# 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).

👙 Genevar 0.5.0 - Connection: Sanger	Institute (Web Service)	
Connection Tools Help		
ExpressionManager     GenotypeManager     GenotypeManager     GenotypeManager	Submit SNP IDs	
CIS-eQTL - GENE dis-eQTL - SNP eQTL - SNP-Gene	1. Select a data set: Group Group 2. Annotation source: HapMap3 Geneva GenCovd	
	3. dbSNP ID (rs number):	
	or upload a list of SNPs from a file: Browse	
	Expression-Genotype Pairs	
	Population         Expression         Génotype           Image: Comparison of the second	
	Analysis Parameters         1. Correlation and regression:         Spearman's rank correlation coefficient (rho)         2. Distance seperation upper limit:         1,000,000	
	3. P-value upper limit filter: 0.001	
	Reset All Run	

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

🚔 Genevar 0.5.0 - Connection: Sange	r Institute (Web Service)	
Connection Tools Help		
	Submit SNP IDs	
cis-eqTL - SNP	1. Select a data set: Hapmap3	
eQTL - SNP-Gene	2. Annotation source: Ensembl	
	3. dbSNP ID (rs number):	
	or unload a list of SNDs from a file.	
	Expression-Genotype Pairs	
	Group: Hapmap3 / Humina HumanWG-6 VZ	
	CEU CEU / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9 CEU / HapMap Phase 3.1 / REBUILT-NO-I	
	CHB CHB / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9 CHB / HapMap Phase 3.1 / REBUILT-NO-I	
	GIH GIH / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9 GIH / HapMap Phase 3.1 / REBUILT-NO-IN	
	3 J V J J Mainina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9 J LWK / HapMap Phase 3.1 / REBUILT-NO-I	
	MEX MEX / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9 MEX / HapMap Phase 3.1 / REBUILT-NO-I	
	MKK MKK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9 MKK / HapMap Phase 3.1 / REBUILT-NO-I           VPI         VPI / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9 VPI / HanMan Phase 3.1 / REBUILT-NO-I	
	Analysis Parameters	
	1. Correlation and regression: Spearman's rank correlation coefficient (rho)	
	2. Distance seperation upper limit: 1,000,000	
	3. P-value upper limit filter: 0.001	
	Reset All Run	

- Choose reference source (e.g. Ensembl).
   Type in SNPs here (e.g. rs13277113).
   Select populations of interest.
   Set parameters (e.g. Spearman's rho, 1 Mb, p < 0.001).</li>
   Submit to get transcript lists.

👙 Genevar 0.5.0 - Connection: Sange	er Institute (Web Service)	- D ×
Connection Tools Help		
ExpressionManager     GenotypeManager     GenotypeManager     GenotypeClassec     eQTL Analysis     is-eQTL - Gene     Gs-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: Evences ion. Concerve Pairs	
	Group:       HapMap3 / Illumina HumanWG-6 v2         Population       Expression         CEU       CEU / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         CEU       CEU / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         CHB       CHB / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         CHB       CHB / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         CHB       GIH / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         V       SIH         GIH / JIlumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         V       VK         V       VK         VK       Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         VK       Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         VK       Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         VK       MKK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         VK       MKK         VK       MKK         VRI       YRI / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         VRI       YRI / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         VRI       YRI / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         VRI       YRI / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9         VRI	
	Analysis Parameters          1. Correlation and regression       Spearman's rank correlation coefficient (rho)         2. Distance seperation upper limit:       1,000,000         3. P-value upper limit filte:       0.001         Image: Control of the seperation o	8

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

Senevar 0.5.0 - Connection: Sange Connection Tools Help	r Institute (Web Service)	<u>-0×</u>
ExpressionManager GenotypeManager	Submit SNP IDs	
Piere equit Analysis IsseqTL - Gene IsseqTL - SNP	1. Select a data set: HapMap3	
• rs13277113_rho_1Mb_p0.001	2. Annotation source: Ensembl	
	9 3. dbSNP ID (rs number):	
	or upload a list of SNPs from a file: Browse	
	Expression-Genotype Pairs	
	Group: HapMap3 / Illumina HumanWG-6 v2	
	Image: Second	
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	Image: WKK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9       MKK / HapMap Phase 3.1 / REBUILT-NO-I         Image: WKK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9       YRI / HapMap Phase 3.1 / REBUILT-NO-IN         Image: WKK / Illumina HumanWG-6 v2 / Lymphoblastoid Cell Line / SMV9       YRI / HapMap Phase 3.1 / REBUILT-NO-IN	
	Analusia Davamatana	
	Analysis Parameters     Spearman's rank correlation coefficient (rho)	
	2. Distance seperation upper limit: 1,000,000 (editable, no greater than 1Mb)	
	3. P-value upper limit filter: 0.001	
	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.

ዿ Genevar 0.5.0 - Connection: Sanger Institute (Web Service)										
Connection Tools Help										
ExpressionManager		10,500,0	000 11,00	00,000	11,500	,000	12,0	000,000		
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		ILMN_1806464	ENSG00000184647	8	10420491	10433738	1	10421226	5	
		ILMN_1749047	ENSG00000171060	8	10567557	10595513	1	10568403	C8orf74	
		ILMN_1652521	ENSG00000104643	8	11179410	11223062	1	11186591	MTMR9	
		ILMN_1715988	ENSG00000177710	8	11225905	11227105	1	11226948	8 AMAC1L2	
		ILMN_1713315	ENSG00000177710	8	11225905	11227105	1	11226948	AMAC1L2	
		ILMN_1789248	ENSG00000154316	8	11234597	11263371	1	11235697	TDH	
		ILMN_1668277	ENSG00000136573	8	11388930	11459516	1	11391304	BLK	
		ILMN_1749866	ENSG00000136574	8	11599162	11654918	1	11602424	I GATA4	
		ILMN_1774884	ENSG00000197615	8	11656174	11658141	1	11657693	3 C8orf49	
		ILMN_1715680	ENSG00000154328	8	11664666	11682263	1	11667022	2 NEIL2	
		ILMN_1741096	ENSG0000079459	8	11697599	11734226	1	11699119	FDFT1	
		ILMN_1711790	ENSG00000205883	8	11877239	11879508	1	11877250	)	
		ILMN_1652469	ENSG00000205873	8	12338997	12468794	1	12340877	1	
		ILMN_1806044	ENSG00000205873	8	12338997	12468794	1	12340877		
		ILMN_1673987	ENSG0000205882	8	11888846	11891230	-1	11891021		
		ILMN_1655553	ENSG00000205884	8	11868855	11869517	-1	11869427	CTCD	
		ILIMN_1696360	EN5G0000164733	8	11737442	11/63055	-1	11/60082		
		ILMN_1687213	EN5G00000154319	8	11316391	11369636	-1	1135///9	C80rr13	
		ILMN_1699065	EN5G00000215346	0	1021390	11025155	-1	11023119	VIDC	
		ILIMN_1000440	ENSG0000171044	0	10791075	11096258	-1	11095561		
		TLMN_1724762	ENSG00000171044	0	10791075	11096236	-1	11095561	VVD4	
		TLMN 1700200	ENSC000001/1044	0	10/910/5	10724706		10724124		
		ILMN 1815032	ENSG00000171056	8	10618688	10625432	-1	10622781	50Y7	
		ILMN 1657040	ENSG00000183638	8	10501269	10607107	-1	10542344	RP1L1	
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	Analysis	Parameters —			40					
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	A. S. S. S. S. S.						1		12	<u> </u>
								2-SESSAMATE -		Eb.

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 I	nstitute (Wo	eb Service)								<u> </u>
		10 500 0	11.00	0.000	11 500	000	12			
GenotypeManager		10,000,0	11,00	Chrony	11,000	tion (bp)	127	500,000		
eQTL Analysis										
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E								-		
🖻 🖳 rs13277113_rho_1Mb_p0.001						_			· · · · ·	
<ul> <li>ILMN_1668277 (rho, padj)</li> </ul>	De	select All Exp	ort List			Link to: Ei	nsembl	НарМар	UCSC	
ILMN_1687213 (rho, padj)		Probe Id	Gene Name	chr	Gene Start	Gene End	Strand	Probe Start	Gene Symbol	
eQTL - SNP-Gene		ILMN 1806464	ENSG00000184647	8	10420491	10433738	1	10421226	dene symbol	
. 1.		ILMN 1749047	EN5G00000171060	8	10567557	10595513	1	10568403	C8orf74	
	- >=	ILMN 1652521	ENSG00000104643	8	11179410	11223062	1	11186591	MTMR9	
	17	ILMN 1715988	ENSG00000177710	8	11225905	11227105	1	11226948	AMAC1L2	
		ILMN_1713315	ENSG00000177710	8	11225905	11227105	1	11226948	AMAC1L2	
	Г	ILMN_1789248	ENSG00000154316	8	11234597	11263371	1	11235697	TDH	
	V	ILMN_1668277	ENSG00000136573	8	11388930	11459516	1	11391304	BLK	
		ILMN_1749866	ENSG00000136574	8	11599162	11654918	1	11602424	GATA4	
		ILMN_1774884	ENSG00000197615	8	11656174	11658141	1	11657693	C8orf49	
		ILMN_1715680	ENSG00000154328	8	11664666	11682263	1	11667022	NEIL2	
		ILMN_1741096	ENSG0000079459	8	11697599	11734226	1	11699119	FDFT1	
		ILMN_1711790	ENSG00000205883	8	11877239	11879508	1	11877250	1	
		ILMN Processing				× 12468794	1	12340877		
		ILMN				12468794	1	12340877	1	
			1			11891230	-1	11891021		
		ILMN_1655553	ENSG00000205884	8	11868855	11869517	-1	11869427		
		ILMN_1696360	ENSG00000164733	8	11/3/442	11/63055	-1	11/60082	CISB COmf10	
		ILMN_1667213	ENSG00000154319	0	11021200	11369636	-1	11022110	Coorf13	
		TLMN_1655446	ENSC00000171044	0	1021390	11025155	-1	11025119	YKD6	
	-	ILMN 1724762	ENSG00000171044	8	10791075	11096258	-1	11095561	YKR6	
		ILMN 1731175	ENSG00000171044	8	10791075	11096258	-1	11095561	XKR6	
	- Ê	ILMN 1790309	ENSG00000104637	8	10659885	10734796	-1	10734126		
	Ē	ILMN 1815032	ENSG00000171056	8	10618688	10625432	-1	10622781	SOX7	
		ILMN_1657040	ENSG00000183638	8	10501269	10607107	-1	10542344	RP1L1	
	Analysis	Parameters			· · · · · · · · · · · · · · · · · · ·					
	1. Corr	elation and regression	: Spearman's rank corre	elation coeffi	icient (rho)		*	]		
	2, Num	ber of permutations:	10,000			<b>Y</b>				
K F	Remove	Node					5	SNP-Gene As	sociation Plot	_

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.

Chromosome 1 (H_	_sapiens_Mar_2006) - 1	Integrated Genome Browser				
File View Bookmarks	Tools Help					
Pick Genome	Sequence: chr1	chr1: 66,425,598 - 68,425	5,598			Refresh Data
refseq (+)	PDE4B PDE4B PDE4B PDE4B PDE4B	SGIP1 ├──── <del>₩₩₩</del> ₩₩₩₩		123R Hisebe	ť	
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67,425,598						41.8 MB / 508.1 MB