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Case Report

Missed Tetralogy of Fallot in an Elderly Woman With a Known Ventricular Septal Defect

Dinesh Kalra, MD,[a] and Sumit Sohal, MD[b]

[a] Division of Cardiology, Rush University Medical Center, Chicago, Illinois, USA

[b] Department of Internal Medicine, Amita Health Saint Francis Hospital, Evanston, Illinois, USA

ABSTRACT

Tetralogy of Fallot is a congenital heart disease comprised of a tetrad of ventricular septal defect, pulmonary stenosis, overriding aorta, and right ventricular hypertrophy. In developed countries, most cases are diagnosed in babies; mortality is high if not surgically corrected in a timely manner. We describe herein a woman who was diagnosed at age 73 years. Several factors accounted for her unusual longevity. We highlight the importance of multimodal imaging to look for other associated anomalies of tetralogy of Fallot in cases of apparent simple ventricular septal defect when the echocardiographic images are either suggestive or suboptimal.

RÉSUMÉ

La tétralogie de Fallot est une maladie cardiaque congénitale, tétrade comprenant une communication inter-ventriculaire, une sténose pulmonaire, une dextroposition de l'aorte et une hypertrophie du ventricule droit. Dans les pays développés, la plupart des cas sont diagnostiqués chez des nourissons; la mortalité est élevée si elle n'est pas corrigée chirurgicalement en temps opportun. Nous rapportons le cas d'une femme qui a été diagnostiquée à l'âge de 73 ans. Plusieurs facteurs expliquent sa longévité inhabituelle. Nous soulignons l'importance de l'imagerie multimodale pour rechercher d'autres anomalies associées à la tétralogie de Fallot dans les cas apparemment simples de communication inter-ventriculaire lorsque les images échocardiographiques sont soit révélatrices, soit imparfaites.

Tetralogy of Fallot (TOF) accounts for 7% of congenital heart disease occurring in 0.05% of live births; it is the most common cause of cyanotic congenital heart disease. It results from anterocephalad displacement of the infundibular septum during embryogenesis, leading to a tetrad of malalignment ventricular septal defect (VSD), right ventricular outflow (RVOT) obstruction, overriding aorta, and right ventricular hypertrophy. TOF may be accompanied by right-sided aortic arch, patent ductus arteriosus, atrial septal defect, MAPCAs (major aortopulmonary collateral arteries), pulmonary stenosis/hypoplasia/atresia, aortic regurgitation, and coronary artery anomalies. For untreated cases, the mortality beyond age 40 years is 95%. Thus, it is rare to encounter undiagnosed TOF in elderly patients.[1]

Case

A 73 year-old female with hypertension and a known restrictive VSD since age 5 years presented to the cardiology clinic with pedal edema for 3 months. Exam was notable for a blood pressure of 145/88, harsh pansystolic murmur at the left lower sternal border, and a 3/6 systolic crescendo–decrescendo murmur in the pulmonic area. There was no palpable heave, and S1 and S2 were normal. Pulse oximeter readings at rest and after a 3-minute hallway walk were 98% and 96%, respectively. An electrocardiogram showed sinus rhythm and left atrial enlargement (Fig. 1). Hemoglobin was 14 g/dL. A transthoracic echocardiogram showed a left ventricular ejection fraction of 65%, increased RVOT gradient (peak: 66 mm Hg; mean: 36 mm Hg), mild pulmonic regurgitation, and a 0.8 cm perimembranous VSD with a left-to-right shunt (peak gradient: 79 mm Hg) (Fig. 1). Cardiac magnetic resonance imaging (CMR) was done for better characterization and showed mild left ventricular hypertrophy (LVH), a 0.8 cm misalignment VSD, an overriding aorta, left ventricular ejection fraction of 72%, and right ventricular ejection fraction of 57% (Fig. 2; Videos 1 and 2, view video online). It also showed moderate right ventricular hypertrophy, infundibular hypertrophy, and moderate RVOT (subpulmonic/infundibular) systolic narrowing/stenosis as well as mild pulmonary valvar stenosis and annular hypoplasia but without pulmonary branch stenosis or any MAPCAs, an

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Corresponding author: Dr Dinesh K. Kalra, Director of Advanced Cardiac Imaging, Director of Lipid Clinic & Infiltrative Cardiac Disease Program, Division of Cardiology, Rush University Medical Center, 1717 W. Congress Parkway, Kellogg Suite 320, Chicago, Illinois 60612, USA. Tel: +1-312-942-4601; fax: +1-312-942-6334.

E-mail: dinesh_kalra@rush.edu

See page 698 for disclosure information.

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Novel Teaching Points

- This case highlights the importance of using multimodal imaging to look for the other associated anomalies of TOF in cases of apparent simple VSDs when the echocardiographic images are either suggestive or suboptimal.
- CMR has advantages in detecting associated congenital anomalies due to its superior spatial and temporal resolution, lack of radiation, and multiplanar imaging with wide field of view.
- CMR provides hemodynamic information to guide treatment and can inform regarding prognosis.
- Cardiac computed tomography has excellent spatial resolution and is useful in defining associated coronary anomalies and in surgical planning in congenital heart disease.

overriding aorta, and a right-sided aortic arch. Cardiac output was 4.1 L/min and Qp:Qs was 1.2. No late gadolinium enhancement to indicate scar was present. These features were consistent with acyanotic TOF—she was previously diagnosed with only a membranous VSD; the other 3 features of TOF had gone undetected for almost 70 years. Cardiac computed tomography (Fig. 2) also showed the VSD and infundibular hypertrophy and stenosis. There was only mild coronary arterial stenosis. Her symptoms improved with diuretics and antihypertensive adjustment. She has been following in the cardiology clinic and has remained stable. No surgical intervention was deemed necessary due to absence of pulmonary hypertension, normal right ventricular size and function, and her age.

Discussion

In most cases, echo can be used to diagnose TOF, but CMR provides additional morphologic and functional information about the heart and pulmonary vasculature.[2] Undiagnosed TOF may be mistaken for a small VSD in the rare asymptomatic patient. In developed countries, most patients are diagnosed as newborns or in the first few years of life when they present with cyanosis and/or heart failure. Less than 3% of patients reach 40 years without intervention. Median unoperated survival is 12 years. Very rarely, patients remain asymptomatic—these patients have slow development of infundibular pulmonary stenosis such that hemodynamics are balanced, ie, a situation that prevents severe pulmonary hypertension (Eisenmenger's) from a large left-to-right shunt, yet maintains enough pulmonary

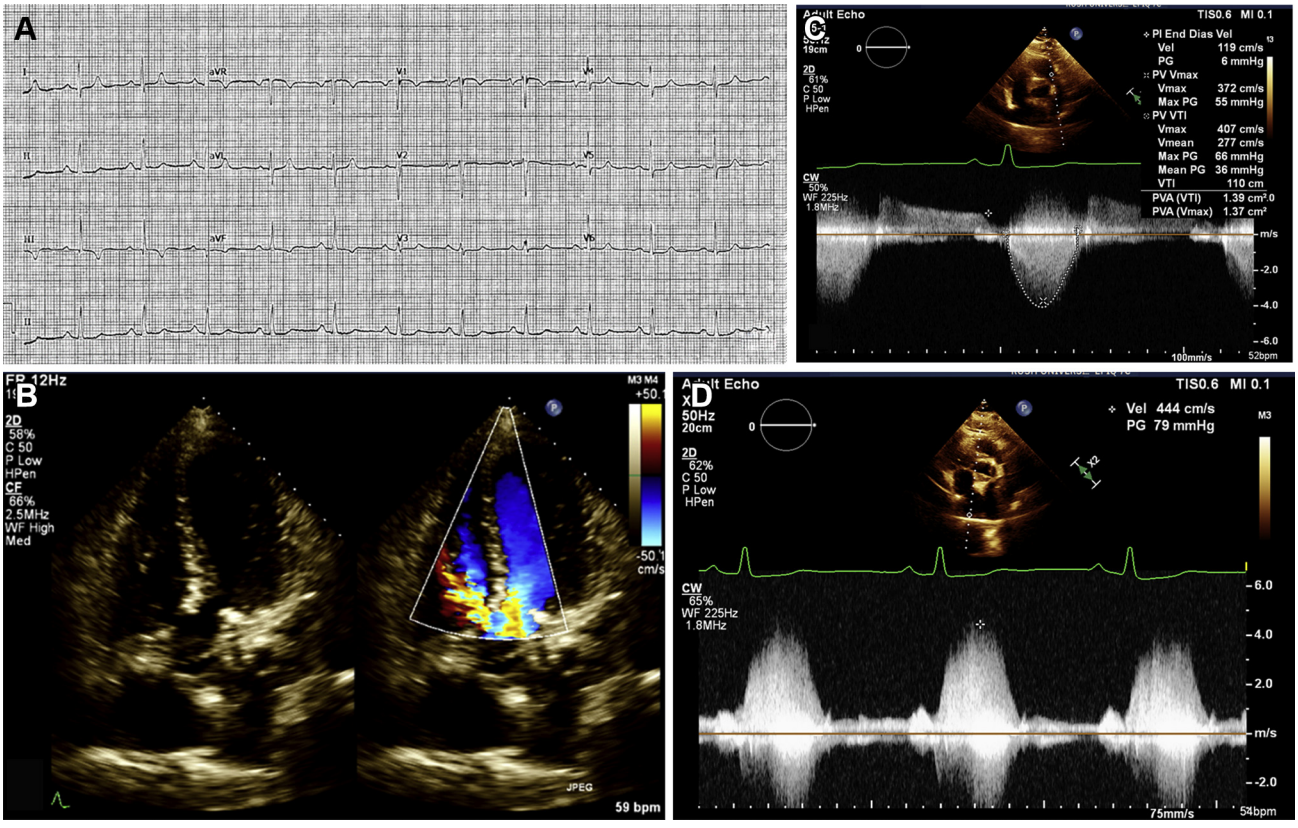

Figure 1. (**A**) A 12-lead electrocardiogram showing sinus rhythm and left atrial enlargement. (**B**) Apical 4-chamber view on echocardiogram showing ventricular septal defect: left-to-right color flow in the membranous septum region. (**C**) Continuous wave Doppler ultrasound across right ventricular outflow shows stenosis from infundibular muscular hypertrophy, pulmonary valvular restriction, and mild regurgitation. (**D**) Continuous wave Doppler ultrasound across the small, restrictive ventricular septal defect.

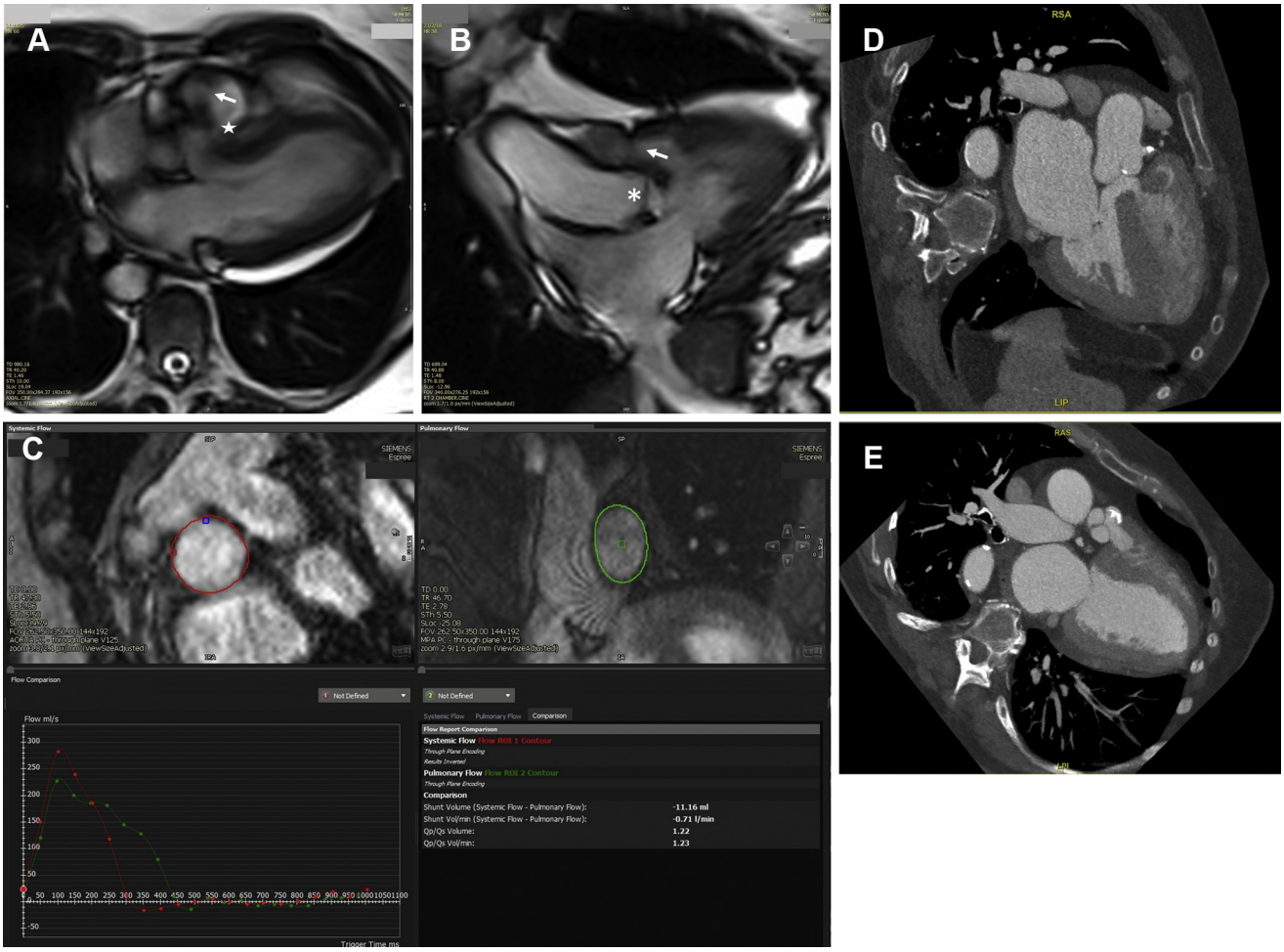

Figure 2. (**A**) Axial cine steady-state free precession magnetic resonance imaging still-frame showing aortic override, misalignment ventricular septal defect (**star**), and infundibular stenosis (**arrow**). (**B**) Right-sided 2-chamber steady-state free precession magnetic resonance imaging still-frame showing right ventricular hypertrophy, right ventricular outflow stenosis (**arrow**) and overriding aorta (**asterisk**). (**C**) Phase-contrast cardiac magnetic resonance image of through-plane flow across aortic root (**red**) and main pulmonary artery (**green**). Flow curves indicate a pulmonary flow to systemic flow ratio (Qp:Qs) of 1.2. (**D**) Cardiac computed tomography showing misalignment ventricular septal defect, right ventricular hypertrophy, and overriding aorta. (**E**) Narrow subpulmonic tract with mild calcification and stenosis of this region is evident.

blood flow to avoid cyanosis. The slow development of LVH/elevated left ventricular end-diastolic pressure also ameliorates against right-to-left ventricular shunting. In children with severe obstruction to RVOT, MAPCAs or systemic-to-pulmonary shunting (occurring via internal mammary arteries or a patent ductus arteriosus) may mitigate against pulmonary hypoperfusion and ensuing cyanosis, such that some of these patients may also survive to adulthood without surgical repair. Our patient had hypertension and LVH, and mild RVOT stenosis with a restrictive VSD, which kept her essentially asymptomatic due to a fine balance in only mildly increased pulmonary blood flow (Qp :Qs [pulmonary flow:systemic flow] of 1.2) that prevented Eisenmenger's physiology, yet protected her against the sequalae of cyanotic heart disease. To our knowledge, this case is one of the oldest patients in the literature, reporting survival into the 70s without surgical intervention.[1]

TOF is characterized by early-onset cyanosis, the severity of which is dependent on the degree of pulmonary obstruction and the magnitude of the VSD shunt. Some patients have heart failure symptoms when the left-to-right shunt predominates. As the RVOT obstruction increases, right-to-left shunting across the VSD increases, causing cyanosis. Surgical repair (transatrial/transpulmonary) involves VSD closure and right ventricular muscle resection/infundibulotomy/patch enlargement of the RVOT, and of the pulmonary arteries as needed. Repair is usually performed in infancy or childhood. However, surgery at an older age is also well tolerated—in a small cohort of patients who underwent surgery between ages 40 and 60 years at the Mayo Clinic, postoperative survival at 5 and 10 years was 92% and 74%, respectively.[3] The unique pathophysiology of our patient's case, consisting of a restrictive small VSD, slowly progressive infundibular muscular stenosis, and LVH developing later on in life, accounted for her compensated hemodynamics.

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Disclosures

The authors have no conflicts of interest to disclose.

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Supplementary Material

To access the supplementary material accompanying this article, visit *CJC Open* at https://www.cjcopen.ca/ and at https://doi.org/10.1016/j.cjco.2020.06.011.