Software / IQI v1.0 (Java)

Introduction

IQI v1.0 (Java) has the following main functions.

- 1、Calculate the degree (IQI) of regulation between SNP and the quantitative phenotype for each single sample.
- 2. Perform the standard hypothesis testing method DQ test, to accurately detect the QTL regulation changes in disease state.

Note: The two functions are implemented by one command line.

Pre-installation

IQI v1.0 (Java) is implemented by Java. Before using it, please install <u>Java programing environment</u> (Java 8 or update) first.

Download

IQI v1.0 (Java, Command-line running)

Usage

command-line example:

java -jar IQI.jar -DQ:test -input.pheno_1 <u>phenotype_1.txt</u> -input.pheno_0 <u>phenotype_0.txt</u> -input.geno_0 <u>genotype_0.txt</u> -input.geno_1 <u>genotype_1.txt</u> -output.IQI_0 <u>IQI_0.txt</u> -output.IQI_1 <u>IQI_1.txt</u> -output.eQe_test <u>DQ_test_result.txt</u>

parameters:

- -DQ.test: Perform the DQ test;
- -input.pheno 1: Input the phenotype information of the case group;
- -input.pheno 0: Input the phenotype information of the control group;
- -input.geno 0: Input the SNP genotype data of the case group;
- -input.geno 1: Input the SNP genotype data of the control group;
- -output.IQI_0: Specify the file name of the IQI values of the SNP-phenotype pairs of all samples in the control group;
- -output.IQI_1: Specify the file name of the IQI values of the SNP-phenotype pairs of all samples in the case group;
- -output.eQe_test: Specify the file name of the results of DQ test.

Input phenotype file

please see phenotype_1.txt and phenotype_0.txt for more information

```
sample1>sample2>sample3>sample4>sample5>sample6>sample7>sample8>samp
Gene
          →7.277892592→7.131781215→4.739206133→3.293924424→5.519652169→7.263166
Gene1
Gene2 \longrightarrow -0.749986321 \longrightarrow 0.486092578 > 0.440564831 > -0.967294794 \longrightarrow -0.022983214
        \longrightarrow1.607443978>1.89183677\longrightarrow2.371212104>1.705601189>2.30635606\longrightarrow1.876922
Gene3

ightarrow 1.554325189 
angle 1.882135295 
angle 2.905419185 
angle 2.14552173 
ightarrow 2.78595273 
ightarrow 1.996336
Gene4
         \longrightarrow2.455953556>0.325004256>-1.646837322\longrightarrow-1.313823954\longrightarrow-0.90075176>
Gene5
Gene6 \longrightarrow 0.854339273 \nearrow 7.590108599 \nearrow 2.640719759 \nearrow 0.692701539 \nearrow 3.941547344 \nearrow 4.541120
Gene7\longrightarrow6.290496647>5.798508771>6.111434888>7.280799496>2.830754116>6.086360
Gene8 \longrightarrow -1.932175133 \longrightarrow -4.349342775 \longrightarrow -0.759426544 \longrightarrow -1.550274264 \longrightarrow 0.61
Gene9\longrightarrow2.079321501\Rightarrow4.661192454\Rightarrow4.72414281\longrightarrow3.634468298\Rightarrow2.392000371\Rightarrow2.543756
Gene10 \rightarrow -0.333984057 \longrightarrow 0.974384607 > 0.724378605 > 1.175572621 > 2.219008096 > -1.0
```

column 1: (e.g. Gene1) is the ID or name of a phenotype.

column 2 to the last: (e.g. 7.277892592) is the phenotype information of each sample.

Input genotype file

please see genotype 1.txt and genotype 0.txt for more information

```
SNP ALLELE - MINOR. ALLELE -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               >sample1>sample2>sample3>sample4>sample5>sample6>
	ext{rs}10001 > 	ext{G}/	ext{A} > 	ext{AA} \longrightarrow 	ext{AA}
rs10002 T/C T \rightarrow CT \rightarrow CC \rightarrow CC \rightarrow TT \rightarrow CC \rightarrow CT \rightarrow CC \rightarrow CC \rightarrow CC \rightarrow CC \rightarrow CC \rightarrow CC \rightarrow CC
| \mathtt{rs}10003 
angle \mathtt{T}/\mathtt{C} 
angle \mathtt{C} \longrightarrow \mathtt{TT} \longrightarrow \mathtt{TT} \longrightarrow \mathtt{TT} \longrightarrow \mathtt{TT} \longrightarrow \mathtt{TC} \longrightarrow \mathtt{TC} \longrightarrow \mathtt{TC} \longrightarrow \mathtt{TC} \longrightarrow \mathtt{TT} \longrightarrow \mathtt{TT} \longrightarrow \mathtt{TC} \longrightarrow 
| 	ext{rs}10004 
angle 	ext{T}/	ext{C} 	ext{T} 
ightharpoons 	ext{CT} 
ightharpoons 	ext{CC} 
ightharpoons 	ext{CC} 
ightharpoons 	ext{CT} 
ightharpoons 	ext{CC} 
ightharpoons 	ext{CT} 
ightharpoons 	ext{CC} 
ightharpoons 	ext{
| 	ext{rs}10005 
angle 	ext{T}/	ext{C} 	ext{T} 	o 	ext{CC} 	o 	ext{CT} 	o 	ext{CC} 	o 	ext{CC} 	o 	ext{CC} 	o 	ext{CC} 	o 	ext{CT} 	o 	ext{CC} 	o 	ext{CC} 	o 	ext{CC}
rs10007 A/GA-

ightarrowGG
ightarrowGA
ightarrowGA
ightarrowGA
ightarrowGA
ightarrowGG
rs10008>A/G>G-
                                                                                                                                                                                                                                                                                                                                                                                      \rightarrow AA \rightarrow AA \rightarrow AA \rightarrow AG \rightarrow GG \rightarrow AG \rightarrow AA \rightarrow AA \rightarrow AA \rightarrow AG \rightarrow AA \rightarrow AA \rightarrow AA
  rs10009>G/A>A-

ightarrow \mathsf{GG} \! 
ightarrow \! \mathsf{GA} \! \sim \! \mathsf{GA} \! \sim \! \mathsf{GA} \! \sim \! \mathsf{CA} \! \sim \! \mathsf{CA}
rs10010>G/A>A

ightarrowGG
ightarrowAA
ightarrowGG
```

column 1: (e.g. rs10001) is the name of an SNP.

column 2: (e.g. G/A) is the two alleles.

column 3: (e.g. G) is the minor allele.

column 4 to the last: (e.g. AA) is the genotype of each sample.

Output IQI file

please see IQI 1.txt and IQI 0.txt for more information

```
sample1>sample2>sample3>sample4>sample5>sample6>sample7>sample8>:
rs10010 Genel
                                                                                                                                                                                                                                                                                      \longrightarrow -0.677544 \longrightarrow -1.131207 \longrightarrow -0.659939 \longrightarrow 0.930822 \longrightarrow
rs10008 Gene1\longrightarrow-0.707659\longrightarrow1.056176\longrightarrow-0.689271\longrightarrow-0.300311\longrightarrow1.099180
rs10004 \ Gene1 \longrightarrow -0.690956 \longrightarrow 1.031248 \longrightarrow 1.997642 \longrightarrow 0.288569 \longrightarrow

ightarrow 0.974955
rs10005 Gene1\longrightarrow-0.703929\longrightarrow-1.129078\longrightarrow0.736847\longrightarrow
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 \rightarrow -0.298728 \rightarrow 1.084661
rs10006 Gene1 \longrightarrow -0.707205 \longrightarrow 1.055499 \longrightarrow -0.688829 \longrightarrow 0.302529 \longrightarrow -0.688829 \longrightarrow 0.302529 \longrightarrow -0.688829 \longrightarrow 0.302529 \longrightarrow -0.688829 \longrightarrow -0.302529 \longrightarrow -0.688829 \longrightarrow -0.68899 \longrightarrow -0.68899 \longrightarrow -0.68899 \longrightarrow -0.68899 \longrightarrow -0.68899 \longrightarrow -0.688990000000000000000000000000
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       \rightarrow-1.013977\longrightarrow0
rs10007 \ Gene1 \longrightarrow -0.695570 \longrightarrow -1.142820 \longrightarrow 2.169127 \longrightarrow -0.295181 \longrightarrow -0.997295 -0.0007
rs10002 Gene1 \longrightarrow 1.507467 \longrightarrow 1.010818 \longrightarrow -0.659670 \longrightarrow -0.287414 \longrightarrow 5.293809 \longrightarrow -0.659670 \longrightarrow -0.287414 \longrightarrow -0.28741414 \longrightarrow
rs10003 Gene1 \longrightarrow -0.683192 \longrightarrow -1.044428 \longrightarrow -0.665439 \longrightarrow -0.289928 \longrightarrow -0.979547
rs10010 Gene2\longrightarrow2.374625\longrightarrow1.285338\longrightarrow
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        \rightarrow -1.690329 \rightarrow -0.352866 \rightarrow 0.491422
rs10008 Gene2\longrightarrow2.480169\longrightarrow-1.200084\longrightarrow-1.765458\longrightarrow0.113845\longrightarrow
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 \rightarrow0.497062
rs10009 Gene2 \longrightarrow 2.615788 \longrightarrow -1.265706 \longrightarrow 1.847159 \longrightarrow 0.120070 \longrightarrow 0.480060 -1.265706 \longrightarrow 0.480060 -1.265706 \longrightarrow 0.120070 \longrightarrow 0.480060 -1.265700 \longrightarrow 0.120070 \longrightarrow 0.1200700000 \longrightarrow 0.120070 \longrightarrow 0.120070 \longrightarrow 0.120070 \longrightarrow 0.120070 \longrightarrow 0.120070 \longrightarrow 0.1
rs10004 Gene2 \longrightarrow 2.421631 \longrightarrow -1.171759 \longrightarrow 5.116644 \longrightarrow -0.109394 \longrightarrow 0.440886 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.109394 -0.10940 -0.10940 -0.10940 -0.10940 -0.10940 -0.10940 -0.10940 -0.10940 -0.10940 -0.10940 -0.10940 -0.10940 -0.109
rs10005 Gene2 \longrightarrow 2.467096 \longrightarrow 1.282919 \longrightarrow 1.887317 \longrightarrow 0.113245 \longrightarrow 0.490496 - 0.490496 \longrightarrow 0.490496 - 0.490496 \longrightarrow 0.490496 \longrightarrow 0.490496 - 0.490496 \longrightarrow 0.49
rs10006 Gene2 \longrightarrow 2.478579 \longrightarrow -1.199315 \longrightarrow -1.764326 \longrightarrow -0.114686 \longrightarrow -0.458532 -0.0006 Gene2 \longrightarrow -0.458579 \longrightarrow -0.0006 Gene2 \longrightarrow -0.0006 Gene2 \longrightarrow -0.00006 Gene2 \longrightarrow -0.0000
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        \rightarrow 1.298533 \longrightarrow 5.555876 \longrightarrow 0.111900 -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            \rightarrow -0.450989
rs10007 Gene2 -- 2.437801 --
\rightarrow -0.466068
rs10002 Gene2 \longrightarrow -5.283299 \longrightarrow -1.148546 \longrightarrow -1.689639 \longrightarrow 0.108956 \longrightarrow -1.689639 \longrightarrow -1.68969 \longrightarrow -1.6899 \longrightarrow -1.6899 \longrightarrow -1.68969 \longrightarrow -1.6899 
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              →2.393922-
```

column 1: (e.g. rs10010_Gene1) is the name of the SNP-phenotype pair.

column 2 to the last: (e.g. -0.677544) is IQI of each sample.

DQ test results

please see DQ test result.txt for more information

```
\rightarrow change_type\Rightarrow beta_1\rightarrowSE_1-
                                                                                                                                                                                                                                                                                                                                                                                                                           \Rightarrowp_1\Rightarrowbeta_0\RightarrowSE_0\longrightarrowp_0\Rightarrowdx1_x0\RightarrowSx1_x0\Rightarrowdf\RightarroweQe_mean_1\RightarroweQe_mean_0\Rightarrowsample_num_
 rs10001 \; Gene1 \longrightarrow GOE, \cdot -/0 \longrightarrow -2.037791 \longrightarrow 0.073848 \longrightarrow 2.1228E - 102 > 0.029417 \longrightarrow 0.077993 \longrightarrow 0.077993 \longrightarrow 0.07993 \longrightarrow 0
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         0.706205-
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  →-2.067208-
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              \rightarrow0.16143
 rs10001 Gene2 \longrightarrow LOE, \cdot0/+-
                                                                                                                                                                                                                                                                              \rightarrow -0.076519 \rightarrow 0.116169 \rightarrow 0.510398 \rightarrow 0.232988 \rightarrow 0.080879 \rightarrow 0.004139 \rightarrow
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      \rightarrow -0.309507 \rightarrow 0.14015
 rs10002\_Gene2 \longrightarrow GOE, \cdot +/O \longrightarrow 1.969310 -
                                                                                                                                                                                                                                                                                                                                                                                                                         \rightarrow 0.074698 \longrightarrow 1.5567E-96 \rightarrow 0.049207 -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          \rightarrow 0 \cdot 085511 -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        →0.565248-
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  \rightarrow1.920103-
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      0.15184
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      rs10003 Gene3\longrightarrowLOE, \cdotO/--
                                                                                                                                                                                                                                                                          \rightarrow 0.027906
                                                                                                                                                                                                                                                                                                                                                                                                             \rightarrow 0.065411 \longrightarrow 0.669837 \longrightarrow -1.519056 \longrightarrow 0.063573 \longrightarrow
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      0.13111
 rs10004\_Gene4 \longrightarrow LOE, \cdot O/+
                                                                                                                                                                                                                                                                                        →0.023216-
                                                                                                                                                                                                                                                                                                                                                                                                                      \rightarrow 0.067263 \longrightarrow 0.730122 -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       \rightarrow 0.901411 \longrightarrow 0.066669 -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      \rightarrow1.054E-35\longrightarrow-0.878195
 rs10005 Gene5 \longrightarrow ROE, \cdot+/--
                                                                                                                                                                                                                                                                                        →1.825745-
                                                                                                                                                                                                                                                                                                                                                                                                                        \rightarrow 0.065499 \longrightarrow 9.9926E-104 \rightarrow -1.736576 \longrightarrow 0.072568 
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        \rightarrow 8.2097E-85 \rightarrow 3.562321 -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      0.17528
  \texttt{rs}10004\_\texttt{Gene}6 \longrightarrow \texttt{LOE}, \cdot \texttt{O}/+ \longrightarrow -0.075922 \longrightarrow 0.111471 \longrightarrow 0.496129 \longrightarrow 0.243735 \longrightarrow 0.108733 \longrightarrow 0.025428 \longrightarrow -0.319656 \longrightarrow 0.15770 
 rs10006_Gene6\longrightarrowROE, \cdot-/+-
                                                                                                                                                                                                                                                                                        \rightarrow -1.965373 \longrightarrow 0.070023 \longrightarrow 1.2194E-104>2.000344 -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          →0.065844-
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        →1.8389E-115>-3.965717-
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      0.18904
 rs10007_Gene7\longrightarrowSOE, \cdot--/-\longrightarrow-1.947513
                                                                                                                                                                                                                                                                                                                                                                                                     \longrightarrow 0.068132 \longrightarrow 4.4395E-107>-0.945750 \longrightarrow 0.066752 -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    \rightarrow1.5783E-38\rightarrow-1.001762\longrightarrow
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      0.1618
rs10008_Gene8 -> SOE, ++/+ -> 1.971503 -
                                                                                                                                                                                                                                                                                                                                                                                                                 \rightarrow 0.067759 \longrightarrow 1.764E-109 \rightarrow 1.081841 \longrightarrow 0.068378 \longrightarrow 5.6507E-46 \rightarrow 0.889661 \longrightarrow
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      0.16522
 rs10005_Gene8\longrightarrowGOE, \cdot+/0\longrightarrow0.288813-
                                                                                                                                                                                                                                                                                                                                                                                                                        \rightarrow 0.103515 \longrightarrow 0.005472 \longrightarrow -0.092004 \longrightarrow 0.083323 \longrightarrow
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    →0.270047-
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                \rightarrow 0.380818 -
                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      0.13388
 rs10009\_Gene9 \longrightarrow \texttt{WOE}, \cdot -/-- \longrightarrow -0.910509 \longrightarrow 0.064703 \longrightarrow 4.1685E - 38 \rightarrow -1.953974 \longrightarrow 0.065978 \longrightarrow 6.5642E - 112 \times 1.043465 \longrightarrow 0.15671 \longrightarrow 0.15671 \longrightarrow 0.064703 \longrightarrow 0.064703 \longrightarrow 0.064703 \longrightarrow 0.064703 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.064703 \longrightarrow 0.064703 \longrightarrow 0.064703 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.064703 \longrightarrow 0.064703 \longrightarrow 0.064703 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.065978 \longrightarrow 0.064703 \longrightarrow 0.064703 \longrightarrow 0.064703 \longrightarrow 0.065978 \longrightarrow 0.065979 \longrightarrow 
 \texttt{rs}10010\_\texttt{Gene}10 \rightarrow \texttt{WOE}, \cdot + / + + \longrightarrow 0.981220 \longrightarrow 0.070475 \longrightarrow 1.8835E - 37 \rightarrow 2.020827 \longrightarrow 0.068436 \longrightarrow 1.6678E - 111 \rangle - 1.039607 \longrightarrow 0.17364 \longrightarrow 0.068436 \longrightarrow
```

```
It contains 18 columns:
  SNP Gene -> the name of SNP-phenotype pair;
  change type -> change type of QTL regulation;
  beta_1 -> the traditional regression based QTL analysis results of the case group;
  SE_1 -> the standard error in the case group;
  p \bar{1} -> the p value in the case group;
  beta 0 -> the traditional regression based QTL analysis results of the control group;
  SE 0 -> the standard error in the control group;
  p \ \bar{0} -> the p value in the control group;
  dx1 x0 -> the mean difference between case and control groups;
  Sx1 x0 -> the standard error between case and control groups;
  df -> the degree of freedom;
  eQe mean 1 -> the average IQI in the case group;
  eQe mean 0 -> the average IQI in the control group;
  sample num 1 -> the sample size of the case group;
  sample num 0 -> the sample size of the control group;
  fold_change -> the fold change value;
  t -> the DQ statistics;
  tp -> the p value.
```

The simulated data and results were offered to test: **Download**