AMSER Case of the Month
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Newborn Female with abdominal distension

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Patient Presentation

• HPI: Newborn female, delivered at 33 and 4/7 weeks via spontaneous vaginal delivery at outside facility. Found to be limp and cyanotic, intubated shortly after birth. Significant abdominal distension noted. Mother with routine prenatal care, notably with 20-week anatomy ultrasound negative.
Patient Presentation

Physical Exam:
- Afebrile
- General: Intubated, in moderate distress
- Cardiovascular: Heart with regular rate and rhythm, no murmurs, good pulses in all extremities
- Respiratory: Coarse breath sounds, substernal retractions noted
- Abdomen: Soft, normal bowel sounds present, three-vessel cord, marked abdominal distension with firm bilateral masses palpated
Early chest radiograph obtained due to respiratory distress, intubation.
Findings (labeled)

Protuberant abdomen, as noted on physical exam

Lungs are under inflated but not hypoplastic.
What Imaging Should We Order?
Select the applicable ACR Appropriateness Criteria

American College of Radiology
ACR Appropriateness Criteria®
Palpable Abdominal Mass-Suspected Neoplasm

<table>
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<th>Variant 1:</th>
<th>Palpable abdominal mass, Suspected intra-abdominal neoplasm, Initial imaging.</th>
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<tr>
<td>Procedure</td>
<td>Appropriateness Category</td>
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<tr>
<td>CT abdomen with IV contrast</td>
<td>Usually Appropriate</td>
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<td>US abdomen</td>
<td>Usually Appropriate</td>
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<tr>
<td>MRI abdomen without and with IV contrast</td>
<td>May Be Appropriate</td>
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<tr>
<td>CT abdomen without IV contrast</td>
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<td>CT abdomen without and with IV contrast</td>
<td>Usually Not Appropriate</td>
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<td>FDG-PET/CT skull base to mid-thigh</td>
<td>Usually Not Appropriate</td>
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<td>Radiology abdomen</td>
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<td>Fluoroscopy enema</td>
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<td>Fluoroscopy upper GI series</td>
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<tr>
<td>Fluoroscopy upper GI series with small bowel follow-through</td>
<td>Usually Not Appropriate</td>
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Originally ordered at outside facility
Findings (labeled)

Enlarged kidneys with innumerable small cysts and ectatic tubules.

Liver adjacent to the right kidney appears normal.
Findings (unlabeled)
Findings (labeled)

Enlarged kidneys, which nearly touch at midline.

Renal hilum seen bilaterally.
Final Dx:
Autosomal Recessive Polycystic Kidney Disease (ARPKD)
ARPKD Disease

- Approximately 1:20,000 births affected (1)
- *PKHD1* mutation leading to dysfunctional primary cilia causing non-obstructive fusiform collecting duct dilation (1)
- Associated with varying degrees of concomitant congenital hepatic fibrosis, often with pulmonary hypoplasia (seen radiographically in this patient) (1)
ARPKD — Prognosis and Progression

- Variable disease severity and progression (1, 4)
- Increased neonatal survival in patients without oligohydramnios or renal enlargement (2)
- Faster disease progression with lower baseline glomerular filtration rate (GFR) (3)
- Older patients (≥ 10 years) with higher rates of decline than younger (3)
- Extrarenal manifestations more common in those who survive the neonatal period (4)
High resolution ultrasound superior to standard resolution ultrasound, especially in patients with milder disease. (1)
Sonographic abnormalities usually detected at approximately 30 weeks of gestation; can be found as early as 16-18 weeks (2).
Fetal liver anomalies are not reliably diagnosed (2)
Ultrasound shows diffusely echogenic kidneys with ectatic collecting ducts
References:


