

S2 Table. Different parameters for variant calling in Ion Torrent Variant Caller for Ion PGM and Ion PGM Dx

Parameter (Torrent Variant Caller Settings)		Ion PGM	Ion PGM Dx	Recommended values
snp_min_allele_freq	snp_Minimum allele frequency	0.15	0.1	0.01-0.2
snp_min_variant_score	snp_Minimum quality	10	10	≥ 10
snp_strand_bias	snp_Maximum strand bias	0.9	0.95	1
hotspot_min_allele_freq	hotspot_Minimum allele frequency	0.15	0.1	NA
hotspot_min_variant_score	hotspot_Minimum quality	10	10	≥ 10
hotspot_strand_bias	hotspot_Maximum strand bias	0.9	0.95	1
indel_min_allele_freq	indel_min_allele_freq	0.15	0.1	0.05-0.2
indel_min_variant_score	indel_Minimum quality	10	10	≥ 10
indel_strand_bias	indel_Maximum strand bias	0.9	0.95	1
heavy_tailed	T-distribution tails	3	default	NA
outlier_probability	prior probability that a read comes from some other distribution	0.01	default	0.005-0.01
do_snp_realignment	realign reads in the vicinity of SNP candidates	0	0	0: germline
prediction_precision	number of pseudo-data-points suggesting our predictions match the measurements without bias	1	default	1
downsample_to_coverage	reduce coverage in over-sampled locations to this value	400	400	400: germline
suppress_recalibration	ignore the base recalibration	1	default	1: ignore base calibration

	values from pipeline in TVC			values in TVC
FreeBayes module setting				
allow_indels	Enable indels	1	1	1
allow_mnps	Enable MNPs	1	1	1
allow_SNPS	Enable SNPs	1	1	1
gen_min_alt_allele_freq	Early-on filter for allele frequency	0.15	default	0.02-0.15
gen_min_coverage	Early-on filter for minimum coverage	6	default	6
gen_min_indel_alt_allele_freq	An early-on filter for allele frequency for indel callings	0.15	default	0.02-0.15
min_base_qv		4	default	NA
min_mapping_qv	Minimum mapping quality	4	default	4
read_max_mismatch_fraction	Maximum fraction of mismatches allowed in the length of read	1	default	1
read_mismatch_limit	Number of mismatches allowed	10	default	10

NA, not available.