

AB009. Pitfalls in mediastinal masses diagnosis

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Abstract: Mediastinum is a complex anatomic space in which different malignancies can occur, originating from different cell types, such as epithelial thymic, lymphoid, germinal cell and mesenchymal cell. Their diagnosis requires dedicated knowledge and expertise, due to heterogeneous features. Indeed, there is also a frequent overlap between different histologies leading to uncertainty in the diagnostic process. Here, we present an uncommon mediastinal mass challenging in its characterization. A thirty-year-old woman performed thoracic CT scan for dyspnea and persistent cough. Imaging showed a solid mass of 14×11 cm involving the left thorax with mediastinal deviation to the right side. Patient underwent resection en bloc of left endothoracic mass and segmental resection of the left upper lung lobe, and of the 3rd, 4th and 5th ribs. Initial histological examination was suggestive for B3 thymoma/thymic carcinoma due to the positivity for p63, CD99, CD117 and vimentin. Pathological stage pT1b. Patient was then referred to our rare tumor Reference Center. Histological review showed small-medium sized tumor cells. The immunohistochemical panel showed negativity for cytokeratins (CKMNF116, CKAE1/AE3, CK19), as well as PAX8, CD56 NSE and CD117; while CD99 was positive, excluding the diagnosis of thymic/thymoma neoplasms; c-kit and other thymoma related genes resulted negative for mutations at a next generation

sequencing (NGS) analysis. On the other hand, the negativity of different types of cytokeratins and the elevated mitotic index associated with the sporadic positivity for p63 was not specific for an epithelial neoplasm. Morphological pattern and positivity for CD99 led to a diagnosis of undifferentiated sarcoma. A third revision, was performed at the National Sarcoma Center and included the assessment of markers for sarcoma cluster differentiation. Tumor cells were characterized by the positivity for CD99 and NKX2.2 consistent with a diagnosis of Ewing's sarcoma, despite the negativity for ETV4. The evaluation of expert centers was critical to establish a correct diagnosis in this complex case. Taking into account the time lasting from the diagnosis and the aggressiveness of this kind of neoplasm, the patient was candidate for chemo-radiotherapy after a multidisciplinary discussion.

Keywords: Differential diagnosis; sarcoma; thymoma

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Footnote

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