QC Functions User Manual

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February 6, 2017

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1 Introduction

This manual shows you how to use the quality control functions and give an example for demonstration.

2 Prerequest

To make the functions run successfully, please ensure you have installed PLINK in previous.
After you installed PLINK, please copy it from defult folder and paste it to /usr/local/bin.

```
$ cp ~/Bin/plink /usr/local/bin
```

3 Caution

Do not modify any content arbitrarily of ouput files generated from PLINK!

4 QC Functions

4.1 Per-individual QC

- sex_check(input.name, output.name)
  Place the name of your PLINK binary files(BED/BIM/FAM) at ”input.name” argument, and give the output name at ”output.name” argument. After the analysis, you will get a plot(”sex_distribution.pdf”) which shows the distribution of individual’s sex and a ”output.name_sex_problem.list” file which records individuals with discordant sex information. Defult homozygosity rates of identifying individual as male is above 0.8, and recognizing as female is below 0.2.

- missing_het_ind(input.name, pop.list, output.name)
  You still need to give input and ouput names for the analysis. You also need to give a list of individuals with their populations at ”pop.list” argument, the example format shows below:

1	TDC13	Paiwan
2	TDC117	Amis
3	TDC18	Bunun
4	TDC129	Amis
5	TDC49	Amis
6	TDC497	Puyuma

  You will get a plot(”imiss-vs-het.pdf”) which shows the distribution of missingness and heterozygosity scores and a ”output.name_miss_het_problem.list” file which records individuals do not pass criteria. Default cuttoffs of genotype failure rates are equal or larger than 0.03 and heterozygosity rates deviate more or less 3 s.d. from the mean.

- IBD(input.name, output.name)
  You still need to give input and ouput names for the analysis. After the analysis, you will get a plot(”IBD.pdf”) which shows the propotion of the different IBD between pairs of individuals. You will also get a ”output.name_ibd_problem.list” that records the individuals do not pass the criterion. Default value of IBD we intend to remove is higher than 0.1875.

- ind_qc_rm(input.name, output.name)
  After you finish the steps of per-individual QC, you can use ind_qc_rm function to output the

list("output.name_fail_ind_qc.txt") which contains all the problem lists that are generated by previous QC steps. You can use following PLINK command to remove the individuals easily:

```
plink --bfile your.PLINK.bfile --remove output.name_fail_ind_QC.txt --make-bed --
out output.name
```

4.2 Per-SNP QC

- missing_snp(input.name, output.name)
  You still need to give input and ouput names for the analysis. You will get a plot("snpmiss_plot.pdf") which shows the distribution of missing genotype rate and a threshold for extreme genotype failure rate. Default missing genotype rate threshold is equal to 0.03.
- hwe_test(input.name, output.name)
  You still need to give input and ouput names for the analysis. You will get a plot("hwe_p_value.pdf") which shows the distribution of Hardy-Weinberg Equilibrium test's p-value and a threshold for extreme high p-value. Default extreme p-value threshold is equal to 0.00001.

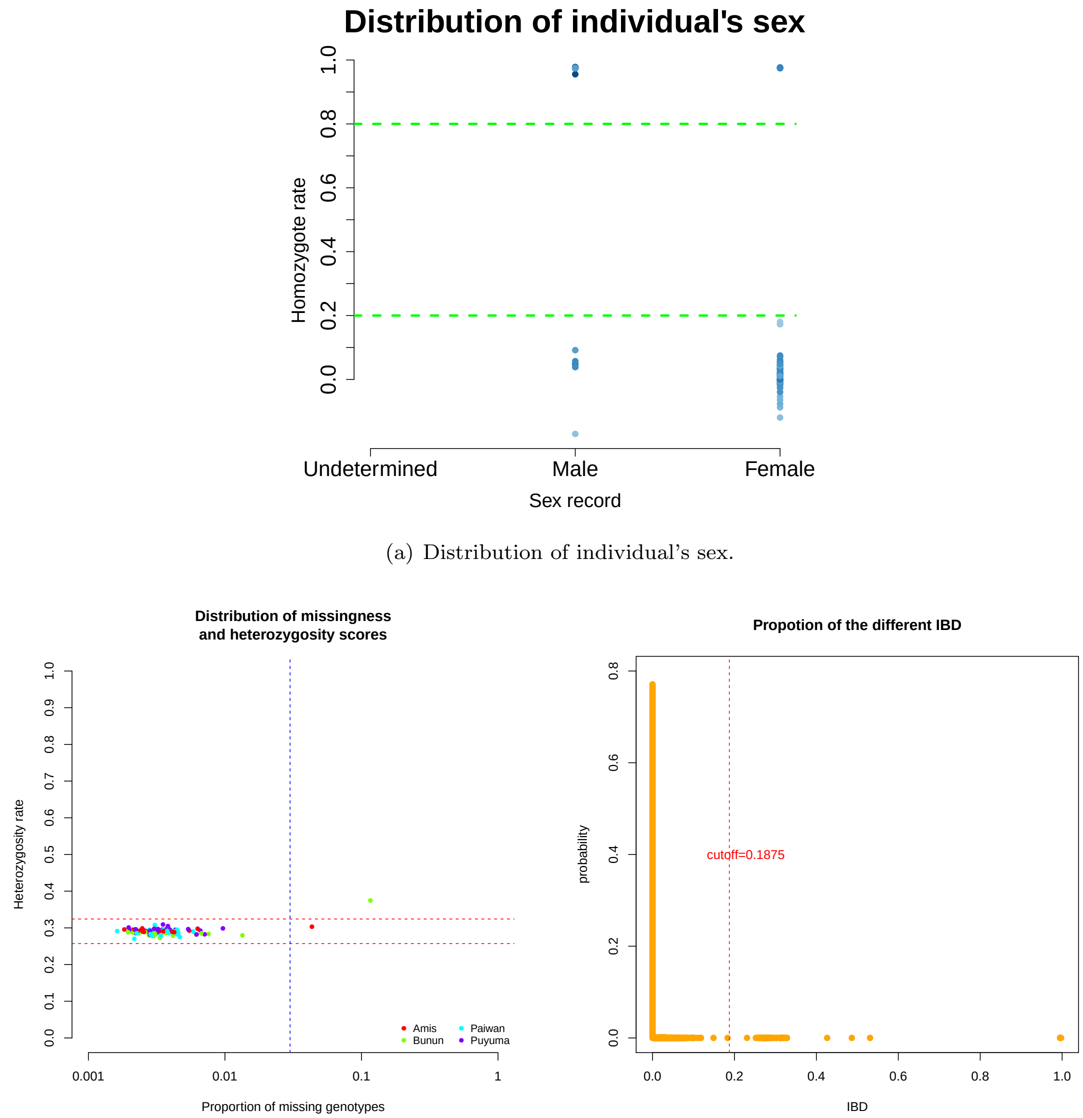
Two per-SNP QC steps show above are aim to show the distribution of missing genotype rate and Hardy-Weinberg Equilibrium test's p-value. If you want to further exclude those SNPs, please use following PLINK commands:

```
plink --bfile your.PLINK.bfile --maf 0.01 --geno 0.03 --hwe 0.00001 --make-bed --out out-
put.name
```

5 Example

Here we give you an example of applying the functions we have illustrated. The example data is consists of 96 individuals and about 2.5 millions of SNPs.

5.1 Per-individual QC



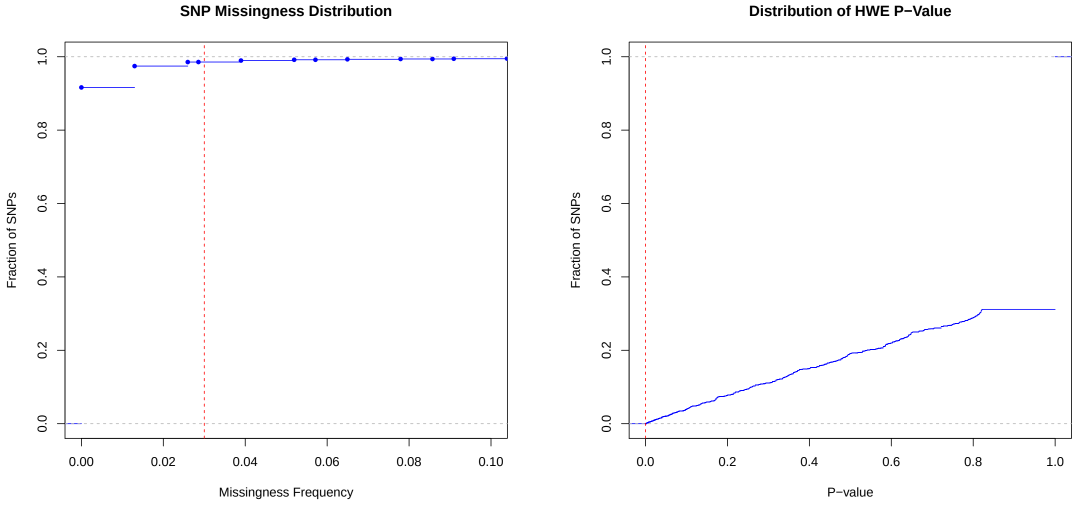
(a) Distribution of individual's sex.

(b) Distribution of missingness and heterozygosity scores.

(c) Propotion of the different IBD

Figure 1: Three analysis in per-individual QC.

5.2 Per-SNP QC



(a) Distribution of missing genotype rate.

(b) Distribution of Hardy-Weinberg Equilibrium test's p-value.

Figure 2: Two analysis in per-SNP QC.

References

[1] Anderson CA, Pettersson FH, Clarke GM, Cardon LR, Morris AP, Zondervan KT. Data quality control in genetic case-control association studies. *Nature protocols.* 2010;5(9):1564-1573. doi:10.1038/nprot.2010.116.

[2] Purcell S, Neale B, Todd-Brown K, et al. PLINK: A Tool Set for Whole-Genome Association and Population-Based Linkage Analyses. *American Journal of Human Genetics.* 2007;81(3):559-575.