User Manual for

QTL.gCIMapping.GUI

QTL genome-wide Composite Interval Mapping GUI

(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) and **QTL IciMapping V4.1** (http://www.isbreeding.net/software/?type=detail&id=18).

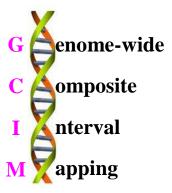
Download website:

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

References

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- Zhang Ya-Wen, Jim M. Dunwell, Zhang Yuan-Ming*. QTL.gCIMapping.GUI v2.0: An R software for detecting small-effect and linked QTLs for quantitative traits in bi-parental segregation populations. *Computational and Structural Biotechnology Journal*, in revision

Quantitative Trait Loci





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INTRODUCTION

1.1 Why GCIM?

QTL.gCIMapping.GUI v2.0 (QTL Genome-wide Composite Interval Mapping Graphical User Interface) is an R package for multi-QTL mapping of quantitative traits in bi-parental segregation populations, including backcross (BC), doubled haploid (DH) lines, recombinant inbred lines (RIL), F₂, and immortalized F₂ (IF₂). QTL.gCIMapping.GUI v2.0 works well on the R environment on Windows, Linux (desktop) and MacOS.

1.2 Getting started

The software package QTL.gCIMapping.GUI v2.0 can be freely downloaded from https://cran.r-project.org/web/packages/QTL.gCIMapping.GUI/index.html, or request from the maintainer, Dr Yuan-Ming Zhang at Crop Information Center, College of Plant Science & Technology, Huazhong Agri Univ (soyzhang@mail.hzau.edu.cn).

1.2.1 One-Click online installation

On R environment and network connection, the command install.packages(pkgs="QTL.gCIMapping.GUI") is used to directly install the software package QTL.gCIMapping.GUI v2.0.

1.2.2 Step-by-step offline installation

1.2.2.1 Install the add-on packages

First, users download thirty-nine R packages, including

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"cmprsk", "corpcor", "data.table", "digest", "doParallel", "Epi", "etm", "fdrtool", "foreach", "GeneNet", "glmnet", "htmltools", "httpuv", "iterators", "jsonlite", "later", "longitudinal", "magrittr", "MASS", "mime", "numDeriv", "openxlsx", "parcor", "plyr", "ppls", "promises", "QTL.gCIMapping", "qtl", "R6", "Rcpp", "shiny", "sourcetools", "stringi", "stringr", "testt hat", "utf8", "xtable", "zip", "zoo",
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from CRAN, github (https://github.com/), or google search.

On the R environment, then, users select all the 39 packages and install them offline.

1.2.2.2 Install QTL.gCIMapping.GUI v2.0

On R GUI environment, users first select "Packages"—"Install package(s) from local files...", then find the software package QTL.gCIMapping.GUI v2.0 on user's desktop computer or mobile device, and launch QTL.gCIMapping.GUI v2.0.

1.2.3 Run QTL.gCIMapping.GUI v2.0

Once the software package QTL.gCIMapping.GUI v2.0 is installed, users may run it using two commands:

library(QTL.gCIMapping.GUI)

QTL.gCIMapping.GUI()

If users re-use the software QTL.gCIMapping.GUI v2.0, users use the above two commands as well.

User Manual file Users can decompress the QTL.gCIMapping.GUI package and find the User Manual file (name: **Instruction.pdf**) in the folder of ".../QTL.gCIMapping.GUI/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 or line, while the first row shows the name of the individual or line. For the genotypes of each marker, the coding criteria are shown as Table 1.

Table 1. Coding criteria for GCIM format

Marker genotype	Code	Meaning
AA	A	Homozygous genotype (P ₁)
Aa	Н	Heterozygous genotype (F ₁)
aa	В	Homozygous genotype (P ₂)
Not AA (Aa + aa)	С	Dominance to P ₂
Not aa (AA + Aa)	D	Dominance to P ₁
Missing	-	Missing or unclear genotype

The genotypic, phenotypic and covariate datasets are located on the upper, middle, lower sections, and each covariate or trait is presented on one row. On each row, the

first column is empty followed by "**trait1**", "real trait name", and "phenotypic values for all the individuals or lines". If there are multiple traits, these traits occupy multiple lines. If there are covariates, the content lies below the trait dataset. The format is seen in Table 2. If there is no covariate, users should delete the last row in Table 2.

Table 2. The GCIM format of the dataset

marker	chr	pos	DH6-10	DH6-101	DH6-102
RGA3(1)	1	0	В	-	В
wPt-6358	1	3.034	В	-	-
Hplc2	1	8.8291	A	A	В
wPt-9752	1	10.1452	A	-	-
abc156a	1	41.3408	A	A	В
:	:	:	:	:	:
gwm437	21	162.5218	A	В	-
gwm121	21	180.2878	A	В	-
wmc157	21	197.9196	A	В	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	В	В

The format of ICIM dataset If users have the QTL IciMapping dataset, these files are also available in our software. Details can be found in the folder of ".../QTL.gCIMapping.GUI/inst/extdata", i.e., WheatDH_QTLIciMapping_Format.xlsx.

The format of WinQTLCart dataset If users have the WinQTLCart dataset, its file is also available in our software. Details can be found in the folder of ".../QTL.gCIMapping.GUI/inst/extdata", i.e., env1-jun3_WinQTLCart_Format.mcd.

The format of ICIM covariate dataset If users use the ICIM dataset and there are covariates, users need 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 indicated by such as 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	В
DH6-165	В
DH6-166	В
DH6-170	В
DH7-124	С
DH7-125	С

4. Operation process

3.1 The graphical interface of QTL.gCIMapping.GUI v2.0

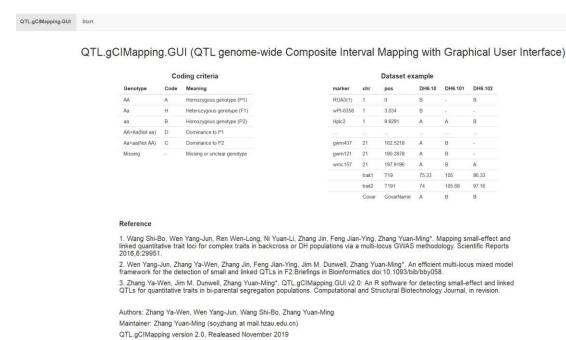
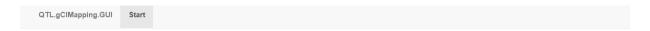


Figure 1. The Graphical User Interface of QTL.gCIMapping.GUI v2.0

3.2 Input dataset

Users must upload the dataset files with three kinds of formats (Figs 2 to 4). If users select the QTLIciMapping format and there are the covariates, users should upload the covariate matrix (Fig 5).



QTL.gClMapping.GUI

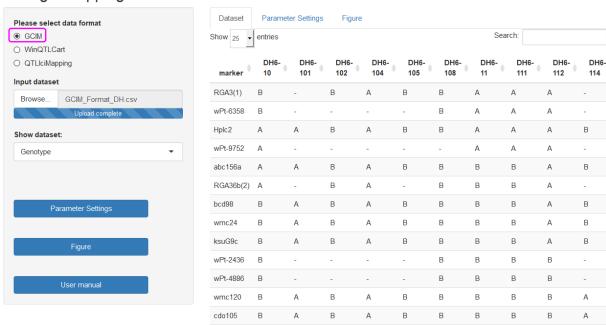
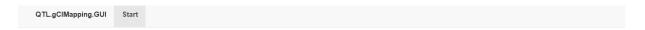


Fig 2. The GCIM dataset format



QTL.gCIMapping.GUI

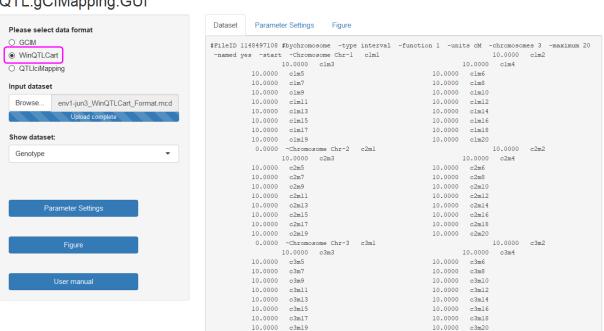


Fig 3. The WinQTLCart dataset format

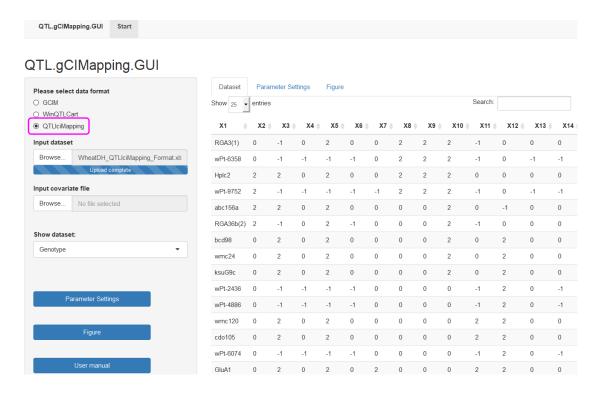


Fig 4. The QTLIciMapping dataset format

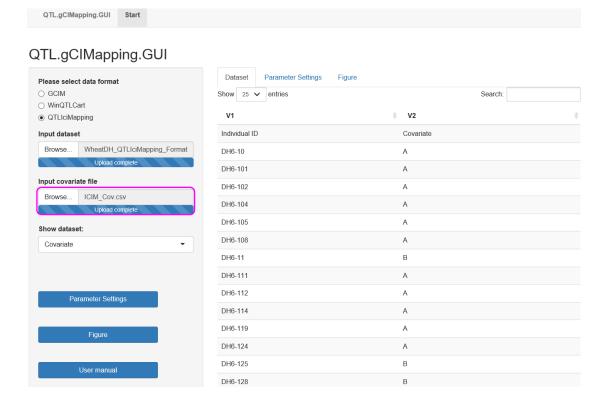


Fig 5. Covariate input in the QTLIciMapping dataset format

3.3 Parameter settings (Fig 6)

Select population: BC1 (F1×P1), BC2 (F1×P2), DH, RIL, and 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, for example, 2.5 or 3.0.

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 F₂ 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" and run again.

Traits analyzed: "2:2" or "2" indicates the analyses from the second trait, "2:4" indicates the analyses from the second to fourth traits, and "2,4" indicates the analyses of the second trait and the fourth trait.

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

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.

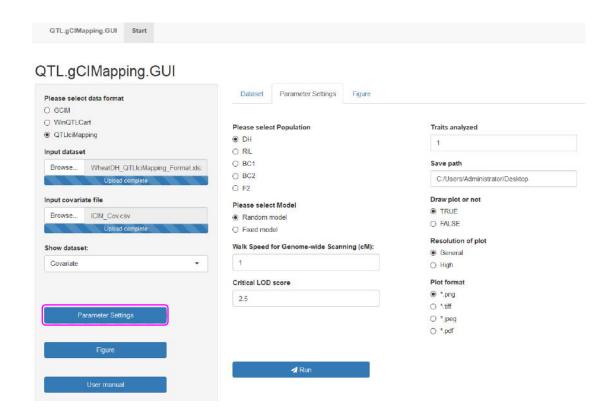


Fig 6. Parameter setting in the mapping of QTL for quantitative traits

3.4 Run the software

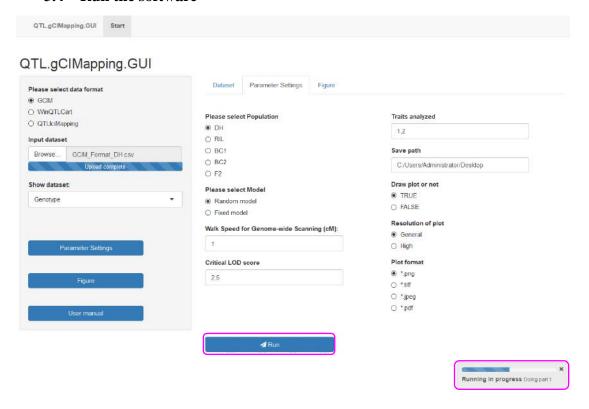


Fig 7. Run the software package QTL.gCIMapping.GUI v2.0

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).

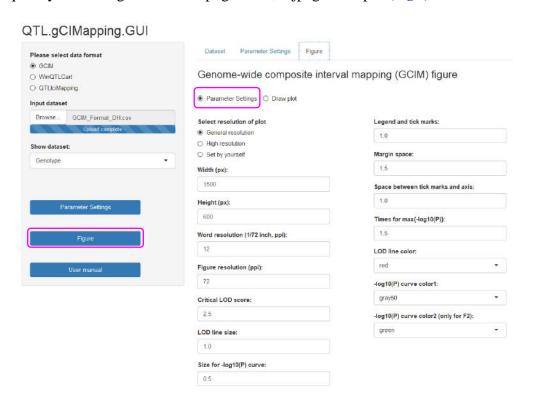


Figure 8. Parameter settings

QTL.gCIMapping.GUI Dataset Parameter Settings Figure Please select data format GCIM Genome-wide composite interval mapping (GCIM) figure O WinQTLCart O QTLIciMapping Input dataset GCIM Format DH.csv Input file to draw plot Select population F2 Browse 1_resultforplot.xlsx ● *.png ○ *.tiff ○ *.jpeg ○ *.pdf Logarithm of odds (LOD) 0

Fig 9. How to draw the LOD score figure in QTL mapping

5. 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.

r² (%): 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_2 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. In F_2 population, QTLs include additive (**Effect.a**) and dominant (**Effect.d**) effects.