# Snp HeRitability Estimation Kit (SHREK) Development Log

Sam Shing Wan, Choi choishingwan@gmail.com

Johnny Sheung Him, Kwan shkwan@hku.hk

Henry Chi-Ming, Leung cmleung2@cs.hku.hk

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### 1 Assumption in implementation

To make life easier, I made a number of assumption when I implement the programme and they are listed as follow

- 1. The intervals within each individual bed files are non-overlapping
- 2. SNPs filtered out when reading the p-value file will not be included in the output no matter what (avoid storing unnecessary information)
- 3. We will remove all SNPs that are in perfect LD with each-other
  - (a) Realistically, we can not remove all SNPs that are in perfect LD with each-other without first computing the LD matrix for the whole genome. However, that will incur a large speed penalty to the algorithm. To compromise, we remove SNPs that are in perfect LD with each-other within the same window. Take into consideration of the calculation of variance, we will need to plan forward and consider SNPs within 4 windows. That's definitely overkill but that avoid problem of crazy perfect LD interactions (to be honest, I would consider those as error, yet it is easier for me to just remove them)
- 4. We do not consider a SNP ambiguous unless we need to perform flipping

#### 2 January 25, 2016

We are going to re-write SHREK starting from today. There are a number of main goal in this re-work

- 1. The whole parameter parsing
- 2. Better help messages
- 3. Allow filtering by imputation score (0-1)
- 4. Filtering of SNPs that have different reference/alternative allele with reference
- 5. Use Armadillo instead of EIGEN to improve speed

- 6. Filter all SNPs that are in perfect LD
- 7. Implement the advance variance calculation
- 8. Start documenting the codes

#### 2.1 Finished today

- 1. Finished the error message for different modes
- 2. Finished the parsing for binary trait
- 3. Finished the parsing for quantitative trait
- 4. Disabled risk prediction, should focus on the heritability estimation first

I have not perform debugging, there can be error

### 3 January 26, 2016

Continue on where I left yesterday Assumptions we made in the programme

- 1. p-value file has header
- 2. the numbers return from Command class are index (0 based)

I really want to write a more compacted code. Will try to restructure the command class

#### 3.1 Finished today

- 1. Finished refining the usage messages
- 2. Finished the basic parameter parsing (Still haven't finish the index based parameter though)

### 4 January 27, 2016

#### 4.1 Aim

We want to at least finish the region parsing and SNP class update today

#### 4.2 Finished today

- 1. Completed the update of the Command class (compiled without problem)
- 2. Completed the update of the Region class (compiled without problem)
- 3. Completed the update of the Snp class (compiled without problem)

I also haven't test run the programme, so there might be additional syntax / logic error that are not picked up at the current stage.

### 5 January 28, 2016

#### 5.1 Finished today

- 1. Finished the checking of reference panel
- 2. Found a different algorithm for popcount, which should be platform independent
- 3. Installed armadillo and OpenBLAS
  - (a) I have to add the OpenBLAS to the PATH and LD\_LIBRARY\_PATH for armadillo to detect it
- 4. Updated the makefile accordingly
  - (a) However, I have not updated the decomposition class, therefore there will be problem when we try to compile the programme
- 5. Updated the  $r^2$  calculation function such that it is using the correct sample size
  - (a) The sample size when calculating the  $r^2$  is defined as the number of samples containing both SNPs
  - (b) Because of how complicated the genotype class is, I really don't want to modify it
  - (c) The main bottleneck for the computation is in the decomposition anyway, so it should be fine

# 6 January 29, 2016

Need to at least finish the getSNP function. Must be careful with it as this is not only complicated, but also extremely important for the whole programme.

I spent whole day in annotating the document of the class to make sure I don't overlook any problem.