Journal of Radiology Nursing 38 (2019) 164–167

Contents lists available at ScienceDirect

Journal of Radiology Nursing

journal homepage: www.sciencedirect.com/journal/journal-of-radiology-nursing

Tetralogy of Fallot: Basic Imaging Findings and Management

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Keywords:
Tetralogy of Fallot
Congenital
Malformation
Right ventricular hypertrophy

A B S T R A C T

Tetralogy of Fallot (TOF) is one of the most common cyanotic, congenital heart disease with complex anatomic malformations and unknown etiology. With medical advancements, most patients in this cohort require early total correction and survive into adulthood. This review addresses various imaging modalities that help diagnose TOF, methods to correct and repair TOF-associated cardiac malformations, and the sequelae and long-term complications of this disease.

Background

Tetralogy of Fallot (TOF) is one of the most common causes of cyanotic cardiac disease and occurs in 3 of every 10,000 births, accounting for about 10% of all congenital heart diseases. TOF is a cardiac malformation that has a ventricular septal defect, obstruction of right ventricular outflow tract, aortic root overriding the ventricular septum, and right ventricular hypertrophy (see Figure 1) (Bailliard & Anderson, 2009). It was first described by Danish physician Dane Niels Stenson in 1671 with later correlation of pathologic and clinical manifestations by French physician Etienne-Louis Arthor Fallot in 1888 (Marios et al., 2014). The etiology of this condition remains elusive; however, TOF has been associated with many genetic abnormalities such as 22q11 deletion syndrome (DiGeorge syndrome) (Apitz et al., 2009), Down syndrome, trisomy 13 and 18 (Bailliard & Anderson, 2009). TOF is associated with various congenital anomalies including right aortic arch in about 25% of patients, atrial septal defect, multiple ventricular septal defect (VSD), patent ductus arteriosus, aortopathy, and aortic regurgitation (Dabizzi et al., 1990). Anomalous coronaries is another important association that occurs in 5-12% of patients which can impact the surgical repair especially when the left anterior descending artery originates from the right cusp and crosses over the right ventricular outflow tract. The clinical presentation is variable and depends on the degree of right ventricular outflow tract obstruction (RVOTO) with earlier presentation occurring in patients with more severe obstruction with significant cyanosis as the deoxygenated venous blood is not adequately reaching the lungs for proper oxygenation.

Imaging findings

Electrocardiogram/Chest X-ray

Initial evaluation of TOF includes obtaining an electrocardiogram, which will typically demonstrate right axis deviation with right bundle branch block and right ventricular hypertrophy with large R waves in anterior precordial leads and large S wave in the lateral precordial leads. Chest X-ray will demonstrate the boot-shaped heart due to displacement of the right ventricular apex secondary to right ventricular hypertrophy and narrowing of mediastinal silhouette given hypoplastic pulmonary outflow tract (Bailliard & Anderson, 2009).

Echocardiogram

Transthoracic echocardiogram or transesophageal echocardiogram (see Figure 2) should be obtained in all patients with TOF, with specifically suprasternal and parasternal views. Parasternal views can be used in evaluating right ventricular outflow tract, pulmonary valve, ventricular septal defect, and ascending aorta. Continuous wave Doppler is more useful than color Doppler as it allows assessment of pressure gradients across RVOTO. Suprasternal view can be used to evaluate aortic arch and systemic-pulmonary shunts. Echo also plays a pivotal role in assessing postsurgical changes after TOF repair, especially pulmonary regurgitation (PR), right ventricle (RV) dilation/dysfunction, assess residual VSD shunting, aortic root dilation, and any residual anomalies (Motta & Miller-Hance, 2012; Pooja et al., 2015; Snider & Silverman, 1981). Tricuspid annular peak systolic velocity is used to measure RV systolic function and correlates with CMR (Koestenberger et al., 2012). However, given that patients with TOF have diminished RV outflow tract contractility, global RV

Disclosures: None.

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https://doi.org/10.1016/j.jradnu.2019.07.003
1546-0843/$36.00/

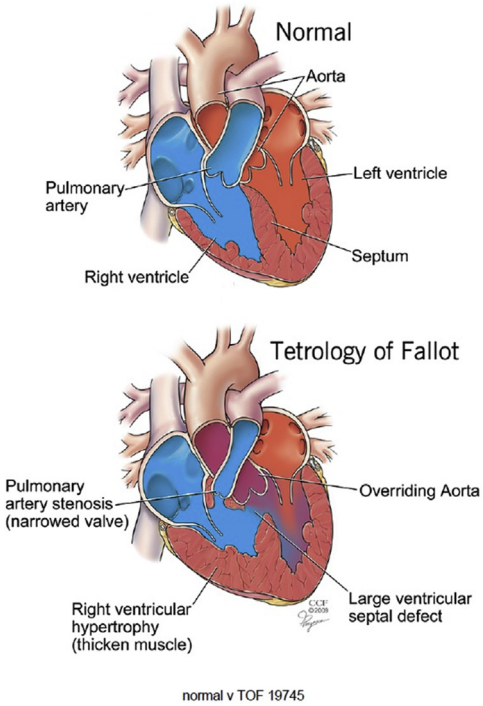

Figure 1. Comparison of the normal heart (top) vs TOF heart (bottom). The normal heart has the pulmonary artery that arises from the right ventricle and aorta that arises from the left ventricle with no mixture of oxygenated and deoxygenated blood. The TOF heart shows ventricular septal defect allowing for mixing of oxygenated and deoxygenated blood, an overriding aorta, pulmonary artery stenosis, and severe right ventricular hypertrophy. Reprinted with permission. TOF = Tetralogy of Fallot.

performance does not accurately reflect RV function and even 3D echo seems to underestimate RV volumes and may overestimate right ventricular ejection fraction (RVEF). Disadvantages to echo include low resolution and inability to perform real-time volume acquisition (Crean et al., 2011). Nonetheless, a small study conducted by Grewal et al. (2010) demonstrated that in 25 patients with severe PR secondary to TOF or pulmonary valvotomy, end diastolic volumes and end systolic volumes were similar when comparing 3D ultrasound to MRI. Axial measurements were used as a comparison for 3D ultrasound as they have less observer variability. The authors concluded that RVEF measurement with 3D

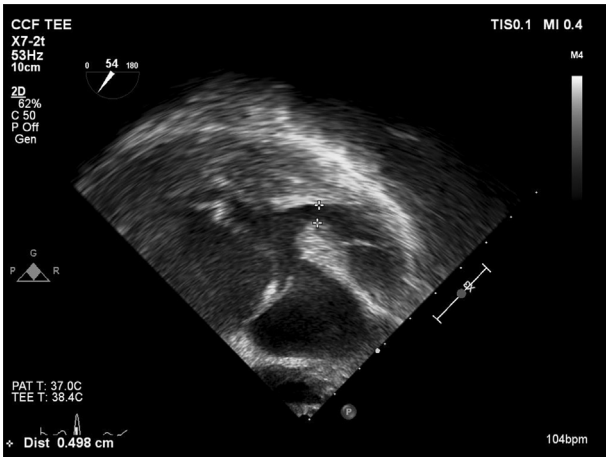

Figure 2. TEE of TOF showing the ventricular septal defect and subpulmonary stenosis, seen as narrowing below the pulmonary valve. Right ventricular hypertrophy can also be visualized. TOF = Tetralogy of Fallot; TEE = transesophageal echocardiogram.

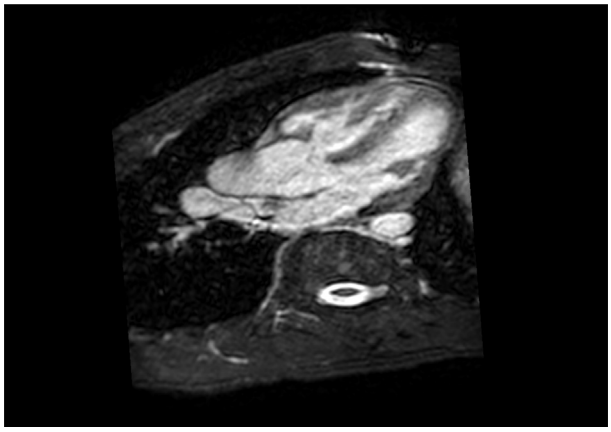

Figure 3. Cardiac MRI showing prominent right ventricle with overriding aorta and VSD. VSD = ventricular septal defect.

ultrasound was accurate and reproducible, as long as the entire RV was visualized (Grewal et al., 2010). As transducer technology continues to improve, 3D echo might be more useful in evaluating cardiac function in more complex cardiac anomalies.

Cardiac MRI

Cardiac MRI (CMR) is the gold standard in quantitative assessment of RV size and function and visualization of extracardiac structures in TOF (see Figures 3 and 4) (Mooij et al., 2008). It can be used in measuring pulmonary regurgitant fraction, ventricular volumes, great vessels, ratio of pulmonary and systemic blood flow through VSD, and myocardial viability (Geva, 2011; Kilner et al., 2010). CMR is also being used as a tool to assess postsurgical outcomes and risk stratification with studies such as those by Valente et al., which shows that addition of CMR measured RVEF and mass to volume ratio showed improved prediction of adverse outcomes such as mortality and sustained VT in patients with TOF (Kilner et al., 2010). One major disadvantage is that it cannot be performed in those with pacemakers or defibrillators. The 2018 American Heart Association and American College of Cardiologist guidelines for adults with congenital heart disease recommend CMR as diagnostic study for patients with TOF to evaluate ventricular size and function, pulmonary valve function, and pulmonary artery. Routine surveillance with CMR is recommended every 12-36 months based on patient's physiological stage (Stout et al., 2019). Cardiac MRI can be performed with cardiac devices, albeit with some limitations. Cardiac CT is also useful in evaluating similar features seen in cardiac MRI and can be used in those where MRI cannot be performed. However, major disadvantage includes contrast and radiation exposure and the temporal resolution is lower than in cardiac MRI.

Treatment and complications

It is estimated that >81% of patients with TOF reach adulthood after surgical repair (Cuypers et al., 2014). Without surgical repair, 25% of affected infants die in the first year of life and mortality increasing up to 40% by age 3 years and 70% by 10 years, with hypoxic spells as a major cause of death (Pooja et al., 2015). Most patients have palliative surgery in childhood such as systemic to pulmonary shunts to augment pulmonary circulation and thus oxygenation or complete repair. Systemic to pulmonary shunts include the Blalock-Thomas-Taussig shunt (Arnaz et al., 2018), Waterston shunt (Jaumin et al., 1979), and Potts shunt (Boshoff

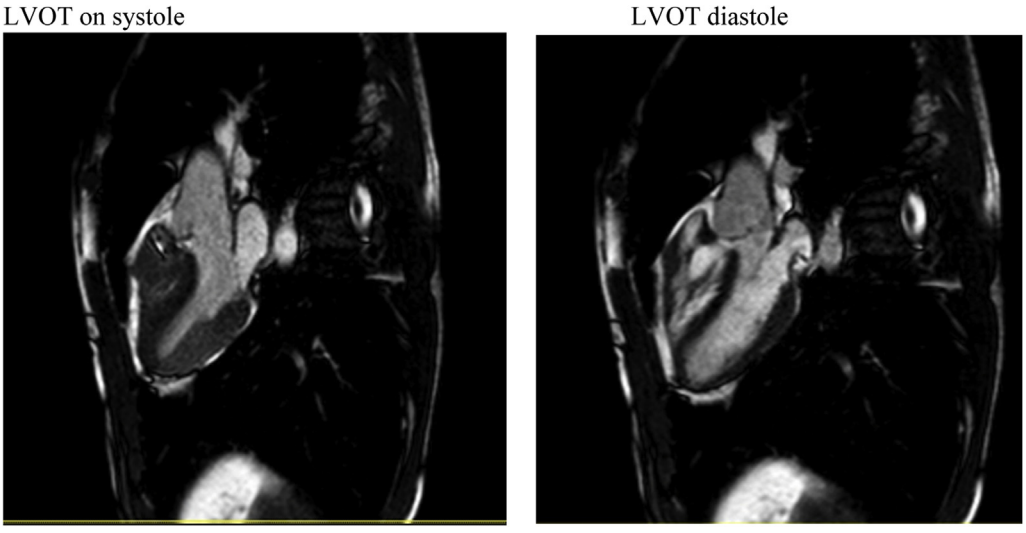

Figure 4. Ventricular septal defect and overriding aorta can be seen. Aorta is dilated getting blood from both the left ventricle and right ventricle.

et al., 2005). Complete repair entails patch closure of the ventricular septal defect and relieving the RVOTO. The latter may involve resection of subpulmonary stenosis, widening the right ventricular outflow tract, pulmonary valvotomy/valvectomy, or placing a conduit between the RV and pulmonary artery (Bove, 2017). Most patients require left heart catheterization to further assess underlying cardiac anatomy before surgery (see Figure 5). Complications of surgery include pulmonary valve regurgitation, tricuspid valve regurgitation with right ventricular enlargements, residual shunt, atrial arrhythmias, ventricular arrhythmias, and sudden cardiac death (Nieminen et al., 2007; Oechslin et al., 2000; Walsh, 2014). Studies have shown that the RV anatomy in TOF increases the chance of macroreentry circuits near the outflow tract leading to increased atrial and ventricular tachyarrhythmia (Kriebel et al., 2007; Khairy et al., 2010; Zeppenfeld et al., 2007). In addition, hemodynamic changes in TOF lead to RV and LV dysfunction, increasing the risk of polymorphic VT and ventricular fibrillation. The combination of tachycardia in patients with depressed ventricular function can lead to sudden cardiac death in this population (Walsh, 2014). In patients with sustained episodes of VT or at high risk for VT, ICD placement is preferred over drug therapy, with improved outcomes also seen in surgical or catheter ablation (Zeppenfeld et al., 2007). PR is the most common indication for reoperation, with tricuspid regurgitation secondary to RV dilation from PR as common finding (Holst et al., 2011; Warnes et al., 2008). Percutaneous techniques can be used in those with postsurgical residual lesions such as pulmonary artery stenosis, pulmonary regurgitation, and residual RV outflow tract obstruction. RV tract stenting and angioplasty for RVOTO, transcatheter balloon dilation, and percutaneous valve replacement are some techniques most commonly used (Quandt et al., 2017; Rothman et al., 1990).

Conclusion

Tetralogy of Fallot is a complex, congenital heart disease with four main anatomic abnormalities: right ventricular hypertrophy, ventricular septal defect, overriding aorta, and right ventricular outflow tract obstruction. Diagnosis of this disease is typically made with an echocardiogram; however, most recent guidelines also recommend cardiac MRI as a way of routine surveillance. Without surgical repair, mortality in this population increases steeply with hypoxia as a major cause of death. Latest advances in surgical and percutaneous techniques have improved mortality and quality of life in this population. As these patient cohorts continue to age and survive into adulthood, continued advancements in diagnosis, surveillance, and therapeutic interventions are needed to improve the care of TOF patients.

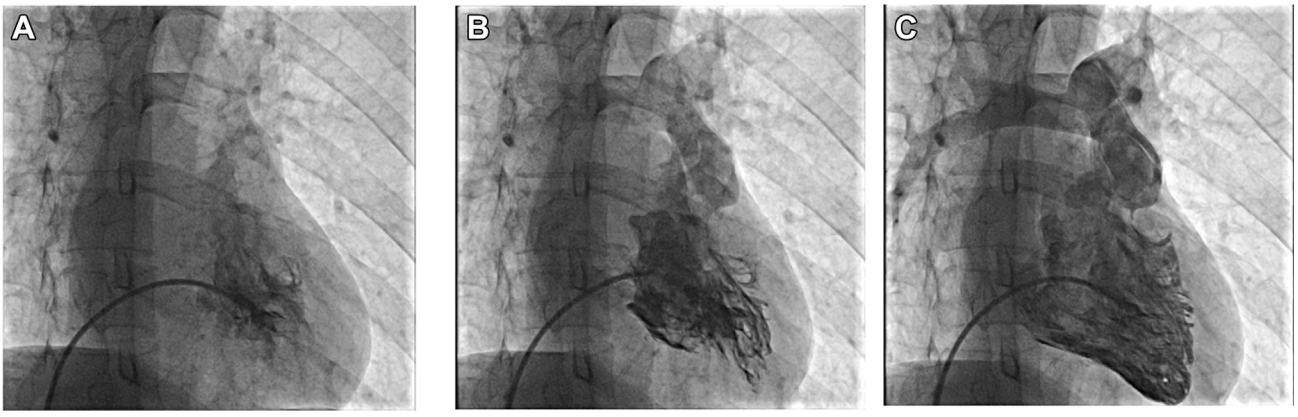

Figure 5. Angiogram showing trabeculated right ventricle with VSD and tricuspid regurgitation visualized by injection of dye into the right ventricle causes spillage of dye in the left ventricle and right atrium. (A) Injection of dye in the right ventricle. (B) Trabeculated right ventricle with VSD and tricuspid regurgitation visualized by injection of dye. (C) Dye from right ventricle causes spillage of dye in the left ventricle and right atrium. VSD = ventricular septal defect.

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