Template: Method and Template for Documenting and Describing Studies Performed in the Validation Phase

As referenced in Jennings et al, 2017

Sample	Targeted region with known sequence [bp]	Targeted region meeting minimum quality criteria [%]	Known variants in the region#	Known variants detected (TP)	Known variants not detected (FN)	FP	PPA	PPV
1								
2								
3								
…								
60								
					Mean			
					SE of the mean			
					95% lower CI			
					95% lower TI			

Legend: During validation, for each sample the true positives (TP), false positives (FP) and false negatives (FN) should be determined for the targeted regions meeting the minimum quality requirements. The positive percent agreement and positive predictive value can be determined as indicated in Figure 4 of Jennings *et al*, 2017. The mean, standard error of the mean, 95% lower confidence interval and 95% tolerance interval should be calculated by variant type. #These are defined as identified in regions meeting minimum QC requirements.

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

Abbreviations: QC, quality control; TP, true positive; FP, false positive; FN, false negative; PPA, positive percent agreement; PPV, positive predictive value; SE, standard error; CI, confidence interval; TI, tolerance interval.