

Mitral regurgitation in a patient with Noonan syndrome: re-do mitral valve repair via mini-thoracotomy

*Noonan sendromlu bir hastada mitral yetmezlik: mini-torakotomi ile
mitral kapağın yeniden tamiri*

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In this article, we present a 38-year-old male case with Noonan syndrome who underwent repair for an atrial septal defect (ASD) at the age of 14. Severe mitral regurgitation, mild tricuspid regurgitation, and residual ASD were detected on echocardiography. As repeated sternotomy was hindered by a severely dilated cardiac chamber with retrosternal adhesion and a suprasternally located aortic arch, we performed a right anterolateral thoracotomy approach with peripheral cannulation. This minimally invasive approach may be an optimal alternative for early recovery in patients with these characteristics.

Keywords: Minimally invasive; mitral regurgitation; Noonan syndrome.

Noonan syndrome (NS) is an autosomal dominant genetic disorder characterized by a number of congenital heart defects. Cardiac defects are the major reason for medical intervention in patients with NS, but mitral valve prolapse also rarely occurs. Herein, we report the case of a patient with NS who underwent a reoperation via minimally invasive cardiac surgery. To our knowledge this is the first report of this kind in the medical literature.

CASE REPORT

A 38-year-old man with a three-month history of severe dyspnea and general edema was referred to our facility for further evaluation of heart failure. He had been diagnosed with a chromosomal abnormality just after birth but had not been seen by a physician for 20 years. He had also undergone spinal surgery for scoliosis at age 14 and cardiac surgery to repair an atrial septal defect (ASD) at age 18.

Bu yazıda, 14 yaşında atriyal septal defekt (ASD) tamiri yapılan Noonan sendromlu 38 yaşında bir erkek olgu sunuldu. Ekokardiyografide şiddetli mitral yetmezlik, hafif triküspid yetmezlik ve rezidüel ASD tespit edildi. Retrosternal adezyona bağlı şiddetli düzeyde dilate olmuş kalp odacığı ve suprasternal yerleşimli aort arkının sternotomi tekrarına engel olması nedeniyle, periferik kanülasyon ile sağ anterolateral torakotomi yaklaşımı uygulandı. Bu minimal invaziv yaklaşım, bu özelliklere sahip hastalarda erken iyileşme için iyi bir alternatif olabilir.

Anahtar sözcükler: Minimal invaziv; mitral yetmezlik; Noonan sendromu.

A physical examination revealed that the patient was shorter than normal (159 cm) and did not weigh much (46 kg). It also showed that he had pectus carinatum, scoliosis, a webbed neck, a low hairline at the nape of the neck, orbital hypertelorism, ptosis, an upturned nose, and low-set ears (Figure 1). In addition, his blood pressure was 110/70 mmHg, and his pulse was irregular. Cardiac auscultation also showed a systolic murmur at the cardiac apex, and electrocardiography (ECG) identified atrial fibrillation (AF) with a controlled ventricular response.

A chest X-ray detected severe cardiomegaly and thoracolumbar scoliosis, and preoperative echocardiography found a 79 mm left atrium, a severely dilated mitral annulus, mild A2 prolapse with severe (+4) mitral regurgitation, mild tricuspid regurgitation with resting pulmonary hypertension (systolic pulmonary artery pressure= 60 mmHg), and a 3.8 mm remnant ASD with a left-to-right



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Figure 1. (a) View of the Noonan syndrome patient showing a typical face with a broad forehead, orbital hypertelorism, ptosis, upturned nose, sparse scalp hair, and low-set ears. (b) Posterior view of the same patient showing the short stature, scoliosis, webbed neck, and low hairline at the nape of the neck. (The patient gave his written consent to publish these images).

shunt (Figure 2). Magnetic resonance imaging (MRI) of the brain revealed diffuse brain atrophy with ventriculomegaly and hypoplasia of both posterior cerebral arteries while enhanced computed tomography (CT) showed a superficially located and

cranially displaced left brachiocephalic vein and right brachiocephalic artery (Figure 3). A repeated sternotomy was hindered by the severely dilated heart chambers with retrosternal adhesion, an abnormally located aortic arch, and an unrepaired pericardium

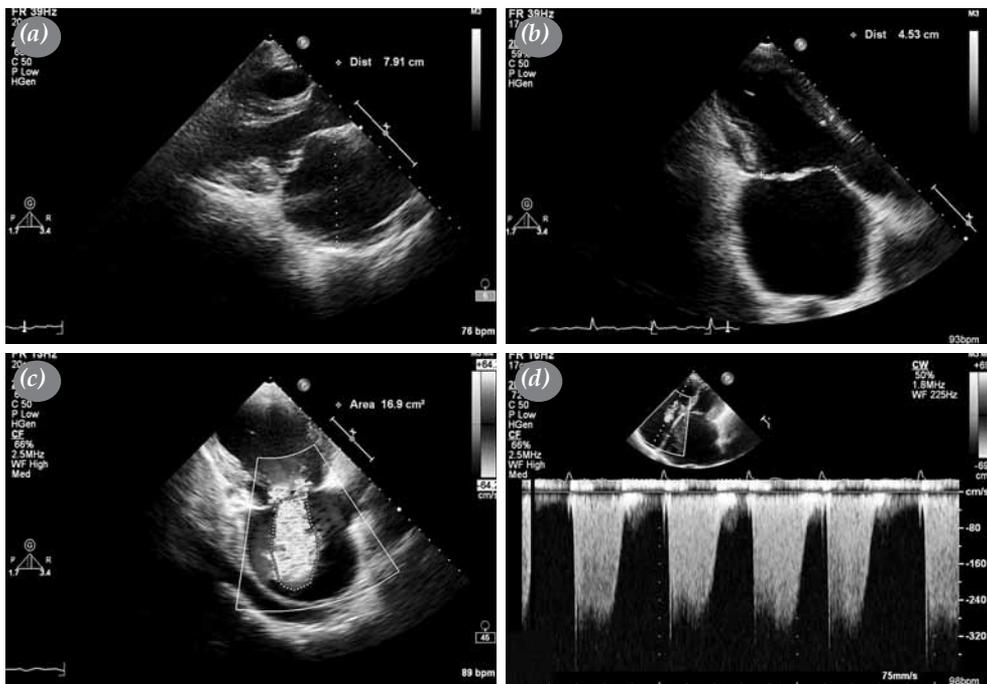


Figure 2. (a) Preoperative echocardiography showing the enlarged left atrium, (b) the severely dilated mitral annulus, (c) severe mitral regurgitation, and (d) mild tricuspid regurgitation with resting pulmonary hypertension.



Figure 3. (a) Enhanced computed tomography showing the superficially and cranially located left brachiocephalic vein (arrow) and right brachiocephalic artery (arrow head) along with (b) the severely dilated cardiac chambers with broad retrosternal adhesion.

from previous surgery. Therefore, we used a right anterolateral thoracotomy approach to perform a mitral valve repair and tricuspid annuloplasty as well as a biatrial cryomaze procedure and the primary repair of the ASD.

The patient was placed right side up in a supine position at 30 degrees, and a 6 cm anterolateral thoracotomy incision was made along the right fourth intercostal space. Because the patient was relatively

small and there was a mild adhesion in the right pleural cavity, all of the procedures were done under direct visualization without thoracoscopic assistance. The right internal jugular vein was cannulated percutaneously, and the right femoral artery and vein were cannulated via a 2 cm skin incision. Intrapericardial dissection around the right atrium was feasible without cardiopulmonary bypass (CPB), but bypass was initiated to facilitate the dissection

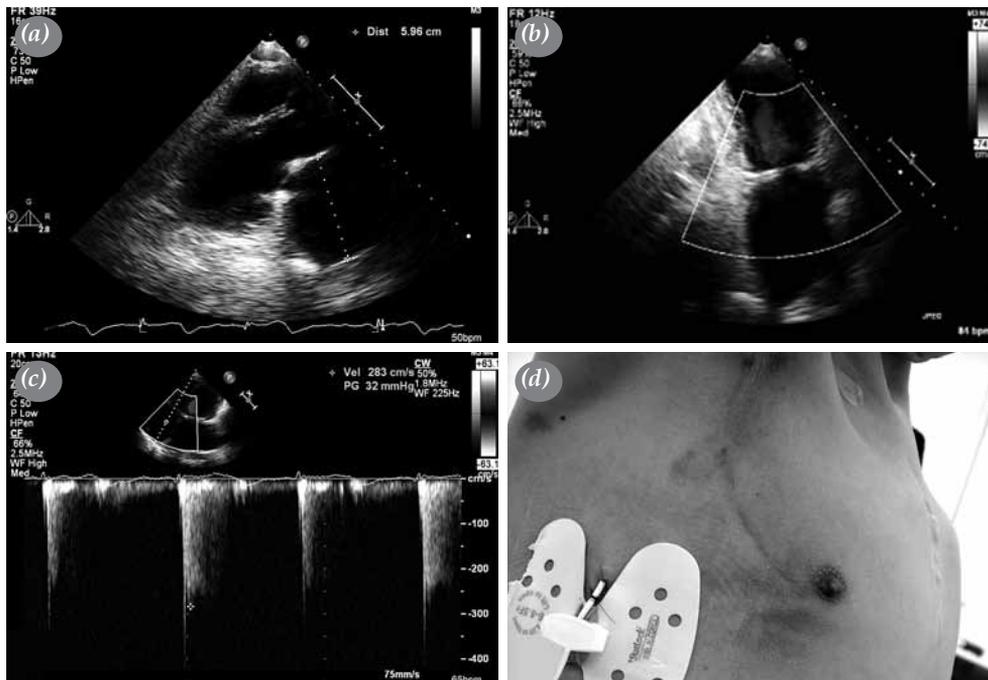


Figure 4. (a) Postoperative echocardiogram showing the decreased diameter of the left atrium, (b) the competent mitral valve, (c) improved pulmonary hypertension, and (d) the postoperative surgical wound.

of a periarotic adhesion. After transthoracic aortic cross-clamping with a Chitwood clamp (Scanlan International Inc., St. Paul, MN, USA) via the third intercostal space and antegrade cardioplegic arrest, the mitral valve was exposed via a left atrial incision along the interatrial groove. The interatrial septum was then retracted anteriorly with a Chitwood hand-held atrial septal retractor (Scanlan International, Inc., Saint Paul, Minnesota, USA) to facilitate the approach to the subvalvular apparatus of the mitral valve. The mitral repair consisted of two 5-0 Gore-Tex sutures (W. L. Gore & Associates, Inc., Flagstaff, AZ) for the new chordae formation of the A2 portion and restrictive annuloplasty with a 30 mm Physio® ring (Edward Life Science, Irvine, CA, USA). The tricuspid valve was repaired using a bicuspidization technique with 2-0 pledgeted polyester sutures, and a 4-0 polypropylene suture was used for direct closure of the ASD. A concomitant biatrial cryomaze procedure was performed with an argon-based cryocatheter system (Cryocath™; Medtronic, Montreal, Quebec, Canada). To facilitate de-airing, intrathoracic carbon dioxide insufflations were applied with a 16GA angiocatheter near the operation window, and intraoperative transesophageal echocardiography was applied to confirm the de-airing process and the positive surgical results. The CPB and aortic clamping times were 138 and 78 minutes, respectively.

Postoperative mechanical ventilation was needed for 18 hours, and the patient recovered in the intensive care unit (ICU) for 48 hours. Echocardiography on the sixth postoperative day showed no residual mitral or tricuspid regurgitation or ASD shunt as well as improved pulmonary hypertension (Figure 4). The patient was then discharged on the seventh postoperative day with normal sinus rhythm and no surgical complications. Because there was no breakdown in the patient's bony thorax, he was able to begin a rehabilitation program two weeks after the surgery, and at the 12-month follow-up, he had maintained Class I functional capacity.

DISCUSSION

Noonan syndrome was first described by Noonan in 1968, who called it "hypertelorism with Turner phenotype".^[1] It is a genetic disorder with an estimated prevalence rate of one in 2,000 births.^[2] Typical characteristics include an abnormally short stature, heart defects, mild mental retardation, and dysmorphic facial features such as a broad forehead, orbital hypertelorism, and a webbed neck. Bleeding diathesis, cryptorchidism, ectodermal anomalies, and lymphatic

dysplasias are also relatively common features of this syndrome.^[3]

Cardiac defects are the major reason for the need for medical attention in patients with NS, and 50% have heart defects, with the most frequent being pulmonary stenosis due to dysplastic valvular tissue.^[4] Atrial septal defects and hypertrophic cardiomyopathy are also commonly reported cardiac problems in NS patients, but mitral valve prolapse is a rare occurrence. In addition, it has been suggested that there may be a pathogenetic link between NS and the partial atrioventricular canal with left ventricular outflow obstruction due to mitral valve anomalies.^[5]

The etiology of mitral regurgitation in patients with NS is not well known. One case study showed severe mitral regurgitation, left ventricle dilation, and severe left ventricle dysfunction in a 21-year-old NS patient, and an intraoperative biopsy of this patient's left ventricle myocardium identified dilated cardiomyopathy.^[6] In our case, the main pathology of mitral regurgitation was annular dilatation and mild prolapse of the middle portion of the anterior mitral valve leaflet. Therefore, we performed a complete ring annuloplasty to prevent further dilation of the mitral annulus and the recurrence of mitral regurgitation.

Recently, a right thoracotomy approach has been suggested as a useful alternative for patients requiring a mitral valve procedure after a previous cardiac operation.^[7] However, in our case, because the repeated sternotomy was impeded by the severe adhesion of the right ventricle to the sternum and an abnormally located aortic arch, we performed a right anterolateral thoracotomy to minimize the risk of injury to the heart chamber close to the sternum. Since 2005, we have used this approach as our procedure of choice in patients requiring mitral valve surgery, and a review of the literature revealed no other cases of a patient with NS undergoing a reoperation via minimally invasive cardiac surgery.

In conclusion, mitral regurgitation can develop in a patient with NS as a consequence of left ventricle dilation. In NS patients with a severely dilated cardiac chamber and an abnormally located aortic arch, the right anterolateral thoracotomy approach can provide a valuable alternative for the cardiac reoperation.

Declaration of conflicting interests

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