

# eQTL/sQTL detection using Nextflow based workflow

Training on methodology for biology-driven selection

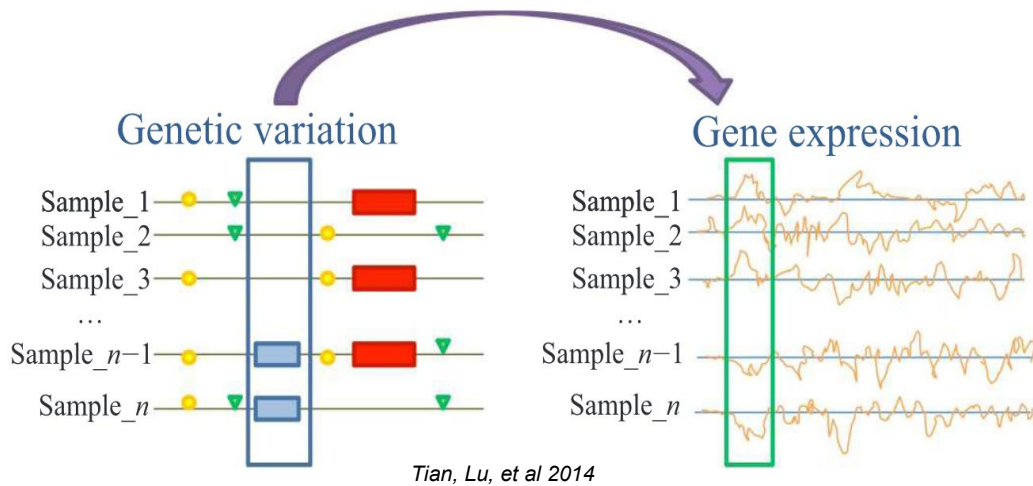
Praveen



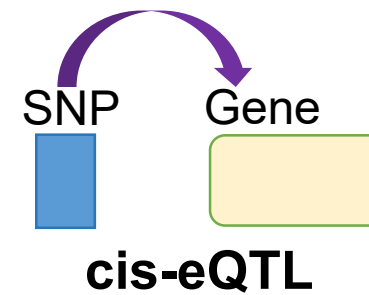
# Introduction

eQTL ( Expression Quantitative Trait Loci ) association study: identifies the genetic variants that affect the gene expression

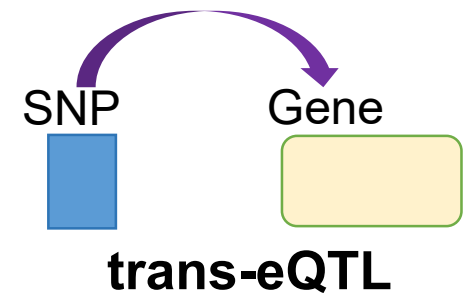
eQTLs explains about 70% of heritability due to DNA variants changing gene expression and RNA splicing via cis and trans effects. *Xiang et al., 2023, Cell Genomics*



Distance  $\leq$  1MB

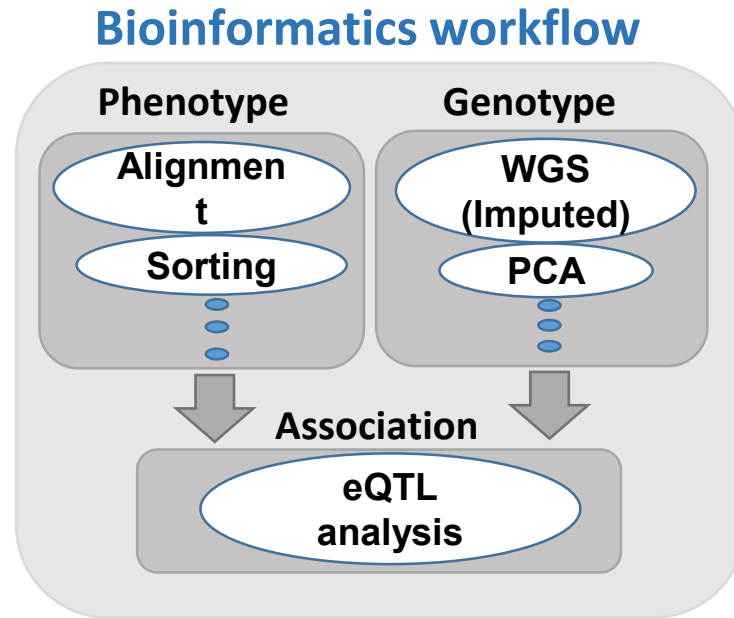


Distance  $>$  1MB



Association between **millions** of genetic variants and **thousands** of genes typically measured in **hundreds** of individuals

# Introduction



```
#!/bin/bash

gtf = bovine_arsuCD.gtf

hisat2_extract_splice_sites.py $gtf >
gtf.bovine.hisat2_splice_sites.txt
```

## nextflow

```
#!/usr/bin/env nextflow

process makeHisatSplicesites {
  container = 'dceoy/hisat2'
  input:
  path gtf
  output:
  path "${gtf.baseName}.hisat2_splice_sites.txt"
  script:
  """
  hisat2_extract_splice_sites.py $gtf >
  ${gtf.baseName}.hisat2_splice_sites.txt
  """
}
```

## Workflow managers

- Scalable and reproducible.

- Simple declaration for calling the input data, executing the process and standard guidelines for installing all the required software tools.

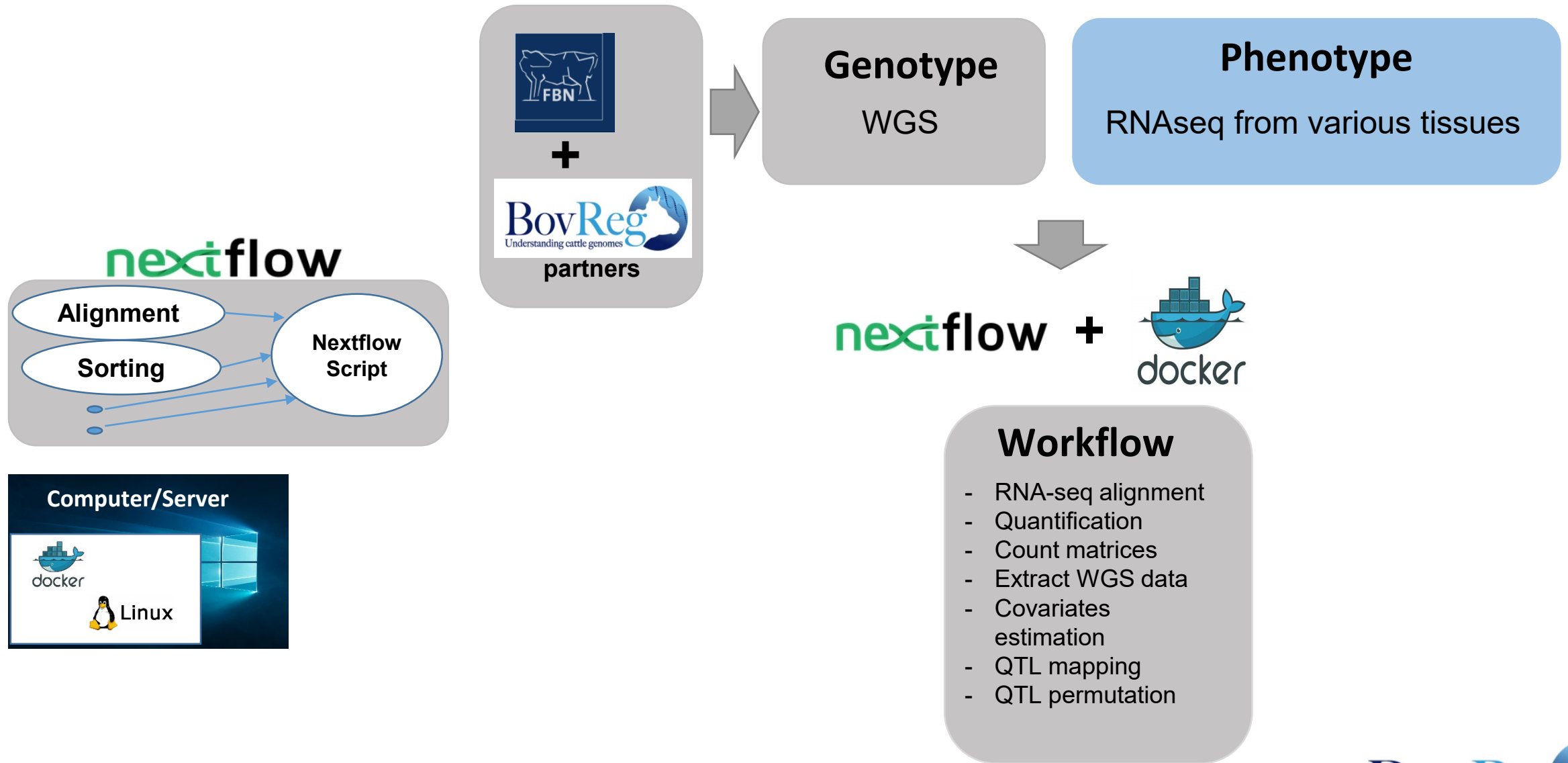


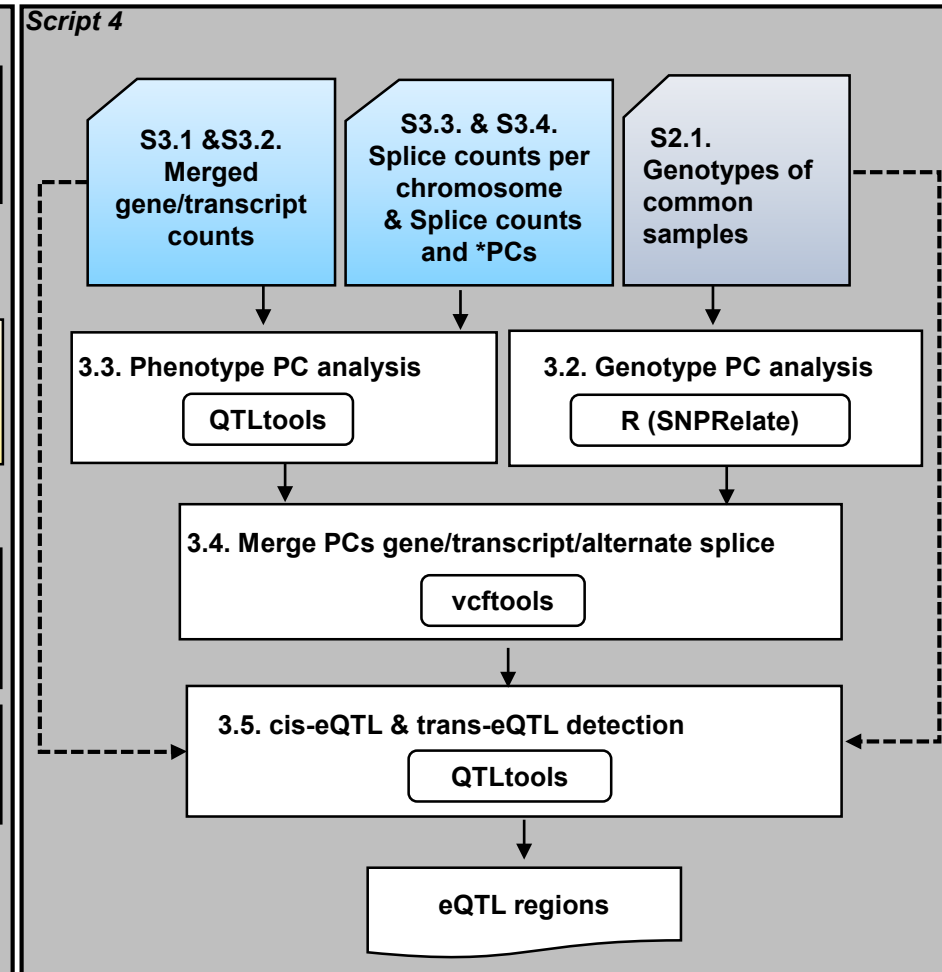
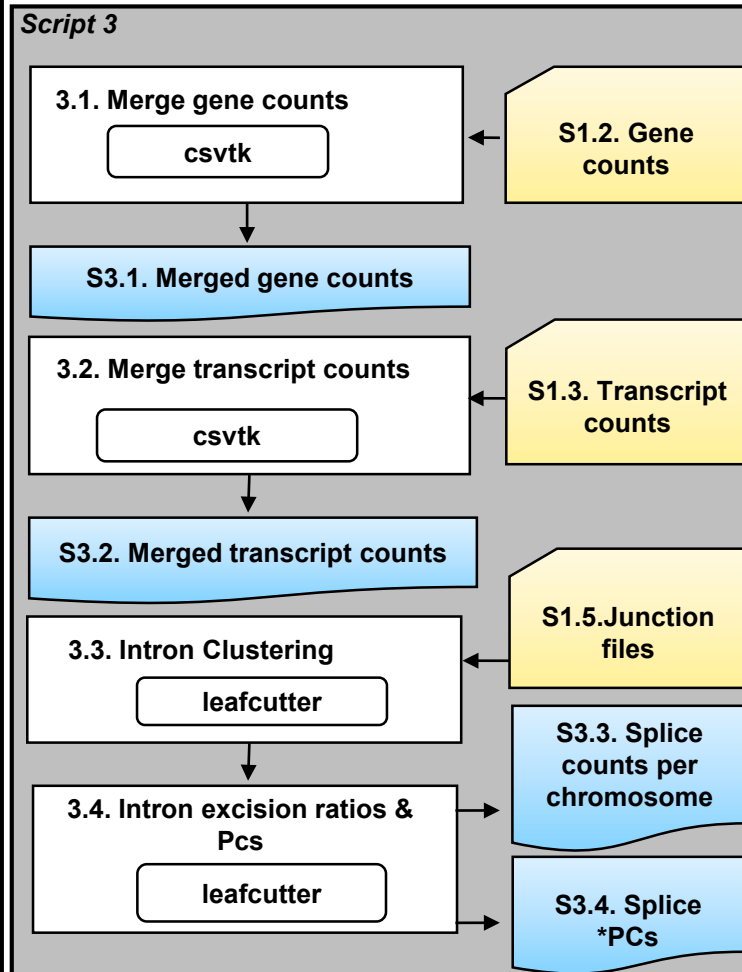
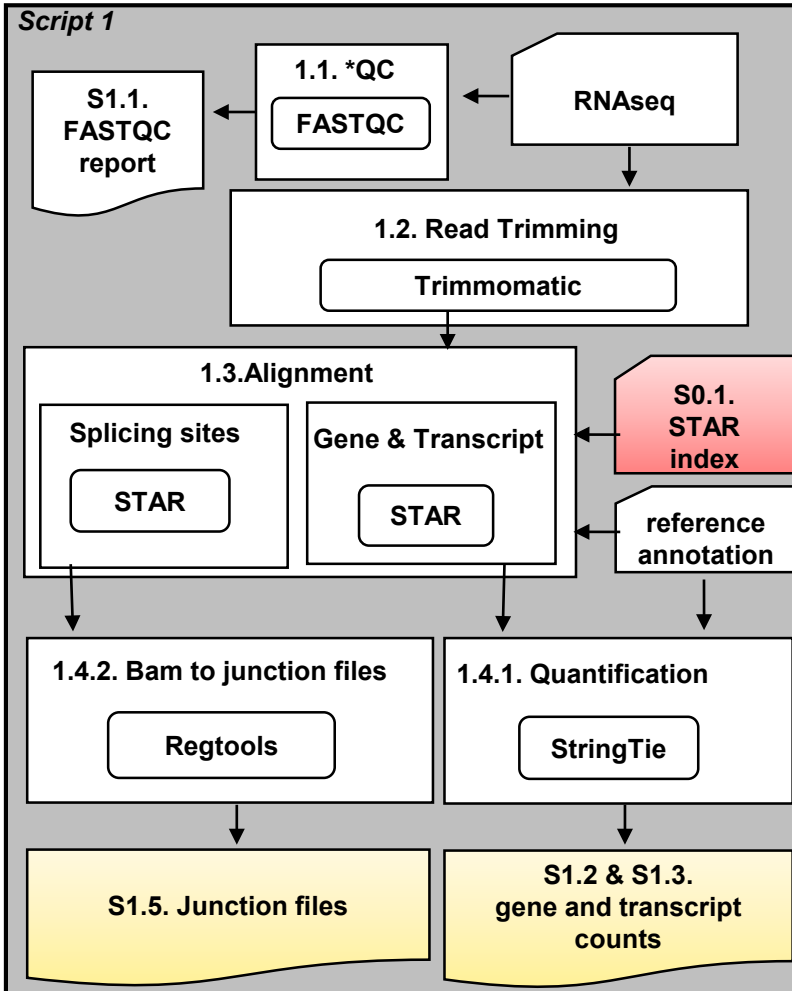
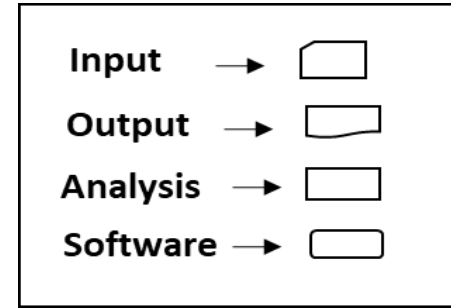
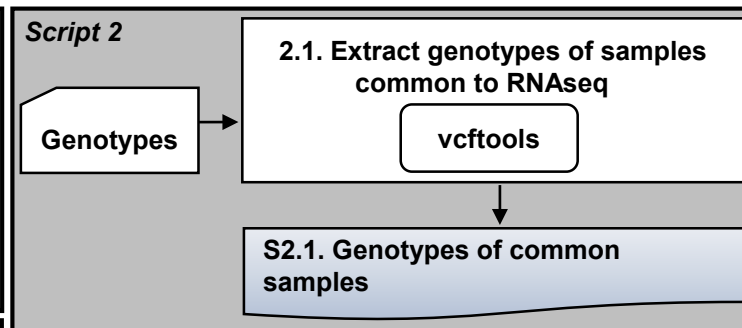
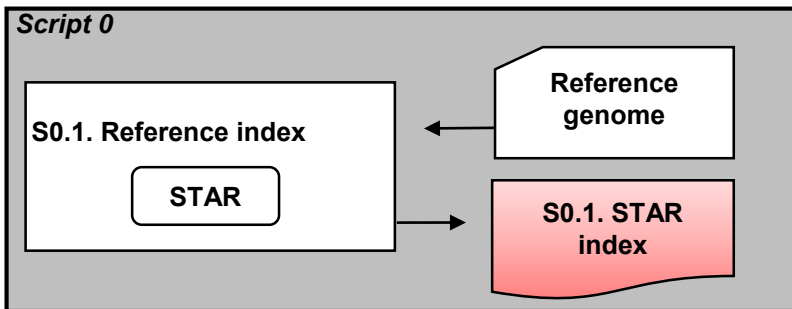
## Containers

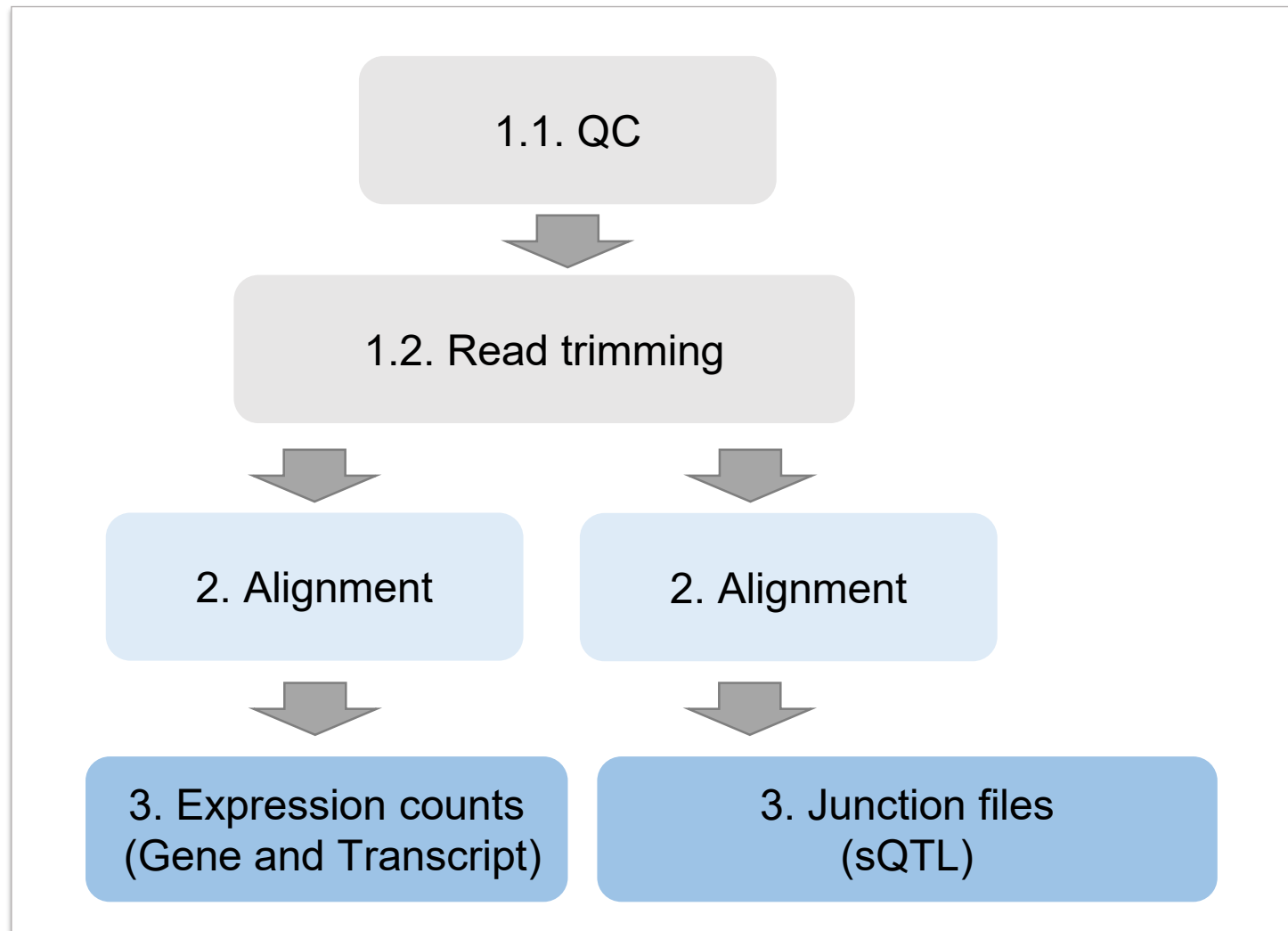
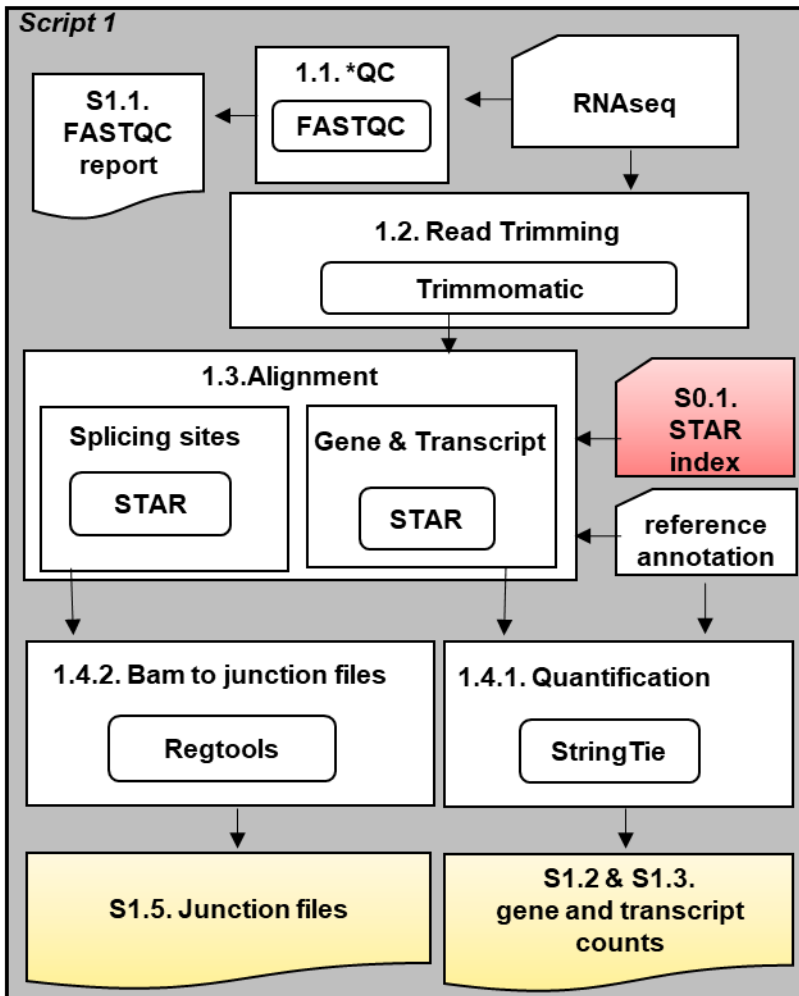
- A standard unit of software that contains all of the necessary elements to run a bioinformatics tool in any computational environment.

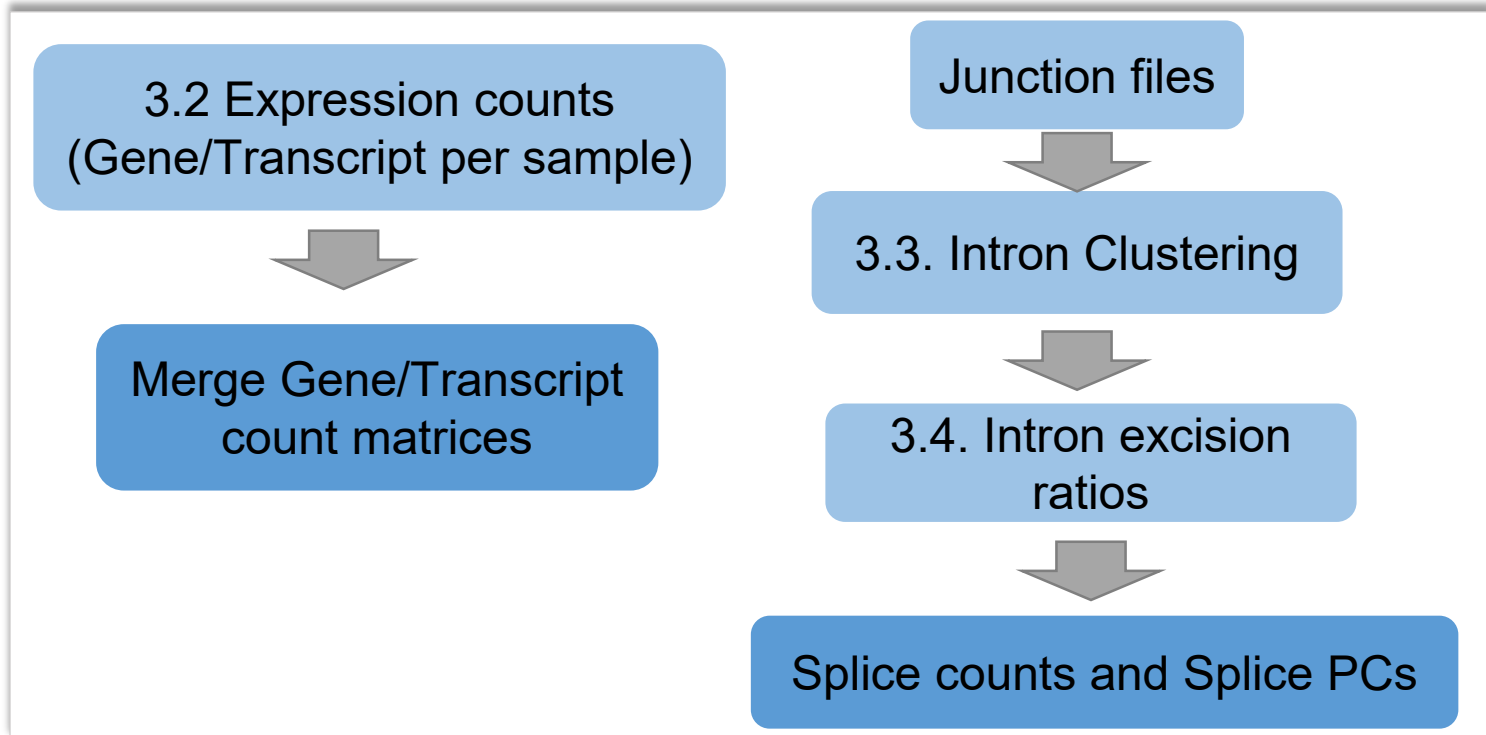
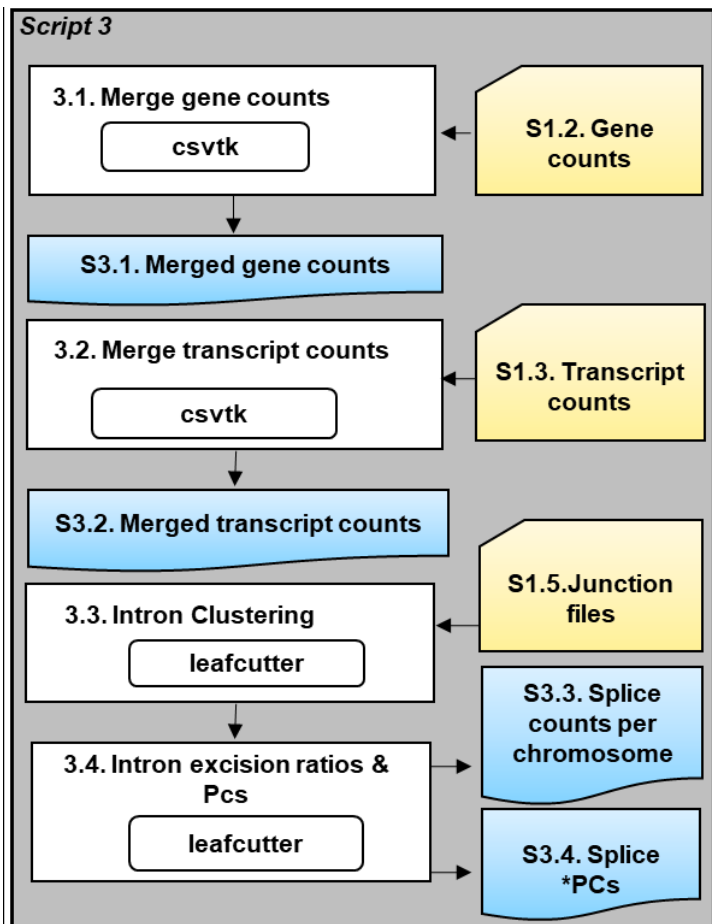
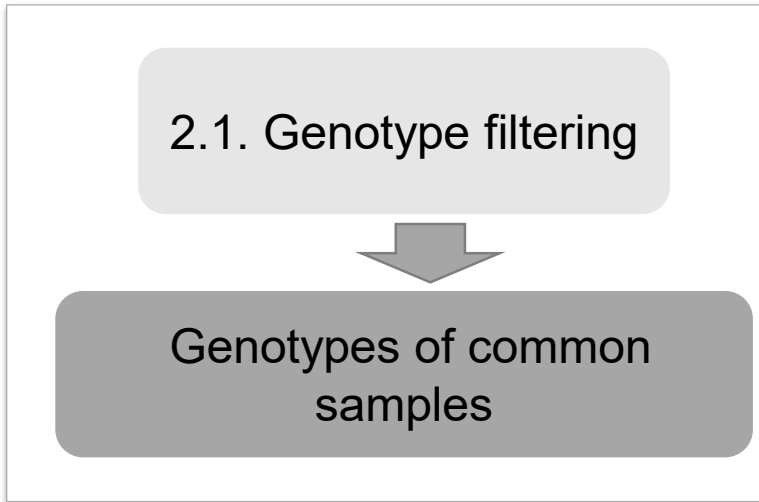
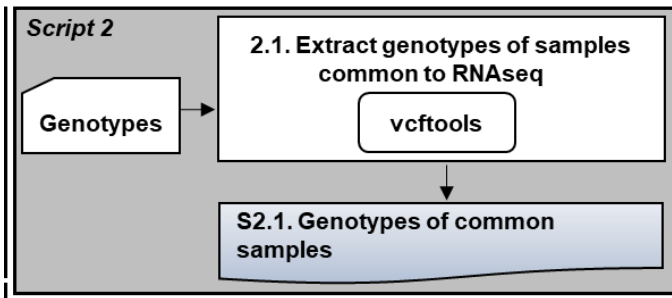


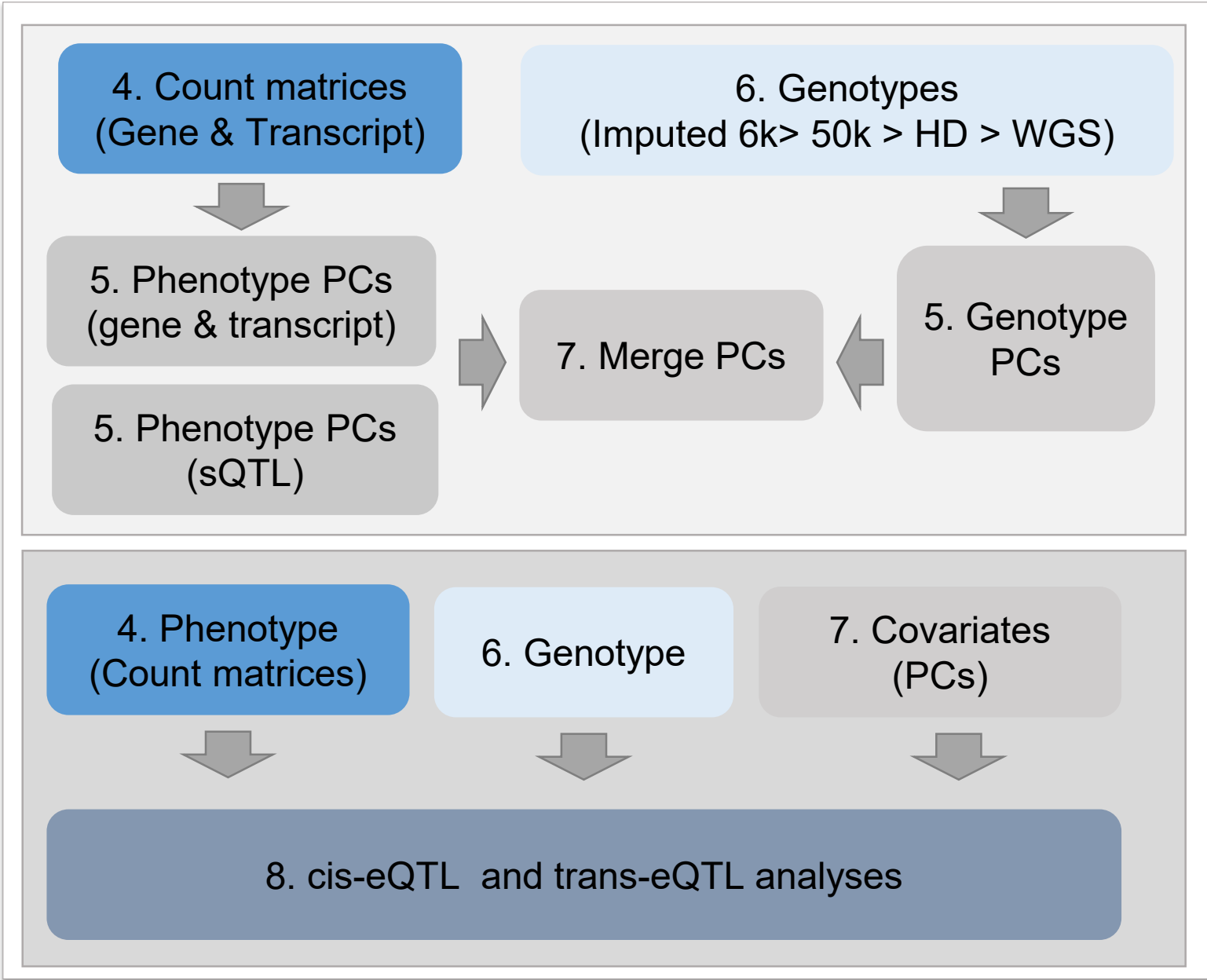
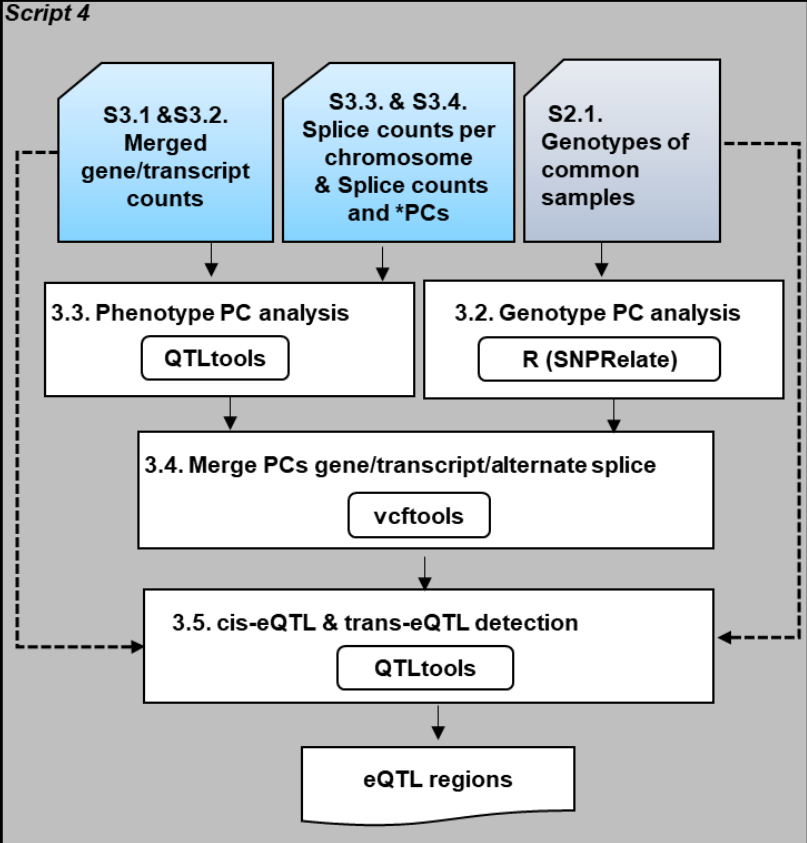
# eQTL detection strategy







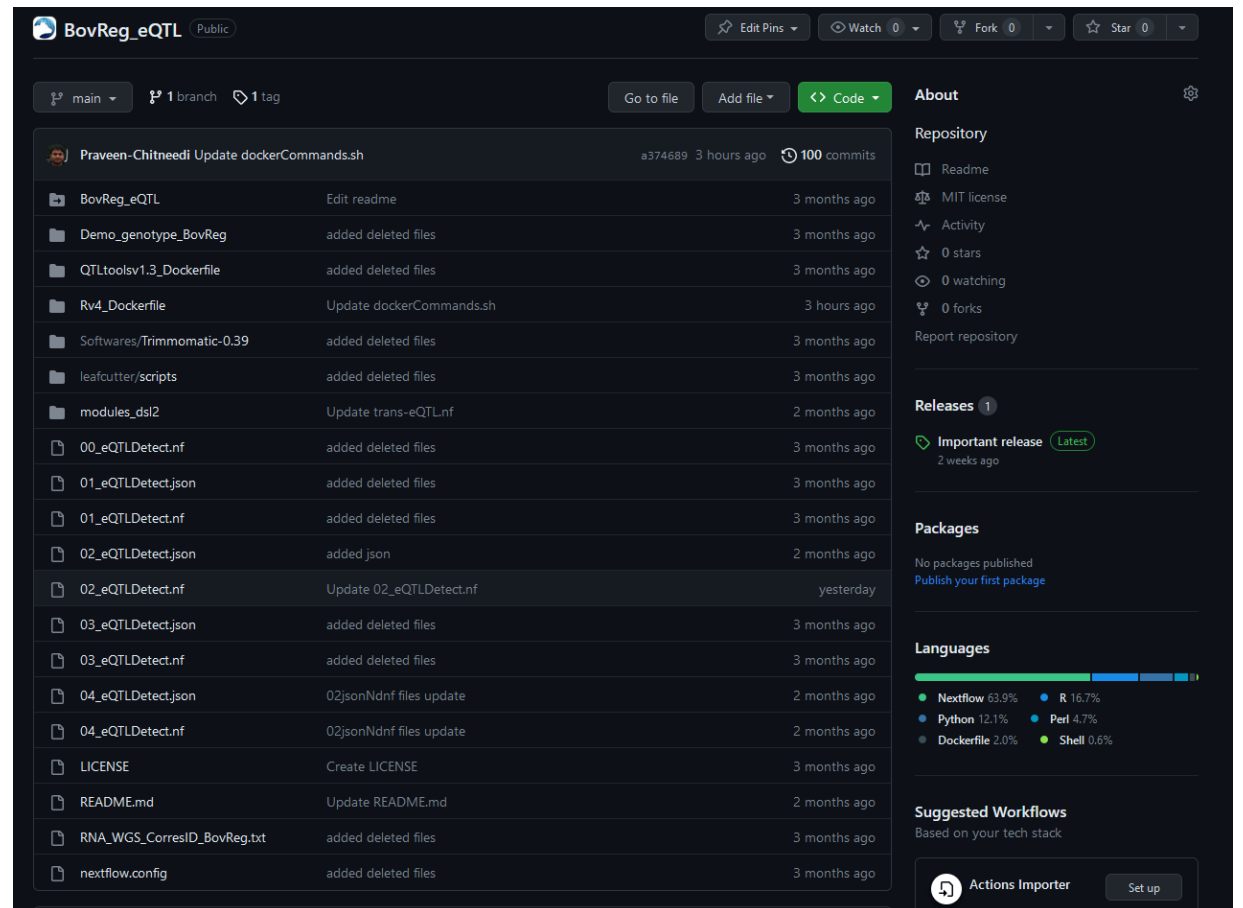






# source code

[https://github.com/BovReg/BovReg\\_eQTL](https://github.com/BovReg/BovReg_eQTL)



The screenshot displays the GitHub repository page for **BovReg\_eQTL**, which is a public repository. The interface includes a navigation bar at the top with options for 'Edit Pins', 'Watch' (0), 'Fork' (0), and 'Star' (0). Below the repository name, there are buttons for 'Go to file', 'Add file', and 'Code'. The main content area shows a list of files and folders, each with a commit message and a timestamp. The right sidebar provides an 'About' section with repository statistics (0 stars, 0 watching, 0 forks) and a 'Releases' section showing one 'Important release' (Latest) from 2 weeks ago. The 'Packages' section indicates no packages are published. The 'Languages' section shows a bar chart for the repository's language composition: Nextflow (63.9%), Python (12.1%), Dockerfile (2.0%), R (16.7%), Perl (4.7%), and Shell (0.6%). At the bottom, there is a 'Suggested Workflows' section with an 'Actions Importer' button.

| File/Folder                 | Commit Message           | Time Ago     |
|-----------------------------|--------------------------|--------------|
| BovReg_eQTL                 | Edit readme              | 3 months ago |
| Demo_genotype_BovReg        | added deleted files      | 3 months ago |
| QTLtoolsv1.3_Dockerfile     | added deleted files      | 3 months ago |
| Rv4_Dockerfile              | Update dockerCommands.sh | 3 hours ago  |
| Softwares/Trimomatic-0.39   | added deleted files      | 3 months ago |
| leafcutter/scripts          | added deleted files      | 3 months ago |
| modules_dsl2                | Update trans-eQTL.nf     | 2 months ago |
| 00_eQTLDetect.nf            | added deleted files      | 3 months ago |
| 01_eQTLDetect.json          | added deleted files      | 3 months ago |
| 01_eQTLDetect.nf            | added deleted files      | 3 months ago |
| 02_eQTLDetect.json          | added json               | 2 months ago |
| 02_eQTLDetect.nf            | Update 02_eQTLDetect.nf  | yesterday    |
| 03_eQTLDetect.json          | added deleted files      | 3 months ago |
| 03_eQTLDetect.nf            | added deleted files      | 3 months ago |
| 04_eQTLDetect.json          | 02jsonNdnf files update  | 2 months ago |
| 04_eQTLDetect.nf            | 02jsonNdnf files update  | 2 months ago |
| LICENSE                     | Create LICENSE           | 3 months ago |
| README.md                   | Update README.md         | 2 months ago |
| RNA_WGS_CorresID_BovReg.txt | added deleted files      | 3 months ago |
| nextflow.config             | added deleted files      | 3 months ago |