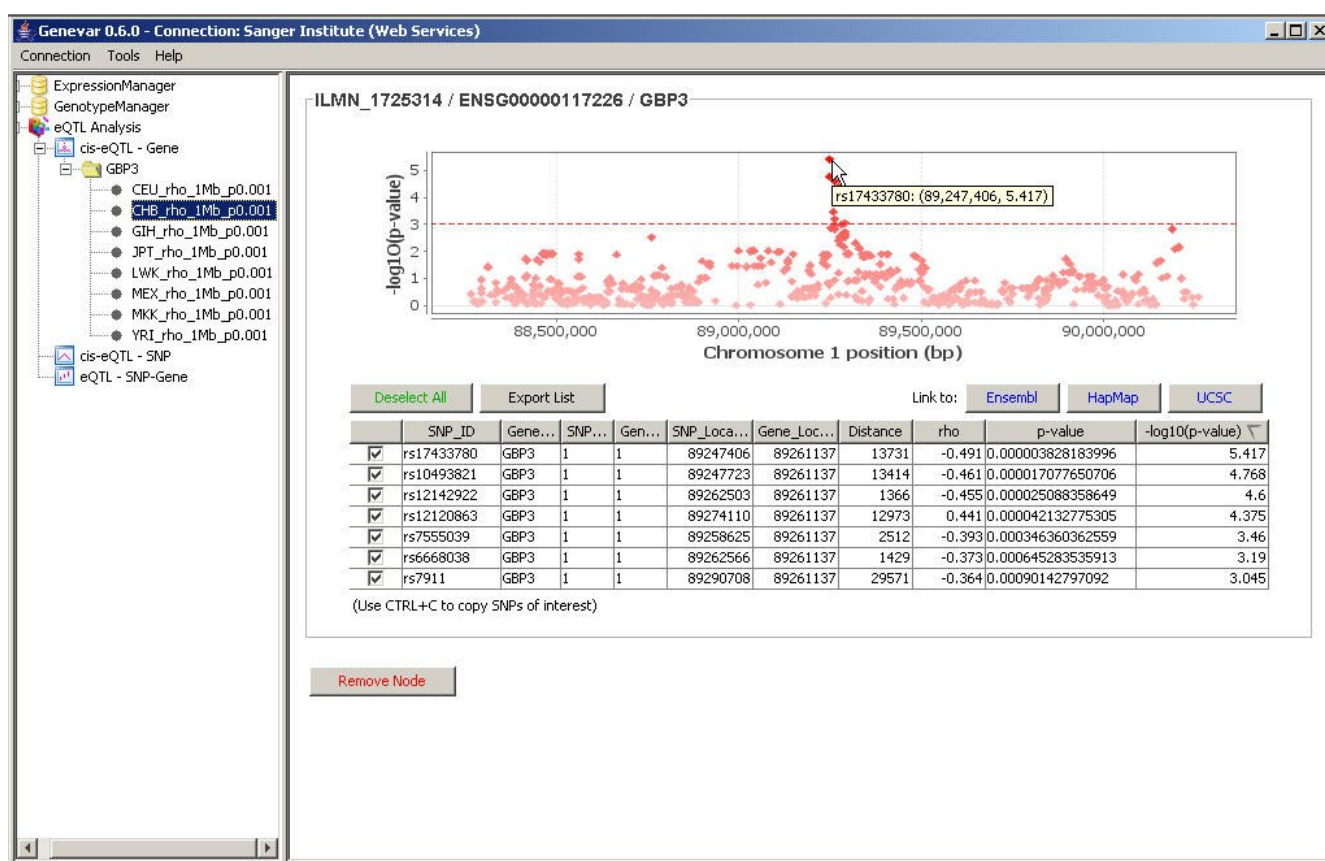


Quick Start: cis-eQTL – Gene

Snapshot

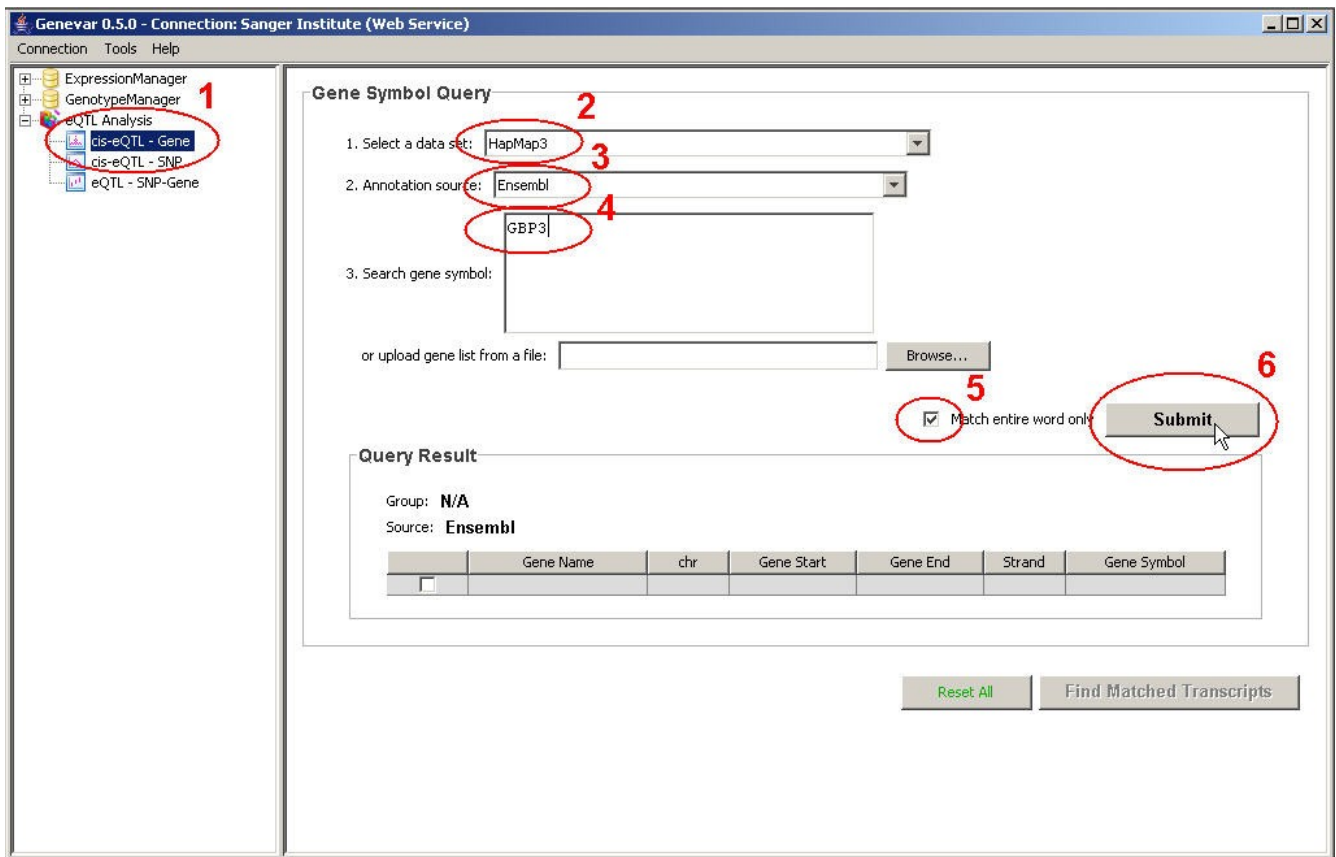
In this example

- Observed eQTLs in a 2-Mb window around the *GBP3* locus in HapMap3 CHB
- Significant SNPs within the chromosome region are listed below
- Dotted line represents the 0.001 p -value threshold
- Information of SNPs will be displayed in a tip text when hovering the cursor over the SNP of interest
- Results can be saved as a PNG diagram or exported as a tab-delimited list
- External links to three major genome browsers



Step-by-Step Guide

1. Click on "cis-eQTL – 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. **GBP3**).
5. Match entire word (optional)?
6. Submit and search from the selected dataset and reference.



7. Query results will be listed here.
8. Select genes of interest.
9. Submit to 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: GBP3

or upload gene list 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
<input checked="" type="checkbox"/>	ENSG00000117226	1	89244948	89261137	-1	GBP3

7 **8** **9**

Reset All **Find Matched Transcripts**

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

Genevar 0.5.0 - Connection: Sanger Institute (Web Service)

Connection Tools Help

ExpressionManager
GenotypeManager
eQTL Analysis
 cis-eQTL - Gene
 GBP3
 cis-eQTL
 eQTL - 9

Gene Symbol Query

1. Select a data set: HapMap3

2. Annotation source: Ensembl

3. Search gene symbol: GBP3

or upload gene list from a file: Browse...

Match entire word only

Processing...

Query Results

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

	Gene Name	chr	Gene Start	Gene End	Strand	Gene Symbol
<input checked="" type="checkbox"/>	ENSG00000117226	1	89244948	89261137	-1	GBP3

11. Select populations of interest.
12. Select probes of interest.
13. Set parameters (**e.g. Spearman's ρ , 1 Mb, $p < 0.001$**).
14. Submit to get mapping results.

Genevar 0.5.0 - Connection: Sanger Institute (Web Service)

Connection Tools Help

ExpressionManager
GenotypeManager
eQTL Analysis
cis-eQTL - Gene
 • GBP3
cis-eQTL - SNP
eQTL - SNP-Gene

Expression-Genotype Pairs

Group: **HapMap3**

<input checked="" type="checkbox"/>	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**

<input checked="" type="checkbox"/>	Probe Id	Gene Name	chr	Gene Start	Gene End	Strand	Probe Start	Gene Symbol
<input checked="" type="checkbox"/>	ILMN_1725314	ENSG00000117226	1	89244948	89261137	-1	89258319	GBP3

Analysis Parameters

- Correlation and regression: Spearman's rank correlation coefficient (rho)
- Distance separation upper limit: 1,000,000 (editable, no greater than 1Mb)
- P-value upper limit filter: 0.001 (editable)

Remove Node **Run**

15. New tree nodes containing mapping results for each population will be added here. Click on one of them.

Genevar 0.5.0 - Connection: Sanger Institute (Web Service)

ExpressionManager
GenotypeManager
eQTL Analysis
cis-eQTL - Gene
 ● GBP3
 ● CEU_rho_1Mb_p0.001
 ● CHB_rho_1Mb_p0.001
 ● GIH_rho_1Mb_p0.001
 ● JPT_rho_1Mb_p0.001
 ● LWK_rho_1Mb_p0.001
 ● MEX_rho_1Mb_p0.001
 ● MKK_rho_1Mb_p0.001
cis-eQTL - SNP
eQTL - SNP-Gene

15

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: IIII

	Probe Id	Gene Name	chr	Gene Start	Gene End	Strand	Probe Start	Gene Symbol
<input checked="" type="checkbox"/>	ILMN_1725314	ENSG00000117226	1	89244948	89261137	-1	89258319	GBP3

Analysis Parameters

- Correlation and regression: Spearman's rank correlation coefficient (rho)
- Distance separation upper limit: 1,000,000 (editable, no greater than 1Mb)
- P-value upper limit filter: 0.001 (editable)

[Remove Node](#) [Run](#)

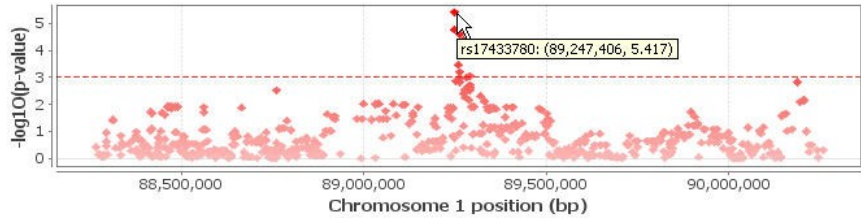
All eQTLs within this 2-Mb region will be plotted on a chromosome view; only significant SNPs are listed below.

Genevar 0.6.0 - Connection: Sanger Institute (Web Services)

Connection Tools Help

- ExpressionManager
- GenotypeManager
- eQTL Analysis
 - cis-eQTL - Gene
 - GBP3
 - CEU_rho_1Mb_p0.001
 - CHB_rho_1Mb_p0.001**
 - GIH_rho_1Mb_p0.001
 - JPT_rho_1Mb_p0.001
 - LWK_rho_1Mb_p0.001
 - MEX_rho_1Mb_p0.001
 - MKK_rho_1Mb_p0.001
 - YRI_rho_1Mb_p0.001
 - cis-eQTL - SNP
 - eQTL - SNP-Gene

ILMN_1725314 / ENSG00000117226 / GBP3



Link to: [Ensembl](#) [HapMap](#) [UCSC](#)

	SNP_ID	Gene...	SNP...	Gen...	SNP_Loca...	Gene_Loc...	Distance	rho	p-value	$-\log_{10}(p\text{-value})$
<input checked="" type="checkbox"/>	rs17433780	GBP3	1	1	89247406	89261137	13731	-0.491	0.000003628183996	5.417
<input checked="" type="checkbox"/>	rs10493821	GBP3	1	1	89247723	89261137	13414	-0.461	0.000017077650706	4.768
<input checked="" type="checkbox"/>	rs12142922	GBP3	1	1	89262503	89261137	1366	-0.455	0.000025088358649	4.6
<input checked="" type="checkbox"/>	rs12120863	GBP3	1	1	89274110	89261137	12973	0.441	0.000042132775305	4.375
<input checked="" type="checkbox"/>	rs7555039	GBP3	1	1	89258625	89261137	2512	-0.393	0.000346360362559	3.46
<input checked="" type="checkbox"/>	rs6668038	GBP3	1	1	89262566	89261137	1429	-0.373	0.000645283535913	3.19
<input checked="" type="checkbox"/>	rs7911	GBP3	1	1	89290708	89261137	29571	-0.364	0.00090142797092	3.045

(Use CTRL+C to copy SNPs of interest)