Echocardiography to the Rescue in Adult-Onset Partial Shone Complex

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INTRODUCTION

Shone complex (or Shone syndrome) is a rare congenital disorder, which consists of supravalvular mitral ring, parachute mitral valve (MV), subaortic stenosis, and coarctation of the aorta (CoA).[1] While at least 4 lesions are required to diagnose complete Shone complex, partial Shone complex has been described when patients have only 2 or 3 of these left-sided obstructive lesions.[2] This disorder is typically diagnosed in childhood, with adult presentation being rare. Accordingly, we describe a case of partial Shone complex detected in adulthood.

CASE PRESENTATION

A 39-year-old man with a medical history of obesity and uncontrolled hypertension was referred to the cardiology clinic for evaluation of shortness of breath. The patient previously played sports in high school but is now limited solely to activities of daily living due to profound dyspnea. While their activity level had significantly decreased from their early adulthood, they remained asymptomatic for most of their adult life, although they rarely saw physicians. Thus, aside from their uncontrolled hypertension, they reported no other prior medical history or evaluation for any other condition. They also reported being compliant with their antihypertensive regimen, but despite this, blood pressure readings remained elevated. Their vitals in clinic showed a blood pressure of 155/71 mm Hg and a heart rate (HR) of 96 bpm. Their physical exam revealed an obese man with a II/IV diastolic murmur radiating to the apex and diminished lower extremity pulses.

A transthoracic echocardiography (TTE) was obtained, which revealed severe left ventricular (LV) hypertrophy (LV interventricular septum thickness in diastole 1.8 cm and LV posterior wall thickness in diastole 1.7 cm) and mildly reduced LV ejection fraction of 41%. Diastolic dysfunction was abnormal with left atrial dilation and significant LV hypertrophy. Tissue Doppler imaging parameters were uninterpretable on the initial TTE due to tachycardia. There was abnormal morphology of the MV with doming of the anterior leaflet and significant restriction of the posterior leaflet resulting in severe mitral stenosis (MS) with a mean transmitral gradient of 13 mm Hg at an HR of 100 bpm (Figure 1). Many of the chordae appeared to attach to one papillary muscle, suggesting the presence of a parachute MV (Videos 1 and 2). Given these findings, a transesophageal echocardiogram (TEE) was obtained to better characterize the MV abnormalities. The TEE confirmed the presence of abnormal MV morphology with thickening, doming, and restriction of the leaflets resulting in flow acceleration and MS (Figure 2A, Videos 3 and 4). Two closely spaced papillary muscles were seen on transgastric imaging, suggesting parachute-like MV morphology (Figure 2B); it was not considered a true parachute morphology due to the presence of 2 papillary muscles. There was no evidence of a supravalvular mitral ring on TEE imaging; the mitral annulus measured 3.4 × 2.9 cm. Long-axis views with color-flow Doppler revealed a trileaflet aortic valve with severe aortic regurgitation (AR) with a pressure half time of 213 ms (Video 3) and severe aortic root dilation up to 5.7 cm (Figure 2C) without any evidence of subaortic membrane. The mechanism of AR was found to be due to central leaflet malcoaptation likely from aortic root dilation (Video 5). Echocardiographic imaging of the descending aorta with pullback toward the aortic arch revealed a segment of discrete narrowing with color-flow Doppler flow acceleration, consistent with CoA (Figure 2D).

As part of a comprehensive workup, a cardiovascular magnetic resonance (CMR) angiography was ordered, which confirmed the presence of parachute-like MV pathology and severe CoA just distal to the left subclavian artery origin with an extensive collateral network (Figure 3, Video 6).

Given the presence of both a parachute-like MV and CoA, a diagnosis of partial Shone complex was made. The patient was referred to both cardiac surgery and the adult congenital heart disease clinic for further management. They also completed a computed tomography angiography of the head with contrast, which ruled out any intracranial aneurysms. Given the severity of their cardiac lesions, a staged procedure was considered to be the safest way to address their coexisting lesions. The patient first underwent successful percutaneous stenting of the CoA in the cardiac catheterization lab. Follow-up cardiac computed tomography (CCT) showed a well-placed coarctation stent with significant improvement in the lumen size (Figure 4, Video 7) in addition to a postoperative chest radiograph that demonstrated rib notching classically associated with CoA (Figure 5). Weeks later, the patient was taken to the operating room for mechanical MV replacement, mechanical aortic valve replacement, and ascending aorta replacement. The patient tolerated the procedure and was eventually discharged to a rehabilitation facility.

DISCUSSION

Shone complex is a rare congenital cardiac malformation—first described by Dr. John Shone in 1963—characterized by a series of 4

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VIDEO HIGHLIGHTS

Video 1: Two-dimensional TTE, parasternal long-axis view, demonstrates LV hypertrophy, mildly reduced LV global systolic function, anterior leaflet doming, and posterior leaflet restriction with most chordae appearing to attach to one papillary muscle.

Video 2: Two-dimensional TTE, apical 4-chamber, left heart focused view, demonstrates the restricted mitral leaflet motion with apparent attachment of most chordae to a single papillary muscle.

Video 3: Two-dimensional TEE, midesophageal 3-chamber view (135°) with color-flow Doppler, demonstrates the domed mitral leaflets with flow acceleration consistent with MS, mild mitral regurgitation, and severe AR with a dilated ascending aorta.

Video 4: Three-dimensional TEE, photorealistic volume-rendered image from the LV perspective, demonstrates the dysmorphic MV with domed and restricted leaflet motion.

Video 5: Two-dimensional TEE, midesophageal aortic valve short-axis view (46°), demonstrates trileaflet aortic valve morphology with central leaflet malcoaptation as the mechanism of severe AR.

Video 6: Cardiovascular magnetic resonance angiography, three-dimensional volume-rendered reconstruction, cine-rotational display, demonstrates the dilated ascending aorta and the CoA with a robust network of intercostal collateral vessels.

Video 7: Cardiac computed tomography, three-dimensional volume-rendered reconstruction, cine-rotational display, demonstrates the dilated ascending aorta and the coarctation following percutaneous repair with stenting.

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left-sided obstructive lesions.[1] Partial Shone complex is diagnosed when less than 4 of these obstructions are present.[2] While the incidence of this condition is quite low (approximately 0.6% of all cases of congenital heart disease), this rare condition has substantial morbidity and mortality.[2] Most cases are diagnosed in childhood, but in rare cases patients may remain undiagnosed and present with obstructive symptoms in adulthood as was seen in our patient.

Often, in adult patients with Shone complex such as ours, the first presenting symptom is dyspnea on exertion.[3] The degree of dyspnea and disability is proportional to the degree of MS from the structurally abnormal MV. Indeed, MV obstruction and stenosis is thought to be the first pathological event in this syndrome. There is also an association with a supramitral ring and subaortic stenosis, neither of which were seen on TEE imaging in our patient. The initial TTE demonstrated a significant transmitral gradient of 13 mm Hg at an HR of 100 bpm. Tachycardia can increase the transmitral gradient in MS patients and worsen symptoms of dyspnea. Prompt initiation of a beta-blocker drug was crucial in lowering their HR and alleviating some of their symptoms. Repeat TTE after covered stenting of the CoA and before their MV replacement demonstrated a lower mean transmitral pressure gradient of 8.0 mm Hg at an HR of 83 bpm, highlighting the contribution of their HR to their preprocedural gradients. The severity of MS is the most important indicator of mortality and long-term prognosis.[3] In a retrospective study of 50 Shone complex patients, certain MV morphologies (thickened leaflets, shortened MV chordae, and turbulent flow below the MV plane) were associated with significant MS and poor outcomes.[4] Classically, the MV morphology associated with Shone complex is that of a true parachute MV and a supramitral ring. A parachute MV is defined as a single, unifocal attachment of the chordae tendineae to a single papillary muscle. This leads to significant MV inflow obstruction.[5] However, a parachute-like MV can also occur. Unlike a true parachute MV, a parachute-like MV can have 2 or more papillary muscles that are morphologically abnormal and closely approximated. The unifocal attachment of the chordae results in significant MS.[6] Prior reviews have found a high association between MV abnormalities and CoA.[7] In the original Shone and colleagues review, 4 of the original 8 cases had a focal CoA.[1] Coarctation of the aorta is defined as a focal, discrete segment of aorta that is constricted and displays evidence of obstruction. These lesions are associated with delayed or absent femoral pulses, difficult to treat hypertension, and rib notching on chest radiograph, all of which were present in our patient. We hypothesize that their delayed presentation may have in part been due to the absence of serial obstructive lesions seen in complete Shone (absence of supravalvular mitral ring and subaortic membrane) and the development of an extensive collateral network. It is also possible that despite significant LV hypertrophy and diastolic dysfunction, they were able to maintain normal to near-normal coronary perfusion pressure due to the coarctation, and once the aortic root dilated and the AR became severe, symptoms developed. Certainly, their illness may have been detected earlier had a thorough screening for secondary causes of hypertension been performed in their youth.

Due to the often-undifferentiated presentation, TTE is often the first imaging test obtained in the evaluation of these patients, as it allows for assessment of LV function and significant valvular abnormalities. Our patient's initial TTE demonstrated a mildly abnormal LV systolic function likely due to long-standing hypertension and severe AR. The initial TTE was also crucial in identifying the abnormal MV structure and MS as the potential cause of the patient's symptoms. The presence of dysmorphic MV features ultimately prompted the pursuit of the TEE, which made the complete diagnosis. Although not present in our patient, TTE may also aid in identifying associated subaortic stenosis, supramitral ring, and CoA. The TTE failed to detect the CoA in our patient due to lack of performance of suprasternal views. Our case highlights the importance of careful interrogation of the descending aorta with suprasternal views to assess for aortic coarctation whenever dysmorphic mitral leaflets or bicuspid aortic valve are present. Additionally, our case highlights the importance of having diligent echocardiographers and sonographers with knowledge and experience in congenital heart disease to obtain the comprehensive images needed to establish a diagnosis. Failure to detect any of the serial obstructive lesions could have resulted in an inadequate operation and worse outcomes in this patient.

Additional imaging (i.e., with TEE, CMR, or CCT) is often required to assess papillary muscle morphology and to evaluate for aortic coarctation. A subsequent TEE confirmed the presence of 2 closely approximated papillary muscles resulting in parachute-like mitral subvalvular morphology. The TEE was also crucial in identifying significant aortic valve insufficiency, aortic root dilation, and a CoA. In the presence of known or suspected CoA, cross-sectional imaging with CMR or CCT may be useful to define the location, length, and diameter of the coarctation to guide operative or transcatheter interventions.

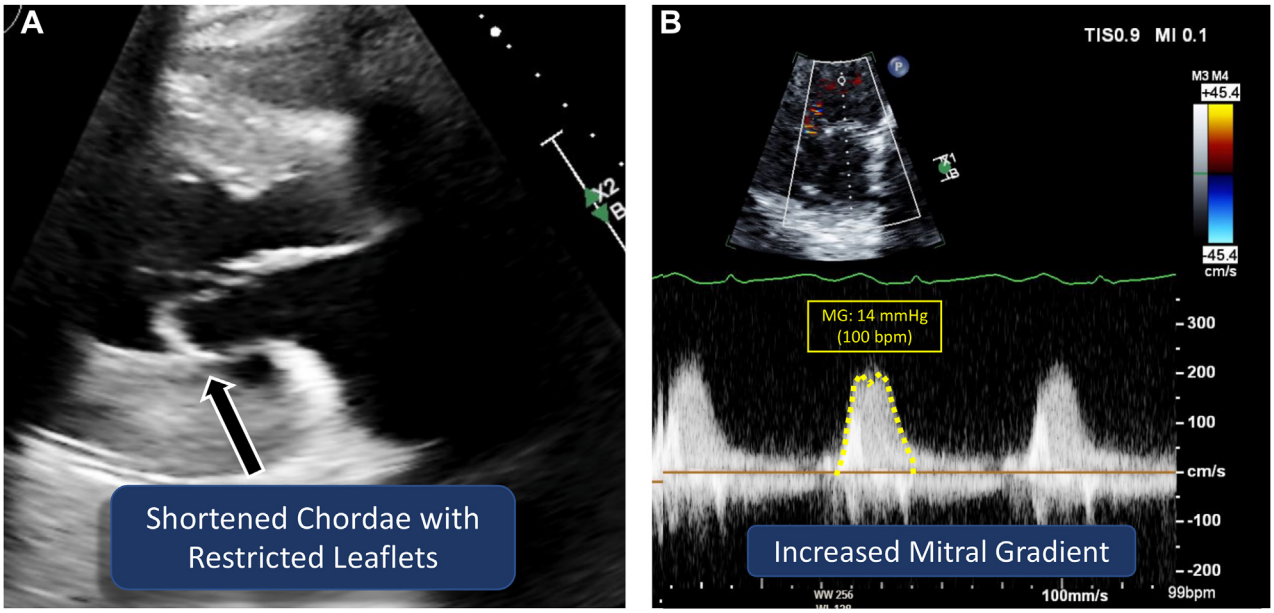

Figure 1 **(A)** Two-dimensional TTE, parasternal long-axis view, diastolic phase, demonstrates shortened chordae with restricted and domed mitral leaflets. **(B)** Two-dimensional TTE, apical 4-chamber view, continuous-wave Doppler spectrum, demonstrates severe MS with a mean diastolic pressure gradient of 14 mm Hg (at HR 100 bpm).

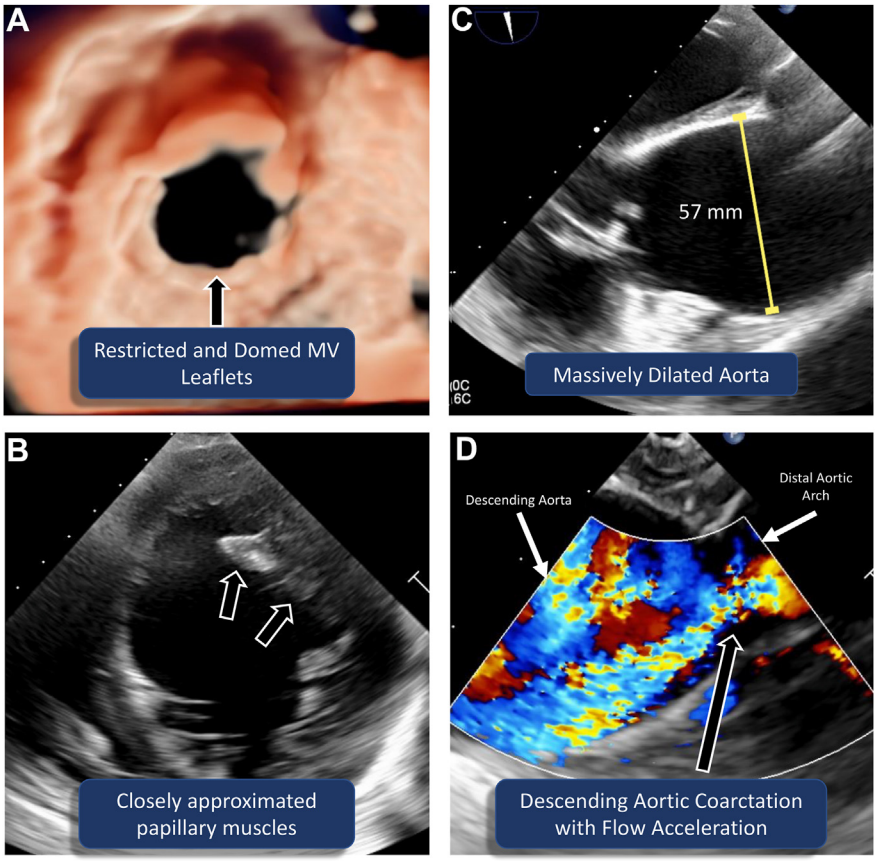

Figure 2 **(A)** Three-dimensional TEE, photorealistic volume-rendered image, diastolic phase from the LV perspective, demonstrates the dysmorphic MV with domed leaflets. **(B)** Two-dimensional TEE, transgastric short-axis mid-LV view, demonstrates the closely approximated papillary muscles with parachute-like morphology. **(C)** Two-dimensional TEE, midesophageal diastolic long-axis view, demonstrates the severe ascending aortic dilation. **(D)** Two-dimensional TEE with color-flow Doppler, long-axis view of the descending aorta in systole, demonstrates flow acceleration consistent with CoA.

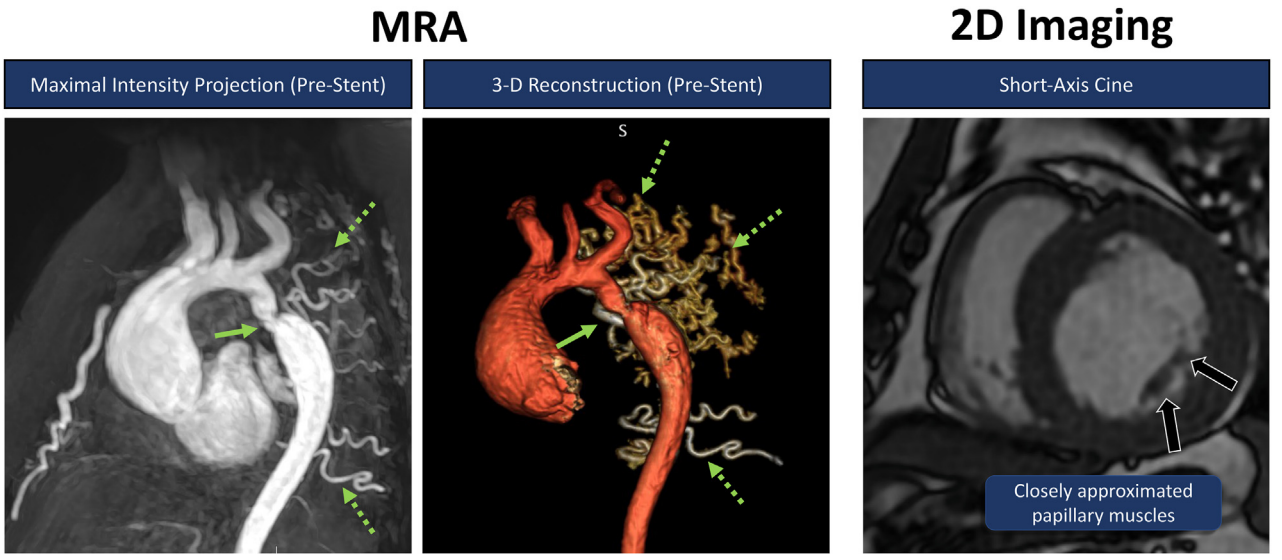

Figure 3 Cardiovascular magnetic resonance angiography (MRA), oblique sagittal view, multiplanar reconstruction (*left panel*) and three-dimensional volume-rendered reconstruction (*middle panel*), demonstrates the CoA just distal to the left subclavian artery (*solid arrows*) with an extensive network of collateral vessels (*dashed arrows*). Steady-state free precession cine sequence, diastolic mid-LV short-axis view, demonstrates the close approximation of the papillary muscles consistent with parachute-like morphology (*right panel*). *2D*, two-dimensional; *3-D*, three-dimensional.

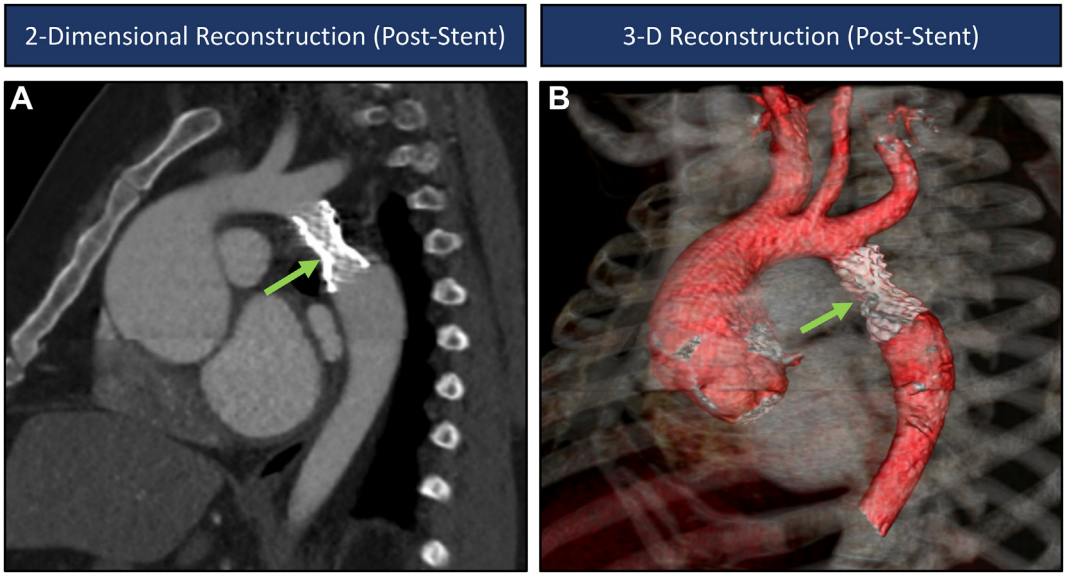

Figure 4 Cardiac computed tomography, oblique sagittal view, multiplanar **(A)** and volume-rendered reconstruction **(B)** of the aorta, demonstrates the coarctation following percutaneous repair with stenting (*arrows*) and improved patency. *3-D*, Three-dimensional.

Partial Shone complex is a largely underrecognized entity that carries significant morbidity and mortality.[8] Although there are no studies that have focused on adult outcomes, early diagnosis and treatment is crucial to lower the mortality associated with this condition. The treatment involves addressing the left-sided obstructive lesions by either surgical intervention or percutaneous approaches. Cardiac obstructive lesions are typically repaired surgically by median sternotomy, whereas aortic coarctation typically requires a thoracotomy approach; this may make it difficult to address all the obstructive lesions in a single cardiac surgery. Thus, several reports have been published describing a staged intervention to address all obstructive lesions. Although an aortic valve–sparing operation was considered, ultimately the adult congenital and cardiac surgical teams opted for mechanical aortic valve replacement given the uncertain repairability of the valve and need for lifelong anticoagulation due to mechanical MV.

Several reports have described the operative approach to address the parachute MV in partial Shone complex. Mitral valve replacement is the most common intervention among adult patients with severe MS and parachute MV. Mitral valve repair is also cited as an alternative

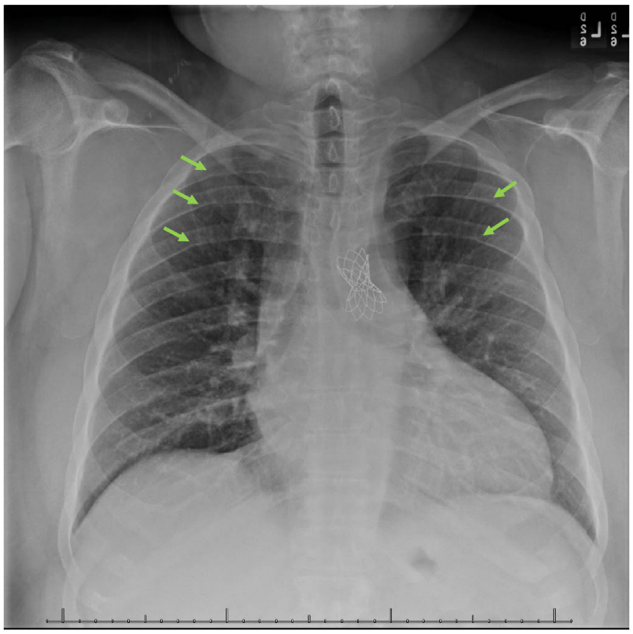

Figure 5 Chest x-ray, anterior-posterior view, demonstrates rib notching (*arrows*) from the hemodynamically significant CoA with a robust network of intercostal collateral vessels.

to complete replacement in selected cases.[9] However, after careful examination of the case, our congenital and cardiothoracic teams opined that the MV and subvalvular apparatus were too dysmorphic for repair, opting instead for valve replacement.

CONCLUSION

We describe a case of an adult patient presenting with dyspnea with exertion who was ultimately diagnosed with partial Shone complex. This case underscores the important role of echocardiography for the identification of congenital valvular and left-sided obstructive lesions associated with this rare disease. Additionally, we highlight the complementary role of advanced imaging modalities such as CCT and CMR. Prompt identification and recognition of this constellation of abnormalities allowed for a swift surgical evaluation and management of the obstructive lesions associated with this condition. Our patient represents a unique case of partial Shone complex diagnosed in adulthood and managed with a novel strategy that involved a staged percutaneous and surgical intervention approach.

ETHICS STATEMENT

The authors declare that the work described has been carried out in accordance with The Code of Ethics of the World Medical Association (Declaration of Helsinki) for experiments involving humans.

CONSENT STATEMENT

The authors declare that since this was a noninterventional, retrospective, observational study utilizing de-identified data, informed consent was not required from the patient under IRB exemption status.

FUNDING STATEMENT

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DISCLOSURE STATEMENT

The authors report no conflict of interest.

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SUPPLEMENTARY DATA

Supplementary data related to this article can be found online at https://doi.org/10.1016/j.case.2024.01.002.

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