RADY 401 Case Presentation

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Focused patient history and workup

- 5 hour old male born at 40w3d via uncomplicated SVD to G2P1011 mom with pregnancy complicated by fetal atrioventricular septal defect and dextrocardia on prenatal echo
- APGARS 8 and 9, no resuscitation required
- Vitals: T 37.2°C, HR 176, RR 60, BP 54/46 with MAP 50, SaO2 95%
- Physical exam with mild acrocyanosis and fixed split S2 with heart sounds primarily heard on right. Otherwise warm and well perfused with lungs CTAB and no respiratory distress. Pulses 2+ bilaterally. Abdominal exam benign. Neurologically intact.
- Normal karyotype and prenatal / postnatal microarray
- Admitted to NICU for further workup and management given inadequate visualization of dextrocardia and AVSD on prenatal echo
Dextrocardia

- Isolated dextrocardia (rare)
  - No associated vital organ abnormalities

- Dextrocardia with partial situs inversus
  - Some organs are on opposite side of body but usually function normally

- Dextrocardia with situs inversus totalis
  - All vital organs in chest and abdomen are on opposite side of the body and may be abnormally functioning

- Dextrocardia with heterotaxy
  - Some or all vital organs are misplaced and possibly absent or partially developed. Commonly associated with cardiac malformations and considerable morbidity and mortality

- Dextrocardia with Kartagener syndrome

- Associated congenital heart defects
  - Double outlet right ventricle, Endocardial cushion defect, Pulmonary stenosis, Tetralogy of Fallot, Transposition of the great arteries, Ventricular septal defect
List of imaging studies

- Chest radiograph
- Abdominal radiograph
- Abdominal ultrasound
- Upper GI series
Imaging studies from PACS 1: AP supine portable CXR

- Trachea midline
- Right sided cardiac silhouette, consistent with dextrocardia. Appears prominent but possibly due to hypoinflation or AP technique
- Possible right sided gastric bubble, concerning for situs inversus
- Paracentral opacities: atelectasis vs. mild vascular congestion
- Borderline hypoinflation
- Sharp costophrenic angles bilaterally
Imaging studies from PACS 2: AP supine portable AXR

Right sided cardiac silhouette

Possible right sided gastric bubble

Streaky perihilar and bibasilar opacities, consistent with pulmonary vascular congestion

Liver in left upper quadrant, suggestive of heterotaxy and situs inversus totalis

Non-obstructive abdominal bowel gas
Imaging studies from PACS 3: Abdominal ultrasound

Enlarged liver, mostly present in LUQ but crosses midline and present to a lesser extent in RUQ, more consistent with heterotaxy than situs inversus totalis

Left sided gallbladder, no intraluminal gallstones or sludge

Hepatic veins
Right atrium
IVC
Aorta
Common bile duct without dilation
Imaging studies from PACS 3: Abdominal ultrasound

- Pylorus, present in RUQ
- Gastric antrum in RUQ
- Small RUQ spleen, closer to asplenia. More consistent with right sided heterotaxy than situs inversus totalis
- Right kidney, normal in size, shape, and echogenicity. No hydronephrosis, renal mass, or calcification
- Right adrenal gland, size within normal limits for age
Imaging studies from PACS 4: Upper GI series with fluoroscopy and radiography

Esophagus with normal contour and peristalsis. No gastroesophageal reflux.

Stomach in right hemiabdomen, empties readily.

Duodenum and duodenojejunal junction in right hemiabdomen, consistent with malrotation. No evidence of obstruction or volvulus.
Patient treatment or outcome: Heterotaxy with malrotation

• Echo (not shown here): Hypoplastic LV and aorta, complete AV canal defect with common AV valve, TAPVR, double outlet RV, dilated RV. Normal RV and LV systolic function

• Diagnosed with heterotaxy with malrotation given dextrocardia with dextroversion, TAPVR, complete atrioventricular canal defect, right sided stomach, intestinal malrotation, RUQ spleen functionally asplenic, hepatomegaly with left sided liver, LUQ gallbladder
  
  • Complex congenital heart defects -> Lasix and staged cardiac reconstruction
    • Palliative pulmonary artery banding to decrease pulmonary circulation with ultimate goal of single ventricle circulation
    • TAPVR repair
    • Complete atrioventricular septal defect repair and pulmonary artery debanding (to provide normal 2 ventricle circulatory system)

• Malrotation with risk of volvulus -> Ladds procedure

• Functional asplenia -> Amoxicillin prophylaxis

• Severe GERD with feeding difficulty -> Temporary NG / PEG tube feeds

• Dextrocardia of unknown etiology -> Pulmonology referral for workup of possible primary ciliary dyskinesia
Heterotaxy

- Heterotaxy: Abnormal arrangement of internal thoracic – abdominal organs across the left-right axis of the body. Commonly associated with cardiac malformations and considerable morbidity and mortality
  - Right atrial isomerism: duplication of right sided structures with absence of left sided structures (two right atria, hepatomegaly, asplenia)
  - Left atrial isomerism: duplication of left sided structures with absence of right sided structures (two left atria, discontinuity of IVC, polysplenia)
- Usually suspected prenatally with routine antenatal screening ultrasound and confirmed with fetal echocardiography
- Presentation at birth varied from asymptomatic to gravely ill with cyanosis depending on severity of cardiac malformations
- Management
  - Surgical cardiac reconstruction
  - Medical management to optimize cardiac function and pulmonary blood flow
Heterotaxy
Imaging discussion 1: Chest radiograph

- Appropriateness: unclear
- True indication: dextrocardia, confirmed congenital cardiac malformation on prenatal echo – initial postnatal imaging
- Sensitivity / specificity of chest radiograph for dextrocardia: no data
- Cost: ~$55 (operating cost), cost to patient largely varied based on geographic location, imaging center, and insurance
- Radiation: <0.03 mSv
Imaging discussion 2: Abdominal radiograph

- **Appropriateness**: unclear
- **True indication**: dextrocardia with concern for visceral situs inversus, poor feeding without vomiting at 1 day of life
- **Sensitivity / specificity of abdominal radiograph for situs inversus**: no data
- **Cost**: ~$55 (operating cost), cost to patient largely varied based on geographic location, imaging center, and insurance
- **Radiation**: 0.03-0.3 mSv
### Imaging discussion 3: Abdominal ultrasound

- **Vomiting within the first 2 days after birth. Poor feeding or no passage of meconium. Initial imaging.**
  - Appropriateness: unclear
  - True indication: dextrocardia and situs inversus / heterotaxy with concern for malrotation. Poor feeding without emesis
  - Pooled sensitivity 94% (82%-100%), pooled specificity 100% (55%-100%) for malrotation with or without midgut volvulus on abdominal ultrasound
  - Cost: $260 but varies based on location, imaging institution, and insurance
  - Radiation: 0 mSv

<table>
<thead>
<tr>
<th>Scenario Id</th>
<th>Procedure</th>
<th>Adult RRL</th>
<th>Peds RRL</th>
<th>Appropriateness Category</th>
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</thead>
<tbody>
<tr>
<td>3191958</td>
<td>Radiography abdomen</td>
<td>0.1-1 mSv</td>
<td>0.03-0.3 mSv [ped].</td>
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<td>3185451</td>
<td>US abdomen (UGI tract)</td>
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<td>0 mSv [ped] O</td>
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<tr>
<td></td>
<td>Fluoroscopy contrast enema</td>
<td>1-10 mSv</td>
<td>3-10 mSv [ped].</td>
<td>Usually not appropriate</td>
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<tr>
<td></td>
<td>Fluoroscopy upper GI series</td>
<td>1-10 mSv</td>
<td>0.3-3 mSv [ped].</td>
<td>Usually not appropriate</td>
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<tr>
<td></td>
<td>US abdomen (UGI tract)</td>
<td>0 mSv O</td>
<td>0 mSv [ped] O</td>
<td>May be appropriate</td>
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### Imaging discussion 4: Upper GI series

<table>
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<th>Scenario</th>
<th>Procedure</th>
<th>Adult RRL</th>
<th>Peds RRL</th>
<th>Appropriateness Category</th>
</tr>
</thead>
<tbody>
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<td>Vomiting, bilious, malrotation suspected, initial imaging</td>
<td>Fluoroscopy upper GI series</td>
<td>1-10 mSv</td>
<td>0.3-3 mSv [ped]...</td>
<td>Usually appropriate</td>
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<tr>
<td></td>
<td>US abdomen (UGI tract)</td>
<td>0 mSv O</td>
<td>0 mSv [ped] O</td>
<td>May be appropriate</td>
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<td>Radiography abdomen</td>
<td>0.1-1mSv</td>
<td>0.03-0.3 mSv [ped]..</td>
<td>May be appropriate</td>
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<tr>
<td></td>
<td>Fluoroscopy contrast enema</td>
<td>1-10 mSv</td>
<td>3-10 mSv [ped]..</td>
<td>Usually not appropriate</td>
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<td></td>
<td>Nuclear medicine gastroesophageal reflux scan</td>
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</table>

- Appropriateness: likely appropriate
- True indication: dextrocardia and heterotaxy with concern for malrotation. Poor feeding without emesis
- Pooled sensitivity 91% (40%-100%), pooled specificity 94% (20%-100%) for malrotation on upper GI series
- Cost: $600 but varies based on location, imaging institution, and insurance
- Radiation: 0.3-3 mSv
Heterotaxy is the abnormal arrangement of internal thoracic–abdominal organs across the left-right axis of the body and is associated with significant morbidity and mortality due to associated cardiac malformations.

Heterotaxy is usually suspected prenatally with screening ultrasound and confirmed with fetal echocardiogram.

Management of heterotaxy depends on severity but commonly involves medical therapy and staged cardiac reconstruction.
References


