*Cardiology in the Young*

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# Successful transcarotid transcatheter aortic valve replacement in a 34-kg patient with Schimke immuno-osseous dysplasia and severe biscuspid aortic stenosis

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**Brief Report**

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## Abstract

We present the case of transcatheter aortic valve replacement in a 20-year-old woman with severe bicuspid aortic stenosis and Schmike immuno-osseous dysplasia who was unfit for surgical aortic valve replacement. Meticulous pre-procedural planning and a multidisciplinary team approach can enable successful transcatheter aortic valve replacement in complex patients with genetic syndromes.

## Background

We present the case of transcatheter aortic valve replacement in a 20-year-old woman with severe bicuspid aortic stenosis and Schmike immuno-osseous dysplasia who was unfit for surgical aortic valve replacement. Schmike immuno-osseous dysplasia is an autosomal recessive multisystem disorder. Typically, the disorder is characterised by steroid resistance, nephrotic syndrome, immunodeficiency, and osseous dysplasia.

Reports of cardiac pathologies associated with this syndrome are uncommon but can include sub-aortic stenosis and atherosclerosis[1]. There is no clear association with bicuspid aortic valve disease or aortopathy in the reported literature. To our knowledge, this is the first reported application of transcatheter aortic valve replacement in a patient with Schmike immuno-osseous dysplasia. The decision to undertake transcatheter aortic valve replacement was complicated, with a number of anticipated clinical and technical challenges. These included low body weight (34 kg), severe left ventricular systolic dysfunction, severe pulmonary hypertension, small-sized femoral and subclavian access vessels precluding both trans-femoral and trans-subclavian access, aberrant aortic arch anatomy, and small aortic annulus size with pre-existing sub-aortic stenosis.

## Case

A 20-year-old woman with Schmike immuno-osseous dysplasia (*SMARCAL1* gene positive) was referred to our unit for management of severe calcific aortic stenosis. In the 6 months before presentation, she was largely wheelchair bound owing to severe exertional shortness of breath and required maintenance oxygen therapy. Her past medical history was notable for surgical repair of a ventricular septal defect at the age of 2 years, end-stage renal disease requiring dialysis, prior right frontal cerebral infarct, and hypothyroidism. Her weight was 34.3 kg and height was 134.3 cm. Transthoracic echocardiography showed severe left ventricular hypertrophy with severely reduced left ventricular systolic function, peak and mean gradients of 95 and 55 mmHg across the aortic valve, and severe tricuspid regurgitation with an estimated right ventricular systolic pressure of 90 mmHg.

After an extensive discussion with the heart team, the patient was considered to be at a prohibitive risk for surgical aortic valve replacement, with a risk of mortality of 13.68% as predicted by Society of Thoracic Surgeons, and transcatheter aortic valve replacement was planned. CT showed small femoral (3 mm) and subclavian arteries (5 mm) precluding access using these sites. The common carotid arteries arose a single trunk from the aortic arch, with the diameters of the left and right common carotid arteries measuring 6 and 5 mm, respectively (Supplementary material). The perimeter- and area-derived mean diameters of the aortic annulus were 17.2 and 17.4 mm, respectively. CT intracranial angiography was performed to verify a complete Circle of Willis before undertaking a transcarotid approach. On the basis of these data, a left transcarotid approach was planned.

The procedure was performed in a hybrid operating room under general anaesthesia. The patient had a 20-g cannula placed in the left hand, an ultrasound-guided arterial line in the left



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femoral artery, and a left femoral vein central venous catheter also under ultrasound guidance. After induction of general anaesthesia with sevoflourane in oxygen and placement of a 6.0 endotracheal tube, the patient required significant vasopressor support to maintain a reasonable systolic blood pressure and cardiac output. Surgical exposure of the left common carotid artery was commenced. However, the low systemic blood pressure made identification of arterial and venous structures difficult. In addition, the patient required increasing amounts of vasopressor support and became increasingly haemodynamically unstable with fast atrial flutter requiring cardioversion and amiodarone. It was then elected to proceed with emergent aortic valvuloplasty. A 7Fr sheath was placed in the right common femoral artery for balloon delivery, and a 6Fr sheath was placed in the right common femoral vein for delivery of a temporary pacing catheter into the right ventricle. Under rapid pacing, a 14 mm × 5 cm valvuloplasty balloon was inflated across the aortic valve. This resulted in immediate haemodynamic improvement, facilitating continuation of left common carotid artery surgical exposure. An 18Fr trans-apical sheath (Edwards Lifesciences, Irvine, California, United States of America) was delivered through the left common carotid artery into the ascending thoracic aorta. A 20-mm Edwards Sapien XT valve (Edwards Lifesciences) was delivered through this sheath to the aortic position and deployed under rapid ventricular pacing (Fig 1). A degree of frame under-expansion was noted at the time of deployment; however, owing to the complicated nature of the index procedure, challenges encountered, and concern regarding valve dislodgement with post-dilation maneuvers, it was felt safer to proceed with post-dilation at a later time if needed.

After the surgery, the patient was transferred to the ICU in stable condition and was successfully extubated the same day without neurocognitive deficit. Transthoracic echo 6 days after the procedure demonstrated good recovery of left ventricuar systolic function. The valve was in satisfactory position. However, elevated gradients, such as peak and mean of 61 and 30 mmHg, respectively, were noted. Symptomatically, she reported significant improvement in her exertional capacity. She was discharged on day 10 and no longer required home oxygen therapy.

Although clinically well, the aortic transvalvular gradient remained persistently elevated. This gradient was felt to be attributable to the relatively small left ventricular outflow tract in combination with a transvalvular gradient. It had been noted at the time of the index procedure that the valve stent frame was not completely expanded. It was therefore elected to schedule balloon valvuloplasty to help remedy the valvular component of the residual gradient.

The patient returned to the catheterisation laboratory 8 months after the index procedure. Under general anaesthesia, valvuloplasty of the transcateter valve was performed using a 7 F 14 mm × 4 cm Atlas Gold balloon (Bard Peripheral Vascular Inc., Temple, TX, United States of America) through the right common femoral artery access (Fig 2). This was associated with a reduction in the peak-to-peak gradient across the aortic valve from 35 to 11 mmHg (Supplementary material). The patient is now 20 months past her initial procedure and remains clinically well. Ejection fraction before the procedure was 30%, and after the procedure it was 55%; in addition, pre-operative systolic pulmonary arterial pressure was 82 mmHg, and after balloon valvuloplasty this was measured at 40 mmHg.

## Discussion

This case outlines the first reported execution of transcatheter aortic valve replacement in a 20-year-old patient with Schmike immuno-osseous dysplasia. Specific challenges in this case were difficult vascular access, a bicuspid aortic valve requiring use of a Sapien XT prosthesis, and significant haemodynamic instability requiring a balloon valvuloplasty after a number of months. Despite these challenges in this unique case, the patient had a marked clinical response.

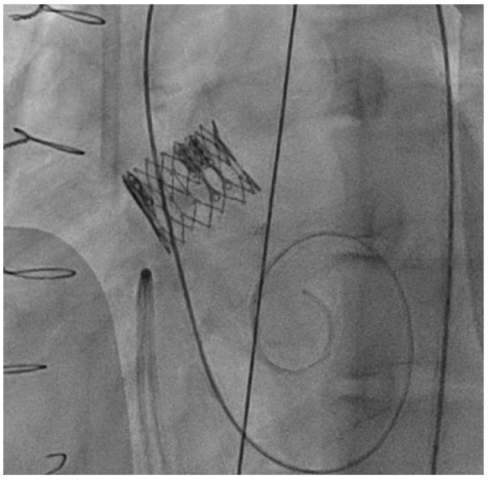

**Figure 2.** Valve following balloon valvuloplasty 8 months after the initial procedure. Note the more symmetrical appearance of the valve.

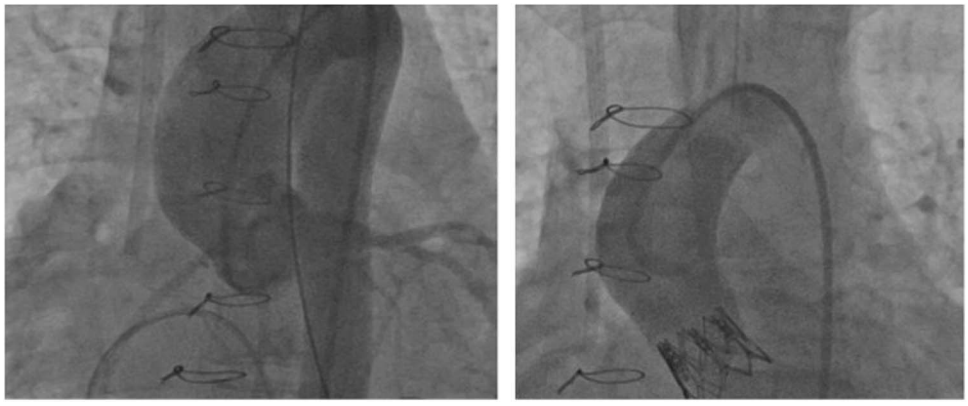

**Figure 1.** (**a**) Angiography showing all leads in place. Temporary pacing device in the right ventricular, trans-carotid wire passing into left ventricular and angiography catheter in the aorta. (**b**) Valve deployed with angiography checking position and function.

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Schmike immuno-osseous dysplasia is an autosomal recessive genetic multisystem disorder characterised by T-cell immunodeficiency, steroid-resistant nephrotic syndrome, and osseous dysplasia[2,3]. In up to 50% of cases, SIOD has been linked to a mutation in the *SMARCAL1* – *SWI/SNF*-related, matrix-associated, actin-dependent regulator of chromatin, subfamily a-like 1 – gene, which encodes a protein of the same name[2–4]. *SMARCAL1/HARP* seems to function at stalled or collapsed deoxyribonucleic acid replication forks to promote their stability or to directly facilitate the restart of replication and as such plays a significant role in the deoxyribonucleic acid damage response[5]. There is a range in severity; in some cases, growth retardation begins *in utero* with death occurring in the first few years of life, whereas others have a delayed onset and a more progressive course enabling survival into adulthood[2,4]. The mean age of death is 10.3 years owing to opportunistic infections, stroke, congestive heart failure, renal failure, and others[2,3]. Approximately half of the patients develop atherosclerosis, which is characterised by hyperplasia of the intima, media, and smooth muscle cells, as well as fragmented and disorganised elastin fibres. A proposed mechanism for its development is an impairment of elastogenesis[1]. In published literature to date, there is no clear association with aortic stenosis as seen in this case. This is perhaps because the majority of these patients fail to reach adulthood.

The clinical and technical challenges presented by this case were considerable. There are no guidelines for the appropriateness of transcatheter aortic valve replacement in such unique cases. Current models that estimate the risk for surgical aortic valve replacement cannot allow for rare serious genetic abnormalities such as this. In addition, the rate of transcatheter valve degeneration in very young patients, and the performance of transcatheter valves in patients with very small annulus size and co-existent sub-aortic stenosis, is not defined. Coronary height can be a concern in such cases and was an important consideration. Existing data demonstrate that the 30-day incidence of major or disabling stroke is 3.4 to 8% and most events occur within the first 24–48 hours[6]. Specific rates associated with transcarotid TAVI are unknown; in one series of transcarotid implantations involving common carotid cross-clamping, no stroke was reported in their series of 19 patients[7].

The appropriateness of transcatheter aortic valve replacement in this situation was largely based on the clinical judgement of the Heart Team. The futility of this procedure was considered; however, it was deemed on balance reasonable to undertake following meticulous planning and in consultation with the patient and her family. Full disclosure of the potential limitations of transcatheter aortic valve replacement in unique cases such as this is a necessary component of the consent process. In retrospect, this patient clearly benefited from the procedure. She recovered a normal ejection fraction, experienced a marked drop in pulmonary hypertension, and showed improvement in NYHA class from IV to II. However, it must be noted that the vigilance of the cardiac anaesthetist and the performance of an emergent aortic valvuloplasty to stabilise the patient before the surgical exposure of the carotid artery were probably life-saving. The importance of meticulous pre-procedural planning and an expert team of interventional cardiologists, anaesthetists, and cardiothoracic surgeons for such complex cases is clear[8–10].

Vascular access proved to be one of the most challenging components of this case. The patients' low body weight and growth retardation contributed to the small calibre of her peripheral arteries. The left common carotid access for valve delivery was chosen on the basis of the pre-procedural CT scan[11]. Trans-apical access was avoided because of the known issues with wound healing associated with the Schmike immuno-osseous dysplasia genetic abnormality and the patient's known severe left ventricular systolic dysfunction. It should be noted that the left common carotid access site did require surgical repair following removal of the valve sheath at the completion of the case. This was probably caused by the small calibre of the artery relative to the valve sheath size, and highlights the need for surgeons with the ability to repair the vascular access site for such complicated transcatheter cases.

The decision to perform a repeat valvuloplasty after the procedure in this patient was based on the hope that this would improve the long-term durability of the valve. The high gradients after valve deployment were probably a function of multiple factors – small valve size owing to the small annulus size, co-existent sub-aortic stenosis, and stent valve under-expansion. The valvuloplasty did successfully reduce the gradient and removed the restriction seen at the proximal end of the stent frame. The risk of trauma to the aortic valve leaflets and annulus with this manoeuvre was balanced against the desire to reduce the gradient and enable complete frame expansion. Distortion of leaflets resultant from frame under-expansion is known to reduce the durability of TAVI prostheses[12].

## Conclusion

Meticulous pre-procedural planning and a multidisciplinary team approach can enable successful transcatheter aortic valve replacement in complex patients with genetic syndromes that fall outside the typical patient demographic and application of the technology.

**Supplementary material.** To view supplementary material for this article, please visit https://doi.org/10.1017/S1047951118000410

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**Conflicts of Interest.** None.

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