Input/output files

Type	Numbering	
Input	1	FASTQ files
Reference	2	hs37d5
Capture kit	3	20130108.exome.targets.bed
fastQC	4	Raw QC Output directory
Fastp	5	Fastp HTML report
Fastp	6	Fastp Json report
fastQC	7	Pre-processed QC Output directory
fastp	8	Pre-processed paired reads
fastp	9	Pre-processed unpaired R1 reads
fastp	10	Pre-processed unpaired R2 reads
BAM files	11	Sorted aligned BAM files
BAM files	12	Filtered sorted de-duplicated BAM files

Workflows

Step	Program + version	Parameters	files?	docker?
Quality Control (raw data)	FastQC v0.11.8		[1] > [4]	Y
Raw Data trimming	Fastp v0.20.0	-h [5] -j [6] ‘--’cut_right;cut_right_window_size:5 ;cut_right_mean_quality:24;trim_tail 1:1;length_required:70	[1] > [5,6,8, 9,10]	Y
Quality Control (pre-processe d)	FastQC v0.11.8		[8] > [7]	Y
Alignment to reference	Bwa v0.7.16a Samtools v1.5	-R (Bwa mem) -bS (Samtools view) (samtools sort) (samtools merge) (samtools index)	[2,8,9, 10] > [11]	Y
Mark duplicates	Picard v2.10.10	MarkDuplicates	[11] > [int1]	Y
BAM filtering	Samtools v1.5	view -q 10 -F 4 (samtools index)	[Int1] > [12]	

CNV calling	Manta v1.6.0	-exome (PHRED>20)*	[12] > [int5]	N (https://github.com/Illumina/manta)
CNV calling	GRIDSS v2.9.3	(QC>999)*	[12] > int6	N (https://github.com/PapenfussLab/GRIDSS)
CNV calling	ExomeDepth v1.1.12	(BF>5.5)*	[12] > int7	N (https://github.com/vplagnol/ExomeDepth)
CNV calling	CODEX v2	(lenght<200, lratio>40)*	[12] > int8	N (https://github.com/yuchaojiang/CODEX2)
Final filtering	Bedtools v2.26.0	merge (DEL) merge (DUP) subtract (DEL, DUP) subtract (DUP, DEL)	[int5,int6,int7,int8] > finalOutput	Y

*) included in filtered version