REVIEW ARTICLE

Congenital unicuspid aortic valve in adults: Minireview and case series

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ABSTRACT

A unicuspid aortic valve (UAV) in adults is a very rare form of aortic valve (AV) malformation. UAV has two distinct subtypes, acommissural UAV and unicommissural, and can be differentiated by anatomical features, imaging modalities, and clinical presentation. With the development of significant AV lesion (s), surgical or transcatheter intervention will be required. The first part is a summarized review of UAV (anatomical features, clinical presentation, diagnostic modalities, and management). In the second part, we present a series of four patients diagnosed with UAV (3 unicommissural and 1 acommissural). The first case underwent balloon aortic valvuloplasty during childhood and surgical AV replacement later, with the progression to severe aortic stenosis (AS). The second case underwent a Ross procedure. The third and fourth cases were asymptomatic with moderate AS and mild-to-moderate AR and were kept on follow-up. In all the cases, transesophageal echocardiography confirmed the diagnosis of UAV with detailed morphological and functional assessment of AV.

Keywords: Aortic stenosis, echocardiography, unicuspid aortic valve

INTRODUCTION

Unicuspid aortic valve (UAV) is a very rare congenital anomaly, first reported in 1958. The true prevalence of UAVs may be underestimated because the data describing the UAV relied on the earlier echocardiographic modalities. Based on these data, the estimated prevalence of UAV is 0.019% in adults undergoing echocardiography[1] and approximately 4%–5% in patients undergoing surgery for aortic stenosis (AS) with a mean age at presentation of 34 ± 10 years.[2] UAV shows male preponderance (male-to-female ratio of 4:1).[3] Until now, hereditary nature or genetic abnormalities have not been identified for UAVs.

ANATOMICAL BACKGROUND

Two anatomical subtypes of UAV have been described: the acommissural type, which is shaped as a pinhole central opening and presents in early childhood with AS, and the unicommissural type, which has different shapes of its eccentric opening, such as slit-shaped, oval, or triangular [Figure 1]. Due to the relatively larger aortic orifice in the unicommissural UAV, patients have stable hemodynamics and a less aggressive course, and they can live up to the fifth decade without any surgical intervention. The acommissural UAV has no lateral attachment to the aorta and thus is known to cause AS in infants and neonates.[3,4] In unicommissural UAV, the location of the commissure between the anatomic noncoronary and left coronary cusp is most common, while the junction of the left and right coronary cusps is less common, and the junction of right coronary and noncoronary cusps is the least common.[5] The

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differences between both subtypes are summarized in Table 1. The fetal development of the aortic valve (AV) occurs through the excavation of three tubercules and sinuses of the valve. Failure of the 3 aortic cusps to separate before birth causes UAV.[2]

AORTIC VALVE LESION AND ASSOCIATED ANOMALIES

The most common lesion with UAV was isolated AS in 41% of the patients, followed by concomitant AS and aortic regurgitation (AR), while isolated AR is uncommon.[3] The calcification of UAV occurs earlier, more accelerated, and more severe than bicuspid or tricuspid AV, which leads to severe AS more rapidly.[6] UAV could also be accompanied by aortopathy, including dilation of the aortic root or ascending aorta (14%), aortic dissection (2%), aortic annulus aneurysm, and coarctation of the aorta (0.4%). Other congenital anomalies can be associated with UAV, including anomalies of coronary artery (0.8%), ventricular septal defect (0.8%), and patent ductus arteriosus.[3]

CLINICAL PRESENTATION

Clinical presentations of UAVs are also quite different with nonspecific symptoms. In general, it is a bimodal presentation with either AS in infants and neonates with acommissural UAV or it can present as mixed AS and AR in the third to fifth decade of life with unicommissural type.[7] The most common symptoms of UAV in adults include dyspnea (41%), angina (17%), dizziness, and syncope (11%), while in children, heart failure and failure to thrive are always present.[2,8] The prominent auscultatory finding is the presence of a cardiac murmur at the aortic areas due to AS and/or AR.

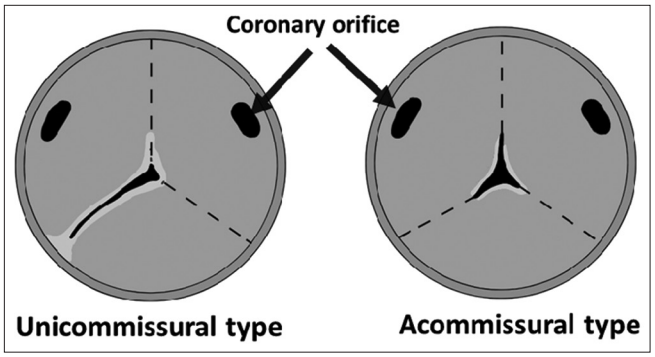

Figure 1: Illustrated drawing of the morphology of the 2 subtypes of unicuspid aortic valve

DIAGNOSIS

The accurate morphological and functional assessment of AV in congenitally malformed AV is important for selecting treatment.[9] The diagnosis of UAV remains a challenge to the extent that intraoperative surgical inspection and echocardiography had 52% sensitivity and 51% specificity compared with pathologic evaluation.[10]

Echocardiography

Echocardiography is the first imaging tool used to diagnose UAVs. Ewen *et al.* defined the major echocardiographic criteria to diagnose UAV as follows: single commissural attachment zone, rounded, leaflet-free edge on the opposite side of the commissural attachment zone, eccentric valvular orifice during systole, and patient age <20 years and mean transvalvular gradient >15 mmHg. The minor criteria were defined as an associated thoracic aortopathy and age <40 years. Three of the four major or two major and one minor criterion are adequate for the diagnosis, with a high sensitivity (94.7%–100%) and specificity (98.1%–100%).[11]

Two-dimensional transthoracic echocardiography (2D-TTE) has a low sensitivity of 27% and a specificity of 50% due to the difficulty and inaccuracies in identifying UAV, especially in adults due to raphes or leaflet calcifications. Transesophageal echocardiography (TEE) overcomes these limitations with a higher sensitivity of 75% and a specificity of 86%.[12] Visualization of AV in short axis view has to be obtained during systole to avoid misinterpretation of raphes or leaflet calcifications as a true commissure.[13] In addition to the morphological assessment, echocardiography can be used to grade the lesion severity of AS and/or AR and to identify other associated congenital heart disease. Both three-dimensional transthoracic echocardiography (3D-TTE) and 3D-TEE can aid in a comprehensive assessment of UAV morphology due to their ability to scan the AV from different angles with multiple cut sections. Visualization of AV from both

Table 1: Main differences between subtypes of unicuspid aortic valve

Character	Acommissural UAV	Unicommissural UAV
Orifice shape	Pinhole shaped	Teardrop-shaped, linear exclamation mark-shaped, triangular, or oval-shaped
Orifice position	Central	Eccentric
Effective orifice area	Small	Relatively larger
Time of presentation	Earlier in childhood	Adulthood
Lateral attachment at or above the level of the coronary ostium	0	1
Number of commissures	No	One lateral commissural attachment to the aorta
Raphes	No	May have at most two such raphes

UAV: Unicuspid aortic valve

aortic and ventricular aspects using 3D echocardiography helps to define the relationship of the commissures to the coronary ostium, in addition to an accurate assessment of UAV thickening and calcification.[14] Computerized multi-slice tomography and magnetic resonance imaging may be required for additional information, e.g. quantification of AV calcification score, identification of aortopathy, and diagnosis of associated coronary anomalies.[15,16]

MANAGEMENT

Different therapeutic strategies are described, and an appropriate modality is chosen based on the patient's age, aortoannular anatomy, and associated cardiac conditions.[17]

Surgical management

The main surgical techniques for UAV in adults were AV replacement in 82%, bicuspidization in 10%, and Ross procedure in 1%.[3,17] Surgical valvotomy or commissurotomy is usually performed in pediatric groups with acommissural UAV.[18] Concomitant aortic surgery, such as replacement of aortic annulus/aortic root/ascending aorta and repair of the aortic coarctation, was performed in 23% of UAV cases.[19] In young patients with AR caused by UAV and root aneurysm, AV bicuspidization and root remodeling can be applied with satisfactory hemodynamic results.[20]

Catheter-based intervention

In children presenting with severe AS, balloon valvuloplasty is usually the primary choice as a palliative procedure. The procedure is possibly associated with residual or recurrent valve dysfunction and re-intervention (due to residual AS and/or severe AR).[21] Transcatheter AV implantation in UAV patients was reported in two cases.[22,23]

CASE SERIES

Case 1

A 39-year-old male referred to the cardiology clinic underwent balloon aortic valvuloplasty at the age of 10 years. He was presented to another hospital 7 months earlier with progressive shortness of breath associated with dizziness and fatigue. There was no palpitation, chest pain, dyspnea, or syncopal attack. Clinically, the patient was alert, conscious, oriented, and not in distress with normal vital signs (pulse: 103 bpm, blood pressure: 110/72 mmHg, and oxygen saturation: 98% on room air). He was admitted with a definitive diagnosis of infective endocarditis and received a complete course (6 weeks) of antibiotics. He was referred to our center in a stable condition for further evaluation. Cardiac auscultation showed an ejection systolic murmur at the aortic area radiating to the left carotid. The electrocardiogram (ECG) showed normal sinus rhythm with no ST or T wave changes. Blood culture, biochemical, and hematological laboratory results were normal. 2D-TTE showed normal left ventricular (LV) size (end-diastolic volume: 125 mL/end-systolic volume 50 mL) and systolic function with no regional wall motion abnormalities. In both parasternal long and short-axis views, AV was thickened and calcified with an eccentric

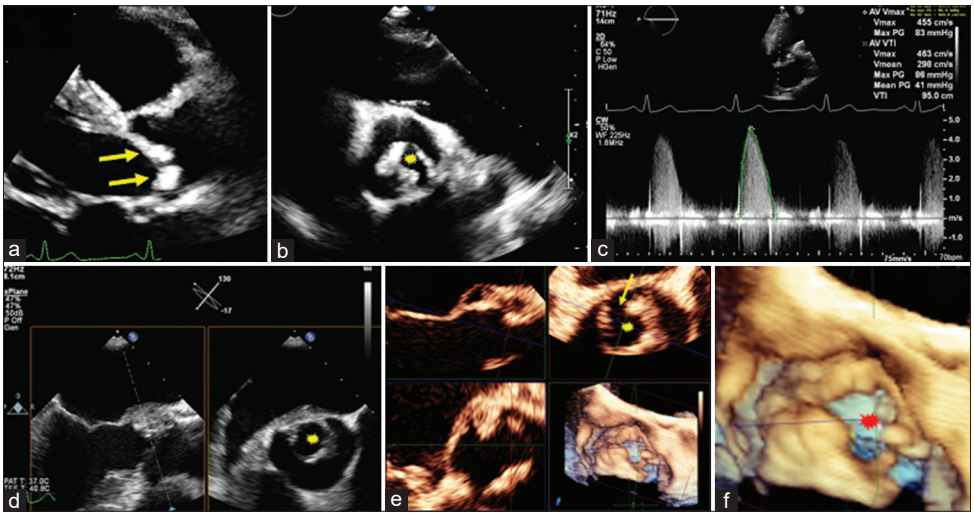

Figure 2: Echocardiographic images for the first patient, (a) Parasternal long axis view using transthoracic echocardiography (TTE) showing thickened and calcified aortic valve (AV) (yellow arrows), (b) Parasternal short axis view showing unicommissural type of unicuspid aortic valve (UAV) with an eccentric small orifice (yellow star), (c) Continuous wave Doppler across the AV obtained from right parasternal view showing high maximum and mean pressure gradient, (d) X-plane visualization of the AV showing eccentric AV orifice (yellow star) using three-dimensional TEE (3D-TEE), (e) Multiplanar reconstruction of the 3D-TEE image showing the AV orifice (yellow star) and commissure (yellow arrow) and (f) zoomed view for the AV orifice (red star). VTI: Velocity time integral, PG: Pressure gradient, AV: aortic valve

orifice, but the leaflets and commissures could not be identified [Figure 2a and b]. Continuous-wave Doppler indicated significant AS (maximal pressure gradient: 86 mmHg, mean pressure gradient: 41 mmHg, AV area: 0.9 cm^2) [Figure 2c]. Three-dimensional TEE confirmed the diagnosis of congenital UAV (unicommissural type) with eccentric orifice. X-plane view clarified the location of commissure between the anatomical left and noncoronary cusps [Figure 2d]. Multiplanar reconstruction of the 3D-TEE image identified the thickened UAV and its orifice and commissure [Figure 2e and f]. The aortic root abscess at the mitral aortic junction, visualized by X-plane at AV level, measured 9 mm × 3 mm [Figure 3a] communicating to the LV outflow tract with color flow [Figure 3b and c]. Color Doppler showed mild- to-moderate AR through the commissure [Movie 1- short axis view of AV with color-Doppler showing the mosaic color of AR jet through the commissure]. The patient was decided for mechanical AV replacement, and the surgical inspection during cardiac surgery confirmed the diagnosis of UAV. The postoperative hospital course was uneventful, and the patient was discharged for regular cardiology clinic follow-up.

Case 2

An 18-year-old male known to have sickle cell disease presented with a history of witnessed recurrent syncopal attacks for a year. Each syncopal attack lasts a few seconds and is associated with mild shortness of breath. There was no history of chest pain, palpation, or dyspnea. Clinical examination was unremarkable except for high blood pressure (147/90 mmHg). Cardiac auscultation revealed an ejection systolic murmur at the aortic area radiating to the carotid. Resting ECG, chest X-ray, and biochemical and hematological laboratory results were normal. A 2D-TTE showed normal LV size and systolic function. Mildly thickened AV with systolic doming, high-pressure gradient (maximum: 50 mmHg and mean: 25 mmHg), and no AR. The morphology of AV cusps and commissures was not clearly defined. A TEE clarified the acommissural type of UAV with a circular and central orifice [Figure 4a and b] with accelerated forward flow [Figure 4c] and significant AS (AV area: 0.7 cm^2 and mean gradient 56 mmHg) and trace AR. The patient underwent a modified Ross procedure (replacement of the UAV with pulmonary autograft and the root of the aorta preserving the two coronary buttons). Intraoperative surgical inspection confirmed the diagnosis of UAV. Postoperatively, the patient's symptoms improved and discharged in stable condition.

Case 3

A 16-year-old male, asymptomatic with infrequent follow-up at family medicine due to a history of congenital heart disease. Clinical examination, resting ECG, chest X-ray, and laboratory investigations were normal. A 2D-TTE showed normal LV wall thickness, size, and systolic function. The AV was markedly thickened with moderate AS (maximum gradient: 46 mmHg and mean gradient: 26 mmHg) and mild-to-moderate AR [Figure 4d]. A 3D-TEE revealed a unicommissural type of UAV with a shaped orifice. The commissure was located between the anatomical left and noncoronary [Figure 4e and f] cusps.

Case 4

A 43-year-old female, asymptomatic, was referred by family medicine for echocardiography due to an ejection

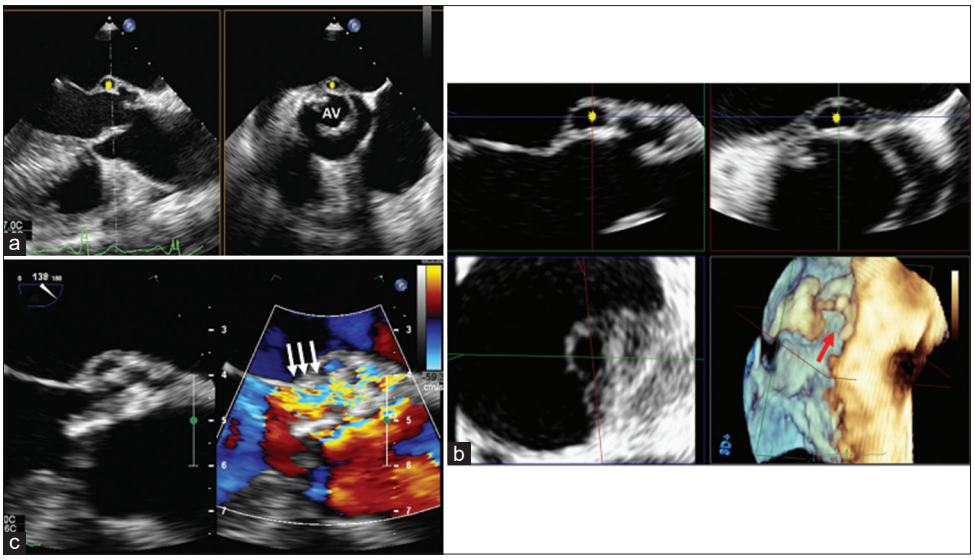

Figure 3: (a) X-plane visualization of the aortic (AO) root abscess (yellow star) with its relation to aortic valve orifice, (b) Multiplanar reconstruction of the three-dimensional transthoracic echocardiography image of aortic root showing an oval-shaped abscess (yellow star) which is communicating with left ventricular (LV) outflow tract (red arrow), and (c) Color flow Doppler showing the flow from the AO root abscess to the LV outflow tract (white arrows). AV: Aortic valve

systolic murmur. Baseline hemodynamics, resting ECG, chest X-ray, and laboratory investigations were normal. A 2D-TTE showed normal LV wall thickness, size, and systolic function. The AV orifice was oval and eccentric with a high-pressure gradient (maximum: 40 mmHg and mean: 20 mmHg) and mild AR [Figure 5a]. 2D and 3D TEE showed a unicommissural type of UAV with an oval-shaped orifice and a single commissure between the anatomic left coronary and noncoronary cusps. Raphe was seen between the anatomic right coronary and noncoronary cusps [Figure 5b and c].

DISCUSSION

Congenital UAV is a very rare cardiac anomaly in adults. The clinical presentation and associated AV lesion are variable and related to its anatomical subtypes (unicommissural or acommissural). Our case series included 4 patients (3 unicommissural UAV and 1 acommissural UAV). The clinical presentation, grading of AV lesions, and management are summarized in Table 2. Usually, UAV in adults is discovered accidentally due to the presence of a cardiac murmur with variable degrees

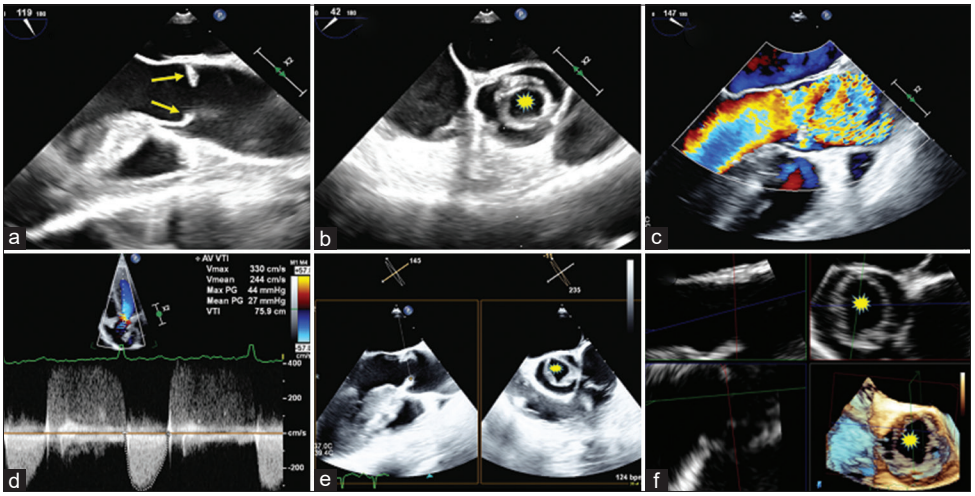

Figure 4: The upper panel has two-dimensional transthoracic echocardiography views for case no. 2, showing, (a) long axis view of the aortic valve (AV) with the doming and restricted opening, (b) Central orifice of unicuspid aortic valve (yellow star), and (c) Accelerated forward color flow indicating aortic stenosis (AS). Lower panel images for case no. 3, (d) Continuous wave Doppler recording of high-pressure gradient across the AV, (e) X-plane visualization of the AV with a small eccentric orifice (yellow star), and (f) Multiplanar reconstruction of the three-dimensional transthoracic echocardiography image showing the AV orifice (yellow star). AV: Aortic valve, VTI: Velocity time integral, PG: Pressure gradient

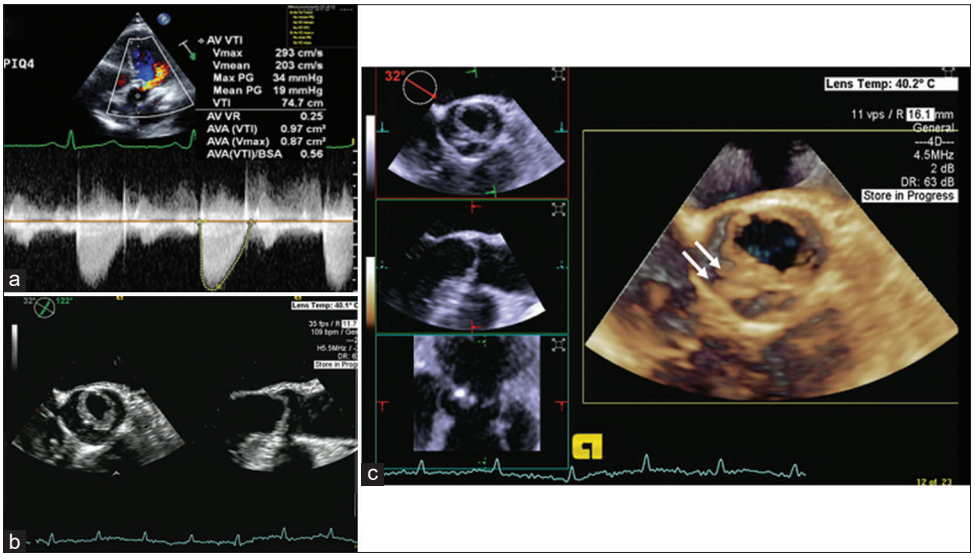

Figure 5: Echocardiographic images for Case no. 4 showing (a) continuous wave Doppler recording of pressure gradient across the aortic valve (AV), (b) X-plane visualization of AV with eccentric orifice and (c) Multiplanar reconstruction of the three-dimensional transthoracic echocardiography image showing the AV orifice with calcified Raphe between the anatomic right coronary and noncoronary cusps (white arrows). AV: Aortic valve, VR: velocity ratio, AVA: Aortic valve area, VTI: Velocity time integral, BSA: Body surface area, PG: Pressure gradient

Table 2: Summary of case series

Case number	UAV type	Age (years)	Clinical presentation	AV lesion	Management
1	Unicommissural	39	Progressive shortness of breath associated with dizziness and fatigue	Severe AS	AV replacement
2	Acommissural	18	Recurrent syncopal attacks and mild shortness of breath	Severe AS	Modified Ross procedure
3	Unicommissural	16	Asymptomatic	Moderate AS, mild to moderate AR	Conservative and follow-up
4	Unicommissural	43	Asymptomatic	Mild AS, mild AR	Conservative and follow-up

AS: Aortic stenosis, AV: Aortic valve, AR: Aortic regurgitation, UAV: Unicuspid AV

of AS and/or AR. It may be associated with nonspecific symptoms, including shortness of breath, angina, dizziness, and syncope.[2] The first case presented with progressive shortness of breath associated with dizziness and fatigue, the second presented with syncope, and the third and fourth were asymptomatic. The diagnostic accuracy of the 2D-TTE is low compared with TEE.[12] In our case series, 2D-TTE helped in the assessment of AV function and hemodynamics, but the detailed valve morphology was defective. In the four cases, TEE, especially 3D, enabled visualization of AV from different angles at multiple levels, which guided the diagnosis of UAV. The findings of TEE were confirmed with the surgical inspection in cases 1 and 2, who underwent AV surgery.

Development of infective endocarditis in patients with UAV is uncommon (11%), as reported in an autopsy study.[7] Our first case developed infective endocarditis complicated with abscess formation with communication to the LV outflow tract. Many factors affect the therapeutic strategy selection, including the patient's age, aortoannular anatomy, and associated cardiac conditions.[17] The first case underwent balloon valvuloplasty in early childhood and mechanical AV replacement 30 years later due to severe AS and mild-to-moderate AR. The second case underwent AV replacement with pulmonary autograft (Ross procedure). The third and fourth cases are still not candidates for surgical intervention.

CONCLUSIONS

Diagnosis of UAVs is still a challenge. A detailed assessment of AV morphology and function is important for identifying them, mainly through 2D and 3D TEE. Management will be planned according to the hemodynamics and function of the UAV, as well as clinical information. Surgical intervention may be necessary for many UAV patients at a young age.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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