Abstract

We are in the early years of one of the most potentially transformative shifts in diagnostics: using blood or urine to replace invasive procedures. This first began commercially with non-invasive prenatal testing (NIPT) and now we are having laboratories and companies introducing non-invasive tests for oncology. Liquid biopsies are a diagnostic test designed to replicate invasive biopsies or imaging procedures, although only requiring a blood draw or urine sample – a non-invasive biopsy. Liquid biopsies have many other advantages over traditional care including, for prenatal testing, reducing risky amniocentesis. For oncology, liquid biopsy could deliver earlier cancer diagnosis, identifying previously unknown tumor heterogeneity, enabling real-time treatment monitoring, and detecting recurrence before significant tumor formation or metastasis. Studies revealed that liquid biopsies could revolutionize cancer treatment and accurately identify targeted treatment therapies and then monitoring tumor response in real-time.

Recently we established as part of routine clinical care, screening both hematologic and solid tumors for a wide spectrum of mutations using two next-generation sequencing (NGS) panels: a novel and rapid library prep was designed for use with both formalin-fixed paraffin-embedded tissue and plasma samples that targets 131 genes commonly mutated in cancer. Our workflow includes the review of the tissue biopsy to ensure there is adequate amount of tumor for the assay followed by customized DNA extraction for both tissue and plasma specimen. The resulting data is analyzed through an in-house bioinformatics pipeline and the variants are reviewed and interpreted for pathogenicity. Here we provide a snapshot of the utility of the panel workflow to provide insight into how a well-designed NGS workflow can contribute to optimizing clinical outcomes.