CORE DIAGNOSTICS[™]

Since T790M mutation confers sensitivity to the third generation EGFR TKI, Osimertinib, the patient has been started on Osimertinib which gained FDA approval in EGFR T790M positive metastatic NSCLC patients based on the results of the AURA3 trial. Recently, Osimertinib also gained FDA approval as first line treatment in EGFR mutation positive (EGFR L858R and EGFR del 19) metastatic NSCLC patients based on the results of the FLAURA trial [PMID: 27959700, 29151359].

CONCLUSION

The above case study highlights the importance of ultra-sensitive techniques such as NGS and ddPCR for the identification of variants which occur at a very low frequency and can still drastically affect treatment decisions. In the said case study, had RT-PCR been the only technique of choice, the EGFR T790M mutation would have been completely missed and the patient might have been subjected to more commonly available first and second-generation EGFR TKIs, which would have been ineffective. The above study also highlights the co-occurrence of EGFR T790M with other activating mutations in the EGFR gene in a treatment naïve patient in whom, the third generation TKI, Osimertinib may offer the best chance for clinical response.