



SCAN

SNP and CNV Annotation Database

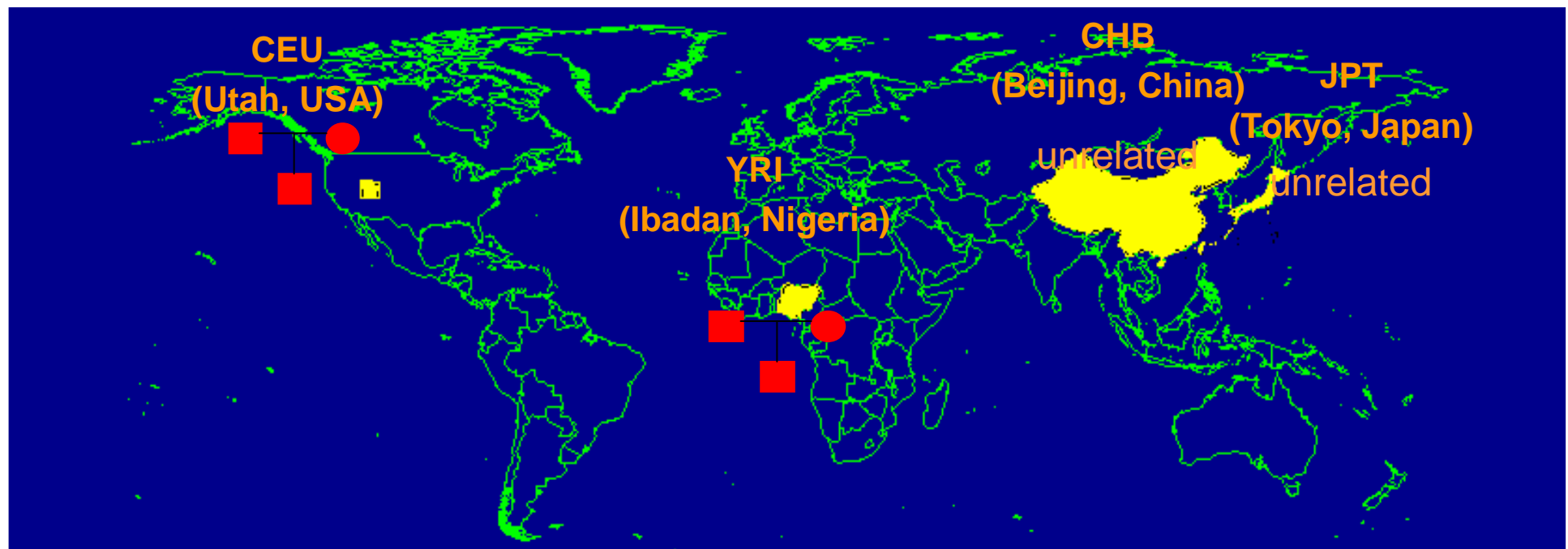
What is SCAN?

- For each SNP, the SCAN database provides:
 - (1) Summary information from eQTL mapping of HapMap SNPs to gene expression (evaluated by the Affymetrix exon array) in the full set of HapMap CEU (Caucasians from Utah, USA) and YRI (Yoruba people from Ibadan, Nigeria) samples;
 - (2) LD information, including what genes have variation in strong LD (pairwise or multilocus LD) with the variant and how well the SNP is covered by different high-throughput platforms;
 - (3) Summary information available from public databases (e.g., physical and functional annotations); and
 - (4) Summary information from other GWAS.
- For each gene, SCAN provides annotations on:
 - (1) eQTLs for the gene (both local and distant SNPs); and
 - (2) The coverage of all variants in the HapMap at that gene on each high-throughput platform.
- For each genomic region, SCAN provides annotations on:
 - (1) Physical and functional annotations of all SNPs, genes, and known CNVs within the region; and
 - (2) All genes regulated by the eQTLs within the region.



What are the HapMap Samples?

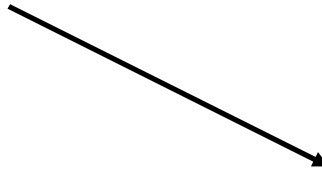
Epstein Barr Virus-transformed lymphoblastoid cell lines derived from apparently healthy individuals of African (YRI), European (CEU) and Asian (CHB and JPT) ancestry



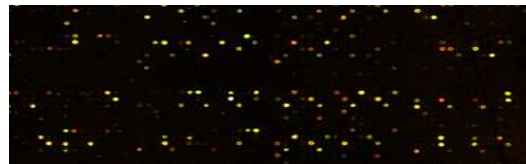
Profiling of mRNA Expression

Affymetrix Human Exon Array 1.0ST
(~18,000 Genes)

87 CEU samples in trios
89 YRI samples in trios



mRNA expression
(~10,000 expressed gene-level transcripts)



Published, GSE9703



SCAN

SNP and CNV Annotation Database

Home

Gene

SNP

Region

LD Annotations

Enter a gene or a set of genes:

CAPN10

or choose a file with a list of genes:

include gene start, end, and chromosome

include SNPs inside gene and up to kb from the gene

include SNPs that predict expression with p-value less than

and frequency greater than

for population

Restrict to eQTLs on the same chromosome

Enter list of genes (white space or comma separated) or upload a file with list of genes. Use official gene symbol (assigned by HUGO).



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Enter a gene or a set of genes:

CAPN10

or choose a file with a list of genes:

Browse...

- include gene start, end, and chromosome
- include SNPs inside gene and up to 2 kb from the gene
- include SNPs that predict expression with p-value less than 0.0001 and frequency greater than .10 for population

All

Restrict to eQTLs on the same chromosome

HTML table output

Submit

Select options indicating what to include in the results set.



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Enter a gene or a set of genes:

CAPN10

Specify p-value threshold.

or choose a file with a list of genes:

Browse...

include gene start, end, and chromosome

include SNPs inside gene and up to 2 kb from the gene

include SNPs that predict expression with p-value less than 0.0001

and frequency greater than .10

for population

All

Restrict to eQTLs on the same chromosome

HTML table output

Submit



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Home Queries

Click on the gene for more info.

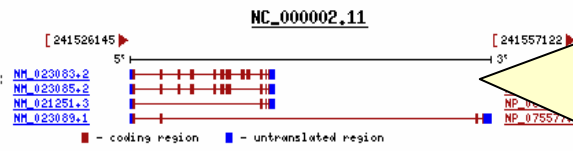
Download the result. Here is a list of SNPs that predict the expression of the gene in LCLs and associated p-value.

gene_name	start	end	chromosome	SNPs	expression_SNPs
CAPN10	241174817	241205794	2	rs56244291	rs716181 CEU 2e-05
				rs55650580	rs716183 CEU 3e-05
				rs11556469	rs7069690 CEU 4e-05
				rs60704894	rs10886046 CEU 0.0001
				rs7596933	rs11197911 CEU 7e-05
				rs4676420	rs11591353 CEU 2e-05
				rs11692507	rs11819305 CEU 7e-05
				rs12619657	rs12763760 CEU 7e-05
				rs55999124	rs17095748 CEU 5e-05
				rs10680220	rs1983265 CEU 0.0001
				rs35428509	rs4945441 CEU 8e-05
				rs10684680	rs7112239 CEU 0.0001
				rs35883707	rs1006496 CEU 9e-05
				rs55893551	rs2682783 CEU 5e-05
				rs55976923	rs2730750 CEU 0.0001
				rs56351903	rs4238055 CEU 0.0001
				rs12620571	rs7969675 CEU 4e-05
				rs61210947	rs11061820 CEU 3e-05
				rs3792263	rs524566 CEU 0.0001
				rs12620622	rs11627062 CEU 0.0001
				rs12475968	rs7165671 CEU 8e-05
				rs56267113	rs29869 CEU 9e-05
				rs41266967	rs12185206 CEU 0.0001
				rs34871679	rs12917862 CEU 5e-05
				rs41266969	rs12923102 CEU 4e-05
				rs2975756	rs1558080 CEU 0.0001
				rs2975757	rs12606900 CEU 8e-05
				rs2975758	rs352815 CEU 0.0001
				rs2975759	rs352818 CEU 0.0001

General Information

Search Gene: Go

Coding Region/Exon/Intron, RNA product, protein product:



GeneID: 11132
 Name: CAPN10
 Gene Type: protein-coding
 Description: calpain 10
 Status: 0
 Chromosome: 2
 Start: 241174817
 End: 241205794
 Strand: +
 Map Location: 2q37.3
 Locus Tag: -
 Other Designations: OTTHUMP00000164509|calcium-activated neutrophil
 Nomenclature: calpain 10
 Chr Accession Ver: [NC_000002.10](#)

Queried gene (in red) and neighboring genes (in grey), including orientation (using NCBI's API):



Image of gene showing alternative splicing, coding regions, and gene products

Map of gene relative to neighboring genes. Queried gene in red.

Platform Coverage

Summary Information

Platform	Population	Avg. Multi-locus LD
Affy100K	CEU	0.37
Affy100K	YRI	0.421
Affy6	CEU	0.874
Affy6	YRI	0.452
ill1M	CEU	0.894
ill1M	YRI	0.545
ill650K	CEU	0.863
ill650K	YRI	0.477

HapMap SNPs (up to 2KB of CAPN10)

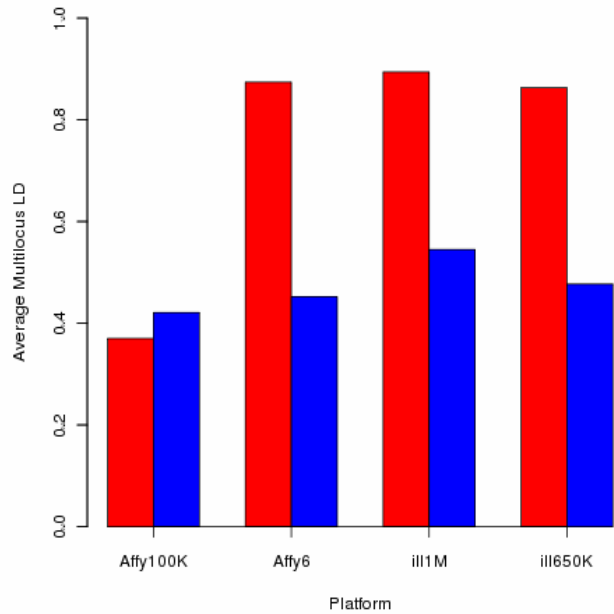
[rs3749164](#) [rs2975757](#) [rs2975758](#) [rs2975759](#) [rs2975760](#) [rs3792267](#) [rs3792269](#) [rs3792270](#) [rs2975761](#) [rs2975762](#) [rs2953166](#) [rs4234123](#) [rs7556712](#) [rs4676348](#) [rs2953161](#) [rs2975770](#) [rs10933620](#) [rs2953147](#) [rs10933621](#)

Measures of (high throughput) platform coverage using multilocus LD

Md Distribution of CAPN10 on Select Platforms

Platform	Population	Average Multi-locus LD	Q1 Multi-locus LD	Median Multi-locus LD	Q3 Multi-locus LD	No. of HapMap SNPs (MAF > .05)	No. of HapMap SNPs (MAF <= .05)
Affy6	CEU	0.874	0.747	0.995	1	26	2
Affy6	YRI	0.452	0.264	0.434	0.526	16	12
ill1M	CEU	0.894	0.729	1	1	26	2
ill1M	YRI	0.545	0.279	0.478	0.747	16	12

Comparison of Platforms for CAPN10 (CEU = Red, YRI = Blue)



Enter list of SNPs to query or upload a file containing list of SNPs.

Enter SNPs (rs numbers):

```
rs7069690
rs10886046
rs11197911
rs11591353
```

or choose a file with a list of SNPs:

Browse...

include SNP info
 include host gene and SNP function
 include left- and right- flanking genes
 include genes that SNP predicts expression for with p-value less than

0.0001

HTML table output format

Submit

Select options indicating what annotations to include in result set.

rsnum	chromosome	position	alleles	gene	function	left_gene	right_gene	expression_gene_(population_a
rs7069690	10	27298646	C/T	NA	NA	LOC100131912	ANKRD26	PLOD3 CEU 0.0001 ENO1 CEU 9e-05 UBE2D2 CEU 8e-05 JAGN1 CEU 8e-05 SARS CEU 0.0001 PH-4 CEU 9e-05 TP53RK CEU 0.0001 YWHAG CEU 2e-05 CBS CEU 0.0001 HIST3H2A CEU 0.0001 PRMT2 CEU 4e-05 YIF1A CEU 5e-05 C11orf2 CEU 3e-05 C11orf3 CEU 3e-05 NEU1 CEU 0.0001 RDBP CEU 0.0001 MFN2 CEU 3e-05 C9orf51 CEU 3e-05 MGC12966 CEU 3e-05 VLDLR CEU 5e-05 B4GALT2 CEU 0.0001 FTS1 CEU 0.0001 C8orf58 CEU 9e-05 NIN1 CEU 0.0001 MON1A CEU 5e-05 IFRD2 CEU 3e-05 NAT6 CEU 3e-05 CAPN10 CEU 4e-05 GPR35 CEU 4e-05 EEF2 CEU 0.0001 SNORD37 CEU 0.0001 ASB8 CEU 0.0001 ATP6Y0B CEU 6e-05 TMED4 CEU 1e-05 PEMT CEU 0.0001 CD151 CEU 2e-06 ABCF3 CEU 8e-05 TRIM27 CEU 9e-05 NT5DC2 CEU 6e-05
rs10886046	10	118905850	A/G	NA	NA	YAX1	KCNK18	CDK3 CEU 4e-05 CAPN10 CEU 0.0001 GPR35 CEU 0.0001 ZNF70 CEU 3e-05 IMMT CEU 8e-05

The list of genes for which the given SNP is an eQTL and the p-value of the SNP-expression association



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A region can be specified as a chromosome:start:end
or by a pair of SNPs from the same chromosome as snpid1:snpid2

Enter a region or a set of regions:

2:100:2000 5:400000:800000

or choose a file with a list of regions:

Browse...

- include SNPs
 - include genes
 - include left- and right- flanking genes
 - include genes regulated by SNPs in region, with p-value less than
- 0.0001
- HTML table output format

Submit

Enter list of regions or upload a file with list of regions. A region may be specified as chromosome:start:end or as rsNum1:rsNum2 for a pair of SNPs on the same chromosome.

More Information

Citation info:

SCAN: SNP and CNV Annotation

Eric R. Gamazon¹, Wei Zhang¹, Anuar Konkashbaev¹, Shiwei Duan¹, Emily O. Kistner⁴, Dan L. Nicolae^{1,2}, M. Eileen Dolan¹, Nancy J. Cox^{1,3*}

Department of Medicine¹, Department of Statistics², Department of Human Genetics³, and Department of Health Studies⁴, The University of Chicago, Chicago, IL 60637, USA.

Technical Questions: egamazon@medicine.bsd.uchicago.edu

Request of Reprints: ncox@medicine.bsd.uchicago.edu