

PedsCases Podcast Scripts

This is a text version of a podcast from PedsCases.com on "Tetralogy of Fallot." These podcasts are designed to give medical students an overview of key topics in pediatrics. The audio versions are accessible on iTunes or at www.pedscases.com/podcasts.

Tetralogy of Fallot

Developed by Katie Girgulis, Dr. Andrew Mackie, and Dr. Karen Forbes for PedsCases.com.
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Introduction

Hello, my name is Katie Girgulis and I am a medical student at the University of Alberta. This podcast was developed with the help of Dr. Andrew Mackie and Dr. Karen Forbes. Dr. Mackie is a pediatric cardiologist at the Stollery Children's Hospital, and Dr. Forbes is a pediatrician and medical educator at the Stollery Children's Hospital. This podcast is about the cardiac condition Tetralogy of Fallot (ToF). For teaching on the general approach to pediatric heart murmurs, please check out the 'Evaluation of a Heart Murmur' podcast on PedsCases.com.

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Learning Objectives

By the end of this podcast, the learner will be able to:

- 1) Recognize the clinical presentations of ToF
- 2) Describe the four anatomical characteristics of ToF
- 3) Describe the pathophysiology of the murmur in ToF
- 4) Formulate initial steps when ToF is suspected
- 5) Delineate the treatment of hypercyanotic episodes
- 6) Summarize the definitive treatment for ToF

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Case – Baby Josh

Let's start with a clinical case: You are working with Dr. Smith, a family physician, during your family medicine rotation. Josh is a 4-month-old infant who is here for a well-baby check. Dr. Smith encourages you to start the history and physical exam on your own before she joins you in the room. All is going well until you notice a murmur while auscultating Josh's chest. Oh no! You are struggling to remember cardiology and murmurs. Let's quickly remind ourselves of some key points about heart murmurs.

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Brief Review of Innocent vs. Pathologic Murmurs

Before getting into the details of the case, let's do a brief review of how we distinguish innocent murmurs from pathological ones. A good mnemonic for features of innocent murmurs is the "Seven S's": **sensitive, short, single, small, soft, sweet, and systolic**. I will explain each of these briefly:

- Sensitive - the murmur changes with position or respiration
- Short duration - the murmur is not pansystolic
- Single - there are no clicks or gallops

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- Small - the murmur is limited to a small area and is non-radiating
- Soft - the murmur is of low intensity
- Sweet - the murmur is not harsh in quality
- Systolic - the murmur is limited to systole. Remember: diastolic and pansystolic murmurs are always pathological, whereas systolic ejection murmurs may or may not be pathological

In addition to the features of the murmur on auscultation, we need to look for symptoms and signs on history and physical exam, such as the following, that would raise concern for a pathological murmur:

- respiratory difficulties
- cyanosis
- poor feeding
- poor growth
- syncope
- family history of congenital heart disease or sudden cardiac death
- abnormal vital signs
- diminished or absent femoral pulses
- diastolic or pansystolic murmur
- high intensity murmur (grade ≥ 3)
- increased murmur intensity when the child is in an upright position
- harsh quality of the murmur
- abnormal S2 (i.e. not physiologically split)
- extra heart sounds (such as clicks or gallops)

Now, back to the case

Dr. Smith enters the room and asks you for a brief presentation about Josh.

From the history, you learned that Josh' parents have no big concerns. However, they have noticed that he seems to turn blue in the lips when crying very hard. These episodes resolve spontaneously but are occurring every few days. Josh is their fourth child. It was an uncomplicated pregnancy with routine prenatal care. He was born one week later than his due date from an induced vaginal delivery. He breastfeeds every 3 hours, generally well, but sometimes takes a long time to feed. Josh's parents report that he was a small baby (born at the 10th percentile), but is tracking along his length and weight growth curves. His immunizations are up to date.

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Thus, the main finding from the history is central cyanosis - Josh's parents described this as turning blue around the lips. Central cyanosis is concerning, as it suggests that deoxygenated blood is being pumped from the heart into circulation – somehow the blood is not getting fully oxygenated in the lungs. Feeding problems in an infant should also raise alarm, particularly if there is a history of prolonged feeding, diaphoresis with feeding, or an infant needing to stop frequently to catch their breath while feeding. These may be indicative of congestive heart failure, with pulmonary over-circulation, leading to shortness of breath and tachypnea. Or, it could be associated with lower oxygen saturation of the blood and therefore reduced exercise tolerance. It is useful to think about feeding as an infant's form of 'exercise'.

Let's move onto the physical exam. You and Dr. Smith work together to examine Josh, and your findings are as follows:

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Josh is content and well-appearing. His vital signs are normal other than a low oxygen saturation of 81%. There is a grade 3/6 harsh systolic crescendo-decrescendo murmur heard best along the left sternal border. The murmur radiates to the axillae and back. The examination of all other systems is normal.

Altogether, Josh's associated symptoms (cyanotic episodes) and abnormal vital signs (low O₂ saturation), as well as features of the murmur (harsh quality, high intensity, large radiation) should point you towards a pathological murmur rather than an innocent murmur. In fact, what has been described is a common presentation of Tetralogy of Fallot (ToF).

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Definition

'What is Tetralogy of Fallot?' - you might ask? ToF is a cyanotic congenital heart disease that arises due to problems during embryological development as the heart undergoes septation. It involves four anatomical characteristics:

1. **ventricular septal defect (VSD)**
2. **pulmonary stenosis**
3. **overriding aorta**
4. **right ventricular hypertrophy**

Next, we should explore why and how these anatomic features occur.

Normally, the ventricles are divided by a muscular septum that develops up from the floor of the ventricles and a membranous septum that develops down from the tissue that divides the outflow tract into the aorta and pulmonary trunk. These two join together to form the ventricular septum. In ToF, the outflow tract is divided unequally such that the aorta is much larger than the pulmonary tract, leading to features of an 'overriding' aorta and pulmonary stenosis. This shifts the membranous septum so it does not align properly with the muscular septum, creating a ventricular septal defect (VSD).

These anatomical characteristics create a system wherein the pulmonary circulation has high resistance, due to the stenosis, and the systemic circulation has relatively lower resistance. Remember that blood will follow the path of least resistance. As a result, the VSD allows de-oxygenated blood from the right ventricle to shunt directly to the aorta, to the lower-resistance circulation. It is important to note that you do not hear a murmur due to the VSD - the hole is typically very large, allowing pressures in the RV and LV to nearly equalize. Moreover, there is not a large pressure gradient driving flow across the VSD, which would cause an audible murmur. Instead, the murmur is due to the pulmonary stenosis – as blood is ejected through the narrow pulmonary tract during systole, a murmur is audible due to the turbulent flow. If the obstruction becomes more severe, the murmur may actually become quieter because more blood shunts across the VSD.

Lastly, the RV hypertrophy is not present at birth. It develops overtime as the right ventricle is exposed to systemic pressures due to the VSD.

History and Physical Exam for ToF

Let's review what we would expect on history and physical examination for an infant with ToF.

ToF is often diagnosed prenatally. However, when it is not, ToF commonly presents within the first **few months of life** as a **systolic murmur** with or without associated **hypercyanotic (Tet) spells** and **low oxygen saturation** – just like Josh. The Tet spells can occur when there is increased right ventricular outflow tract obstruction or pulmonary vascular resistance, or when there is decreased systemic vascular resistance – these promote more shunting from the right

ventricle to left ventricle, causing more mixing of oxygenated and deoxygenated blood, which is pumped out into the systemic circulation and leads to cyanosis.

Uncommonly, children with ToF can present with signs and symptoms of congestive heart failure if there is very minimal outflow obstruction and thus L-to-R shunting across the VSD, leading to pulmonary over-circulation. Symptoms and signs such as **respiratory difficulties, difficulty feeding, failure to thrive, tachycardia, tachypnea**, and **hepatomegaly** are suggestive of congestive heart failure. These patients are often referred to as “**pink-tets**”, as they do not experience significant cyanosis.

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Overall, your approach for ToF is the same as that for any other pediatric heart murmur. Through the history and physical exam, you should be looking for any features that are concerning for a pathological murmur. If you suspect congenital heart disease, you should try to classify the clinical presentation into one of three categories: cyanotic, congestive heart failure, or low cardiac output. Cyanotic suggests a right-to-left shunt; congestive heart failure suggests a left-to-right shunt; low cardiac output suggests an obstructive lesion. ToF fits into the cyanotic category.

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Investigations

Let's think about Josh again. We have established that his clinical presentation is highly suspicious for ToF. But what should you do now? What are the next steps?

Regardless of underlying diagnosis, suspected cyanotic congenital heart disease or concerns regarding a pathologic murmur require prompt **evaluation by pediatric cardiology** and evaluation with an **echocardiogram (ECHO)**. An ECHO allows for identification of the cardinal features, including the degree of obstruction of the RV outflow tract. Electrocardiograms (ECGs) are valuable tools in assessment of congenital heart disease, but are not sufficient alone for diagnosis. Please note: a chest x-ray does not need to be performed for children with a murmur, unless there is respiratory distress, as it is neither sensitive nor specific.

Case

So, what happened to Josh? As was previously mentioned, you and Dr. Smith agreed that Josh's had concerning features for a pathological murmur. Thus, you help write a referral to pediatric cardiology at the Stollery Children's Hospital. Josh's parents book an appointment in 2 weeks. In the meantime, you send Josh for an ECG. When the results become available, you and Dr. Smith sit down in her office to take a look. The ECG shows right axis deviation and right ventricular hypertrophy. A week later, the clinic receives a letter from the pediatric cardiologist. Josh underwent an ECHO, which confirmed the diagnosis of ToF.

Treatment

Let's move on to management.

Now that the diagnosis of ToF has been confirmed, Josh's parents have a lot of questions. They want to know about the long-term treatment of his ToF, but first, they want to know what they should do when Josh turns blue.

The management for Tet spells can be divided into two categories – supportive measures performed by the parent/guardian and medical interventions provided by a physician:

- **Supportive measures** that can be performed by a parent/caregiver:
 1. Place the child in a knees-to-chest position. This increases systemic resistance, reducing the right to left shunting

2. Have a caregiver hold the child. This prevents further agitation and may help him/her calm down
 - **Medical interventions:**
 1. Supply oxygen. This is important because low oxygen saturation is causing the cyanosis
 2. Morphine can help calm the child and also reduce pulmonary vascular resistance
 3. Phenylephrine, an alpha-1 adrenergic receptor agonist, is sometimes used in the hospital setting to increase systemic vascular resistance, therefore promoting blood flow through the pulmonary trunk to the lungs for oxygenation.

In terms of the long-term management of ToF, we need to consider **surgical repair** and **endocarditis prophylaxis**. Let's briefly discuss each one of these.

- First, surgical repair usually takes place within a few months of the diagnosis. This is the definitive treatment. It involves closing the VSD and enlarging the RV outflow tract. The procedure may be valve-sparing if the stenosis is mild to moderate, but in cases of moderate or severe stenosis, surgical management may affect the pulmonary valve and result in regurgitation post-operatively. These children will often need a pulmonary valve replacement later in life
- Second, Josh will require infective endocarditis prophylaxis before dental, surgical, or other invasive procedures, as his ToF and surgical repair puts him at risk for bacteria adhering to his cardiac endothelium. According to the American Heart Association and Canadian Dental Association, infective endocarditis prophylaxis is required for patients with:
 - prosthetic valve or material
 - history of infective endocarditis
 - specific types of congenital heart disease
 - unrepaired or incompletely repaired cyanotic congenital heart disease
 - completely repaired congenital heart defect with prosthetic material or device during the first six months after the procedure
 - any repaired congenital heart defect with residual defect at the site or adjacent to the site of a prosthetic patch or a prosthetic device
 - Thus, children with ToF require infective endocarditis prophylaxis preoperatively and for at least the first 6 months postoperatively (in some cases the prophylaxis is required lifelong).

Case conclusion

Let's conclude with our case. You are back with Dr. Smith for a family medicine elective, and are excited to see that Josh is coming in for his 1-year checkup. Josh's ToF was successfully repaired at 6 months of age. He is no longer having hypercyanotic Tet spells. He is growing well and meeting all his developmental milestones. Josh's parents hand you a letter from their most recent appointment with the pediatric cardiologist, which states that Josh is doing very well from their perspective. The cardiologist recommends stopping the endocarditis prophylaxis 6 months post-surgery. Josh's parents are very impressed with your clinical skills and wish you all the best in your future career (they suggest that you should become a pediatrician!).

Summary

Thank you for listening to the podcast, I hope you now have a better understanding of ToF. You should be able to:

- 1) Recognize the clinical presentations of Tetralogy of Fallot (ToF)
- 2) Describe the four anatomical characteristics of ToF
- 3) Describe the pathophysiology of the murmur in ToF
- 4) Formulate initial steps when ToF is suspected

5) Delineate the treatment of hypercyanotic episodes

6) Summarize the definitive treatment for ToF

Next, check out the companion case about ToF on PedsCases.com. Here you can test your knowledge by working through the case and answering practice questions.

This concludes the podcast on ToF.

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References

1) Up to Date

- Approach to the infant or child with a cardiac murmur
- Pathophysiology, clinical features, and diagnosis of Tetralogy of Fallot
- Management and outcome of Tetralogy of Fallot

2) http://www.rch.org.au/clinicalguide/guideline_index/Cyanotic_Episodes_Spells/

3) <http://bestpractice.bmj.com/best-practice/monograph/701/treatment.html>

4) https://www.cda-adc.ca/en/about/position_statements/InfectiveEndocarditis/

5) http://rourkebabyrecord.ca/pdf/RBR2014Nat_Eng_aqua.pdf

6) <http://www.aafp.org/afp/2011/1001/p793.html>