HISTORY

15-year-old male.

CHIEF COMPLAINT: Decreasing exercise tolerance.

PRESENT ILLNESS: A heart murmur was noted in childhood, but subsequent medical care was sporadic. Easy fatigability and slight blueness of the lips and nail beds have been present all of his life.

Question: What type of heart disease is suggested by this history?
**Answer:** Congenital heart disease. A murmur early in life and the presence of cyanosis suggest a right-to-left shunt.

**PHYSICAL SIGNS**

a. **GENERAL APPEARANCE** - A normally developed male with mild generalized cyanosis. His fingers are shown below.

![Fingers Image]

**Question:** What is this abnormality?
**Answer:** Clubbing of the fingers.

Clubbing is characteristic of central cyanosis as well as pulmonary disease with hypoxemia. It takes years to develop in cases of congenital cyanotic heart disease. There is obliteration of the normal angle between the base of the nail and the skin, and in the most severe forms there are bony changes, i.e. hypertrophic pulmonary osteoarthropathy.

**Proceed**
b. **VENOUS PULSE** - The CVP is estimated to be 6 cm of H$_2$O.

**Question:** How do you interpret the venous pulse?
**Answer:** The estimated central venous pressure and wave form are normal.

c. **ARTERIAL PULSE - (BP = 90/60 mm Hg)**

**Question:** How do you interpret the arterial pulse?
**Answer:** The arterial pulse is normal. The blood pressure is also normal in this young patient.

d. PRECordial MOVEMENT

**Question:** How do you interpret the apical impulse?
**Answer:** The apical impulse is normal.

d. **PRECORDIAL MOVEMENT (continued)**

![Diagram of ECG and MID LEFT STERNAL EDGE]

**Question:** How do you interpret the impulse at the left sternal edge?
**Answer:** There is a systolic impulse that is sustained, reflecting a hypertrophied right ventricle. The absence of a palpable presystolic impulse suggests that right ventricular compliance is not significantly decreased. There is no impulse in the second left interspace suggesting that the pulmonary artery pressure is not elevated.

**e. CARDIAC AUSCULTATION**

![Audio waveform and ECG with annotations S1 and S2](image)

**Question:** How do you interpret the acoustic events at the upper right sternal edge?
**Answer:** Immediately following S1, there is an ejection sound (arrow). The ejection sound does not vary with respiration. This fact, and its location, identify it as aortic. This sound is followed by a medium-to-high frequency, early peaking, systolic crescendo-decrescendo murmur that extends to S2.

e. CARDIAC AUSCULTATION (continued)

![Cardiac Auscultation Diagram](image)

**Question:** How do you interpret the acoustic events at the upper left sternal edge?
**Answer:** There is a crescendo-decrescendo murmur that peaks early in systole and ends prior to a single second sound. P2 is inaudible due to low pulmonary artery pressure.

**e. CARDIAC AUSCULTATION (continued)**

![Cardiac auscultation waveform](image)

**Question:** How do you interpret the acoustic events at the lower left sternal edge?
Answer: The ejection sound (arrow) is well heard. The murmur is most intense in this area, consistent with right ventricular infundibular obstruction below the level of the pulmonic valve.

This murmur may be confused with that due to a ventricular septal defect (VSD).

f. PULMONARY AUSCULTATION

Question: How do you interpret the acoustic events in the pulmonary lung fields?
**Answer:** In all lung fields, there are normal vesicular breath sounds.

**ELECTROCARDIOGRAM**

**Question:** How do you interpret this electrocardiogram?
Answer: This ECG shows severe right ventricular hypertrophy, as evidenced by the presence of right axis deviation and an abnormally tall R wave in V1. Peaking of the P waves in leads II and V2 suggests right atrial enlargement. Note the change from an isolated R in V1 to an R/S deflection in V2, characteristic of equal right and left ventricular systolic pressure and wall thickness.

Proceed
Question: How do you interpret this chest X ray?
Answer: The PA chest X ray shows decreased pulmonary arterial vascularity consistent with a right-to-left shunt. The aortic arch is on the right (single arrow). Overall heart size is normal. The apex is uplifted, indicating right ventricular hypertrophy, and the left heart border is straight (double arrows), suggesting that the right ventricular outflow tract and main pulmonary artery segments are small.

Question: Based on the history, physical findings, electrocardiogram and chest X rays, what is your initial diagnostic impression?
**Answer:** The most likely diagnosis is tetralogy of Fallot, a condition characterized by right ventricular outflow tract obstruction and a large VSD.

Cyanosis in a teenager suggests this diagnosis. The murmur is consistent with outflow tract obstruction and, when coupled with the parasternal impulse, suggests it is right sided in origin. The maximum intensity of the murmur at the lower left sternal edge indicates that there is obstruction at the level of the infundibulum. The ejection sound is caused by a large volume of flow into the aorta reflecting the ventricular right-to-left shunt. The second sound is single, because right ventricular infundibular obstruction causes low pulmonary artery pressure, resulting in an inaudible P2.
Answer (continued): Routine laboratory studies further support the diagnosis of tetralogy of Fallot. The chest X ray demonstrates a right aortic arch, as seen in 25% of such patients, and the ECG shows right ventricular hypertrophy that is consistent with this diagnosis.

Proceed
LABORATORY

This patient’s hematocrit is 54%, and the hemoglobin is 18 gm/dl. Both values are elevated, indicating systemic arterial desaturation. These findings are typical of congenital heart disease with a right-to-left shunt.

**Question:** What noninvasive test would you now select to further define the diagnosis?
Answer: An echocardiogram.

TWO DIMENSIONAL STUDY

Question: How do you interpret this echocardiogram?

PARASTERNAL LONG AXIS VIEW

RV = Right Ventricle
Ao = Aorta
LV = Left Ventricle
LA = Left Atrium
S = Septum
**Answer:** The aorta is mildly enlarged and displaced anteriorly, so that it overrides the ventricular septum where a large ventricular septal defect is located. There is encroachment of the infundibular septum on the right ventricular free wall causing subpulmonary stenosis.

The patient’s Doppler study follows.
COLOR FLOW DOPPLER

PARASTERNAL LONG AXIS

Proceed

RV = Right Ventricle
Ao = Aorta
LV = Left Ventricle
LA = Left Atrium
S = Septum
LABORATORY  (continued)

The blue color indicates flow from the right ventricle to the left ventricle. The red color in the left ventricle indicates normal flow to the aorta.

Cardiac catheterization and angiography are not required in all patients but were performed prior to surgery in our patient.
**LABORATORY (continued): CATHETERIZATION DATA**

<table>
<thead>
<tr>
<th>Site</th>
<th>Pressure (mm Hg)</th>
<th>Oxygen Saturation (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Right Atrium</td>
<td>4 (mean)</td>
<td>65</td>
</tr>
<tr>
<td>Right Ventricle (Body)</td>
<td>90/8 (N=20/5)</td>
<td>67 (N=70)</td>
</tr>
<tr>
<td>Right Ventricle (Outflow)</td>
<td>40/8</td>
<td>67</td>
</tr>
<tr>
<td>Pulmonary Artery</td>
<td>15/3 (N=20/7)</td>
<td>69</td>
</tr>
<tr>
<td>Left Atrium</td>
<td>6 (mean)</td>
<td>98</td>
</tr>
<tr>
<td>Left Ventricle</td>
<td>90/7</td>
<td>93 (N=98)</td>
</tr>
<tr>
<td>Aorta</td>
<td>90/60</td>
<td>89</td>
</tr>
</tbody>
</table>

**Question:** How do you interpret these data?
**Answer:** Right ventricular systolic pressure is elevated and equal to that in the aorta, while pulmonary artery pressure is less than normal. Right sided pressures indicate two levels of obstruction, subvalvular and valvular. Equal right and left ventricular pressures reflect the large VSD. Oxygen saturations confirm a significant right-to-left shunt at the ventricular level. These data also suggest a small left-to-right shunt. Such bidirectional shunting is frequently seen in defects that cause predominant right-to-left shunts.

**Proceed**
Question: How do you interpret this angiogram?
**Answer:** There is significant right ventricular outflow tract narrowing (arrows). Contrast can also be seen in the aorta, indicating the right-to-left shunt.

These findings are indicative of tetralogy of Fallot, wherein four pathologic components are present: right ventricular outflow tract obstruction, large VSD, dextroposition (overriding) of the aorta and right ventricular hypertrophy.

Left heart angiography can further define the VSD, identify additional defects and identify coronary artery abnormalities. None were present in this case.

**Question:** How would you treat this patient?
**Answer:** This patient requires surgical correction to avoid the risks of hypoxemia, brain abscess, stroke and progressive right ventricular dysfunction. Lifelong infective endocarditis prophylaxis is required.

The results of surgery are excellent, with an expected operative mortality of less than 2% in a patient such as this.

**Proceed for Summary**
SUMMARY

Tetralogy of Fallot presents a spectrum of severity that is related to the degree of right ventricular outflow tract obstruction. In the most severe form, the right ventricular outflow tract is completely occluded (pulmonary atresia). These patients present early in infancy, usually shortly after birth, with severe cyanosis. At the other end of the spectrum, obstruction may be mild and, therefore, little or no cyanosis is present (pink tetralogy). Most often the obstruction is both subvalvular and valvular, although it may be only subvalvular. The VSD is always large, resulting in equal left and right ventricular pressures.

Proceed
SUMMARY (continued)

In tetralogy of Fallot, a drop in systemic arteriolar resistance can increase the right-to-left shunt. This suddenly decreases systemic oxygenation and may cause a hypoxic spell. The patient demonstrates hyperpnea, a decreased level of consciousness and increased cyanosis. Mild spells may cause a child to squat. Squatting increases systemic peripheral resistance, diminishing the degree of right-to-left shunt. In severe spells, unconsciousness, brain damage and death can occur.

Proceed
Hypoxic spells are more common in infants and toddlers. They are even more common in those with mild cyanosis or iron deficiency anemia. Patients with such spells demand immediate attention to alleviate the episode followed by surgical correction. If a contraindication to total correction exists, a systemic artery to pulmonary artery shunt (e.g., Blalock - Taussig procedure) should be carried out.

The typical gross pathology of tetralogy of Fallot follows.
Note the narrow orifice of the right ventricular outflow tract surrounded by hypertrophied muscle.

**PATHOLOGY**

**RVOT** = Right Ventricular outflow tract  
**CS** = Crista Supra-ventricularis  
**VSD** = Ventricular Septal Defect

Proceed for Case Review
To Review This Case of Tetralogy of Fallot:

The **HISTORY** is typical, with a heart murmur since childhood, easy fatigability and cyanosis.

**PHYSICAL SIGNS:**

a. The **GENERAL APPEARANCE** reveals a cyanotic young male with clubbing of his fingers.

b. The **JUGULAR VENOUS PULSE** is normal in mean pressure and wave form.

c. The **CAROTID ARTERIAL PULSE** is normal, as is the blood pressure.

Proceed
d. **PRECORDIAL MOVEMENTS** reveal a normal apical impulse, and there is a sustained systolic impulse over the hypertrophied right ventricle.

e. **CARDIAC AUSCULTATION** reveals an aortic ejection sound followed by a medium-to-high frequency, early peaking, systolic crescendo-decrescendo murmur heard best at the LLSE. The murmur suggests right ventricular outflow tract obstruction below the level of the pulmonic valve.

   The second heart sound is single because P2 is inaudible.

f. **PULMONARY AUSCULTATION** reveals normal vesicular breath sounds in all lung fields.

   Proceed
The **ELECTROCARDIOGRAM** shows right ventricular hypertrophy.

The **CHEST X RAY** shows decreased pulmonary arterial vascularity and a right aortic arch. The overall heart size is normal. The uplifted apex suggests right heart enlargement.

**LABORATORY STUDIES** reveal an increased hematocrit and hemoglobin. The echocardiogram shows an enlarged aorta overriding a large VSD and right ventricular infundibular stenosis. There is a right-to-left shunt across the VSD.

**CATHETERIZATION** shows equal left and right ventricular pressures, valvular and subvalvular pulmonic obstruction, and a right-to-left ventricular level shunt. Angiography shows significant narrowing of the right ventricular infundibulum and a right-to-left shunt across a large VSD.

**TREATMENT** is surgical correction. Infective endocarditis prophylaxis is required.