

Challenges in molecular testing: overcoming sample limitations in NSCLC



Context

L.G. is a 71-year-old retired accountant who began experiencing symptoms associated with lung cancer, including shortness of breath and persistent cough. Concerned about his symptoms, he sought medical help from his primary care physician, who noted a large pleural effusion and pleural nodularity by imaging. A pleural fluid sample was aspirated, and a cell block was made; however, it was deemed insufficient for molecular studies due to low cellularity. Histology and immunohistochemistry analysis of the cell block were consistent with lung cancer (non-small cell carcinoma). A ctDNA test detected a TP53 variant at low variant allele frequencies, indicating a low tumor fraction and reducing the likelihood of identifying actionable mutations.

Following this, a pleural biopsy was performed, but the sample was found to be Quantity Not Sufficient (QNS) by pathology and was not sent for Next-Generation Sequencing (NGS). The pathologist's report noted, "Pleura with scattered malignant clusters consistent with lung primary (TTF-1 positive tumor cells)" and added, "Most of the tissue consists of pleura, fibrin, and chronic inflammation with minimal tumor, most likely not sufficient for molecular studies." With no actionable results and limited options, the patient was started on chemotherapy, which he did not tolerate well.

Seeking an alternative, the medical oncologist referred the pleural biopsy to Biofidelity, recognizing the lab's expertise in working with small and challenging sample types.

Results

Biofidelity's CLIA-certified lab received the sample on Thursday morning, and the report was finalized by Friday afternoon — a turnaround time of just 34 hours. The results revealed an EGFR L858R mutation, enabling the patient to start targeted therapy with an EGFR inhibitor. Starting EGFR treatment sooner would have provided L.G. with a significantly better quality of life, allowing him to avoid the debilitating side effects of chemotherapy and maintain the ability to engage in daily activities and the things he enjoys.

Conclusion

This case illustrates the significant advantages of using Biofidelity's Aspyre Clinical Test for Lung in managing non-small cell lung cancer (NSCLC). If the Aspyre Clinical Test for Lung had been utilized from the start, L.G. could have initiated targeted therapy much sooner, sparing him from the detrimental effects of chemotherapy and delays in effective treatment. The Aspyre Clinical Test for Lung's ability to detect actionable mutations in small, low tumor cellularity, and low-quality samples underscores its value in clinical settings where sample limitations often pose significant challenges. Biofidelity's Aspyre technology not only overcame these barriers but also demonstrated the power to enhance patient outcomes by improving quality of life through faster and more accurate diagnostic precision.