

# **User Manual for**

# **QTL.gCIMapping**

**QTL genome-wide Composite Interval Mapping**

**(version 2.0)**

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**Disclaimer:** While extensive testing has been performed by Yuan-Ming Zhang's Lab (Statistical Genomics Lab) at Huazhong Agricultural University, the results are, in general, reliable, correct or appropriate. However, results are not guaranteed for any specific datasets. We strongly recommend that users validate the GCIM results with other software packages, such as [Windows QTL Cartographer V2.5\\_011](https://brcwebportal.cos.ncsu.edu/qtlcart/WQTLCart.htm) (<https://brcwebportal.cos.ncsu.edu/qtlcart/WQTLCart.htm>) and [QTL IciMapping V4.1](http://www.isbreeding.net/software/?type=detail&id=18) (<http://www.isbreeding.net/software/?type=detail&id=18>).

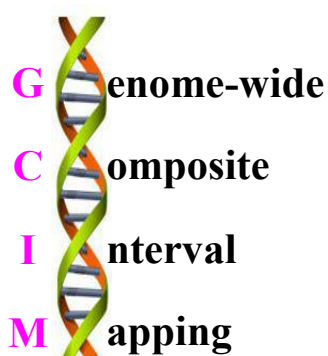
**Download website:**

<https://cran.r-project.org/web/packages/QTL.gCIMapping/index.html>

**References**

1. Wang Shi-Bo, Wen Yang-Jun, Ren Wen-Long, Ni Yuan-Li, Zhang Jin, Feng Jian-Ying, Zhang Yuan-Ming\*. Mapping small-effect and linked quantitative trait loci for complex traits in backcross or DH populations via a multi-locus GWAS methodology. *Scientific Reports* 2016, 6: 29951.
2. Wen Yang-Jun, Zhang Ya-Wen, Zhang Jin, Feng Jian-Ying, Jim M. Dunwell, Zhang Yuan-Ming\*. An efficient multi-locus mixed model framework for the detection of small and linked QTLs in F<sub>2</sub>. Submitted

## Quantitative Trait Loci



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## INTRODUCTION

### 1.1 Why GCIM?

**QTL.gCIMapping**(**QTL** **G**enome-wide **C**omposite **I**nterval **M**apping **G**raphical **U**ser **I**nterface) is an R package for multi-QTL mapping of quantitative traits in bi-parental segregation populations.

QTL.gCIMapping v1.0 is able to work on the popular platforms, like Windows, Linux (desktop) and MacOS.

### 1.2 Getting started

QTL.gCIMapping is a package that runs in the R software environment, which can be freely downloaded from <https://cran.r-project.org/web/packages/QTL.gCIMapping/index.html>, or request from the maintainer, Dr Yuan-Ming Zhang at Huazhong Agricultural University ([soyzzhang@mail.hzau.edu.cn](mailto:soyzzhang@mail.hzau.edu.cn) or [soyzzhang@hotmail.com](mailto:soyzzhang@hotmail.com)).

#### 1.2.1 One-Click installation

Within R environment, the QTL.gCIMapping software can be installed directly using the below command:

```
install.packages(pkgs="QTL.gCIMapping")
```

#### 1.2.2 Step-by-step installation

##### 1.2.2.1 Install the add-on packages

**Online installation** Within R environment on the internet, the QTL.gCIMapping package can be installed online, using the below command:

```
install.packages(pkgs=c("shiny","qtl","doParallel","foreach","iterators","openxlsx","MASS","stringr","parcor","data.table"))
```

**Offline installation** Users should download the below 35 packages from CRAN, github (<https://github.com/>), or google search:

```
"cmprsk","corpcor","data.table","digest","doParallel","Epi","etm","fdrtool","foreach","GeneNet","glmnet","htmltools","httpuv","iterators","jsonlite","longitudinal","magrittr","MASS","mime","numDeriv","openxlsx","parcor","plyr","ppls","qtl","R6","Rcpp","shiny","sourcetools","stringi","stringr","testthat","utf8","xtable","zoo"
```

Then, install them offline (under the R environment, select all the 35 packages and

install them offline).

### 1.2.2.2 Install QTL.gCIMapping

Open R GUI, select **"Packages"**—**"Install package(s) from local files..."** and then find the QTL.gCIMapping package which you have downloaded on your desktop.

Within R environment, launch the QTL.gCIMapping by command:

```
library(QTL.gCIMapping)
```

**User Manual file** Users can decompress the QTL.gCIMapping package and find the User Manual file (name: **Instruction.pdf**) in the folder of ".../QTL.gCIMapping/inst/doc".

## 2. Dataset format

**GCIM format for Dataset** The first three columns, named **"marker"**, **"chr"** and **"pos"**, stand for marker name, chromosome and marker position (cM) on the chromosome, respectively. Among the remaining columns, each column lists all the genotypes of one individual while the first row shows the individual name. For the genotypes of each marker, the coding criteria are shown as [Table 1](#). The phenotype and covariate information are followed the marker genotypes, and each covariate or trait are listed on one row. On each row, the first column is empty followed by **"trait1"**, "real trait name", and "phenotypic values for all the individuals". If multiple traits exist, more rows will be added. If covariates exist, all the information for the covariates list after the trait information. The format is seen in [Table 1](#). If there is no covariate, users should delete the last row in [Table 2](#).

**Table 1. Coding criteria for GCIM format**

Marker genotype	Code	Meaning
AA	A	Homozygous genotype (P <sub>1</sub> )
Aa	H	Heterozygous genotype (F <sub>1</sub> )
aa	B	Homozygous genotype (P <sub>2</sub> )
Not AA (Aa + aa)	C	Dominance to P <sub>2</sub>
Not aa (AA + Aa)	D	Dominance to P <sub>1</sub>
Missing	-	Missing or unclear genotype

**Table 2. The GCIM format of the dataset**

marker	chr	pos	DH6-10	DH6-101	DH6-102
RGA3(1)	1	0	B	-	B
wPt-6358	1	3.034	B	-	-
Hplc2	1	8.8291	A	A	B
wPt-9752	1	10.1452	A	-	-
abc156a	1	41.3408	A	A	B
:	:	:	:	:	:
gwm437	21	162.5218	A	B	-
gwm121	21	180.2878	A	B	-
wmc157	21	197.9196	A	B	A
*stm1actc	21	200.4216	-	-	-
	trait1	T19	75.33	105	96.33
	trait2	T191	74	105.68	97.16
	trait3	T192	75.37	104.67	95.55
	Covar1	CovarName	A	B	B

**ICIM format for Dataset** If users have the dataset files for QTL IciMapping format, these files are also available in our software. Details can be seen in the folder of “.../QTL.gCIMapping/inst/extdata”, i.e., [WheatDH\\_QTLIciMapping\\_Format.xlsx](#).

**WinQTLCart format for Dataset** If users have the dataset file for WinQTLCart format, its file is also available in our software. Details can be seen in the folder of “.../QTL.gCIMapping/inst/extdata”, i.e., [env1-jun3\\_WinQTLCart\\_Format.mcd](#).

**The dataset fileICIMcov format** If users select ICIM format and the covariate exists in the dataset, it needs to input a covariate file. In the file, the first column indicates individual name and the second column is the covariate information ([Table 3](#)). In [Table 3](#), the covariate values are A, B and C.

**Table 3. The covariate file format**

Individual ID	Covariate
DH6-10	A
DH6-101	A
DH6-102	A
DH6-104	A
DH6-164	B
DH6-165	B
DH6-166	B
DH6-170	B
DH7-124	C
DH7-125	C

### 3. Operation process

#### 3.1 The graphical interface of QTL.gCIMapping

GCIM
Start

### QTL.gCIMapping (QTL genome-wide Composite Interval Mapping)

#### Coding criteria

Genotype	Code	Meaning
AA	A	Homozygous genotype (P1)
Aa	H	Heterozygous genotype (F1)
aa	B	Homozygous genotype (P2)
AA+Aa/(not aa)	D	Dominance to P1
Aa+aa/(not AA)	C	Dominance to P2
Missing	-	Missing or unclear genotype

#### Dataset example

marker	chr	pos	DH6.10	DH6.101	DH6.102
RG43(1)	1	0	B	-	B
wf4-6358	1	3.034	B	-	-
Hpk2	1	8.8201	A	A	B
---	---	---	---	---	---
gwm437	21	162.5218	A	B	-
gwm121	21	160.2878	A	B	-
wmc157	21	197.9196	A	B	A
trait1	T19	75.33	105	96.33	
trait2	T191	74	105.68	97.16	
Covar	Covariate		A	B	B

#### Reference

1. Wang Shi-Bo, Wen Yang-Jun, Ren Wen-Long, Ni Yuan-Li, Zhang Jin, Feng Jian-Ying, Zhang Yuan-Ming\*. Mapping small-effect and linked quantitative trait loci for complex traits in backcross or DH populations via a multi-locus GWAS methodology. Scientific Reports 2016;6:29951.
2. Wen Yang-Jun, Zhang Ya-Wen, Zhang Jin, Feng Jian-Ying, Jim M. Dunwell, Zhang Yuan-Ming\*. An efficient multi-locus mixed model framework for the detection of small and linked QTLs in F2. Submitted

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Maintainer: Zhang Yuan-Ming (soy Zhang at mail.hzau.edu.cn)  
QTL.gCIMapping version 1.0, Released April 2018

**Figure 1. The Graphical User Interface of QTL.gCIMapping**

#### 3.2 Input dataset

Users must upload the dataset files with three formats (Figs 2 to 4). If users select the QTL IciMapping format and the covariate exists in the dataset, users should upload the covariate matrix (Fig 5).

QTL.gCIMapping

Please select data format

☒ GCIM

☐ WinQTLcart

☐ QTLciMapping

Input dataset

Browse... GCIM\_Format\_DH.csv

Upload file

Show dataset:

Genotype

Parameter Settings

Figure

User manual

Dataset

Parameter Settings

Figure

Show 25 entries

Search

marker	DH6-10	DH6-101	DH6-102	DH6-104	DH6-105	DH6-108	DH6-11	DH6-111	DH6-112	DH6-114	DH6-119	DH6-124	DH6-125	DH6-128	DH6-129
RG43(1)	B	-	B	A	B	B	A	A	A	-	B	B	B	A	B
WP6-6308	B	-	-	-	-	B	A	A	A	-	B	-	-	A	-
hpk2	A	A	B	A	B	B	A	A	A	B	B	B	B	A	B
WP6-6752	A	-	-	-	-	-	A	A	A	-	B	-	-	A	B
adx156a	A	A	B	A	B	B	B	B	A	B	-	B	B	A	B
RG436b(2)	A	-	B	A	-	B	B	B	A	-	B	B	B	A	B
bc986	B	A	B	A	B	B	B	B	A	B	A	B	B	A	B
wmx24	B	A	B	A	B	B	B	B	A	B	A	B	B	A	B
kou59k	B	A	B	A	B	B	B	B	A	B	A	B	B	A	B
WP6-2436	B	-	-	-	-	B	B	B	B	-	A	B	-	B	-
WP6-4886	B	-	-	-	-	B	B	B	B	-	A	B	-	B	B
wmx120	B	A	B	A	B	B	B	B	B	A	A	B	B	B	B
cds105	B	A	B	A	B	B	B	B	B	A	A	B	B	B	B
WP6-6074	B	-	-	-	-	B	B	B	B	-	A	B	-	B	-
GluA1	B	A	B	A	B	A	B	B	B	A	A	B	B	B	B
bc980b	B	A	A	B	B	A	B	A	B	B	A	B	B	B	A

Fig 2. Dataset GCIM format

QTL.gCIMapping

Please select data format

☒ WinQTLcart

☐ GCIM

☐ QTLciMapping

Input dataset

Browse... winqtlcart\_WinQTLcart\_Format.ncs

Upload file

Show dataset:

Genotype

Parameter Settings

Figure

User manual

Dataset

Parameter Settings

Figure

Show 25 entries

Search

marker	DH6-10	DH6-101	DH6-102	DH6-104	DH6-105	DH6-108	DH6-11	DH6-111	DH6-112	DH6-114	DH6-119	DH6-124	DH6-125	DH6-128	DH6-129
RG43(1)	B	-	B	A	B	B	A	A	A	-	B	B	B	A	B
WP6-6308	B	-	-	-	-	B	A	A	A	-	B	-	-	A	-
hpk2	A	A	B	A	B	B	A	A	A	B	B	B	B	A	B
WP6-6752	A	-	-	-	-	-	A	A	A	-	B	-	-	A	B
adx156a	A	A	B	A	B	B	B	B	A	B	-	B	B	A	B
RG436b(2)	A	-	B	A	-	B	B	B	A	-	B	B	B	A	B
bc986	B	A	B	A	B	B	B	B	A	B	A	B	B	A	B
wmx24	B	A	B	A	B	B	B	B	A	B	A	B	B	A	B
kou59k	B	A	B	A	B	B	B	B	A	B	A	B	B	A	B
WP6-2436	B	-	-	-	-	B	B	B	B	-	A	B	-	B	-
WP6-4886	B	-	-	-	-	B	B	B	B	-	A	B	-	B	B
wmx120	B	A	B	A	B	B	B	B	B	A	A	B	B	B	B
cds105	B	A	B	A	B	B	B	B	B	A	A	B	B	B	B
WP6-6074	B	-	-	-	-	B	B	B	B	-	A	B	-	B	-
GluA1	B	A	B	A	B	A	B	B	B	A	A	B	B	B	B
bc980b	B	A	A	B	B	A	B	A	B	B	A	B	B	B	A

Fig 3. Dataset WinQTLcart format

QTL.gCIMapping

Please select data format

☐ GCIM

☒ QTLciMapping

☐ WinQTLcart

Input dataset

Browse... winqtlcart\_QTLciMapping\_Format.ncs

Upload file

Input covariate file

Browse... No file selected

Show dataset:

Genotype

Parameter Settings

Figure

User manual

Dataset

Parameter Settings

Figure

Show 25 entries

Search

marker	X1	X2	X3	X4	X5	X6	X7	X8	X9	X10	X11	X12	X13	X14	X15	X16	X17	X18	X19	X20	X21
RG43(1)	0	-1	0	2	0	0	2	2	2	-1	0	0	0	2	0	2	2	2	0	2	
WP6-6308	0	-1	-1	-1	-1	0	2	2	2	-1	0	-1	-1	2	-1	-1	2	2	0	2	
hpk2	2	2	0	2	0	0	2	2	2	0	0	0	0	2	0	2	2	2	0	2	
WP6-6752	2	-1	-1	-1	-1	2	2	2	2	-1	0	-1	-1	2	0	-1	2	2	-1	2	
adx156a	2	2	0	2	0	0	0	0	2	0	-1	0	0	2	0	2	2	0	0	0	
RG436b(2)	2	-1	0	2	-1	0	0	2	-1	0	0	0	2	0	2	2	0	0	0	0	
bc986	0	2	0	2	0	0	0	2	0	2	0	0	2	0	2	0	2	0	2	0	
wmx24	0	2	0	2	0	0	0	2	0	2	0	0	2	0	2	0	2	0	2	0	
kou59k	0	2	0	2	0	0	0	2	0	2	0	0	2	0	2	0	2	0	2	0	
WP6-2436	0	-1	-1	-1	-1	0	0	0	0	-1	2	0	-1	0	-1	2	0	2	0	0	
WP6-4886	0	-1	-1	-1	-1	0	0	0	0	-1	2	0	-1	0	0	-1	2	0	2	0	
wmx120	0	2	0	2	0	0	0	0	2	2	0	0	0	0	2	2	0	2	0	0	
cds105	0	2	0	2	0	0	0	0	2	2	0	0	0	0	2	2	0	2	0	0	
WP6-6074	0	-1	-1	-1	-1	0	0	0	-1	2	0	-1	0	-1	-1	-1	0	2	0	0	
GluA1	0	2	0	2	0	2	0	0	2	2	0	0	0	0	2	0	0	2	0	0	
bc980b	0	2	2	0	0	2	0	2	0	0	2	0	0	0	2	2	0	0	0	2	
WP6-2	0	2	2	0	0	2	0	2	0	-1	2	0	2	0	2	2	0	-1	2	0	

Fig 4. Dataset QTLciMapping format

QTL.gCIMapping

Please select data format

☐ QCM

☐ WinQTLcart

☒ QTL IciMapping

Input dataset

Browse... WheatDH\_QTL IciMapping\_Format.txt

Upload complete

Input covariate file

Browse... GCRL\_Cov.csv

Upload complete

Show dataset:

Covariate

Parameter Settings

Figure

Show 25 entries

V1	V2
Individual ID	Covariate
DH6-10	A
DH6-101	A
DH6-102	A
DH6-104	A
DH6-105	A
DH6-108	A
DH6-11	A
DH6-111	A
DH6-112	A
DH6-114	A
DH6-118	A
DH6-124	A
DH6-125	A
DH6-128	A
DH6-129	A
DH6-13	A

Parameter Settings

Figure

User manual

**Fig 5. Covariate input in the QTL IciMapping dataset format**

### 3.3 Parameter settings (Fig 6)

**Select population:** BC1 ( $F1 \times P1$ ), BC2 ( $F1 \times P2$ ), DH, RIL, F2.

**Select model:** Random or Fixed model for QTL effects.

**Walk Speed for Genome-wide Scanning (cM):** Set walk speed for Genome-wide Scanning (centi-Morgan, cM), for example, 1 cM.

**Critical LOD score:** Critical LOD scores for significant QTL.

**Likelihood function:** This parameter is only for F2 population, including restricted maximum likelihood (REML) and maximum likelihood (ML).

**Completing CIM in one neighborhood:** This parameter is only for F<sub>2</sub> population. In the first running, please set "FALSE". If the other software detects only one QTL in a neighborhood but the current software finds two linked QTLs (one with additive effect and another with dominant effect) in the neighborhood, please set "TRUE".

**Draw plot or not:** This parameter setup includes FALSE and TRUE. "FALSE" indicates no figure output, and "TRUE" indicates the output of QTL mapping curve, for example, the LOD score [or  $-\log_{10}(P\text{-value})$ ] curve against genome position.

**Resolution of plot:** Low or High: the low or high resolution for the figure file.

**Plot format:** Users can download the picture for different file formats: \*.jpeg, \*.png,



\*.tiff and \*.pdf.

**Select trait ID:** “2:2” indicates the analyses from the second trait, and “2:4” indicates the analyses from the second to fourth traits.

**Save path:** The result will be written to the path in your computer.

The screenshot shows the QTL.gCIMapping software interface. On the left, under 'Please select data format', 'GCM' is selected. Below it, 'Input dataset' has a 'Browse...' button and 'No file selected'. 'Show dataset:' is set to 'Genotype'. At the bottom left are buttons for 'Parameter Settings', 'Figure' (highlighted with a red box), and 'User manual'. The main area has three tabs: 'Dataset', 'Parameter Settings', and 'Figure'. Under 'Please select Population', 'DH' is selected. 'Critical LOD score' is 2.5. 'Draw plot or not' is 'TRUE'. 'Select trait ID' has 'From' as 1 and 'To' as 1. Under 'Please select Model', 'Random model' is selected. 'Likelihood function (only for F2):' has 'REML' selected. 'Resolution of plot' has 'Low' selected. On the right, 'Walk Speed for Genome-wide Scanning (cM):' is 1. 'Completing CIM in the neighborhood (only for F2)' has 'TRUE' selected. 'Plot format' has '.png' selected. 'Save path' is 'C:/Users/Administrator/Desktop/'. A 'Run' button is at the bottom center.

Fig 6. Parameter setting in the mapping of QTL for quantitative traits

### 3.4 Run the software

This screenshot shows the same QTL.gCIMapping interface as Figure 6, but with additional changes. In the 'Input dataset' section, 'GCM\_Format\_DH.csv' is selected and 'Upload complete' is shown. The 'Run' button at the bottom center is highlighted with a red box. On the right side, a 'Running in progress' status bar is visible, also highlighted with a red box.

Fig 7. Run the software QTL.gCIMapping

### 3.5 Re-draw the plot according to your own requirement

When users finish the running, users get the resultforplot.xlsx file. With this file information, users may redraw the curve figure {LOD score or  $-\log_{10}(P\text{-value})$  }. With this Figure module, users may set all the figure parameters (Fig 8), including

**Legend and tick marks:** the size of the words in axis.

**LOD line size:** the size of the LOD line, the larger the coarse.

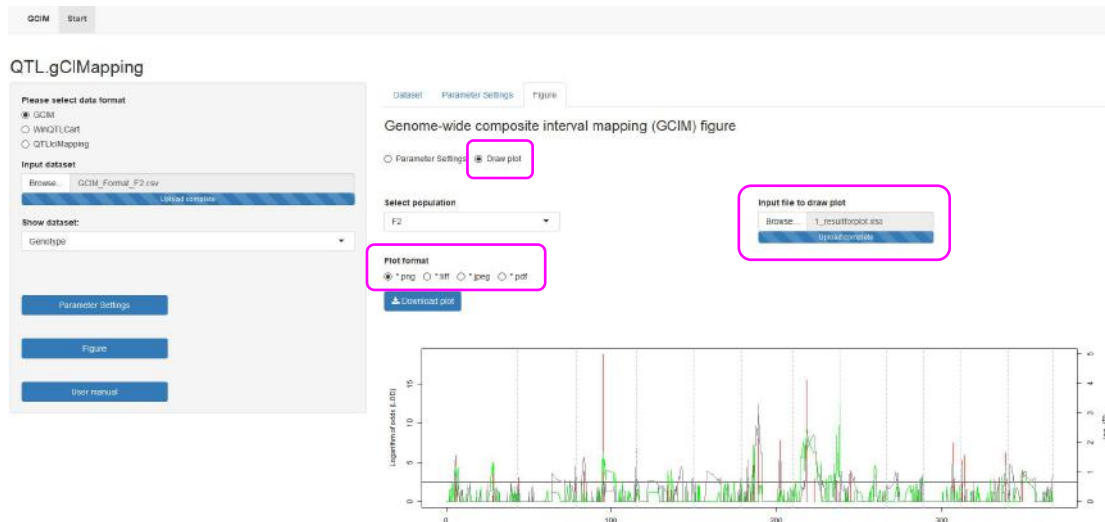
**Size for  $-\log_{10}(P\text{-value})$  curve:** the size of  $-\log_{10}(P\text{-value})$  curve, the larger the coarse.

**Margin space:** the space between the figure and the margin of the paper.

**Critical LOD score:** The critical LOD score for significant QTL.

Before saving this Figure, please set the related parameters: **width** and **height** [with the unit of pixel (px)], **word resolution** [with the unit of 1/72 inch, being pixels per inch (ppi)], and **figure resolution** [with the unit of pixels per inch (ppi)]. Users may set the colors for the LOD line color and  $-\log_{10}(P\text{-value})$  curve, with a drop-down option. Use Download plot button to choose a path and to save the Figure, with four frequently used image formats: \*.png, \*.tiff, \*.jpeg and \*.pdf (Fig 9).

**Figure 8. Parameter settings**



**Fig 9. How to draw the QTL mapping figure**

## 4. Result

For BC1, BC2, DH and RIL populations, the **Results** file has ten columns, as shown below.

**Trait:** The trait name analyzed.

**Chr:** Chromosome, represented by an integer number.

**Position (cM):** The QTL position (cM) on the chromosome.

**Additive Effect:** Additive effect for significant QTL.

**LOD:** LOD score for significant QTL.

**Left\_Marker:** Left flanking marker name for significant QTL.

**Right\_Marker:** Right flanking marker name for significant QTL.

**Var\_Genet:** Genetic variance for each significant QTL.

**$r^2$  (%):** Proportion of phenotypic variance explained by single QTL.

**Var\_Error:** residual variance for the full model.

**Var\_Phen (total):** Phenotypic variance in the analyzed population.

For F<sub>2</sub> population, the **Results** file has eleven columns. Trait, Chr, Position (cM), Left\_Marker, Right\_Marker, Var\_Genet, LOD,  $r^2$  (%), Var\_Error and Var\_phen are same as those in the above populations. The different columns are as follows.

**Effect.a** and **Effect.d:** Additive and dominant effects for significant QTL.