AB014. A case of Li-Fraumeni syndrome associated thymoma

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Abstract: Li-Fraumeni syndrome (LFS) is a rare genetic cancer syndrome caused by loss of function mutations of the tumor suppressor gene TP53. We present a case of LFS associated thymoma, a very rare occurrence in the population of LFS patients. A 57-year-old woman presented to the emergency room with swelling in her neck, face and right arm. The patient had no previous personal history of cancer but family history was significant and genetic consultation had revealed a missense mutation of TP53. Chest X-ray showed a large superior mediastinal mass. A CT scan confirmed an aggressive anterior mediastinal mass overlying the sternum and anterior left pleural space. It invaded and obstructed the superior vena cava and left brachiocephalic vein. Enlarged mediastinal lymph nodes and a small pericardial effusion was evident. Pathology on core needle biopsy of the mass revealed a thymoma consisting of large, atypical epithelial cells with admixed background thymocytes and intersecting dense, fibrous bands. The level of atypia was unusual, with cells showing pleomorphic nuclei with a high nuclear to cytoplasmic ratio. Immunohistochemistry was performed for the p53 protein. The malignant epithelial cells stained with strong nuclear positivity, however, there was no staining of background normal cells. The patient was treated with radiation and 6 cycles of Cisplatin-Etoposide chemotherapy with reduction in the size of the primary mass, mediastinal lymph nodes and pericardial effusion. Treatment relieved symptoms but her disease remained unresectable. Patient presentation, genetic consultation and pathological analysis was consistent with LFS associated thymoma. Due to the degree of atypia, the final diagnosis was thymoma with anaplasia. This case demonstrates that thymoma can be a rare consequence of LFS and must be assessed for when appropriate. Prior research shows sporadic p53 mutated thymoma is rare, however the mutation carries a worse prognosis and it is unknown what the implications are for the LPS population. The pathological findings of elevated p53 levels in the context of LFS is intriguing and may be a target of future research.

Keywords: Thymoma; Li-Fraumeni syndrome (LFS); p53

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Footnote

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