JACC: CASE REPORTS VOL. 2, NO. 15, 2020

MINI-FOCUS ISSUE: CHEST WOUNDS

ADVANCED

CASE REPORT: CLINICAL CASE

Incidental Diagnosis After a Car Accident

A Rare Case of Asymptomatic Uncorrected Tetralogy of Fallot

Monica Verdoia, MD, PhD,[a,b] Rocco Gioscia, MD,[b] Pier Luigi Soldà, MD,[a] Federica Marrara, MD,[a] Borejda Xhyheri, MD,[a] Stefano Leuzzi, MD,[a] Giuseppe De Luca, MD, PhD,[b] Caterina Masia,[c] Ermanno Spagarino,[c] Mimii Emanoela Pascu,[c] Umberto Colageo, MD,[c] Marco Marcolongo, MD[a]

ABSTRACT

Tetralogy of Fallot (TOF) is a heterogeneous congenital heart disease that is occasionally diagnosed during adulthood. However, although they are often asymptomatic, adult patients with uncorrected TOF often have a poor prognosis. Poor outcomes indicate the importance of the identification and management of these patients, especially in the context of intercurrent disease or noncardiac surgery. We describe a case of clinically silent TOF in a 51-year-old woman. TOF was unmasked during a major noncardiac surgery for a polytrauma and successfully treated with the cooperation of a multidisciplinary team. (**Level of Difficulty: Advanced.**) (J Am Coll Cardiol Case Rep 2020;2:2289–94) © 2020 The Authors. Published by Elsevier on behalf of the American College of Cardiology Foundation. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).

A 51-year-old woman presented to the emergency department with polytrauma after a car accident, as the pedestrian victim who was hit while crossing the road.

Vital signs and physical examination on admission documented a heart rate of 82 beats/min and blood pressure of 90/60 mm Hg; no heart murmurs or signs of pulmonary congestion were reported.

Neurologic examination and state of consciousness were normal. She underwent immediate total body computed tomography (CT) scan, which revealed multiple costal and vertebral fractures; bilateral lung contusion; a displaced fracture of the distal part of the left humerus bone, of the acetabulum and left femur; and a detachment of the left renal calyx. She underwent emergency intervention of ureteral stenting and leg fracture fixation with an intramedullary nail.

While the orthopedic procedure was performed, the patient experienced transient hypotension and subsequent severe desaturation despite ventilation with maximal oxygen concentration. The patient had an unremarkable past medical history.

LEARNING OBJECTIVES

- To achieve awareness of atypical presentations of uncorrected CHD in adulthood.
- To understand the underlying pathophysiological mechanism and its impact on the management of these patients.

DIFFERENTIAL DIAGNOSIS

Due to suspected pulmonary fatty or thrombotic embolization, a dedicated pulmonary CT scan was performed immediately after the intervention. Interstitial edema and partial atelectasis of the inferior pulmonary lobes were observed. Moreover, enlargement of the ascending aorta (37 mm) and hypertrophy

From the [a]Division of Cardiology, Ospedale degli Infermi, ASL Biella, Italy; [b]Division of Cardiology, Ospedale Maggiore della Carità, Università del Piemonte Orientale, Novara, Italy; and the [c]Intensive Care Unit, Ospedale degli Infermi, ASL Biella, Italy.
The authors attest they are in compliance with human studies committees and animal welfare regulations of the authors' institutions and Food and Drug Administration guidelines, including patient consent where appropriate. For more information, visit the Author Center.

Manuscript received July 22, 2020; revised manuscript received August 18, 2020, accepted September 1, 2020.

ISSN 2666-0849 https://doi.org/10.1016/j.jaccas.2020.09.017

ABBREVIATIONS AND ACRONYMS

CHD = congenital heart disease

CT = computed tomography

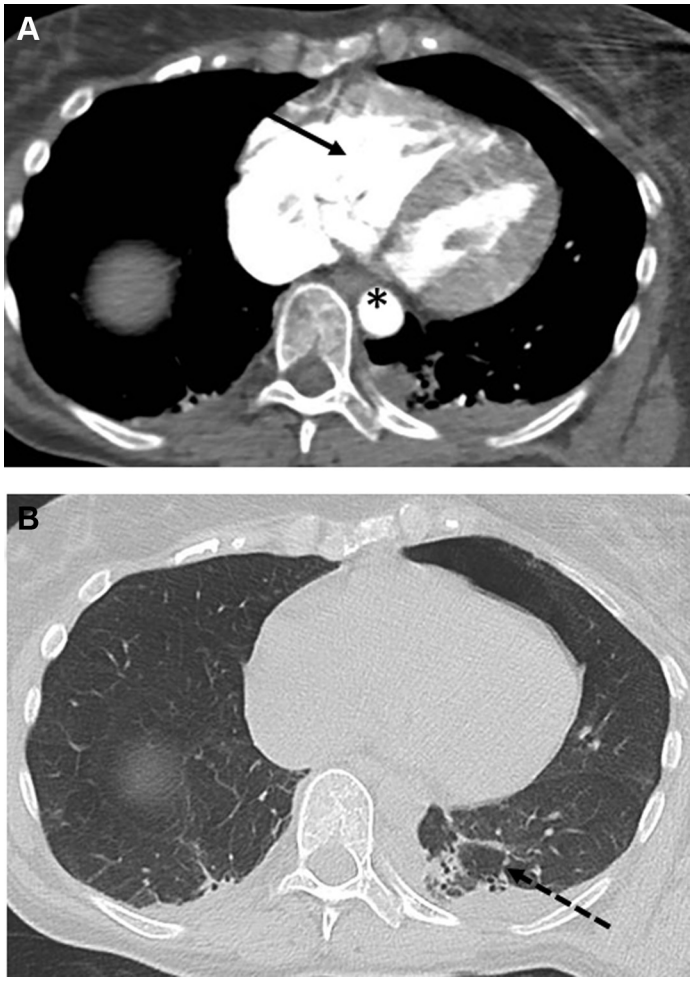
RV = right ventricle

SVR = systemic vascular resistance

TOF = tetralogy of Fallot

and dilatation of the right heart chambers were indicated (Figures 1A and 1B), the latter being confirmed by the QRS axis at 102 degrees as seen in the electrocardiogram (Figure 2). In the following hours, repeated episodes of sudden desaturation and hypotension were observed, and the patient required hemodynamic support with high-dose inotropes and vasopressors (norepinephrine 1 μg/kg/min). After the care team provided a report on the patient's critical condition, her relatives remembered that the patient had been diagnosed during childhood with a complete atrioventricular canal defect; however, her parents had refused surgery. Moreover, the family reported that the patient was asymptomatic in her daily activity, with modest dyspnea only in case of uphill walking. Therefore, being convinced of having no treatment option and without complaint of severe symptoms or limitations, she did not seek any medical evaluation during adulthood. No cognitive disability or syndromic disorder was reported. Only at the time of the accident was cardiovascular evaluation required.

FIGURE 1 Angio-Computed Tomography Scan



Pulmonary embolism was excluded, showing the enlargement of the ascending aorta (37 mm, *) and hypertrophy and dilatation or the right heart chamber **(A, arrow)** and interstitial pulmonary edema with atelectasis of the inferior lobes **(B, dashed arrow)**.

INVESTIGATIONS

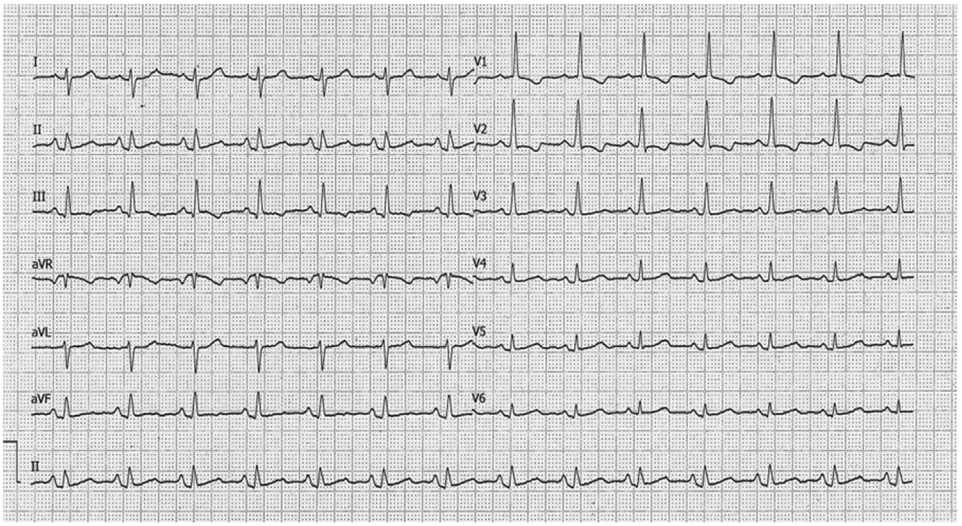
It was not possible to perform transthoracic echocardiogram because of subcutaneous emphysema.

A transesophageal echocardiogram was then performed and revealed a displacement and enlargement of the right chambers, biventricular hypertrophy, and an interventricular membranous septal defect, with a modest shunt (Figure 3). A trans-tricuspid gradient of more than 90 mm Hg was measured, leading the care team to hypothesize that the patient could have uncorrected congenital heart disease (CHD) complicated by pulmonary hypertension (Eisenmenger syndrome) (Figure 4). However, physical examination and chest radiograph at admission did not support this hypothesis, because no splitting of the second heart sound or adjunctive diastolic heart sound could be heard and no prominence of the left second arch could be observed, as shown in Figure 5. To better define the diagnosis and to eventually start a treatment with sildenafil or prostanoids, right heart catheterization was then performed. The examination documented normal values of pulmonary arterial pressure (20/8 mm Hg, mean 14 mm Hg) and wedge pressure (16 mm Hg), but elevated right ventricular (RV) pressures (116/6 mm Hg, mean 51 mm Hg) with no unexpected oxygen fall at the oximetric run (O_2 saturation 82% in the pulmonary artery, 76% in the RV, and 69% in the right atrium). Cardiac index by thermodilution estimation was 5 l/min/m^2.

A new echocardiogram finally demonstrated the combination of interventricular septal defect with pulmonary infundibular stenosis and overriding the aorta (Figures 6A to 6C). Focused revision of the images of the first CT scan with contrast medium confirmed the presence of a subpulmonary muscular sleeve (Figure 4D).

Despite the fact that no arrhythmic event was documented, the hypotension induced by sedatives and the impact of mechanical ventilation on the endothoracic pressures favored the inversion of the shunt. Thus, this required an attempt to elevate systemic blood pressure with norepinephrine in order to warrant oxygen saturation and rendered it impossible

FIGURE 2 Electrocardiogram at Admission



to wean the patient off inotropic support or mechanical ventilation.

MANAGEMENT

Tracheostomy was performed, and the patient was then transferred to the intensive care unit at a larger center, where she underwent surgical correction of TOF by infundibular resection and closure of the septal defect with a Dacron patch. The patient was rapidly hemodynamically stabilized with no more need of pharmacological support. The patient was subsequently discharged to a cardiac rehabilitation center after the humerus fracture was surgically corrected with external fixation nails.

Post-pericardiectomy pericardial effusion developed in subsequent weeks (**Figure 7**), that resolved with corticosteroids.

FOLLOW-UP

At the 6-month follow-up, the patient remained well compensated and underwent successful elective intervention for the removal of her forearm fixation device.

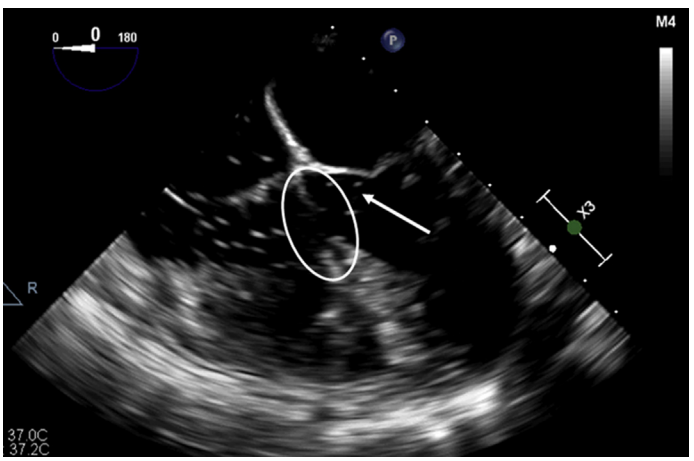
DISCUSSION

We have described a rare case of an adult woman with clinically silent TOF that was unmasked by major noncardiac surgery for a polytrauma and requiring urgent correction due to hemodynamic decompensation.

TOF is the most common form of cyanotic CHD worldwide (35 per 100,000 patients alive at birth [1,2]). This form of CHD is most frequently associated with patients reaching adulthood before the occurrence of symptoms.

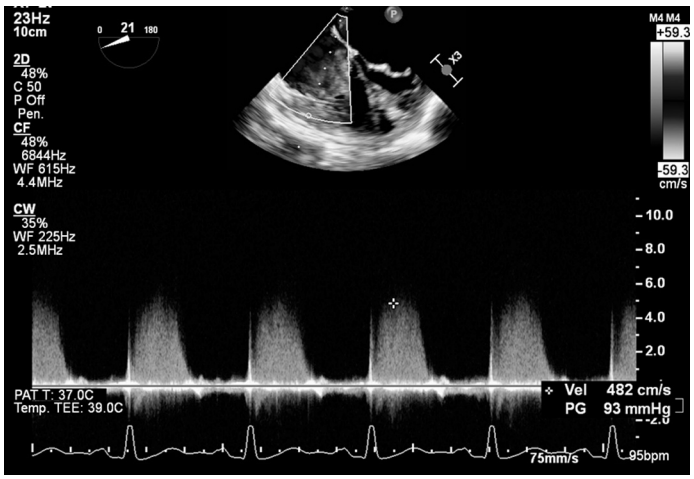
Despite the fact that life expectancy without surgery is usually short, rare silent cases in which TOF is detected in adults by chance have been previously described in the literature (3-5).

FIGURE 3 Transesophageal Echocardiogram



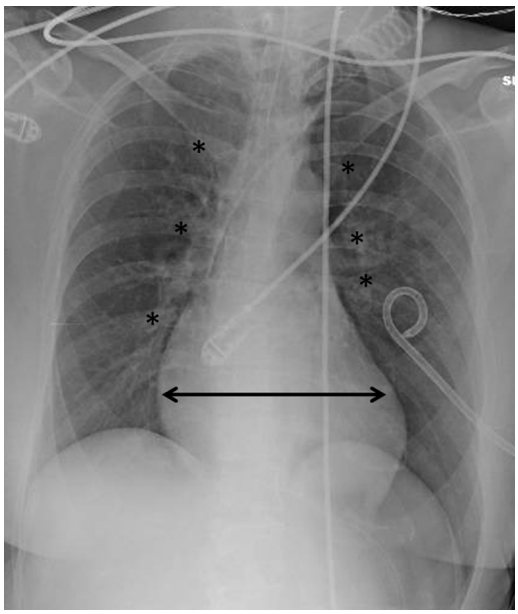
Ventricular septal defect with a modest passage of sonicated contrast is shown **(arrow)**.

FIGURE 4 Elevated Transtricuspid Gradient



Continuous Doppler measurement at transesophageal echocardiogram is shown.

FIGURE 5 Chest Radiograph at Admission



A normal distribution of the pulmonary vessels **(*)** and no enlargement of the cardiac shape **(arrow)**, hindering the hypothesis of pulmonary hypertension, is represented.

In fact, the presentation of the disease is quite heterogeneous, with severity of the cyanosis being related to the degree of the right-to-left shunting. The amount of blood shunted from right-to-left heart is dynamic, depending on the relative resistance in the systemic and pulmonary vasculatures. In some individuals, the pulmonary artery obstruction is fixed, most commonly involving the pulmonary valve, so the degree of shunt predominantly depends on the systemic vascular resistance (SVR). In other cases, the muscular outflow obstruction is more dynamic and delays the presentation of dyspnea only when RV failure has already occurred. Nevertheless, mortality of untreated TOF is very high, with patients having only a 3% chance of surviving until the age of 40 years (6).

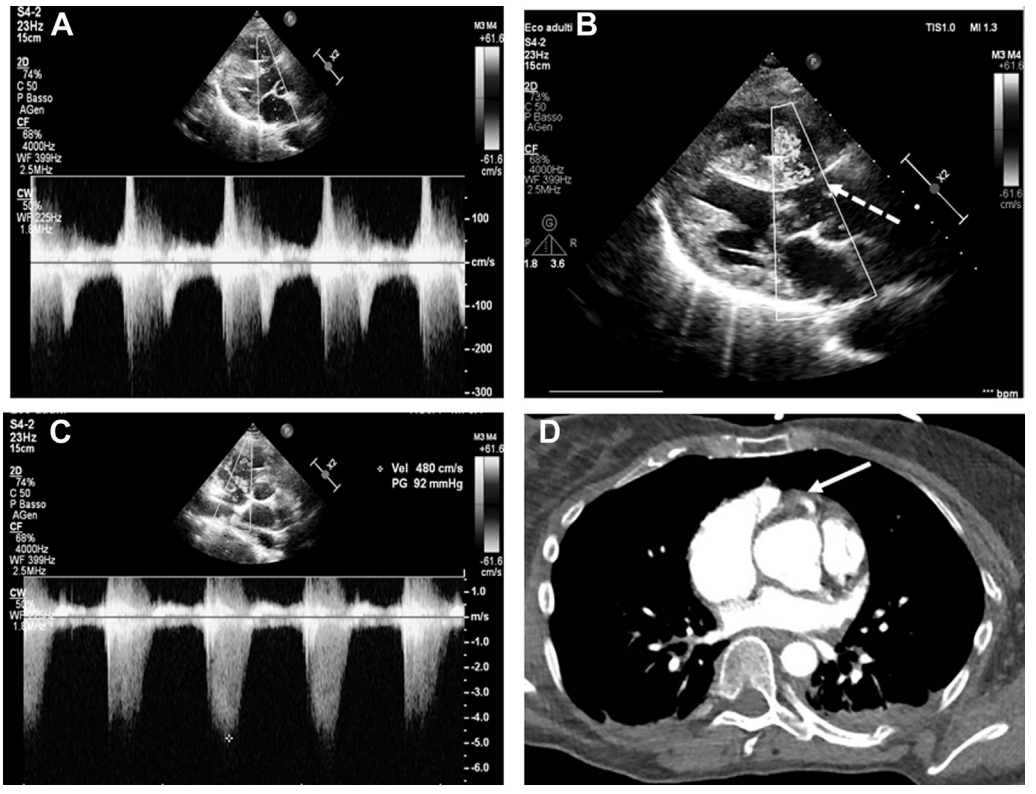
Even in the absence of clear symptoms, conditions affecting the hemodynamic balance can lead to an acute heart decompensation and extremely impaired prognosis.

In particular, general anesthesia for noncardiac surgery might unveil an uncorrected CHD and lead to severe complications, if not correctly managed (7). In effect, such procedures can modify the shunt through the septal defect by inducing peripheral vasodilatation and hypotension and by increasing endothoracic pressure with mechanical ventilation. In our case, the acute decompensation could have been ascribed to a vasospasm of the subpulmonary cuff or an imbalance between the systemic and pulmonary pressures, induced by hemorrhage, anesthesia, and pulmonary trauma. In fact, the reduction of systemic blood pressure and vascular resistances, as well as the obstacle to blood flow induced by positive pressure ventilation, could have increased the magnitude of the intracardiac shunt that was unexpected at the moment of the intervention. Indeed, despite the suspicion of TOF, which could have already been raised from the first CT scan, the patient's urgent condition that required the care team to focus on other issues did not allow for the early identification of the pulmonary outflow obstruction.

In fact, attentive preoperative evaluation of these patients is generally required for their safe surgical management; however, this can be impossible in an emergency procedure, as it was in this case. In effect, it is well described that patients with unrepaired cyanotic CHD often self-limit their activities and underreport their symptoms that are identifiable only through an objective assessment of the functional status via stress test.

Moreover, previous reports (4) have documented the safe peri-interventional management of adult

FIGURE 6 **Diagnostic Findings Consistent With Diagnosis of Tetralogy of Fallot**



Transthoracic echocardiogram showing the left-right shunt at continuous Doppler **(A)**, the color Doppler flow through the septal defect **(B, dashed arrow)**, the pulmonary outflow gradient at continuous Doppler **(C)**, and the presence of a subpulmonary muscular sleeve at CT scan **(D, arrow)**.

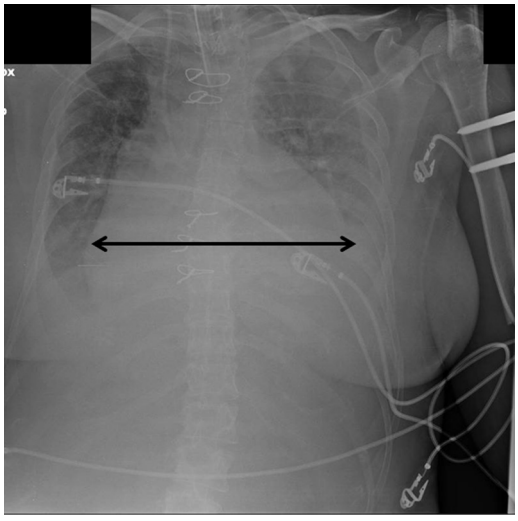
patients with clinically inapparent TOF, by an anesthetic strategy aimed at the maintenance of SVR and the reduction of pulmonary vascular resistance, including induction with ketamine or etomidate and the use of beta-blockers in order to lower the degree of pulmonary obstruction (8).

In fact, Faraoni et al. (5) clearly demonstrated in a large-scale registry that preoperative markers of critical illness (emergency procedure, inotropic support, mechanical ventilation, preoperative cardiopulmonary resuscitation, and acute or chronic kidney injury), the type of lesion (i.e., single-ventricle physiology), and the functional severity of the heart disease were strong predictors of in-hospital mortality in children undergoing noncardiac surgery, criteria that can probably apply also to adults with uncorrected CHD. In the latter, however, the prolonged RV overload can lead to unfavorable RV remodeling and dysfunction, being associated with an increased occurrence of symptoms, arrythmias, and poor prognosis (9,10).

Cardiac magnetic resonance imaging has become increasingly used as routine screening to evaluate RV function, dilation, scar, and interstitial fibrosis in adult patients with TOF (11,12). In this case, the presence of bone nails and hemodynamic instability prevented this kind of evaluation.

Regarding the correction of TOF, although early repair is well established in children (13), the indication for adults is still debated. Indeed, the onset of symptoms or signs of heart decompensation can certainly point in favor of a correction of the CHD, although requiring the attentive balance of operative risk, especially in the case of RV failure that could severely increase the perioperative risk without offering significant benefits on the prognosis or functional status. In effect, previous studies have shown that the main negative predictors of survival after surgical correction in adult TOF cases were polycythemia (14), RV dilatation (15), increasing age (16), and increased RV/left ventricular systolic pressures (16).

FIGURE 7 Pericardial Effusion



Chest X-ray showing the enlargement of pericardial shadow **(arrow)**.

CONCLUSIONS

Adult patients with TOF represent a challenging subset of patients due to the modest experience of most clinicians and the heterogeneity of the disease, which make understanding the patients' individual pathophysiology essential to customize the best therapeutic approach. In this case, we demonstrated that management by a multidisciplinary team, including anesthetists, surgeons, cardiologists, and cardiac surgeons, allowed us to safely discharge the patient with no residual disability.

AUTHOR DISCLOSURES

The authors have reported that they have no relationships relevant to the contents of this paper to disclose.

ADDRESS FOR CORRESPONDENCE: Dr. Monica Verdoia, Ospedale degli Infermi, ASL Biella, Eastern Piedmont University, Via dei Ponderanesi, 13900 Biella, Italy. E-mail: monica.verdoia@aslbi.piemonte.it.

REFERENCES

1. Moodie DS. Diagnosis and management of congenital heart disease in the adult. Cardiol Rev 2001;9:276–81.

2. Apitz C, Webb G, Redington A. Tetralogy of Fallot. Lancet 2009;374:1462–71.

3. Fairley SL, Sands AJ, Wilson CM. Uncorrected tetralogy of Fallot: adult presentation in the 61st year of life. Int J Cardiol 2008;128:e9–11.

4. Marulasiddappa V, Raghavavendra BS. Anesthesia for a rare case of uncorrected pentalogy of Fallot undergoing craniotomy and drainage of brain abscess. J Clin Diagn Res 2015;9:UD01–2.

5. Shaff DA, Raines DE, Vidal Melo MF, King ME, Misra S, Chen LL. Anesthetic management for transurethral resection of the bladder in a 74-year-old man with uncorrected tetralogy of Fallot. J Clin Anesth 2005;17:198–201.

6. Bertranou EG, Blackstone EH, Hazelrig JB, Turner ME, Kirklin JW. Life expectancy without surgery in tetralogy of Fallot. Am J Cardiol 1978;42:458–66.

7. Faraoni D, Vo D, Nasr VG, DiNardo JA. Development and validation of a risk stratification score for children with congenital heart disease undergoing noncardiac surgery. Anesth Analg 2016;123:824–30.

8. Stoelting RK, Dierdorf SF. Anesthesia and Coexisting Disease. 3rd ed. New York, NY: Churchill Livingstone, 1993:42–5.

9. Wald RM, Haber I, Wald R, et al. Effects of regional dysfunction and late gadolinium enhancement on global right ventricular function and exercise capacity in patients with repaired tetralogy of Fallot. Circulation 2009;119:1370–7.

10. Babu-Narayan S, Kilner P, Li W, et al. Ventricular fibrosis suggested by cardiovascular magnetic resonance in adults with repaired tetralogy of Fallot and its relationship to adverse markers of clinical outcome. Circulation 2006;113:405–13.

11. Geva T. Repaired tetralogy of Fallot: the roles of cardiovascular magnetic resonance in evaluating pathophysiology and for pulmonary valve replacement decision support. J Cardiovasc Magn Reson 2011;13:9.

12. Valente A, Gauvreau K, Assenza G, et al. Contemporary predictors of death and sustained ventricular tachycardia in patients with repaired tetralogy of Fallot enrolled in the INDICATOR cohort. Heart 2014;100:247–53.

13. Van Arsdell G, Maharaj G, Tom J, et al. What is the optimal age for repair of tetralogy of Fallot? Circulation 2000;102:Iii123–9.

14. Horer J, Friebe J, Schreiber C, et al. Correction of tetralogy of Fallot and of pulmonary atresia with ventricular septal defect in adults. Ann Thorac Surg 2005;80:2285–91.

15. Amorim S, Cruz C, Macedo F, Bastos PT, Goncalves FR. Tetralogy of Fallot: prognostic factors after surgical repair. Rev Port Cardiol 2005;24:845–55.

16. Murphy JG, Gersh BJ, Mair DD, et al. Long-term outcome in patients undergoing surgical repair of tetralogy of Fallot. N Engl J Med 1993;329:593–9.

KEY WORDS congenital heart defect, cyanotic heart disease, hemodynamics, tetralogy of Fallot