

Transcatheter aortic valve implantation in a 13-year-old child with end-stage heart failure: a case report

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Background

Left ventricular non-compaction cardiomyopathy (LVNC) has been reported in association with almost all types of congenital heart valve disease. The presence of LVNC-related ventricular dysfunction increases the perioperative risk in these patients. The advantages of transcatheter treatment modalities outweigh those of surgical strategies, as they avoid cardioplegic arrest and myocardial trauma. To our knowledge, there have been no reports on transcatheter treatment of pure aortic regurgitation in patients with a bicuspid aortic valve (BAV) and concomitant LVNC.

Case summary

In this article, we present the case of a 13-year-old boy with a regurgitant BAV and concomitant LVNC who presented with end-stage heart failure and severe pulmonary hypertension. As a bridge to definitive therapy, the patient underwent an uneventful transcatheter aortic valve implantation (TAVI) using a 26-mm balloon-expandable prosthesis. Device success without paravalvular regurgitation was achieved. At 17 months of follow-up, a steady reduction in pulmonary arterial pressure, persistent normalization of systolic left ventricular function and a tremendous improvement in the patient's physical resilience was observed. The initially considered heart–lung transplantation was avoided and will not be necessary.

Discussion

To the best of our knowledge, this is the first case performed with TAVI for BAV regurgitation in the context of LVNC. With technical modifications and appropriate planning, TAVI in paediatric patients with a non-calcified BAV is feasible. Different imaging modalities revealed an intriguing relationship between aortic regurgitation and morphological signs of a left ventricular non-compaction myocardium.

Keywords

Transcatheter aortic valve implantation or replacement • Aortic regurgitation • Bicuspid aortic valve • Left ventricular non-compaction cardiomyopathy • End-stage heart failure • Heart transplantation • Case report

Introduction

Left ventricular non-compaction cardiomyopathy (LVNC) is considered a rare heterogeneous myocardial disorder characterized

by a thick trabeculated endocardial myocardial layer leading to heart failure. Therapeutic strategies are focused on preserving or improving cardiac efficiency and preventing sudden death, but may result in cardiac transplantation when end-stage heart failure

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Learning points

- Different imaging modalities are necessary to discriminate between valvular and ventricular causes of end-stage heart failure in congenital cardiac disease.
- Congenital bicuspid aortic valve (BAV) disease might mimic morphological signs of a left ventricular non-compaction myocardium.
- With technical modifications and appropriate planning, transcatheter aortic valve implantation in paediatric patients with a non-calcified BAV is feasible.

occurs.¹ The coincidence of LVNC and the congenital malformation of a bicuspid aortic valve (BAV) has been described, but represents a great rarity.² The treatment of BAV disease is defined in current guidelines³ and is based on surgical or catheter-based interventions. Whereas transcatheter aortic valve implantation (TAVI) is an established alternative to surgery in elderly patients with calcified BAV stenosis,⁴ only casuistics on the implementation

of TAVI in children exist with limited device success in pure aortic regurgitation.⁵

This article describes the unusual case of a 13-year-old boy with BAV disease and concomitant LVNC who presented with end-stage heart failure and severe pulmonary hypertension. Instead of the initially considered heart–lung transplantation, definitive therapy was postponed and TAVI performed.

Timeline

| Timeline | Events |
|-----------------|---|
| (Patient's age) | |
| 0 years | Diagnosis of critical bicuspid aortic valve stenosis at birth Balloon aortic valvuloplasty on the day of birth and on the 49th day |
| 11 years | New York Heart Association (NYHA) functional class I Diagnosis of dilated left ventricular non-compaction cardiomyopathy (ratio of non-compacted: compacted layers $\geq 2.0:1$) Mild aortic valve stenosis and mild-to-moderate aortic regurgitation; moderate functional mitral regurgitation Haemodynamic parameters: left ventricular end-diastolic volume index (LVEDVi) 113 mL/m ² , left ventricular end-systolic volume index (LVESVi) 49 mL/m ² , left ventricular stroke volume index (LVSVi) 64 mL/m ² , left ventricular ejection fraction (LVEF) 57%, left ventricular end-diastolic pressure (LVEDP) 27 mmHg, mean pulmonary arterial pressure (mPAP) 30 mmHg |
| 13 years | NYHA class III Recurrent episodes of pneumonia Increase in pulmonary artery pressure and increase in systolic and diastolic left ventricular dysfunction; moderate aortic regurgitation Right ventricular endomyocardial biopsy: persistence of parvovirus B19 genotype 1 without signs of active myocarditis, storage diseases, or dilated cardiomyopathy First course of levosimendan therapy NYHA class III–IV despite optimal medical therapy and after several courses of dobutamine, milrinone, and levosimendan therapy Haemodynamic parameters: LVEDVi 103 mL/m ² , LVESVi 65 mL/m ² , LVSVi 38 mL/m ² , LVEF 37%, LVEDP 25 mmHg, mPAP 47 mmHg Referral to our institution for a second opinion after refusal of aortic valve surgery and heart transplantation and consideration of heart–lung transplantation N-terminal pro-brain natriuretic peptide (NT-proBNP) 2288 pg/mL Transfemoral transcatheter aortic valve implantation—successful procedure |
| 14 years | NYHA class I Normalization of left ventricular systolic function and increase in left ventricular compacted layer thickness (ratio of non-compacted: compacted layers = 1.5:1) NT-proBNP 185 pg/mL Haemodynamic parameters: LVEDVi 111 mL/m ² , LVESVi 53 mL/m ² , LVSVi 58 mL/m ² , LVEF 53%, LVEDP 17 mmHg, mPAP 34 mmHg |
| 15 years | Normalization of the transpulmonary gradient: mPAP 30 mmHg, pulmonary capillary wedge pressure 18–20 mmHg, cardiac index 3.0–3.5 L/min/m ² , Pulmonary vascular resistance 4–5 WU m ² Absence of complications according to the Valve Academic Research Consortium-2 criteria |

Case presentation

A 13-year-old boy was referred to our institution for a second opinion after heart–lung transplantation was being considered due to end-stage heart failure and fixed pulmonary hypertension. He presented with a BAV and pre-diagnosed LVNC. The boy was admitted with progressive fatigue and shortness of breath consistent with New York Heart Association (NYHA) class III–IV.

The physical examination produced the following findings: 13-year-old boy in a good general and nutritional condition (height 159 cm, weight 44.8 kg). No cyanosis, but shortness of breath on light exertion; transcutaneous oxygen saturation 98% at ambient air. Heart rate of about 90 b.p.m., systolic (2/6) and diastolic (1/6) murmur; prominent second heart sound during auscultation. No blood pressure side-to-side differences; 73/42 (51) mmHg. Vesicular breath sound across the lungs. Soft abdomen without signs of hepatosplenomegaly. No neurological abnormalities.

Critical stenosis of the BAV was diagnosed at birth and the initial systemic perfusion was found to be dependent on the patent ductus arteriosus. The patient underwent balloon valvuloplasty on the day of birth and on the 49th day.

At the age of 11, he was diagnosed with left ventricular (LV) dilation in combination with diastolic dysfunction. Transthoracic echocardiography and magnetic resonance imaging (MRI) confirmed mild stenosis and moderate regurgitation of the BAV (aortic valve area 1.8 cm², regurgitant fraction 28–30%). Concomitant cardiomyopathy was suspected and classified as LVNC with a thin layer of compacted LV myocardium measuring 3–5 mm and a two-fold thicker layer of non-compacted myocardium (Figure 1). MRI revealed LV dilation as well as systolic and diastolic dysfunction (Supplementary material online, Slide set). Different imaging modalities (Supplementary material online, Slide set) identified areas of spongy myocardium reaching from the basal parts of the left ventricle to the apex, corresponding to the dilated subtype of LVNC.¹ The ratio of non-compacted and compacted layers (NC/C) was measured as 2.0 at the end of diastole. The non-compacted areas were identified at the ventricular wall reflection points of the diastolic jet stream originating from the regurgitant BAV.

Despite optimal medical therapy, the patient experienced recurrent episodes of pneumonia and cardiac decompensation. Further LV deterioration with haemodynamic compromise and an increase in pulmonary hypertension above 2/3 of the systemic arterial blood pressure were observed. A right ventricular endomyocardial biopsy showed persistence of parvovirus B19 genotype 1 without signs of active myocarditis and absence of inflammation or fibrosis in MRI. Genetic testing was not performed. The electrocardiogram (ECG) showed an incomplete left bundle branch block with a QRS of about 100 ms; cardiac arrhythmias were excluded.

Different therapeutic options were discussed intensively. Primary heart transplantation was rejected because of the risk of right heart failure in the presence of severe pulmonary hypertension. LV assist device implantation was considered as a bridge-to-transplant concept to allow heart transplantation after achieving a marked reduction in pulmonary vascular resistance. This treatment would require simultaneous surgical correction of BAV regurgitation. The possibility of eliminating the aortic regurgitation first and without myocardial ischaemic trauma by performing TAVI was discussed. This concept

was driven by the hope of achieving at least LV unloading and postponing definitive treatment until adulthood. The following concerns were discussed: TAVI is not recommended for BAV without leaflet calcification and is not approved in children.³

In agreement with the parent, the multidisciplinary heart team opted for a TAVI procedure. Computed tomography confirmed a hostile device landing zone with a very eccentric annular shape and no calcifications of the BAV leaflets (Figure 2). A 26-mm SAPIEN-3 prosthesis (Edwards Lifesciences Corp., Irvine, CA, USA) was chosen with the intention to allow for later valve-in-valve implantation if necessary. The following step was added to the otherwise standard transfemoral TAVI procedure (Figure 3): to verify the anticipated anchoring of a 26-mm prosthesis, balloon sizing was performed prior to valve deployment using a 24-mm percutaneous transluminal valvuloplasty catheter. Device success was achieved without a residual leak (Figure 4, Supplementary material online, Slide set) and low transvalvular pressure gradients; dP_{mean} 3.3 mmHg.

The post-procedural course was uneventful according to the Valve Academic Research Consortium-2 criteria.⁶ The patient was discharged on the 4th post-procedural day under optimal heart failure medication (enalapril, carvedilol, and spironolactone) and a 3 months course of phenprocoumon followed by a regimen of aspirin 100 mg daily. In the ECG, the QRS interval persisted at about 100 ms, whereas the spatial QRS-T angle normalized from 80° to 50°. At the 6-month follow-up, the boy reported a drastic improvement in symptoms (NYHA class I). He was able to re-attend school sports without any restrictions. Various clinical and imaging tests in conjunction with computational fluid dynamics⁷ confirmed restoration of haemodynamics and flow profiles, accompanied by complete normalization of systolic LV function, whereas diastolic dysfunction and LV dilation persisted (Figure 4, Table 1, Video 1, Supplementary material online, Slide set). At the 17-month follow-up, further haemodynamic normalisation was observed with a transpulmonary gradient of 10 mmHg.

Discussion

Our case presents a patient in whom the benefits of a transcatheter strategy allowed for myocardial recovery under critical circumstances with the potential advantages of less myocardial trauma by avoiding cardioplegic arrest during valve replacement. All concerns regarding the performance of TAVI in children and in non-calcified BAV were considered. Until today, the isolated elimination of BAV has had a tremendous benefit on the boy's physical resilience.

Regarding the diagnosis of LVNC, we detected the non-compaction layer at the reflection site of the regurgitant jet stream originating from the BAV. During growth and cardiac morphogenesis, these strong regurgitant jets may have provoked the morphological alterations of the LV myocardium that mimicked the pathology of LVNC. In a case series of 109 BAV patients aged between 14 and 56 years, 12 patients met the criteria of concomitant LVNC.² Two of the adult patients underwent surgical aortic valve replacement, following which normalization of the LV ejection fraction was observed in one. In our patient, we observed only a slight reduction in the NC/C ratio but a rapid normalization of the LV systolic function, whereas LV dilation persisted. In this context, it has been described that the physiological embryonic process of myocardial maturation

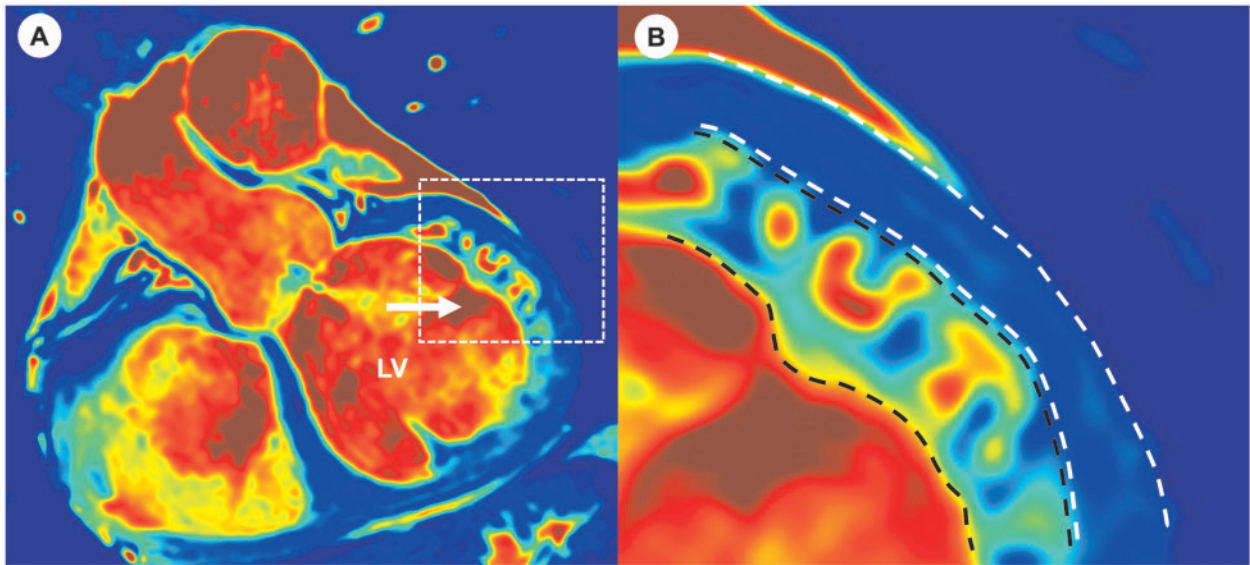


Figure 1 Colourized cardiac magnetic resonance imaging. Coronal view at end-diastole (A): a jet stream (white arrow) originating from the regurgitant bicuspid valve is directed towards the spongy left ventricular wall (dashed box). Enlarged section (B): a thick inner layer of non-compacted myocardium (black dashed lines) can be separated from a thin outer compacted layer (white dashed lines).

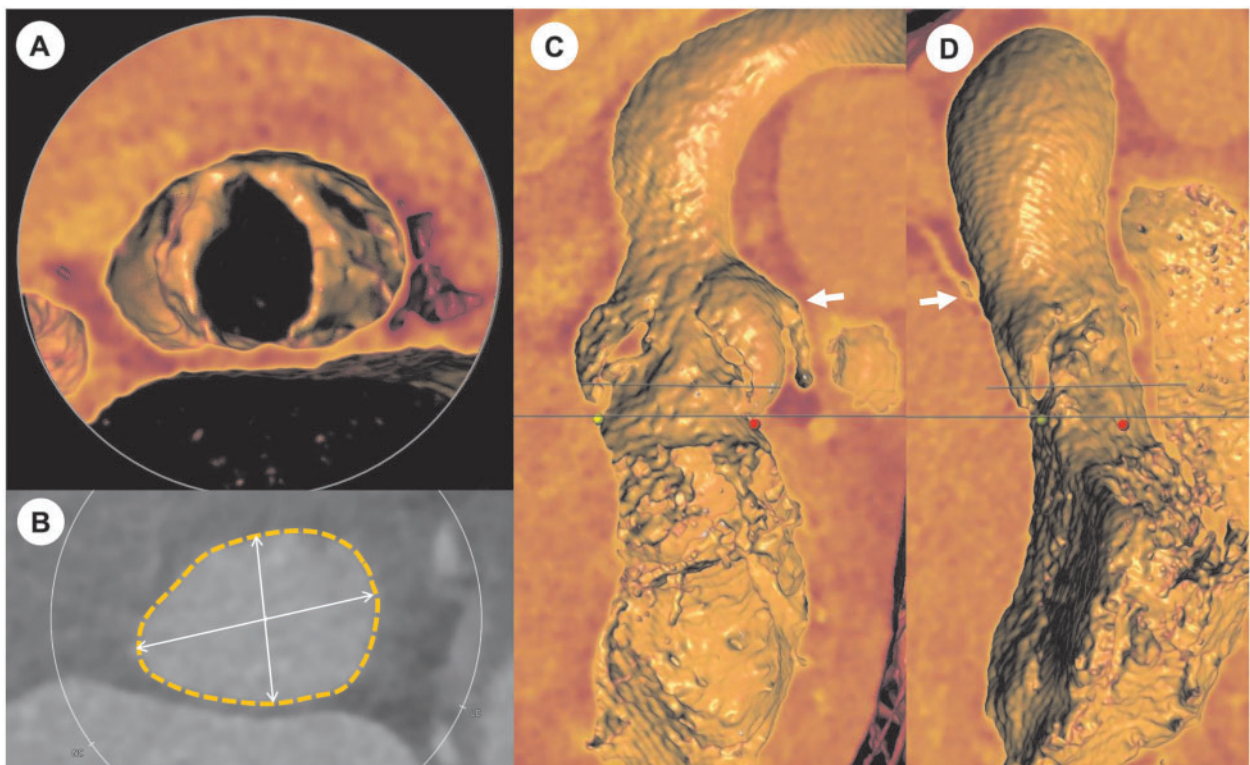


Figure 2 Device landing zone: 3D reconstruction of computed tomography. (A) View on the bicuspid aortic valve; (B) virtual aortic valve annulus: 19.3 × 30.4 mm; (C) oblique coronal view with left coronary artery (white arrow); (D) oblique sagittal view with right coronary artery (white arrow).

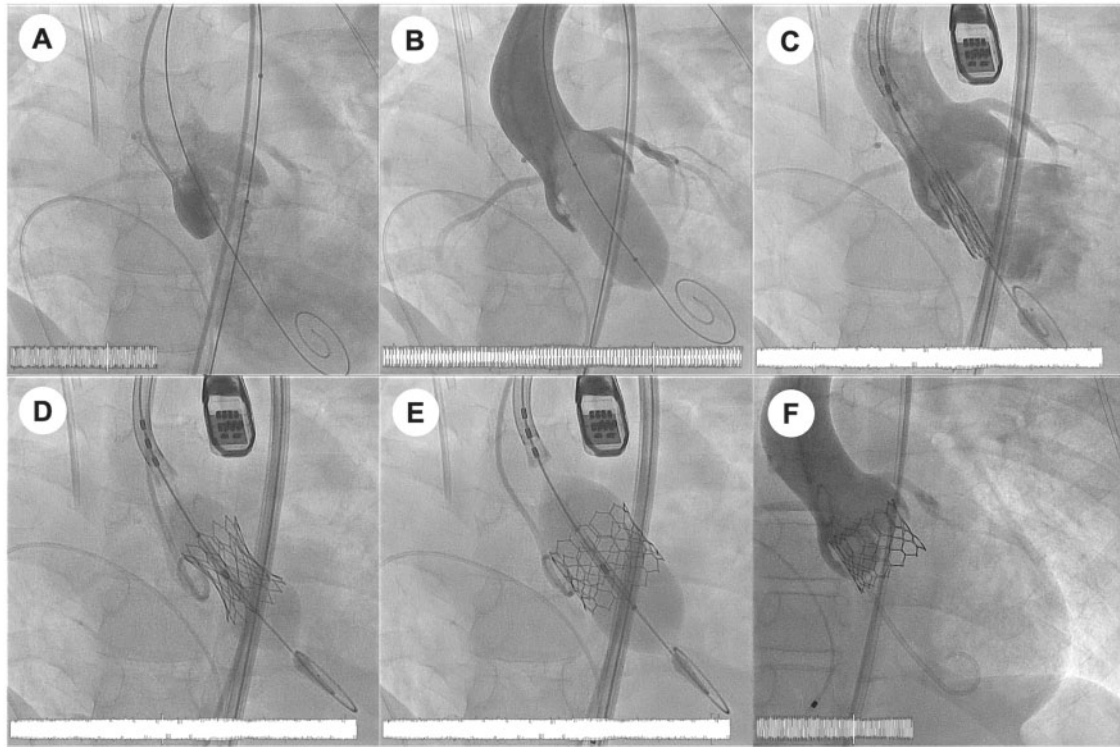


Figure 3 Transcatheter aortic valve implantation. Sequence of transcatheter aortic valve implantation with aortic regurgitation (A), balloon sizing (B), stepwise valve deployment (C–E), and final result (F).

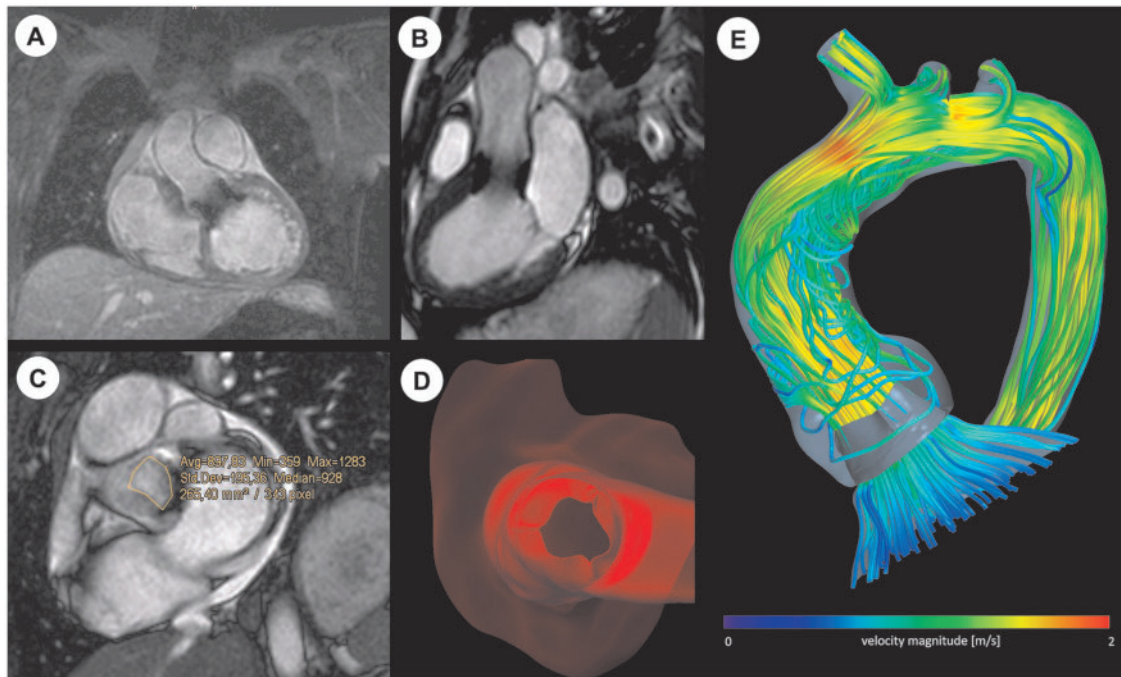
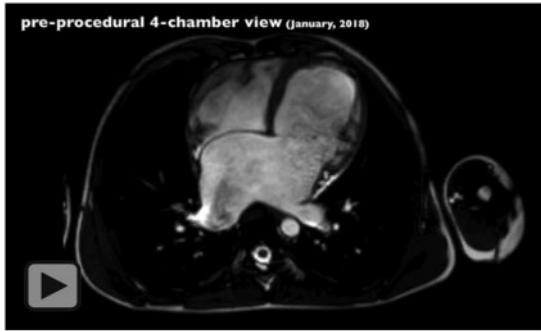


Figure 4 Cardiac magnetic resonance imaging after transcatheter aortic valve implantation. Coronal view (A) and 3-chamber view (B). End-systolic orifice area of the SAPIEN-3 prosthesis (C). Whole-heart 3D imaging with virtual SAPIEN-3 geometry (D). Computational fluid dynamic simulation (E) shows colour-coded streamlines illustrating a laminar flow profile without regurgitation.



Video 1 Cardiac magnetic resonance imaging before and after transcatheter aortic valve implantation. Four-chamber view and short-axis view are shown.

Table 1 Clinical, laboratory, and haemodynamic parameters 6–11 months before and 4 months after transcatheter aortic valve implantation

| Parameter | Before TAVI | After TAVI |
|-----------------------------|-------------|------------|
| BSA (m ²) | 1.36 | 1.43 |
| NYHA class (1–4) | 3 | 1 |
| NT-proBNP (pg/mL) | 2288 | 185 |
| LVEDVi (mL/m ²) | 103 | 111 |
| LVESVi (mL/m ²) | 65 | 53 |
| LVSVi (mL/m ²) | 38 | 58 |
| LVEF (%) | 37 | 53 |
| AV RF (%) | 30 | 2 |
| AV Vmax (m/s) | 2.5 | 1.6 |
| CI (L/min/m ²) | 2.6 | 4.0 |
| mPAP (mmHg) | 47 | 34 |
| PCWP (mmHg) | 21 | 15 |
| TPG (mmHg) | 26 | 19 |
| LVEDP (mmHg) | 25 | 17 |
| PVR (WE m ²) | 7.3 | 5.3 |
| mAAP (mmHg) | 71 | 72 |
| mPAP/mAAP | 0.66 | 0.47 |
| Grade of AR (0–4) | 3 | 0 |

AR, aortic regurgitation; AV RF, aortic valve regurgitant fraction; AV Vmax, peak aortic valve velocity; BSA, body surface area; CI, cardiac index; LVEDP, left ventricular end-diastolic pressure; LVEDVi, left ventricular end-diastolic volume index; LVEF, left ventricular ejection fraction; LVESVi, left ventricular end-systolic volume index; LVSVi, left ventricular stroke volume index; mAAP, mean aortic arterial pressure; mPAP, mean pulmonary arterial pressure; NT-proBNP, N-terminal pro-brain natriuretic peptide; NYHA, New York Heart Association; PCWP, pulmonary capillary wedge pressure; PVR, pulmonary vascular resistance; TAVI, transcatheter aortic valve implantation; TPG, transpulmonary gradient.

(compaction) is influenced by the pressure/volume load.⁸ The transient appearance of LV hypertrabeculation in response to LV loading conditions has been observed in pregnant women.⁹ As a consequence, we ask ourselves whether early elimination of a combined pressure/volume load and altered blood flow jets may also, to a

certain extent, facilitate restoration of LV morphology during childhood. In contrast to our paediatric patient, the characteristic LVNC features remained unchanged after aortic valve replacement in adult casuistics.^{2,10} Further studies in this very rare paediatric patient population should also include genetic analyses to discriminate between a hereditary genesis of LVNC-induced heart failure and BAV-induced morphological signs mimicking LVNC.

Conclusion

To the best of our knowledge, this is the first case performed with TAVI for BAV regurgitation in the context of LVNC. We consider our strategy a bridge to definitive therapy until our patient reaches adulthood. Whether surgical valve replacement with a mechanical prosthesis or heart transplantation will be favoured in the future will depend on the extent of LV recovery. We will continue to monitor the teenage patient.

Lead author biography



Axel Unbehaun is a cardiac surgeon. He is responsible for the institutional transcatheter heart valve programme.

Supplementary material

Supplementary material is available at *European Heart Journal - Case Reports* online.

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Slide sets: A fully edited slide set detailing this case and suitable for local presentation is available online as [Supplementary data](#).

Consent: The author/s confirm that written consent for submission and publication of this case report including image(s) and associated text has been obtained from the patient in line with COPE guidance.

Conflict of interest: A.U. and J.K. serve as proctors to Edwards Lifesciences Corp. The other authors have no disclosures regarding the manuscript.

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