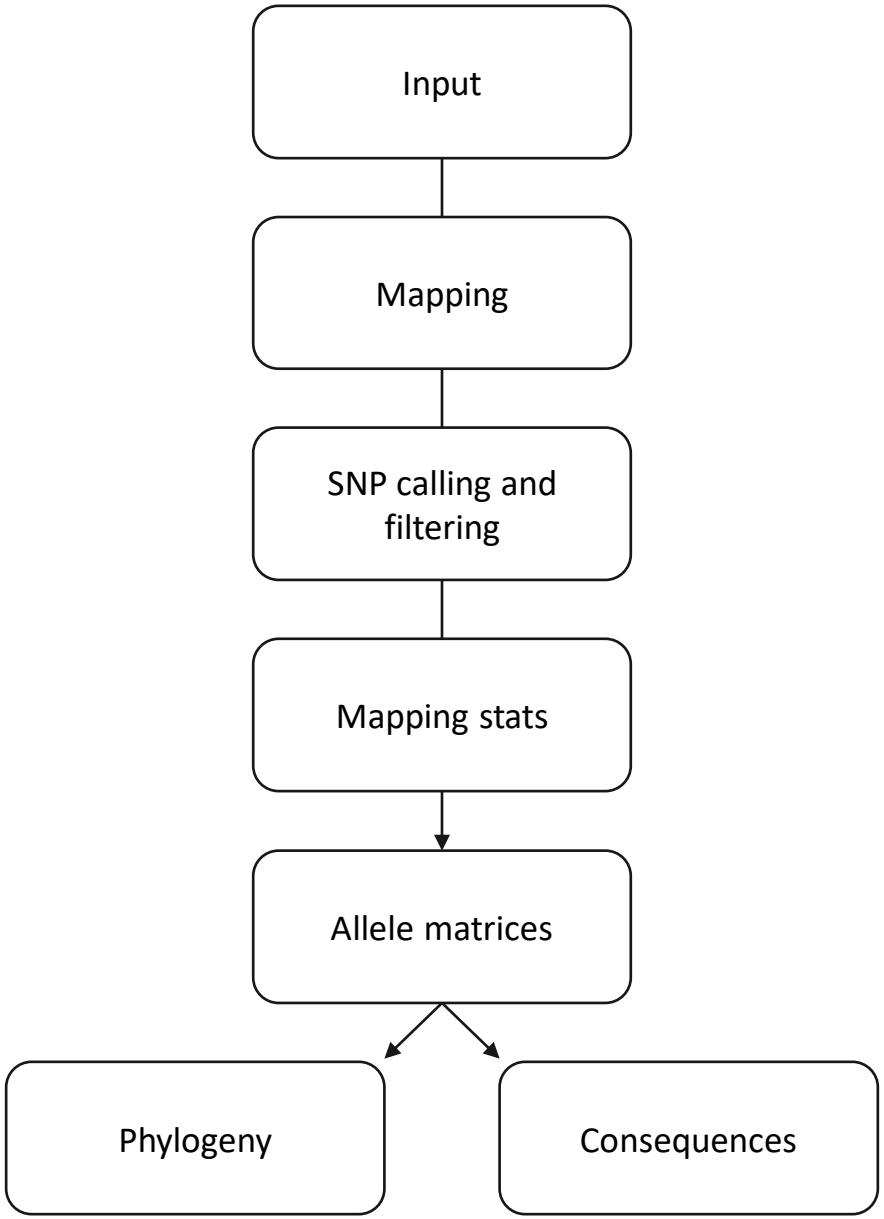
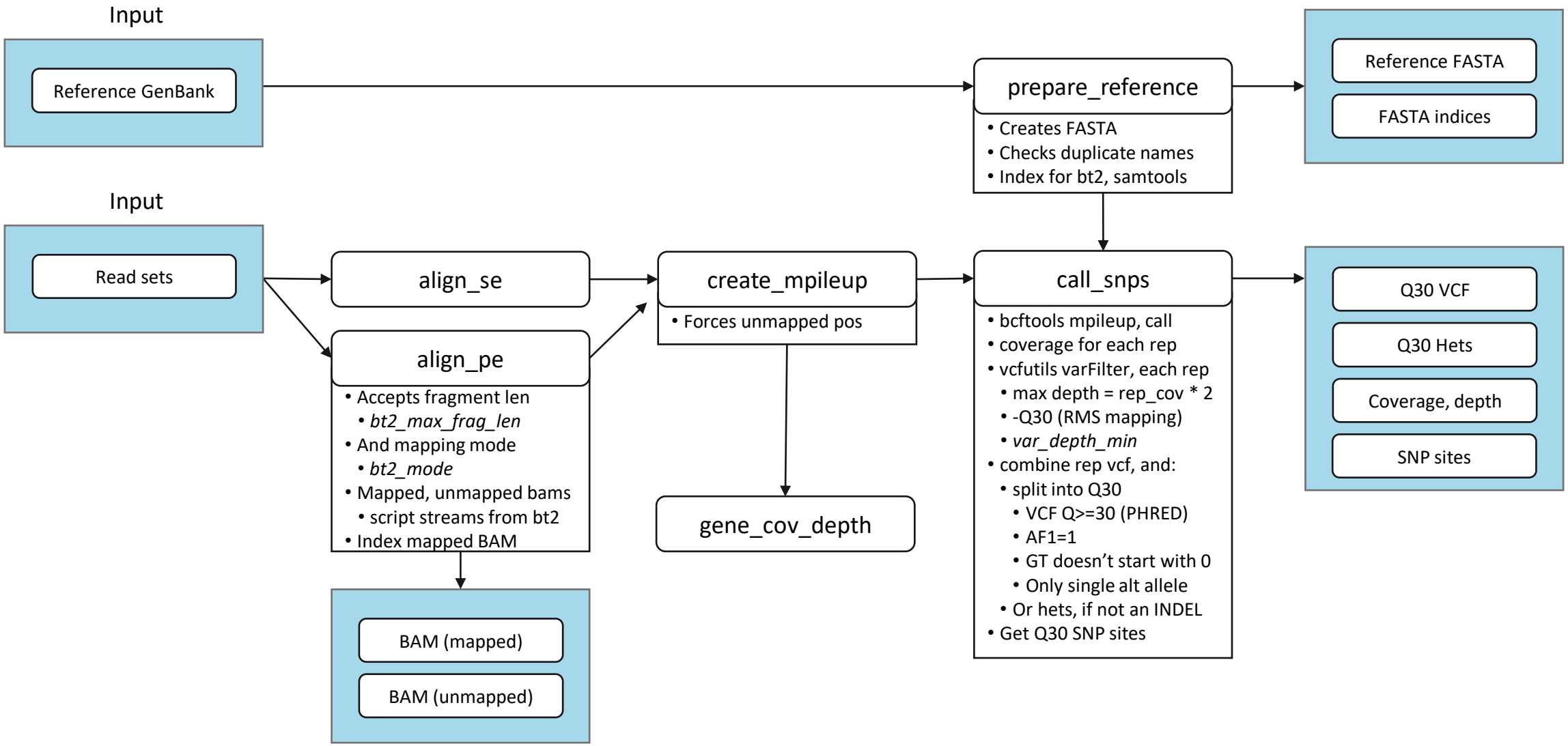
Implementation

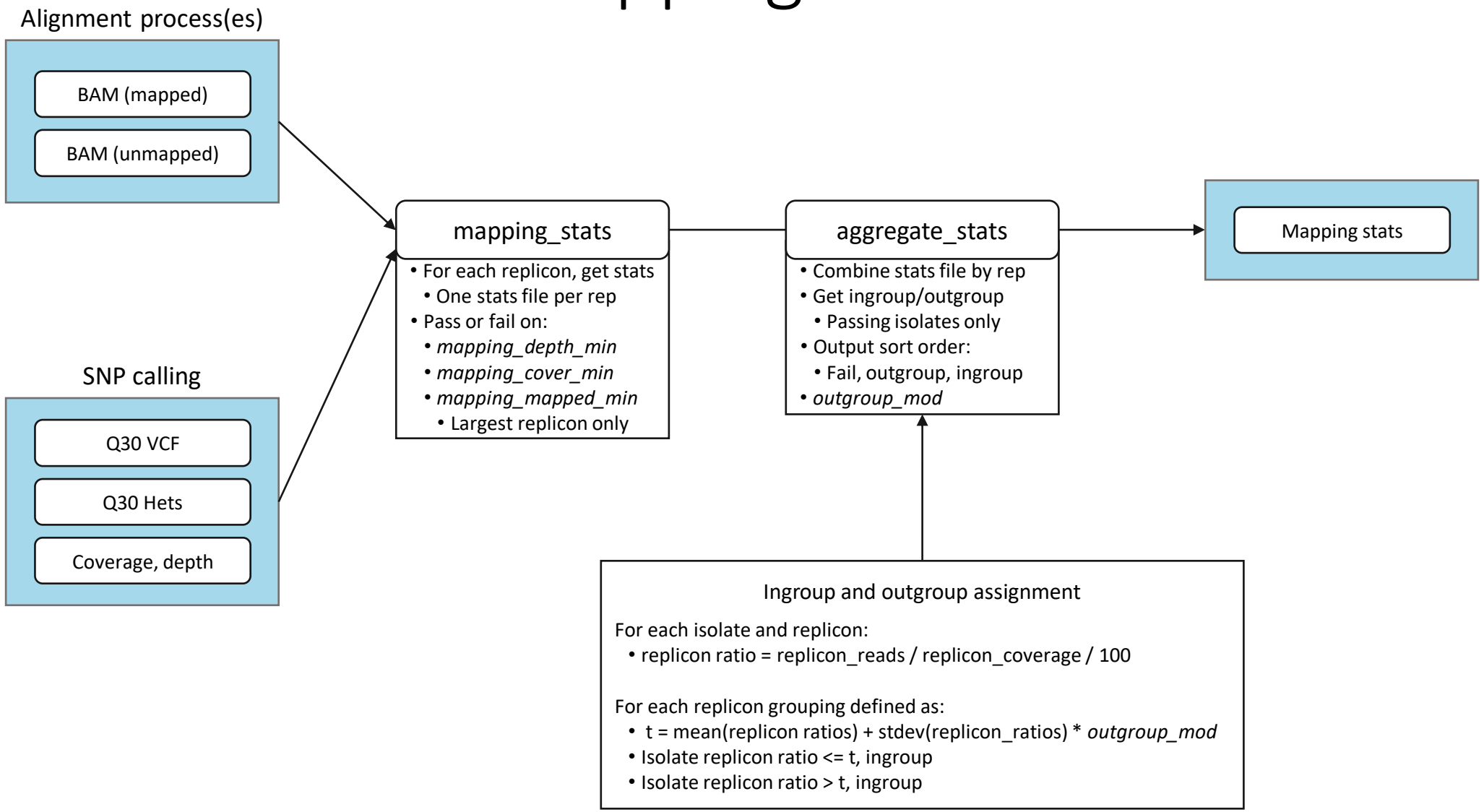
High level overview

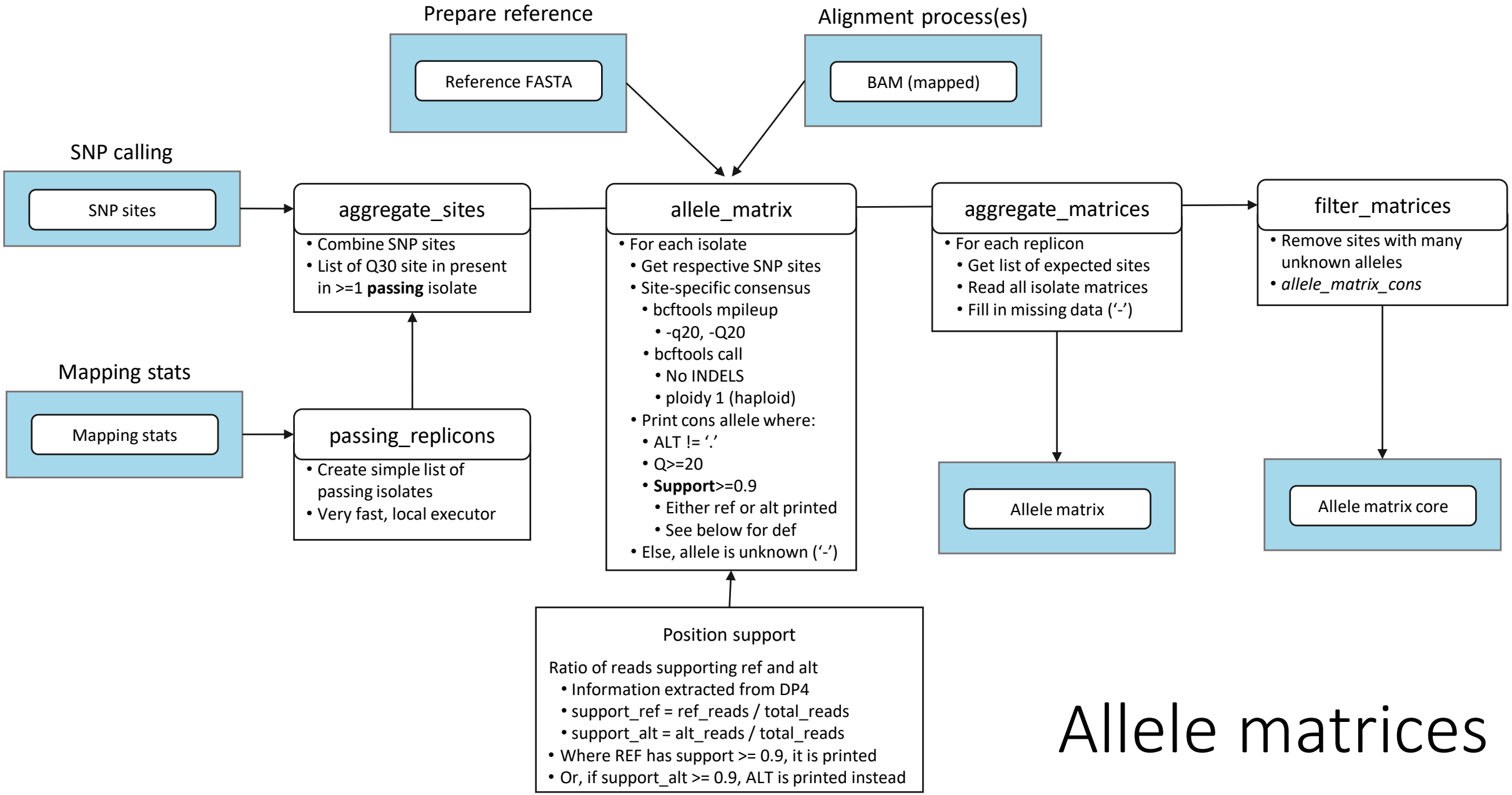


Mapping and SNP calling



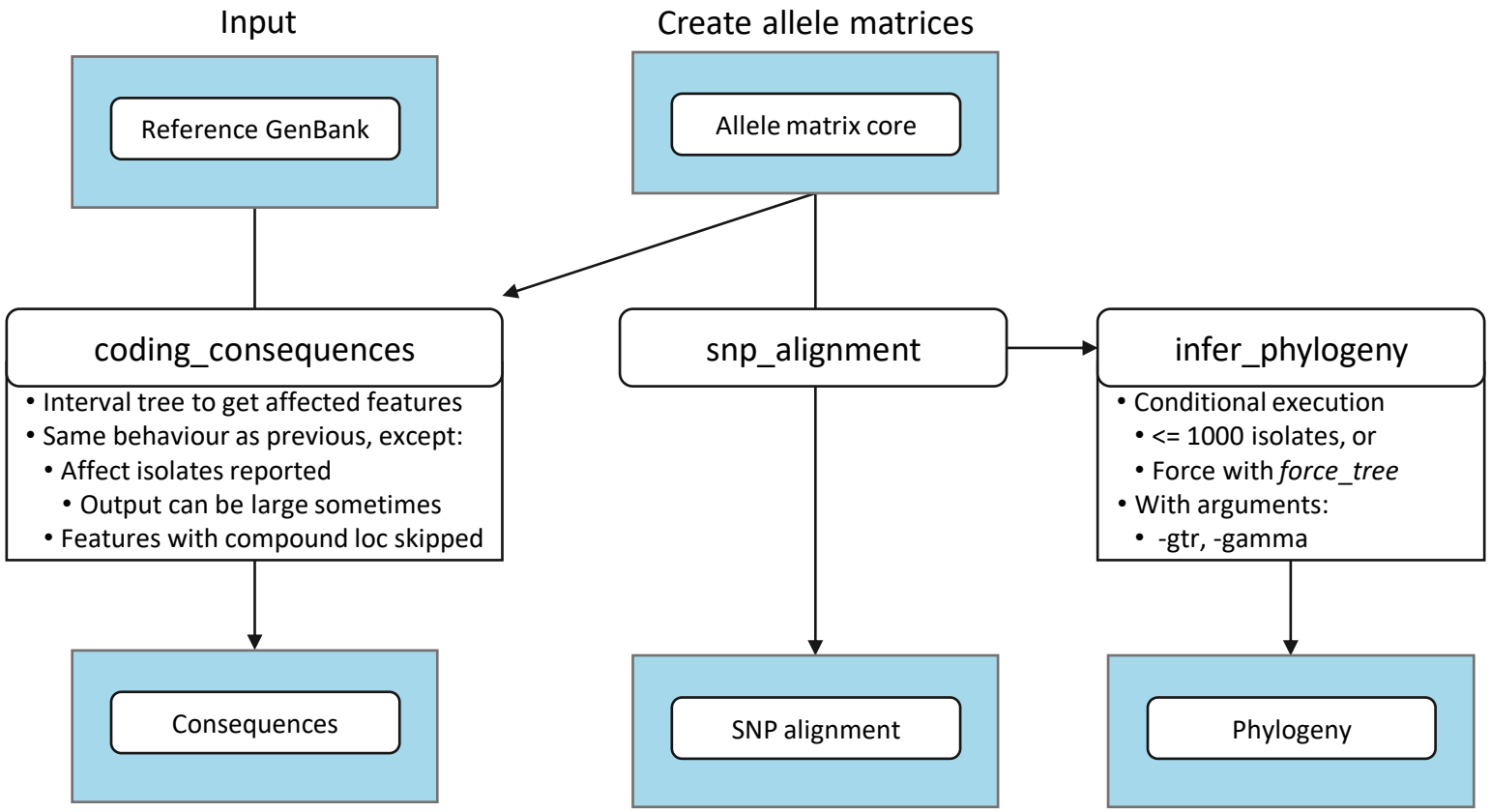
Mapping stats



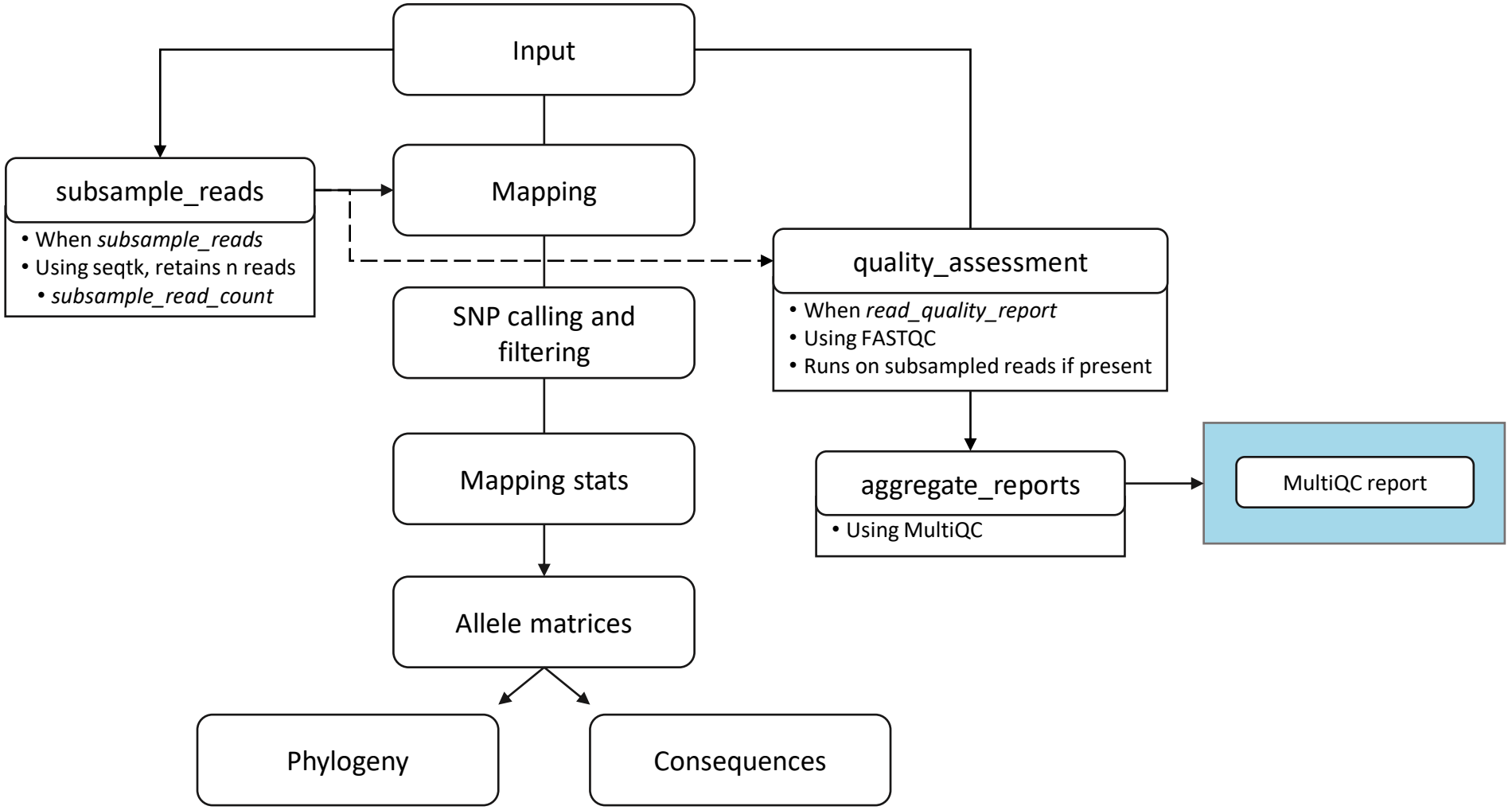


Allele matrices

Consequences, phylogeny



Optional stages

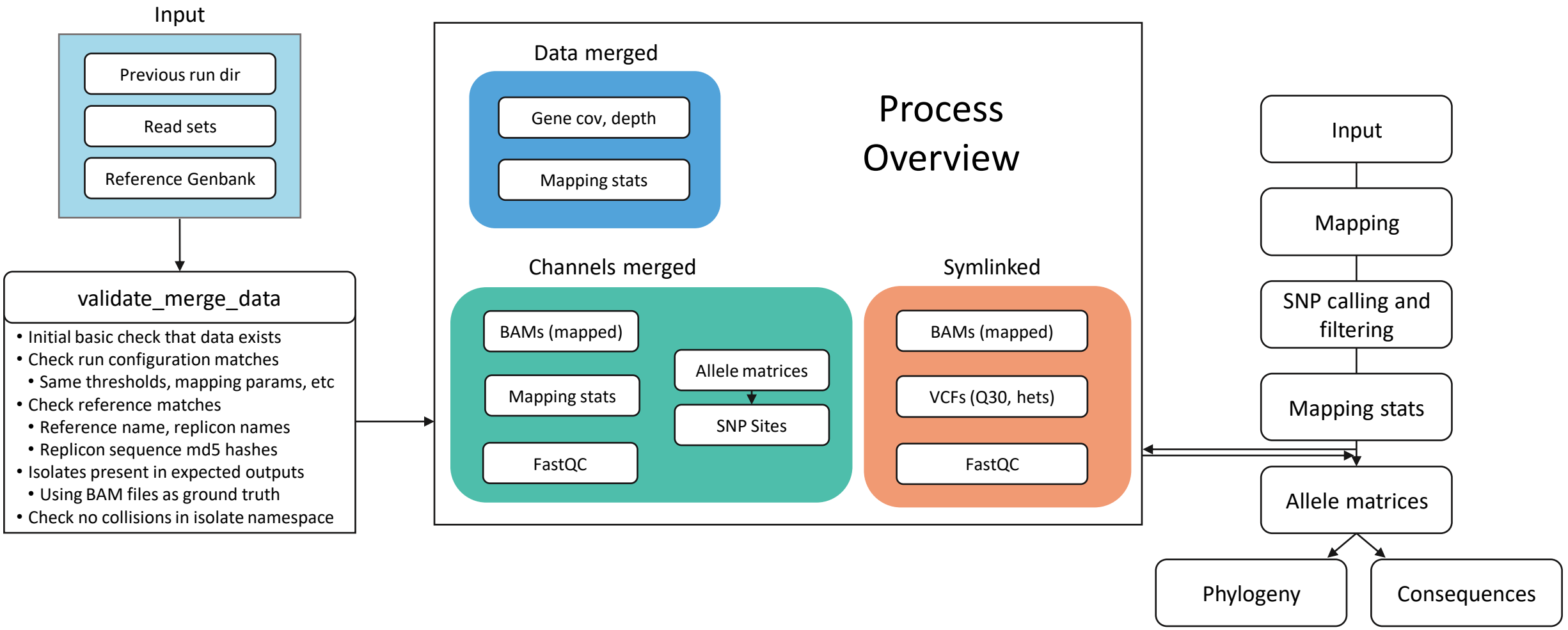


Merge run settings

merge_run: enables a merge run
output_dir: output directory
previous_run_dir: data to merge

merge_ignore_errors: ignore merge data validation errors

Merge runs



Testing

Dataset simulator

Create readsets from single specification file and reference

Isolate spec section

- Name
- Mean depth
- Read length and outer length (for PE)
- Unmappable proportion of reads

Example:

```
# Defaults
metric  value   type
mean_depth      15      int
outer_length    750     int
read_length     250     int
# Readsets
isolate_name    data
## Coding consequences test isolate
isolate_1       read_type:pe
isolate_2       read_type:pe
## Mapping stats pass/ fail test isolates
## Pass
isolate_6       unmapped:0.5;read_type:pe
isolate_7       unmapped:0.45;read_type:pe
isolate_8       mean_depth:11;read_type:pe
isolate_9       mean_depth:25;read_type:pe
isolate_10      mean_depth:11;unmapped:0.45;read_type:pe
```

Modification spec section

- Homozygous SNPs
- Heterozygous SNPs, at specific ratio
- INDELs
- Low quality positions, appear as ‘-’ in allele matrix

Example:

```
# Variants
isolate_name    replicon        type    data
## SNPs to testing coding consequences - start and stop codons, and various others
## Non-synonymous
isolate_1       contig_1        hom     position:1158;alt:a;note:sul1_G2R
isolate_1       contig_1        hom     position:1644;alt:g;note:sul1_T164A
isolate_1       contig_1        hom     position:2081;alt:t;note:sul1_*309Y
isolate_1       contig_1        hom     position:2092;alt:t;note:secA_M1L
## Heterozgyous SNPs
## Both different toreference allele
isolate_3       contig_1        het     position:672;alt_1:t;alt_2:c;ratio:0.5
isolate_3       contig_1        het     position:3181;alt_1:t;alt_2:c;ratio:0.4
## Site with <5% unknown - retained (1/22 unknown (4.5%))
isolate_18      contig_1        hom     position:4820;alt:g;note:secD_S5A
isolate_19      contig_1        low_quality     position:4820
## Site with >5% unknown - filtered (2/22 unknown (9.1%))
isolate_20      contig_1        hom     position:4850;alt:a;note:secD_L15M filtered
isolate_21      contig_1        low_quality     position:4850
isolate_22      contig_1        low_quality     position:4850
```

Automated testing

Tests expected output data from pipeline run on simulated dataset

- Fast turn around for debug loop
- Runtime for 37 isolates on 31 kpb reference (two reps) of ~60 seconds

Compares data specification file to:

- Mapping stats
- Allele tables
- Consequences

Merge run testing approach is to:

- Split simulated dataset into two groups
- Execute a typical run the first group
- Execute merge run with second group
- Run automated comparison

Secondary to simulated datasets, we cat test previously know good runs

- Requires previous run output
- Script compares all major output files
- Performs ordering of data for correct comparison