Pulmonary Embryonal Rhabdomyosarcoma: A differential in pulmonary masses

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Introduction

Rhabdomyosarcomas (RMS) are the most common type of soft tissue neoplasm in children. They typically arise from primitive skeletal muscle and are usually observed in either the head and neck or the genitourinary region. We report a rare case of an embryonal rhabdomyosarcoma lung malignancy and a formulated histopathological process to differentiate these lesions from its main differentials. We aim to bring attention to this case in an effort to assist physicians in making the correct diagnosis, should they be presented with a similar case.

Case Description

Here we present a case of a 56-year-old female with a past medical history of COPD and a 30-pack-year smoking history who was referred to a Regional Cancer Center clinic due to newly diagnosed lung cancer. The patient presented with anorexia, weight loss, cough, dyspnea for two months, and a 30-pound weight loss over the last month. She also has a history of multiple COPD exacerbations that required steroids and antibiotics for treatment. CTA of the chest on 7/26/2021 revealed a 14 cm confluent mass extending throughout the mediastinum and the left hilum with associated narrowing of the left-sided pulmonary arteries and bronchi consistent with neoplasia. Several cavitary nodules in the left lung are present, consistent with metastatic disease. CT-guided biopsy on 8/2/2021 revealed a high-grade neoplasm with neuroendocrine features, and frequent mitotic figures, and tumor necrosis. Immunohistochemical staining was positive for synaptophysin, and CD56. Negative stains include cytokeratin TTF-1, AE1/AE3, CAM 5.2, CK 34 beta E12, CK5/6, CK7, CK20, p40, mart 1, SOX10 and CD45. The patient was then diagnosed with pulmonary embryonal rhabdomyosarcoma.



Figure 1. Samples of Pulmonary Embryonal RMS.



Figure 2. Staining methods associated with RMS

Discussion

In this case report the patient was diagnosed with pulmonary embryonal rhabdomyosarcoma, which characteristically has cells that show variable degrees of skeletal muscle differentiation with spindled morphology and differentiated rhabdomyoblasts. Desmin, myoD1, and myogenin are the key immunohistochemical stains that should be utilized to confirm the suspected diagnosis of a RMS. The staining pattern varies between different RMS subtypes, as the pulmonary subtype staining pattern is more focal compared to the others which tend to stain diffusely. CD56 staining can also be used to identify an alveolar RMS, however, is nonspecific. In this patient's case, CD56 was positive but the FISH analysis confirmed embryonal RMS as the final diagnosis. Previous studies have indicated that RMS can metastasize to the lung, thus this strain can prove to be a useful tool in rare cases such as this one, where the etiology of the cancer is unclear but has progressed to the lung.

Conclusion

This unique case report highlights the diagnostic approach and aims to provide a differential diagnosis for a pulmonary embryonal rhabdomyosarcoma as well as an effective workup. Future research into the origin of the pulmonary embryonal rhabdomyosarcoma is indicated to provide comprehensive treatment for the patient and further understand the pathophysiology of the disease discussed.

References

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