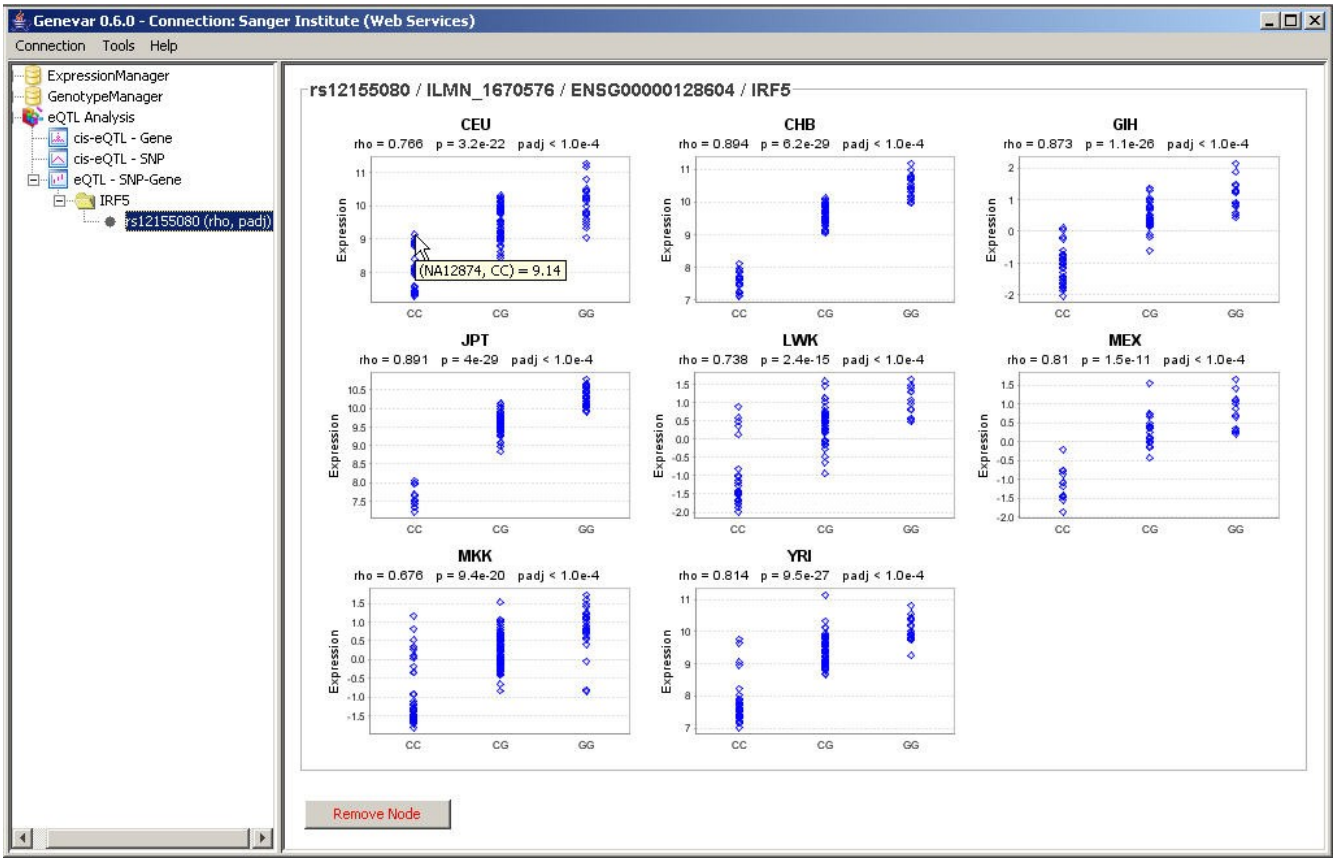
Quick Start: eQTL – SNP-Gene

Snapshot

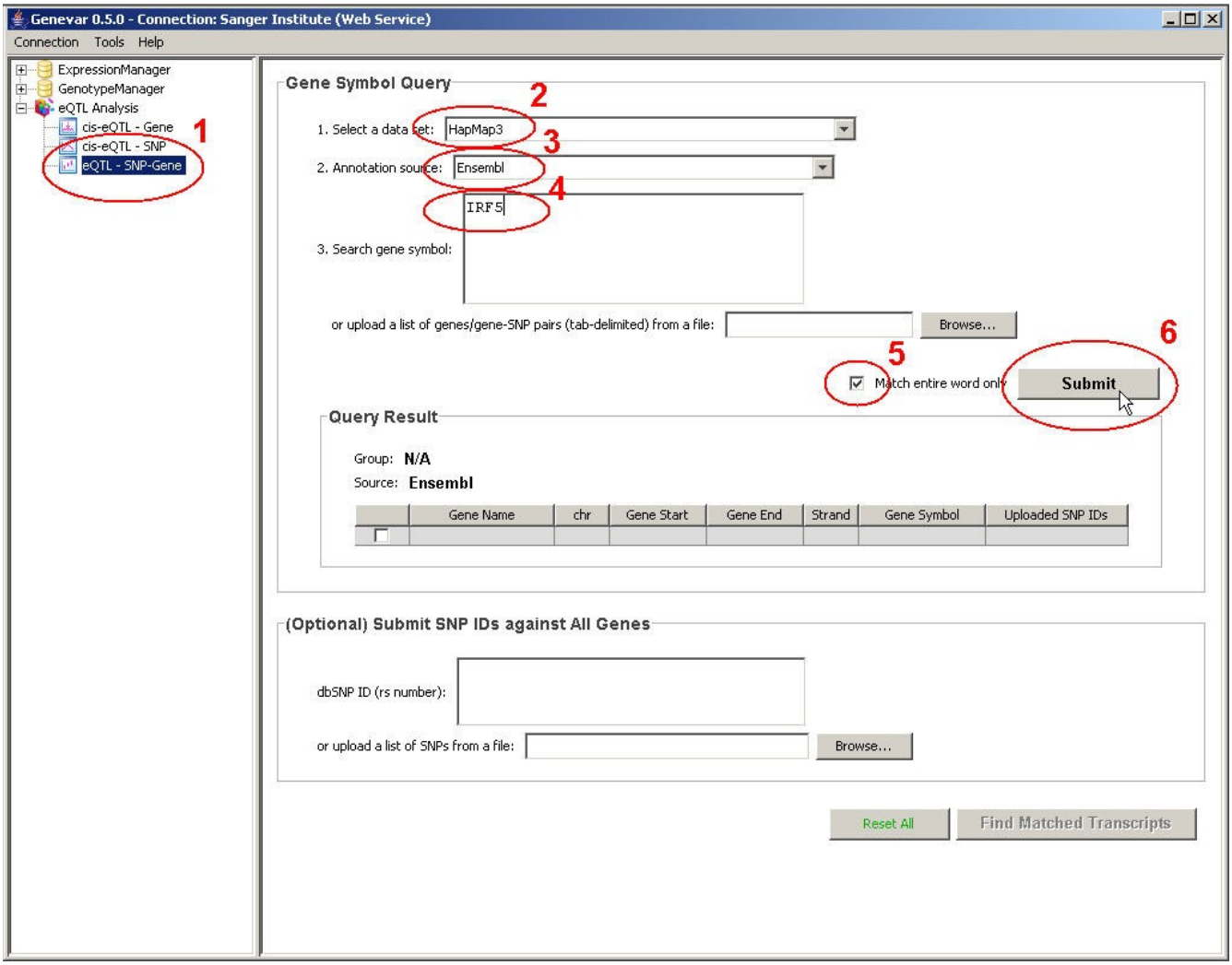
In this example

- SNP-probe association plots in eight HapMap3 populations
- Spearman's *rho*, 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

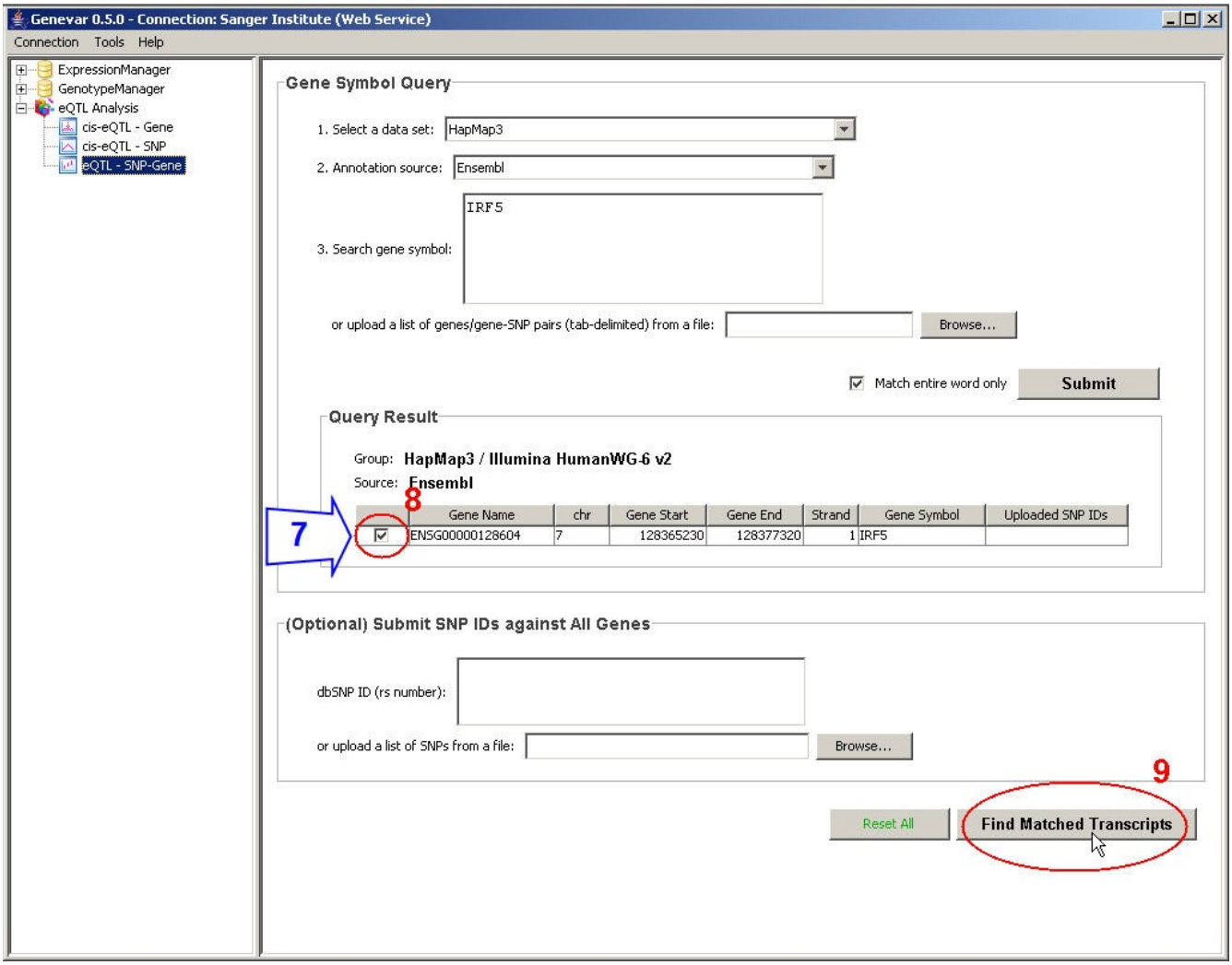


Step-by-Step Guide

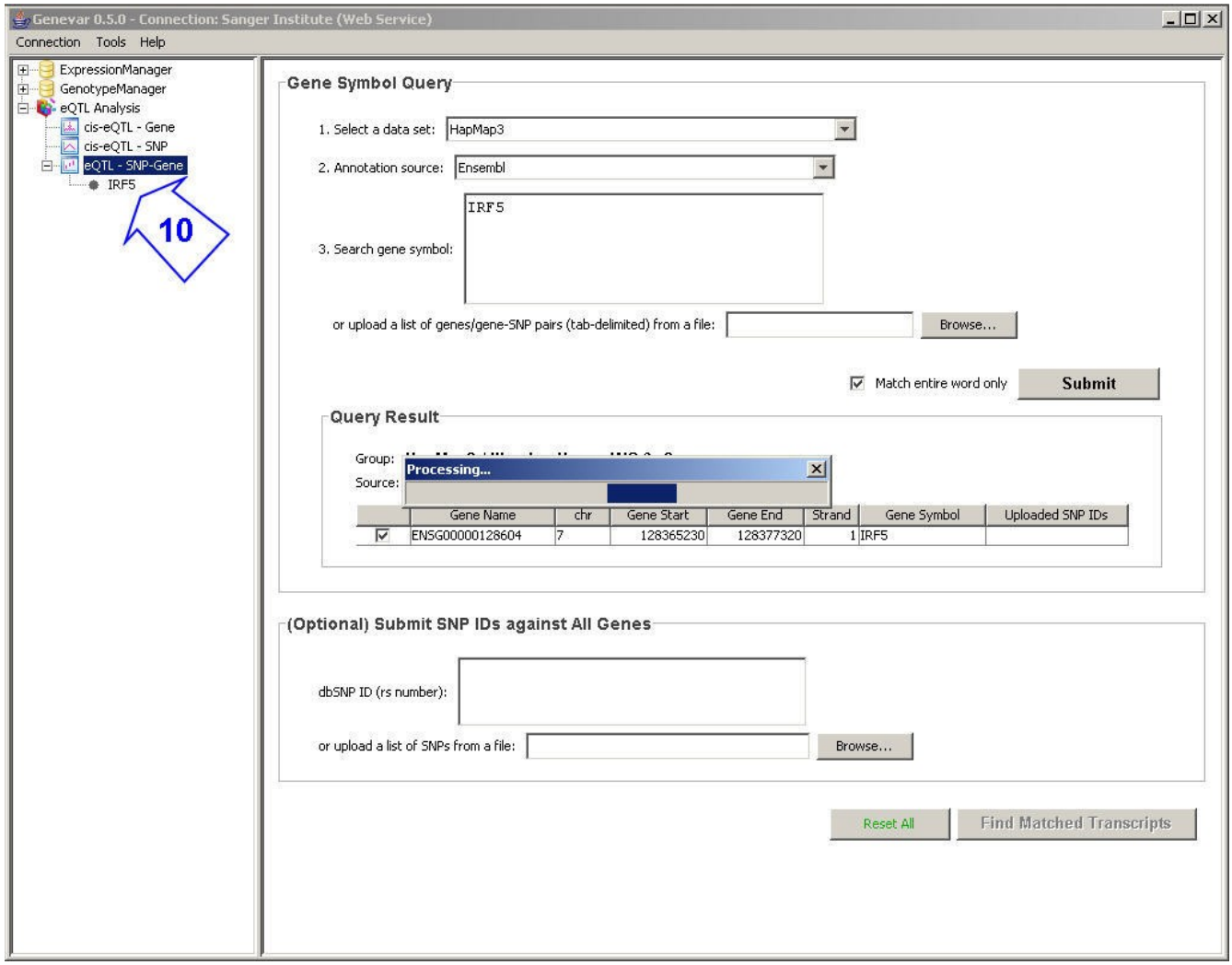
1. Click on "eQTL – SNP-Gene".
2. Select a dataset of interest (**e.g. HapMap3**).
3. Choose reference source (**e.g. Ensembl**).
4. Type in gene symbol of interest (**e.g. *IRF5***).
5. Match entire word (optional)?
6. Submit and search from the selected platform and source.



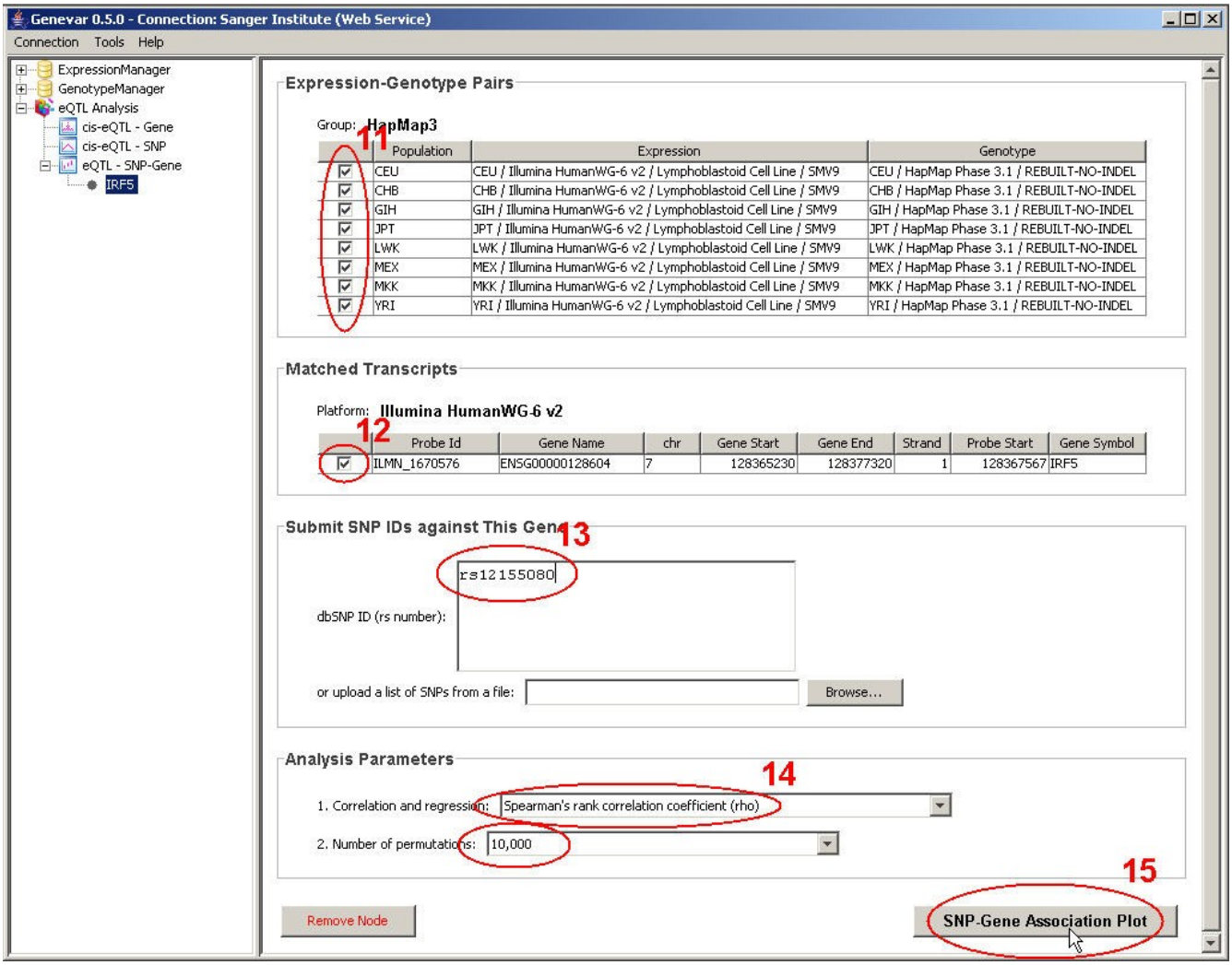
7. Query results will be listed here.
8. Select genes of interest.
9. Submit and find matched transcripts.


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
8
7
Gene Name
chr
Gene Start
Gene End
Strand
Gene Symbol
Uploaded SNP IDs
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...
9
Reset All
Find Matched Transcripts

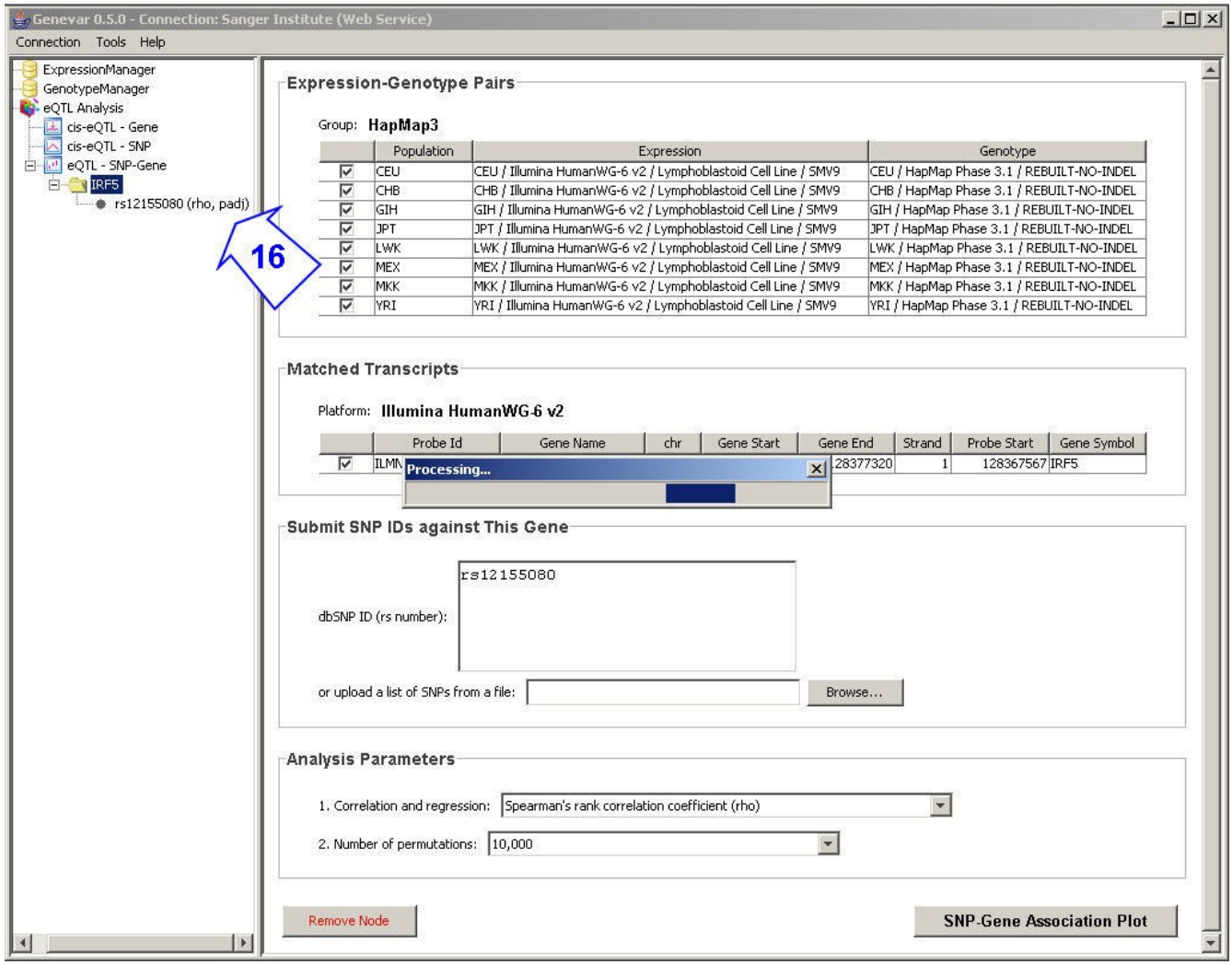
10. A new tree node containing matched trascripts will be added here. Click on this node.



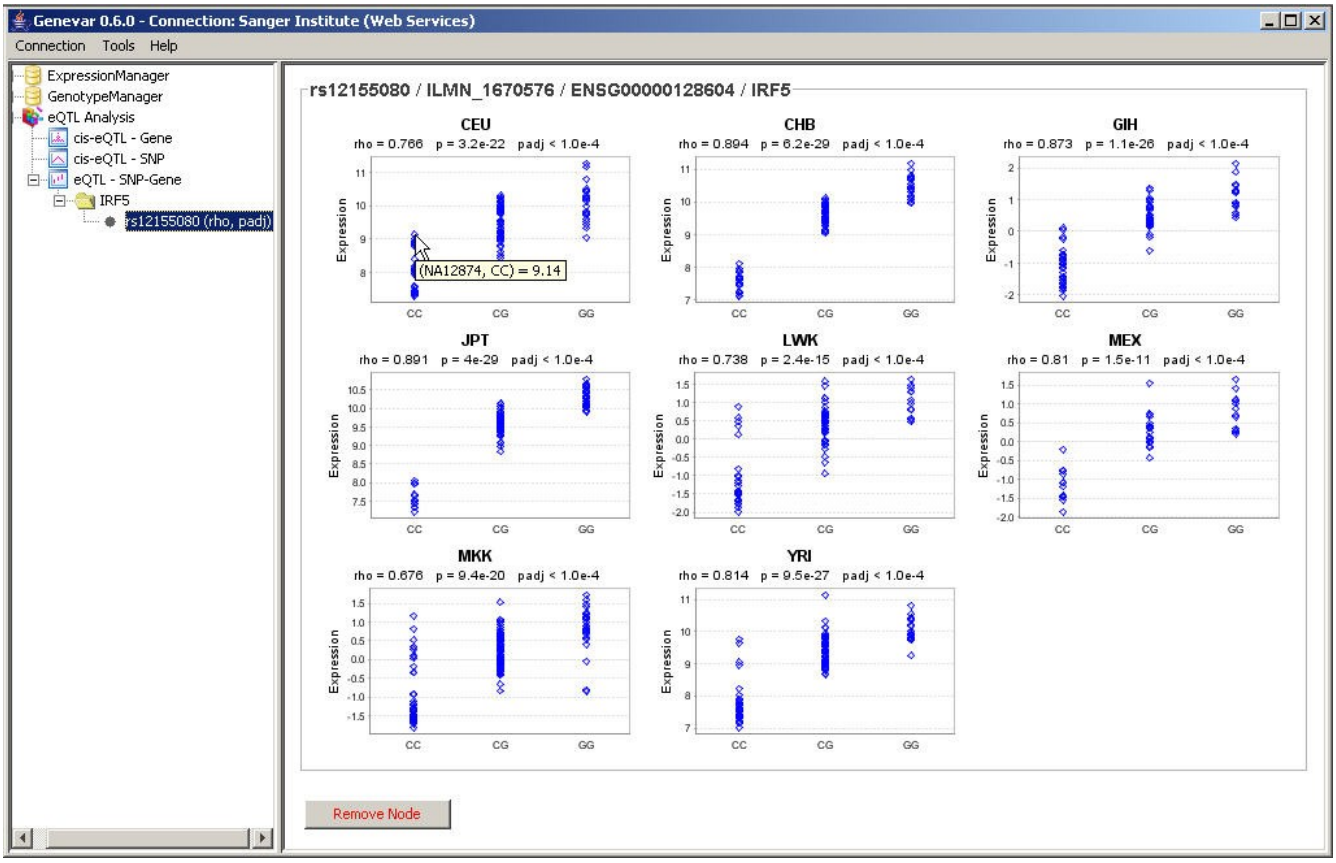
11. Select populations of interest.
12. Select probes of interest.
13. Type in SNP IDs here (**e.g. rs12155080**).
14. Set parameters (**e.g. Spearman's *rho*, 10,000 permutations**).
15. Submit to get SNP-probe association plots.


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
IRF5
Expression-Genotype Pairs
Group: HapMap3
11
Population
Expression
Genotype
CEU
CEU / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9
CEU / HapMap Phase 3.1 / REBUILT-NO-INDEL
CHB
CHB / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9
CHB / HapMap Phase 3.1 / REBUILT-NO-INDEL
GIH
GIH / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9
GIH / HapMap Phase 3.1 / REBUILT-NO-INDEL
JPT
JPT / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9
JPT / HapMap Phase 3.1 / REBUILT-NO-INDEL
LWK
LWK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9
LWK / HapMap Phase 3.1 / REBUILT-NO-INDEL
MEX
MEX / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9
MEX / HapMap Phase 3.1 / REBUILT-NO-INDEL
MKK
MKK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9
MKK / HapMap Phase 3.1 / REBUILT-NO-INDEL
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
12
Probe Id
Gene Name
chr
Gene Start
Gene End
Strand
Probe Start
Gene Symbol
ILMN_1670576
ENSG00000128604
7
128365230
128377320
1
128367567
IRF5
Submit SNP IDs against This Gen
13
rs12155080
dbSNP ID (rs number):
or upload a list of SNPs from a file:
Browse...
Analysis Parameters
14
1. Correlation and regression:
Spearman's rank correlation coefficient (rho)
2. Number of permutations:
10,000
15
Remove Node
SNP-Gene Association Plot

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



SNP-probe association plots.


Genevar 0.6.0 - Connection: Sanger Institute (Web Services)
Connection
Tools
Help
ExpressionManager
GenotypeManager
eQTL Analysis
cis-eQTL - Gene
cis-eQTL - SNP
eQTL - SNP-Gene
IRF5
rs12155080 (rho, padj)
rs12155080 / ILMN_1670576 / ENSG00000128604 / IRF5
CEU
rho = 0.766 p = 3.2e-22 padj < 1.0e-4
(NA12874, CC) = 9.14
CHB
rho = 0.894 p = 6.2e-29 padj < 1.0e-4
GIH
rho = 0.873 p = 1.1e-26 padj < 1.0e-4
JPT
rho = 0.891 p = 4e-29 padj < 1.0e-4
LWK
rho = 0.738 p = 2.4e-15 padj < 1.0e-4
MEX
rho = 0.81 p = 1.5e-11 padj < 1.0e-4
MKK
rho = 0.676 p = 9.4e-20 padj < 1.0e-4
YRI
rho = 0.814 p = 9.5e-27 padj < 1.0e-4
Expression
CC
CG
GG
Remove Node