CASE STUDY
STANFORD UNIVERSITY

Empire Genomics Helps Establish Viability of Performing NGS on Malignant Effusions in Patients with MLA

Background

Lung cancer is the leading cause of cancer-related morbidity. As new therapeutic biomarkers are discovered, targeted therapy for genomic alterations has become clinically relevant. Tumor genotyping and specific molecular treatments have become standard of care. Molecular testing in lung adenocarcinoma has relied on tissue sections from surgical specimens, but more recently, the use of cytology samples has emerged as a desirable alternative, due to high accessibility and minimal invasiveness. The suitability of malignant body fluids for high-throughput sequencing, however, remains unexplored.

Objectives

In this study, the accuracy and utility of performing targeted next-generation sequencing (NGS) on malignant effusions from patients with metastatic lung adenocarcinoma was investigated. NGS is a high throughput technology, which utilizes a small sample input, but the suitability of NGS analysis for tissue-free cytology samples such as malignant body fluids is not well understood.

Approach

In order to investigate the adequacy, reliability, and utility of NGS on liquid biopsies of metastatic lung adenocarcinoma, targeted NGS was performed using custom target panels consisting of many oncogenes. Additional non-NGS molecular tests were performed on the effusion samples, including fluorescence in situ hybridization (FISH) analysis for the oncogene ROS1, using a break-apart FISH probe manufactured by Empire Genomics.

Results

As a result of the analyses carried out, including FISH analysis using Empire Genomics' break-apart probe, it can be concluded that targeted NGS can be performed on malignant effusions from patients with metastatic lung adenocarcinoma, given adequate amounts of DNA input and tumor cellularity. This comprehensive genomic profiling can help to establish mutational status at diagnosis, and inform response to treatment. The results of this study are clinically important, as malignant effusions are accessible via minimally invasive techniques, a preferable alternative to surgical procedures.

Comprehensive Genomic Profiling of Malignant Effusions in Patients with Metastatic Lung Adenocarcinoma

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Lead Organization

Stanford University

Diseases

- Metastatic Lung Adenocarcinoma

Biomarkers Mentioned

- EGFR
- ALK
- ROS1

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