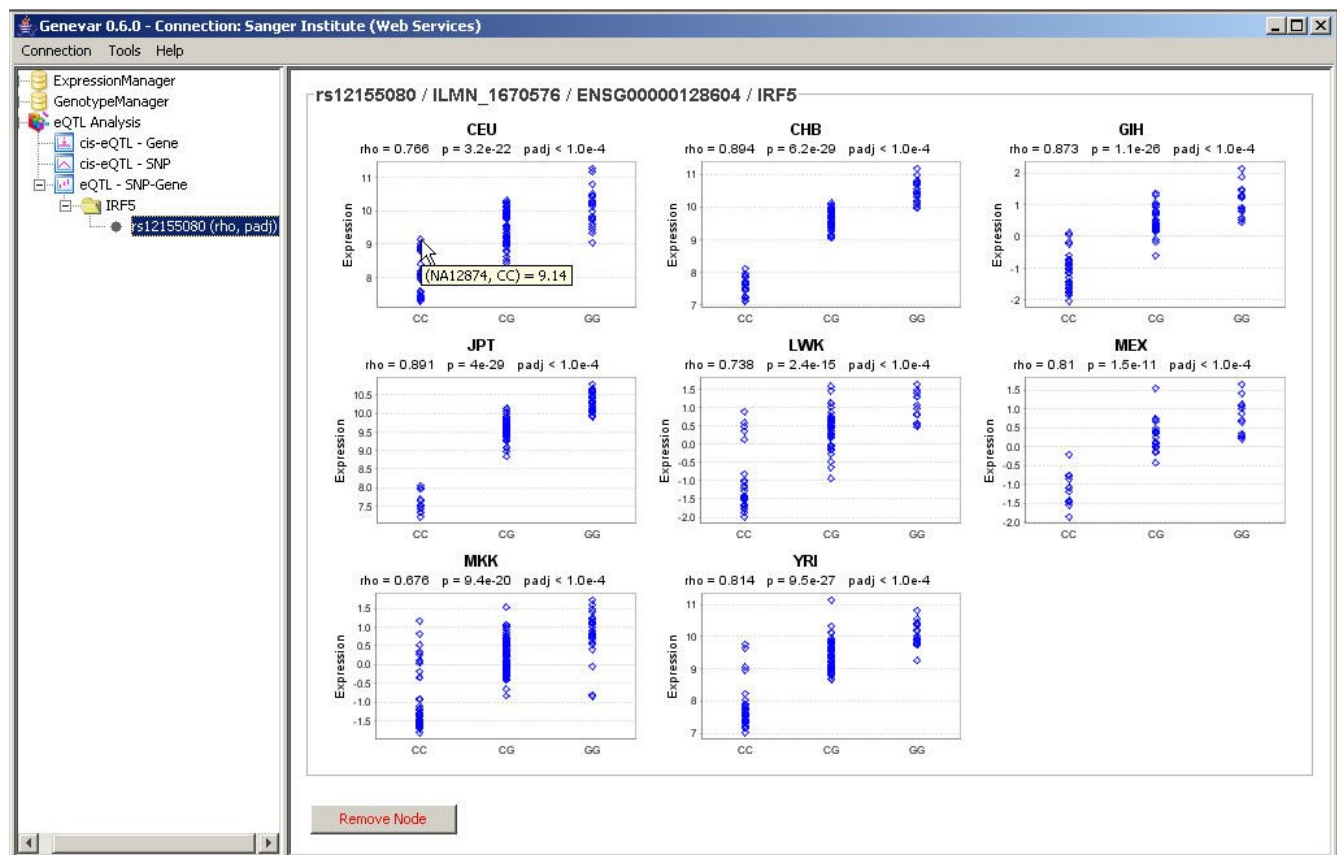


Quick Start: eQTL – SNP-Gene

Snapshot

In this example

- SNP-probe association plots in eight HapMap3 populations
- Spearman's ρ , nominal p -value and permutation p -value are shown above each plot
- Information of SNP-probe pairs will be displayed in a tip text when hovering the cursor over individual of interest



Step-by-Step Guide

1. Click on "eQTL – SNP-Gene".
2. Select a dataset of interest (**e.g. HapMap3**).
3. Choose reference source (**e.g. Ensembl**).
4. Type in gene symbol of interest (**e.g. IRF5**).
5. Match entire word (optional)?
6. Submit and search from the selected platform and source.

Genevar 0.5.0 - Connection: Sanger Institute (Web Service)

Connection Tools Help

ExpressionManager
GenotypeManager
eQTL Analysis
cis-eQTL - Gene
cis-eQTL - SNP
eQTL - SNP-Gene

Gene Symbol Query

1. Select a data set: HapMap3

2. Annotation source: Ensembl

3. Search gene symbol: IRF5

or upload a list of genes/gene-SNP pairs (tab-delimited) from a file: Browse...

Match entire word only

Submit

Query Result

Group: N/A
Source: Ensembl

	Gene Name	chr	Gene Start	Gene End	Strand	Gene Symbol	Uploaded SNP IDs
<input type="checkbox"/>							

(Optional) Submit SNP IDs against All Genes

dbSNP ID (rs number):

or upload a list of SNPs from a file: Browse...

Reset All Find Matched Transcripts

7. Query results will be listed here.
8. Select genes of interest.
9. Submit and find matched transcripts.

Genevar 0.5.0 - Connection: Sanger Institute (Web Service)

Connection Tools Help

- ExpressionManager
- GenotypeManager
- eQTL Analysis
 - cis-eQTL - Gene
 - cis-eQTL - SNP
 - eQTL - SNP-Gene

Gene Symbol Query

1. Select a data set: HapMap3

2. Annotation source: Ensembl

3. Search gene symbol: IRF5

or upload a list of genes/gene-SNP pairs (tab-delimited) from a file: Browse...

Match entire word only **Submit**

Query Result

Group: **HapMap3 / Illumina HumanWG-6 v2**
Source: **Ensembl**

	Gene Name	chr	Gene Start	Gene End	Strand	Gene Symbol	Uploaded SNP IDs
<input checked="" type="checkbox"/>	ENSG00000128604	7	128365230	128377320	1	IRF5	

(Optional) Submit SNP IDs against All Genes

dbSNP ID (rs number):

or upload a list of SNPs from a file: Browse...

Reset All **Find Matched Transcripts**

7

8

9

10. A new tree node containing matched transcripts will be added here. Click on this node.

The screenshot shows the Genevar 0.5.0 web interface. On the left sidebar, a tree structure is visible with nodes for ExpressionManager, GenotypeManager, eQTL Analysis, cis-eQTL - Gene, cis-eQTL - SNP, eQTL - SNP-Gene, and IRF5. A blue arrow points to the IRF5 node, which is labeled with the number 10. The main content area is titled 'Gene Symbol Query' and contains the following fields:

- 1. Select a data set: HapMap3
- 2. Annotation source: Ensembl
- 3. Search gene symbol: IRF5

Below these fields, there is a checkbox for 'Match entire word only' and a 'Submit' button. The 'Query Result' section displays a table with the following data:

	Gene Name	chr	Gene Start	Gene End	Strand	Gene Symbol	Uploaded SNP IDs
<input checked="" type="checkbox"/>	ENSG00000128604	7	128365230	128377320	1	IRF5	

At the bottom of the interface, there is an '(Optional) Submit SNP IDs against All Genes' section with a 'dbSNP ID (rs number):' field and a 'Browse...' button. At the very bottom, there are 'Reset All' and 'Find Matched Transcripts' buttons.

11. Select populations of interest.
12. Select probes of interest.
13. Type in SNP IDs here (**e.g. rs12155080**).
14. Set parameters (**e.g. Spearman's rho, 10,000 permutations**).
15. Submit to get SNP-probe association plots.

Genevar 0.5.0 - Connection: Sanger Institute (Web Service)

Connection Tools Help

- ExpressionManager
- GenotypeManager
- eQTL Analysis
 - cis-eQTL - Gene
 - cis-eQTL - SNP
 - eQTL - SNP-Gene
 - IRFS

Expression-Genotype Pairs

Group: **HapMap3**

	Population	Expression	Genotype
<input checked="" type="checkbox"/>	CEU	CEU / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	CEU / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	CHB	CHB / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	CHB / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	GIH	GIH / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	GIH / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	JPT	JPT / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	JPT / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	LWK	LWK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	LWK / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	MEX	MEX / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	MEX / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	MKK	MKK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	MKK / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	YRI	YRI / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	YRI / HapMap Phase 3.1 / REBUILT-NO-INDEL

Matched Transcripts

Platform: **Illumina HumanWG-6 v2**

	Probe Id	Gene Name	chr	Gene Start	Gene End	Strand	Probe Start	Gene Symbol
<input checked="" type="checkbox"/>	ILMN_1670576	ENS5G00000128604	7	128365230	128377320	1	128367567	IRFS

Submit SNP IDs against This Gene

dbSNP ID (rs number):

or upload a list of SNPs from a file:

Analysis Parameters

1. Correlation and regression:

2. Number of permutations:

16. A new tree node containing association plots will be added here. Click on this node.

The screenshot shows the Genevar 0.5.0 web interface. On the left, a tree view shows the navigation structure: ExpressionManager, GenotypeManager, eQTL Analysis, cis-eQTL - Gene, cis-eQTL - SNP, eQTL - SNP-Gene, and IRFS. A blue arrow labeled '16' points to the 'rs12155080 (rho, padj)' node under IRFS.

The main content area is titled 'Expression-Genotype Pairs' and shows a table for the 'HapMap3' group. The table has columns for Population, Expression, and Genotype. Below this is the 'Matched Transcripts' section, showing the platform 'Illumina HumanWG-6 v2' and a table with columns for Probe Id, Gene Name, chr, Gene Start, Gene End, Strand, Probe Start, and Gene Symbol. A 'Processing...' dialog box is overlaid on the transcript table.

The 'Submit SNP IDs against This Gene' section contains a text input field with 'rs12155080' and a 'Browse...' button. The 'Analysis Parameters' section has two dropdown menus: '1. Correlation and regression: Spearman's rank correlation coefficient (rho)' and '2. Number of permutations: 10,000'. At the bottom, there are 'Remove Node' and 'SNP-Genotype Association Plot' buttons.

Group:	HapMap3		
	Population	Expression	Genotype
<input checked="" type="checkbox"/>	CEU	CEU / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	CEU / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	CHB	CHB / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	CHB / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	GIH	GIH / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	GIH / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	JPT	JPT / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	JPT / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	LWK	LWK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	LWK / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	MEX	MEX / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	MEX / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	MKK	MKK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	MKK / HapMap Phase 3.1 / REBUILT-NO-INDEL
<input checked="" type="checkbox"/>	YRI	YRI / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9	YRI / HapMap Phase 3.1 / REBUILT-NO-INDEL

Platform:	Illumina HumanWG-6 v2							
	Probe Id	Gene Name	chr	Gene Start	Gene End	Strand	Probe Start	Gene Symbol
<input checked="" type="checkbox"/>	ILMA	Processing...			28377320	1	128367567	IRFS

Submit SNP IDs against This Gene

dbSNP ID (rs number):

or upload a list of SNPs from a file:

Analysis Parameters

1. Correlation and regression:

2. Number of permutations:

SNP-probe association plots.

