

# International Journal of Clinical Pathology and Diagnosis

Boulle G, et al. Int J Clin Pathol Diagn: IJCP-118.

DOI: 10.29011/2577-2139.000018

## Case Study

### A Rare and Unexpected Pulmonary Tumor in a Young Woman

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**Citation:** Boulle G, Dupeux M, Charpentier MC, Larousserie F, Régnard JF, et al. (2018) A Rare and Unexpected Pulmonary Tumor in a Young Woman. Int J Clin Pathol Diagn: IJCP-118. DOI: 10.29011/2577-2139.000018

**Received Date:** 13 June, 2018; **Accepted Date:** 25 June, 2018; **Published Date:** 03 July, 2018

#### Clinical Question

A 21-year-old woman presented with an upper left lobe mass, discovered in a context of asthenia, dyspnea, wheezing, flushes and evening fever. Initial CT scan revealed a heterogeneous and predominantly endobronchial mass, which extends into the bronchus of the lingula with latero-aortic and peri-hilar lymphadenopathy. PET-CT scan detected hypermetabolic (SUV<sub>max</sub> = 10) mediastinal lymph nodes (SUV = 2.2). Initial fibroscopy performed found a stenosing endobronchial tumor. Left superior lobectomy and mediastinal lymph node dissection were performed.

#### Q1: What is your diagnosis? HES Slide 1:

- A. Carcinoid tumor.
- B. Pulmonary myxoid sarcoma.
- C. Inflammatory myofibroblastic tumor.
- D. Myxoid liposarcoma.
- E. Pulmonary hamatoma.

#### Q 2: What additional test (s) to order?

- A. Fluorescence in situ hybridization (FISH) for EWSR1.
- B. Fluorescence in situ hybridization (FISH) for ALK.
- C. Immunohistochemistry synaptophysin.
- D. Molecular analysis for lung carcinoma (EGFR, ALK, ROS1).
- E. None.

The correct answers are after the discussion.

#### Discussion

Pulmonary Myxoid Sarcoma (PMS) is a very rare low-grade tumor, approximately fifteen cases described in the literature [1-2].

According to the literature, this disease presents in patients between 28 to 68 years old with no male or female predominance. The clinical presentation is variable, the patients can be asymptomatic or may present with cough, chest pain, or haemoptysis. The macroscopic examination of the left upper lobe of the lung revealed a well-defined proximal tumor measuring 7 x 6 x 4 cm, growing in the upper lobar bronchus, following the bronchial tree with minimal areas of pulmonary infiltration. Microscopically, the tumoral cells appeared monomorphic, fusiform, and dispersed in a myxoid substance. There were very few mitoses, no atypia and necrosis. The lymph node showed follicular hyperplasia without tumor metastasis. Tumoral cells were positive for EMA (80%) and negative for AE1/AE3, actin, desmin and CD34. The FISH analysis performed on formalin-fixed paraffin embedded tissue found a translocation of EWSR1, which was confirmed by the Next-generation sequencing (NextSeq 550 System, illumina) to the fusion transcript EWSR1/ATF1. The common genetic aberration in PMS is t(2;22)(q33;q12) translocation with the EWSR1-CREB1 fusion gene. EWSR1 gene is found in most chromosomal translocations of sarcomas, with nearly 15 types of reported fusion products [3]. EWSR1/ATF1 fusion transcript is rarely seen in this type of tumor. ATF1 gene encodes for a cyclic AMP protein (cAMP) responsive element, which is a constitutively product after the translocation with EWSR1 [4]. EWSR1/ATF1 fusion transcript is not pathognomonic for myxoid sarcoma, since it is also found in other diseases, such as clear cell sarcoma and angiomatoid fibrous histiocytoma [2-5]. The differential diagnosis is mainly pulmonary mesenchymal chondrosarcoma and other myxoid tumors, such as myxoid liposarcoma [6-7]. The immediate post-operative follow-up was uncomplicated, and the patient received no further treatment. At 6 months' follow-up, the patient had no evidence of recurrence by clinical and radiologic evaluation. The risk of recurrence is low; however, there are rare reported cases of metastasis to the lung, brain and kidney.

## Answers

Pulmonary myxoid sarcoma with EWSR1 translocation.

## Take Home Messages

Mesenchymal tumors of the lung are very rare; the most common entity is pulmonary hamartoma, a benign tumor. Pulmonary Myxoid Sarcoma (PMS) is a low-grade tumor, which is often localized and partially endobronchial lesion, which rarely metastasize. The diagnosis of PMS is based on histologic evaluation and confirmed by the presence of EWSR1 translocation.

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