Large Pleuropulmonary Blastoma Mimicking Congenital Pulmonary Airway Malformation in an Infant: A Case Report

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INTRODUCTION

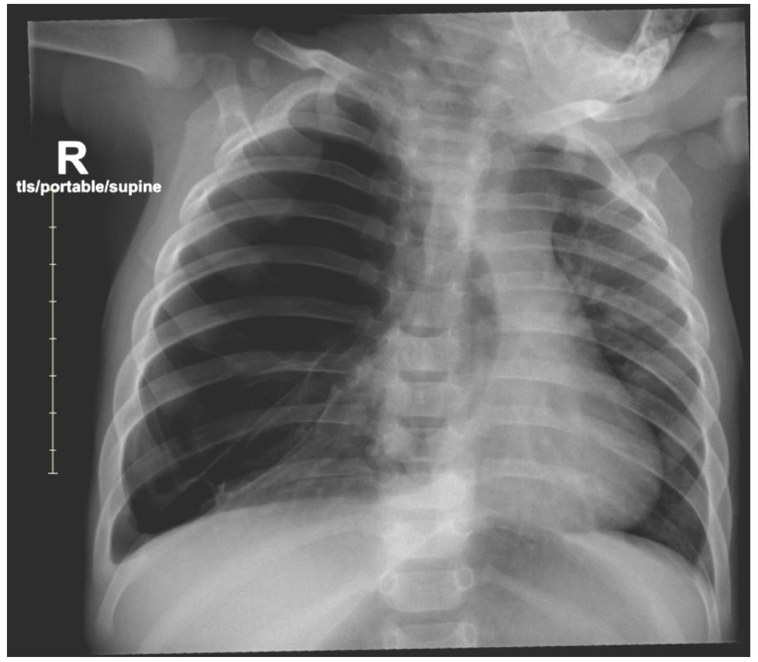
- Pleuropulmonary blastoma (PPB) is a rare pediatric lung neoplasm that occurs in children under 5 years old, frequently misdiagnosed as congenital pulmonary airway malformation (CPAM) due to similar cystic imaging appearances. Early and accurate diagnosis is critical for prognosis and management.
- This case presents a COVID-positive infant with initially suspected CPAM. Surgery removed the mass, and histopathology confirmed PPB type Ir. No further intervention was needed, and the patient was back at baseline shortly thereafter.

CASE PRESENTATION

- A 10-month-old male presented to the emergency department (ED) with a 3-day history of upper respiratory symptoms
- A viral panel tested positive for COVID-19
- At arrival, blood pressure was 110/94, heart rate 184, temperature 40.1 C (rectal), O_2 saturation mid-90's to high 80s.
- Physical exam showed subcostal retractions, decreased right-sided breath sounds, prominent use of accessory muscles, and greater left chest wall excursion than right.
- CXR (right) revealed a large multicystic lesion on the right, causing significant mediastinal shift. CT angiogram showed a 11x11x4 cm lesion consistent with CPAM or neoplasm. Two hypodense lesions were also noted in the right kidney and thyroid.
- Pediatric Surgery was consulted and surgery was initially delayed 5–6 weeks due to active COVID infection. However, repeat imaging showed worsening mediastinal shift, warranting urgent surgical intervention. He was started on Remdesivir for COVID. He underwent thoracoscopic resection of the lesion.
- On hospital day 2, postoperative imaging confirmed lung re-expansion and resolution of mediastinal shift. He was discharged on day 3 with no complications. Parents reported prompt return to baseline activity.
- Gross pathology suggested PPB rather than CPAM

HISTOPATHOLOGY AND GENETIC FINDIINGS

- Pathology showed a mass with multilobulated, thin-walled cysts with an absence of primitive mesenchymal cells.
- Immunohistochemistry showed <15% p53+, patchy desmin, and absent myogenin. Collectively, this confirmed PPB type Ir.
- Genetcic testing of the tumor was positive for DICER1 somatic pathogenic variants. Germline variant suspected.
- The family moved locations due to insurance complications. The new oncology team found that the lesions on the kidney and thyroid were benign. The family was advised to obtain subsequent genetic testing under the care of a new team.



CXR of large multicyclic lesion in right. Note mediastinal shift due to mass effect

DISCUSSION

- PPB is often confused with CPAM on imaging; CT sensitivity/specificity is 58%/83%. Prognosis for PPB is dependent on type, with a near 100% 5-year survival rate for type Ir. For types I–III, survival decreases to 91%, 71%, and 53%, respectively.
- Pathology and IHC are vital to distinguish type I vs. Ir. The mass mimicked a CPAM on imaging, but on resection, was very similar to a type I mass because of its size. Both types have a multilocular cystic nature, but type Ir tumors will begin regressing. Pathology and IHC were able to confirm type Ir, despite the size
- DICER 1 germline pathogenic variants are found in ~70% of PPB cases. Genetic testing is especially recommended patients' siblings. Such pathogenic variants put patients at risk for PPB, cystic nephroma, multinodular goiter, thyroid carcinoma, and other neoplasms. Monitoring is necessary if patient is positive for DICER1 germline pathogenic variants.
- Due to poor insurance coverage, this patients family had to transfer care to another state (where they had lived previously) to pursue genetic testing and monitoring, including monitoring the thyroid and kidney lesions. This highlight the struggles many families have with underinsurance and the need for cross-team communication.

CONCLUSION

PPB should be considered in infants with cystic lung lesions and respiratory distress. Imaging alone is not diagnostic—histopathology is essential. Surgical resection is curative in Type Ir. Importantly, genetic testing for DICER1 pathogenic variants is recommended for patients and first-degree relatives.

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