Clinical Case Study

A 78-Year-Old Woman with Brain Metastases

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CASE DESCRIPTION

The patient was a 78-year-old woman, a nonsmoker, with a history of a stage IIIC2 low-grade endometrial carcinoma, endometrioid type, in 2011, which was treated with surgery, vaginal cuff brachytherapy, and adjuvant chemotherapy. Her medical history also included hyperthyroidism and hypertension.

In December 2013 she presented to another hospital with nausea and a posterior occipital headache. Head computed tomography demonstrated a cerebellar mass and she was transferred to our institution for further workup. Imaging also showed an approximately 5-cm left lung perihilar mass suspicious for a primary bronchogenic malignancy, an ill-defined approximately 3-cm right hepatic lobe mass concerning for malignancy, either primary or metastatic, and multiple sclerotic osseous metastases in the thoracolumbar spine. A brain MRI was performed to further characterize the metastatic lesions. There was a 3.0-cm mixed solid and cystic mass in the medial and inferior right cerebellar hemisphere with marked mass effect upon the fourth ventricle. An additional enhancing mass was present in the left occipital lobe (1.7 cm).

The right cerebellar tumor was excised and on pathology showed a moderately differentiated adenocarcinoma, TTF-1 (thyroid transcription factor 1)-positive and PAX-8 negative, consistent with a metastatic adenocarcinoma of pulmonary origin.

PATIENT FOLLOW-UP

The patient's tumor was genotyped on the Ion AmpliSeq™ Cancer Hotspot Panel v2 and was found to harbor an epidermal growth factor receptor exon 20 9-bp insertion (c.2311_2312insGCGTGGACA, p.D770_N771insSVD) and an incidental tumor protein p53 (*TP53*) mutation (c.403C>T, p.R135W); another testing method was not used to confirm the genotype.

Questions to Consider
• What is the role of genotyping lung adenocarcinomas for selection of therapy?
• Describe the difference between primary and secondary resistance.
• Which gene mutations should routinely be tested for in lung adenocarcinoma in a molecular pathology laboratory?

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the April 2015 issue of *Clinical Chemistry*. To view the case and comments online, go to http://www.clinchem.org/content/vol61/issue4 and follow the link to the Clinical Case Study and Commentaries.

Educational Centers

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