Pleuropulmonary blastoma in combination with cystic nephroma

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Patient: 21 months, male

Clinical History:

A previously healthy 21-month-old Caucasian boy presented with a history of vomiting and a left-sided abdominal mass. The antenatal history was unremarkable. He was born in the 39th week of pregnancy by caesarean section. His parents had no medical history and no family history of tumours and other genetic disorders.

Imaging Findings:

Abdominal ultrasound showed two multilocular cystic lesions in the left kidney with multiple anechoic spaces traversed by thin septations. Abdominal MRI demonstrated two multilocular cystic masses in the left kidney, without evidence for liver or lymph node metastasis or invasion in inferior vena cava.

Chest radiograph showed a large radiolucent lesion in the left upper and middle hemithorax without definite bronchovascular markings.

CT demonstrated one large and a few small cystic air-containing cavities with no fluid levels of the left upper lobe. A few small cystic lesions were also found in the left lower lobe. The right lung was normal and there was mild mediastinal shift to the right side.

The patient underwent thoracoscopic excision of the lesion, and nephrectomy of the left kidney. Histopathology revealed an intrapulmonary cystic lesion with pleural adhesions, covered partially with respiratory-like and unclassified epithelium, in addition to cell-rich blastomatous areas beneath the epithelium.

Discussion:

Pleuropulmonary blastoma (PPB) is an embryonal tumour of the lung, which only occurs in young children under 6 years of age at diagnosis [1]. PPB is related to family tumour susceptibility syndrome associated with pulmonary cysts, cystic nephroma (CN) and other embryonal neoplasms, including rhabdomyosarcoma and Wilms’ tumour [2].

A review of the International Pleuropulmonary Blastoma Registry (IPPBR) (www.ppregistry.org) revealed that cystic nephroma (CN) or related tumours were found in 9.2% of 152 registry-reviewed PPB cases [3, 9]. The registry identified 18 patients with PPB associated with 20 renal tumours (15 CN), either in the same patient or a family member. Eleven children had both PPB and renal tumours (7 CN).

The data from (IPPBR) indicate that 40% of the cases with PPB may have a genetic predisposition [4]. Recently,
PPB has been linked to mutations in the DICER-1 gene. This gene encodes an enzyme that negatively regulates gene expression [5, 8].

PPB has been classified into 3 groups based upon the characteristics of the tumour: type 1, purely cystic; type 2, intermediate cystic/solid; and type 3, predominantly solid [6, 10].

Current data support the assertion that type 1 PPB may progress to the more aggressive types 2 or 3 PPB. [1].

Most cases of PPB present as recurrent respiratory infections, respiratory distress and in some cases as recurrent pneumothorax [7].

Radiograph is the first imaging modality for respiratory symptoms. CT is typically used for evaluation of the initial extent of the disease and monitoring after resection and treatment.Appearances of PPB depend on the type. Type 1 appears as air-filled lung cysts associated with multiple septations, type 2 is an air-filled cystic mass with variable soft tissue component and type 3 is a large, heterogeneous, solid mass.

PPB Type 1 is indistinguishable from benign lung cysts, based on imaging findings, especially Congenital Pulmonary Airway Malformation (CPAM) type 1 and 4 [1, 2]. Therefore many congenital cystic lung lesions are removed surgically because of the risk of recurrent infection and possible underlying neoplasm.

Treatment of PPB type 1-3 includes surgical resection +/- chemotherapy. Benefit of local radiation in type 2 and 3 PPB is controversial. Lesions that are not removed should be monitored closely.

Five-year survival is 85-90% in type 1 and 45-60% in type 2 and 3 [2].

In conclusion PPB type 1 should be considered in the differential diagnosis in children with renal tumours and cystic lung lesions.

**Differential Diagnosis List:** Pleuropulmonary blastoma in combination with cystic nephroma, CPDN (Cystic Partially Differentiated Nephroblasoma) with congenital lobar emphysema, Pleuropulmonary blastoma in combination with cystic nephroma, Wilms’ tumour with pneumothorax, Mesoblastic nephroma with congenital pulmonary airway malformation (CPAM), Renal cyst with cavitary necrosis complicating pneumonia

**Final Diagnosis:** Pleuropulmonary blastoma in combination with cystic nephroma

**References:**


Description: Large radiolucent lesion in the left upper and middle hemithorax without definite bronchovascular markings. Origin: Department of Radiology Heidelberg/ Germany
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**Description:** The left upper lobe shows one large and a few small cystic air-containing cavities. Few small cystic lesions were also found in the left lower lobe. Mild mediastinal shift to the right side. **Origin:** Department of Radiology Heidelberg/ Germany
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Figure 3

a

Description: Two multilocular cystic masses in the left kidney, without evidence for liver or lymph node metastasis or invasion in inferior vena cava. Origin: Department of Radiology Heidelberg

b

Description: Two multilocular cystic masses in the left kidney, without evidence for liver or lymph node metastasis or invasion in inferior vena cava. Origin: Department of Radiology Heidelberg
Description: Two multilocular cystic lesions in the left kidney with multiple anechoic spaces traversed by thin septations. Origin: Department of Radiology