



# Aerophagia and subcutaneous emphysema in a patient with Rett syndrome

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## BACKGROUND

Subcutaneous emphysema is a rare condition in which air accumulates within soft tissue planes. It typically occurs secondary to an identifiable trauma or iatrogenic insult. When an inciting event cannot be identified, it is described as “spontaneous.”<sup>1</sup>

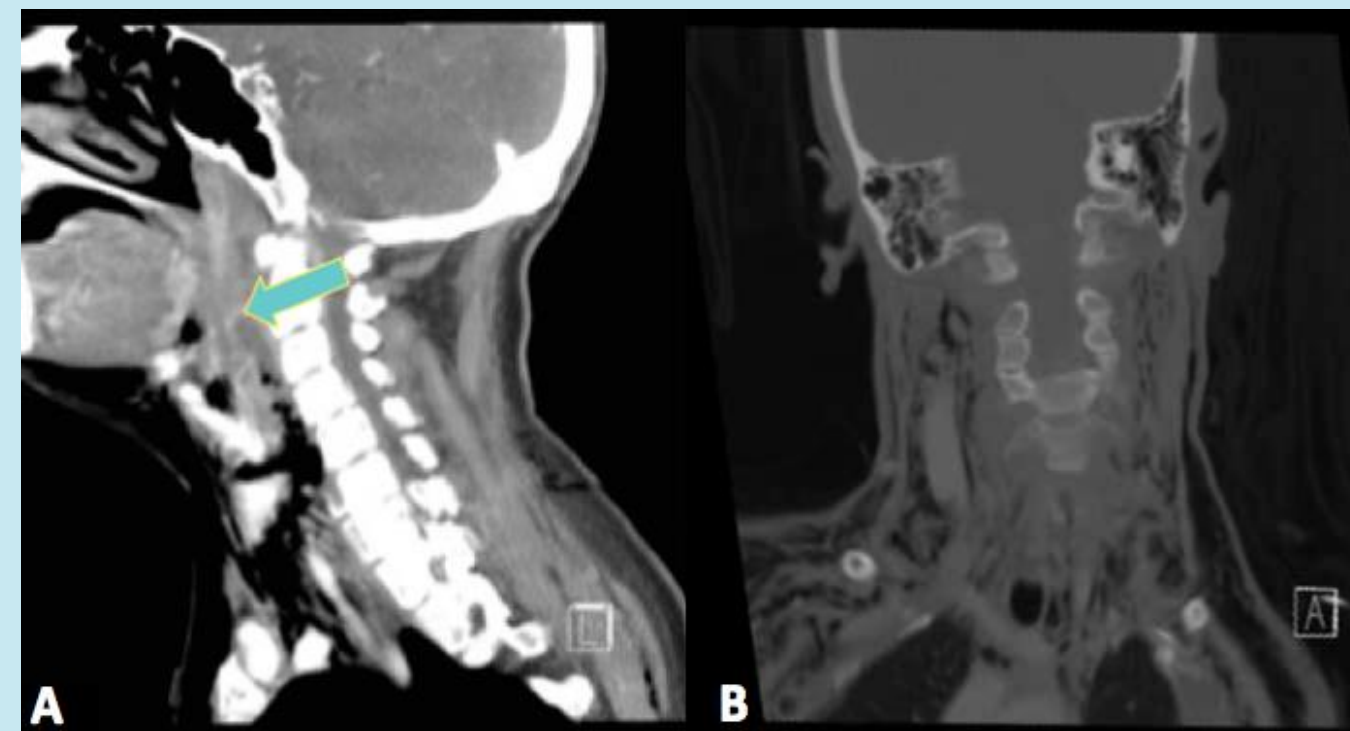
## CASE DESCRIPTION

A 43 year-old female with Rett syndrome presented to our ED with her parents for worsening neck swelling discovered two days prior by her caretakers. Her parents reported that she had a tendency to swallow air in a gulping fashion at rest; however, to their knowledge, she had not experienced any recent illnesses or trauma.

Diffuse subcutaneous air and crepitus were observed along the anterior aspects of her neck and upper chest wall bilaterally. CT of the neck and chest revealed a faint tubular ring-enhancing fluid collection and air in the left pharyngeal region and extensive subcutaneous emphysema involving the entire anterior, posterior, and lateral chest walls extending into the bilateral upper extremities and mediastinum (Figure 1).

An EGD revealed a small left-sided oropharyngeal tear just inferior to the uvula with mucosal edema and white purulence (Figure 2).

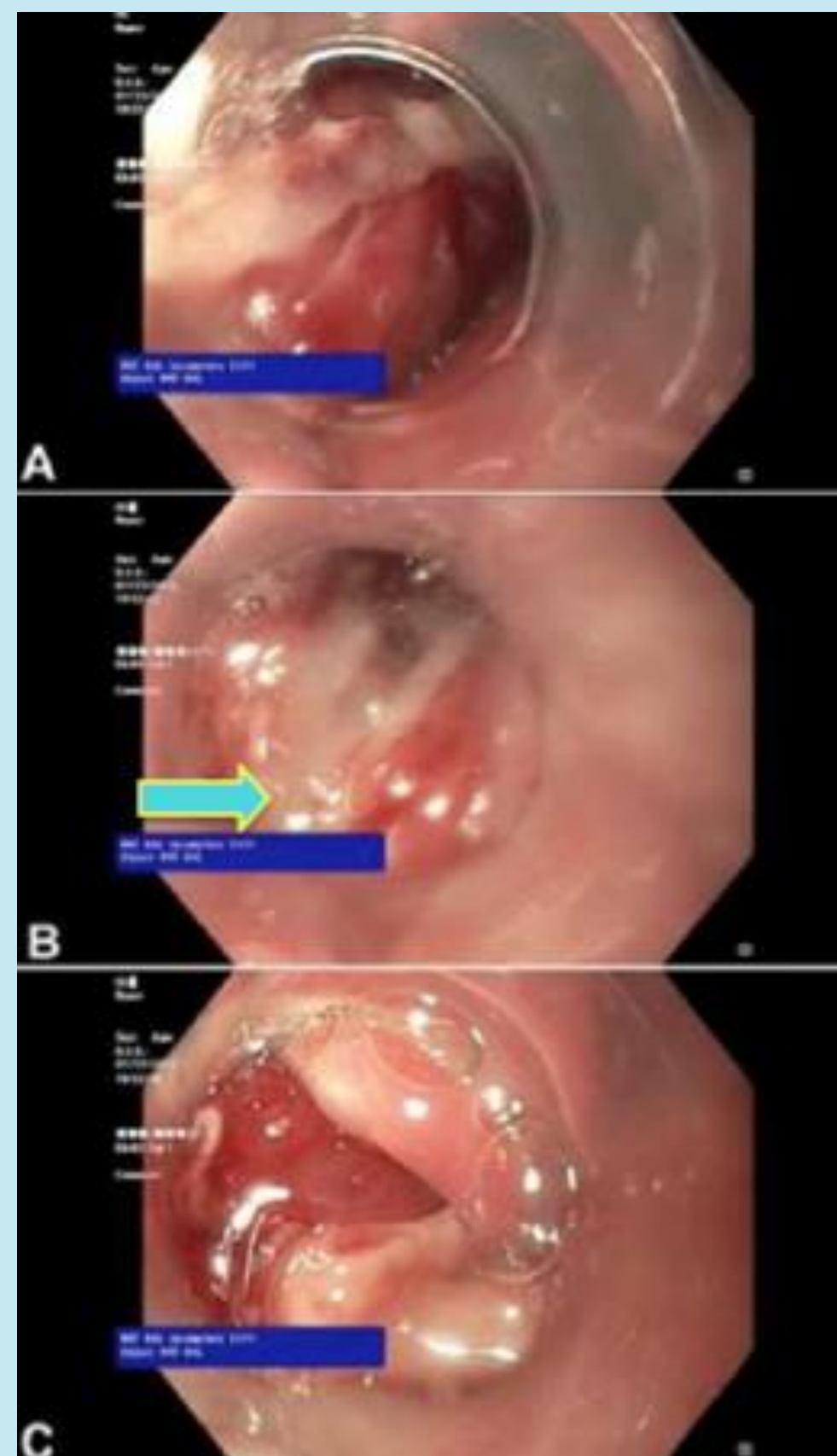
Otolaryngology was consulted, and she underwent DL under anesthesia, which revealed mucosal injury consistent with EGD findings. Flexible bronchoscopy revealed no evidence of tracheal or bronchial injury.



**Figure 1.**

(A) Sagittal CT image to the right of midline demonstrating retropharyngeal fluid collection (arrow) and air tracking

(B) Coronal CT image demonstrating extent of cervical and upper thoracic subcutaneous emphysema



**Figure 2.**

(A & B) Frank white purulence draining down posterior oropharyngeal wall (arrow) from oropharyngeal tear

(C) The tear demonstrated with a transparent cap tip

## CASE DESCRIPTION (cont'd.)

The patient remained in the hospital for 6 additional days. She was kept NPO and fed through an NG tube during this time and placed on 100% oxygen via face tent. Throughout her admission, staff noted intermittent aerophagia. Repeated CT on post-operative day 3 showed resolving crepitus. Outpatient follow-up at 2 weeks demonstrated symptom resolution.

## DISCUSSION

Subcutaneous emphysema is typically encountered in the head and neck due to the proximity of the airway.<sup>2</sup> Patients present with clinically palpable crepitus in affected areas.

Although it can result in fatal complications such as airway compromise and tension pneumothorax, subcutaneous emphysema is typically self-resolving and managed with bedrest and analgesics. Supplemental oxygen can be given to alter pressure gradients and improve the clinical course.<sup>3</sup>

In this case, we believe that our patient’s extensive subcutaneous emphysema of the cervical and mediastinal regions was a consequence of her tendency toward accentuated aerophagia, a known sequela of her neurodevelopmental disorder.

Patients with Rett syndrome exhibit six months of normal development followed by regression. Postnatal decelerations in head growth, loss of purposeful hand control with subsequent stereotypic movements, and impairments in gait and spoken language are common. Up to 50% of affected adults have been found to exhibit aberrant breathing patterns, including accentuated aerophagia, in which air is habitually swallowed at rest or during feeding.<sup>4</sup>

## DISCUSSION (cont'd.)

Our patient was noted to exhibit gulping of air at rest by her caregivers and hospital staff. Although no precipitating history of instrumentation was elicited, we believe that the most likely cause of the defect observed in her pharyngeal wall was still trauma of some nature, which served as a point of air entry. However, we cannot eliminate the possibility that her tendency towards accentuated aerophagia caused spontaneous subcutaneous emphysema. Regardless, her habitual air swallowing likely acted as a positive pressure source, promoting and worsening her subcutaneous emphysema.

Therefore, it is important to consider unique mechanisms, such as accentuated aerophagia, as possible etiologies and exacerbating factors for subcutaneous emphysema of unknown etiology.

## REFERENCES

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