

AMSER Case of the Month

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A 19-year-old male with Trisomy 21 presents
with chest wall pain and weight loss

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Patient Presentation:

- **HPI:** 19-year-old male with history of Trisomy 21 complicated by chronic otitis media, obstructive sleep apnea presented to an outside care facility for 10lb weight loss over 3 months. Parents also note that patient has been rubbing his chest in discomfort. Chest radiograph and cardiac workup were normal. Patient was referred to GI for additional workup.
- **PMHx:** Hypothyroidism, celiac disease, mild IgM deficiency, obstructive sleep apnea, chronic suppurative otitis media
- **PSHx:** Repaired membranous VSD at age of 2, multiple tympanostomy tube placements
- **Meds:** Levothyroxine 816mcg total weekly dose, Omeprazole 40mg po daily
- **Allergies:** NKDA
- **FHx/SHx:** Noncontributory

What test should we order?

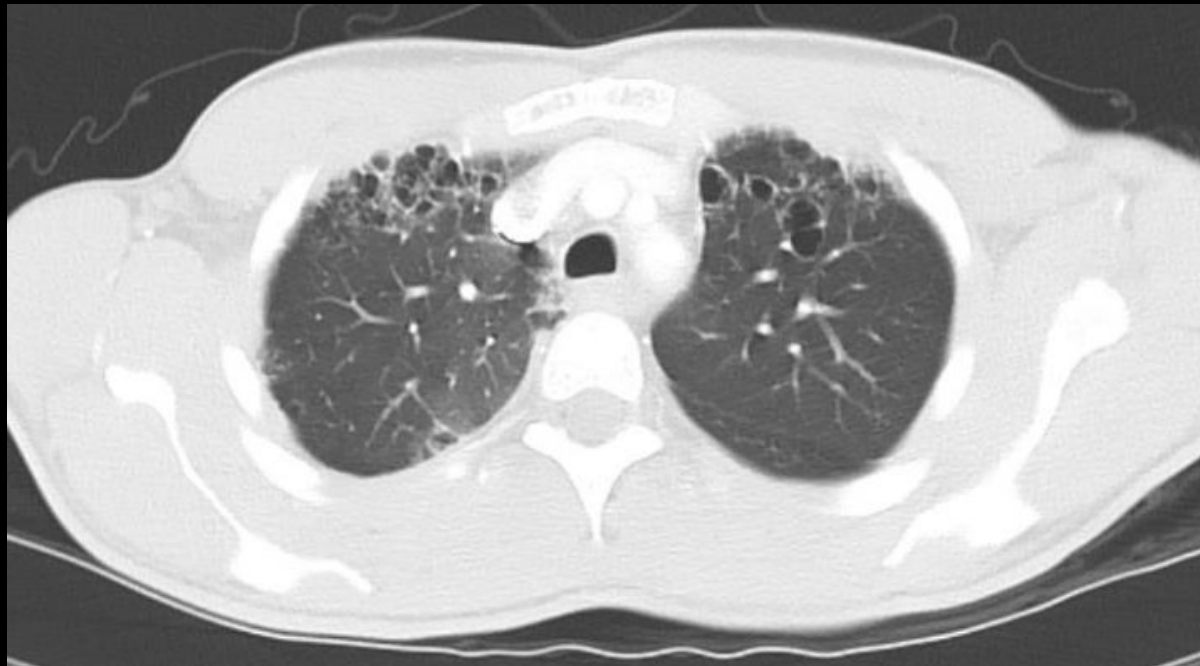
Select the applicable ACR Appropriateness Criteria

Scenario	Scenario Id	Procedure	Adult RRL	Peds RRL	Appropriateness Category	
chest wall pain, nontraumatic, inflammatory condition suspected, chest radiography normal, next imaging study	3194252	CT chest with IV contrast	1-10 mSv ☼☼☼	3-10 mSv [ped]..	Usually appropriate	●
		CT chest without IV contrast	1-10 mSv ☼☼☼	3-10 mSv [ped]..	Usually appropriate	●
		US chest	0 mSv ○	0 mSv [ped] ○	May be appropriate	●
		MRI chest without IV contrast	0 mSv ○	0 mSv [ped] ○	May be appropriate	●
		MRI chest without and with IV contrast	0 mSv ○	0 mSv [ped] ○	May be appropriate	●
		Bone scan whole body	1-10 mSv ☼☼☼	3-10 mSv [ped]..	May be appropriate	●
		FDG-PET/CT skull base to mid-thigh	10-30 mSv ☼☼☼☼	3-10 mSv [ped]..	May be appropriate	●
		WBC scan chest	10-30 mSv ☼☼☼☼	Not Assigned	May be appropriate	●
		Radiography rib views	1-10 mSv ☼☼☼	Not Assigned	Usually not appropriate	●
		CT chest without and with IV contrast	1-10 mSv ☼☼☼	3-10 mSv [ped]..	Usually not appropriate	●



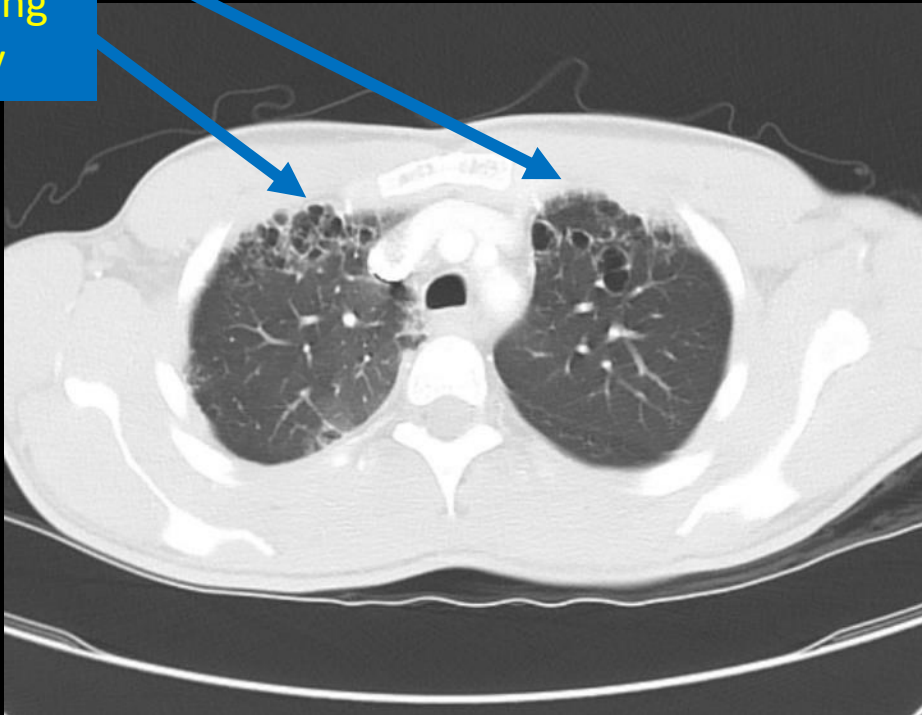
This imaging modality was ordered by the GI physician

Findings (unlabeled)

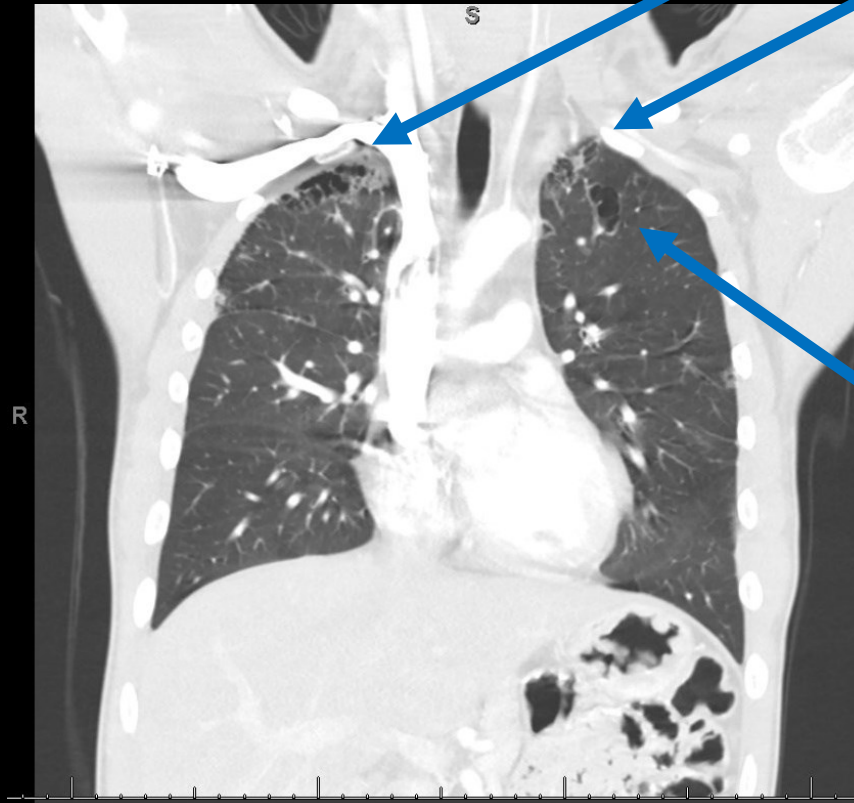


Findings (labeled)

Multiple bilateral lung cysts which are primarily centered in subpleural spaces/lung periphery



Cysts clustered within the anterior and medial aspects of the upper lobes with a maximal size of 1.2cm.



A small number of cysts are centered along the bronchovascular bundles

Final Dx:

Trisomy 21 Associated Interstitial Lung Disease

Trisomy 21 Interstitial Lung Disease

Definition:

- Interstitial (diffuse) lung diseases of infants and children (also known as chILD) include a heterogeneous group of parenchymal lung disorders with a wide range of etiologies including developmental, genetic, infectious, and inflammatory.
- Disorders of alveolar growth are the most common form of chILD, and typically occur in infants as a result of a superimposed condition or event that results in the abnormal development of the lungs.
- Trisomy 21 can be associated with an alveolar growth disorder characterized by the presence of small subpleural cysts (SPCs), that most commonly involve the anteromedial lung regions.
- These SPCs are seldom seen on chest radiography and are usually only appreciated on chest computed tomography.
- The clinical relevance of this imaging finding is not well understood. However, SPCs are more often found in pediatric Trisomy 21 patients with congenital heart disease.

Trisomy 21 Interstitial Lung Disease :

Epidemiology/Etiology:

- Trisomy 21 ILD is a known childhood phenomenon with several postmortem studies revealing a prevalence of up to 36% in pediatric patients with Trisomy 21.
- The formation of SPCs in Trisomy 21 ILD is likely a sequela of pulmonary hypoplasia as subpleural alveoli develop at a later stage compared to central lung regions after birth.
- This hypothesis is also supported by the fact that stillborn infants with Trisomy 21 do not have SPCs on autopsy.
- It is currently unclear whether SPCs spontaneously resolve with age in patients with Trisomy 21.

Trisomy 21 Interstitial Lung Disease :

Other Considerations:

- Additional differential diagnoses for pediatric lung cysts on chest CT include chronic lung disease/bronchopulmonary dysplasia (an additional chILD alveolar growth disorder), congenital pulmonary airway malformation, pulmonary lymphangiectasia, or regional scarring. These can often be differentiated from Trisomy 21 ILD by cyst distribution and the clinical history.
- These radiological findings of Trisomy 21 ILD can be alarming to providers when found on imaging and can be misinterpreted as findings concerning for cystic fibrosis or even honeycombing associated with end-stage lung disease.
- It is important to not confuse Trisomy 21 ILD with these other etiologies of cystic lung findings in this patient population as this may unnecessarily lead to additional imaging, laboratory testing, and patient/parental anxiety.

References:

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