Template: Performance Characteristics from Mixing Studies to Detect Copy Number Variations and Translocations

As referenced in Jennings et al, 2017

Tumor samples or cell lines	Variant type	Variant allele frequency (VAF)	Known variants in the region[#]	Known variants detected (TP)	Known variants not detected (FN)	FP	PPA	PPV
Ploidy <4, CN ≥8	Copy number alteration-Amplification	≥30% tumor nuclei						
Ploidy <4, CN ≥8	Copy number alteration-Amplifications	20-30% tumor nuclei						
Ploidy <4, CN =0	Copy number alteration-Homozygous Deletions	≥30% tumor nuclei						
Ploidy <4, CN =0	Copy number alteration-Homozygous Deletions	20-30% tumor nuclei						
Gene translocation	Structural Variants	≥20% tumor nuclei						
Gene translocation	Structural Variants	≥10% tumor nuclei						

Legend: This represents a sample method or template for documenting and describing the mixing studies that are performed in the Optimization & Familiarization phase. [#]These are defined as identified in regions meeting minimum QC requirements.

PPV=TP/(TP+FP)*100
PPA=TP/(TP+FN)*100

Abbreviations: CN, copy number; QC, quality control; TP, true positive; FP, false positive; FN, false negative; PPA, positive percent agreement; PPV, positive predictive value.