**Template: Performance Characteristics from Mixing Studies to Detect Different Types of Variants**

*As referenced in Jennings et al, 2017*

| Cell line mixture* | Variant type | Allele burden | Known variants in the region# | Known variants detected (TP) | Known variants not detected (FN) | FP | PPA | PPV |
|---|---|---|---|---|---|---|---|---|
| | SNV | 5% | | | | | | |
| | SNV | 10% | | | | | | |
| | indel (<5 bp) | 5% | | | | | | |
| | indel (<5 bp) | 10% | | | | | | |
| | indel (5-40 bp) | 5% | | | | | | |
| | indel (5-40 bp) | 10% | | | | | | |

**Legend:** This represents a sample method or template for documenting and describing the mixing studies that are performed in the Optimization & Familiarization phase. *For example, Coriell cell lines NA12878 and NA12877 could be used in a mixing study as per the example below. #These are defined as identified in regions meeting minimum QC requirements.

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

**Abbreviations:** SNV, single nucleotide variant; indel, insertion/deletion; QC, quality control; TP, true positive; FP, false positive; FN, false negative; PPA, positive percent agreement; PPV, positive predictive value.

**Example template:**

| Cell line mixture[*] | Variant type | Allele burden | Known variants in the region# | Known variants detected (TP)# | Known variants not detected (FN) | FP | PPA | PPV |
|---|---|---|---|---|---|---|---|---|
| 90% NA12878/ 10% NA12877 | SNV | 5% | | | | | | |
| 80% NA12878/ 20% NA12877 | SNV | 10% | | | | | | |
| 90% NA12878/ 10% NA12877 | indel (<5 bp) | 5% | | | | | | |
| 80% NA12878/ 20% NA12877 | indel (<5 bp) | 10% | | | | | | |
| 90% NA12878/ 10% NA12877 | indel (5-20 bp) | 5% | | | | | | |
| 80% NA12878/ 20% NA12877 | indel (5-20 bp) | 10% | | | | | | |