Focused patient history and workup

- 14 yo previously healthy female who presented 6/5 from OSH with shortness of breath, orthopnea, and epigastric pain
- Sleeping upright on 2-3 pillows for the last week
- Family history negative for cardiac and respiratory disease
- D-dimer 622 and BNP 1,725 at OSH
- EKG demonstrated sinus tachycardia and CTA chest with and without contrast notable for cardiomegaly, interstitial pulmonary edema, and small bilateral pleural effusions from OSH
Focused patient history and workup

- Physical exam remarkable for hyperdynamic heart sounds and desaturations to low 90s
- CBC, BMP, Troponin unremarkable on admission
- Pro-BNP elevated to 13,800
- EKG unremarkable
List of imaging studies

- XR Chest (portable AP)
- Echocardiogram
- MRI Cardiac Morphology with and without contrast
AP Portable Chest X-ray

Findings?
AP Portable Chest X-ray

Findings are concerning for heart failure.

- Pulmonary vascular congestion
- Enlarged cardiac silhouette
- Small bilateral pleural effusions
Transthoracic Echocardiogram (parasternal long axis)

Findings?
Transthoracic Echocardiogram (parasternal long axis)

Echo findings: Severe dilated cardiomyopathy, severely diminished LV systolic function (EF 29%), severely dilated LV, moderately dilated LA, moderately dilated inferior vena cava
MRI Cardiac Morphology With and Without Contrast (coronal view)

Findings?
MRI Cardiac Morphology With and Without Contrast (coronal view)

MRI Findings:
- Dilated cardiomyopathy
- Global hypokinesis
- No evidence of ischemia
- Mitral insufficiency
- Ejection fraction 19%
Patient treatment and outcome

- Work up negative for inborn errors of metabolism
- Infectious work up remarkable for positive EBV IgG, otherwise negative
- Diagnosis: Idiopathic dilated cardiomyopathy
  - Complicated by HFrEF
- Treated with furosemide, enalapril, carvedilol, spironolactone
- 6/28: Left ventricular assist device placed
- 7/18: Discharged
- 8/26: Started high school!
- 10/1: Underwent heart transplant
- 10/10: Discharged!
  - Per Genetics, found to have pathogenic variant in *TNNT2*, which is associated with dilated and hypertrophic cardiomyopathy
AP Portable Chest X-ray: Post LVAD placement

- ET tube
- Swan g anz catheter
- Chest tube
- Sternotomy wire
- Left ventricular assist device
AP Portable Chest X-ray: Post cardiac transplantation

- ET tube
- Swan ganz catheter
- Mediastinal drain
- Chest tube
- Heart transplant
Discussion: Dilated Cardiomyopathy

• **Diagnosis:**
  - Evidence of both dilation and impaired systolic function of the left or both ventricles
  - Imaging: chest x-ray, echocardiography
    - Consider MRI or cardiac catheterization

• **Causes:**
  - Idiopathic – 50%
  - Other – 10%
  - Myocarditis – 9%
  - Ischemic heart disease – 7%
  - Infiltrative disease – 5%
  - Others: Peripartum cardiomyopathy, hypertension, HIV infection, connective tissue disease, substance abuse, Doxorubicin
Discussion: Dilated Cardiomyopathy

- Chest x-ray
  - Sensitivity for cardiomegaly: 40%
  - Specificity for cardiomegaly: 91%
  - Radiation dose: 0.1 mSv
  - Cost:
    - With health insurance: $0-$50
    - Without health insurance: $200-$400

- Echocardiogram
  - Sensitivity: 100% (if 3D)
  - Specificity: 100% (if 3D)
  - Radiation dose: None
  - Cost:
    - With health insurance: $0-$500
    - Without health insurance: $1,000-$3,000
Common Findings on Chest X-ray?

- Pulmonary vascular congestion
- Enlarged cardiac silhouette
- Small bilateral pleural effusions
- Kerley B lines
- Peribronchial cuffing
- Fluid in fissures
Common Findings on Transthoracic Echocardiogram?

Echo findings: Dilated cardiomyopathy, diminished LV systolic function (ie. reduced ejection fraction)
Discussion: Dilated Cardiomyopathy

• Epidemiology
  • ~10,000 deaths and 46,000 hospitalizations yearly in US
  • Pediatric cardiomyopathy incidence: 1 in every 100,000 children annually
    • Dilated cardiomyopathy- 50-60%
    • Hypertrophic cardiomyopathy- 25-40%
    • Left ventricular noncompaction- 9%
    • Restrictive/other- 3%

• Top indication for cardiac transplant
• Most patients are 20-60 years old
Discussion: Dilated Cardiomyopathy

• Presentation:
  • Incidental finding
  • Heart failure – dyspnea, orthopnea, paroxysmal nocturnal dyspnea, lower extremity edema, etc
  • Arrhythmia, thromboembolism, sudden death

• Heart failure presentation by age group:
  • Infants – poor weight gain, low volume feeds, tachypnea while feeding, easily fatigued
  • Young children – chronic cough, wheezing, abdominal pain, nausea/vomiting, failure to thrive, inability to keep up with peers
  • Older children – dyspnea, peripheral edema, chest pain, palpitations, abdominal pain, reduced appetite
## Discussion: Dilated Cardiomyopathy

### Stages of heart failure in infants and children and recommended therapy

<table>
<thead>
<tr>
<th>Stage</th>
<th>Definition</th>
<th>Examples</th>
<th>Therapy</th>
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</table>
| A     | Patients with increased risk of developing HF, but with normal cardiac function and chamber size | Exposure to cardiotoxic agents  
Family history of heritable cardiomyopathy  
Univentricular heart  
Congenitally corrected transposition of the great arteries | None |
| B     | Patients with abnormal cardiac morphology or function, with no symptoms of HF, past or present | Aortic insufficiency with LV enlargement  
History of anthracycline exposure with decreased LV systolic function | ACE inhibitor for patients with systemic ventricular dysfunction |
| C     | Patients with structural or functional heart disease, and past or current symptoms of HF | Symptomatic cardiomyopathy  
Congenital heart defect with ventricular pump dysfunction | ACE inhibitor and an aldosterone antagonist; oral diuretic therapy as needed for fluid overload; low-dose digoxin if needed for additional symptom relief  
After a few weeks of stability, a beta blocker is added in patients with persistent LV dilation and dysfunction |
| D     | Patients with end-stage HF requiring specialized interventions | Marked symptoms at rest despite maximal medical therapy | Pharmacologic therapy consists of intravenous diuretics and/or inotropes  
Other interventions may include positive pressure ventilation, cardiac resynchronization therapy, mechanical circulatory support, and heart transplantation |

HF: heart failure; LV: left ventricular; ACE: angiotensin-converting enzyme.
Wrap Up

• When there is concern for dilated cardiomyopathy, the initial imaging modalities include a chest x-ray to evaluate for signs of heart failure and an echocardiogram to evaluate for dilation and systolic dysfunction.

• Heart failure in the pediatric population can primarily present with GI symptoms:
  • Eg. Nausea/vomiting, reduced appetite, anorexia, abdominal pain.

• Dilated cardiomyopathy is the top cause of cardiomyopathy in this population.
References


References


