

Neuro-endocrine cell hyperplasia of Infancy: clinical presentation and diagnosis

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Introduction

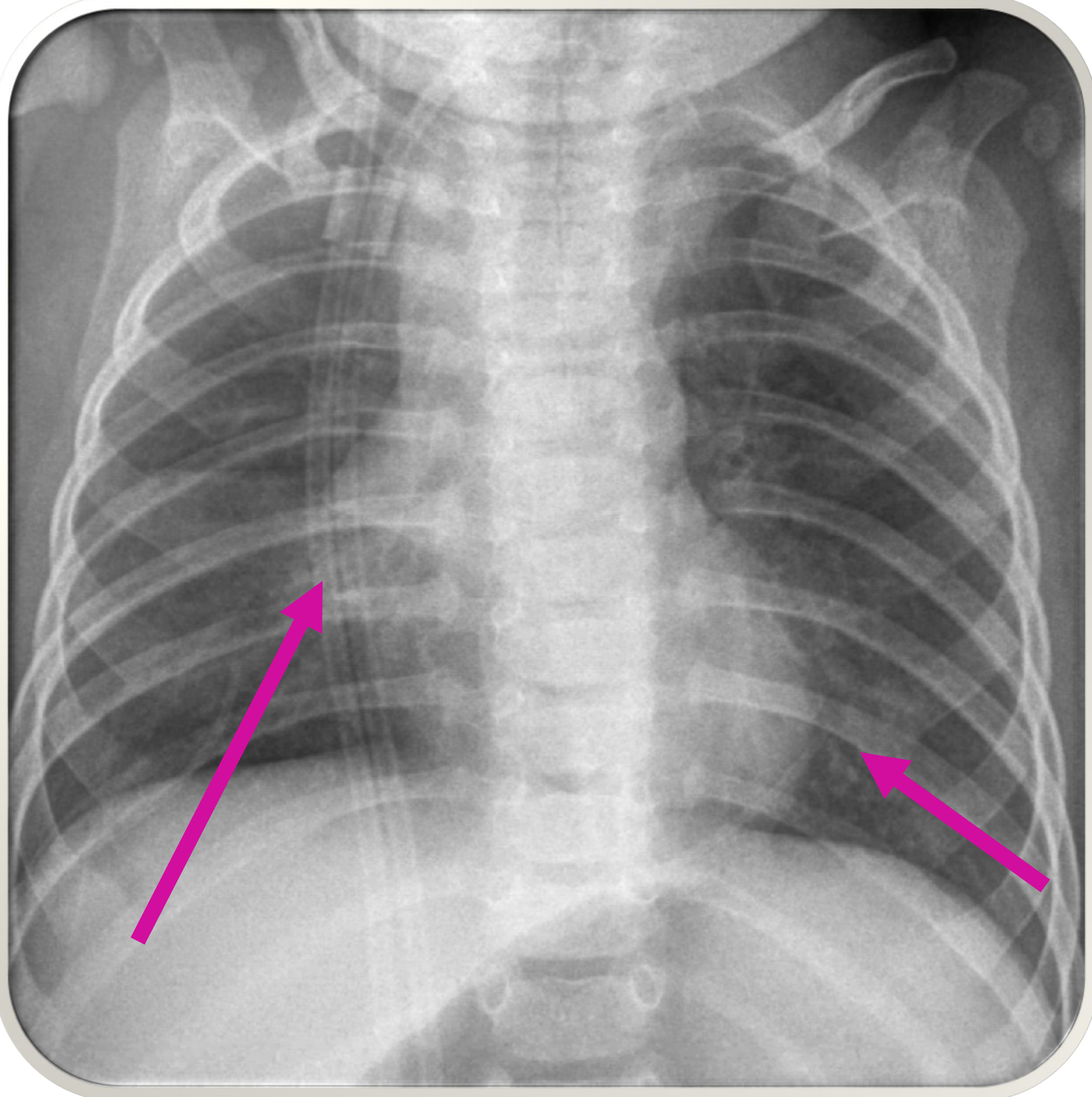
- **Neuro-endocrine cell hyperplasia pf infancy (NEHI) is a childhood interstitial lung disease (chILD)**
- First described in 2005 as persistent tachypnea of infancy associated with neuroendocrine cell hyperplasia in lung biopsy by Deterding et al.
- Most common symptoms in infancy include persistent tachypnea, hypoxia and chest retractions. Presentation may also involve failure to thrive and few respiratory symptoms (no baseline cough, wheezing, or clubbing).
- **Clinical score by Liptzin et al. can assist in making the diagnosis**

Case report:

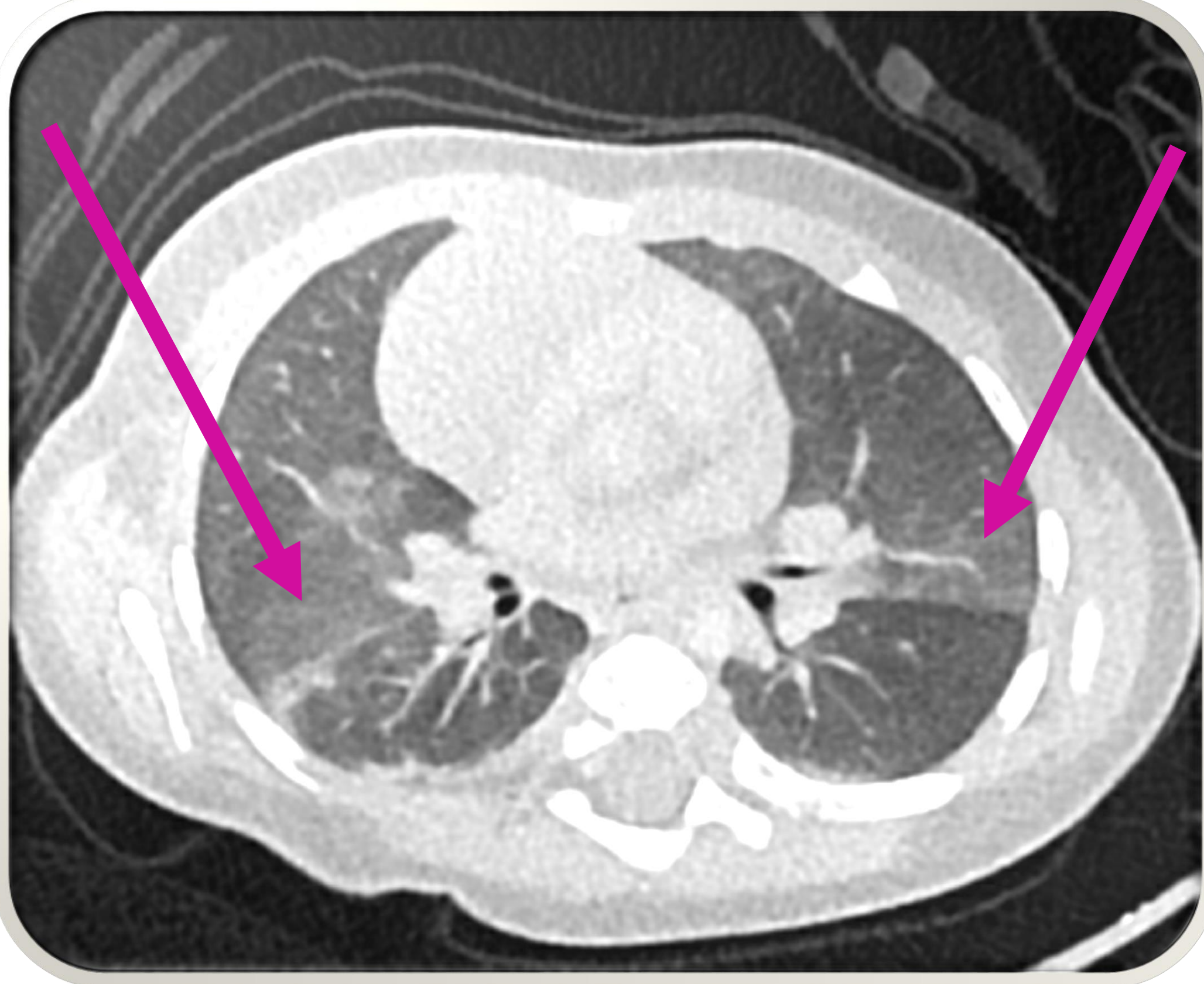
- **A 5 –month old boy** with **recurrent episodes of respiratory distress, with signs of increasing intermittent tachypnea and chest retractions over 2 months**
- **Clinical picture:**
 - Respiratory distress with no specific trigger,
 - No history of respiratory infection,
 - Antenatal history: born at term by cesarian section for maternal indication, no respiratory distress at birth
- **Physical examination:**
 - Severe tachypnea, chest retractions and hypoxia (oxygen saturation at 88%), discrete crackles heard on superior lobe
 - No failure to thrive but difficulty in starting diversification.
 - Cardiovascular and ENT examination was normal, normal physical exam.
- **Liptzin Score: 8/10**
- **Radiological findings**
 - **Chest X-ray:** Chest hyperinflation and **ground glass opacification** involving the right middle lung and the left lingular area.
 - **Hight Resolution Computed Tomography:** **Mosaic pattern of ground glass opacities and in the superior lobes, right middle lobe and left lingula, with air trapping in the inferior lobes**
- **Biological analysis:**
 - **Full hematological workup, renal, liver, thyroid fonctions:** normal
 - **Immunological workup:** no immunodeficiency
- **Guthrie test:** negative for cystic fibrosis
- **Broncho-alveolar lavage:** no signs of cellular damage, infections or neoplasia
- **Biopsy:** risks outweigh benefits, deemed unnecessary as high chest CT specificity
- **Treatment:** Short courses of i.v corticosteroids and home oxygen supplementation with a favourable response

Liptzin Score: NEHI Clinical Score		
Chest retractions		/1
Tachypnea		/1
Hypoxemia		/1
Chest wall abnormality		/1
Failure to thrive		/1
Crackles		/1
No clubbing		/1
No baseline wheezing		/1
Symptoms before 12 months		/1
Chest wall abnormality		/1
>7/10: Consistent with NEHI		

Liptzin, D.R., Pickett, K., Brinton, J.T., et al. 2020. Neuroendocrine Cell Hyperplasia of Infancy. Clinical Score and Comorbidities. Ann. Am. Thorac. Soc. 17, 724–728.



Ground-glass opacification involving the right middle lung and left lingular area



Mosaic pattern of ground glass opacities in the right middle lobe and left lingula

Conclusion

- **Rare form of childhood interstitial lung disease with a clinical score and radiological diagnosis**
- High resolution Computed tomography (chest CT) with a typical ground-glass pattern contributes to the diagnosis and the estimation of severity of neuro-endocrine cell hyperplasia of infancy.