Newborn with VACTERL...with a Twist

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Our Patient: History

36yo G2P1 at 30w1d with bicornuate uterus, 2-vessel cord, polyhydramnios (AFI 25), intermittent absent end diastolic flow is admitted for nonreactive NST and 6/8 BPP. Follow up US…
Our Patient: US Report at 30w2d
Showing IUGR

BIDMC PACS

Note the asymmetric nature of the IUGR observed on the US.
**Our Patient: Further Findings on US Report at 30w2d**

<table>
<thead>
<tr>
<th><strong>Fetal Anatomy:</strong></th>
<th>Normal</th>
<th>Abnormal</th>
<th>Suboptimal</th>
</tr>
</thead>
<tbody>
<tr>
<td>Head</td>
<td>X</td>
<td>0</td>
<td>o</td>
</tr>
<tr>
<td>Brain</td>
<td>o</td>
<td>0</td>
<td>X</td>
</tr>
<tr>
<td>Face</td>
<td>o</td>
<td>0</td>
<td>X</td>
</tr>
<tr>
<td>Spine</td>
<td>o</td>
<td>0</td>
<td>o</td>
</tr>
<tr>
<td>Neck/Skin</td>
<td>o</td>
<td>0</td>
<td>X</td>
</tr>
<tr>
<td>Thorax</td>
<td>X</td>
<td>o</td>
<td>o</td>
</tr>
<tr>
<td>Heart</td>
<td>See Details</td>
<td>o</td>
<td>0</td>
</tr>
</tbody>
</table>

**Abdominal Wall**

**GI Tract**

**Kidneys**

**Bladder**

**Extremities**

**Skeletor**

**Genitalia**

**Details:**
- Heart: Visualized and normal appearance: four-chamber view, inferior vena cava, superior vena cava.
- Not visible: left outflow tract, right outflow tract, Ductal arch.
- Cardiac axis: Dextrocardia. Aortic arch: Unable to determine.

**Gastrointestinal Tract:** Stomach: abnormal. Dextroposition.

**Summary of Ultrasound Findings:**

**Fetal Wellbeing Assessment:**

**Maternal Structures:**
- Right Ovary: normal.
- Left Ovary: not visible.
Our Patient: Fetal MRI confirming situs inversus totalis

T2-weighted sagittal MRI shows cephalic presentation of fetus

T2-weighted axial MRI image shows the fetal heart facing away from abdominal wall, reflecting dextrocardia
Situs Inversus Totalis: Facts

- Condition in which the major thoracic and abdominal organs are rotated around the sagittal plane.
- Incidence of situs inversus is about 1:10,000. 20-25% of pts have primary ciliary dykinesia (PCD)
- No serious adverse health affects per se, and situs patients often present incidentally on CXR or other imaging.
- PCD has 200-fold higher risk of cardiac abnormalities—fetal echocardiography indicated.

Celebrities who have situs inversus totalis include NBA player Randy Foye (above) and actress Catherine O’Hara (right) who starred in Home Alone.
Our Patient: Further Course

- Pt’s mother, now at 34w1d comes for decreased fetal movement.
- BPP is 0/8, nonreactive NST. Taken for emergent c-section.
- Infant emerged limp, with slow heart rate and O2 sat of 60. CPAP improved ventilation to 90s. Apgars were 2, 7, and 8 at 1, 5 and 10 min of life. Pt taken to NICU. ET and NG tubes are placed.
- Babygram is ordered…
Our Patient: CXR with a new finding

This CXR shows a NG tube coiled in the esophagus, reflecting esophageal atresia. The air inside the stomach demonstrates that there is a tracheoesophageal fistula, allowing air to enter the distal GI tract.
• Tracheoesophageal Fistula (TEF) – 1:3500 live births, and 95% of time also with Esophageal Atresia (EA)
• Several types but C is by far the most common.
• Polyhydramnios is present in 66% of pregnancies.
• Infants present with excessive drooling, choking, respiratory distress, inability to feed
• Treatment is ligation of the fistula and anastamosis of proximal and distal esophagus

Source: UpToDate
Our patient underwent thoracotomy and surgical ligation of the TEF. Unfortunately the proximal and distal esophagi were not able to be anastamosed and a **G-tube** had to be placed for feeding.
Prenatally, TEF/EA can present with small stomach and esophageal pouch on US and MRI.

Images source: Carol E. Barnewolt, *Seminars in Roentgenology*, 2004
Now that we learned our patient has TEF/EA, we have to evaluate for other congenital anomalies often associated with it. These cluster of anomalies are known as VACTERL.
VACTERL: Basic Facts

- Association of congenital abnormalities (no known cause to tie them together into a syndrome).
- Originally named VATER in 1970s
- Frequency of 1-9/100k infants
- Diagnosis requires 3+ of 7 defect categories
- Also associated with single umbilical artery (like our pt)

Vertebral defects
Anal atresia
Cardiac defects
Tracheoesophageal fistula
Esophageal Atresia
Renal defects
Limb defects
Schematic drawing of hemivertebra (A), butterfly vertebra (B), fused (C), and consecutive hemivertebra (D).

Companion patients. In set A, a transabdominal US shows a fetus which has hemivertebra (white arrow). Set B shows a CXR of an infant with several hemivertebra and fused ribs.
VACTERL: Cardiac Anomalies, e.g. TAPVR

Total Anomalous Venous Return

- Pulmonary veins do not return to left atrium.
- Instead return to systemic venous circulation e.g. SVC, IVC, or hepatic veins or directly to RA
- Always associated with ASD
- Infradiaphragmatic kind often are obstructed, leading to pulmonary congestion
- Imaging: echo to identify anomalous connection and to see if there is obstruction. If obstruction is suspected but cannot be confirmed with echo, can do cardiac cath.

Source: UpToDate
In the schematic figure above, the normal anatomy of the venous drainage of the liver is shown, with the right, middle and left hepatic veins flowing into the IVC at about the same level.

This transverse US with doppler of Companion Patient 2 shows normal venous drainage of the liver.
Schematic above shows TAPVR. On the left, the pulmonary veins connect via an anomalous ‘vertical vein’ subdiaphragmatically to the hepatic veins. On the right, the pulmonary veins connect via a vertical vein into the IVC above the diaphragm.

Image source: Katre, Seminars in Ultrasound, CT and MRI, 2012

Transverse abdominal US with doppler shows an anomalous vertical vein connecting the pulmonary veins to middle hepatic vein.

Image source: CHB PACS
Our Patient: Doppler Echo showing TAPVR

* Large ASD

Transthoracic echo showing an enlarged right atrium and a large ASD. The doppler image on the right shows flow from the R atrium, through the ASD and into the L atrium then L ventricle. There is no other source of flow into the L atrium.
VACTERL: Limb Defects, e.g. Companion Pt 3 with radial ray defects

Image A shows Companion Patient 3 fetal US at 20wk with radial aplasia and hypoplastic ulna. Image B is from the same US and shows the contralateral forearm of Companion patient 3, which has normal anatomy. Image C is an X-ray of a different patient who also has radial ray defects. Here, the infant as an absent radius and thumb and hypoplastic ulna.

# VACTERL: Review of Work Up

<table>
<thead>
<tr>
<th>Condition</th>
<th>Work Up</th>
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<tbody>
<tr>
<td>Vertebral defects</td>
<td>X-ray. US or MRI for spine</td>
</tr>
<tr>
<td>Anal atresia</td>
<td>PE. Abd US for GU defects</td>
</tr>
<tr>
<td>Cardiac defects</td>
<td>Echo. Cardiac CT or MRI for further details</td>
</tr>
<tr>
<td>TEF/Eosophageal atresia</td>
<td>PE/Coiled NG. contrast studies rarely required</td>
</tr>
<tr>
<td>Renal abnormalities</td>
<td>Renal US. VCUUG if abnl US.</td>
</tr>
<tr>
<td>Limb defects</td>
<td>PE, X-ray</td>
</tr>
</tbody>
</table>
References

• *StudyBlue:* http://2.bp.blogspot.com/_8a64_UUPAh0/SFLchyHWncI/AAAAAAAAAzY/j00by30enSg/s1600-h/Spine+-+failure+of+formation.jpg. Date accessed 2/24/13
Acknowledgements

• Dr. Liebermann for all her teaching and support
• Dr. Deborah Levine for helping retrieve and interpret OB images
• Claire Odom for her support
• Michelle-Marie Peña for technical support
• Nina Mann for helping retrieve CHB images