



config.yml

Pipeline script





.bed + .bim+ .fam files



Database set up

conda

Α

Set up packages,tools

docker

docker image

Download reference data

Internal QC



plink

Removal of very poor quality SNPs Check and removal of poor quality samples Make report

.html report, QC plots, bfiles

D

VCF + .fam file

Human Genome

Build Conversion

LiftOver

b37 to b38 and

plink VCF to binary plink

b38 to b37 conversion

Population Stratification

plink2

Internal QC of 1,000 human genome data

Merge user's + 1,000 human genome dataset

PCA

eigensoft

Removal of ethnic outliers

Make report



.html report, QC and PCA plots, bfiles



Ε Variant QC

plink

Check and removal of poor quality SNPs

Make covariates

PCA with only user's data Make report



Pre-Imputation

plink2, snpflip

Remove duplicated and ambiguous SNPs Flip SNPs that are on

the reverse strand bcftools

Plugin fixref



VCF file



G **Imputation**

В

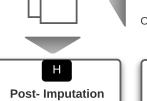
x1

Α

shapeit4 Phasing

impute5 Imputation







VCF + .fam file

bcftools

Filtering of poorly imputed variants

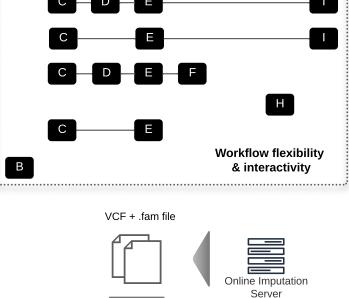
plink2

Handling duplicates and multi-allelics Update phenotypes

Imputed VCF files



plink bfiles







.html report, Manhattan and QQ-plot, GWAS results

