Cardiology in the Young

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Unrepaired tetralogy of Fallot in the adult: an unexpected diagnosis

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Brief Report

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Abstract

A 40-year-old female is admitted for paroxysmal episodes of exertional dyspnoea, with associated cyanosis, improving with squatting, and a holosystolic murmur radiating to the interscapular area. Echocardiography showed a subaortic ventricular septal defect with left-to-right shunt and overriding aorta. The characteristic murmur prompted us to seek right ventricular outflow tract obstruction. Magnetic resonance was performed, confirming Tetralogy of Fallot, and corrective surgery was performed.

The authors present the case of a 40-year-old female patient born in Angola, with 2 previous uncomplicated pregnancies and a history of iron-deficiency anaemia, who had recently immigrated to Portugal. The patient was admitted to the Cardiology unit with frequent paroxysmal episodes of exertional dyspnoea with progressive worsening in the last months, associated with cyanosis and oxygen desaturation.

These episodes alleviated with squatting position or prolonged rest. The oxygen saturation was 97% in ambient air. The symptoms had been present since childhood, but the patient had not received regular medical care and had no known cardiac condition. The physical examination was notable for a harsh intense (IV/VI) holosystolic murmur at the left sternal border, with a sternal heave, radiating to the interscapular area. The blood pressure was 100/58 mmHg and heart rate of 62 beats per minute, and there was no evidence of digital clubbing, peripheral oedema nor an abnormal jugular venous pulse.

The patient had previously performed a 12-lead electrocardiogram revealing sinus rhythm with right axis deviation, RSR pattern in lead V1, and suggestive of right ventricular hypertrophy. The chest x-ray did not show significant findings.

Laboratory studies showed a haemoglobin level of 11.8 g/dL, a creatinine level of 0.77 mg/dL, normal liver enzymes, thyroid function, and urinalysis, and there was a mild elevation of the N-terminal pro-b-type natriuretic peptide of 210 pg/mL (reference range <125 pg/mL).

A transthoracic echocardiogram was performed, revealing a non-dilated left ventricle with preserved systolic function and a 10mm subaortic ventricular septal defect, with left-to-right shunt (50 mmHg gradient), as well as an overriding aorta (30–50% of the interventricular septum) (Supplementary Fig S1). The presence of a characteristic murmur with the concomitant finding of right ventricular hypertrophy on echocardiography prompted us to seek the presence of a right ventricular outflow tract obstruction, which was present in the pulmonary infundibulum with a 99 mmHg gradient. 24-hour Holter revealed sinus rhythm with a mean heart rate of 61 beats per minute and no significant ventricular ectopy or sinus pauses.

During the hospital admission, there was recurrence of the exertional dyspnoea and oxygen desaturation episodes (minimum oxygen saturation of 66% by pulse oximetry) with cyanosis, improving with oxygen, intravenous propranolol and morphine, reverting to an oxygen saturation of 97%. Beta-blocker oral treatment with propranolol was started, with decreasing frequency of the hypoxemia episodes.

Cardiac MRI confirmed the final diagnosis of tetralogy of Fallot, showing an overriding aorta (40–50% of the interventricular septum) with moderate aortic regurgitation, a 12 mm subaortic ventricular septal defect (Fig 1), and a smaller muscular septal defect (5 mm), with a Qp/Qs of 1.37:1. Furthermore, cardiac magnetic resonance detailed right ventricular hypertrophy and hypertrabeculation and showed thick muscle bundles in the pulmonary infundibulum (Fig 2) conditioning a severe obstruction during systole (Supplementary Figs S1 and S2). No pulmonary artery branch stenosis was present.

Right heart catheterisation showed a Qp/Qs of 1.1:1 and excluded pulmonary hypertension (mean pulmonary artery pressure of 10 mmHg). There were no obstructive coronary lesions.

After multidisciplinary discussion with the adult CHD and cardiac surgical teams, the patient underwent tetralogy of Fallot surgical repair with closure of the septal defect, resection of muscle bundles in the right ventricular outflow tract, and augmentation of the infundibulum, with no complications.

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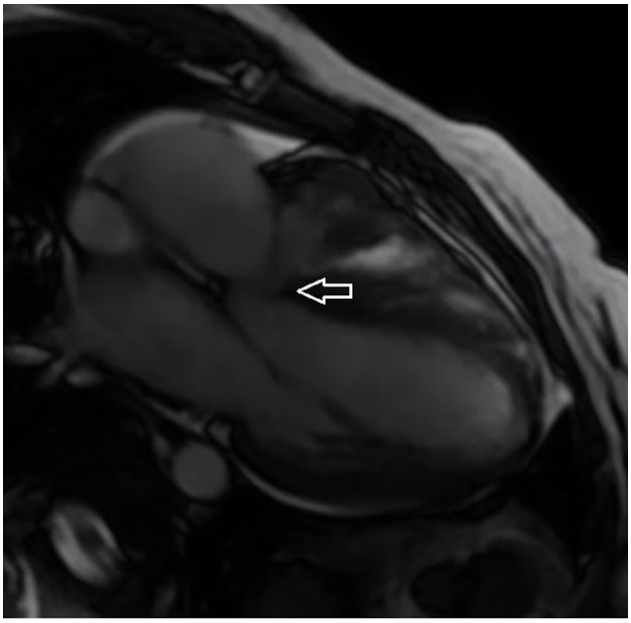

Figure 1. Cardiac MRI. White arrow: subaortic ventricular septal defect. Note the overriding aorta.

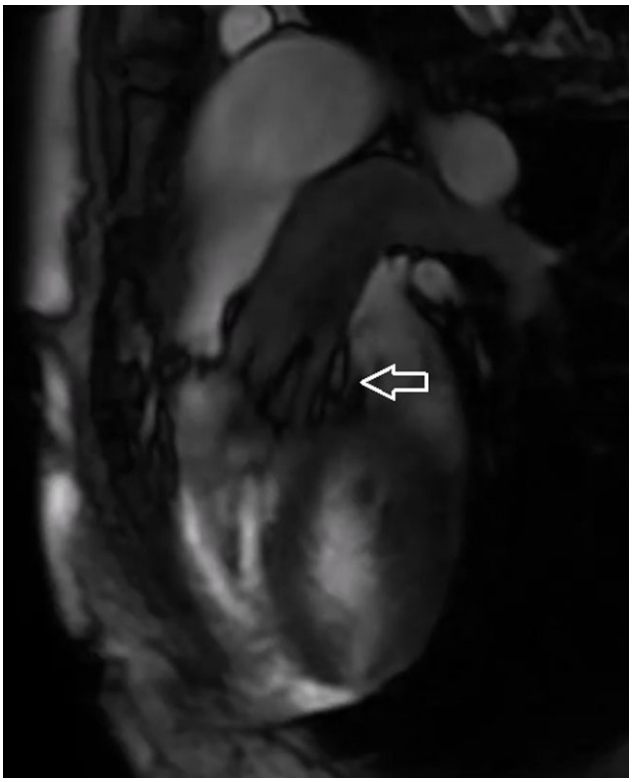

Figure 2. Cardiac MRI. White arrow: Thick muscle bundles in the pulmonary infundibulum conditioning a severe obstruction during systole.

The post-operative transthoracic echocardiogram showed two residual small restrictive ventricular septal defects – the largest with an 84 mmHg gradient – a mild pulmonary infundibulum gradient (15 mmHg) and mild aortic regurgitation.

In a 6-month follow-up, there was no recurrence of dyspnoea or hypoxemia episodes, in NYHA class I with no further hospitalisations.

Tetralogy of Fallot is characterised by a non-restrictive ventricular septal defect; an overriding aorta; infundibular, valvular, supravalvular right ventricular outflow tract obstruction, and/or branch pulmonary artery stenosis; and consequent right ventricular hypertrophy. The majority of tetralogy of Fallot patients are non-syndromic, although it can be associated with microdeletion 22q11, trisomy 21, Alagille, Noonan, Williams, and Klippel–Feil, among other syndromes.[1]

This case details an unexpected diagnosis of CHD in a 40-year-old patient, reminding us that one should never overlook the possible diagnosis of CHD in adult patients, particularly in patients without previous regular medical care. Likewise, this case highlights the health disparities regarding adult CHD care in several African countries, with limited access to centres with adult CHD expertise.

The symptoms improving with squatting was a key diagnostic clue as squatting manoeuvre increases systemic vascular resistance, resulting in reversal of shunting at the ventricular septal defect and therefore a reduction in symptoms.[2]

The initial diagnostic hypothesis was a ventricular septal defect; however, the presence of the harsh holosystolic murmur radiating to the interscapular area, characteristic of right ventricular outflow tract obstruction, prompted us to seek the presence of additional congenital anomalies or a complex CHD, uncovering the final diagnosis of tetralogy of Fallot.

Cardiac magnetic resonance is a key complementary exam in the evaluation of complex CHD, providing us crucial anatomical information regarding the VSD, overriding aorta, and RVOT obstruction, and thus guiding an optimal surgical corrective intervention.[3,4] CT can also provide insightful information on cardiac anatomy in adult CHD, allowing for accurate imaging of smaller structures due to its high spatial resolution.[5]

Supplementary material. To view supplementary material for this article, please visit https://doi.org/10.1017/S1047951122001779

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Ethical standards. The patient has provided written consent for the submission of this manuscript.

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