperative eighth day.

## DISCUSSION

Ellis-van Creveld syndrome is a rare congenital disease that has an autosomal recessive pattern of inheritance,[2] and the real incidence rate and etiology have not yet been determined. This syndrome is commonly (1/5000) seen in the Amish population in the United States,[3] with no difference in frequency between men and women. However, as with other autosomal recessive diseases, it is seen much more with intermarriage,[3-7] as was the case with our anamnestic patient (mother-father first degree relationship). An early prenatal diagnosis can be performed after the 18$^{th}$ week with ultrasonography to determine whether there are the characteristic signs of EVC syndrome, such as a narrow thorax, polydactyly (fingers and toes), disorder of the long bones, and cardiac defects, but the diagnosis most often occurs after birth.[8]

The EVC phenotype is labile and affects many organ systems. Clinical features after birth can be summarized as a disproportionate, short body type, polydactyly that affects the hands and sometimes the feet, and congenital heart defects. Congenital heart defects are found in 50-60% of patients, with ventricular septal defect, patent ductus arteriosus, mitral and tricuspid valve disorder, hypoplastic left

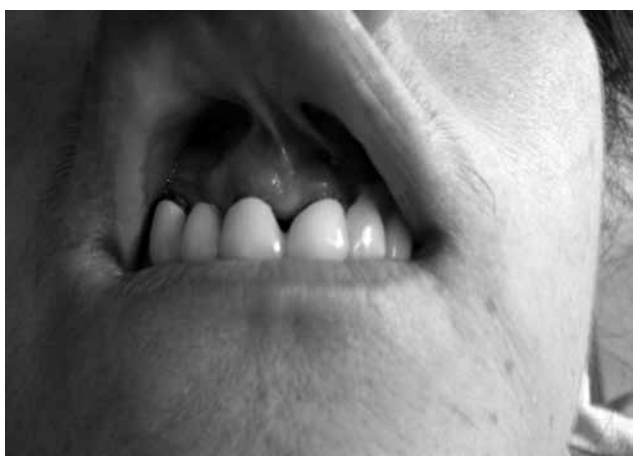

**Figure 3.** Labiogingival frenulum in upper lip.

**Figure 4.** High palate.

heart syndrome, single ventricle, atrioventricular channel defects, transposition of the large arteries, aortic coarctation and a totally abnormal pulmonary vein being exhibited.[4,8] Almost half of the patients die from cardiac and respiratory complications in childhood.[5,6]

The most prominent symptoms in the skeleton system are the shortness of the long bones and ribs (disproportionate dwarfism), nail and teeth disorders, and polydactyly. Patients have been reported with extra digits ranging between 1.15 and 1.50 cm in length.[6]

Mouth symptoms of EVC syndrome, such as multiple frenula, a short upper lip, and broad alveolar ridges, are common. Dental anomalies, including partial anodontia, neonatal teeth, small teeth, and enamel hypoplasia, are also possible.[9] Most patients have normal intelligence.[5,6,10]

As far as the molecular genetic diagnosis is concerned, it is understood that these patients have a mutation in two genes called EVC1-EVC2 that is located in the short part of the fourth chromosome,[11] but few research centers conduct a genetic analysis for a differential diagnosis because of the rarity of the disease and the high cost. Our patient was not tested.

In the literature, most patients are treated for EVC in childhood, and the number of adults who have surgery for this condition is very limited. Our case presented a very rare occurrence of this syndrome in an adult and demonstrates that adults with EVC and concurrent ostium primum defects can be safely operated on for total correction.

### Declaration of conflicting interests

The authors declared no conflicts of interest with respect to the authorship and/or publication of this article.

### Funding

The authors received no financial support for the research and/or authorship of this article.

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