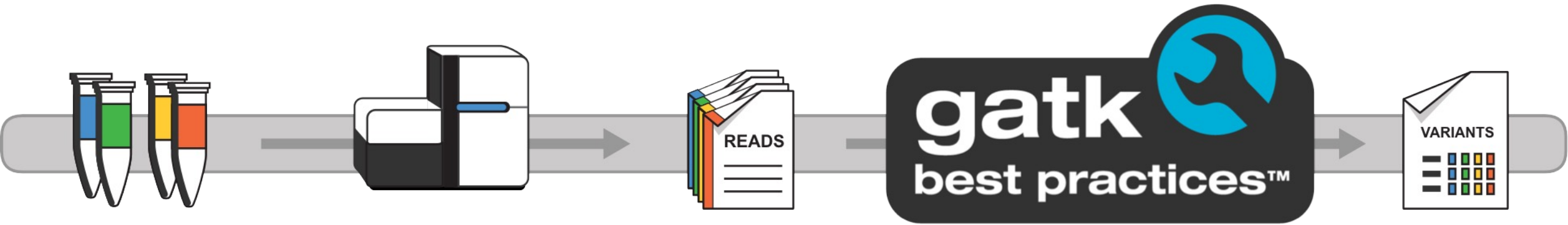


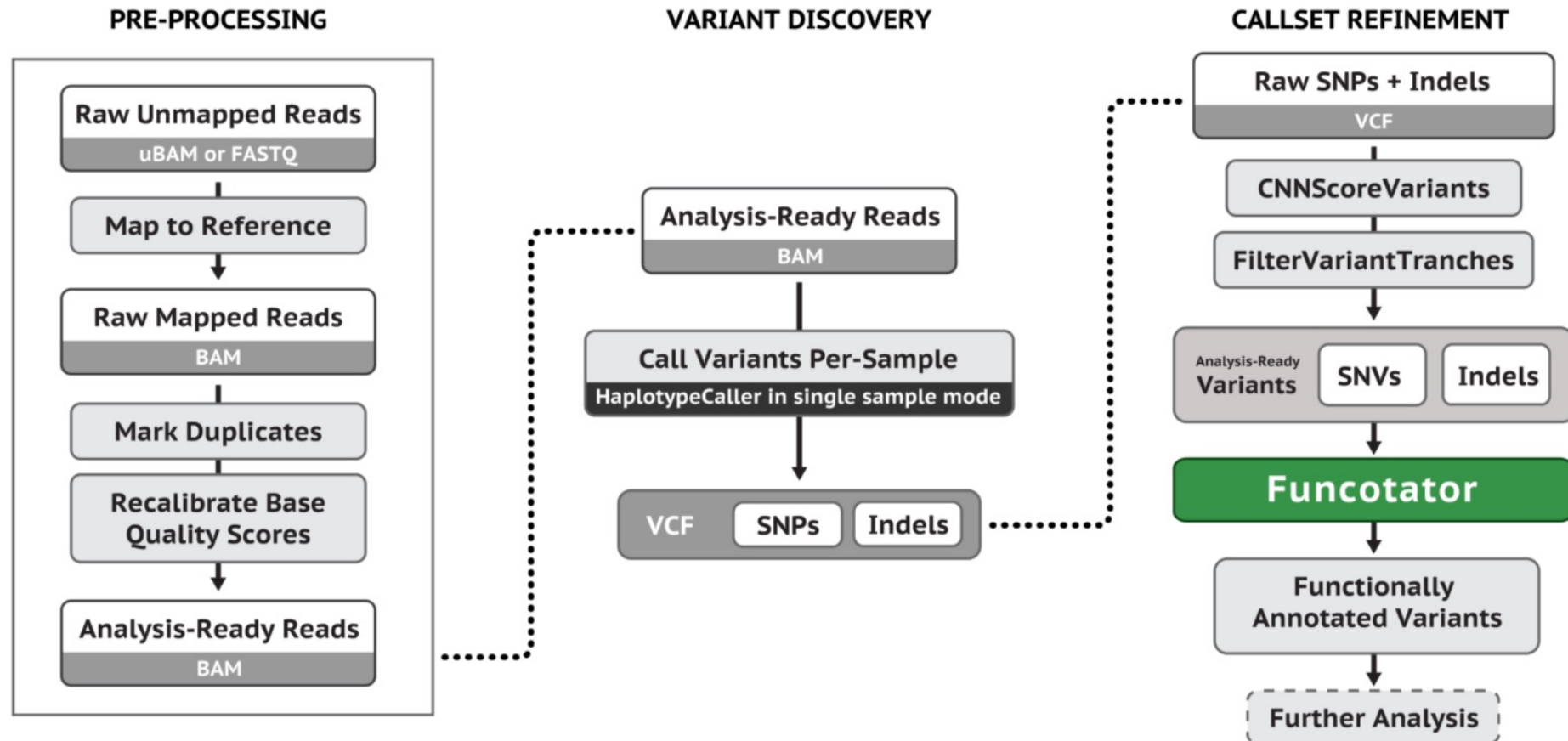
Variant Calling with GATK: a tutorial



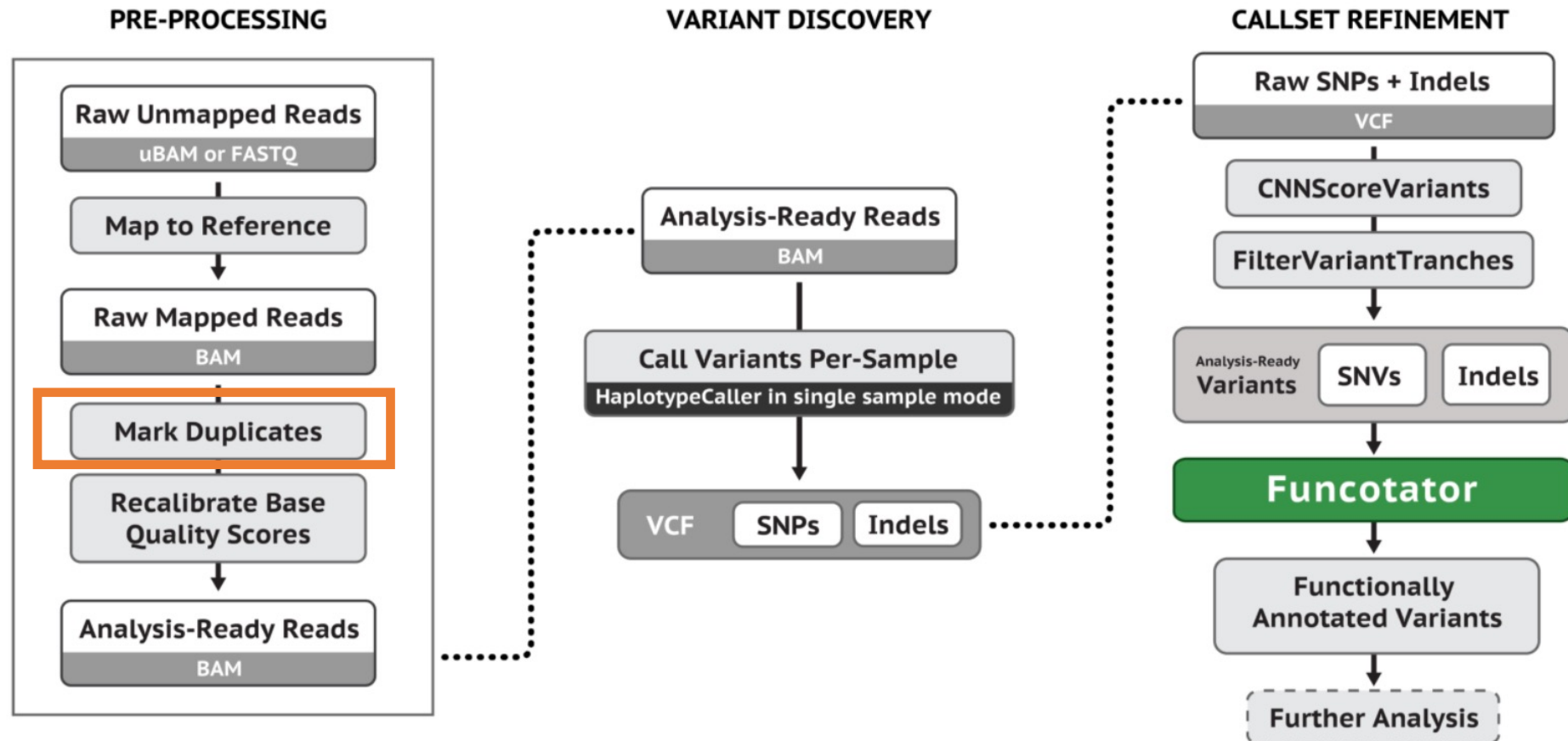
BIOL435/535: Bioinformatics

April 12th, 2021

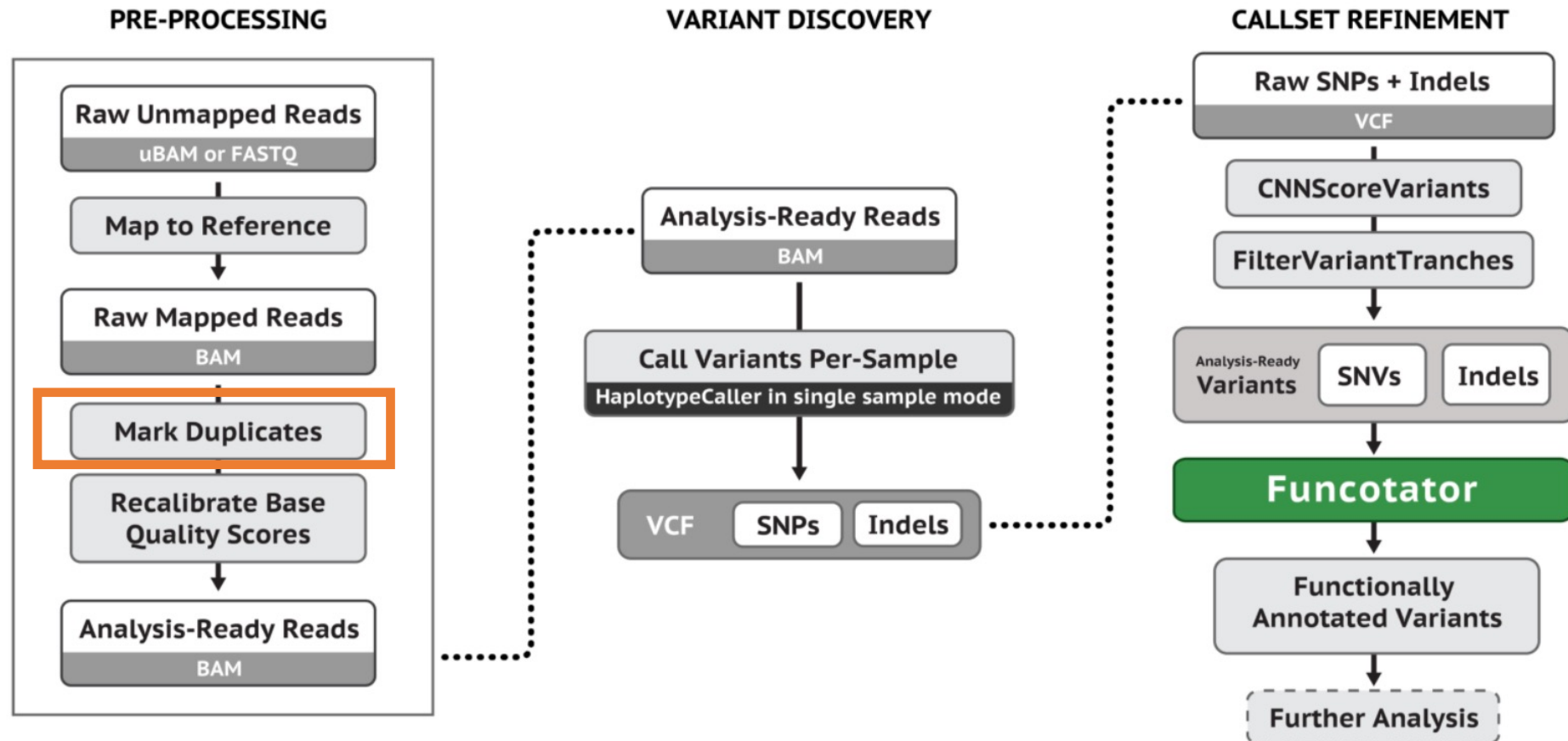
GATK Pipeline



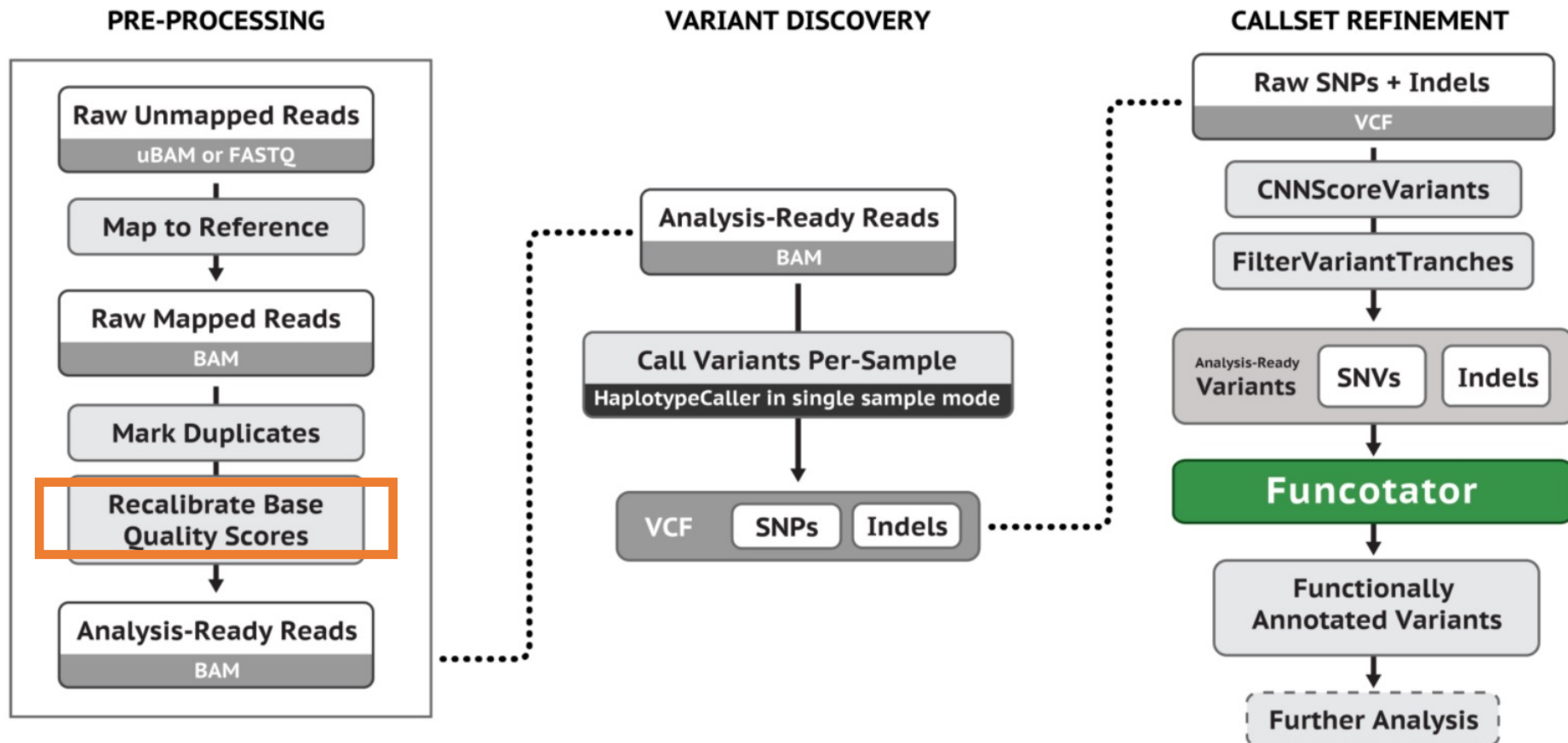
GATK Pipeline: MarkDuplicates



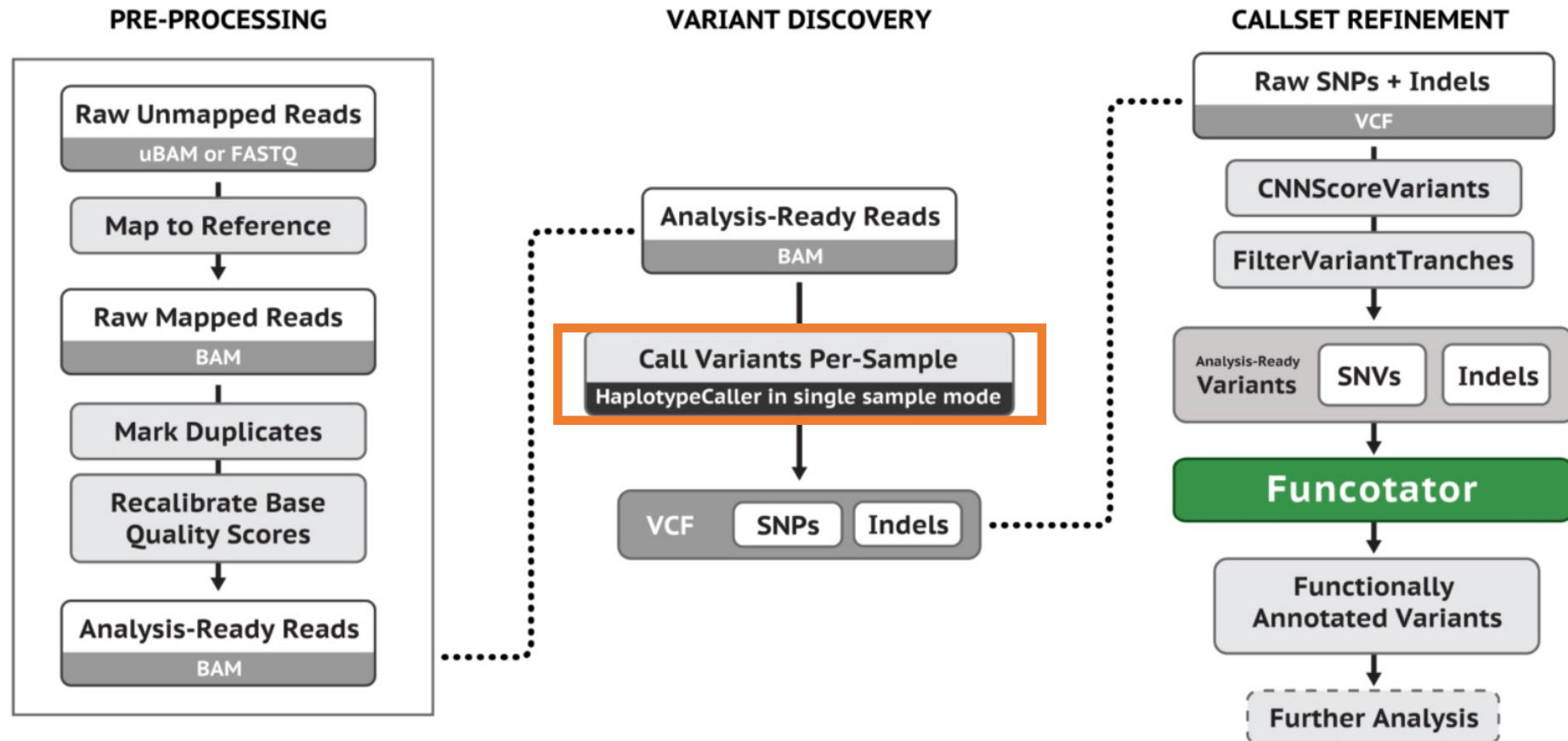
GATK Pipeline: MarkDuplicates



GATK Pipeline: Recalibrate Base Quality Scores (only use with high-quality polymorphisms database)



GATK Pipeline: HaplotypeCaller



VCF Files

1. CHROM – Chromosome
2. POS – Position
3. ID
4. REF – Reference allele
5. ALT – Variant allele
6. QUAL – Variant confidence
7. FILTER
8. INFO – DP, DP4, GT:0/1, GL

Samtools/bcftools variant calling

```
bcftools mpileup
```

```
\-f competitive_mtDNA.fasta
```

```
\-O b
```

```
\ERR2039570.competitive_mtDNA.duplicatesRemoved.bam > ERR2039570.competitive_mtDNA.bcftools.bcf.gz
```

```
bcftools call
```

```
\-m
```

```
\--ploidy 1
```

```
\-v
```

```
\-O v
```

```
\ERR2039570.competitive_mtDNA.bcftools.bcf.gz > ERR2039570.competitive_mtDNA.bcftools.variants.vcf
```