CASE REPORT

UDC: 616.12-089
DOI: 10.2298/VSP1506557M

Transcatheter closure of atrial septal defect in a patient with Noonan syndrome after corrective surgery

Transkatetersko zatvaranje atrijalnog septalnog defekta kod bolesnice sa Nunanovim sindromom nakon hirurškog zatvaranja defekta

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Abstract

Introduction. Transcatheter atrial septal defect (ASD) closure is considered to be a gold standard for patients with the suitable anatomy as compared to cardiac surgery. Reocurrence of ASD after surgical closure is a very rare late complication which can be successfully managed with transcatheter procedure. **Case report.** We reported a female patient with Noonan syndrome who presented with hemodinamically significant ASD 37 years after the corrective cardiac surgery. Due to numerous comorbidities which included severe kyphoscoliosis, pectus excavatum and multiple surgeries we decided to perform transcatheter closure of ASD. The procedure itself was very challenging due to the patient's short stature and heart's orientation in the chest, but was performed successfully. The subsequent follow-up was uneventful and the patient reported improvement in the symptoms. **Conclusion.** Transcatheter closure of ASD in a patient with Noonan syndrome with the history of surgically corrected ASD can be performed successfully, despite challenging chest anatomy.

Key words:
noonan syndrome; heart septal defects; congenital abnormalities; cardiovascular surgical procedures; treatment outcome.

Apstrakt

Uvod. Transkatetersko zatvaranje atrijalnog septalnog defekta (ASD) smatra se zlatnim standardom kod bolesnika sa pogodnom anatomijom za ovaj pristup, u poređenju sa hirurškim zatvaranjem ovog defekta. Ponovna pojava ASD nakon hirurške korekcije defekta je retka komplikacija koja se uspešno može rešiti transkateterskom intervencijom. **Prikaz bolesnika.** Prikazali smo bolesnicu sa Nunanovim sindromom i hemodinamski značajnim ASD 37 godina nakon hirurške korekcije. S obzirom na brojne komorbiditete koji su uključivali tešku kifoskoliozu, *pectus excavatum,* kao i brojne hirurške intervencije, odlučili smo da izvedemo transkatetersko zatvaranje ASD. Sama procedura bila je veoma zahtevna zbog komorbiditeta, niskog rasta i poremećene orijentacije srca u grudnom košu, ali je uspešno izvedena. Period oporavka prošao je uredno, a bolesnica je navela i značajno poboljšanje prvobitnih tegoba. **Zaključak.** Transkatetersko zatvaranje ASD kod bolesnice sa Nunanovim sindromom može se uspešno izvesti, uprkos poremećenim anatomskim odnosima u grudnom košu.

Ključne reči:
nunanov sindrom; srce, atrijumski septumski defekti; anomalije; hirurgija, kardiovaskularna, procedure; lečenje, ishod.

Introduction

Noonan syndrome is an autosomal, dominant, variably expressed, multisystem disorder with an estimated prevalence of 1 in 1000–2500 newborns [1]. This syndrome occurs in both genders and is associated with the normal karyotype (46XX or 46XY) with identified missense mutations in the protein tyrosine phosphatase non-receptor type 11 gene (PTPN11), located on chromosome 12 [2, 3]. Characteristic findings include distinctive facial features, short stature, chest deformity and congenital heart disease [4]. It is the second most common syndromic cause of congenital heart disease, exceeded in the prevalence only by trisomy 21. The most common cardiovascular phenotypes of Noonan syndrome include pulmonary stenosis, hypertrophic cardiomyopathy and *secundum* atrial septal defect (ASD) [1].

Transcatheter closure of *secundum* ASD is a safe procedure with a high success rate and low morbidity as compared to cardiac surgery [5]. However, the outcomes of these procedures in patients with previous surgical closure of ASD, with reoccurrence of the same defect are unknown. We presented a patient with surgically corrected congenital heart

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disease and transcatheter closure of ASD, 37 years after the surgery.

Case report

A 39-year-old female with Noonan syndrome was referred to our Center because of the shortness of breath and New York Heart Association (NYHA) Functional Class 2, accompanied by palpitations. Her body height was 139 cm, and body weight 35 kg. Her medical history included surgically closed *secundum* ASD (direct suture with Tycron 4.0) and resection of the right ventricular infundibulum at the age of two, and percutaneous dilatation of pulmonary valve stenosis at the age of 16, multiple surgeries of her right foot because of congenital malformations, breast surgery for the fibroadenoma, kyphoscoliosis and pectus excavatum. Chest X-ray showed enlarged cardiac silhouette with severe kyphoscoliosis. Transthoracic echocardiogram (TTE) and subsequent transesophageal echocardiogram (TEE) revealed the preserved left and right ventricular systolic function, normal dimensions of the left ventricle and the atrium, severe pulmonary valve regurgitation, hypoplastic right ventricular outflow tract, dilated main pulmonary artery and its branches, as well as ASD with calculated Qp/Qs of 2 : 1. Injection of contrast into the cubital vein resulted in bubbles passing into the left atrium. Interestingly, cardiac magnetic resonance (CMR) imaging did not show any communication between the two atria (Figures 1 and 2).

The patient was presented to the international heart team for grown up congenital heart diseases and the decision to perform transcatheter closure of ASD was made, due to numerous comorbidities. It was estimated that the surgical risk of reoperation was unacceptably high for this patient, with a huge range of possible complications that could occur.

Right femoral venous access was obtained. A multipurpose catheter with a stiff guidewire passed through ASD and was placed into the left upper pulmonary vein. A lot of difficulties occurred in terms of heart`s orientation in the chest as the heart was rotated and the position of the interatrial septum was shifted towards the right side of the patient's chest, almost parallel to the diaphragm. Sizing was made with 25/40 mm balloon, which showed a defect of 10 mm in diameter, with continuous fluoroscopic and 3D TEE imaging. TEE probe passing through the esophagus was very demanding because of the almost 90 degrees bend in the proximal third, but was successful ultimately. Size 15 Figulla® Flex ASD (Occlutech®, Jena, Germany) occluder was delivered *via* 12 French introducer sheath with excellent stability and

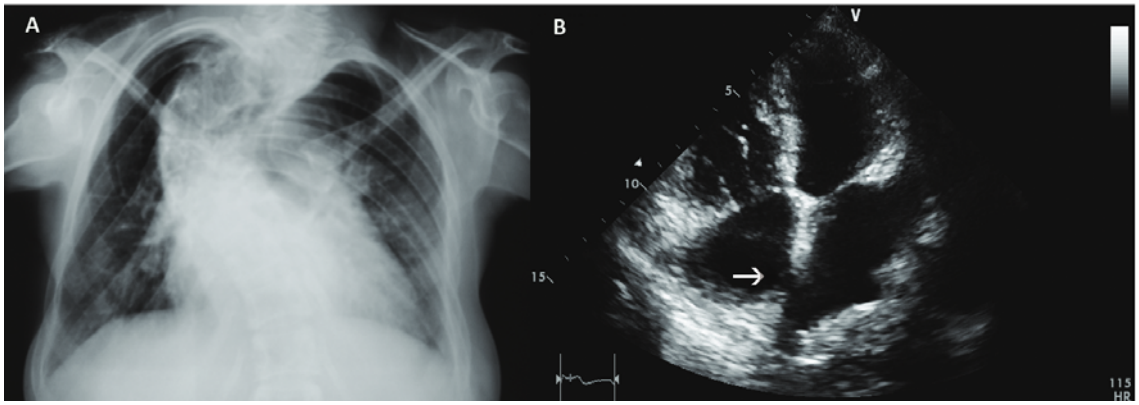

Fig. 1 – A) Chest x-ray showing the enlarged cardiac silhouette and severe kyphoscoliosis; B) Transthoracic echocardiogram showing atrial septal defect (arrow).

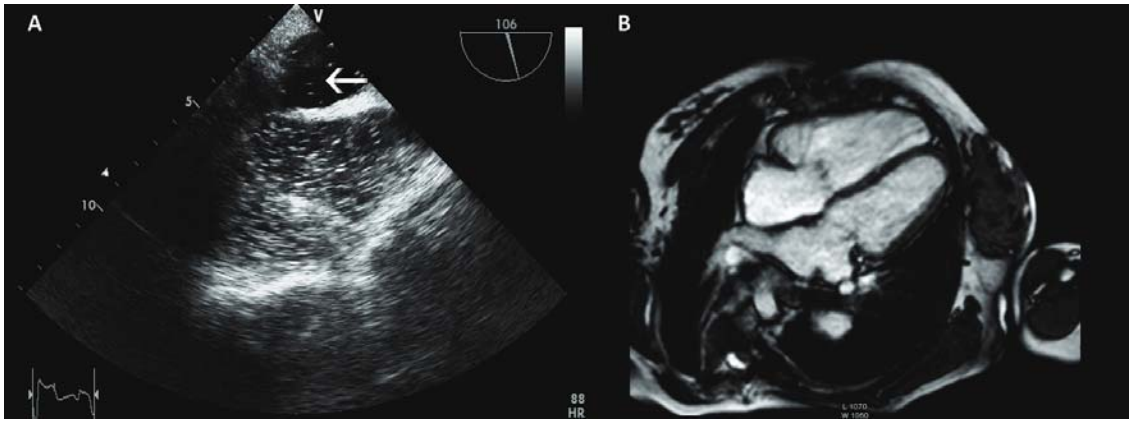

Fig. 2 – A) Transesophageal echocardiogram showing passage of the contrast from the right to the left atrium (arrow); B) Cardiac magnetic resonance imaging showing no visible communication between the two atria.

with exclusion of any shunt at the level of interatrial septum (Figure 3).

A 24 h-postprocedure TTE exam showed no shunt at the level of interatrial septum and the patient was discharged two days after the procedure in good condition, without complications and on dual antiplatelet therapy. One- and six-month clinical follow-ups were done and the patient reported significant improvement in her NYHA class from 2 to 1. TTE examination confirmed good apposition of the device without the evidence of residual shunt (Figure 4).

later with hemodinamically significant ASD, with calculated Qp/Qs of 2 : 1. Decision was made to perform transcatheter closure of ASD because of the patient's numerous comorbidities. The procedure itself was very demanding in terms of patient's short stature and low body weight as well as her kyphoscoliosis which was disturbing normal anatomy of the chest. Occluder device was successfully delivered, and complete closure of ASD was achieved, with excellent positioning and stability.

The follow-up period of 6 months was uneventful and the patient reported significant improvement in her symptoms.

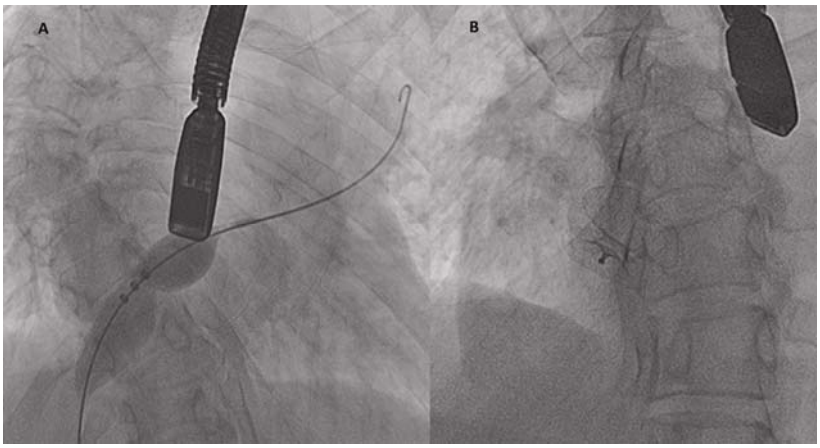

Fig. 3 – A) Fluoroscopy imaging showing the sizing of atrial septal defect with 25/40 mm balloon; B) Fluoroscopy imaging demonstrating atrial septal defect occluder positioned in the atrial septum.

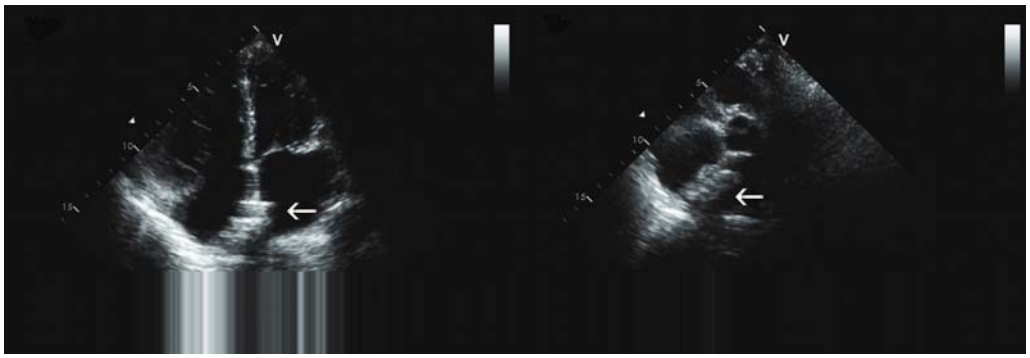

Fig. 4 – Transthoracic echocardiogram showing the good aposition of atrial septal defect occluder device on 6-month follow-up (arrows).

Discussion

Noonan syndrome is an autosomal dominant multisystem disorder which also affects the cardiovascular system. The most common cardiovascular phenotypes include pulmonary stenosis, hypertrophic cardiomyopathy and *secundum* ASD [1,4]. Some patients will require surgery or transcatheter procedures early in the childhood including surgery of the pulmonary valve, closure of ASD or resection of hypertrophied myocardium [6]. Reoccurrence of the defect after surgical closure of ASD is a rare complication occurring in less than 1% in the late postoperative period [7].

In this case report we presented a female patient diagnosed with Noonan syndrome in her early age, who underwent surgical closure of ASD and resection of the hypertrophied right ventricular infundibulum at the age of two. She presented 37 years

Conclusion

Transcatheter atrial septal defect closure in a patient with Noonan syndrome with the history of surgically corrected this defect can be performed successfully, despite the challenging chest anatomy. Although rare, significant atrial septal defect reoccurrence may potentially have deleterious effects on hemodynamics if not treated in a timely manner. Transcatheter closure of such defects could represent a reasonable treatment option in these patients.

Acknowledgements

The authors would like to thank Dr. Tamara Ilisić from the University Children`s Hospital, Belgrade, Serbia, for CMR images.

R E F E R E N C E S

1. *Roberts AE, Allanson JE, Tartaglia M, Gelb BD*. Noonan syndrome. Lancet 2013; 381(9863): 333−42.
2. *Opitz JM*. The Noonan syndrome. Am J Med Genet 1985; 21(3): 515−8.
3. *Tartaglia M, Mehler EL, Goldberg R, Zampino G, Brunner HG, Kremer H*, et al. Mutations in PTPN11, encodes the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. Nat Genet 2001; 29(4): 465−8.
4. *Romano AA, Allanson JE, Dahlgren J, Gelb BD, Hall B, Pierpont ME,* et al. Noonan syndrome: clinical features, diagnosis, and management guidelines. Pediatrics 2010; 126(4): 746−59.
5. *Cowley CG, Lloyd TR, Bove EL, Gaffney D, Dietrich M, Rocchini AP*. Comparison of results of closure of secundum atrial septal defect by surgery versus Amplatzer septal occluder. Am J Cardiol 2001; 88(5): 589−91.
6. *Webb GD, Smallhorn JF, Therrien J, Redington AN*. Congenital heart disease. In: *Bonow RO, Mann DL, Zipes DP, Libby P*, editors. Braunwald's Heart Disease: A Textbook of Cardiovascular Medicine. Philadelphia, Pa: Saunders Elsevier; 2011. chap 65. Available from: http://www.nlm.nih.gov/medlineplus/ency/article/001114.htm
7. *Murphy JG, Gersh BJ, McGoon MD, Mair DD, Porter CJ, Ilstrup DM,* et al. Long-term outcome after surgical repair of isolated atrial septal defect. Follow-up at 27 to 32 years. N Engl J Med 1990; 323(24): 1645−50.

Received on January 15, 2014.
Revised on March 3, 2014.
Accepted on March 5, 2014.