Ebsteinova anomalija – prikaz slučajeva

Ebstein's Anomaly: A Case Report

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SAŽETAK: Ebsteinova anomalija rijetka je prirođena srčana greška trikuspidnog zalistka i desne klijetke koja se, ovisno o anatomskoj varijanti i stupnju težine trikuspidne regurgitacije, prezentira u različitoj životnoj dobi i različitim simptomima. Ovdje smo prikazali slučajeve dvoje bolesnika u kojih je Ebsteinova anomalija otkrivena u odrasloj dobi, kada su se prezentirali simptomima srčanog zatajivanja. Ehokardiografski je u obama slučajevima verificiran apikalno pomaknut trikuspidni zalistak uz tešku regurgitaciju i znatno dilatiranu desnu stranu srca. S obzirom na to da su unatoč medikamentnoj terapiji bili simptomatski, učinjena je kirurška korekcija trikuspidnog zalistka s implantacijama bioloških proteza. U kontrolama šest mjeseci nakon zahvata, u obama slučajevima, prati se znatno poboljšanje funkcionalnoga statusa.

SUMMARY: Ebstein's anomaly is a rare congenital heart defect of the tricuspid valve and the right ventricle that presents at different ages and with different symptoms, depending on the anatomical variant and the severity of the tricuspid regurgitation. We report two cases of Ebstein's anomaly discovered at an adult age when the patients presented with symptoms of heart failure. In both cases, echocardiography verified apical displacement of the tricuspid valve, severe tricuspid valve regurgitation, and significant dilatation of the right side of the heart. Since the patients remained symptomatic despite medication therapy, surgical correction of the tricuspid valve with bioprosthetic valve implantation was performed. At follow-up after 6 months, both patients showed significantly improved functional status.

KLJUČNE RIJEČI: Ebsteinova anomalija, prirođene srčane greške.

KEYWORDS: Ebstein's anomaly, congenital heart defects.

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Ebsteinova je anomalija malformacija trikuspidnog zalistka i desne klijetke. Postoje različite varijante greške koje uključuju: 1. poremećaj delaminacije trikuspidnog zalistka; 2. apikalni i stražnji pomak funkcionalnoga trikuspidnog prstena (septalni > stražnji > prednji); 3. proširenje „atrijaliziranog" dijela desne klijetke s različitim stupnjem hipertrofije i stanjenja same stijenke; 4. fenestracije, obilatost i napinjanje prednjeg kuspisa; 5. dilatacija pravoga atrio-ventrikulskog spoja (pravog trikuspidnog prstena).

Incidencija je približno 1 : 200 000 živorođene djece i čini manje od 1 % svih prirođenih srčanih grešaka. Često su uz nju pridružene druge srčane greške kao npr. otvoreni foramen ovale, atrijski septalni defekt (ASD), ventrikulski septalni defekt, jedan ili više akcesornih provodnih putova.

Ebstein's anomaly is a malformation of the tricuspid valve and the right ventricle. There is a wide variation of abnormalities that include: 1) failure of delamination of the tricuspid valve; 2) apical and backward displacement of the functional tricuspid junction (septal>back>forward); 3) dilation of the "atrialized" part of the right ventricle with different levels of hypertrophy and thinning of the wall; 4) fenestration, redundancy, and tension of the anterior cusp; 5) dilation of the true atrioventricular junction.

The incidence of this disease is about 1: 200,000 newborns and makes for less than 1% of all congenital heart disease. It often appears in conjunction with other heart defects such as for instance open foramen ovale, atrial septal de-

Klinička slika ovisi o stupnju apikalnog pomaka i funkciji trikuspidnog zalistka, a najčešći simptomi u adolescenata i odraslih jesu umor, slabo podnošenje fizičkog napora, dispneja u naporu, cijanoza, palpitacije i paroksizmalne atrijske aritmije.

Auskultacijski su često prisutni široko cijepani I. i II. ton, pokatkad uz čujne tonove punjenja klijetke (S3 i S4) te sistolički šum trikuspidne regurgitacije.

U 12-kanalnom elektrokardiogramu često je prisutan inkompletni blok desne grane, pokatkad preekscitacija te supraventrikulska tahikardija i fibrilacija atrija.

Radiološki, veličina sjene srca varira od gotovo normalne do za Ebsteinovu anomaliju tipične znatno uvećane, loptaste, zbog proširenja desne pretklijetke.

Ehokardiografija omogućuje točnu procjenu trikuspidnog zalistka i subvalvularnog aparata (stupanj apikalnog pomaka, displazije, nedostatka kuspisa), veličinu desnog atrija i atrijaliziranog dijela desne klijetke te funkciju desne i lijeve klijetke. Glavno ehokardiografsko obilježje u odnosu prema drugim poremećajima trikuspidnog zalistka jest apikalni pomak septalnog kuspisa > 0.8 cm/m^2 ukupne površine tijela.

U današnje vrijeme sve se češće, dodatno za procjenu veličine i funkcije klijetki, primjenjuje magnetna rezonancija.

Kateterizacija uglavnom nije potrebna jer je tlak u plućnoj arteriji obično normalan.

Medikamentna terapija (diuretici, ACE inhibitori, antiaritmici) i perkutane intervencije (ablacija aritmija, zatvaranje ASD-a) dovode do poboljšanja, ali nakon pojave simptoma, smanjenja funkcionalnog kapaciteta (NYHA III i IV) i/ili progresije proširenja desne klijetke kardiokirurška operacija popravka ili zamjene zalistka pruža jedinu šansu za poboljšanje.

Ako je moguće, reparacija zalistka metoda je izbora. U slučaju nepogodne anatomije i/ili starije životne dobi, dobra je opcija zamjena zalistka biološkom protezom. U istom je aktu potrebno korigirati pridružene srčane anomalije te eventualno provesti antiaritmijske postupke (MAZE), plikaciju atrijaliziranog dijela desne klijetke i redukcijsku plastiku desne pretklijetke. Transplantacija srca rijetko je indicirana, obično u bolesnika sa značajnim biventrikulskim zatajivanjem.

Postoperativna je prognoza dobra i više od 90 % bolesnika živi dulje od 10 godina. Potrebne su redovite godišnje kontrole zbog mogućnosti rezidualne trikuspidne regurgitacije, pojavljivanja srčanih aritmija, kao i znakova srčanog popuštanja.[1,2]

Ebsteinova se anomalija u odrasloj dobi uglavnom liječi konzervativno medikamentnom terapijom (ACE inhibitori, diuretici, antiaritmici). Međutim, u slučajevima teške insuficijencije trikuspidnog zalistka i znatne dilatacije desne klijetke kliničko stanje često nije zadovoljavajuće unatoč konzervativnoj terapiji. U takvim slučajevima kirurška korekcija (reparacija ili zamjena zalistka protezom) može dovesti do značajnog kliničkog poboljšanja. Navedeno se potvrdilo u slučajevima svih petero bolesnika u kojih je tijekom 2015. godine u našoj ustanovi obavljena kirurška korekcija Ebsteinove anomalije s implantacijama bioloških proteza. Slijedi prikaz dvaju slučajeva.

fect (ASD), ventricular septal defect, or one or more accessory conduction pathways.

The clinical picture depends on the amount of apical displacement and the function of the tricuspid valve, and the most common symptoms in adolescents and adults are fatigue, poor exertion tolerance, dyspnea on exertion, cyanosis, palpitations, and paroxysmal atrial arrhythmia.

Auscultation often shows a wide splitting of S1 and S2 sound, sometimes with audible ventricle-filling sounds (S3 and S4) as well as the systolic murmur of tricuspid regurgitation.

Using a 12-channel electrocardiogram often shows an incomplete right bundle branch block and occasionally preexcitation as well as supraventricular tachycardia and atrial fibrillation.

Radiologically, the heart image can vary between nearly normal to the image typical for Ebstein's anomaly of an enlarged ball-like heart due to the dilation of the right atrium.

Echocardiography allows precise assessment of the tricuspid valve and the subvalvular apparatus (the amount of apical displacement, dysplasia, cusp absence), the size of the right atrium and the atrialized part of the right ventricle, and the function of the right and left ventricles. The main echocardiographic characteristic in comparison with other diseases of the tricuspid valve is the apical displacement of the septal cusp >0.8 cm/m^2 of total body surface area.

Today, magnetic resonance imaging is being used increasingly commonly in the assessment of the size and function of the ventricles.

Catheterization is generally not necessary since the pressure in the pulmonary artery is usually normal.

Medication (diuretics, angiotensin-converting-enzyme (ACE) inhibitors, antiarrhythmic agents) and percutaneous interventions (cardiac ablation, ASD closure) lead to improvement, but once symptoms have appeared – reduction in functional capacity (New York Heart Association (NYHA) class III or IV) and/or progression of the dilation of the right ventricle – surgical repair or replacement of the valve is the only chance of improvement.

If possible, valve repair is the method of choice. In case of unsuitable anatomy and/or advanced age, a bioprosthetic replacement is a good option. The same procedure must also repair associated heart anomalies and possibly include antiarrhythmic procedures (MAZE), plication of atrialized part of the right ventricle, or reduction of the right atrium. Heart transplants are indicated only rarely, usually in patients with significant biventricular failure.

Postoperative prognosis is good, and more than 90% of patients survive for more than 10 years. Regular annual follow-up is needed because of possible residual tricuspid regurgitation, appearance of arrhythmia, and symptoms of heart failure.[1,2]

Ebstein's anomaly in adult patients is usually treated conservatively using medication (ACE inhibitors, diuretics, antiarrhythmic agents). However, in cases of severe tricuspid valve insufficiency and significant dilation of the right ventricle, the clinical picture does not improve sufficiently under

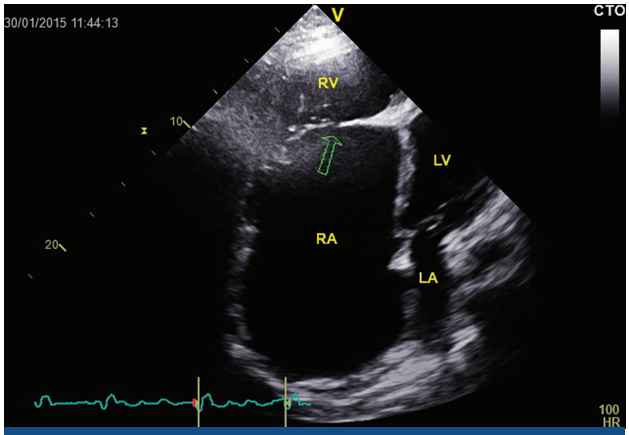

FIGURE 1. Apical four chamber view – dilated right ventricle with apical displacement of the functional tricuspid annulus (arrow), small functional right ventricle and large dilated "atrializied" portion of the right ventricle.

RA = right atrium; RV = right ventricle; LA = left atrium; LV = left ventricle.

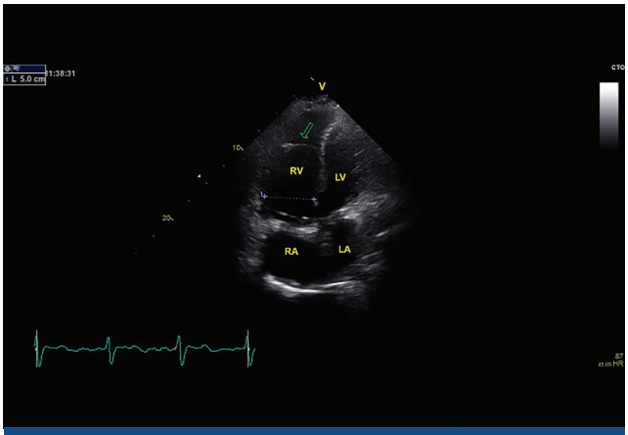

FIGURE 2. Apical four chamber view – bioprosthetic tricuspid valve placed in position of anatomic tricuspid annulus, lesser degree of right ventricle dilatation; arrow shows native tricuspid valve.

RA = right atrium; RV = right ventricle; LA = left atrium; LV = left ventricle.

Slučaj 1.

Bolesnik u dobi od 40 godina liječen je zbog hipotireoze. U dvama navratima, 2000. i 2002. godine, bio je liječen zbog kardijalne dekompenzacije te brze forme fibrilacije atrija. Pri drugoj hospitalizaciji obavljen je ehokardiografski pregled, pri čemu je postavljena dijagnoza Ebsteinove anomalije. Bolesnik je uz medikamentnu terapiju bio bez većih smetnji i nije se javljao na kardiološke kontrole do listopada 2014. godine. Tada su ehokardiografski verificirane znatno dilatirana desna klijetka i pretklijetka te umjerena trikuspidna regurgitacija.

U siječnju 2015. godine bolesnik je bio hospitaliziran radi dodatne kardiološke obrade. Liječen je bisoprololom 2,5 mg, furosemidom 40 mg, levotiroksinom 75 mcg i varfarinom. Bio je kardijalno kompenziran, NYHA II-III stupnja funkcionalnog statusa, dobro regulirane frekvencije permanentne fibrilacije atrija. U 24-satnom kontinuiranom elektrokardiogramu frekvencija srca kretala se od 40 do 150/min, a prosječna je iznosila 75/min.

Ehokardiografski (**slika 1**) je verificirana abnormalna trikuspidna valvula s izrazito apikalno pomaknutim septalnim kuspisom te veliki prednji kuspis s oblikom „jedra" vezan na pretpostavljenom mjestu trikuspidalnog anulusa. Posljedično je bila prisutna atrijalizacija desne klijetke koja je malog kavuma te teška trikuspidna regurgitacija s volumnim opterećenjem desne strane srca. Bez znakova plućne hipertenzije. Lijeva klijetka potisnuta desnom stranom, granične sistoličke funkcije. Bez značajnije druge valvularne greške.

Koronarografijom je isključena koronarna bolest srca. Bolesnik je bio operiran sredinom ožujka 2015. godine. Zbog nepogodne anatomije nije bilo moguće učiniti reparaciju zalistka te mu je implantirana biološka proteza Carpentier Edwards Perimount M 33 mm. Sama operacija i postoperativni tijek protekli su bez komplikacija.

conservative treatment. In these cases surgical correction (repair or replacement of the valve) can lead to significant clinical improvement. This was confirmed in all 5 patients with Ebstein's anomaly that underwent surgical bioprosthetic valve implantation during 2015 in our institution. We report two cases below.

Case 1

This patient was a 40-year-old man, treated for hypothyroidism. He was treated for cardiac decompensation and rapid atrial fibrillation twice, in 2000 and 2002. Echocardiographic imaging was performed during his second hospital stay, and the diagnosis of Ebstein's anomaly was established. The patient did not have significant issues under medication treatment and did not attend cardiologic follow-up until October 2014. It was then that echocardiography verified significant dilation of the right ventricle and atrium and moderate tricuspid regurgitation.

In January 2015, the patient was hospitalized for further cardiologic assessment. He was treated with bisoprolol 2.5 mg, furosemide 40 mg, levothyroxine 75 mcg, and warfarin. The patient had compensated heart failure, was with a NYHA class II-III, and had permanent atrial fibrillation with well-regulated heart rate. Heart frequency was 40-150/min as established by 24-hour continuous electrocardiogram monitoring, with an average of 75/min.

Echocardiographic imaging (**Figure 1**) verified tricuspid valve abnormality with very pronounced apical displacement of the septal cusp and a large anterior "sail-like" cusp attached to the presumed place of the tricuspid ring. Consequently, "atrialization" of the right ventricle had occurred, the cavum was small, and there was severe tricuspid regurgitation with volume overload to the right side of the heart, but with no signs of pulmonary hypertension. The left ventricle was com-

U daljnjim kontrolnim pregledima prati se kliničko poboljšanje. Bolesnik je subjektivno bez tegoba, sada NYHA I. – II. stupnja funkcionalnog statusa. Ehokardiografski (**slika 2**) se prati normalna funkcija biološke trikuspidne proteze sa srednjim dijastoličkim PG od 3 mmHg te sasvim blagom centralnom regurgitacijom. Sama proteza implantirana je nešto bazalnije prema desnom atriju zbog mogućnosti oštećenja AV čvora. Prethodno atrijalizirani dio desne klijetke sada je u funkciji desne klijetke koja je nešto manje dilatirana, ali i dalje teže hipokontraktilne slobodne stijenke. Prema vrijednostima vremena akceleracije nad plućnom valvulom, pacijent je bez znakova plućne hipertenzije.

Slučaj 2.

Bolesnici u dobi od 57 godina 1995. godine postavljena je dijagnoza Ebsteinove anomalije i ASD-a tipa secundum s L-D šantom i protokom kroz plućnu cirkulaciju u odnosu prema sistemnkoj 1,3 : 1. S obzirom na stabilno kliničko stanje, odustalo se od operativnog liječenja.

Subjektivno je bila bez većih tegoba, zbog čega nije bila na kardiološkim kontrolama sve do 2011. godine. Tada je bila hospitalizirana zbog kardijalne dekompenzacije. Uz medikamentnu terapiju nastupilo je kliničko poboljšanje, a s obzirom na znatno dilatiranu desnu stranu srca, zaključeno je da je riječ o krajnjoj fazi Ebsteinove anomalije te da više nije operabilna. Ponovno se ne kontrolira kardiološki. Subjektivno od 2013. godine osjeća kliničko pogoršanje uz sve slabije podnošenje napora. Potkraj 2014. godine provedeno je ergometrijsko testiranje na kojemu je ostvareno opterećenje od 4,3 MET-a, uz subjektivno prisutan izrazit umor i zaduhu. S obzirom na kliničko pogoršanje, bolesnica je upućena u Kliniku radi kardiološke reevaluacije.

Kod prijma je bila kardijalno kompenzirana. Prema anamnestičkim podatcima, do pogoršanja je došlo unatrag dvije godine, a zadnjih mjeseci je NYHA III funkcionalnoga statusa sa zaduhom prisutnom u minimalnom naporu. U terapiji je imala bisoprolol 1,25 mg, digoksin 0,1 mg, furosemid 80 + 40 mg, pantoprazol 20 mg, varfarin. Uz navedenu terapiju slabo su regulirane frekvencije srca u permanentnoj fibrilaciji atrija, a u mirovanju je frekvencija 100/min. Radiološki je verificirana znatno uvećana sjena srca, bez izljeva i akutnih zastojnih promjena. Ehokardiografski (**slika 3**) je nađena izrazito dilatirana desna strana srca (anulus TV 8 cm) s pomakom septalnog kuspisa trikuspidnog zalistka apikalno u odnosu prema mitralnom zalistku za 2 cm i posljedično atrijalizacijom desne klijetke. Prisutna potpuna nekoaptacija kuspisa s masivnom trikuspidnom regurgitacijom. Desna je klijetka dilatirana, reducirane sistoličke funkcije (TAPSE 12 mm). Lijeva je klijetka normalne veličine i funkcije. Nema znakova koarktacije aorte ni na transtorakalnoj ehokardiografiji sigurnih znakova otvorenog formena ovale.

Koronarografijom je isključena koronarna bolest srca, a kateterizacijom srca nađene su normalne vrijednosti tlakova u plućnoj arteriji, bez znakova L-D šanta.

Bolesnica je premještena u Kliniku za kardijalnu kirurgiju. Nekoliko dana poslije implantirana joj je biološka proteza Medtronic Hancock Valve 33 mm na poziciji trikuspidnog anulusa te učinjena redukcijska plastika desne pretklijetke.

pressed by the right side of the heart, with borderline systolic function. There were no other significant valve defects.

The diagnosis of coronary heart disease was elxcluded by coronarography. The patient underwent a surgical procedure mid-March in 2015. Valve repair was not possible due to an unsuitable anatomical configuration, so a bioprosthetic valve was implanted: Carpentier Edwards Perimount M 33 mm. The procedure and recovery was without complications.

Later follow-up examinations indicated clinical improvement. The patient was without subjective issues, now NYHA class I-II. Echocardiography (**Figure 2**) showed normal function of the tricuspid bioprosthesis with a mean diastolic pressure gradient (PG) of 3 mmHg and very mild central regurgitation. The prosthesis itself was implanted in a somewhat more basal location towards the right atrium to avoid damage to the atrioventricular node. The previously atrialized part of the right ventricle had now taken the function of the right ventricle that was somewhat less dilated, but the free wall continued to be hypocontractile. According to the pulmonary acceleration time there were no signs of pulmonary hypertension.

Case 2

A female patient, 57 years of age, was diagnosed in 1995 with Ebstein's anomaly and ASD secundum with a left-to-right shunt and a pulmonary to systemic flow ratio of 1.3:1. Since the patient was stable, no surgical procedure was performed.

The patient was without subjective issues and did not attend cardiologic follow-up until 2011, when she was hospitalized for cardiac decompensation. Medication treatment led to clinical improvement, but significant dilation of the right side of the heart led to the conclusion that this was the final phase of Ebstein's anomaly that was no longer operable. Once more, the patient did not attend cardiologic follow-up. Subjectively, she reported deterioration starting in 2013 with increasingly poor exertion tolerance. In late 2014, ergometric stress testing was performed which demonstrated the load of 4.3 MET, with subjective presence of intensive fatigue and shortness of breath. Due to clinical deterioration, the patient was referred to our Clinic for cardiologic reevaluation.

At admission, she had compensated heart hailure. According to anamnesis, deterioration had happened two years ago, and over the last months the patient had been in the NYHA III functional class with dyspnea even during minimal exertion. She was treated with bisoprolol 1.25 mg, digoxin 0.1 mg, furosemide 80+40 mg, pantoprazole 20 mg, and warfarin. She had permanent atrial fibrillation with poorly regulated heart rate, at rest 100 beats/min. Radiological examination established a significantly enlarged heart shape, without pleural effusion or acute failure changes. Echocardiographic imaging (**Figure 3**) found severe dilation on the right side of the heart (annulus TV 8 cm) with 2 cm apical displacement of the septal cusp of the tricuspid valve in relation to the mitral valve and consequent atrialization of the right ventricle. There was non coaptation of the cusps and massive tricuspid regurgitation. The right ventricle was dilated and the systolic function was reduced (TAPSE 12 mm). The left ventricle was normal in size and function. There were no indications of aortic coarctation,

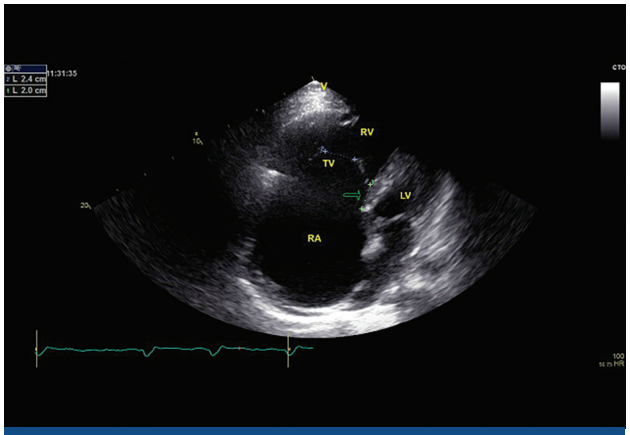

FIGURE 3. Apical four chamber view (modified view of the right ventricle), dilated right ventricle with apical displacement of the functional tricuspid annulus (arrow); failure of tricuspid leaflet coaptation.

RA = right atrium; RV = right ventricle; TV = tricuspid valve; LV = left ventricle.

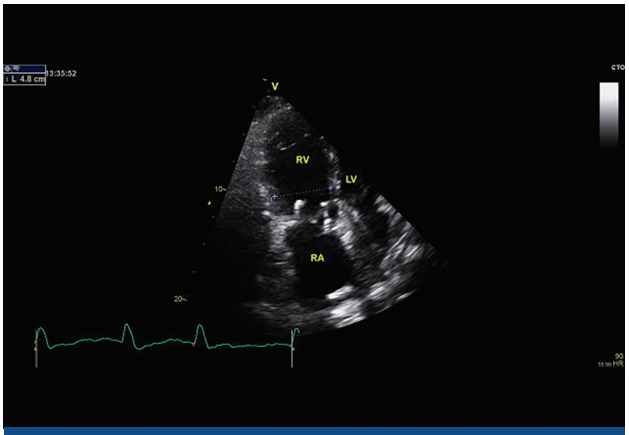

FIGURE 4. Apical four chamber view (modified view of the right ventricle) - bioprosthetic tricuspid valve placed in position of anatomic tricuspid annulus, lesser degree of right ventricle dilatation, reduced size of right atrium.

RA = right atrium; RV = right ventricle; LV = left ventricle.

Sama operacija i postoperativni tijek protekli su bez komplikacija. Do sada je obavljena jedna kontrola. Subjektivno je bolesnica bez tegoba i ograničenja u svakodnevnim aktivnostima. Ehokardiografski (**slika 4**) se prati normalna funkcija biološke trikuspidne proteze uz nešto bolju funkciju dilatirane desne klijetke.

nor did transthoracic electrocardiography find sure signs of an open foramen ovale.

Coronarography was used to exclude coronary heart disease, and coronary catheterization found normal pressure values in the pulmonary artery, with no signs of a left-to-right shunt.

The patient was moved to the Cardiac Surgery Clinic and several days later prosthetic valve replacement was performed with implantation of Medtronic Hancock Valve 33 mm bioprosthesis at the tricuspid annulus position and surgical reduction of the right atrium. One follow-up examination has been performed so far. Subjectively, the patient does not suffer from any issues or limitations in everyday activities. Echocardiography (**Figure 4**) shows normal function of the tricuspid bioprosthesis with somewhat improved function of the dilated right ventricle.

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