

0.1. Mapping (hg38) (lane)
bwa-mem, alt aware

0.2. Select mapped reads (lane)

autosomes + X chromosome

A1. Duplicate
marking (lane)

A2. Indel
realignment (lane)

A4. BQSR
(lane)

A3. HaplotypeCaller
(sample)

A5. HaplotypeCaller
(sample)

VCF

VCF

A6. "triple mask"
BQSR (lane)

A7. Merge and sort
(sample)

A8. Duplicate
marking (sample)

A9. Indel
realignment (sample)

autosomes

A10. HaplotypeCaller
(sample)
GVCF mode, ploidy=2

A11. CombineGVCFs
(batches)

A12. GenotypeGVCFs
(all samples)

A13. SNP VQSR
(all samples)

A14. Indel VQSR
(all samples)

A15. Callset
refinement

- relatedness filter
- mark ambiguous sites
- site missingness filter
- HWE filter

X chromosome

X1. HaplotypeCaller
(sample)
GVCF mode, ploidy=1 or 2

X2. CombineGVCFs
(batches)

X3. GenotypeGVCFs
(all samples)

X4. SNP VQSR
(all samples)

X5. Indel VQSR
(all samples)

X6. Callset
refinement

- relatedness filter
- mark ambiguous sites
- site missingness filter