Table S10. Comparison of three benchmarking studies that used GiaB gold standard variant dataset NA12878

	Present study	Hwang et al., 2015	Highnam et al, 2015	Cornish and Guda, 2015
GiaB Version	GiaB V3.3.2	GiaB high confident integrated pedigree calls V0.2	GiaB V2.18	GiaB V2.15
Genome Version	GRCh37&38	GRCh37	GRCh37	GRCh37
Sequencing Platform	HiSeq2500	HiSeq2000	HiSeq2000	HiSeq2000
Data sets used	NA12878, NA24385, NA24631 and Simulated Reads(WES)	12 Datasets NA12878(WES, WGS) including IonTorrent	NA12878(WES) and Simulated Reads	NA12878(WES)
Aligners	BWA-MEM 0.7.16 Bowtie2 2.2.6 Novoalign 3.08.02 SOAP 2.21 MOSAIK 2.2.3	Bowtie2-2.2.25 BWA-MEM-0.7.10 Novoalign-3.02.12 Tmap (for Ion Proton)	Bowtie2-2.0.0-beta5 BWA-0.7.5a BWA-MEM-0.7.5a Novoalign-3.00.04	Bowtie2-2.1.0 BWA sampe-0.7.5a BWA-MEM-0.7.5a CUSHAW3-3.0.3 MOSAIK-2.2.3 Novoalign-3.02.07
Variant callers	GATK HaplotypeCaller 4 FreeBayes 1.1.0 SAMtools mpileup 1.7 DeepVariant r0.4	FreeBayes-0.9.18 GATK-HaplotypeCaller3.3.0 SAMtools mpileup-1.1 TVC-4.2.3 (for Ion Proton)	GATK-HaplotypeCaller3.1-1 GATK-UnifiedGenotyper3.1-1 SAMtools mpileup v0.1.18 Isaac v01.13.06.20	FreeBayes v9.9.2-19- g011561f GATK-HaplotypeCaller2.7-2 GATK-UnifiedGenotyper2.7-2 SAMtools mpileup-0.1.19 SNPSVM-0.01
Benchmark Method	F-Score	Area under Precision-recall curve	Positive predictive value	Positive predictive value, sensitivity separately
Best Performing Pipeline	BWA/Novoalign_DeepVariant (SNVs and InDels were analyzed separately)	SNVs: BWA_SAMtools InDels: BWA_GATK HaplotypeCaller (SNVs and InDels were analyzed separately)	Novoalign_GATK HaplotypeCaller (SNV and InDel were combined for this analysis)	Novoalign_GATK HaplotypeCaller (Only SNV was analyzed)