



HOLY C.O.W.!

IT'S...

Clinical Question of the Week #11
September 8th, 2008 through September
15th, 2008

Please e-mail your answers to Kuo, Tim, Wendy, and Kevin (klian@mednet.ucla.edu; tprovias@mednet.ucla.edu; wsimon@mednet.ucla.edu; kbreger@mednet.ucla.edu) by 0800 on Monday, September 15th, 2008. The resident or intern with the most correct answers at the end of each month will receive a prize!

Case: An 17-year-old man presents for initial intake evaluation at the Medical Subspecialties Clinic at UCLA. Unfortunately, however, his medical records have not yet been forwarded from his previous physician's clinic and his mother (who knows his medical history best) is wrangling for parking in the Medical Plaza lot. What he can recall is that his mother told him he had "fits" as an infant where he seemed out of breath and grunted and seemed ashen. He is status post a cardiothoracic surgery but can't describe any of the details of the procedure. His pre-operative anatomy is shown below.

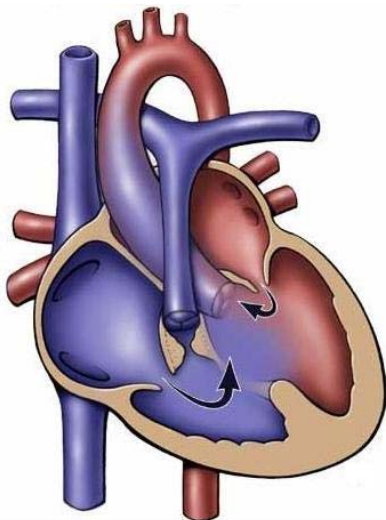


Figure 1 – Black arrows depict blood flow.

Questions:

1. What is the diagnosis?

Tetralogy of Fallot (TOF), named for French physician Etienne-Louis Arthur Fallot, who published a case series of patients with the disorder in 1888, is the most common cyanotic congenital heart defect and consists of the following: a) ventricular septal defect; b) pulmonic stenosis; c) overriding aorta; and right ventricular hypertrophy. The original description of the disease, however, has been credited to Niels Stensen in 1672. The tetralogy is also associated with multiple other congenital anatomic abnormalities

including left pulmonary artery stenosis, bicuspid aortic valve, right aortic arch, coronary artery anomalies, ASD (making the pentalogy of Fallot), and others.

The disorder occurs in approximately 3.9/100,000 live births in the United States, accounting for 7-10% of congenital heart disease cases, and occurs equally in males and females. There is an association with DiGeorge's syndrome and chromosome 22 deletions. Clinical presentation depends on degree of right ventricular outflow obstruction: children with severe obstruction present in the newborn period with severe cyanosis; children with minimal obstruction may present with heart failure due to pulmonary overcirculation; but children with moderate obstruction resulting in balanced pulmonary/systemic flow often do reasonably well and are diagnosed on echo for evaluation of murmur.

Physical examination reveals right ventricular impulse and occasional systolic thrill, the second heart sound is often single, and murmur of pulmonary outflow obstruction is heard. Chest x-ray reveals classic "boot-shaped heart" and EKG reveals RAE and RVH. Echocardiography is diagnostic and reveals the VSD, right ventricular outflow obstruction with severity, and other anatomic anomalies. (0.5)

2. What are the "fits," and how are they treated?

These are known as "tet spells" or "hypercyanotic spells," resulting from a dynamic increase in right ventricular outflow obstruction. Although the etiology of these spells is not entirely clear, there is variability in the degree of right ventricular outflow obstruction, and in these situations, the patient may become severely cyanotic.

Treatment of tet spells includes placing the patient in a knee-chest position to increase SVR, promoting blood flow into the pulmonary circulation rather than the aorta. More severe spells may require morphine (unknown mechanism) and saline bolus (increases RV filling and pulmonary flow). Finally, intravenous beta blockers to relax the right ventricular outflow tract and phenylephrine to increase systemic afterload and increase pulmonary circulation are also used. (1)

3. What is the surgery that the patient is likely to have had?

Blalock and Taussig reported the first palliative surgical correction of TOF in 1945, in which a connection between the aorta and pulmonary artery was made (now made with Gortex graft). Corrective surgery is now performed usually in the first year of life, and may be performed in the first three months of life if necessary. Additional corrections that may be performed in addition to the BT shunt include intracardiac closure of the VSD and reduction of the RV outflow obstruction. Short-term survival with operative repair now approaches 100% at six months. (0.5)

4. Name two long-term complications of this condition.

Complications of operated TOF include: 1) chronic pulmonic regurgitation which may result in CHF and require pulmonic valve replacement; 2) residual right ventricular outflow tract obstruction, which may require stenting or balloon dilation; 3) atrial tachycardias; 4) ventricular tachycardias with risk of sudden cardiac death (the major cause of death in adults, along with CHF); and 5) aortic root dilation, which may require aortic valve replacement. It is noted in long-term follow up of TOF patients that neurodevelopmental outcome is impaired, and may result in lower intelligence, difficulties with language tasks, and mild impairment of fine motor skills. As these patients have complex congenital heart disease with repair, they are also at risk for endocarditis, and also qualify for prophylactic antibiotics for complex dental procedures. (1)

** Notably, 2006 Winter Olympics Gold Medalist Shaun White, aka "The Flying Tomato," was born with Tetralogy of Fallot and is status post two reparative operations.