

## **Five Years from Implementing Next-Generation-Sequencing Based Assays for Molecular Profiling of Tumors in the Clinic – What Did We Learn?**

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### **Abstract**

Next-Generation sequencing technology allows rapid analysis of multiple genes for the detection of germline mutations and a more complete biological characterization of a tumor at the molecular level, as well as the detection of clinically actionable somatic variants. It has not been long since the NGS technology has been introduced into clinical setting, and since then much experience has been gained with applying NGS in the clinic. In this short review, I will briefly review the currently most commonly used sequencing platforms in the clinic; Illumina's and Ion Torrent PGM sequencing technologies and share our experience with some of the considerations that need to be taken into account before and during the implementation of NGS in clinical setting for molecular profiling of tumors, such as the decision making on gene content for gene panels, types of mutations to be detected, the validation studies required for the assays, variant classification and reporting, factors that affect testing turnaround time, factors that affect successful outcome of testing, the diagnostic yield, the challenges associated with germline mutations for cancer-predisposition genes, issues with reimbursement, and finally review the testings that are ready to be offered at the University of Arizona Genetic Core for Clinical Services Laboratory (UAGC- CS).