Detection of an EGFR Kinase Domain Duplication in a Lung Adenocarcinoma Patient by Liquid Biospy using Hybrid Capture-based Next Generation Sequencing

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**TECHNOLOGY:**
- Proprietary NEO Assays:
  - DNA extracted from FFPE and blood

**ASSAY CHARACTERISTICS:**
- hybrid capture-based next generation sequencing technology
- detection of point mutations, InDels, copy number alterations and gene fusions
- provides nucleotide resolution for every genomic lesion
- identification of novel fusion partners
- comprehensive testing of 94 genes for FFPE samples (NEOplus assay) and 39 genes for liquid biopsies (NEOliquid assay).

**CASE:**

**Patient History:**
- 72-year old female non-smoker
- diagnosed with adenocarcinoma of the lung in 2015
- metastatic to the kidney, adrenal glands and nervous system
- tested negative for EGFR, KRAS, EML4-ALK and ROS1

**NEO Findings:**
- **NEOplus** identified an EGFR kinase domain duplication (EGFR-KDD) (REF.2) in primary tissue which was also found by NEOliquid in blood (A)
- EGFR KDD was not present in blood after initiation of Afatinib treatment (B)
- NEOliquid detected EGFR amplification, EGFR kinase “triplication” (B) and the EGFR p.T790M resistance mutation (C) upon relapse (liver and brain metastases)

**REFERENCES:**