AMSER Case of the Month April 2021

Newborn Female with abdominal distension

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Patient Presentation

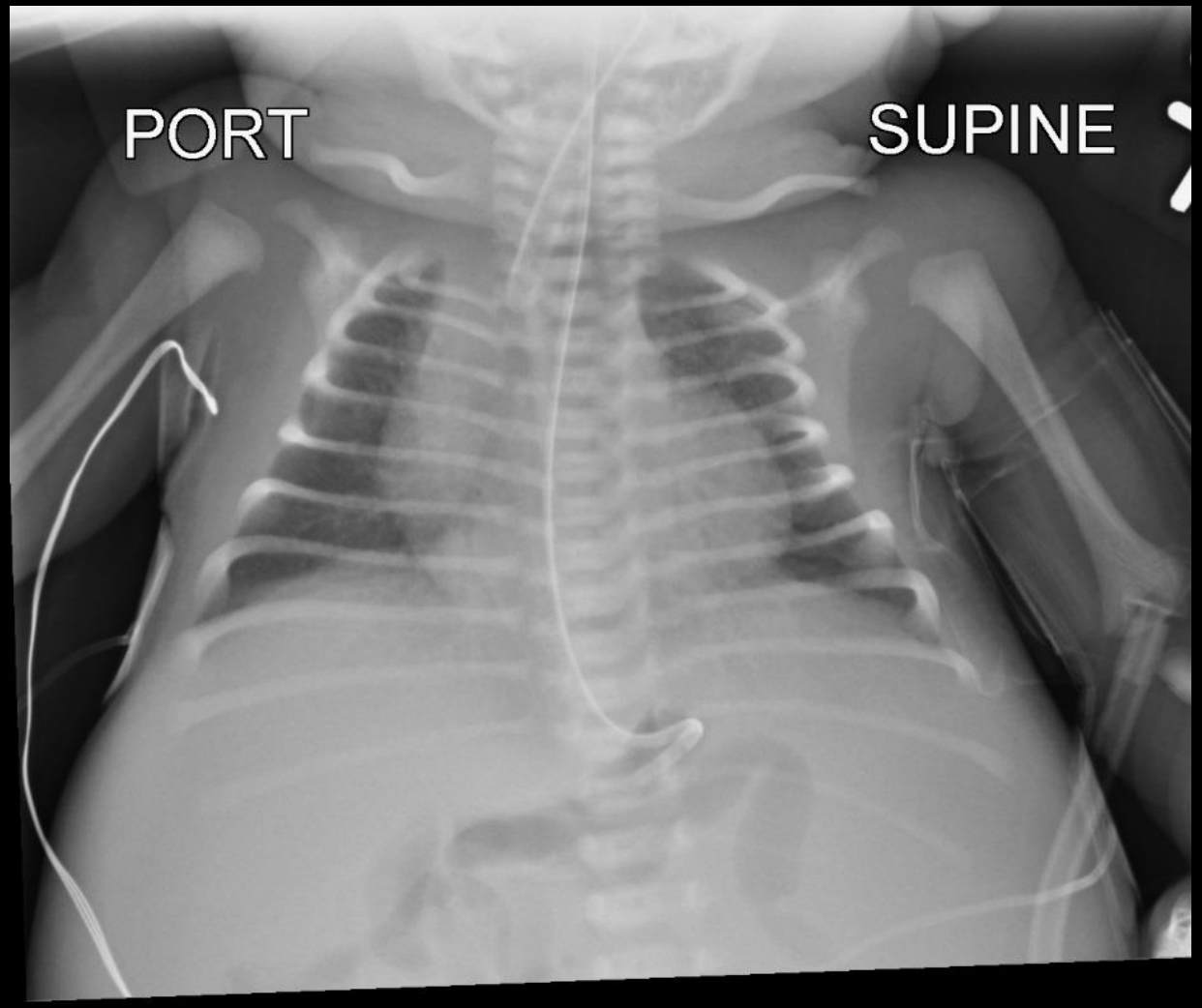
- HPI: Newborn female, delivered at 33 and 4/7 weeks via spontaneous vaginal delivery at outside facility. Found to be limp and cyanotic, intubated shortly after birth. Significant abdominal distension noted. Mother with routine prenatal care, notably with 20-week anatomy ultrasound negative.

Patient Presentation

Physical Exam:

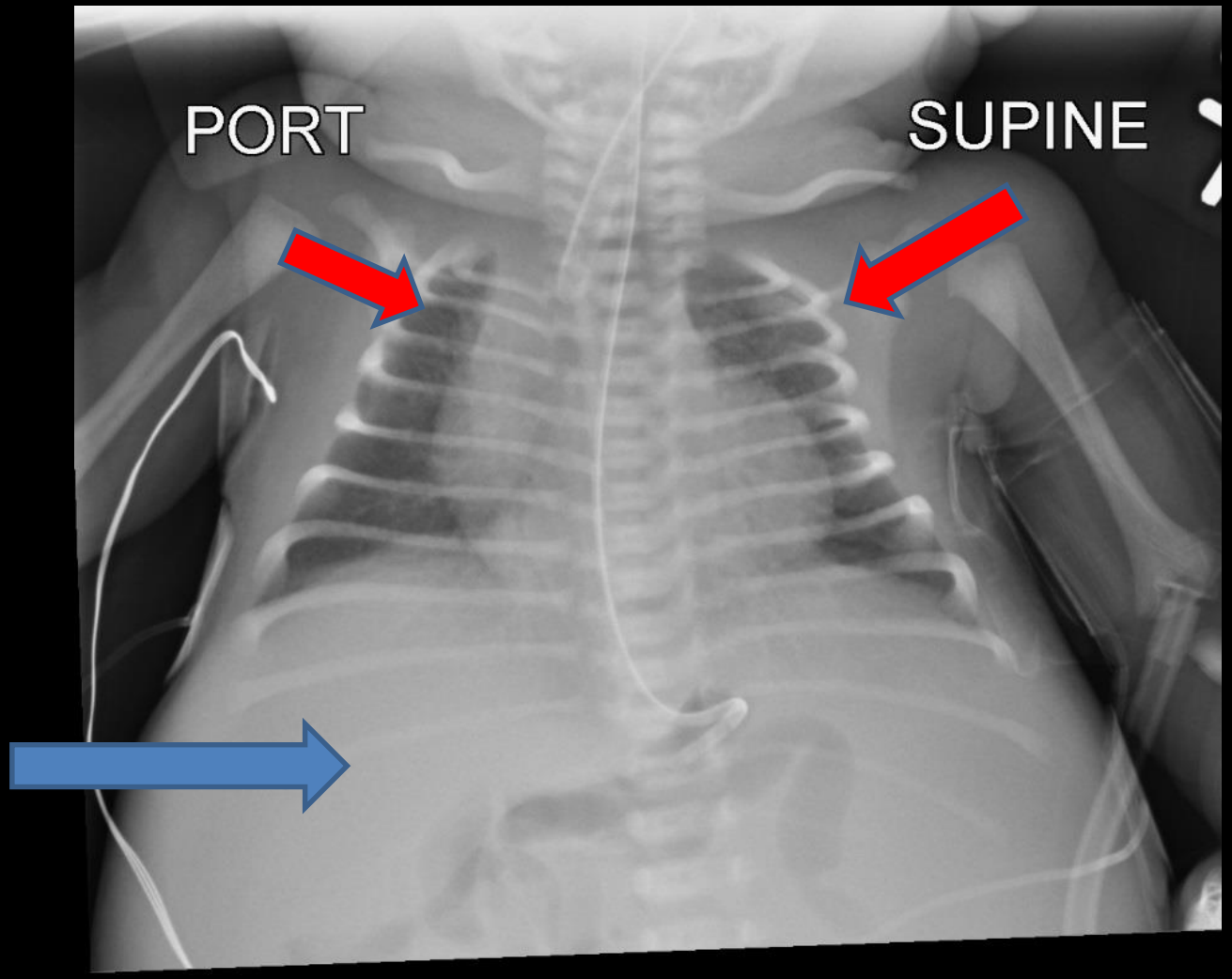
- Afebrile
- General: Intubated, in moderate distress
- Cardiovascular: Heart with regular rate and rhythm, no murmurs, good pulses in all extremities
- Respiratory: Coarse breath sounds, substernal retractions noted
- Abdomen: Soft, normal bowel sounds present, three-vessel cord, marked abdominal distension with firm bilateral masses palpated

Findings (unlabeled)



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

Revised 2019

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

Variant 1: **Palpable abdominal mass. Suspected intra-abdominal neoplasm. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
CT abdomen with IV contrast	Usually Appropriate	☢☢☢
US abdomen	Usually Appropriate	O
MRI abdomen without and with IV contrast	May Be Appropriate	O
CT abdomen without IV contrast	May Be Appropriate	☢☢☢
MRI abdomen without IV contrast	May Be Appropriate	O
CT abdomen without and with IV contrast	Usually Not Appropriate	☢☢☢☢
FDG-PET/CT skull base to mid-thigh	Usually Not Appropriate	☢☢☢☢
Radiography abdomen	Usually Not Appropriate	☢☢
Fluoroscopy contrast enema	Usually Not Appropriate	☢☢☢
Fluoroscopy upper GI series	Usually Not Appropriate	☢☢☢
Fluoroscopy upper GI series with small bowel follow-through	Usually Not Appropriate	☢☢☢

Originally ordered at outside facility

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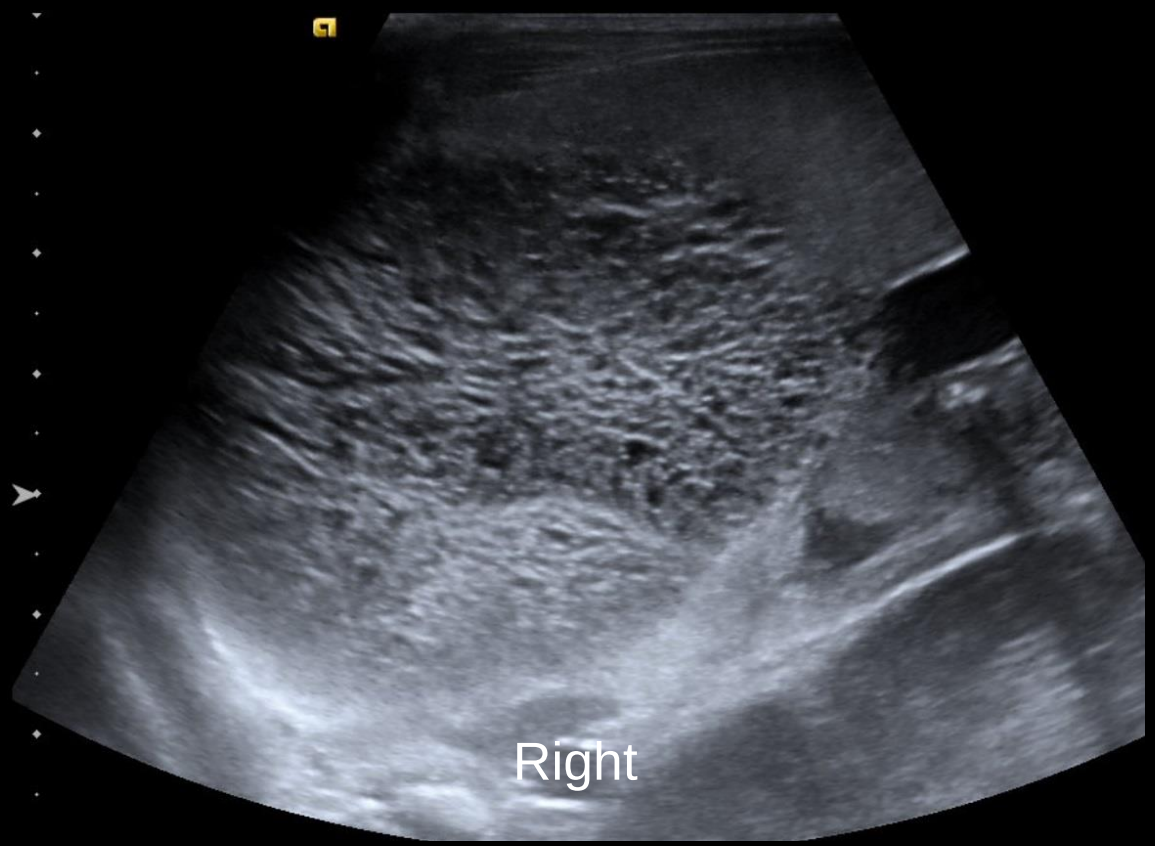
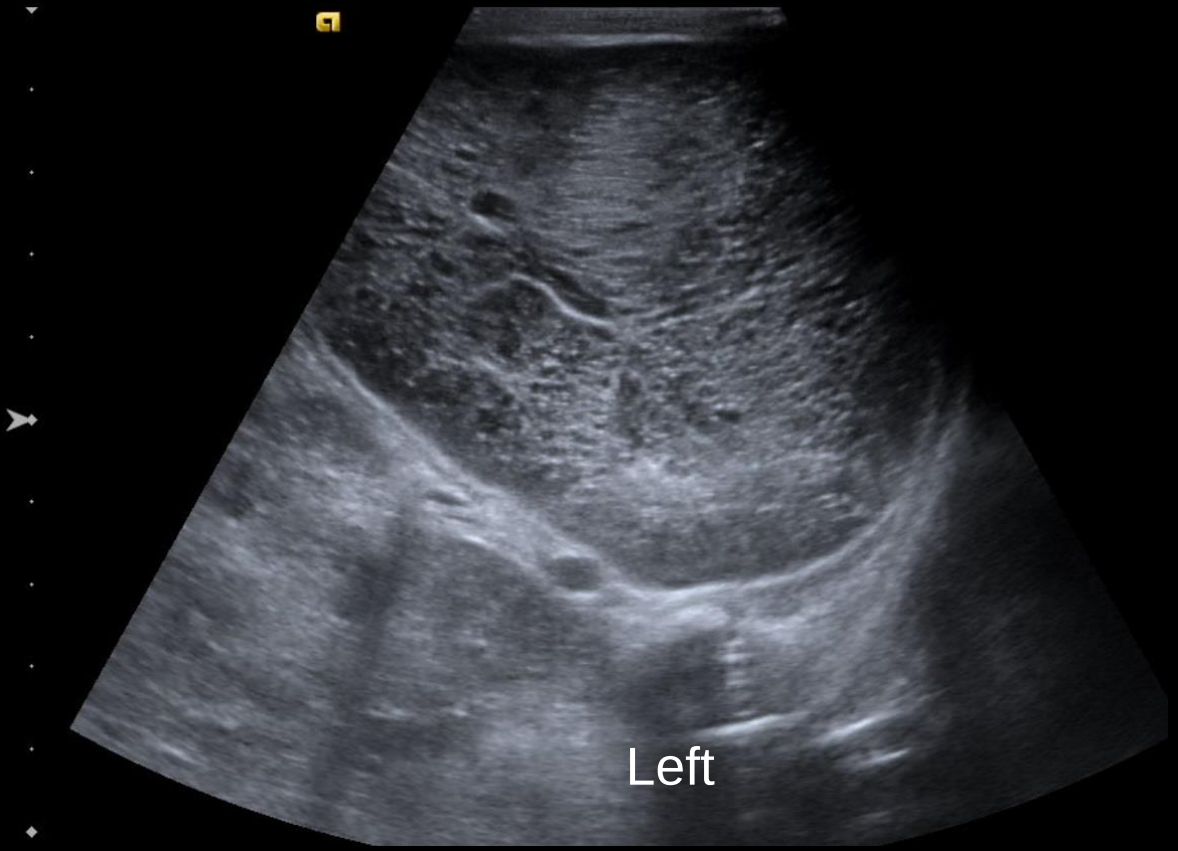
Findings (unlabeled)

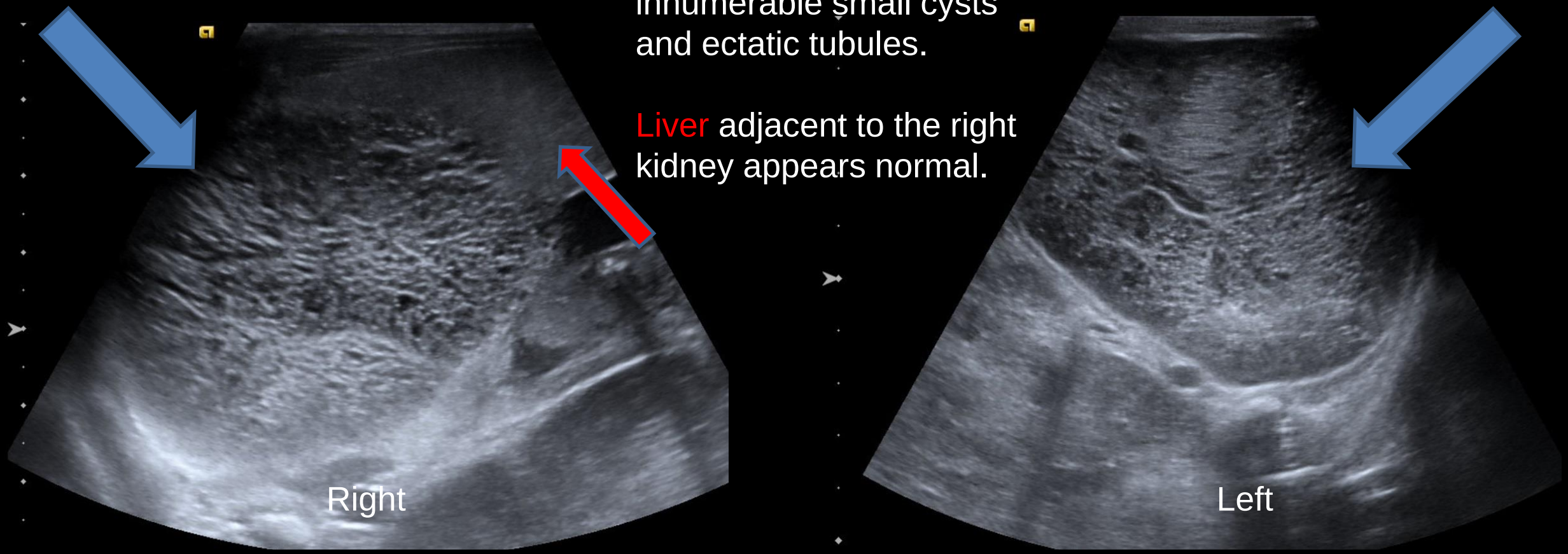




Findings (labeled)

Enlarged kidneys with innumerable small cysts and ectatic tubules.

Liver adjacent to the right kidney appears normal.



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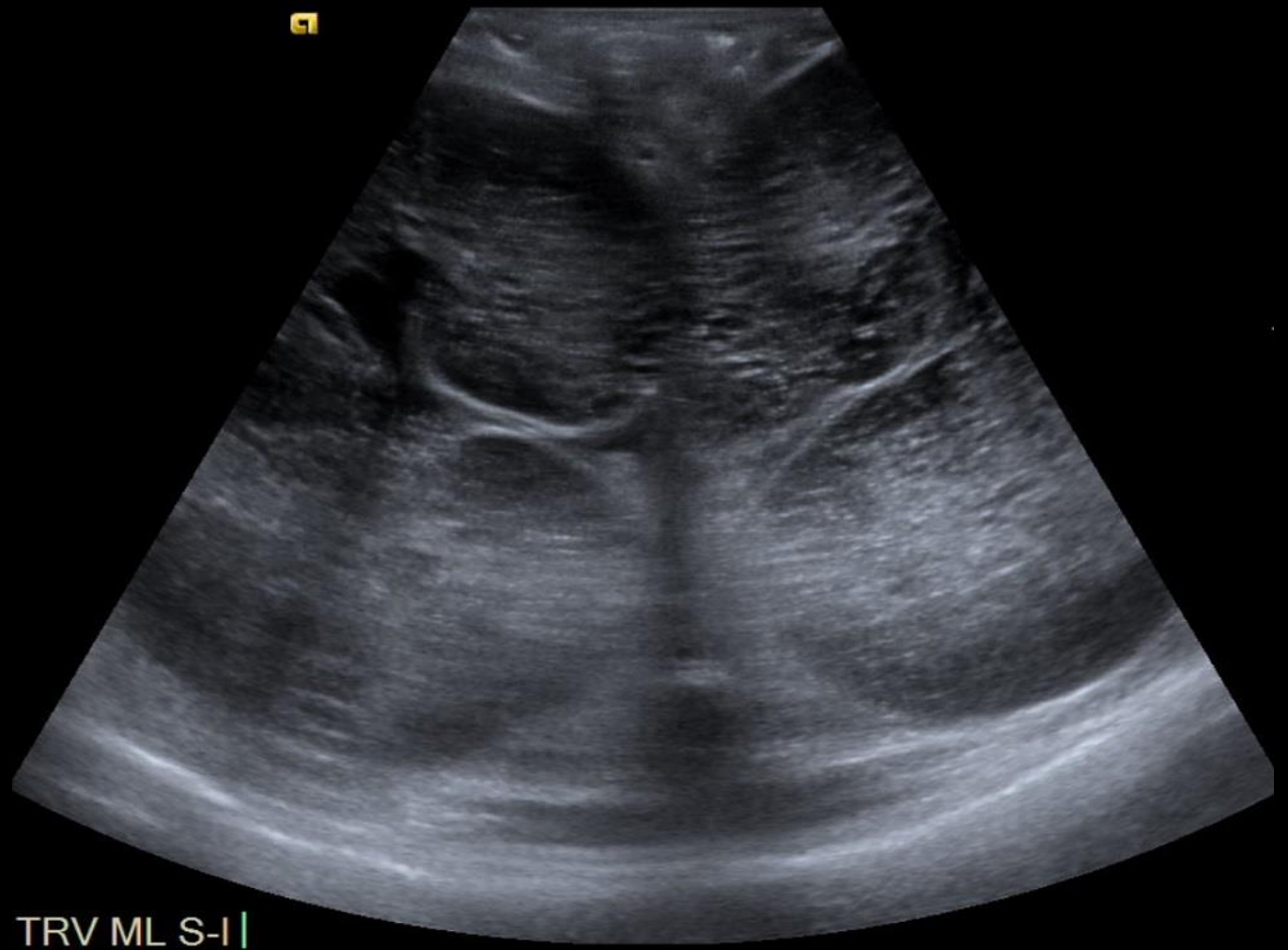

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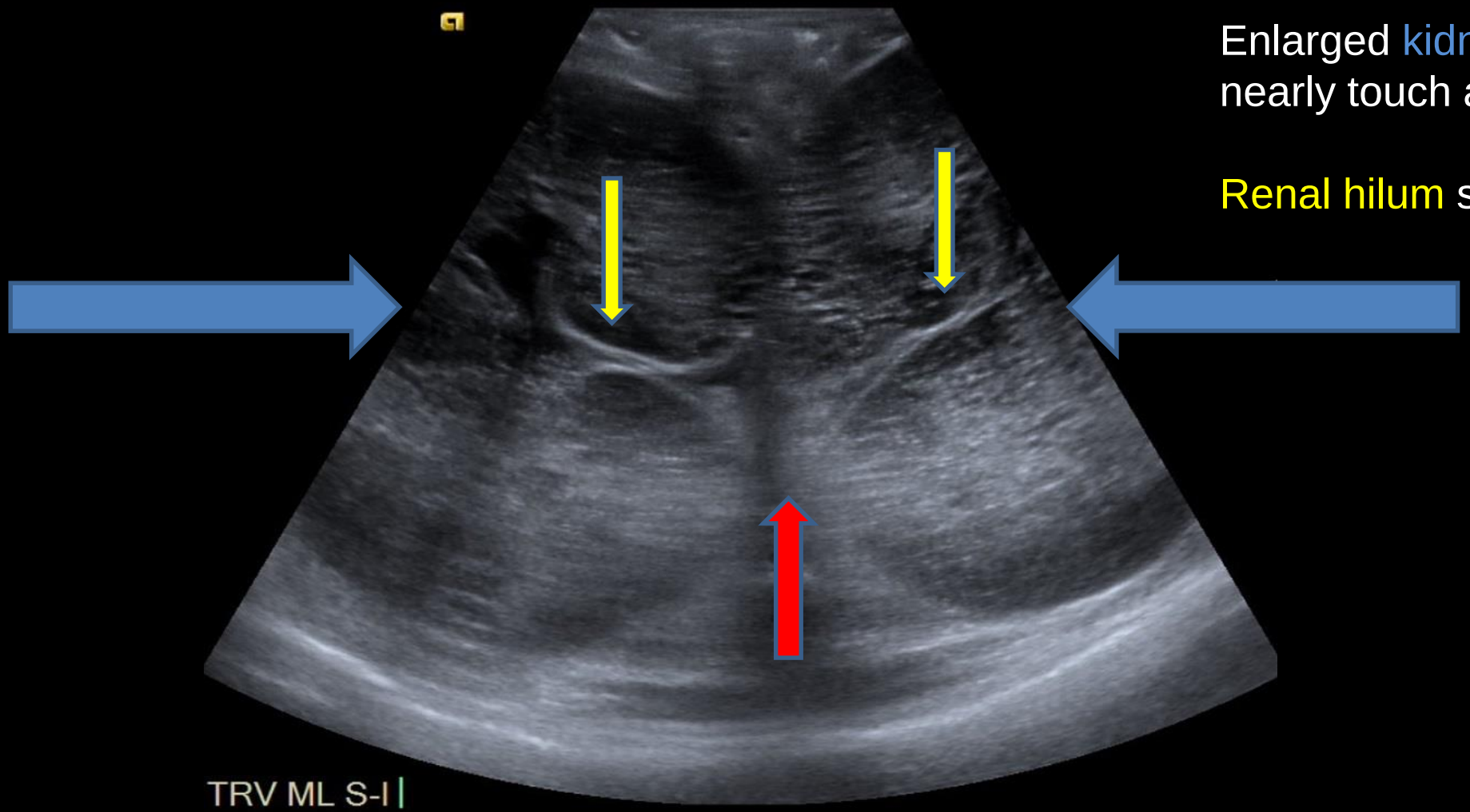

Enlarged kidneys, which nearly touch at midline.

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)

ARPKD — Imaging

- 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:

1)Gunay-Aygun M, Font-Montgomery E, Lukose L, et al. Correlation of kidney function, volume and imaging findings, andPKHD1Mutations in 73 patients with autosomal recessive polycystic kidney disease. *Clinical Journal of the American Society of Nephrology*. 2010;5(6):972-984. doi:10.2215/cjn.07141009

2)Erger F, Brüchle NO, Gembruch U, Zerres K. Prenatal ultrasound, genotype, and outcome in a large cohort of prenatally affected patients with autosomal-recessive polycystic kidney disease and other hereditary cystic kidney diseases. *Archives of Gynecology and Obstetrics*. 2017;295(4):897-906. doi:ref-Erger_2017ref-Erger_201710.1007/s00404-017-4336-6

3) Dell KM, Matheson M, Hartung EA, et al. Kidney disease progression in autosomal recessive polycystic kidney disease. *The Journal of Pediatrics*. 2016;171:196-201.e1. doi:ref-Dell_2016ref-Dell_201610.1016/j.jpeds.2015.12.079

4) Büscher R, Büscher AK, Weber S, et al. Clinical manifestations of autosomal recessive polycystic kidney disease (ARPKD): Kidney-related and non-kidney-related phenotypes. *Pediatric Nephrology*. 2013;29(10):1915-1925. doi:ref-B_scher_2013ref-B_scher_201310.1007/s00467-013-2634-1

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