

## Percutaneous right ventricle outflow tract stenting in a patient with trisomy 18 associated with double outlet right ventricle

### Trizomi 18 ve çift çıkışlı sağ ventrikül tanılı bir bebekte sağ ventrikül çıkım yolu stenti ile perkütan palyasyon

Erkut Öztürk, M.D., Ender Ödemiş, M.D., Neslihan Kıplapınar, M.D.

Department of Pediatric Cardiology, Istanbul Mehmet Akif Ersoy Thoracic and Cardiovascular Surgery Training and Research Hospital, Istanbul

**Summary**– Trisomy 18, or Edwards syndrome, is the second most common chromosome anomaly after trisomy 21. Various types of congenital heart diseases are seen in the majority of trisomy 18 patients. Palliative treatment of right ventricular outflow tract (RVOT) stenosis includes options like balloon dilatation, stenting and surgery. Herein, we present a case with trisomy 18 and double outlet right ventricle, pulmonary stenosis, and ventricular septal defect. During the follow-up, at the age of three months, his saturation dropped to 70% and an interventional procedure was planned. The patient was considered high risk, and after discussing treatment options with the family, RVOT stenting was chosen. The patient was lost on the 8th day of the follow-up.

Trisomy 18, or Edwards syndrome, is seen in 0.3 of every 1000 live births, making it the second most common chromosome anomaly after trisomy 21. Some of the structural defects seen in Edwards syndrome are intrauterine growth retardation, microcephaly, micrognathia, low-set ears, and extremity anomalies. Congenital heart diseases, most commonly ventricular septal defect (VSD), are seen in 90% of cases. Some of them may lead to right ventricular outflow tract (RVOT) stenosis, necessitating medical or surgical intervention.<sup>[1-3]</sup>

Systemic or supra-systemic right ventricular pressure due to pulmonary stenosis as well as increased cyanosis, exercise intolerance, and fatigue due to re-

**Özet**– Trizomi 18 veya Edward sendromu, trizomi 21'den sonra en sık görülen kromozom anomalisidir. Bu hastalarda farklı tiplerde doğumsal kalp hastalığı görülebilmektedir. Bunlardan sağ ventrikül çıkım yolu (RVOT) darlıklarının palyatif tedavisinde balon, stent ve cerrahi yaklaşım gibi farklı tedavi seçenekleri vardır. Bu yazıda trizomi 18 tanısı konmuş olan ve ekokardiyografisinde çift çıkışlı sağ ventrikül, pulmoner stenoz ve ventriküler septal defekt saptanan bir olgu sunuldu. Poliklinik kontrolüne alınan hastanın üç aylıkken oksijen saturasyonunun %70'e düşmesi nedeniyle mevcut kardiyak patolojisine müdahale edilmesi planlandı. Yüksek riskli olarak kabul edilen hastaya, aile ile olası tedavi seçenekleri değerlendirildikten sonra RVOT'ye stent yerleştirilmesine karar verildi. Hasta işlemin sekizinci günü kaybedildi.

duced pulmonary blood flow constitute indications for treatment in patients with RVOT stenosis.

#### Abbreviations:

DORV Double outlet right ventricle  
RVOT Right ventricular outflow tract  
VSD Ventricular septal defect

RVOT stenting is an alternative to surgery that achieves safe and effective palliation, particularly in high-risk cases and in congenital anomalies. This text presents RVOT stenting in a trisomy 18 patient.

#### CASE REPORT

The patient was the first child of a consanguineous father aged 23 and mother aged 20 (cousins on mother's

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Correspondence: Dr. Erkut Öztürk. İstanbul Mehmet Akif Ersoy Eğitim ve Araştırma Hastanesi, İstasyon Mahallesi İstanbul Caddesi Bezirganbağçe Mevki Küçükçekmece 34303 İstanbul.

Tel: +90 212 - 616 54 14 e-mail: erkut\_ozturk@yahoo.com

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side). Born by normal spontaneous delivery, the patient had a birth weight of 2300 g, micrognathia, microcephaly, inspiratory stridor, and pectus carinatum, and was diagnosed as trisomy 18 by chromosome analysis. The patient was referred to us with heart murmurs to be evaluated for possible congenital heart diseases. Echocardiography revealed double outlet right ventricle (DORV), significant pulmonary stenosis (valvular and subvalvular) and VSD. In the 3rd month of outpatient follow-up, the patient's oxygen saturation fell to 70%, necessitating treatment of the existing cardiac pathology. The patient was considered to be at high risk, and after discussing treatment options with the family, RVOT stenting was chosen. The RVOT anatomy was visualized using angiography under general anesthesia (Fig. 1a). As the patient had a narrow RVOT, a 0.014 inch Roadrunner guidewire was used to pass the pulmonary valve, and a 5 mmx13 mm coronary stent (Skylor Co-Cr stent, Invatec, Roncadelle, Italy) was inserted. Follow-up contrast injection revealed a significantly increased amount of contrast material passing through the RVOT (Fig. 1b). At the end of the procedure, saturation increased to 85%. After one day in the intensive care unit, the patient was extubated, observed for four more days and intubated again after developing aspiration pneumonia caused by feeding. Despite broad-spectrum antibiotic support, the patient died on the 8th day of follow-up.

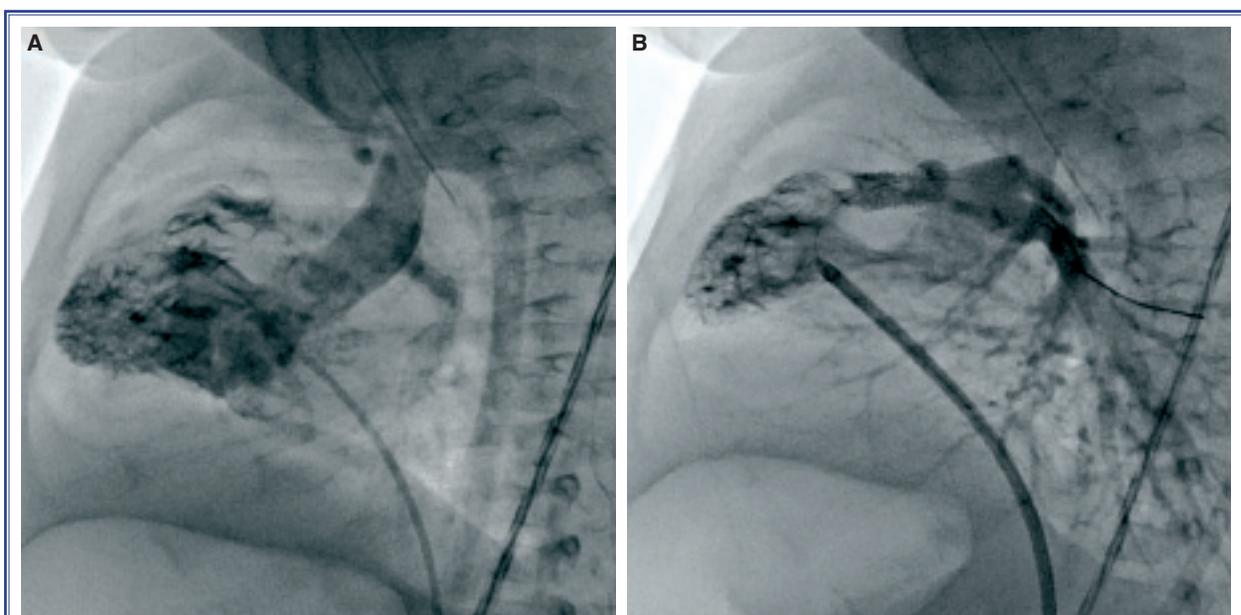
## DISCUSSION

The survival rate for Edwards syndrome is low. Pregnancies end in embryonic or fetal death in 90% of cases, with only 10% of affected children surviving past the first year of life. Death is generally caused by heart and kidney anomalies, feeding problems, sepsis, and apnea secondary to central nervous system defects.<sup>[3-6]</sup>

Congenital heart disease is reported in 95% of trisomy 18 cases.<sup>[5-7]</sup> Kosho et al.<sup>[7]</sup> found congenital heart diseases in 96% of 24 trisomy 18 patients. The most common ones were VSD, atrial septal defect and patent ductus arteriosus, while RVOT stenosis was seen in only three patients.

RVOT stenosis can be a component of congenital heart diseases like tetralogy of Fallot, DORV and hypoplastic right heart. Due to symptoms like increased cyanosis, exercise intolerance and fatigue caused by reduced pulmonary blood flow, these patients require intervention by balloon dilatation, stenting or surgery.

Surgical reconstruction is considered to be the first choice in the management of RVOT stenosis. However, in patients who are not suitable for surgical treatment, balloon dilatation and stent implantation into the RVOT can be used.<sup>[8,9]</sup>



**Figure 1.** (A) Right ventricular outflow tract stenosis and pulmonary artery blood flow as seen on angiogram. (B) The pulmonary artery and branches after RVOT stenting.

Balloon expandable stents can be implanted into the RVOT in cases that are resistant to simple balloon dilatation. Although this strategy is a palliation and does not provide definitive treatment, some patients may benefit from non-surgical improvement in RVOT obstruction.

The major possible complications of stent implantation are stent migration, ventricular arrhythmias, stent breakage or collapse, and subsequent re-stenosis. While these patients develop significant free pulmonary insufficiency after the procedure, there are rarely hemodynamic problems.<sup>[8,9]</sup>

Hausdorf et al.<sup>[10]</sup> noted that stenting might provide good palliation in infundibular stenosis. The authors used stenting into the RVOT after radiofrequency valvotomy for pulmonary atresia. Nevertheless, there may be some risks of such a procedure, including stent compression and associated fracture, pulmonary embolization, and re-stenosis.<sup>[6]</sup>

Compared to balloon dilatation, stenting is more effective in reducing the pressure gradient and decreasing stenosis at the subvalvular and valvular levels, yielding better results in terms of development of pulmonary artery branches.<sup>[6,8,9]</sup>

Stenting may be preferred if the family refuses surgery or if surgery is high risk (neonatal period, severe cyanosis, low birth weight, concomitant systemic diseases, severe acidosis, and additional congenital malformations).<sup>[4-6]</sup>

We chose RVOT stenting as the patient had trisomy 18 with severe cyanosis, and the family had refused surgery.

In conclusion, RVOT stenting is an efficient alternative method of palliative treatment that improves the quality of life in trisomy 18 patients with RVOT obstruction caused by congenital heart diseases. The presented patient was the first trisomy 18 patient to undergo RVOT stenting.

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**Key words:** Child, preschool; heart septal defects, ventricular/surgery; pulmonary artery/abnormalities; right ventricular outflow obstruction; stents; trisomy/diagnosis.

**Anahtar sözcükler:** Çocuk, okul öncesi; kalp septal defekti, ventrikül /cerrahi; pulmoner arter/anomali; sağ ventrikül çıkım yolu; stent; trizomi/tanı.