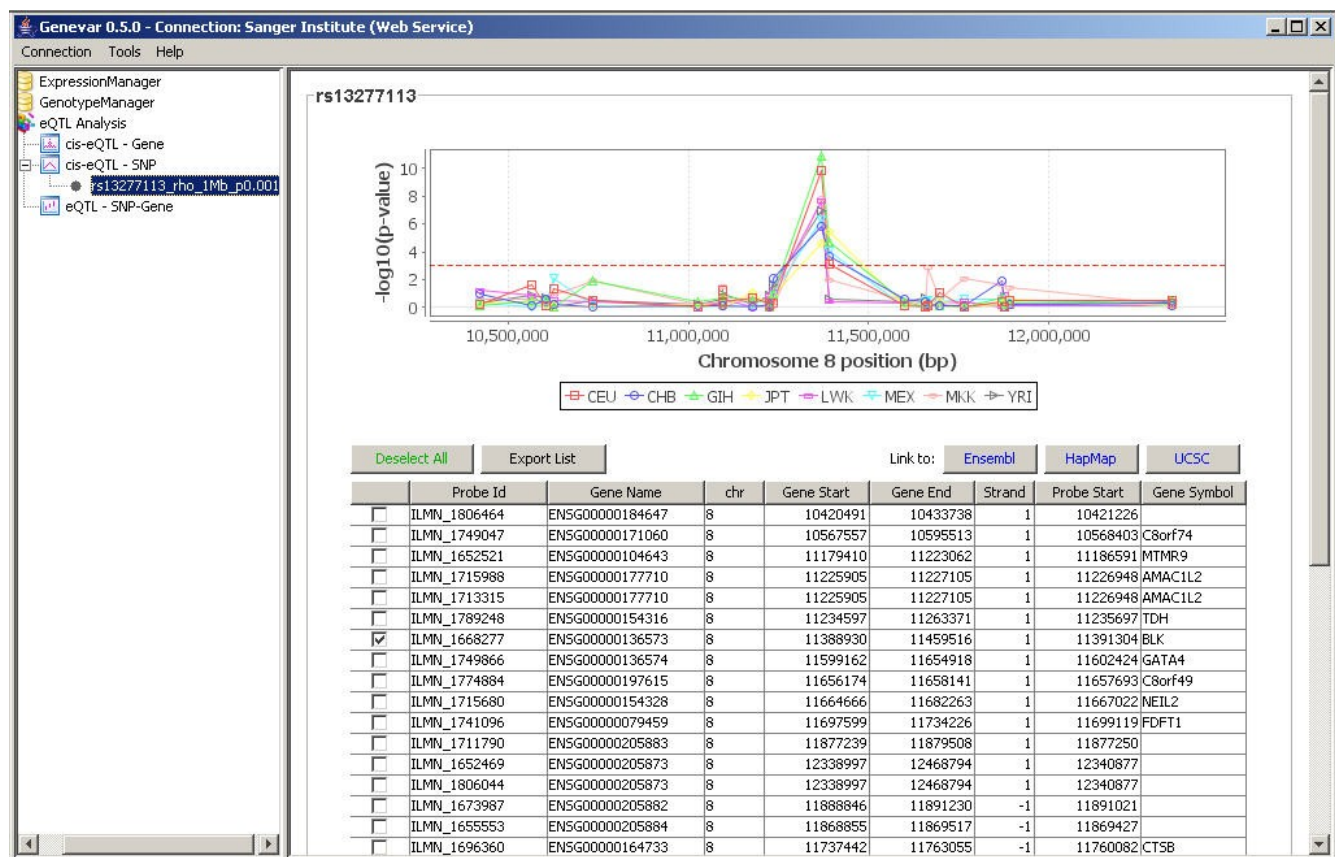


Quick Start: cis-eQTL – SNP

Snapshot

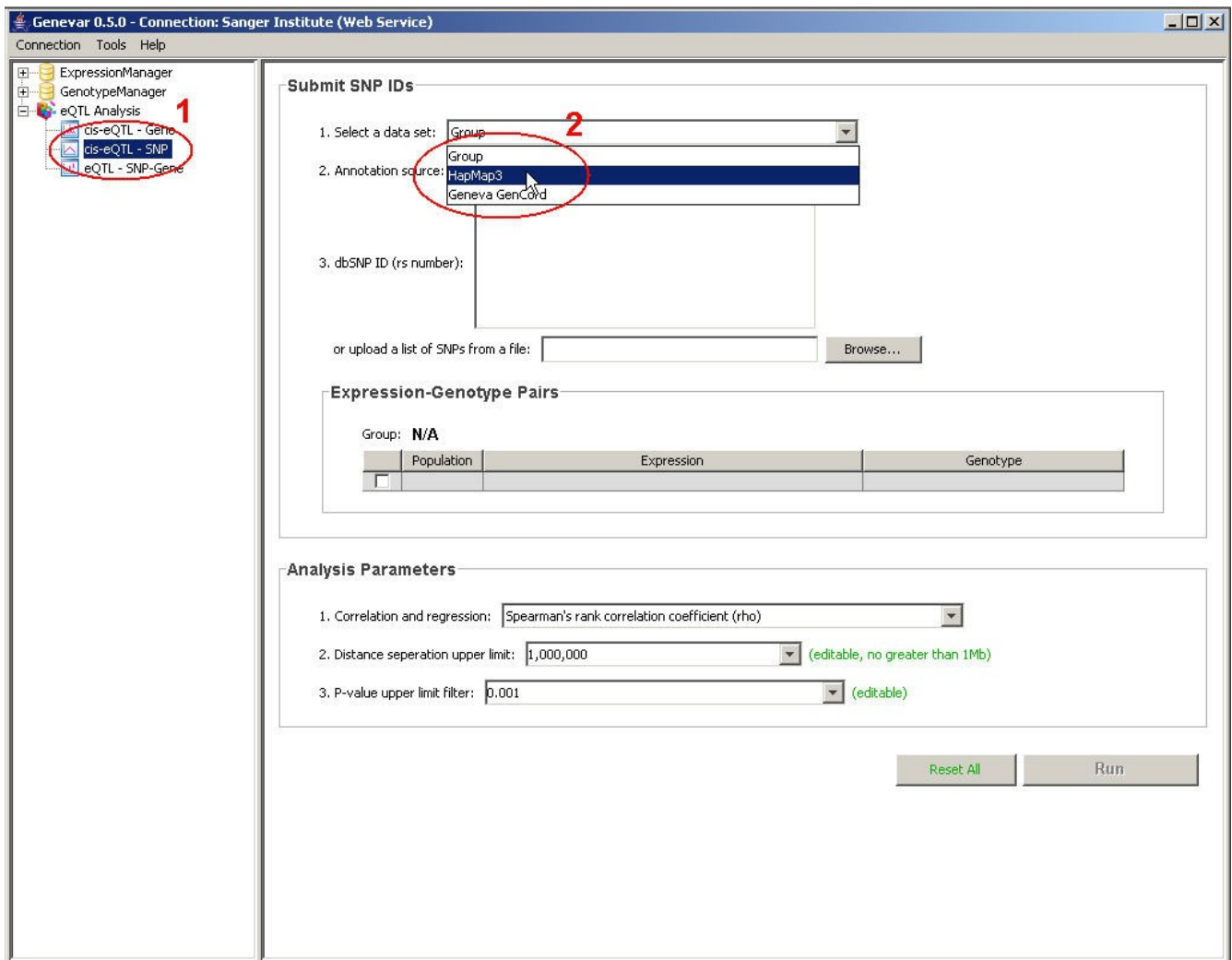
In this example

- Observed SNP-gene associations in a 2-Mb region surrounding rs13277113 SNP in eight HapMap3 populations
- Significant genes within the chromosome region are listed below
- Dotted line represents the 0.001 p -value threshold
- 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 – SNP".
2. Select a group of interest (e.g. HapMap3).



3. Available expression-genotype pairs will be listed here.

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

Submit SNP IDs

1. Select a data set: HapMap3

2. Annotation source: Ensembl

3. dbSNP ID (rs number):

or upload a list of SNPs from a file:

Expression-Genotype Pairs

Group: HapMap3 / Illumina HumanWG-6 v2

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

Analysis Parameters

1. Correlation and regression: Spearman's rank correlation coefficient (rho)

2. Distance separation upper limit: 1,000,000 (editable, no greater than 1Mb)

3. P-value upper limit filter: 0.001 (editable)

4. Choose reference source (**e.g. Ensembl**).
5. Type in SNPs here (**e.g. rs13277113**).
6. Select populations of interest.
7. Set parameters (**e.g. Spearman's rho, 1 Mb, $p < 0.001$**).
8. Submit to get transcript lists.

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

Submit SNP IDs

1. Select a data set: HapMap3 **4**

2. Annotation source: Ensembl **5**
rs13277113

3. dbSNP ID (rs number):

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

Expression-Genotype Pairs

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

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

Analysis Parameters

1. Correlation and regression: Spearman's rank correlation coefficient (rho) **7**

2. Distance separation upper limit: 1,000,000 (editable, no greater than 1Mb)

3. P-value upper limit filter: 0.001 (editable)

8

9. A new tree node containing matched transcripts 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
 cis-eQTL - SNP
 rs13277113_rho_1Mb_p0.001
 eQTL - SNP-Gene

Submit SNP IDs

1. Select a data set: HapMap3

2. Annotation source: Ensembl

rs13277113

3. dbSNP ID (rs number):

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

Expression-Genotype Pairs

Group: HapMap3 / Illumina HumanWG-6 v2

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

Analysis Parameters

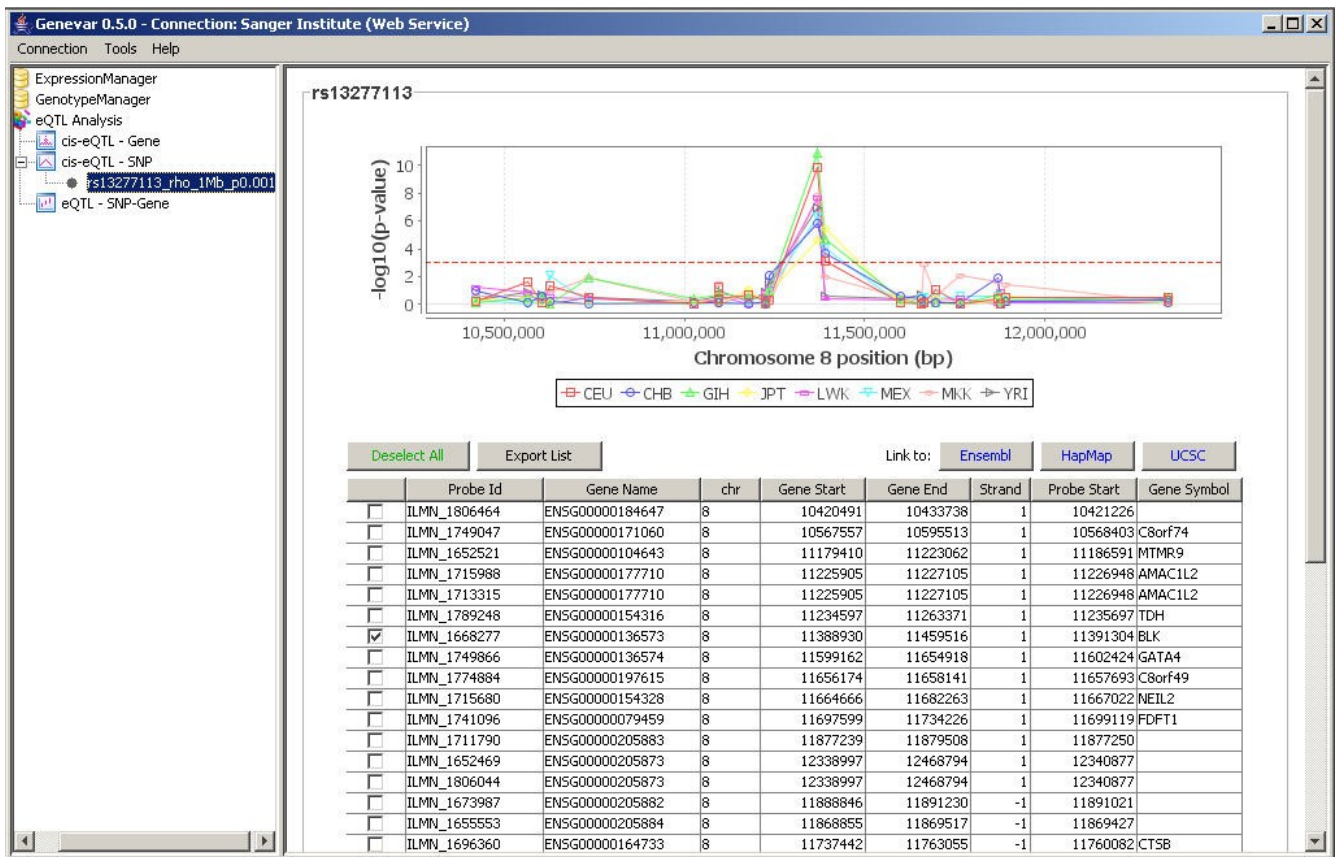
1. Correlation and regression: Spearman's rank correlation coefficient (rho)

2. Distance separation upper limit: 1,000,000 (editable, no greater than 1Mb)

3. P-value upper limit filter: 0.001 (editable)

Reset All Run

Probes within this 2-Mb region will be listed below; significant ones are clicked as "chosen".



10. Set parameters (e.g. Spearman's rho, 10,000 permutations).
11. Submit to get SNP-probe association plots.

Genevar 0.5.0 - Connection: Sanger Institute (Web Service)

Connection Tools Help

10,500,000 11,000,000 11,500,000 12,000,000

Chromosome 8 position (bp)

CEU CHB GIH JPT LWK MEX MKK YRI

Deselect All Export List Link to: Ensembl HapMap UCSC

	Probe Id	Gene Name	chr	Gene Start	Gene End	Strand	Probe Start	Gene Symbol
<input type="checkbox"/>	ILMN_1806464	ENS00000184647	8	10420491	10433738	1	10421226	
<input type="checkbox"/>	ILMN_1749047	ENS00000171060	8	10567557	10595513	1	10568403	C8orf74
<input type="checkbox"/>	ILMN_1652521	ENS00000104643	8	11179410	11223062	1	11186591	MTMR9
<input type="checkbox"/>	ILMN_1715988	ENS00000177710	8	11225905	11227105	1	11226948	AMAC1L2
<input type="checkbox"/>	ILMN_1713315	ENS00000177710	8	11225905	11227105	1	11226948	AMAC1L2
<input type="checkbox"/>	ILMN_1789248	ENS00000154316	8	11234597	11263371	1	11235697	TDH
<input checked="" type="checkbox"/>	ILMN_1668277	ENS00000136573	8	11388930	11459516	1	11391304	BLK
<input type="checkbox"/>	ILMN_1749866	ENS00000136574	8	11599162	11654918	1	11602424	GATA4
<input type="checkbox"/>	ILMN_1774884	ENS00000197615	8	11656174	11658141	1	11657693	C8orf49
<input type="checkbox"/>	ILMN_1715680	ENS00000154328	8	11664666	11682263	1	11667022	NEIL2
<input type="checkbox"/>	ILMN_1741096	ENS00000079459	8	11697599	11734226	1	11699119	FDFT1
<input type="checkbox"/>	ILMN_1711790	ENS00000205883	8	11877239	11879508	1	11877250	
<input type="checkbox"/>	ILMN_1652469	ENS00000205873	8	12338997	12468794	1	12340877	
<input type="checkbox"/>	ILMN_1806044	ENS00000205873	8	12338997	12468794	1	12340877	
<input type="checkbox"/>	ILMN_1673987	ENS00000205882	8	11888846	11891230	-1	11891021	
<input type="checkbox"/>	ILMN_1655553	ENS00000205884	8	11868855	11869517	-1	11869427	
<input type="checkbox"/>	ILMN_1696360	ENS00000164733	8	11737442	11763055	-1	11760082	CTSB
<input checked="" type="checkbox"/>	ILMN_1687213	ENS00000154319	8	11316391	11369636	-1	11357779	C8orf13
<input type="checkbox"/>	ILMN_1699085	ENS00000215346	8	11021390	11025155	-1	11023119	C8orf16
<input type="checkbox"/>	ILMN_1655446	ENS00000171044	8	10791075	11096258	-1	11095561	XKR6
<input type="checkbox"/>	ILMN_1724762	ENS00000171044	8	10791075	11096258	-1	11095561	XKR6
<input type="checkbox"/>	ILMN_1731175	ENS00000171044	8	10791075	11096258	-1	11095561	XKR6
<input type="checkbox"/>	ILMN_1790309	ENS00000104637	8	10659885	10734796	-1	10734126	
<input type="checkbox"/>	ILMN_1815032	ENS00000171056	8	10618688	10625432	-1	10622781	SOX7
<input type="checkbox"/>	ILMN_1657040	ENS00000183638	8	10501269	10607107	-1	10542344	RP1L1

Analysis Parameters

1. Correlation and regression: Spearman's rank correlation coefficient (rho) 10

2. Number of permutations: 10,000

Remove Node 11 SNP-Gene Association Plot

12. New tree nodes containing association plots for each transcript will be added here. Click on one of them.

Genevar 0.5.0 - Connection: Sanger Institute (Web Service)

Connection Tools Help

10,500,000 11,000,000 11,500,000 12,000,000

Chromosome 8 position (bp)

CEU CHB GIH JPT LWK MEX MKK YRI

Deselect All Export List Link to: Ensembl HapMap UCSC

	Probe Id	Gene Name	chr	Gene Start	Gene End	Strand	Probe Start	Gene Symbol
<input type="checkbox"/>	ILMN_1806464	ENS00000184647	8	10420491	10433738	1	10421226	
<input type="checkbox"/>	ILMN_1749047	ENS00000171060	8	10567557	10595513	1	10568403	C8orf74
<input type="checkbox"/>	ILMN_1652521	ENS00000104643	8	11179410	11223062	1	11186591	MTMR9
<input type="checkbox"/>	ILMN_1715988	ENS00000177710	8	11225905	11227105	1	11226948	AMAC1L2
<input type="checkbox"/>	ILMN_1713315	ENS00000177710	8	11225905	11227105	1	11226948	AMAC1L2
<input type="checkbox"/>	ILMN_1789248	ENS00000154316	8	11234597	11263371	1	11235697	TDH
<input checked="" type="checkbox"/>	ILMN_1668277	ENS00000136573	8	11388930	11459516	1	11391304	BLK
<input type="checkbox"/>	ILMN_1749866	ENS00000136574	8	11599162	11654918	1	11602424	GATA4
<input type="checkbox"/>	ILMN_1774884	ENS00000197615	8	11656174	11658141	1	11657693	C8orf49
<input type="checkbox"/>	ILMN_1715680	ENS00000154328	8	11664666	11682263	1	11667022	NEIL2
<input type="checkbox"/>	ILMN_1741096	ENS00000079459	8	11697599	11734226	1	11699119	FDFT1
<input type="checkbox"/>	ILMN_1711790	ENS00000205883	8	11877239	11879508	1	11877250	
<input type="checkbox"/>	ILMN_1655553	ENS00000205884	8	11868855	11869517	-1	11869427	
<input type="checkbox"/>	ILMN_1696360	ENS00000164733	8	11737442	11763055	-1	11760082	CTSB
<input checked="" type="checkbox"/>	ILMN_1687213	ENS00000154319	8	11316391	11369636	-1	11357779	C8orf13
<input type="checkbox"/>	ILMN_1699085	ENS00000215346	8	11021390	11025155	-1	11023119	C8orf16
<input type="checkbox"/>	ILMN_1655446	ENS00000171044	8	10791075	11096258	-1	11095561	XKR6
<input type="checkbox"/>	ILMN_1724762	ENS00000171044	8	10791075	11096258	-1	11095561	XKR6
<input type="checkbox"/>	ILMN_1731175	ENS00000171044	8	10791075	11096258	-1	11095561	XKR6
<input type="checkbox"/>	ILMN_1790309	ENS00000104637	8	10659885	10734796	-1	10734126	
<input type="checkbox"/>	ILMN_1815032	ENS00000171056	8	10618688	10625432	-1	10622781	SOX7
<input type="checkbox"/>	ILMN_1657040	ENS00000183638	8	10501269	10607107	-1	10542344	RP1L1

Processing...

Analysis Parameters

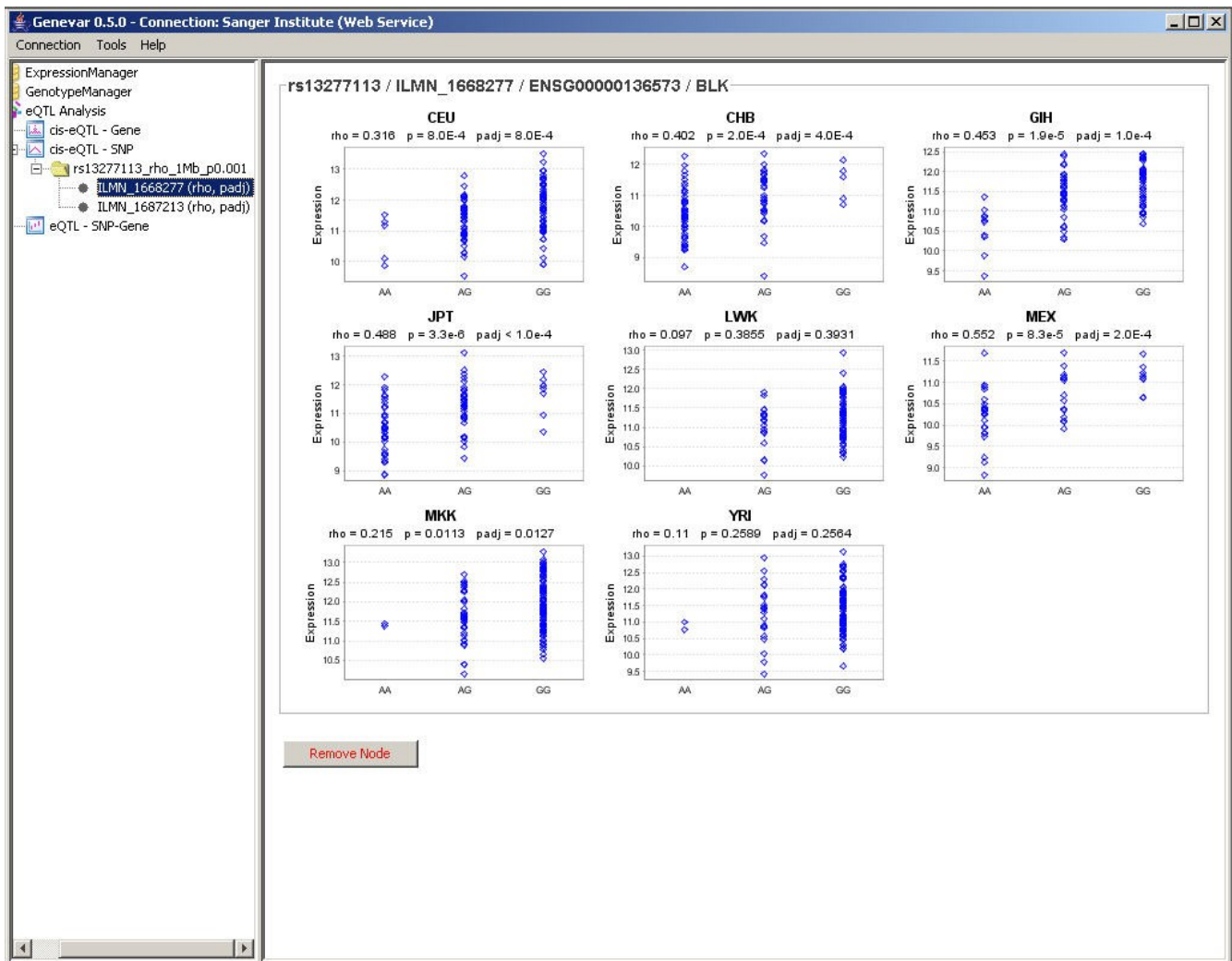
1. Correlation and regression: Spearman's rank correlation coefficient (rho)

2. Number of permutations: 10,000

Remove Node SNP-Gene Association Plot

12

SNP-probe association plots.



Gene Start Sites

Sophisticated SQL query fetches both *PDE4B* (+) and *GPR177* (-) even though their gene starts are outside the 2-Mb region.

