CASE 3.3
Congenital Heart Defects | Level 3

Brady S. Moffett

1. What is the subjective and objective evidence for the diagnosis of Tetralogy of Fallot?

SUBJECTIVE FINDINGS: Patient complained of chest pain and difficulty breathing, which was relieved by knee-to-chest maneuver. Patient reported instances of hypercyanotic ("Tet") spells, and lips and fingertips occasionally have turned blue.

OBJECTIVE FINDINGS: Findings include 89% oxygen saturation, echocardiogram demonstrating the pulmonary stenosis, overriding aorta, ventricular septal defect, and right ventricular hypertrophy. Patient has peri-oral cyanosis, is mildly cyanotic on exam, has a respiratory rate of 38 breaths per minute, and has a murmur.

2. Devise a pharmacologic regimen for prevention of a hypercyanotic spell secondary to Tetralogy of Fallot.

Traditional pharmacotherapy with the goal of preventing hypercyanotic spells in a patient with Tetralogy of Fallot is propranolol. It is theorized that this prevents infundibular spasm, which can block flow through the stenotic pulmonary artery and subsequently prevent right-to-left shunting of deoxygenated blood. Other beta-blockers may be used, but information is limited. The dose of propranolol in a patient with frequent hypercyanotic spells ranges from 1 to 4 mg/kg/day divided 3 to 4 times daily up to a maximum of 60 mg/day. The dose for this patient would be 35 mg orally, 3 times daily. The patient should be maintained on this regimen indefinitely or until propranolol therapy is no longer effective in preventing hypercyanotic spells and surgical intervention is indicated.

Adverse reactions to propranolol therapy include hypoglycemia, bradycardia, and reduced exercise tolerance. Heart rate should be evaluated after patients have been initiated on therapy. Patients should be educated to potential adverse reactions and report symptoms of exercise intolerance, lightheadedness, dizziness, or fainting. Some patients may experience neurologic symptoms such as vivid dreams/nightmares or cognitive dysfunction. If any of these reactions are severe enough to result in discontinuation of propranolol, other beta-blockers may be used. However, no data are available for use of other beta-blockers in prophylaxis of hypercyanotic spells in Tetralogy of Fallot.

3. Devise a pharmacologic regimen for the treatment of a life-threatening hypercyanotic spell secondary to Tetralogy of Fallot.

Nonpharmacologic therapy includes the use of a knee-to-chest maneuver. This increases peripheral vascular resistance and left ventricular pressure preventing right-to-left shunting of deoxygenated blood. Some patients will instinctively perform this maneuver, and they are colloquially known as "squatters." Pharmacotherapy is required if nonpharmacologic measures are ineffective in stopping the hypercyanotic episode and the patient is having life-threatening symptomatology, such as deteriorations in mental status or decreased perfusion to the periphery.

Pharmacotherapy for a hypercyanotic spell should focus on the reversal of right-to-left shunting of deoxygenated blood across the ventricular septal defect. The underlying physiology for decreasing the right-to-left shunt include increasing afterload and left ventricular end systolic pressure, decreasing pulmonary vascular resistance, and relaxing (or decreasing) infundibular spasm. Treatment should be based on the severity of the hypercyanotic spell and the potential for adverse reactions associated with pharmacotherapy. Patients should be admitted to the intensive care unit if the episode is deemed life threatening.

Initially, intravenous fluids at infusion rates to reflect maintenance fluid requirements are used to improve preload and cardiac output. Hypercyanotic spells can occur in patients secondary to other illnesses, which may result in dehydration. Inhaled oxygen via face mask should be used to improve oxygenation of blood and as a pulmonary vasodilator to decrease pulmonary vascular resistance.

If these modalities do not relieve the episode, care can progress to pharmacologic therapy. Morphine (0.05 mg/kg/dose) or ketamine (0.5 mg/kg/dose) can be useful to relieve pain and anxiety and to also lower pulmonary vascular resistance. Beta-blockers (enteral or intravenous) can be used to minimize infundibular spasm, reduce afterload, and promote myocardial relaxation. Propranolol can be administered at 35 mg (1 mg/kg/dose), or, if the patient is critically ill, esmolol intravenous continuous infusion initiated at 50 mcg/kg/min can be used. Phenylephrine (0.02 mg/kg/dose) can be used in severe cases to promote peripheral vasoconstriction thus increasing left ventricular pressure and preventing right-to-left shunting of deoxygenated blood.

For the patient in this case, initiation of normal saline intravenously at 75 mL/hr and oxygen 100% via face mask at 4 L/min would be appropriate first interventions. If the patient is still in pain, a dose of 1.8 mg of morphine intravenously would be appropriate. Pharmacotherapy should escalate in intensity based on the patient status. Ultimately, patients with severe activity restrictions or life-threatening hypercyanotic spells will have to undergo surgical repair.

4. Devise pertinent patient and caregiver education for the prophylaxis and treatment of hypercyanotic spell in Tetralogy of Fallot.

Patients and caregiver should be educated on the signs and symptoms of a hypercyanotic spell and to avoid activities or situations that have precipitated these events in the past, such as vigorous physical activity, dehydration, or fever. If the patient has a hypercyanotic spell, the knee-to-chest maneuver should be performed and the patient should be taken to the nearest emergency department.

Parents and patients should be educated on the indications for propranolol therapy and the importance of compliance to therapy to prevent future hypercyanotic events. Additionally, potential adverse reactions that may be associated with propranolol therapy (hypoglycemia, hypotension, bradycardia, fatigue/decreased exercise tolerance) should be highlighted to parents and patients.