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 lobe and the left lingular area.
  - High 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 functions: 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

<table>
<thead>
<tr>
<th>Liptzin Score: NEHI Clinical Score</th>
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<tbody>
<tr>
<td>Chest retractions</td>
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<tr>
<td>Tachypnea</td>
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<tr>
<td>Hypoxemia</td>
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<tr>
<td>Chest wall abnormality</td>
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<tr>
<td>Failure to thrive</td>
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<tr>
<td>Crackles</td>
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<tr>
<td>No clubbing</td>
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<tr>
<td>No baseline wheezing</td>
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<tr>
<td>Symptoms before 12 months</td>
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<tr>
<td>Chest wall abnormality</td>
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</tbody>
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>7/10: Consistent with NEHI

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.