Case Report

A 10-month-old infant with respiratory distress and hypoxemia

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Abstract

A 10-month-old infant with Prader-Willi Syndrome presented with a 7 month history of increased work of breathing, wheeze, inspiratory crepitations, and mild hypoxemia. Subsequent investigations including chest CT suggested the diagnosis to be neuroendocrine cell hyperplasia of infancy (NEHI). NEHI is a rare cause of children's interstitial lung disease. Childhood interstitial lung disease should be considered in an infant with persistent tachypnea, crepitations, and hypoxemia.

Case Presentation

A 10-month-old girl with Prader-Willi Syndrome presented to hospital with respiratory distress, tachypnea, and a history of three previous emergency department visits and two admissions over seven months with similar symptoms. During her previous visits, she was thought to have acute viral bronchiolitis with increased work of breathing, tracheal and subcostal indrawing, wheeze, inspiratory crepitations, and mild hypoxemia (SpO2 88%). Between visits her mother reported increased work of breathing with intermittent grunting and tracheal and subcostal indrawing.

Other than the diagnosis of Prader-Willi Syndrome made during the neonatal period her previous history had been unremarkable. She had been feeding normally and gaining weight.

On admission she required oxygen by nasal prongs at 0.5 L/min to keep her SpO2 greater than 90%. Her weight was 7.45 kg (10th-25th percentile), heart rate 156 per minute, respiratory rate 48 per minute, temperature 37.9oC. She demonstrated increased work of breathing with tracheal and subcostal indrawing, no cough, and on auscultation, symmetrical breath sounds, no wheeze, but bilateral inspiratory crepitations. She had no finger clubbing and the remainder of the exam was consistent with her Prader-Willi Syndrome.

The nasopharyngeal aspirate was negative for respiratory viruses (as it was on four previous occasions). Capillary gas pH 7.34, pCO2 57.1, base excess 3.5, bicarb 30. Cardiology assessment and echocardiogram were normal. Chest radiograph showed mild increase in the size of the retrosternal airspace with flattening of the diaphragms. There was evidence of mild atelectasis in the region of the right middle lobe. A previous barium/formula feeding study was normal. Newborn screening was negative for Cystic Fibrosis. The clinical findings and investigations were not consistent with viral bronchiolitis, asthma, cystic fibrosis, pulmonary aspiration, or pneumonia. Because of the persisting clinical picture and radiographic evidence of air trapping, a chest computed tomogram (chest CT) was performed.

Chest CT revealed diffuse ground glass opacities of the right middle lobe and minimal subsegmental atelectasis (see figure). The clinical picture and chest CT findings were felt to be most consistent with neuroendocrine cell hyperplasia of infancy (NEHI) in consultation with Pediatric Respirology.

Discussion

NEHI is a rare cause of children's interstitial lung disease (cHILD) previously known as persistent tachypnea of infancy. It manifests in otherwise well infants with tachypnea, retractions, crepitations, hypoxemia, and rarely cough. The pathogenesis is thought to be linked to pulmonary neuroendocrine cell prominence, although the exact process is unknown. When performed, lung biopsy demonstrates staining for neuropeptide bombesin in distal airway cells. High resolution CT (HRCT) findings of ground glass opacities in the right middle lobe and lingula, in some patients,

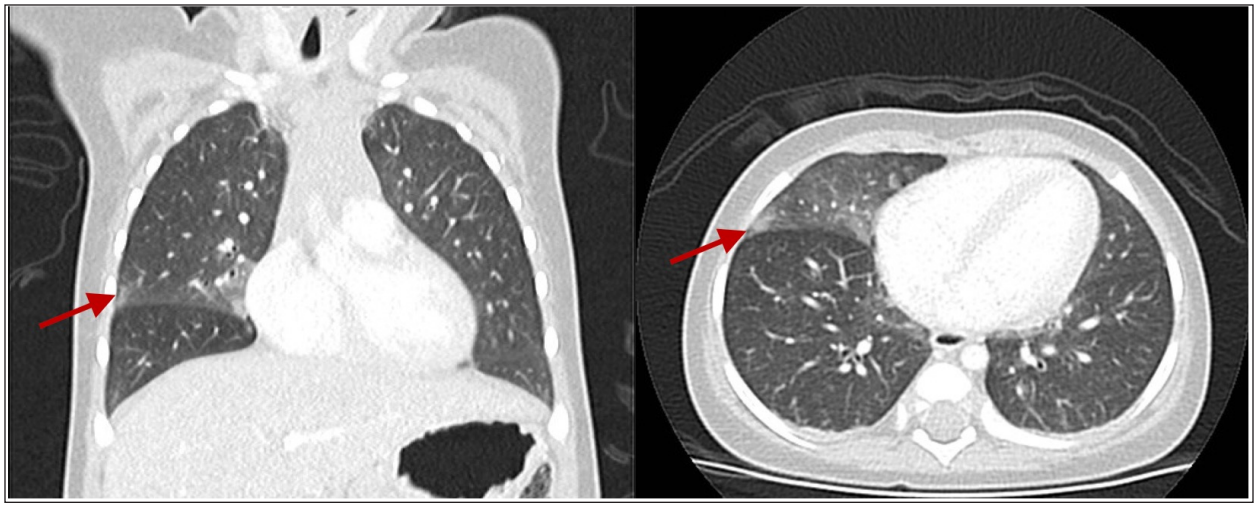

Figure 1.

have been suggested to be diagnostic and avoiding the need for a lung biopsy[1,2].

The treatment of NEHI is supportive; providing extra calories to optimize nutritional status, if failing to thrive, and oxygen supplementation. Caution will be required in our patient because of the weight increase seen around 18-36 months in Prader-Willi Syndrome and the risk of obstructive sleep apnea. Some patients require oxygen 24 hours per day while others only at night and with illnesses. Most patients gradually reduce and cease their need for supplemental oxygen although in follow-up some may develop asthma[3]. Seasonal influenza shots and respiratory syncytial virus prophylaxis are recommended as for all children with significant respiratory problems.

Our patient was initially thought to have viral bronchiolitis but the prolonged course (7 months) made this extremely unlikely. Cardiac assessment was normal and there was no clinical nor imaging evidence of swallowing dysfunction or gastroesophageal reflux.

The etiology of NEHI is unknown but genetic mechanisms may play a role as familial patterns are seen in some cases associated with hypothyroidism. Our patient had normal thyroid function. We are not aware of any association between Prader-Willi Syndrome and NEHI.

In preparation for growth hormone therapy for Prader-Willi Syndrome, a pediatric polysomnogram was performed which showed, in room air, mild hypoxemia and a central apnea-hypoxia index (CAHI) of 16.3 per hour; with supplemental oxygen the CAHI fell to 4 per hour. The fall in this index with oxygen is the usual response in a patient with PWS.

Our patient was discharged on supplemental oxygen for both the central apneas of Prader-Willi Syndrome and the hypoxemia of NEHI and follow-up arranged.

Clinical Pearls

1. Childhood interstitial lung disease should be considered in an infant with persistent tachypnea, crepitations, and hypoxemia.
2. While lung biopsy is the gold standard for NEHI, the classic clinical picture and characteristic CT findings are felt to be diagnostic.

References

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