

Resolving an uncertain diagnosis of adenocarcinoma favoring gastrointestinal origin

Cancer Care Centers of South Texas, South Texas Medical Center, San Antonio, TX

Initial Diagnosis: Unknown primary adenocarcinoma, gastrointestinal favored

Final Confirmed Diagnosis: Metastatic breast cancer

Initial Staining/Scan History:

- MRI Thoracic and Lumbar spine 9/29/2008 – T10/L3 abnormality
- Mammogram 10/6/08 – benign findings, physical exam of breast normal
- Chest X-ray 10/6/08 – normal
- Bone scan 10/6/08 – abnormality at axial skeleton
- Biopsy source, lower spine 10/21/08
- Magnetic resonance cholangiopancreatography (MRCP) 11/5/08 - normal

Case Summary:

A 45-year-old white female nurse with no family history of breast or ovarian cancers and no smoking history presented at the Cancer Care Centers with lower back pain. An initial evaluation by physical examination was normal. An MRI of the top and lower spine demonstrated lytic lesions suspicious for metastatic disease. Both a mammogram and physical exam of the breasts were normal, as was a chest X-ray. Biopsy of a lesion at the bottom of the patient's spine demonstrated adenocarcinoma favoring gastrointestinal origin. Several weeks later, the oncology team performed an MRCP, which was normal.

The presentation for bone metastasis was unusual for gastrointestinal cancers and the patient's oncologist, Dr. Y. Gia Dice, felt the gastrointestinal diagnosis *"didn't seem right."* Despite the negative physical exam and mammogram, Dr. Dice favored a diagnosis of breast primary. She requested ER/PR/Her2/neu on the bone biopsy and simultaneously ordered a THEROS CancerTYPE ID test to help provide a definitive diagnosis.

"When you have an unknown or uncertain cancer, it's obviously very challenging for both the clinician and the patient to plan," said Dr. Dice. *"I was curious to see how a molecular test could help us get to the bottom of this patient's cancer."* Soon after, the THEROS CancerTYPE ID test results revealed a high confidence prediction for breast cancer. Simultaneously, the receptor stains supported that the patient was estrogen-receptor positive.

With this information in hand, Dr. Dice ordered an MRI, which revealed bilateral breast lesions. *"There is no way insurance would have paid for a bilateral MRI without a mass [evident on a mammogram],"* she said.

"We had a much more positive outcome from this case thanks to the THEROS CancerTYPE ID assay," Dr. Dice said. *"A definitive diagnosis gave this patient a sense of direction and more treatment options. She may be able to live years with the presence of 'bone only' disease. If the primary cancer remains an unknown, neither the patient nor the professional can plan well. Now we know who the enemy is."*



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