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CASE REPORT
Sports cardiology

The valuable role of cardio-pulmonary exercise testing in the diagnosis of atrial septal defect in a competitive triathlete: a case report

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Background Atrial septal defect (ASD) is characterized by a diverse clinical presentation influenced by the type, size, and haemodynamics. Endurance athletes with ASD may exhibit higher than normal performance levels, however they face an elevated risk of exercise-induced cardiac volume and pressure strain, potentially expediting a maladaptation of the right heart.

Case summary An asymptomatic 28-year-old female elite triathlete sought a pre-participation sports medical examination. Her past medical history revealed right heart enlargement. Transthoracic echocardiography and magnetic resonance imaging did not ascertain a definitive diagnosis such as shunting. The examination revealed a remarkably high maximum oxygen uptake during cardio-pulmonary exercise testing (CPET), yet an abnormal oxygen uptake/workload slope and a low, plateauing oxygen pulse. The athlete agreed to transoesophageal echocardiography that demonstrated a superior sinus venosus-type ASD. Surgical intervention, conducted with minimally invasive endoscopic robotic technology and a pericardial patch, was performed at a tertiary centre under full cardio-pulmonary bypass. At seven-month follow-up, the patient reported engaging in swim sessions without limitations and participating in high intensity cycling sessions with performances similar to pre-surgery. Cardio-pulmonary exercise testing revealed increased maximum oxygen consumption and normalization of oxygen uptake/workload slope and maximum oxygen pulse.

Discussion Endurance athletes with ASD may have abnormal haemodynamic response during CPET despite an exceptional high maximum oxygen uptake. This underscores the value of CPET in the diagnostic work-up of right heart enlargement.

Keywords Congenital heart disease • Echocardiography • Exercise testing • Sports cardiology • Case report

ESC curriculum 8.1 Sports cardiology • 9.7 Adult congenital heart disease

Learning points

- Cardio-pulmonary exercise testing is a valuable tool for the evaluation of right heart enlargement in endurance athletes.
- The surgical strategy for atrial septal defect closure in competitive athletes needs to be tailored to the patient's characteristics and the aim of maintaining competitive sports participation.

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Introduction

Congenital heart disease stands as the most prevalent birth defect, affecting 8–9 per 1000 live births and encompassing a diverse range of conditions.[1] Among these, atrial septal defect (ASD) emerges as the most frequently diagnosed congenital heart disease in adulthood, resulting in blood shunting between the systemic and pulmonary circulation.[2] The physiological impact and clinical course of ASD in adults vary depending on its type, size, and haemodynamic consequences. This spectrum ranges from asymptomatic cases without structural or functional cardiac anomalies to instances of asymptomatic right heart enlargement, pulmonary hypertension, and a progression of symptoms, mainly exercise intolerance, with age.[3] In asymptomatic adults, the detection of ASD often occurs incidentally through the identification of a heart murmur during auscultation, an abnormal electrocardiogram, or through cardiac imaging conducted for unrelated reasons.[3] Echocardiography, serving as the primary diagnostic tool, permits direct visualization of ASD or indirect signs such as pulmonary hypertension and/or right heart enlargement.

Endurance athletes with ASD comprise a distinct, vulnerable cohort, as the cardiac volume and pressure strain induced by prolonged and/or vigorous exercise may expedite the maladaptation of the right heart associated with ASD.[4] Consequently, early diagnosis and timely intervention, particularly in cases involving right heart enlargement, alongside regular follow-up examinations, are of critical importance for ensuring the continued safe participation of endurance athletes in competitive sports. Pre-participation sports medical screening examinations play a pivotal role in the early identification of conditions like ASD, often incorporating transthoracic echocardiography (TTE) as a point-of-care assessment.[5] However, athletes with ASD may exhibit impressive performance levels that to some extent mask the condition, especially as some ASDs prove elusive during routine TTE, and right heart enlargement is frequently considered a norm in an athlete's heart,[6] complicating ASD diagnostic work-up. The incorporation of cardio-pulmonary exercise testing (CPET), readily available in sports medical centres, can facilitate early assessment of ASD in equivocal clinical cases.

This case report presents an asymptomatic national elite endurance athlete diagnosed with ASD, right heart enlargement, and abnormal findings during CPET.

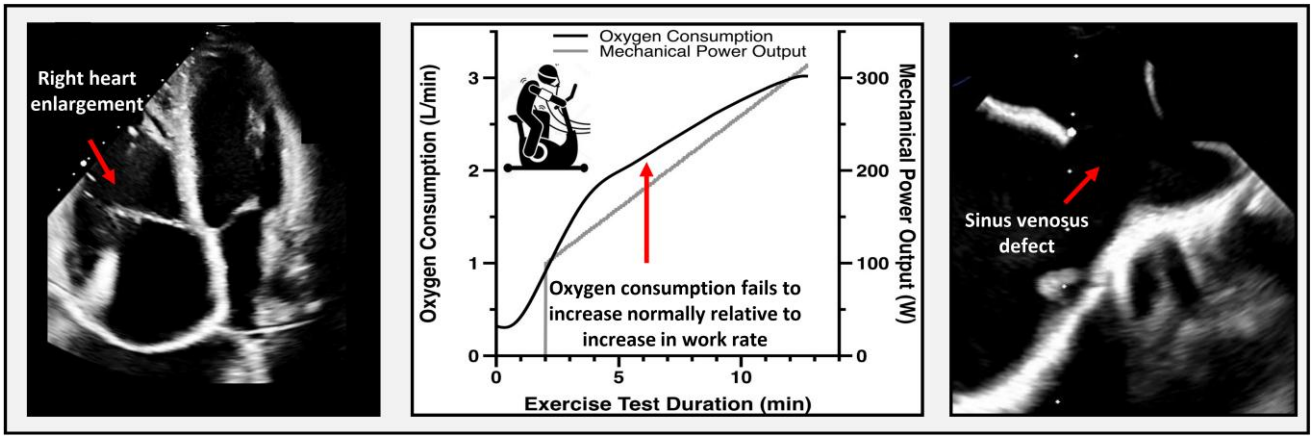
Summary figure

Case presentation

A 28-year-old female triathlete (height: 167 cm; body mass: 54 kg) engaged in 25 h of weekly training sought a pre-participation sports medical examination at our department. Her past medical history revealed a pre-collapse episode during a triathlon race approximately one year ago. A CT scan ruled out pulmonary embolism but revealed right heart enlargement and borderline pulmonary hypertension without objective evidence for shunting on subsequent TTE. The pre-collapse was attributed to dehydration but magnetic resonance imaging was performed for diagnostic work-up of right heart pathologies, which affirmed right heart enlargement but could not ascertain a definitive diagnosis or shunting. Despite recommendations for a right-heart catheter examination, the athlete declined due to the invasive nature of the procedure and her lack of symptoms. The physical examination during the sports medical assessment at our department was unremarkable, and the resting electrocardiogram displayed normal p-wave amplitude (largest amplitude within the inferior leads in II with 1.2 mm), left-axis deviation (−150°), incomplete right bundle branch block with rSR′ configuration, and a persisting S until V6. Transthoracic echocardiography depicted significant right heart enlargement (*Figure 1A*), with additional findings of reversed inter-ventricular septal motion during diastole, suggestive of haemodynamic significance. Spectral Doppler assessment demonstrated a right ventricular to atrial pressure gradient (RVPG) of 29 mmHg (*Figure 1B*).[7] Right ventricular systolic function was normal (global strain −28%). Attempts to visualize an ASD using 2D and colour-Doppler TTE proved inconclusive. Dynamic stress echocardiography, using a supine exercise table, revealed an increased RVPG already at 50 W (45 mmHg; *Figure 1C*), a RVPG of 62 mmHg at maximum exercise and a steep mean pulmonary arterial pressure [mPAP)/cardiac output (CO)] slope (2.7 mmHg/L/min), indicating the potential impact of physical exertion on the condition. Systolic cardiac function did not indicate abnormalities at maximum exercise.

Cardio-pulmonary exercise testing indicated a remarkable maximum oxygen uptake ($\dot{V}O_2$max) of 57 mL/min/kg and mechanical power output (5.8 W/kg), yet an abnormal $\dot{V}O_2$/workload slope (7.5 mL/min/W; *Figure 2A*), plateauing and low maximum oxygen pulse (16 mL/beat; *Figure 2B*), and a drop in peripheral oxygen saturation (92%) at the end of the exercise test indicating abnormal haemodynamic response to exercise. Exercise electrocardiography, heart rate, and blood pressure during exercise were unremarkable.

In light of these inconclusive findings, the athlete agreed to right-heart catheterization, revealing a considerable difference in resting



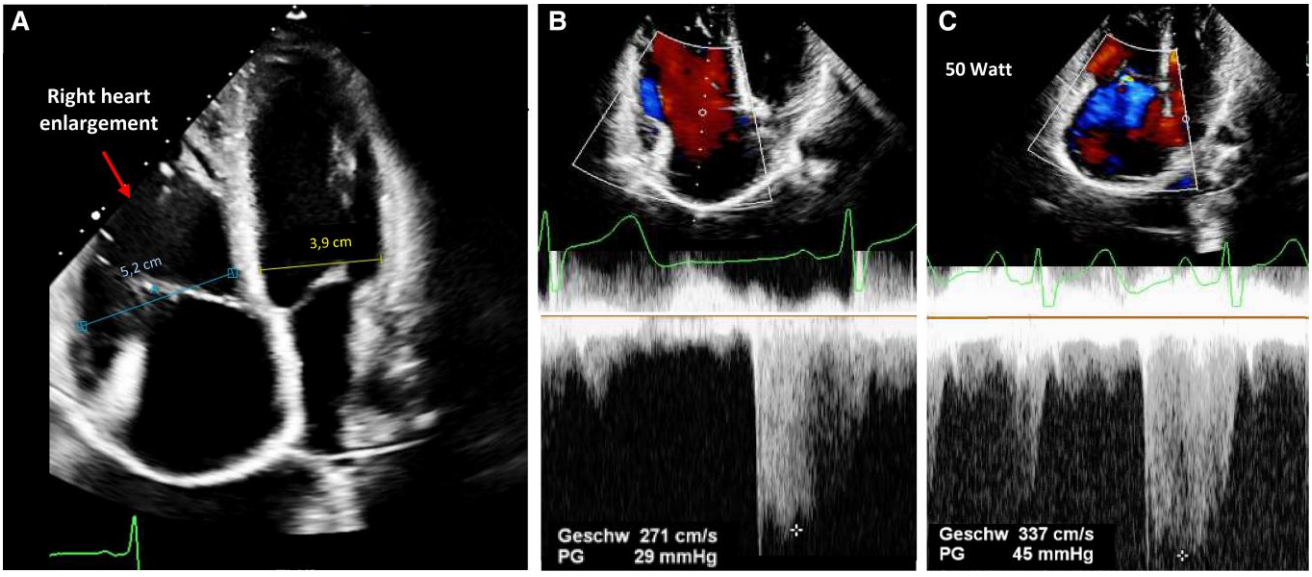


Figure 1 Transthoracic echocardiography pre-atrial septal defect closure. Two-dimensional (*A*), spectral Doppler at rest (*B*), and exercise (50 W, *C*) transthoracic echocardiography.

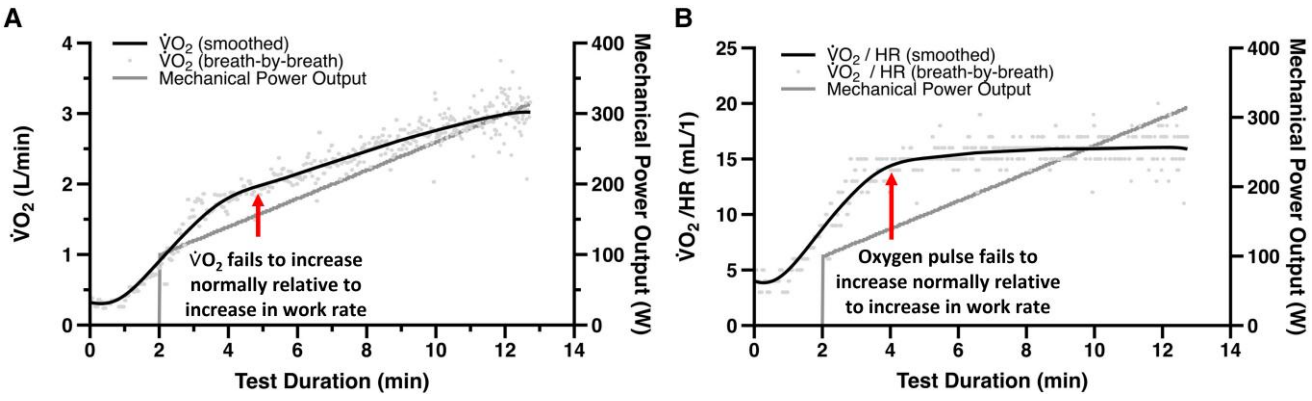


Figure 2 Cardio-pulmonary exercise testing pre-atrial septal defect closure. Oxygen uptake (*A*) and oxygen pulse (*B*); $\dot{V}O_2$, oxygen uptake; HR, heart rate.

oxygen saturation between the superior caval vein (72.2%) and the pulmonary artery (88.6%), indicative of left-to-right shunting. Right-heart catheter assessment during exercise testing confirmed echocardiographic findings with a mPAP increase to 52 mmHg at 200 W. 2D and colour Doppler-enhanced transoesophageal echocardiography demonstrated a superior sinus venosus-type ASD (*Figure 3A and B*; Supplementary material online, *Video S1*), which is a class I indication for surgical repair when considering the enlarged right heart.[8]

Surgical intervention, conducted with minimally invasive endoscopic robotic technology and a pericardial patch was performed at a tertiary centre under full cardio-pulmonary bypass. Post-surgery, the patient faced a complication in the form of pericarditis, successfully managed with non-steroidal anti-inflammatory drugs and colchicine. Physical rehabilitation commenced with walking 10 days after surgery, progressing to cycle ergometer training and inpatient cardiac rehabilitation approximately two months later.

At three-month follow-up examination, the patient was free of symptoms and TTE indicated regression of right-heart dimensions (*Figure 4A*), normalized pulmonary pressures (*Figure 4B*), and normal inter-ventricular septal motion during diastole indicating normal haemodynamics. Dynamic stress echocardiography at 50 W revealed a reduced RVPG of 26 mmHg (*Figure 4C*).

2D and colour Doppler-enhanced transoesophageal echocardiography demonstrated successful ASD closure (*Figure 5A and B*; Supplementary material online, *Video S2*).

At seven-month follow-up, the patient reported engaging in swim sessions without limitations and participating in high intensity cycling sessions with performances similar to those pre-surgery. Cardio-pulmonary exercise testing revealed increased $\dot{V}O_2$max (63 mL/min/kg), increased maximum mechanical power output (5.9 W/kg), normalization of $\dot{V}O_2$/workload slope (10.1 mL/min/W; *Figure 6A*), and maximum oxygen pulse (20 mL/beat; *Figure 6B*). Peripheral oxygen

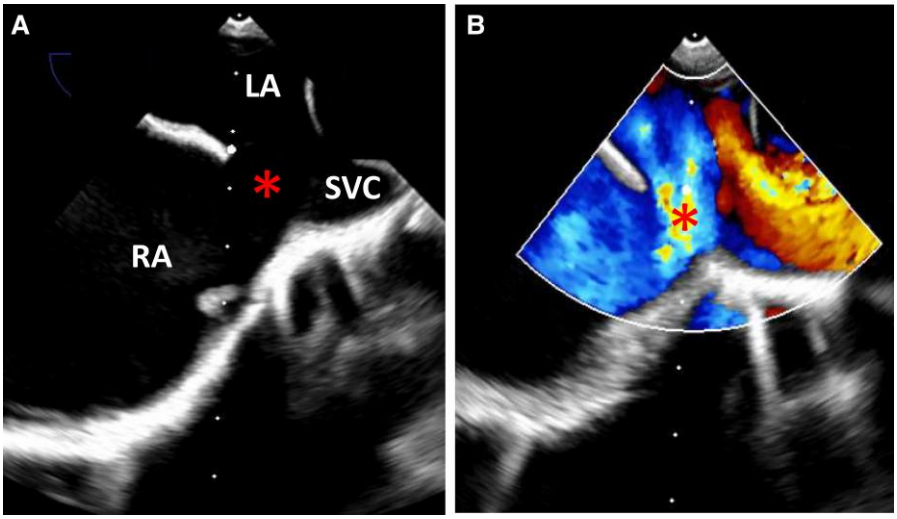


Figure 3 Transoesophageal echocardiography pre-atrial septal defect closure. Two-dimensional (*A*) and colour-Doppler (*B*) transoesophageal echocardiography; LA, left atrium; RA, right atrium; SVC, superior caval vein; asterisk indicates the sinus venosus defect.

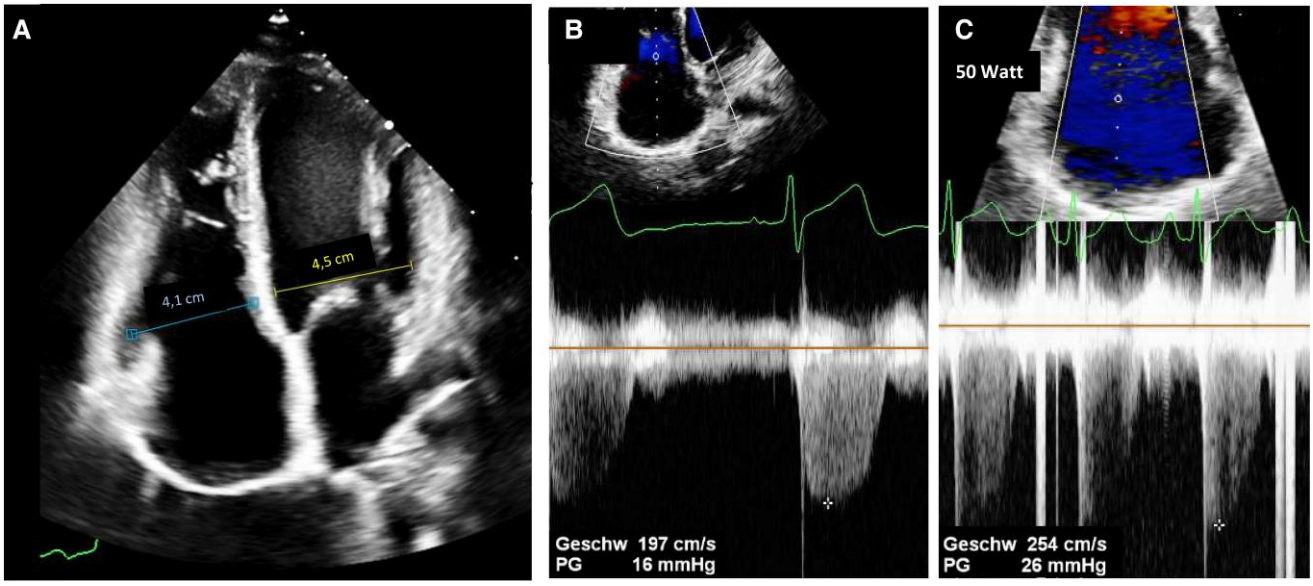


Figure 4 Transthoracic echocardiography post-atrial septal defect closure. Two-dimensional (*A*), spectral Doppler at rest (*B*), and exercise (50 W, *C*) transthoracic echocardiography.

saturation at the end of the exercise test (91%) was similar to that pre-surgery.

Discussion

This case vividly underscores the diagnostic dilemma of differentiating endurance exercise-related right heart enlargement from ASD-related maladaptation in an asymptomatic endurance athlete while emphasizing the valuable role of CPET. Despite a $\dot{V}O_2$max exceeding normal values by 65%, the pattern observed in the abnormal increase of $\dot{V}O_2$ and oxygen pulse relative to the rise in workload during incremental exercise point to a significant impairment in stroke volume during exertion. While dynamic stress echocardiography revealed normal contractility, impairment in left ventricular diastolic mechanics became apparent already during resting TTE by the reversed inter-ventricular septal motion during diastole, which likely resulted from a disruption in ventricular-ventricular interaction secondary to right ventricular enlargement. This disruption, in turn, affects left ventricular preload, contributing to left ventricular stroke volume reduction, especially with increasing heart

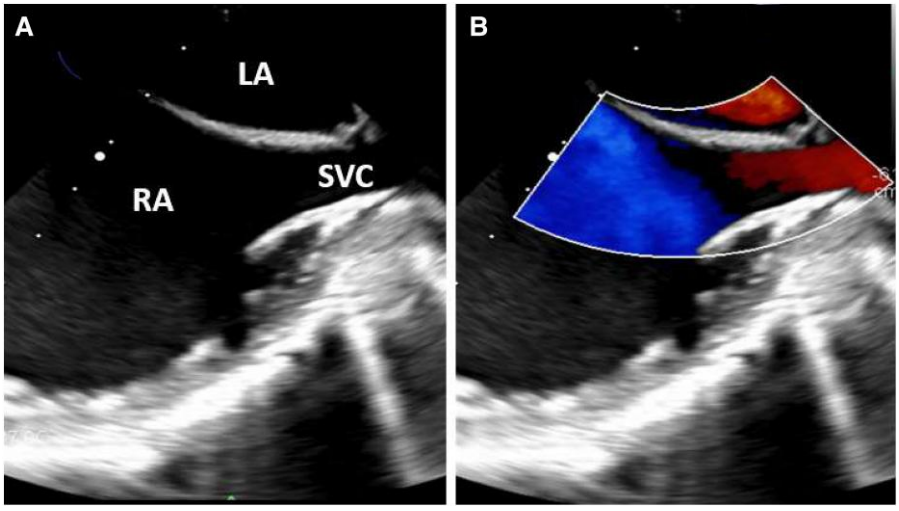


Figure 5 Transoesophageal echocardiography post-atrial septal defect closure. Two-dimensional (*A*) and colour-Doppler (*B*) transoesophageal echocardiography; LA, left atrium; RA, right atrium; SVC, superior caval vein.

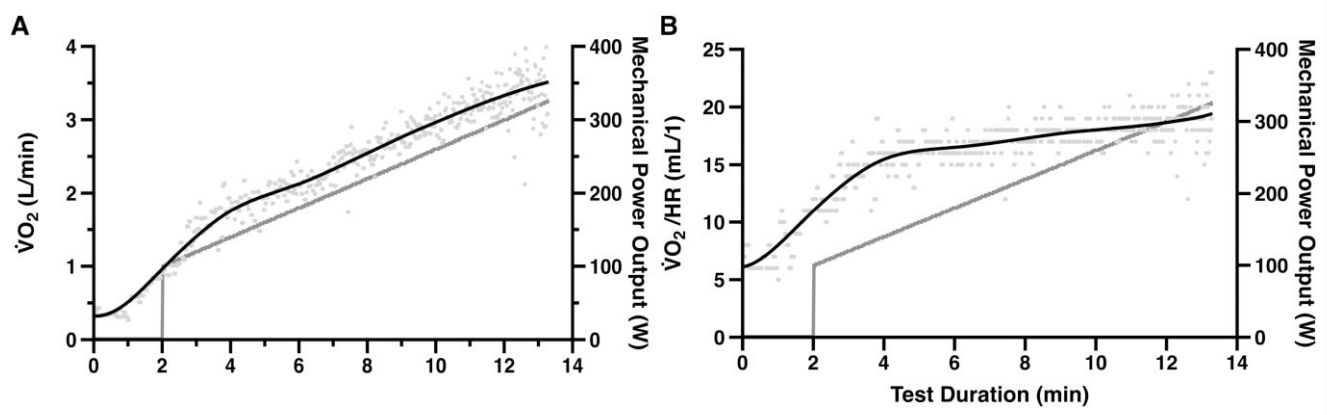


Figure 6 Cardio-pulmonary exercise testing post-ASD closure. Oxygen uptake (*A*) and oxygen pulse (*B*); $\dot{V}O_2$, oxygen uptake; HR, heart rate.

rate during exercise. Furthermore, the steep mPAP/CO slope of 2.7 mmHg/L/min indicates increased right ventricular afterload during exercise.[9] These factors collectively emerge as the most plausible explanations for the abnormal $\dot{V}O_2$ observed during CPET, highlighting the multifaceted pathophysiologic nature and diagnostic challenge in such cases.

Notably, prior to surgery, peripheral oxygen saturation decreased to 92% at the end of the exercise test, a condition commonly known as exercise-induced arterial hypoxaemia (EIAH). This phenomenon can stem from various factors, predominantly affecting female athletes. In these, the occurrence of EIAH is possibly associated with e.g. compromised airway structures, limiting sufficient oxygen supply during periods of excessive ventilatory demand.[10] However, it can also be due to right-to-left heart shunting. Interestingly, the deoxygenation pattern did hardly change in our athlete after ASD closure, indicating EIAH causing mechanisms unrelated to the ASD.

Current guidelines recommend ASD closure if there is (i) right ventricular volume overload, (ii) pulmonary vascular resistance that does not exceed 3 Wood units, and (iii) no left ventricular disease.[8] Surgical closure is typically performed for sinus venosus defect, given their unsuitability for device closure.[2] Traditionally, these procedures involve a median sternotomy, with larger defects addressed using patches, often sourced from homologous or heterologous pericardium or artificial materials. However, contemporary approaches explore alternative techniques, such as partial mini-sternotomy and endoscopic methods. While median sternotomy remains conventional, studies suggest that approaches with limited skin incisions yield similar outcomes,[11] offering specific advantages in terms of athletic performance, particularly in activities like swimming, and addressing cosmetic concerns. In our athlete, the surgical strategy for ASD closure was tailored to the patient's characteristics and the goal of maintaining elite-level participation in triathlon competitions, and thus performed with minimally invasive endoscopic robotic technology.

It is widely recognized that ASD closure induces reverse right ventricular remodelling, with the extent inversely related to patient age.[12] This dynamic is highlighted by our athlete, where ASD closure

led to the normalization of right heart geometry within three months. Additionally, both resting and exercise pulmonary pressures returned to normal three months post-ASD closure.

Successful surgical repair of ASD paves the way for a potential return to all sports in athletes who are then asymptomatic.[13] In our specific case, seven months post-surgery, the patient remained symptom-free, transoesophageal echocardiography confirmed successful closure, and CPET demonstrated a normal cardiovascular response to exercise. Consequently, the athlete received clearance to resume competitive triathlon. Follow-up examinations including CPET were arranged to monitor for potential late complications, such as supraventricular arrhythmias.

In conclusion, endurance athletes with ASD may have an abnormal haemodynamic response during CPET despite an exceptional high maximum oxygen uptake. This underscores the value of CPET in the diagnostic work-up of right heart enlargement, where the surgical strategy must be tailored to the patient's characteristics and needs, the latter include the aim of maintaining competitive sports participation in case of competitive athletes.

Lead author biography

Mahdi Sareban is a senior cardiologist and sports physician at the University Institute of Sports Medicine, Prevention and Rehabilitation in Salzburg, Austria. His main clinical interest and research cover cardiovascular adaptation to exercise training in endurance athletes and patients with cardiovascular diseases.

Supplementary material

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

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Consent: The authors confirm that written consent for submission and publication of this case report including images and associated text has been obtained from the patient in line with COPE guidance.

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Data availability

The data underlying this article will be shared on reasonable request to the corresponding author.

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