



Case Report / Olgu Sunumu

## Our experience on Bentall procedure in an adult patient with Baraitser-Winter syndrome

*Baraitser-Winter sendromlu erişkin bir hastada Bentall prosedürü deneyimimiz*

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### ABSTRACT

Baraitser-Winter syndrome is a very rare genetic disorder caused by cytoplasmic actin-encoding genes defects. Although most patients have similar phenotype, concomitant cardiac anomalies widely vary. In addition to well-described congenital heart diseases, aortic aneurysms occur due to underlying actin gene mutation in these patients in the further years. Herein, we present a 26-year-old male case who underwent Bentall procedure with the diagnosis of new-onset aortic valve regurgitation and an ascending aorta aneurysm.

**Keywords:** Adulthood; ascending aortic aneurysm; Baraitser-Winter syndrome; Bentall procedure.

Baraitser-Winter syndrome (BWS) is a very rare genetic disorder and was first described in 1988 in siblings as a combination of iris coloboma, bilateral ptosis, hypertelorism, broad nasal bridge, prominent epicanthic folds, and growth and mental retardation.<sup>[1]</sup> According to recent literature, BWS is caused by heterozygous missense mutations in ubiquitous cytoplasmic actin-encoding genes ACTB or ACTG1 which encode  $\beta$ - and  $\gamma$ -actins.<sup>[2]</sup>

Over years, several cases with similar resemblance of BWS have been reported worldwide, and the spectrum has been widened with microcornea, microphthalmia, microcephaly, trigonocephaly, gyral malformation, seizures, hypotonia, and cardiac, urogenital, and skeletal defects in addition to aforementioned dysmorphologies.<sup>[3-9]</sup> Although various congenital cardiac anomalies have been reported as clinical features of this syndrome including patent ductus arteriosus, ventricular septal

### ÖZ

Baraitser-Winter sendromu, sitoplazmik aktin kodlayan gen defektinin neden olduğu, oldukça nadir bir genetik bozukluktur. Pek çok hasta benzer fenotipe sahip olmasına rağmen, eşlik eden kalp anomalileri çeşitlidir. İyi tarif edilmiş doğuştan kalp hastalıklarının yanı sıra, ileriki yıllarda altta yatan aktin gen mutasyonuna bağlı olarak, bu hastalarda aort anevrizmaları görülebilir. Bu yazıda, yeni gelişen aort kapak yetmezliği ve çıkan aort anevrizması tanısıyla Bentall prosedürü yapılan 26 yaşında bir erkek olgu sunuldu.

**Anahtar sözcükler:** Erişkinlik; çıkan aort anevrizması; Baraitser-Winter sendromu; Bentall prosedürü.

defect, mitral-tricuspid valve prolapse, and mitral-tricuspid regurgitation, BWS has no precise definition for adult patients.<sup>[5,6,9]</sup>

Herein, we describe a 26-year-old male case who underwent Bentall procedure with the diagnosis of new-onset aortic valve regurgitation and an ascending aorta aneurysm.

### CASE REPORT

A 26-year-old male patient born from unrelated parents was diagnosed with BWS in 2001. The patient was referred to our center with a recent diagnosis of an ascending aortic aneurysm with severe aortic regurgitation. He was suffering from dyspnea and episodic chest pain. In his medical history, he was on medication with nebivolol 5 mg/day due to hypertension for three years and risperidone 1 mg per day ever since childhood. Previously, he underwent

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**Figure 1.** Typical syndromic face features including short neck, broad nasal bridge, ptosis, hypertelorism, and epicanthic folds.

several operations due to other medical conditions such as umbilical hernia, dental diseases, pes planus, and scoliosis. In addition, he was on digoxin treatment from the neonatal period until the age of seven due to cardiomegaly and was under regular follow-up through transthoracic echocardiography (TTE) until three years ago (Figure 1).

The physical examination findings were non-specific, except 3/6 diastolic murmur. In addition, preoperative laboratory test results were normal. Computed tomography (CT) measured the enlargement in the ascending aorta as 6.7 cm, while the arcus and descending aorta were in normal width. Subsequent TTE indicated severe aortic regurgitation and left ventricular global hypokinesis with 50% ejection fraction. Mitral regurgitation was 1-2°. Therefore, the patient was scheduled for the Bentall procedure.

A written informed consent was obtained from the patient. As a cannulation strategy, aortic cannulation was planned through the left common femoral artery and a central two-stage cannula was planned for venous drainage. We accessed mediastinum via median sternotomy (Figure 2). Reaming surgical process was performed as usual. After the excision of the calcified aortic valve and dilated segment of



**Figure 2.** A view of dilated aortic segment and increased cardiac size.

the aorta, a 21 valve-sized composite Bentall graft was inserted. Coronary anastomoses were sutured as in standard Button-Bentall procedure. Total cardiopulmonary circulation time was 129 min and total cross-clamp time was 111 min. Following an uneventful postoperative course, he was extubated in the first day in the intensive care unit and was admitted to ward on the second day.

## DISCUSSION

To the best of our knowledge, the present case was the 14<sup>th</sup> reported patient as of 2001 among the 42 reported patients worldwide until now.<sup>[10]</sup> Our patient's physical appearance and clinical manifestations are consistent with significant phenotypic descriptions of BWS, such as short neck, broad nasal bridge, ptosis, hypertelorism, and epicanthic folds.

Henedy et al.<sup>[11]</sup> published a review in 2010 regarding the phenotypic spectrum of first 20 cases worldwide. According to the results, cardiac anomalies were observed in only six of them, and multiple anomalies were frequent in these cases. In the study of Ramer et al.<sup>[6]</sup> one case presented with a bicuspid aortic valve with mild aortic stenosis and a small patent ductus arteriosus and the other one presented with mitral valve prolapse, mitral regurgitation, tricuspid valve prolapse, and tricuspid regurgitation. Unfortunately, there is a lack of information about cardiac follow-up of such patients in the literature; therefore, we are unable to know how many of them needed surgical approach.

With the help of emerging technologies, as shown by many authors in different studies, defective actin proteins may cause aortic aneurysms and various cardiac diseases.<sup>[12-14]</sup> Baraitser-Winter syndrome is also one of them and aortic aneurysms may occur in these patients.

In conclusion, our case is uniquely the first reported adult patient with Baraitser-Winter syndrome who underwent cardiac surgery with the diagnosis of an aortic aneurysm and aortic regurgitation. We believe that these patients are under the risk of developing aortic and cardiac diseases and, therefore, should be kept in close echocardiographic follow-up.

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