Challenge Cases

Undiagnosed Cardiomyopathy after Pregnancy

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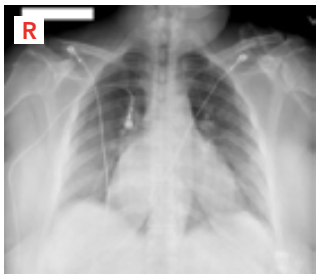
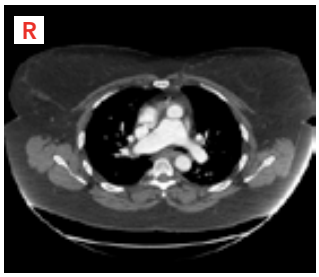
The patient is a 46-year-old woman with a past medical history of hypertension (HTN), congestive heart failure (CHF), chronic obstructive pulmonary disease (COPD), and an unknown congenital heart lesion, that presents by EMS with chest pain and shortness of breath (SOB). The patient reports acute onset of severe chest pain radiating into her back. The pain is described as sharp, pressure pain with a pleuritic component and no aggravating or alleviating factors. Associated symptoms include nausea, diaphoresis, and moderate SOB. The patient notes baseline chronic 3 pillow orthopnea, paroxysmal nocturnal dyspnea, and lower-extremity edema controlled with Lasix. EMS provided nebulized albuterol en route and placed her on continuous positive airway pressure (CPAP). The patient thinks her ejection fraction is 20% and reports that her heart lesion was never surgically corrected. Her current medications include albuterol, Advair, furosemide, lisinopril, metoprolol, and spironolactone.

On exam, the patient is in moderate distress; initial exam is somewhat limited secondary to BiPAP. Vital Signs include: BP 134/92 mmHg, HR 112 bpm, RR of 35/min, Sats 100% on BiPAP (EPAP: 5 cmH2O/IPAP: 12 cmH20). She has pink conjunctiva and appears well perfused. No appreciable jugular venous distension (JVD) is noted. Cardiac exam is regular rate and rhythm with no murmurs, gallops, or rubs. Her lungs have mild end-expiratory wheezing bilaterally, but are otherwise clear. Her abdomen is soft, nontender, and nondistended with no hepatosplenomegaly. She has no cyanosis, clubbing, or edema. Pulses are equal in her bilateral upper and lower extremities. Immediate concerns included acute coronary syndrome (ACS), CHF exacerbation, COPD exacerbation, pulmonary embolism, aortic dissection, and complication from the unknown congenital (acyanotic) heart lesion.

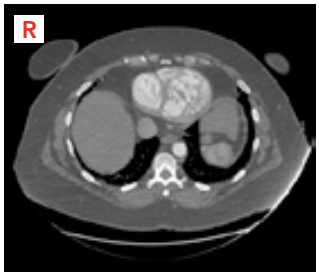
The initial EKG showed a sinus tachycardia with a left bundle branch block (LBBB). Comparison to previous ECG was unchanged. Selected views of her CXR, ECG, and CT chest are shown to the right.



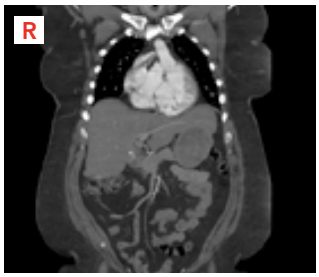
CXR: AP view



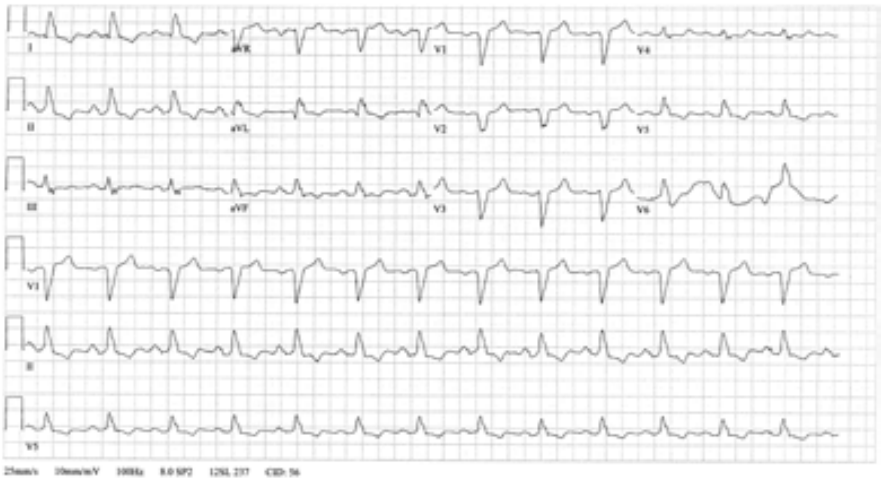
Chest CT: Axial



Chest CT: Axial, Distal



Chest CT: Coronal



ECG showing sinus tachycardia with LBBB

Q1: Based on the patient's history and provided imaging, what acyanotic congenital heart lesion does this patient likely have?

A. Atrioventricular septal defect
B. Coarctation of the aorta
C. Levo-transposition of the great arteries (L-TGA)
D. Pulmonary valve stenosis
E. Aortic stenosis

Q2: Patients with the isolated form of this lesion usually present later in life secondary to:

A. Acute coronary syndrome from anomalous coronary artery anatomy
B. Acute rupture of the tricuspid (systemic atrioventricular) valve
C. Sequela of pulmonary hypertension
D. Ventricular dysfunction of the morphologic right ventricle

The correct answer to Q1 is C.

All of the listed answers are acyanotic congenital heart lesions. The key to this patient's diagnosis of L-TGA is found on the CT scan. The patient's initial imaging study, CXR, is indicative of an undifferentiated cardiomyopathy based on the symmetric enlargement of the cardiopericardial silhouette. Further investigation with a Chest CTA show multiple findings indicating the left ventricle is in fact a morphologic right ventricle, with the aorta arising from this morphologic right ventricle. These findings as they relate to L-TGA will be discussed in more depth in the discussion portion.

The correct answer to Q2 is D.

For isolated L-TGA (no additional associated congenital cardiac defects) the admixture of deoxygenated and oxygenated blood does not occur and therefore no cyanosis is present. Thus, these patients can compensate and do quite well into adulthood with a morphologic right ventricle serving the function of the left ventricle. The problem occurs when the morphologic right ventricle (the systemic ventricle) becomes unable to handle the systemic workload. When this happens, the patient will usually manifest with either heart failure or arrhythmias.

We asked experts in both Cardiology and Radiology to comment on the pathophysiology and clinical course of a patient with Levo-Transposition of the Great Arteries (L-TGA), as well as the pertinent radiologic findings that are associated with the condition.

Discussion

Matthew Belford (Cardiologist)

Levo-Transposition of the great arteries (TGA), also known as "Congenitally Corrected TGA," is a rare condition which may not be diagnosed until well into adulthood. It was first described by Von Rokitansky in 1875.[1] This disease represents <1%[2,3] of all forms of congenital heart disease and represents a diverse patient population due to the presence of other associated cardiac abnormalities. The term "congenitally corrected" refers to the fact that despite anatomic abnormalities, blood flows in the physiologically correct direction. Anatomically, the morphologic left

ventricle (LV) is positioned to the right of the morphologic right ventricle (RV). However, because of atrio-ventricular and ventriculoarterial discordance, oxygenated blood flows from the lungs to the pulmonary veins to the left atrium to the discordant RV (via the tricuspid valve) then returns to the systemic circulation via the aorta (the discordant ventricular-arterial connection). L-TGA is distinct from D-TGA, which is a cyanotic lesion that constitutes 3% of all congenital heart disease.[3] D-TGA is the more common of the two, and is usually picked up at birth.

Many patients with L-TGA have another cardiac lesion[4,5] and estimates range as high as 90% of patients. These associated lesions are often what prompt identification to the clinician. Most common are tricuspid (the systemic atrioventricular) valve lesions (~90%), ventriculoseptal defect (VSD) (70-80%), pulmonary (LVOT) obstruction (~40%), then mitral valve abnormalities. A small percentage of patients (estimated between 10-20%) have isolated L-TGA without other lesions — it is these patients who may be clinically silent into adulthood, as exemplified by the patient in this case.

Bahram Kiani (Radiologist)

In this case, the first clue regarding the radiologic diagnosis of L-TGA is found on the presenting chest radiograph, which demonstrates an enlarged and centrally positioned cardiopericardial silhouette (Figure 1a). A normal chest radiograph is shown for comparison in Figure 1b. Immediate considerations of symmetric enlargement of the cardiopericardial silhouette on a chest radiograph include: pericardial effusion, acquired cardiomyopathy, and congenital cardiac abnormalities with mesocardia.

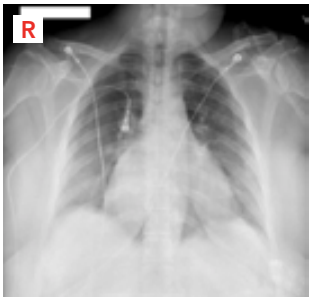


Figure 1a (patient)

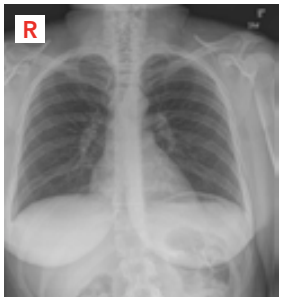


Figure 1b

Subsequent CT chest abdomen pelvis (CAP) with contrast excludes pericardial effusion, but demonstrates several morphologic abnormalities of the heart and great vessels of the mediastinum. The ascending thoracic aorta (Ao) is anterior and left of the main pulmonary artery (PA) (Figure 2a), rather than to the right of the main pulmonary artery as would be seen in a normal patient (Figure 2b).

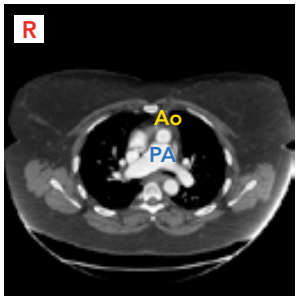


Figure 2a

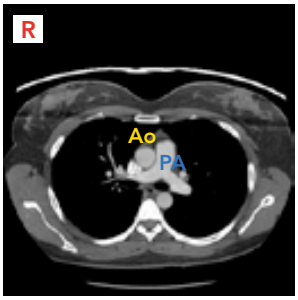


Figure 2b

A coronal reformatted image from the CTs demonstrates the presence of a moderator band (MB) and trabeculations (Tr) in the left-sided ventricle, indicating that this ventricle is, in fact, a morphologic right ventricle (Figure 3a). The aorta arises from this morphologic right ventricle (mRV), and lies superior to the main pulmonary artery that arises from the morphologic left ventricle (mLV) (Figure 3b).

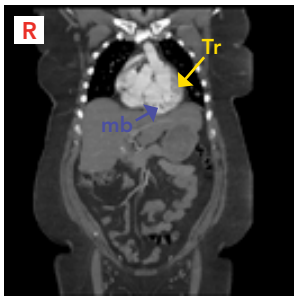


Figure 3a

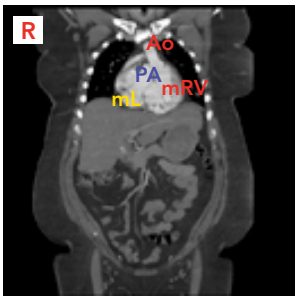


Figure 3b

The patient's right-sided ventricle (morphologic left ventricle) has a thicker lateral wall and essentially no trabeculations (characteristics of a morphologic left ventricle) (Figure 4a),

whereas in a normal patient the right ventricle would have a thinner wall and trabeculations (Figure 4b).

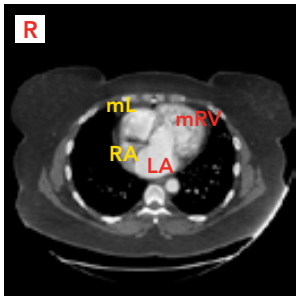


Figure 4a

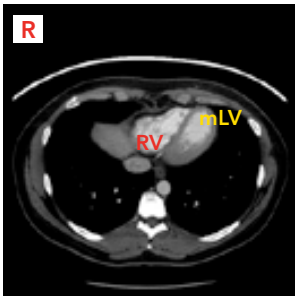


Figure 4b

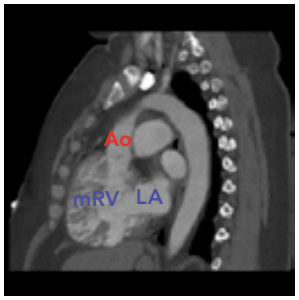


Figure 5

Finally, the sagittal plane reformat image shows the left atrium (LA) draining into the morphologic right ventricle (mRV) in addition to the aorta (Ao) arising from the morphologic right ventricle (mRV) (Figure 5). In this case, cardiomegaly with left atrial (LA) enlargement is also seen, commensurate with the clinical picture of systemic ventricular dysfunction.

Matthew Belford (Cardiologist)

From a clinical perspective, patients with isolated L-TGA will frequently present with one of the sequelae of discordant ventricular positioning—most frequently heart failure or underlying arrhythmia.[2] The cause of this progressive heart failure is not well understood, but it may be due to a mismatch of perfusion and supply/demand. In these patients the morphological right ventricle serves as the systemic ventricle and is subjected to left-sided, systemic blood pressure. However, this ventricle only receives perfusion by a single right coronary artery. These patients progressively develop failure symptoms over decades and are worse in the patients with associated lesions. Even in the group without associated lesions (such as the patient described in this case), 1/3 have heart failure symptoms by their 5th decade of life.[4]

Early work regarding surgical repair for L-TGA centered only on patients with other associated cardiac lesions. Presently, isolated L-TGA is surgically repaired, and the focus is on anatomic correction, structuring the morphologic LV as the systemic pump. This has improved long-term outcomes in these patients. To prevent postoperative LV failure, these patients may require "training" of the LV for it to be adequately hypertrophied to handle systemic pressures. This is accomplished by placing a band on the pulmonary artery to increase the afterload. After a period of time a "double repair" which includes a venous switch procedure, as well as an arterial switch operation, is performed.[2] Concomitant repair of a VSD, LVOT outflow obstruction or a regurgitant tricuspid valve may be performed if needed. As with patients in the preoperative state, worsened tricuspid regurgitation appears to portend worse outcomes.[4]

References:

1. Von Rokitansky K. Pathologisch-anatomische Abhandlung.Vienna: W. Braumuller,1875:83-6.
2. Warnes, C.A., Transposition of the great arteries. Circulation. 2006. 114: p. 2699-2709
3. Connelly M.S., et al., Congenitally Corrected transposition of the great arteries in the adult: functional status and complications. J Am Coll Cardiol, 1996. 27(5): p. 1238-43
4. Graham, T.P. Jr, et al., Long-term outcome in congenitally corrected transposition of the great arteries: a multi-institutional study. J Am Coll Cardiol, 2000. 36(1): p. 255-61.
5. Presbitero P, et al. Corrected transposition of the great arteries without associated defects in adult patients: clinical profile and follow up. Br Heart J 1995. 74(1): p. 57-9.
6. CT Atlas of Adult Congenital Heart Disease by Wojciech Mazur, Marilyn J Siegel, Tomasz Miszalski-Jamka, Robert Pelberg. Publisher: Springer. Publication Date: July 2, 2013.