ROHHAD Syndrome Associated With a Neural Crest Tumor in a Pediatric Patient: A Case Report

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Introduction

Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation (ROHHAD) syndrome is a rare, life-threatening pediatric disorder, with fewer than 200 cases reported as of 2022, suggesting considerable underdiagnosis. The acronym reflects the typical disease progression, with rapid weight gain as the initial manifestation, most commonly presenting between ages 2 and 7 in previously healthy children. Without early intervention, prognosis is poor with a mortality rate of 50-60%, primarily due to cardiorespiratory arrest secondary to central hypoventilation.

Case

A 3-year-old female presented to the ED with shortness of breath, hypoxemia, and fever, requiring intubation for respiratory distress. Initial lab values revealed:

- Hypernatremia (164 mmol/L)
- Hyperprolactinemia (39.7 ng/mL)
- Hyperinsulinemia (23.7 uU/mL)
- Low TSH (0.082 uIU/mL)

Chest x-ray showed no infiltrate, and respiratory PCR was negative. Family history revealed progressive decline over the past year with multiple hospitalizations for similar presentations, rapid weight gain of 25 pounds, and speech regression. The constellation of laboratory findings and clinical history prompted evaluation for ROHHAD syndrome. A noncontrast CT of the abdomen and pelvis was ordered to look for associated neural crest tumors, which revealed a left periaortic retroperitoneal mass adjacent to the left renal hilum with heterogeneous enhancement and fatty density suggesting a ganglioneuroma/ganglioneuroblastoma.

Complications of ROHHAD Syndrome

Cardiorespiratory

- Central hypoventilation
- Cor pulmonale / right ventricular hypertrophy
- Cardiorespiratory arrest

Endocrine

- Hyperprolactinemia
- Hypernatremia
- Hypothyroidism
- Growth hormone deficiency
- Adrenal insufficiency
- Precocious puberty

Autonomic Dysregulation

- Altered pain perception
- Temperature dysregulation
- Profound bradycardia
- Hypotension
- Hypertension

Neuropsychiatric

- Personality changes
- Developmental/speech regression
- Seizures

Clinical Course

After initial stabilization of the patient, a multidisciplinary approach was taken with several discussions between the hospitalist service, rheumatology, neurology, and surgery. IVIG was initiated and surgery successfully resected the tumor. Pathology confirmed a ganglioneuroblastoma with favorable histology. Follow-up with Boston Children's Hospital for ZSCAN1 antibody testing has been coordinated.

Discussion

Early diagnosis and treatment of ROHHAD Syndrome is critical to improve patient outcomes. A high index of suspicion is imperative for any child presenting with rapid-onset obesity, particularly when accompanied by hyperprolactinemia. The most critical factor to improve survival in these patients is early diagnosis and timely initiation of respiratory support. Long-term management requires lifelong multidisciplinary care; most patients require either nocturnal BiPAP or mechanical ventilation for ventilatory support, along with hormone replacement therapy.

References

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