Prikaz slučaja rijetkog ehokardiografskog nalaza – kvadrikuspidne aortalne valvule

Quadricuspid aortic valve – a case report of a rare echocardiographic finding

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SAŽETAK: Kvadrikuspidna aortalna valvula iznimno je rijetka pojavnost. Riječ je o kongenitalnoj anomaliji koja se relativno kasno prepozna, otprilike u petom ili šestom desetljeću života. Premda se radi o anatomski drukčije strukturiranoj valvuli, ona ne znači nužno bolest. Međutim, može biti preduvjet razvoja ozbiljne valvularne greške. S obzirom na dugo očuvan funkcionalni status, upitna je točna incidencija. O tome govori i podatak da se najčešće prepozna pri kardiokirurškim zahvatima. Najčešće opisana greška kvadrikuspidne aortalne valvule jest aortalna regurgitacija, a najčešća konkomitantna anomalija jest aneurizma ascendentne aorte. Prikazujemo slučaj 68-godišnjeg bolesnika s paroksizmalnom fibrilacijom atrija kojemu smo, kao slučajan nalaz, ehokardiografijom dokazali kvadrikuspidnu aortalnu valvulu.

SUMMARY: The presence of a quadricuspid aortic valve is extremely rare. It is a congenital anomaly that is recognized relatively late, approximately in the fifth or sixth decade of life. Although it is an anatomically differently structured valve, it does not necessarily represent a disease. However, it can be a prerequisite for the development of a serious valvular defect. Given the long-preserved functional status of the heart, the exact incidence is unclear. This is evidenced by the fact that it is most often recognized during cardiac surgery. The most commonly described quadricuspid aortic valve defect is aortic regurgitation, and the most common concomitant anomaly is an aneurysm of the ascending aorta. We present the case of a 68-year-old patient with paroxysmal atrial fibrillation for whom we demonstrated a quadricuspid aortic valve by echocardiography.

KLJUČNE RIJEČI: kvadrikuspidna aortalna valvula, aortalna regurgitacija, ehokardiografija.

KEYWORDS: quadricuspid aortic valve, aortic regurgitation, echocardiography.

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Uvod

Četverolisna (kvadrikuspidna) aortalna valvula rijetka je prirođena srčana greška, čija je incidencija manja od 0,05 do 1 %. Prvi opisani slučaj datira još iz 1862. godine, a od tada je, prema literaturi, opisano više od 200 slučajeva. Uvelike je pomogao napredak dijagnostičkih aparata premda se najviše novootkrivenih slučajeva i dalje registrira pri kardiokirurškim zahvatima.

Unatoč brojnim hipotezama, embriogenetski razvoj ostao je nepotpuno razjašnjen. Kao mogući mehanizmi razvoja navode se abnormalna proliferacija mezenhima zajedničkog trunkusa i abnormalna fuzija aortikopulmonalnog septuma s u konačnici razvijenim aberantnim endokardijalnim jastučićem. Iako je riječ o kongenitalnoj

Introduction

A four-leafed (quadricuspid) aortic valve is a rare congenital heart defect with an incidence of less than 0.05 to 1%. The first described case dates back to 1862, and, according to the literature, more than 200 cases have been described since then. The advancement of diagnostic devices has greatly facilitated diagnosis, although most newly diagnosed cases are still registered during cardiac surgery. Despite numerous hypotheses, embryogenetic development has remained incompletely elucidated. Possible mechanisms of development include abnormal proliferation of the common trunk mesenchyme and abnormal fusion of the aorticopulmonary septum with the ultimately developed aberrant endocardial pad.

anomaliji, ona se prepozna kasno, tek u petom ili šestom desetljeću života zbog dugo očuvanoga funkcijskog statusa. Zbog širokoga spektra mogućih komplikacija može se manifestirati i ranije.[1,2]

Prikaz bolesnika

U veljači 2021. godine na Odjel kardiologije zaprimili smo 68-godišnjeg bolesnika zbog recidiva paroksizmalne fibrilacije atrija. Nakon prijma bio je tahikardan, ali hemodinamski stabilan i respiratorno suficijentan, bez znakova volumnog opterećenja. Riječ je o bolesniku s dugogodišnjom arterijskom hipertenzijom (više od 20 godina), optimalno reguliranom uz antihipertenzivnu terapiju i suboptimalno reguliranom dislipidemijom. Pri ambulantnim kardiološkim kontrolama posljednjih 20-ak godina prate se vrijednosti LDL kolesterola više od 4 mmol/L uz napomenu o prekidu uzimanja statina u terapiji. Od 2008. godine kontroliran je u urološkoj ambulanti zbog adenoma prostate i obostrane nefrolitijaze, a 2019. godine izvedena je litotripsija konkremenata mokraćnoga mjehura. Drugih čimbenika kardiovaskularnog rizika nije imao. Na Odjelu kardiologije prvi put je bio hospitaliziran deset godina prije, također zbog paroksizmalne fibrilacije atrija. Tada je sinusni ritam postignut medikamentnom konverzijom s propafenonom. Nakon otpusta zabilježeni su vrlo rijetki paroksizmi fibrilacije atrija unatoč tomu što bolesnik nije uzimao preporučenu antiaritmijsku, a ni antikoagulantnu terapiju. Transtorakalnom ehokardiografijom (TTE) registriran je suboptimalni prikaz aortalne valvule sa suspektnim prolapsom desnog i nekoronarnog kuspisa, a Dopplerom se registrirao blagi stupanj aortalne regurgitacije. Korijen aorte mjerio je 40 mm. Globalna sistolička funkcija lijeve klijetke bila je očuvana.

Pri posljednjem prijmu na kardiološki odjel 2021. godine sinusni je ritam postignut farmakološkom kardioverzijom amiodaronom. Kardioverziji je prethodio nalaz TTE-a koji je bio sličan ranijima. Opisana je lijeva klijetka granične veličine (LVIDd 5,5 cm) i koncentrično hipertrofičnih stijenki (1,4 cm), bez segmentalnih ispada kontraktilnosti s očuvanom sistoličkom funkcijom (LVEF prema 2D izračunu oko 70 %), blago dilatiran lijevi atrij i mlaz centralne aortalne regurgitacije (AR 2-3+) sa stacionarno dilatiranim korijenom aorte. Pregledom transezofagijskim ultrazvukom (TEE) isključeno je postojanje tromba u aurikuli lijevog atrija, a iz višestrukih se projekcija utvrdio prikaz aortalne valvule s četiri gotovo identična listića relativno dobro očuvane koaptacije i morfološki blagog zadebljanja na vrškovima (**slika 1, 2** i **3**). Iz duge je osi prikazan centralni mlaz aortalne regurgitacije umjerena stupnja (**slika 4**). S obzirom na veličinu i distribuciju kuspisa, okarakterizirali smo je kao tip A prema Hurwitzovoj & Robertsovoj klasifikaciji. Također su prikazana su normalna ušća koronarnih arterija. Bolesnik je ubrzo otpušten kući dobroga općeg stanja i normofrekventnog sinusnog ritma. Pri daljnjim je kontrolama subjektivno bez tegoba, nema boli u prsima i dobro podnosi tjelesne napore. Ehokardiografski parametri aortalne regurgitacije i dimenzije korijena aorte ostali su nepromijenjeni. Uzimajući u obzir morfološko i funkcionalno stanje aortalne valvule, stupanj insuficijencije te dilatiran korijen aorte, za sada je indicirano daljnje praćenje.

Although it is a congenital anomaly, it is recognized late, only in the fifth or sixth decade of life, given the long-preserved functional status of the heart. Due to the wide range of possible complications, it can manifest even earlier.[1,2]

Case report

In February 2021, we hospitalized a 68-year-old patient due to recurrence of paroxysmal atrial fibrillation at the Department of Cardiology. The patient was tachycardic upon admission, but hemodynamically stable and without respiratory distress, with no signs of volume overload. The patient had a history of long-term arterial hypertension (more than 20 years according to available medical documentation), optimally regulated with antihypertensive therapy and with suboptimal regulated dyslipidemia. In outpatient cardiac controls, LDL cholesterol levels above 4 mmol/L had been monitored for the last 20 years with a note that the patient discontinued statin therapy. Since 2008, he has been in urologic care for prostate adenoma and bilateral nephrolithiasis, and lithotripsy of bladder concrements was performed in 2019. The patient had no other cardiovascular risk factors. He was first hospitalized at the Department of Cardiology ten years earlier, also for paroxysmal atrial fibrillation. At the time, sinus rhythm was achieved by pharmacological conversion with propafenone. After discharge, very rare paroxysms of atrial fibrillation were observed despite the fact that the patient was not taking the recommended antiarrhythmic or anticoagulant therapy. Transthoracic echocardiography (TTE) registered a suboptimal view of the aortic valve with suspected prolapse of the right and noncoronary cusp, and Doppler showed a mild degree of aortic regurgitation. The aortic root measured 40 mm. Global left ventricular systolic function was preserved.

At the last admission to the cardiology department in 2021, sinus rhythm was achieved by pharmacological cardioversion with amiodarone. Cardioversion was preceded by a TTE exam with findings similar to earlier ones. The left ventricle was of borderline size (LVIDd 5.5 cm) with concentrically hypertrophic walls (1.4 cm), well-preserved segmental contractility and with preserved systolic function (LVEF about 70% according to 2D calculation). The left atrium was mildly dilated, and a central aortic regurgitation jet was described (AR 2-3 +) with a stationary dilated aortic root. Examination by transesophageal echocardiography (TEE) ruled out the presence of a thrombus in the left atrial auricle, and multiple projections revealed an aortic valve with four almost identical leaflets of relatively well preserved coaptation and morphologically mild thickening at the tips (**Figure 1, Figure 2**, and **Figure 3**). A central jet of aortic regurgitation of moderate degree was observed from the long axis (**Figure 4**). Given the size and distribution of the cusps, we characterized the quadricuspid aortic valve as type A according to the Hurwitz and Roberts classification. Normal coronary artery orifices were also observed. The patient was soon discharged in good general condition and with a normal sinus rhythm. In further follow-up, he was subjectively without problems, had no chest pain and tolerated physical exertion well. Echocardiographic parameters of aortic regurgitation and aortic root dimensions remained unchanged. Taking into account the morphological and functional state of the aortic valve, the degree of insufficiency and dilated aortic root, further supervision is indicated for now.

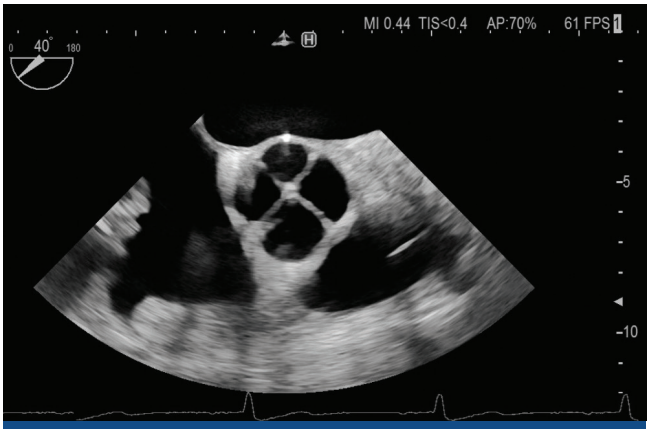

FIGURE 1. Transesophageal echocardiography shows a quadricuspid aortic valve.

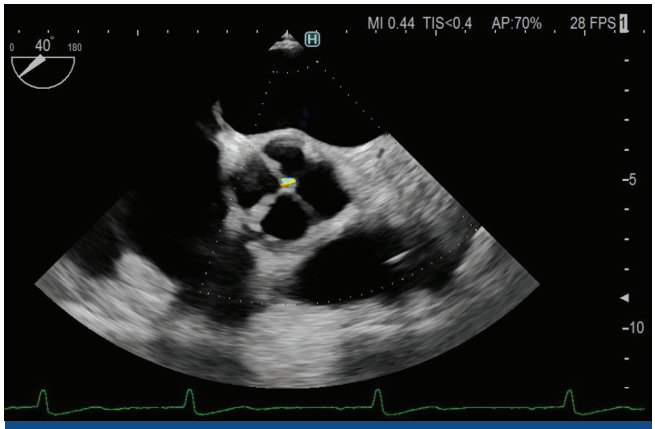

FIGURE 2. Diastolic frame with color Doppler.

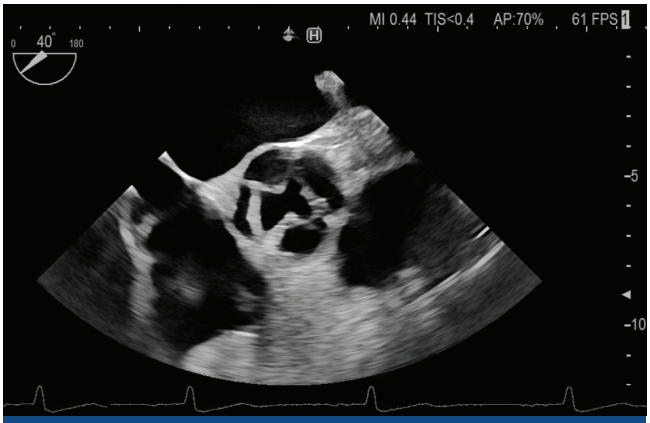

FIGURE 3. Normal systolic opening of all four leaflets.

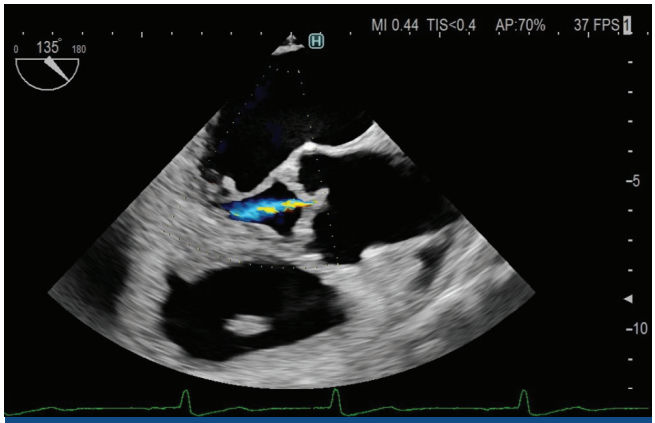

FIGURE 4. Color Doppler examination featuring aortic regurgitation.

Rasprava

Dok Hurwitz i Roberts kvadrikuspidnu valvulu prema veličini i smještaju kuspisa razvrstavaju na 7 tipova (A-G), Nakamura *i sur.* jednostavnije ih kategoriziraju u 4 tipa (I. –IV.), također ovisno o veličini i distribuciji pojedinog kuspisa.[1,3] Prva su dva tipa u objema klasifikacijama jednaka (tip I. i II. odgovaraju tipu A i B), a Tsang *i sur.* kao najčešće dokazali su A (64 %) i B-oblik (32 %) valvule. Predominantna greška ovako anatomski razvijene valvule jest aortalna regurgitacija koja u petine bolesnika progredira do teškoga stupnja, kada je potrebno pristupiti kardiokirurškoj reparaciji ili zamjeni.[4] Retrospektivnim istraživanjem provedenim 2004. godine Tutarel je evidentirao 186 bolesnika s kvadrikuspidnom aortalnom valvulom, od kojih je 74 % imalo aortalnu regurgitaciju, stenozu je imalo 0,7 % bolesnika, a kombiniranu grešku aortalne valvule (regurgitacija sa stenozom) imalo je njih 8,4 %.[5] Ipak, Tsang *i sur.* spominju 8 % slučajeva stenoze u ukupno 50 bolesnika s kvadrikuspidnom aortalnom valvulom. Glavnim krivcem za deterioraciju funkcijskoga statusa ovako anatomski strukturirane valvule smatra se progresivna fibroza listića s po-

Discussion

While Hurwitz and Roberts rank quadricuspid valve defects into 7 types (A-G) according to the size and location of the cusp, Nakamura et al. categorized them more simply into 4 types (I-IV), also depending on the size and distribution of each cusp.[1,3] The first two types in both classifications are equal (types I and II correspond to types A and B), and Tsang et al. have demonstrated that types A (64%) and B (32%) are the most common. The predominant defect of such anatomically developed valves is aortic regurgitation, which progresses to severe regurgitation in one-fifth of patients, which is when cardiac surgery or replacement is required.[4] In a retrospective study conducted in 2004, Tutarel recorded 186 patients with quadricuspid aortic valve, of whom 74% had aortic regurgitation, 0.7% had stenosis, and 8.4% had combined aortic valve regurgitation (regurgitation with stenosis).[5] However, Tsang et al. noted 8% of cases of stenosis in a total of 50 patients with quadricuspid aortic valve. The main culprit for deterioration of the functional status of such an anatomically structured valve is considered to be progressive cusp fibrosis with conse-

sljedično narušenom koaptacijom. Komplementarno fibrozi, ulogu mogu imati i slabo razvijena stijenka listića, miksoidna degeneracija i kalcifikacija listića.

Nije poznato u kojoj je mjeri kvadrikuspidna aortalna valvula podložnija infektivnom zbivanju od trikuspidne ili pak bikuspidne. Postoji nekoliko zabilježenih slučajeva infektivnog endokarditisa kvadrikuspidne aortalne valvule. Dok neki autori predlažu obveznu antibiotsku profilaksu, većina smatra da je antibiotsku profilaksu potrebno ordinirati u slučajevima kada su veličine kuspisa nejednake i postoji jasno dokazano aktivno upalno zbivanje.[1,4]

Važno je napomenuti da je kvadrikuspidna aortalna valvula često izolirana prirođena bolest srca, premda je moguće istodobno postojanje drugih srčanih anomalija (u 18 – 32 % slučajeva). Tsang *i sur.* ehokardiografski su dokazali istodobno dilatiran korijen aorte u 29 % bolesnika, ali je u većine oboljelih bila riječ o blagoj dilataciji. U istraživanju provedenom u Cleveland klinici, od ukupno 19 722 bolesnika koji su pristupili operaciji aortalne valvule u razdoblju od 1989. do 2010. godine, kod 31 (0,0016 %) bolesnika tijekom samog operativnog zahvata vizualizirana je kvadrikuspidna valvula. U 13 od 31 bolesnika (42 %) registrirana je dilatacija ascendentne aorte na >4 cm, a u 10 % bolesnika (3/31) nađeno je anomalno polazište koronarnih ušća.[6] Pregledom literature nađeni su rijetki slučajevi istodobnog postojanja atrijskih i ventrikulskih septalnih defekata, perzistirajući duktus arteriosus, kongenitalna stenoza plućne valvule, ali i hipertrofijska kardiomiopatija.[7]

Tijekom dosadašnjih kardiokirurških zahvata najčešće se pristupalo kompletnoj zamjeni aortalne valvule. Izbor kardiokirurškog liječenja bio je pretežno odabiran intraoperativno primarno zbog uočavanja anomalije tek pri samoj operaciji. Odluka se donosila na temelju ozbiljnosti bolesti valvule, prema inspekcijskom stanju kuspisa i osobnim odabirom operatera. Zamjena aortalne valvule preferirala se dominantno u slučajevima teško deteriorirane kvadrikuspidne aortalne valvule. U 23 % bolesnika izvedena je reparacija valvule u obliku resekcije, što je rezultiralo „trikuspidalizacijom". U ostalih je učinjena konekcija komisura, čime se dobila bikuspidna aortalna valvula, a u jednog bolesnika prijavljena je i procedura po Rossu (zamjena defektne aortalne valvule pulmonalnom).[8] Otprilike petina bolesnika imala je i konkomitantnu reparaciju uzlazne aorte. Nije zabilježen perioperativni mortalitet, ali su zabilježene komplikacije poput neposredne intracerebralne ishemijske atake. Dosadašnja postoperativna praćenja bila su različita trajanja, u razdobljima od tri pa do deset godina tijekom kojeg su evidentirani rijetki slučajevi razvoja umjerene stenoze, endokarditisa bioproteze, i deterioracija reparirane valvule koja je zahtijevala zamjenu.[4,6,8]

Zaključak

Iako kvadrikuspidna aortalna valvula pripada grupi iznimno rijetkih kongenitalnih srčanih defekata, incidencija je u blagom porastu prije svega zahvaljujući boljim dijagnostičkim alatima. Većina bolesnika za njezinu inkompetenciju ipak sazna tek u srednjoj do kasnoj životnoj dobi, a otprilike petina njih zahtijevat će operativni zahvat bilo u smislu reparacije bilo kompletne zamjene valvule. Manji će dio oboljelih biti podvrgnut istodobnoj reparaciji uzlazne aorte.

Zbog suboptimalnoga prikaza s pomoću TTE-a imali smo djelomičan uvid u morfološko i funkcijsko stanje aortalne valquent impaired coaptation. Poorly developed cusp wall, myxoid degeneration, and cusp calcification may also play a role complementary to fibrosis.

It is not known to what extent quadricuspid aortic valves are more susceptible to infectious events than tricuspid or bicuspid valves. There have been several reported cases of infective quadricuspid aortic valve endocarditis. While some authors suggest mandatory antibiotic prophylaxis, most believe that antibiotic prophylaxis should be administered in cases where the size of the cusps is uneven and there is a clearly proven active infection.[1,4]

It is important to note that quadricuspid aortic valves are often an isolated congenital heart defect, although the coexistence of other cardiac anomalies is possible (in 18-32% of cases). Tsang et al. echocardiographically demonstrated concomitant dilated aortic root in 29% of patients, but most had mild dilatation. In a study conducted at the Cleveland Clinic, out a total of 19,722 patients who underwent aortic valve surgery between 1989 and 2010, 31 (0.0016%) had a quadricuspid valve observed during surgery. In 13 of 31 patients (42%) dilatation of the ascending aorta to >4 cm was registered, and an abnormal origin of coronary orifices was found in 10% of patients (3/31).[6] A review of the literature found rare cases of concomitant atrial and ventricular septal defects, persistent ductus arteriosus, congenital pulmonary valve stenosis, as well as hypertrophic cardiomyopathy.[7]

Complete replacement of the aortic valve was most often performed during previous cardiac surgeries. The choice of cardiosurgical treatment was mostly chosen intraoperatively, primarily due to the detection of an anomaly only during the surgery itself. The decision was made based on the severity of the valve disease, according to the condition of the cusp and the personal choice of the surgeon. Aortic valve replacement was predominantly preferred in cases of severely deteriorated quadricuspid aortic valve. In 23% of patients, valve repair was performed in the form of resection, which resulted in "tricuspidalization". In others, a commissure connection was made to obtain a bicuspid aortic valve, and a Ross procedure was reported in one patient (replacement of a defective aortic valve with a pulmonary one).[8] Approximately one-fifth of patients also had concomitant reparation of the ascending aorta. No perioperative mortality has been reported, but complications such as immediate intracerebral ischemic attack have been reported. Previous postoperative follow-ups have been of varying duration, ranging from three to ten years, during which rare cases of moderate stenosis, endocarditis bioprosthesis, and deterioration of the repaired valve requiring replacement have been recorded.[4,6,8]

Conclusion

Although quadricuspid aortic valve belongs to the group of extremely rare congenital heart defects, the incidence is slightly increasing, primarily due to better diagnostic tools. However, most patients discover its incompetence only in middle to late life, and about a fifth of them will require surgery either in terms of reparation or complete valve replacement. A small group will be subjected to simultaneous repair of the ascending aorta. Due to suboptimal imaging by TTE, we had only partial insight into the morphological and functional state of the aortic valve and cardiac structures. The newly established diagnosis using TEE puts in focus more active monitoring of the diseased

vule i srčanih struktura. Novopostavljena dijagnoza primjenom TEE-a stavila je u fokus aktivnije praćenje hemodinamskih performansi promijenjene aortalne valvule. Zaključno, bolesnici s kvadrikuspidnom aortalnom valvulom trebali bi biti podvrgnuti kontinuiranome kardiološkom praćenju radi pravodobnog daljnjeg liječenja. Kontinuirani se nadzor prije svega odnosi na ehokardiografsko praćenje eventualne progresije aortalne greške, praćenje dimenzija aorte i praćenje njihove reperkusije na srčanu hemodinamiku.

valve and its hemodynamic performance. In conclusion, patients with a quadricuspid aortic valve should be subjected to continuous cardiac supervision for timely application of further treatment. Primarily, continuous supervision refers to echocardiographic monitoring of possible progression of aortic regurgitation, monitoring of aortic dimensions, and assessment of their repercussions on cardiac hemodynamics.

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