

Manuscript title: Discovery of expression QTLs using large-scale transcriptional profiling in human lymphocytes

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Supplementary Table 1 Summary of transcript count, detection and density by chromosome

Only autosomal chromosomes are shown. Transcript density was computed based on the chromosomal lengths in NCBI Build 35. See text for details on the statistical test of whether or not a given transcript was significantly detected.

chromosome	all transcripts				RefSeq transcripts				non-RefSeq transcripts			
	# transcripts	# transcripts detected	% transcripts detected	# transcripts detected / Mb	# transcripts	# transcripts detected	% transcripts detected	# transcripts detected / Mb	# transcripts	# transcripts detected	% transcripts detected	# transcripts detected / Mb
1	4,290	1,987	46.3	8.1	2,202	1,408	63.9	5.7	2,088	579	27.7	2.4
2	3,477	1,423	40.9	5.9	1,449	915	63.1	3.8	2,028	508	25.0	2.1
3	2,530	1,098	43.4	5.5	1,238	769	62.1	3.9	1,292	329	25.5	1.6
4	1,949	773	39.7	4.0	849	479	56.4	2.5	1,100	294	26.7	1.5
5	2,405	946	39.3	5.2	1,023	602	58.8	3.3	1,382	344	24.9	1.9
6	2,541	1,095	43.1	6.4	1,181	732	62.0	4.3	1,360	363	26.7	2.1
7	2,359	1,001	42.4	6.3	983	617	62.8	3.9	1,376	384	27.9	2.4
8	1,969	746	37.9	5.1	767	449	58.5	3.1	1,202	297	24.7	2.0
9	1,977	834	42.2	6.0	865	552	63.8	4.0	1,112	282	25.4	2.0
10	2,147	806	37.5	6.0	810	499	61.6	3.7	1,337	307	23.0	2.3
11	2,651	1,203	45.4	8.9	1,370	854	62.3	6.4	1,281	349	27.2	2.6
12	2,229	1,023	45.9	7.7	1,155	744	64.4	5.6	1,074	279	26.0	2.1
13	1,106	381	34.4	3.3	407	224	55.0	2.0	699	157	22.5	1.4
14	1,462	672	46.0	6.3	700	447	63.9	4.2	762	225	29.5	2.1
15	1,273	598	47.0	6.0	683	427	62.5	4.3	590	171	29.0	1.7
16	1,918	912	47.5	10.3	921	622	67.5	7.0	997	290	29.1	3.3
17	2,354	1,152	48.9	14.6	1,261	845	67.0	10.7	1,093	307	28.1	3.9
18	946	327	34.6	4.3	316	183	57.9	2.4	630	144	22.9	1.9
19	2,295	1,291	56.3	20.2	1,476	1,027	69.6	16.1	819	264	32.2	4.1
20	1,409	627	44.5	10.0	708	437	61.7	7.0	701	190	27.1	3.0
21	676	246	36.4	5.2	285	158	55.4	3.4	391	88	22.5	1.9
22	1,117	507	45.4	10.2	526	351	66.7	7.1	591	156	26.4	3.1
autosomes	45,080	19,648	43.6	6.9	21,175	13,341	63.0	4.7	23,905	6,307	26.4	2.2

Supplementary Table 2 Transcript expression phenotypes with the highest *cis* lod scores

Cis lod scores were computed at the integer cM location nearest the genetic map position of a given transcript.

gene symbol	probe ID	chromosome	physical location (bp)	genetic location (cM)	lod	locus-specific heritability estimate
<i>Hs.379903</i>	Hs.379903-S	17	15,820,666	47	52.468	0.775
<i>PPA2</i>	GI_31881619-A	4	106,648,324	112	49.782	0.817
<i>UTS2</i>	GI_12056480-A	1	7,841,942	14	48.192	0.764
<i>UTS2</i>	GI_12056478-I	1	7,841,942	14	44.049	0.718
<i>LR8</i>	GI_21361500-S	7	149,926,039	165	43.206	0.679
<i>UBA52</i>	GI_15451941-S	19	18,543,669	46	43.091	0.762
<i>LOC400566</i>	GI_42661283-S	17	260,331	0	40.087	0.677
<i>Hs.283934</i>	Hs.283934-S	19	11,272,571	33	39.677	0.756
<i>GSTM1</i>	GI_23065543-I	1	109,942,485	132	39.006	0.665
<i>ZNFN1A1</i>	GI_31657112-S	7	50,218,441	74	38.688	0.646
<i>RPS26</i>	GI_15011935-S	12	54,722,194	73	37.659	0.759
<i>GSTM1</i>	GI_23065546-A	1	109,942,485	132	37.157	0.641
<i>TIMM10</i>	GI_6912707-S	11	57,052,513	65	37.019	0.790
<i>LGALS2</i>	GI_5729902-S	22	36,290,767	47	36.335	0.685
<i>RPS26</i>	GI_42657859-S	12	54,722,194	73	36.051	0.691
<i>RPL14</i>	GI_16753224-S	3	40,473,820	66	33.932	0.668
<i>HLA-DRB3</i>	GI_18641371-S	6	32,584,724	53	33.771	0.740
<i>RPS26</i>	GI_42658911-S	12	54,722,194	73	33.639	0.717
<i>HLA-DRB5</i>	GI_26665892-S	6	32,593,130	53	33.119	0.752
<i>HSPC157</i>	GI_7661813-S	1	22,096,990	42	32.714	0.744

Supplementary Table 3 Transcript expression phenotypes with the highest *trans* lod scores

The maximum *trans* lod score is defined as the highest lod score peak obtained in genome-wide linkage analysis, excluding the chromosome on which the gene for a given transcript is located.

gene symbol	probe ID	<i>cis</i> chromosome	maximum <i>trans</i> eQTL				
			chromosome	genetic location (cM)	lod	locus- specific heritability estimate	
<i>MAPK8IP1</i>	GI_20986517-S	11	17	68	22.826	0.621	
<i>LOC401118</i>	GI_42657060-S	4	6	55	18.022	0.682	
<i>SMA3</i>	GI_5803174-S	6	5	87	12.785	0.485	
<i>FLT4</i>	GI_4503752-I	5	11	1	12.708	0.582	
<i>LOC390217</i>	GI_42659696-S	11	12	31	12.185	0.693	
<i>MSI1</i>	GI_25121990-S	12	11	79	11.852	0.480	
<i>KLK3</i>	GI_22208991-A	19	21	67	8.458	0.392	
<i>LOC284233</i>	GI_29744002-S	18	21	8	6.400	0.202	
<i>EPB41L4A</i>	GI_37537711-S	5	6	60	6.390	0.475	
<i>MGC11386</i>	GI_40254998-S	18	8	82	6.280	0.420	
<i>LOC401446</i>	GI_42658811-S	8	12	25	6.061	0.493	
<i>LOC375662</i>	GI_37552401-S	8	7	101	5.636	0.478	
<i>LOC285687</i>	GI_30151807-S	5	6	84	5.569	0.416	
<i>PD2</i>	GI_42476168-S	19	21	48	5.449	0.337	
<i>MPO</i>	GI_4557758-S	17	3	197	5.437	0.282	
<i>BTBD14A</i>	GI_42716306-S	9	20	53	5.424	0.247	
<i>C18orf1</i>	GI_4757883-S	18	4	207	5.420	0.481	
<i>SCRIB</i>	GI_33186909-A	8	15	91	5.414	0.369	
<i>SEPN1</i>	GI_28827777-S	1	6	56	5.410	0.439	
<i>NIPA</i>	GI_21361544-S	7	8	85	5.325	0.395	

Supplementary Table 4 Heritability estimates, *cis* lod scores and maximum *trans* lod scores for all transcripts

The comma-delimited text file contains information on the probes and the corresponding gene symbols, their physical and genetic map locations, as well as the obtained heritability estimates, *cis* lod scores and maximum *trans* lod scores. Information is provided for all 19,648 autosomal transcripts that were found to be significantly expressed. Probes are listed in chromosomal order by the start location of the probe (in base pairs).

[electronic file Supplementary_Table_4.csv]

Supplementary Table 5 Primer sequences

See main text for details on the assays.

a. Amplification of *VNNI* promoter

direction	oligonucleotide sequence
sense	5'-CTAAATCGAATTGGTCCTATGATTG-3'
antisense	5'-AACAGAGCATGTCCTGGTATTTATAG-3'

b. Electrophoretic mobility shift assay of *VNNI* promoter variant

promoter variant	oligonucleotide sequence
<i>VNNI</i> -137T	5' ATCTTCACAAATTACTGCAATAAGGTAGGGTCTTTTGTTATGTAACAATG 3'
<i>VNNI</i> -137G	5' ATCTTCACAAATTACTGCAATAAGGGAGGGTCTTTTGTTATGTAACAATG 3'

Supplementary Figure 1 Relationship of heritability estimate and *cis* eQTL-specific heritability estimate

Note that the probability that message abundance is subject to *cis* regulation is greatly influenced by the estimated heritability. Among transcripts with $h^2 \geq 43\%$, significant evidence for *cis* regulation (based on a lod score threshold of 0.588) is observed for ~50% of transcripts, among those with $h^2 \geq 60\%$ the proportion is ~80%, and all 42 transcripts with $h^2 \geq 70\%$ are *cis*-regulated.

