Diagnosis of double hit lymphoma

Initial Presentation

The patient went to see his PCP for a chief complaint of “neck lumps”, is found on physical exam to have firm R supravacular mass. Given he was a former smoker (15 pack year smoking history, quit in 1987), he was sent for CT scan of the neck which showed mild bilateral cervical lymph node enlargement, right side greater than left with the largest node measuring 12 mm in the short axis in the lower right neck.

Clinical Course – 1 week later, CT chest

The patient receives CT scan of the chest given neck CT findings. This scan shows lytic and sclerotic lesion in the right posterolateral 7th ribs with an adjacent smoothly marginated pleural/subpleural soft tissue mass measuring 1.7 x 0.6 x 1.8 cm concerning for a malignancy arising from within the pleural/chest wall soft tissues or within the bone.

Excisional biopsy – 5 weeks after initial presentation

Given high suspicion for malignancy, more tissue was needed so an excisional biopsy of the right neck lymph nodes was performed. Pathology revealed diffuse large B-cell lymphoma and flow cytometry reported a clonal B cell population. FISH studies were positive for MYC oncogene (8q24) rearrangement in 37% of cells and BCL2 rearrangement in 54% of cells.

Chemotherapy started – 6 weeks after initial presentation

The patient is started on dose adjusted R-EPOCH chemotherapy with 6 cycles planned (one cycle every 21 days) for Stage IV double hit diffuse large B-cell lymphoma.

PET scan – 4 weeks after initial presentation

A PET/CT is performed and shows abnormal hypermetabolic foci in the posterolateral R lung, mediastinum, left hilar regions, bilateral neck, peri-caval, para-aortic region and throughout the axial and appendicular skeleton highly suspicious for a malignant or metastatic process.

Table from http://www.gotoper.com/publications/ajho/2015/2015apr/clinical

<table>
<thead>
<tr>
<th>Gene</th>
<th>T(14;18) (%)</th>
<th>T(14;18) with E2A-Pbx1 (%)</th>
<th>T(14;18) with MYC, E2A-Pbx1 (%)</th>
<th>T(14;18) with MYC, E2A-Pbx1 and ETP rearrangement (%)</th>
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Summary

Double hit lymphoma is an aggressive form of diffuse large B-cell lymphoma where MYC rearrangement is associated with either BCL2 or BCL6 rearrangement thus patients often present with a rapidly growing mass, B symptoms and extra nodal disease. Here we present a case of bilateral lymphadenopathy that was negative for malignancy via needle biopsy but positive on excisional biopsy. High clinical suspicion for malignancy should lead one to obtain additional tissue for diagnostic work-up.

References