Oregon Health and Science University Gene Trails Comprehensive Panel

This panel uses amplification-based next-generation sequencing with gene-specific primers (GSPs) and unique molecular indexes (UMIs). It identifies the two classes of genomic alterations (base substitutions, small insertions and deletions) then sequences DNA of selected exons of CARD11 and the entire coding region of the rest 75 genes to an average depth of approximately 2200x. The sensitivity of this panel is 100% for base substitutions at ≥2% Variant Allele Frequency (VAF) and Small insertions/Deletions (1-84 base pairs) at ≥5 VAF. This panel has 100% Positive Predictive Value (PPV) for base substitutions and small insertion/deletions. The concordance between both inter-batch and intra-batch replicates is 100% while the co-efficiency of both variation inter-batch and intra-batch is 5.4 and 5.9%, respectively.

Ohio State University Hematologic Mutation Neoplasm Panel – Myeloid Indication (50-gene set)

This panel uses amplification-based next-generation sequencing with gene-specific primers (GSPs). It identified two classes of genomic alterations (base substitutions, small insertions and deletions). This panel sequences DNA for the full coding sequence for the following genes: ABL1, ANKRD26, ASXL1, BCOR, BCORL1, BRAF, CEBPA (partial), DDX41,DNMT3A, ELANE, ETV6, EZH2, GATA1, GATA2, JAK2 (including exon 12), KIT, KRAS, MAP2K1, KMT2A (including some MLL-PTD), NOTCH1, NRAS, PHF6, PTEN, RAD21, RUNX1, SF3B1, SH2B3, SMC1A, SMC3, STAG2, TERC, TERT, TET2, TP53, WT1 and ZRSR2 and mutation hotspots in CALR, CBL, CSF3R, FLT3 (TKD and smaller ITDs only), IDH1, IDH2, MPL, MYD88, NPM1, PIK3CA, PTPN11, SETBP1, SRSF2, and U2AF1. Promoter regions of ANKRD26 and TERT also interrogated. Intronic/splice variants are analyzed routinely -8 position into intron. This panel sequences to an average depth of 2100x.

Sensitivity (compared to lab developed/validated bait-probe NGS panel or orthogonal methods such as ddPCR) is 100% for base substitutions at ≥4% VAF and Small insertions/Deletions (1-40 base pairs for ITD) at ≥5% VAF (excluding CEBPA and FLT3 ITD >40 base pairs). The sensitivity is 99% for ≥2% Variant Allele Frequency VAF. The PPV is >99% for base substitutions and 100% for small insertions/deletions. The concordance between both inter-batch and intra-batch replicates is 100% while the co-efficiency of both variation inter-batch and intra-batch is 2.9% and 3.8%, respectively.

Beat AML® Clinical Trial Assay

The Beat AML® Clinical Trial Assay panel sequences the complete coding region of 406 genes, as well as selected introns of 31 genes involved in rearrangements. The platform is capable of detecting all classes of genomic alterations, including base substitutions, insertion and deletions (indels), and copy number alterations (CNAs), though only short variants were reported for Beat AML® treatment decisions.

Sensitivity is >99% for base substitutions at ≥5% mean allele frequency (MAF) and 97% for small insertions/Deletions (1-40 base pairs for ITD) at ≥10% MAF. The PPV is >99% for both base substitutions and small insertions/deletions (Supplementary Table 1).