

AMSER Case of the Month

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A newborn with prenatally diagnosed neck mass

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Patient Presentation

- An ex-37 week male infant, prenatally diagnosed with a neck mass, born via scheduled C-section with planned elective intubation at birth.
- Social/family history: Non-contributory.
- Genetics: Carrier screening: CF neg, SMA neg, Frag X neg. First trimester screen low risk, NIPT low risk XR. Declined chorionic villi sampling and amniocentesis.
- Maternal history: 33-year-old G1P1001
 - Hypothyroidism (on levothyroxine)
 - Obesity (BMI 36)
 - Gestational hypertension (on baby aspirin)
 - Gestational diabetes (on metformin)
 - Bipolar depression (well controlled on bupropion, sertraline and lamotrigine)
 - HSV 1 Ab positive (not on Valtrex)

Pertinent Physical Exam

- Vitals: Afebrile, HR 96-158, RR 34-85, SpO₂ 78-100%, 3.03 kg (25th-ile)
- General: Born vigorous, crying, APGAR 8, 8; CPAP 5, 40% started at 11 min. of life for desaturations and then intubated and sedated on fentanyl and midazolam.
- HEENT:
 - Normocephalic, normal conjunctiva and EOMI
 - Normal set ears
 - Palate intact, tongue noted to be enlarged with blue hue
 - Soft mass on anterior neck across midline that extends to ears and floor of mouth bilaterally

Rest of exam within normal limits, with no other skeletal or skin abnormalities

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

Variant 4: Child. Neck mass(es). Not parotid region or thyroid. Initial imaging.

Procedure	Appropriateness Category	Relative Radiation Level
CT neck with IV contrast	Usually Appropriate	☼ ☼ ☼
MRI neck without and with IV contrast	Usually Appropriate	○
US neck	Usually Appropriate	○
MRI neck without IV contrast	Usually Appropriate	○
CT neck without IV contrast	May Be Appropriate (Disagreement)	☼ ☼ ☼
MRA neck without and with IV contrast	Usually Not Appropriate	○
MRA neck without IV contrast	Usually Not Appropriate	○
CT neck without and with IV contrast	Usually Not Appropriate	☼ ☼ ☼ ☼
CTA neck with IV contrast	Usually Not Appropriate	☼ ☼ ☼
Arteriography cervicocerebral	Usually Not Appropriate	☼ ☼ ☼ ☼
FDG-PET/CT skull base to mid-thigh	Usually Not Appropriate	☼ ☼ ☼ ☼ ☼
FDG-PET/MRI skull base to mid-thigh	Usually Not Appropriate	☼ ☼ ☼

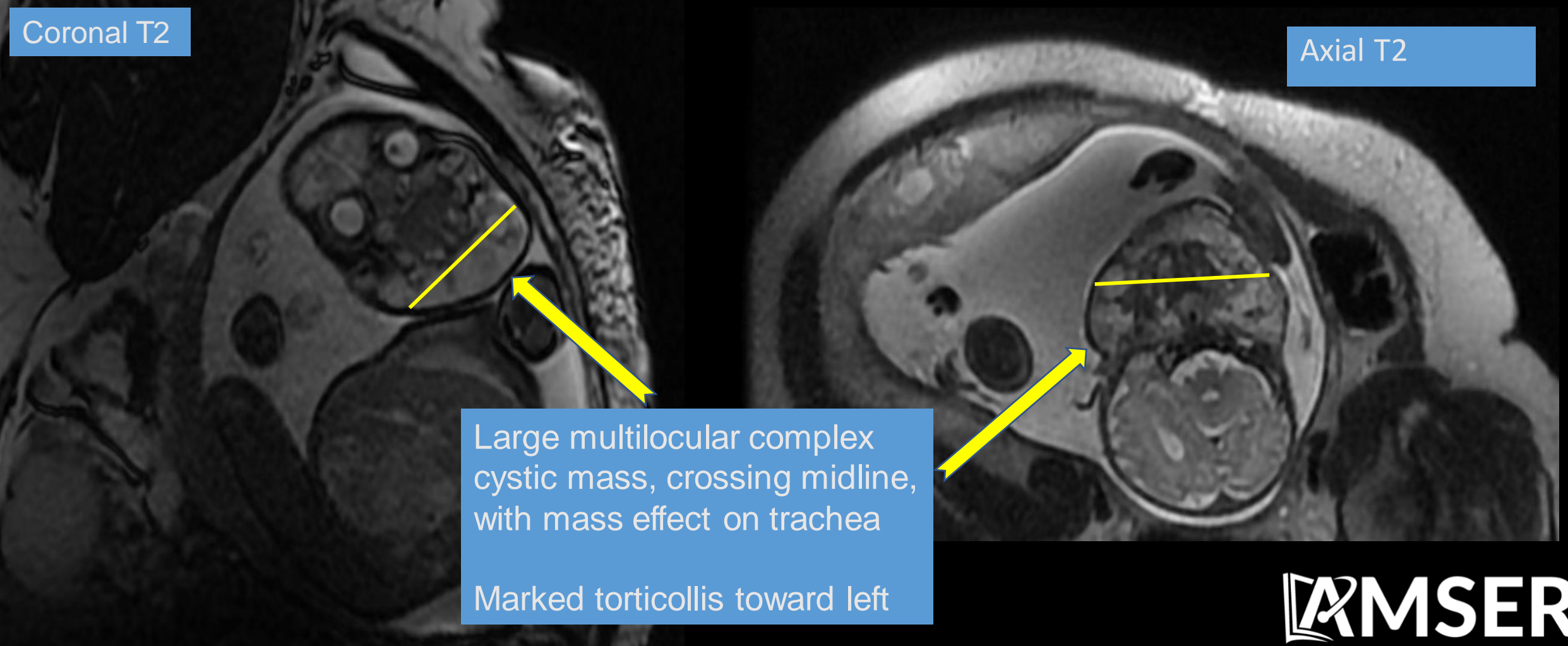
These imaging modalities were ordered by the neonatologist



Prenatal Imaging at 35 weeks - MRI Fetus without contrast

Coronal T2

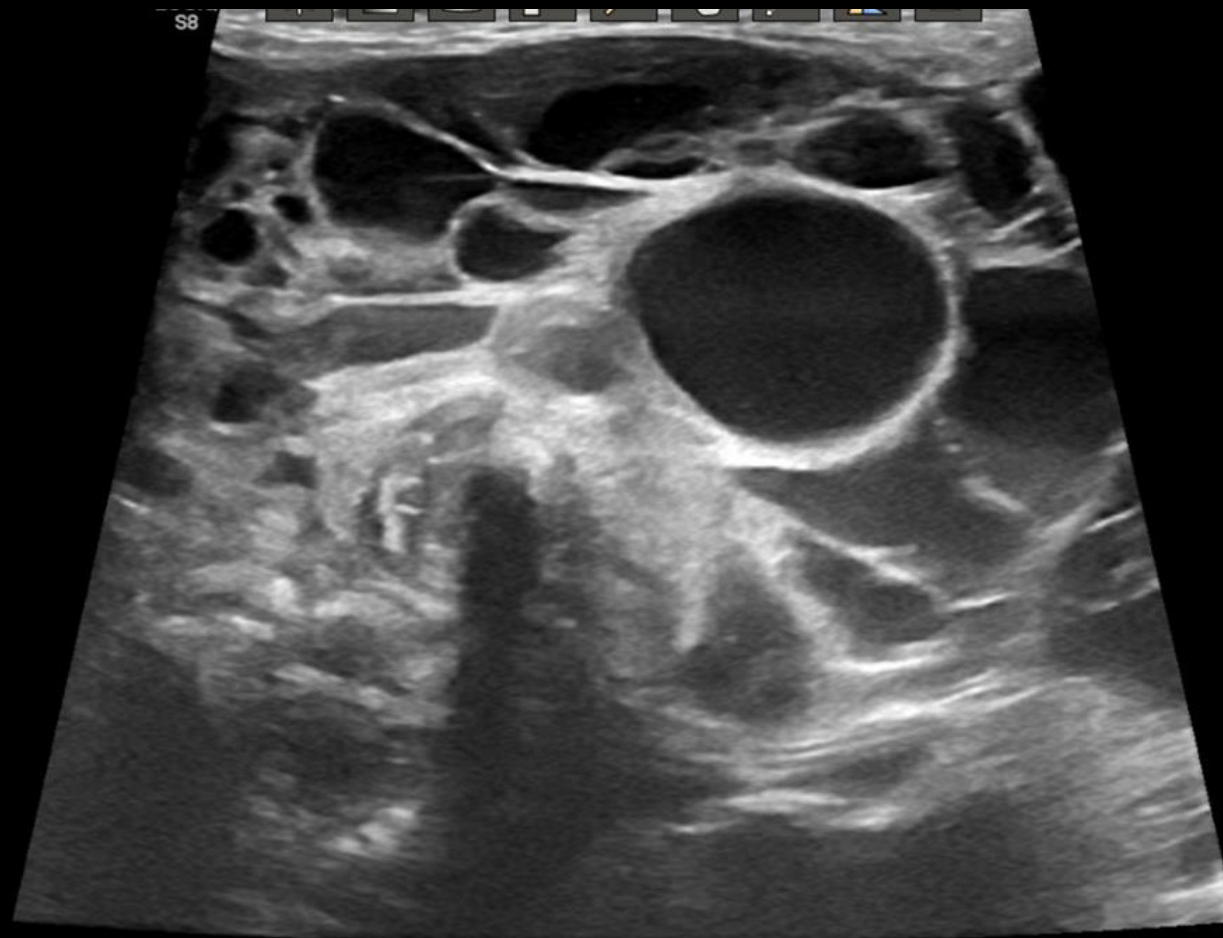
Axial T2



Large multilocular complex cystic mass, crossing midline, with mass effect on trachea

Marked torticollis toward left

Findings (unlabeled)



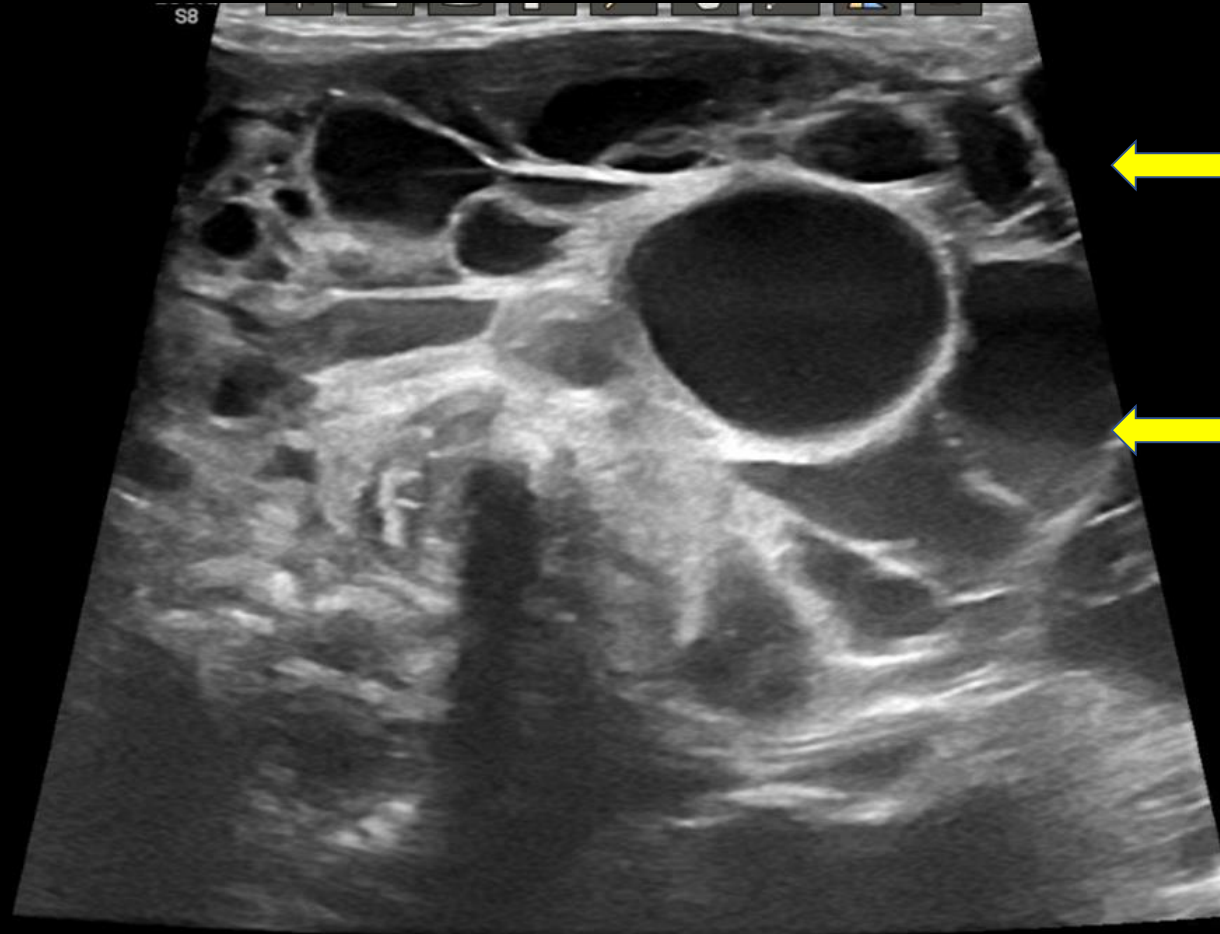
D PORTABLE

RIGHT NECK UPPER CHEEK AREA TRANS FROM SUP TO INF

Leibniz

AMSER

US Findings: (labeled)



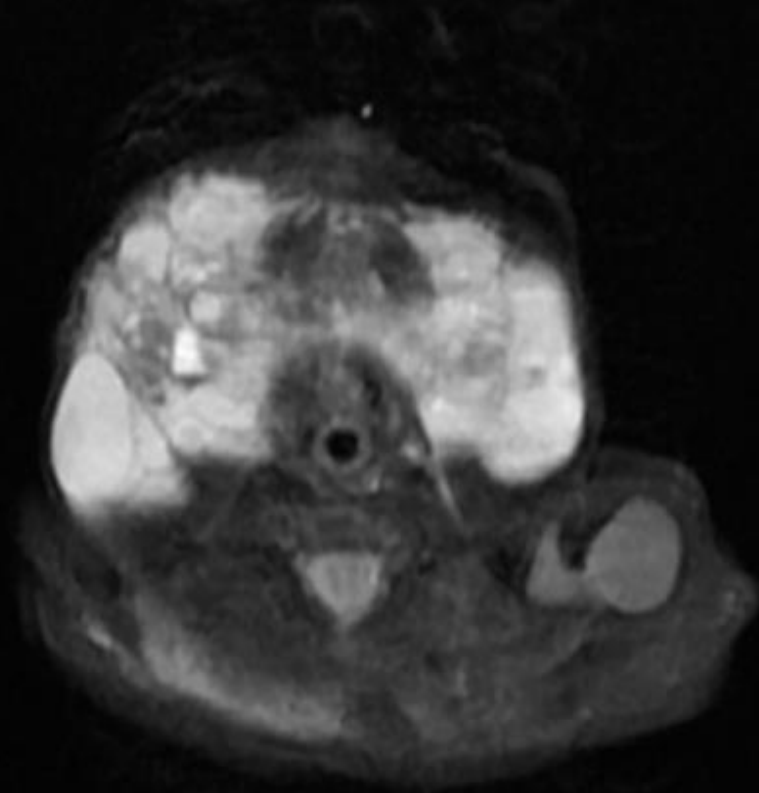
Septated,
predominantly
cystic mass in right
cheek and neck,
crossing midline

Fluid-debris
level

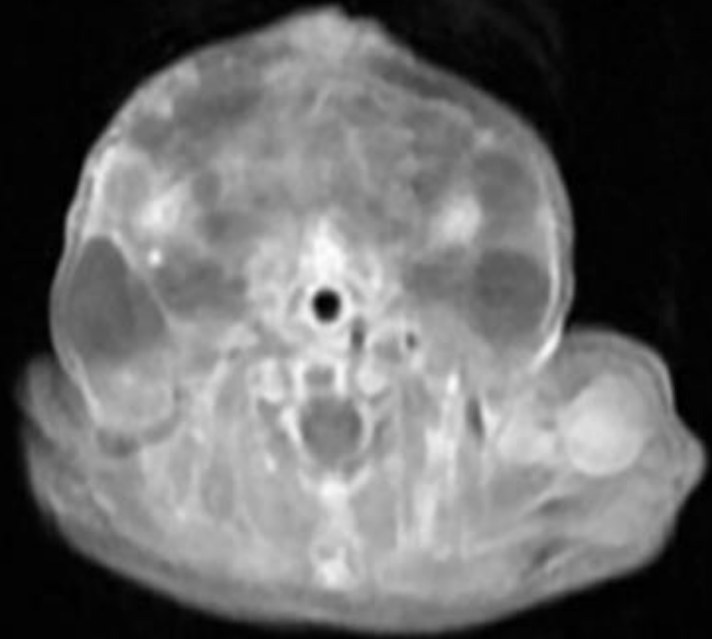
D PORTABLE

RIGHT NECK UPPER CHEEK AREA TRANS FROM SUP TO INF

MRI Findings (unlabeled)

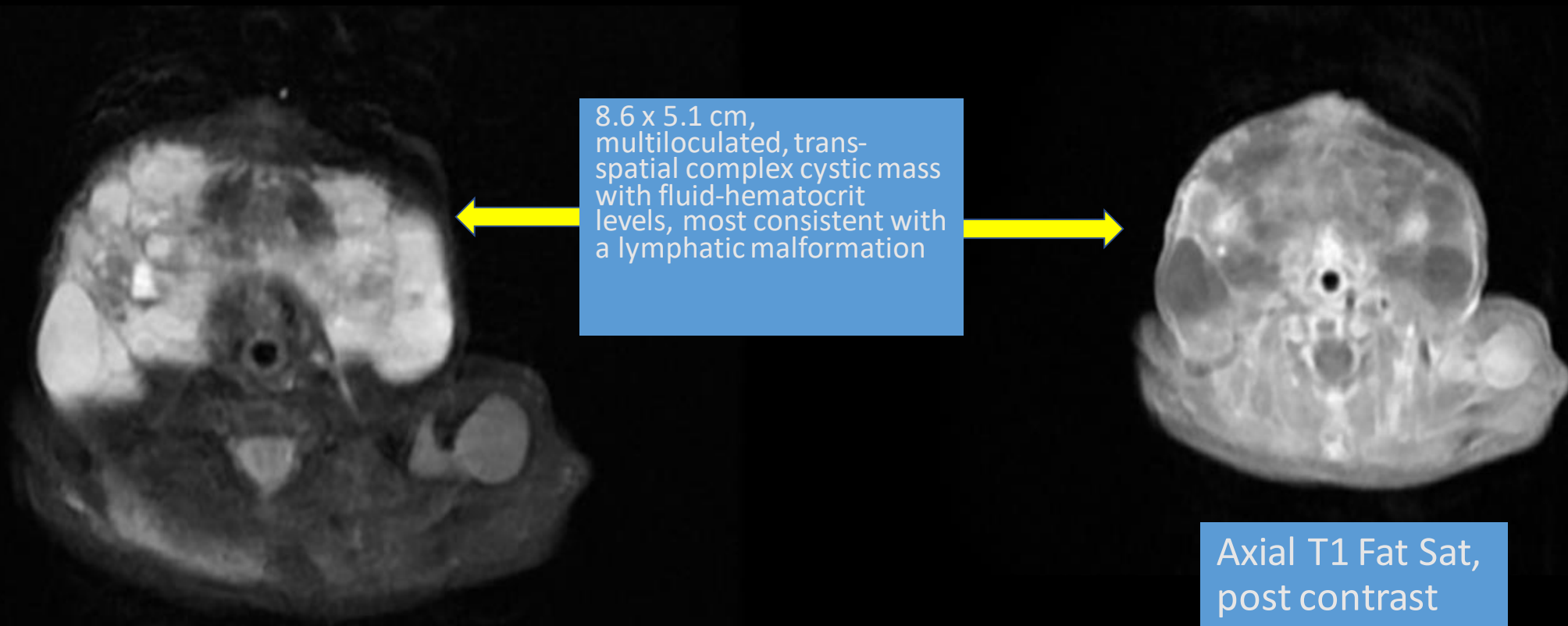


Axial T2



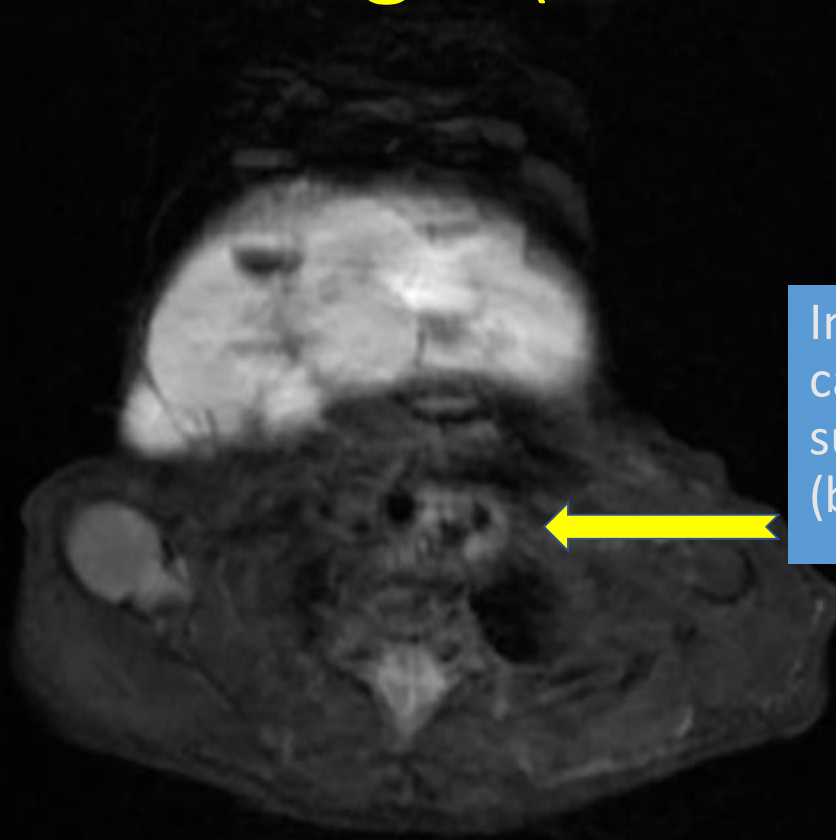
Axial fat sat T1 post contrast

MRI Findings: (labeled)



MRI Findings: (labeled)

Axial T2 Fat Sat



Involvement of left carotid and subclavian arteries (bright T2 signal)

Final Dx: Macrocystic lymphatic malformation

- Incidence: 1/100 fetuses in first trimester, septated: 1/285 fetuses
- Usually presents at birth as large, transspatial, cystic, soft mass covered by normal skin
- Most common in cervicofacial region, axilla or lateral chest wall
- Associated with chromosomal abnormalities (like Down syndrome, Turner syndrome, and Noonan syndrome) in about 50% of cases
- Aneuploidy more common with septated vs. simple lymphatic malformations (57 vs. 21%)
- $\frac{1}{3}$ of euploid fetuses with septated lymphatic malformations have major structural abnormalities, usually cardiac and skeletal

Differential Diagnosis

- **Cystic teratoma:** cystic teratoma consists of both cystic and solid components vs. just cystic. May have mature fat or calcification as part of the lesion. Typically more than just cyst wall or septal enhancement.
- **Hemangioma:** typically highly vascular vs. lymphatic malformation (previously referred to as cystic hygroma), which is more likely to have internal septae and only limited vascular supply
- **Venous malformation:** abnormally enlarged veins that are typically asymptomatic and can have a bluish hue clinically. CT usually demonstrates a hypoattenuating lesion that enhances slowly after bolus injection of contrast material

Case Discussion

- Sclerotherapy is first line treatment for macrocystic lymphatic malformations and reduces risk for future bleeding and infection
- Sclerotherapy relieves symptoms through injection of inflammatory substance to sclerose the lesion
- Surgical resection is indicated if lesion is well localized or sclerotherapy can no longer be performed
- Patient Course:
 - Patient was intubated for critical airway 15 minutes post-delivery, on minimal ventilator settings; Tracheostomy placed at 4 weeks of life
 - Sclerotherapy with doxycycline and sodium tetradecyl sulfate was completed at 6 days of life
 - Round 2 of sclerotherapy after internal hemorrhage into cystic spaces of the malformation was completed approximately 2 weeks later
 - Debulking surgery at 4 weeks of life with plan for sirolimus postoperatively to continue shrinking the malformation

References:

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