

# Genomic projects

Stefano Scansani

March 12, 2024

## Abstract

So far this is the  $\text{\LaTeX}$  version of the README.md.

## Contents

<b>1</b>	<b>Genomic projects tutorials</b>	<b>1</b>
1.1	Contents . . . . .	2
1.2	Tools . . . . .	2
1.3	Example case studies . . . . .	3
1.4	Resources & Data . . . . .	4
1.5	Setup of the working environment . . . . .	4
1.5.1	Get PLINK working in Linux . . . . .	4
1.5.2	Get TASSEL (GUI) on Linux . . . . .	5
1.5.3	Get GEMMA . . . . .	6
1.5.4	Get GAPIT (R) . . . . .	6
<b>2</b>	<b>Project: SNP Profiling of worldwide goat populations</b>	<b>6</b>
2.1	Data . . . . .	6
<b>3</b>	<b>Methods</b>	<b>6</b>
3.0.1	Data import . . . . .	6
<b>4</b>	<b>Results</b>	<b>6</b>

## List of Figures

## List of Tables

## 1 Genomic projects tutorials

:warning: This repository is under construction :warning:

This repository contains a collection of genomic projects that I am working on. GitHub repository of bioinformatic projects revolving around genomics using different tools like Plink through `plinkr` R package, `rTASSEL` and TASSEL 5 (GUI),

GEMMA for mixed models analysis in R, SAMtools to analyze BAM files, gBLUP coming soon.

The repository has been created for testing and self-teaching purposes of biological concept and bioinformatic tools, and make use of other repositories, scripts and data sources, taken or modified as such.

## 1.1 Contents

- Genomic projects tutorials<sup>1</sup>
- Contents<sup>2</sup>
- Tools<sup>3</sup>
- Example case studies<sup>4</sup>
- Resources & Data<sup>5</sup>
- Setup of the working environment<sup>6</sup>
  - Get PLINK working in Linux<sup>7</sup>
  - Get plinkr (R)<sup>8</sup>
  - Get TASSEL (GUI) on Linux<sup>9</sup>
  - Get rTASSEL (R)<sup>10</sup>
  - Get GEMMA<sup>11</sup>
  - Get GAPIT (R)<sup>12</sup>

## 1.2 Tools

- **PLINK 1.90** <https://www.cog-genomics.org/plink2/>
- plinkr R package repository documentation. <https://github.com/AJResearchGroup/plinkr>
- **TASSEL 5** <https://www.maizegenetics.net/tassel>. **Bradbury et al.**, (2007) TASSEL: software for association mapping of complex traits in diverse samples, *Bioinformatics*, Volume 23, Issue 19, Pages 2633-2635 <https://doi.org/10.1093/bioinformatics/btm308>
- rTASSEL R package repository documentation. <br> Vignettes: <https://rtassel.maizegenetics.net/index.html>, Repository: <https://github.com/maize-genetics/rTASSEL>. **Monier et al.**, (2022). rTASSEL: An R interface to TASSEL for analyzing genomic diversity. *Journal of Open Source Software*, 7(76), 4530, <https://doi.org/10.21105/joss.04530>

---

<sup>1</sup>#genomic-projects-tutorials  
<sup>2</sup>#contents  
<sup>3</sup>#tools  
<sup>4</sup>#example-case-studies  
<sup>5</sup>#resources--data  
<sup>6</sup>#setup-of-the-working-environment  
<sup>7</sup>#get-plink-working-in-linux  
<sup>8</sup>#get-plinkr-r  
<sup>9</sup>#get-tassel-gui-on-linux  
<sup>10</sup>#get-rtassel-r  
<sup>11</sup>#get-gemma  
<sup>12</sup>#get-gapit-r

- GEMMA Genome-wide Efficient Mixed Model Association <https://github.com/genetics-statistics/GEMMA>. **Xiang Zhou and Matthew Stephens** (2012). Genome-wide efficient mixed-model analysis for association studies. *Nature Genetics* 44, 821824.
- rMVP A Memory-efficient, Visualization-enhanced, and Parallel-accelerated Tool for Genome-Wide Association Study <https://github.com/xiaolei-lab/rMVP>
- GPtour Genomic Prediction in R using Keras models <https://github.com/miguelperezenciso/GPtour> and [https://keras.posit.co/articles/getting\\_started.html](https://keras.posit.co/articles/getting_started.html)
- GAPIT Genome Association and Integrated Tools <https://github.com/jiabowang/GAPIT>

### 1.3 Example case studies

1. SNP profiling of goat breeds.<br>*Data source:* **Colli et al.** (2018) <https://doi.org/10.1186/s12711-018-0422-x>

  
  
<small>**Multidimensional Scaling (MDS) Plot of a population of 4,653 Individuals from 169 Goat Breeds genotyped with 49,953 SNPs.**</small>

<small>The MDS plot visualizes genetic relationships among 4,653 individuals from 169 goat breeds. Genetic distances were computed using PLINK to generate the distance matrix, and MDS analysis was conducted with the `cmdscale` function based on genotyping data from 49,953 SNPs. Each point represents a goat, and spatial arrangement reflects genetic dissimilarities. This exploratory analysis offers insights into genetic diversity, population structure, and relatedness. </small>

1. a. Manhattan plot of a GWAS on dog population for deafness.*Data source:* **Hayward et al.** (2020) <https://doi.org/10.1371/journal.pone.0232900>

  
  
<small>Manhattan plots showing the genome wide association (GWA) between dog deafness and their genotype. The plot displays the genomic positions of single nucleotide polymorphisms (SNPs) across the genome on the x-axis, with the corresponding  $-\log_{10}$  transformed P-values indicating the strength of association with the trait on the y-axis. The red-dashed lines are representation of the 99.99 percentile threshold of the LOD values.</small>

2. b. Plot of the top significant SNPs identified in the above GWAS.

<small>Points are jittered around their respective chromosome.</small>  
  
and a zoom in the chromosome 3 above the 99.99 percentile (LOD score = 4.71).  


## 1.4 Resources & Data

- **Marees et al.** (2018) A tutorial on conducting genome-wide association studies: Quality control and statistical analysis. *Int J Methods Psychiatr Res.* 27:e1608. <https://doi.org/10.1002/mpr.1608>
- **Marees et al.** (2018) tutorial [https://github.com/MareesAT/GWA\\_tutorial](https://github.com/MareesAT/GWA_tutorial)
- **Gábor Mészáros** (2021) Genomic Boot Camp Book <https://genomicsbootcamp.github.io/book/>
- **Gábor Mészáros** video tutorials <https://www.youtube.com/c/GenomicsBootCamp>
- **Colli et al.** (2018) Genome-wide SNP profiling of worldwide goat populations reveals strong partitioning of diversity and highlights post-domestication migration routes. *Genet Sel Evol* 50, 58. <https://doi.org/10.1186/s12711-018-0422-x>
- DATA: **Colli et al.** (2020). Signatures of selection and environmental adaptation across the goat genome post-domestication [Dataset]. *Dryad*. <https://doi.org/10.5061/dryad.v8g21pt>
- **Decker et al.** (2014) Worldwide Patterns of Ancestry, Divergence, and Admixture in Domesticated Cattle. *PLOS Genetics* 10(3): e1004254. <https://doi.org/10.1371/journal.pgen.1004254><sup>13</sup>,
- DATA: **Decker et al.** (2015) Worldwide patterns of ancestry, divergence, and admixture in domesticated cattle [Dataset]. *Dryad*. <https://doi.org/10.5061/dryad.th092>

## 1.5 Setup of the working environment

Install R: The Comprehensive R Archive Network (CRAN)<sup>14</sup>

IDE: VSCode<sup>15</sup>/RStudio<sup>16</sup><sup>\*</sup>

Install Python: Miniconda 3<sup>17</sup><sup>\*</sup>

OS: Linux<sup>\*</sup>/WSL

<small><sup>\*</sup>Suggested</small>

### 1.5.1 Get PLINK working in Linux

1. Download PLINK 1.90 Linux 64-bit<sup>18</sup>

2. Install PLINK `cd Downloads/ sudo unzip plink_linux_x86_64_20200616.zip -d plink_install`

<sup>13</sup><https://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1004254>

<sup>14</sup><https://cran.r-project.org/>

<sup>15</sup><https://code.visualstudio.com/>

<sup>16</sup><https://posit.co/download/>

<sup>17</sup><https://docs.anaconda.com/free/miniconda/index.html>

<sup>18</sup>[https://s3.amazonaws.com/plink1-assets/plink\\_linux\\_x86\\_64\\_20231211.zip](https://s3.amazonaws.com/plink1-assets/plink_linux_x86_64_20231211.zip)

### 3. PLINK in usr/local/bin

```
cd plink_install sudo cp plink /usr/local/bin sudo chmod 755 /usr/local/bin/plink
```

### 4. Add PLINK to PATH

with bash/zsh/...

```
sudo nano ~/.bashrc
```

and include the line:

```
export PATH=/usr/local/bin:$PATH
```

Save and exit. Refresh the terminal and you should be able to call plink from the terminal at any user position in the system.

```
source ~/.bashrc plink -help
```

### Get plinkr (R)

PLINK directly in R.

refer to the installation guide at <https://github.com/AJResearchGroup/plinkr/blob/master/doc/install.md>

```
library(remotes) install_github("richelbilderbeek/plinkr") remotes::install_github("chng/2.0/pgenlibr") library(plinkr) install_plinks()
```

### 1.5.2 Get TASSEL (GUI) on Linux

1. Go on the website <https://www.maizegenetics.net/tassel> and download the last UNIX version.
2. Download the TASSEL\_{xxx}\_unix.sh and make it executable `chmod +x ~/Downloads/TASSEL_{xxx}_unix.sh`
3. Run the TASSEL installer `~/Downloads/TASSEL_{xxx}_unix.sh`

### Get rTASSEL (R)

#### 1. rJava installation

```
sudo apt install default-jdk sudo R CMD javareconf R install.packages("rJava")
```

#### 2. Installation in R

```
if (!require("devtools")) install.packages("devtools") devtools::install_github(repo = "maize-genetics/rTASSEL", ref = "master", build_vignettes = TRUE, dependencies = TRUE )
```

#### 3. Run rTASSEL

- Allocate job's memory<sup>>1</sup> and start the logger (here at the root of the project):</sup>

<sup>>1</sup>"-Xmx50g" and "-Xms50g", "50g" represents 50 Gigabytes of memory.</sup>

*!! Choose an appropriate value that fits your machine !!*

```
options(java.parameters = c("-Xmx50g", "-Xms50g")) rTASSEL::startLogger(fullPath = NULL, fileName = NULL)
```

- Run & infos

```
library(rTASSEL) ??rTASSEL
```

Useful resource for rTASSEL are the vignettes and tutorials at <https://rtassel.maizegenetics.net/index.html>

### 1.5.3 Get GEMMA

GEMMA can be installed from source at the GitHub repo, but is also available through Bioconda <http://www.ddocent.com/bioconda/>. To install is suggested to have miniconda installed and working, and then added the channel for Bioconda, you should already have defaults and conda-forge.

```
conda config --add channels defaults conda config --add channels
conda-forge conda config --add channels bioconda conda install gemma
And use GEMMA with
gemma -h
```

### 1.5.4 Get GAPIT (R)

R package, here we are going to install it through GitHub. For the manual visit <https://zzlab.net/GAPIT/gapithelpdocument.pdf>

```
R> install.packages("devtools") R> devtools::install_github("jiabowang/GAPIT",
force=TRUE) R> library(GAPIT)
```

---

## 2 Project: SNP Profiling of worldwide goat populations

### 2.1 Data

Data source: Colli et al. (2018) <https://doi.org/10.1186/s12711-018-0422-x>

## 3 Methods

### 3.0.1 Data import

## 4 Results