ABSTRACT

Next-generation sequencing (NGS) technology enables rapid analysis and tumour of multiple genes for clinically actionable somatic variants. A liquid biopsy, based on circulating cell-free DNA (cfDNA), can capture the entire heterogeneity of the disease, and offer what tissue biopsies can’t, the opportunity to take serial samples in order to monitor tumour genomic evolution in real time. We have recently developed a proprietary cfDNA enrichment process that requires only droplet volumes of blood. Here, we presented clinical validation of the blood-drop liquid biopsy NGS assay interrogating 2656 mutation hotspots in 50 cancer-associated genes using the AmpliSeq cancer panel and Ion Torrent Proton sequencer.

OBJECTIVES

The liquid biopsy industry is utilizing an extraction method flawed for clinical application. Clinical validation of Circulogene’s blood-drop liquid biopsy NGS will provide a first-in-class offering – matching each patient’s cancer mutations with specifically targeted therapy and follow-up.

METHODS

Blood was collected in EDTA-containing tubes, centrifuged, and circulating cfDNA was recovered from 20 ul of plasma by Circulogene’s proprietary cfDNA enrichment technology. Targeted sequencing was performed using Ion AmpliSeq Cancer Hotspot Panel v2 on Ion Proton with 1 ng cfDNA input. Sequencing data were analyzed by Variant Caller 4.0 and GenePool software (Station X).

RESULTS

1. NGS Quality: Controls Used in This Validation (Mean Coverage ~500,000)

2. Test Sensitivity: Down to 0.5%.

3. Test Reproducibility: and Repeatability

4. Test Specificity

RESULTS (Continued)

5. Test Accuracy: Inter-Lab Correlation

CONCLUSIONS

Our new-generation liquid biopsy NGS assay, with the capability to multiplex and simultaneously sequence multiple patient samples using finger-stick blood, is a robust and sensitive method for mutation analysis of clinical significance, further expedite treatment decision-making and identify targeted therapies for eligible patients in a time- and cost-efficient manner.

- CLIA-certified
- Minimal input with maximal output
- NGS-grade cfDNA quality
- Most time- and cost-efficient

REFERENCES