



## Adult pulmonary blastoma in an adult with urothelial cancer: A case report

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### ABSTRACT

Pulmonary blastoma is an aggressive lung cancer with incidence ranging from 0.25–0.5 of all the reported lung cancers. Although, pulmonary blastoma is seen commonly in childhood its very rare in adults.

Surgical treatment is often the treatment of choice, but benefits of neoadjuvant chemotherapy are unclear.

People with DICER1 syndrome commonly develop Pulmonary blastoma and do have concomitant or previous history of benign or malignant tumours in extra pulmonary site like kidney, thyroid, ovary cervix testicle and eye.

As per our knowledge, this is the first case of adult pulmonary blastoma previously diagnosed with urothelial cancer and a strong familial predilection of malignancy, with negative genetic test for DICER1 mutations.

### 1. Introduction

Pulmonary blastoma is a rare type of aggressive lung cancer with a poor prognosis and an estimated incidence of 0.5% among all pulmonary neoplasms [1,2]. Tumours are composed of immature malignant epithelial and/or mesenchymal tissue with features that resemble early embryological lung tissue [2,3]. Surgery is the standard treatment and the efficacy of adjuvant chemotherapy and radiotherapy has not yet been established [3]. We describe a rare case of this unusual pulmonary malignancy with previous history of urothelial cancer along with strong familial disposition to cancer, with possibility of DICER1 syndrome.

### 2. Case report

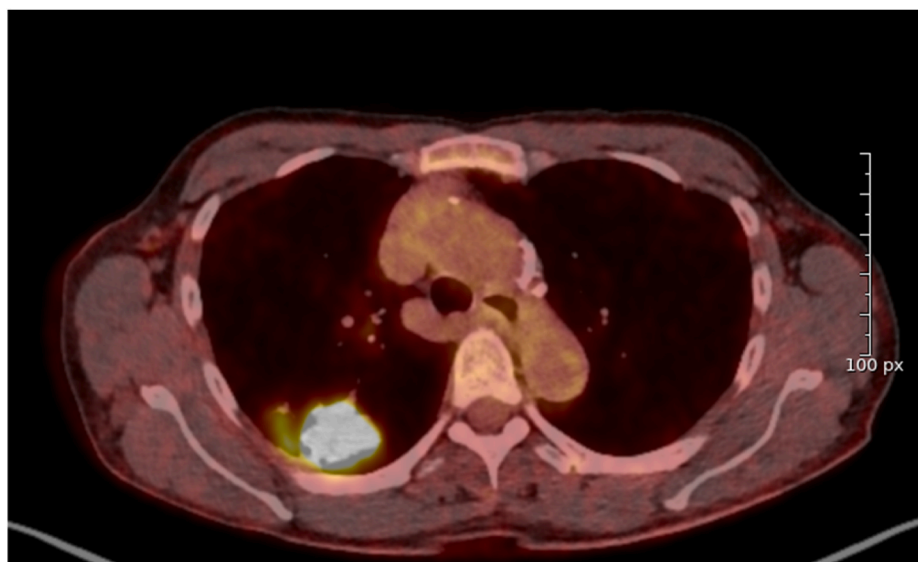
A 64-year-old woman presented with a 2-week history of haemoptysis and a 4-week history of pain in the neck, shoulders, left side of the chest and back. Throughout the preceding few months, the patient had experienced influenza-like symptoms including tiredness, fever and slight weight loss, but without any respiratory symptoms. The patient had been diagnosed with pTa low-grade urothelial cancer of the right

ureter 3 years earlier. She underwent nephroureterectomy, but 5 months later she presented with multiple local pTa low grade recurrences in the urinary bladder and was treated with intravesical chemotherapy using mitomycin. At subsequent control visits, the patient exhibited no signs of recurrence. Her family history included various types of neoplasms: her mother died of jaw cancer, her father died of leukaemia, her sister had Waldenström's disease, and an aunt was treated for medulla spinalis schwannoma. She was a current smoker with 40 pack-years and was being treated for hypothyroidism, but was otherwise in good health, fully functional and still working part-time. On admittance, diagnostic computed tomography of the thorax showed a 25 × 30 mm consolidating mass with peripheral infiltration posteriorly in the upper lobe of the right lung. Positron emission tomography-computed tomography scanning suggested localised malignancy (Fig. 1), and core-needle aspiration biopsy from the lung tumour revealed a malignant tumour of unknown type.

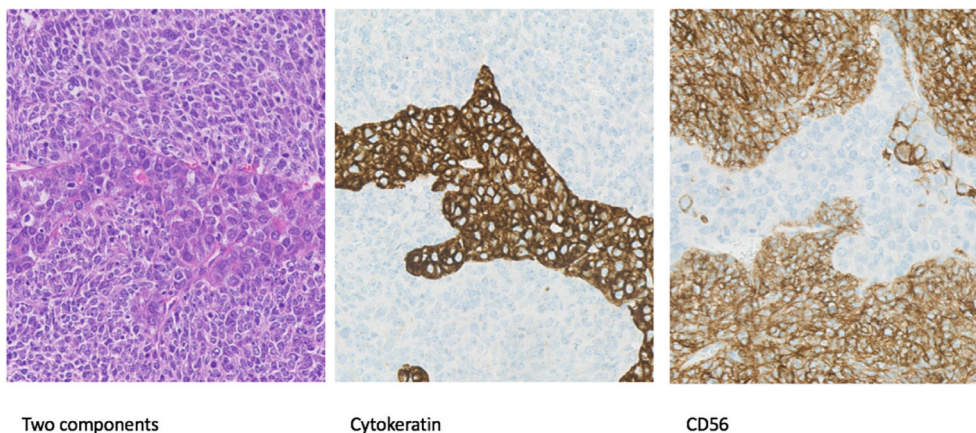
Endosonographic staging revealed no malignancy in the mediastinum, and the lung cancer was classified as cT2aN0M0, stage IB. The patient underwent right upper lobectomy with complete resection and clear margins, revealing classic biphasic pulmonary blastoma (Fig. 2)

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**Fig. 1.** Positron emission tomography-computed tomography scan showing increased fludeoxyglucose uptake within the mass located posteriorly in the right upper lobe.



Two components

Cytokeratin

CD56

**Fig. 2.** Part of the biphasic high-grade tumour with a cytokeratin positive epithelial component and a blastematos component that was positive for CD56 (also known as neural cell adhesion molecule).

and confirming the classification (pTNM CT2aN0M0). However, 8 months later she had developed respiratory symptoms that included wheezing. A new computed tomography scan of the thorax suggested local recurrence, and a right pneumonectomy was performed. The patient died from post operative complications that included pulmonary bleeding 3 days later. Genetic analysis for DICER1 syndrome was performed twice and was negative.

### 3. Discussion

Pulmonary blastoma is an aggressive cancer with a poor prognosis. The symptoms may include a cough, haemoptysis, dyspnoea, fever, chest pain, pleural effusion and pneumo-haemothorax [4–7]. Approximately 40% of cases are asymptomatic and are identified on radiographs performed for other indications [5]. Males are more frequently affected than females and the average age at diagnosis is 40 years [1]. The majority of cases have a history of smoking [1,5]. Recurrence of the disease generally occurs within the first 12 months after a diagnosis or not at all [1]. The 5-year survival rate of patients is only 16% and two-thirds of patients die within 2 years. The prognosis is worse if the tumour is larger than 5 cm at presentation [4,5]. The route of metastasis for pulmonary blastoma has not been determined. However, pulmonary blastoma

metastases have been observed in the brain, bones, liver, pancreas, kidneys, and adrenals, implying a haematogenous route [8]. Because the disease is rare, our current knowledge of optimal treatments is based on case reports and very small cohort studies. For patients who undergo lobectomy rather than pneumonectomy, the prognosis is better [1,3]. When given as a first-line treatment, chemotherapy had a response rate of 26% [1]. Whether adjuvant chemotherapy is beneficial remains uncertain [3]. The medical history of the patient and her relatives suggested the possibility of DICER1 syndrome, a rare genetic disorder that increases the risk of a variety of malignant and benign neoplasms [9,10], but genetic analysis for DICER1 syndrome was negative twice. To the best of our knowledge, this is the first case of pulmonary blastoma previously diagnosed with urothelial cancer and strong family history of malignancy.

### 4. Conclusion

Pulmonary blastoma is a rare type of aggressive lung cancer with a poor prognosis. Our knowledge of this disease is poor because the literature is sparse. Therefore, we believe it is important to describe this patient's unusual case history.

### Financial disclosure and conflicts of interest

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