

## Introduction

Although respiratory illness is quite common in pediatric patients, the importance of diagnosing typical (asthma, respiratory infections, and allergies) versus rare conditions such as Neuroendocrine Hyperplasia of Infancy (NEHI) is imperative. Due to factors such as patient's verbal communication level, minimal medical history due to age, and limited evaluations, NEHI could be overlooked. NEHI is difficult to diagnose because of the wide array of physical exam findings such as tachypnea, wheezing, and hypoxia which are common symptoms in many other diseases. This means that a detailed, physical exam is pertinent, especially lung sounds, to determine if further investigation is necessary.

The corresponding case is an 11-month-old male who presented to the emergency department with hypoxia and increased work of breathing. Due to patient having multiple respiratory symptoms with multiple admissions for acute hypoxic respiratory failure, the patient was transferred to Children's Hospital where a diagnosis of NEHI was made.

## Case Description

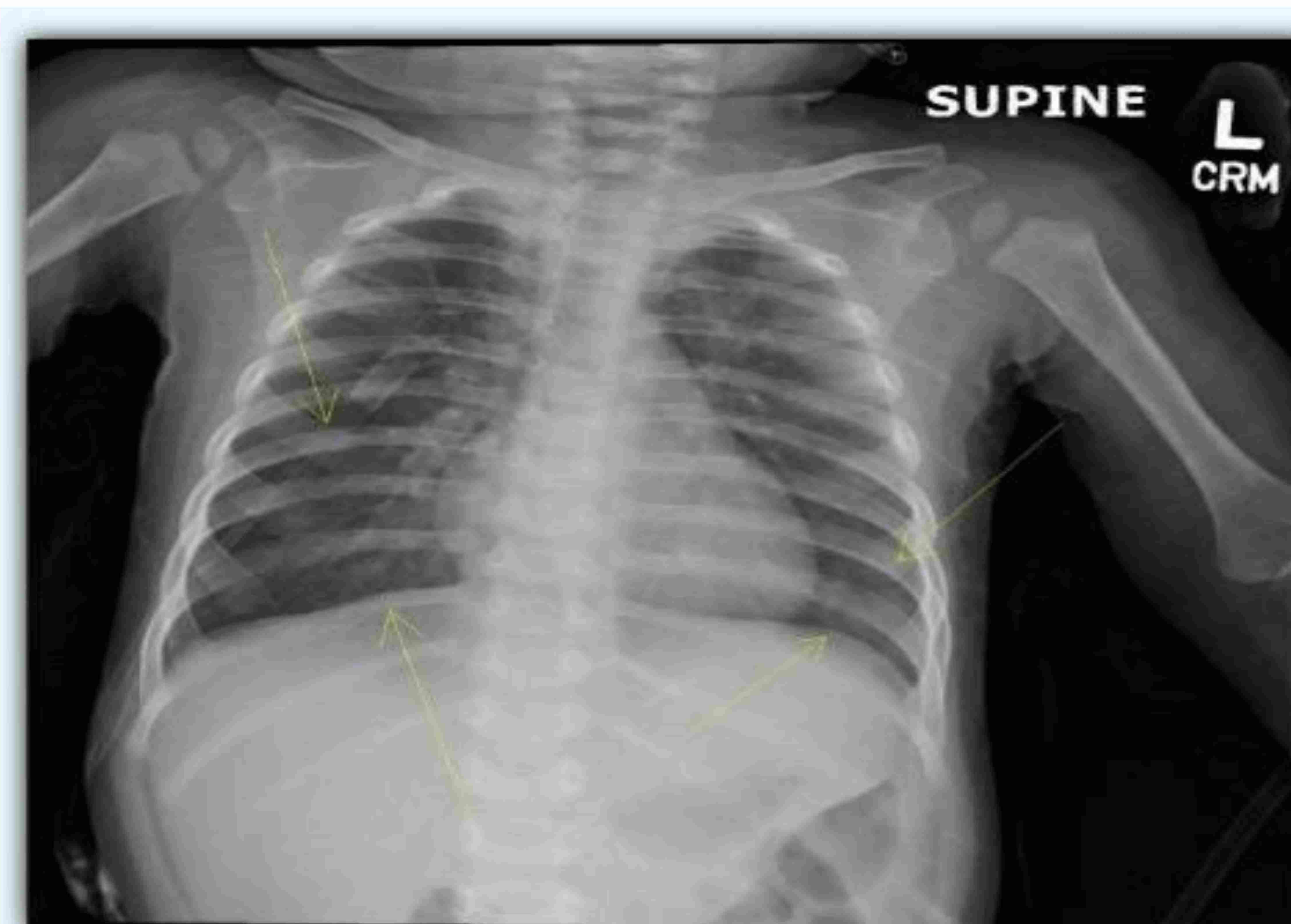
- An 11-month-old, Hispanic male with a past medical history of reactive airway disease who presented to the emergency room for evaluation of hypoxia and increased work of breathing.
- Prior to this admission, patient has had 5 admissions for acute hypoxic respiratory failure and pneumonia with hypoxia.
- Prior to presentation, guardian reports increased work of breathing and measured the oxygen saturation at 84% while sleeping.
- Vital signs:** BP 83/64, HR 132, RR 28, SPO2 98%, TEMP 98.8F
- Physical exam**
  - Increased work of breathing
  - Mild expiratory wheeze
  - Tachypnea
  - Lethargy
- Initial labs**
  - WBC 12.9, IgA 42, IgG 324, IgM 49, IgE <2
- Imaging**
  - CXR: Opacities in the right and left lung bases
  - CT: Bilateral ground glass consolidations predominantly involving the right middle lobe, lingula and Para mediastinal aspect of bilateral upper lobes.
- Plan**
  - Transfer to Children's Hospital

## Differential Diagnoses

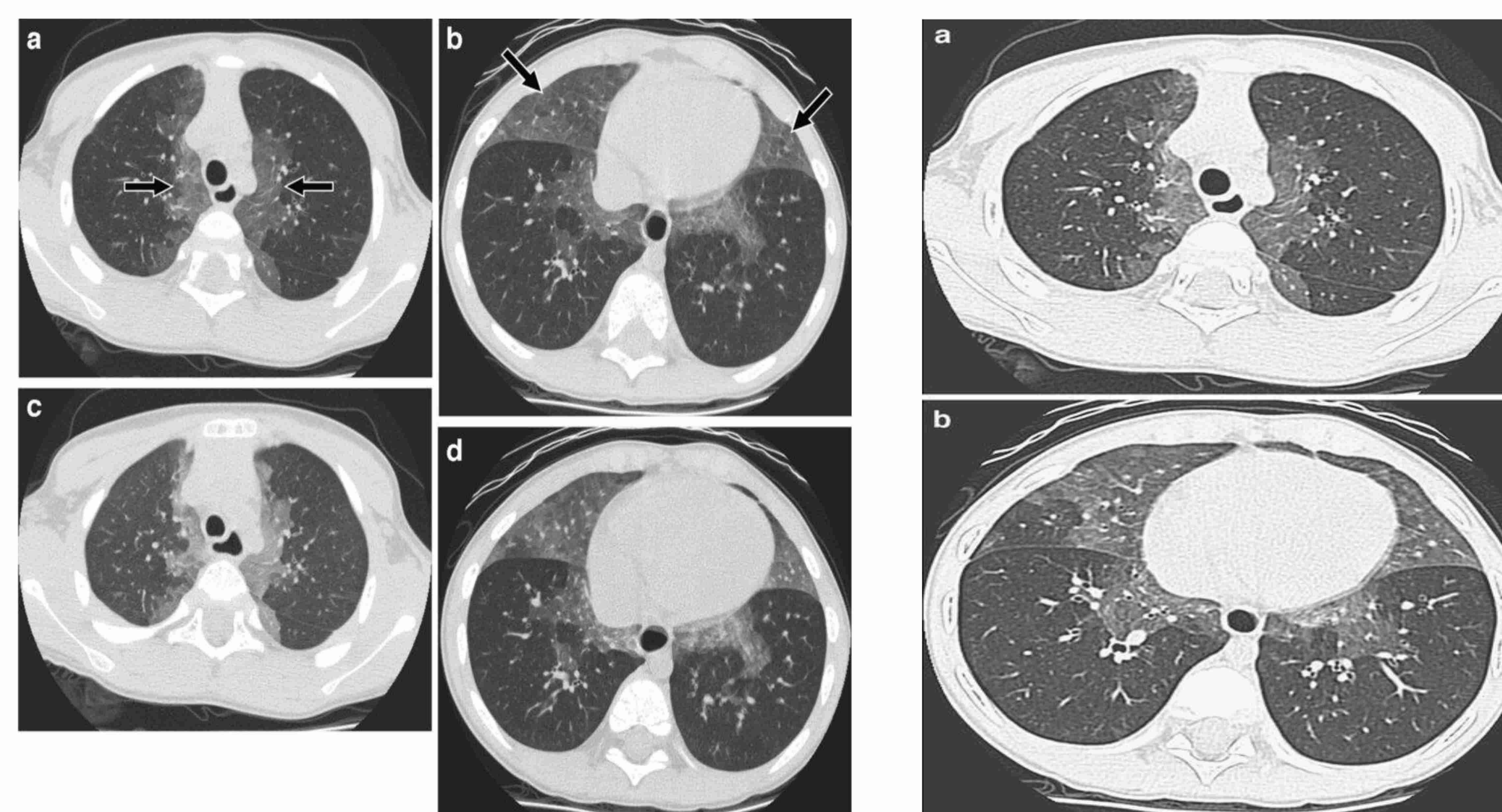
- Asthma
- Reactive airway disease
- Cystic fibrosis
- Bronchopulmonary dysplasia

## Diagnosis/Imaging

- Diagnosis is based on presenting complaints, frequency of symptoms with admissions, and radiological findings.
- The first step in evaluation of patients with persistent tachypnea is to exclude other common causes like asthma, acute or chronic infections, immunodeficiency, cystic fibrosis, allergies, and congenital heart disease.
- A thorough history should always be done.
- Chest x-ray In infants with NEHI may be normal or may reveal hyperinflation and perihilar opacities which are also consistent with viral infection.
- A high-resolution CT scan is key diagnostic tool;** it shows characteristic ground glass opacities centrally and in the right middle lobe and lingula, and air-trapping.



Above picture with arrows shows decrease opacities in the right lung base and possible minimal opacity in the left lung base.  
Below image shows ground glass opacities.



## Discussion

- NEHI, also called persistent tachypnea in infancy, is a rare lung disease found during the first year of life. The incidence and prevalence are unknown.
- The causes are unknown, but there is evidence suggesting that genetic and environmental factors may influence the development of NEHI.
- The symptoms of NEH includes tachypnea, increased work of breathing (retractions), hypoxemia, lethargy, and failure to thrive.
- In most cases, patients usually present to the physician's office with viral URTI. Sometimes, patients are misdiagnosed because their respiratory symptoms are mistakenly attributed to common disorders like GERD.
- On physical exam, tachypnea and retractions are common clinical features. Wheezing may also be heard if there is a superimposed viral infection. These symptoms can be mistaken for common respiratory conditions leading to initial misdiagnosis.
- Many infants with NEHI usually have trouble in gaining weight or failure to thrive.
- Diagnosis is based on presenting complaints and radiological findings. The first step in evaluation of patients with NEHI is to exclude other common causes like asthma, acute or chronic infections, immunodeficiency, allergies, cystic fibrosis, and congenital heart disease through history taking and specific physical exam findings.
- In infants with NEHI, chest x-ray may be normal or may reveal hyperinflation and perihilar opacities which are also consistent with viral infection.
- A HRCT scan is the key diagnostic tool. It shows characteristic ground glass opacities centrally and in the right middle lobe and lingula and air trapping.
- Additionally, infant pulmonary function test can also be done to confirm diagnosis. PFTs in patients with neuroendocrine hyperplasia reveals a mixed physiological pattern including profound air trapping evidenced by reduction in FEV and FVC. There is also a marked elevation in FRC, RV and TLC.
- If these tests are inconclusive, or symptoms are not typical of neuroendocrine hyperplasia, invasive test like lung biopsy can be done. The biopsy is used to identify increased presence of pulmonary neuroendocrine cells.
- There is no specific treatment for NEHI. Management consists of supportive and preventative care. Supplemental oxygen can be used in patients who are hypoxic. Due to failure to thrive, many patients will also require nutritional supplementation.
- Parents should be encouraged on immunization with the influenza and pneumococcal vaccines.

## Conclusion

- Therefore, the importance of having a broadened differential when the clinical picture does not match typical symptoms; it is critical that one must consider NEHI.
- The long-term outcomes for NEHI has not been well-established. In most cases, respiratory symptoms improve with time. Some patients require oxygen therapy for several years. Failure to thrive has been associated with a longer duration of supplemental oxygen use. The prognosis varies as some patients become asymptomatic at rest, and some continue to have exercise intolerance or have an exacerbation of their symptoms during respiratory infections.

## References

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