



ENCODE Use Cases: Using SCREEN to Generate Hypotheses About Disease-associated Variants

Mike Pazin

Program Director, Functional Genomics, Division of Genome Sciences

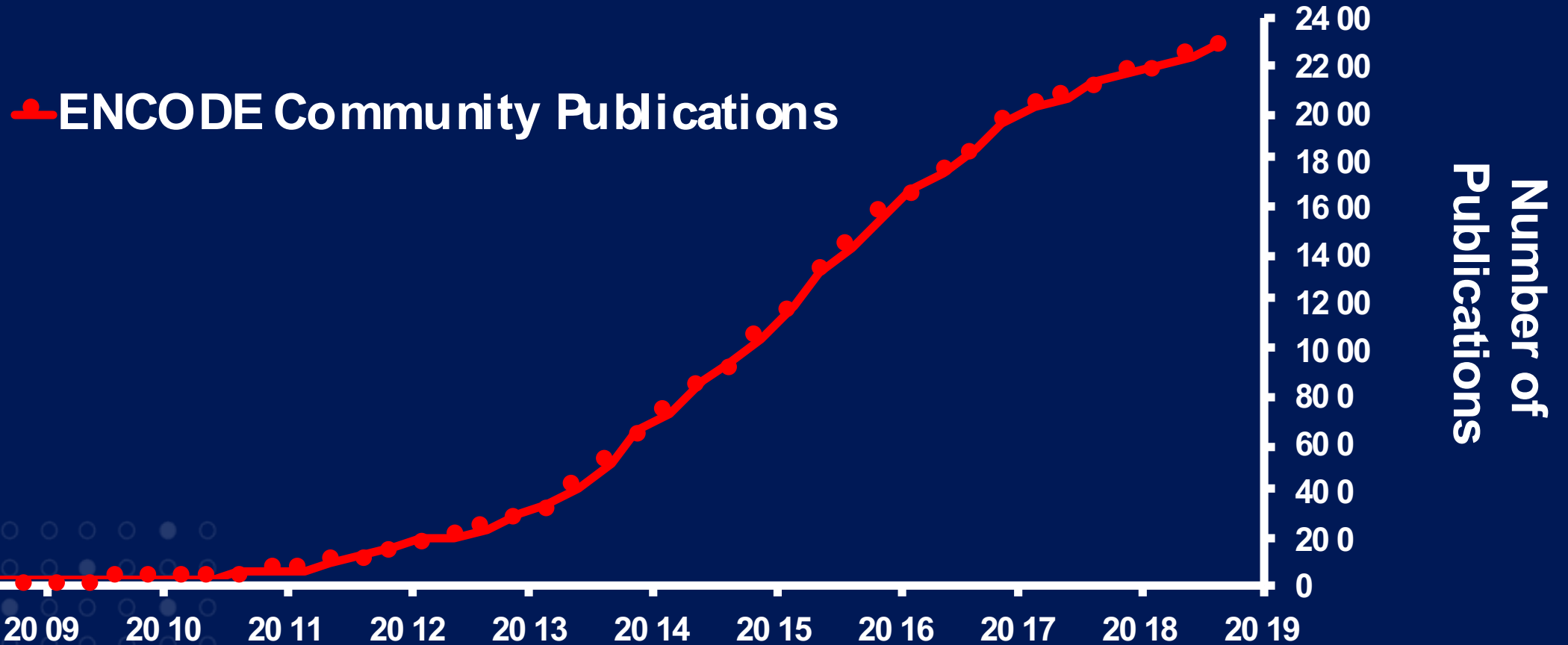
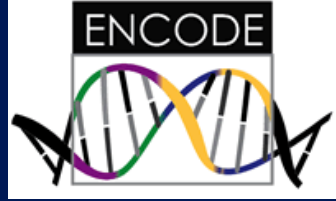
16 October 2018



National Human Genome
Research Institute

The **Forefront**
of **Genomics**

ENCyclopedia Of DNA Elements (ENCODE)



<https://www.encodeproject.org/publications/>



ENCODE Use Cases

- Protocols and Approaches
- Methods development/validation
- Disease association/gene regulation studies
 - Affected cell type
 - Upstream regulators
 - Target gene
 - Causal variant

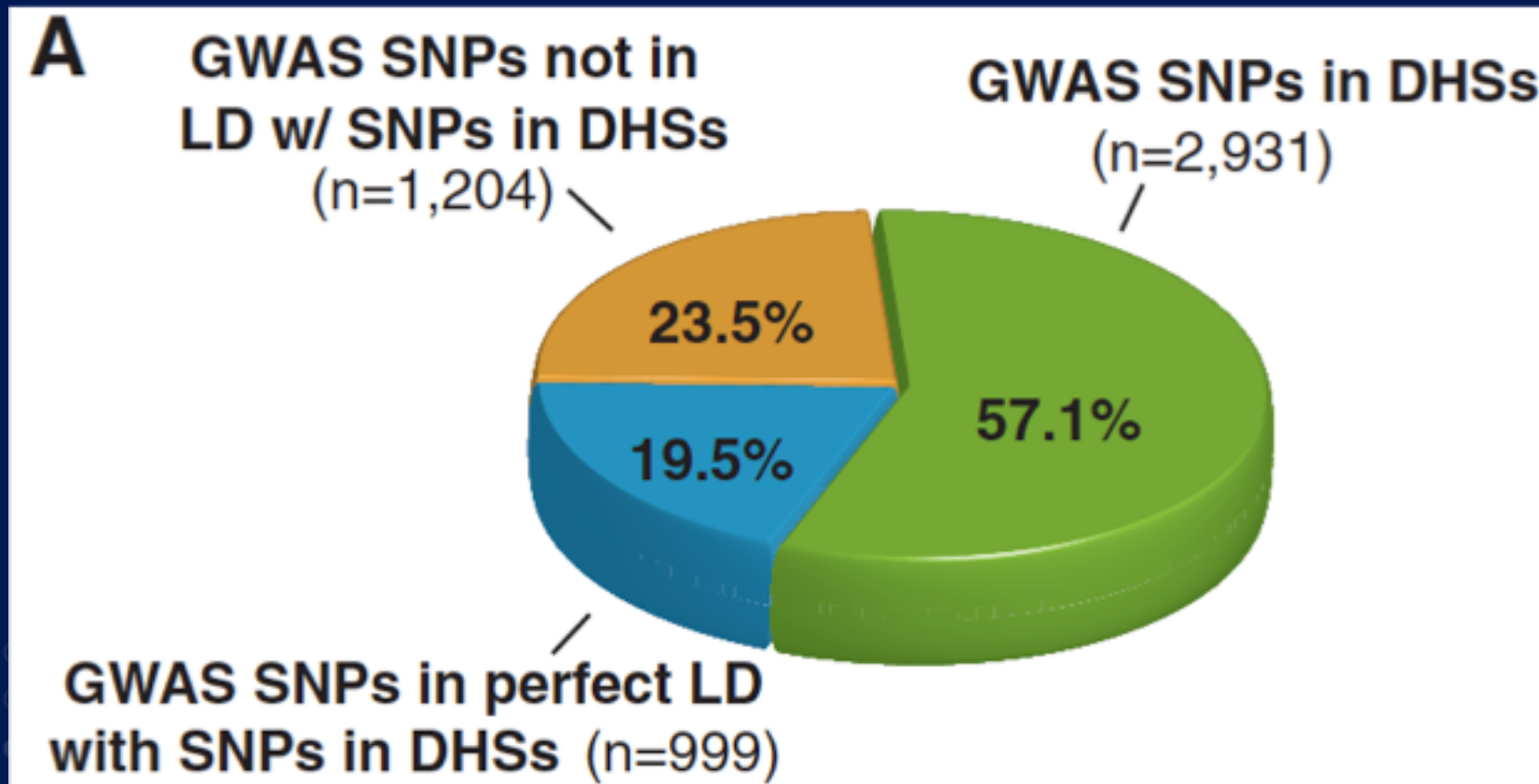
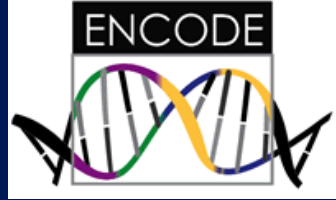


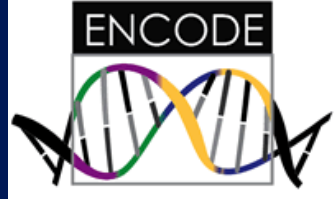
Non-coding DNA Is Important For Disease And Gene Regulation

- Vast majority of common disease associations and heritability appear to lie outside of protein-coding regions
- Non-coding DNA variants are known to cause human diseases and alter human traits (FXS, ALS)

Functional information is needed to interpret the role of genetic variation in human disease, and to apply genomics in the clinic.

Many GWAS Associations Lie In Regions Annotated By ENCODE And Roadmap Epigenomics Data






Using Tools With ENCODE Data

- Portal
- SCREEN
- HaploReg
- RegulomeDB
- Other tools that incorporate ENCODE Data (e.g. CADD, Annovar, JASPAR, Ensembl Variant Effect Predictor, Ensembl Regulatory Build)
- Tracks



ENCODE/Roadmap Data Using HaploReg

HaploReg v4.1 

HaploReg is a tool for exploring annotations of the noncoding genome at variants on haplotype blocks, such as candidate regulatory SNPs at disease-associated loci. Using LD information from the 1000 Genomes Project, linked SNPs and small indels can be visualized along with chromatin state and protein binding annotation from the Roadmap Epigenomics and ENCODE projects, sequence conservation across mammals, the effect of SNPs on regulatory motifs, and the effect of SNPs on expression from eQTL studies. HaploReg is designed for researchers developing mechanistic hypotheses of the impact of non-coding variants on clinical phenotypes and normal variation.


Update 2015.11.05: Version 4.1 GWAS and eQTL have been updated; a simpler pruning strategy is applied when combining GWAS; and links out to other NHGRI/EBI GWAS hits and GRASP QTL hits are provided.

Update 2015.09.15: Version 4.0 now includes many recent eQTL results including the GTEx pilot, four different options for defining enhancers using Roadmap Epigenomics data, and a complete set of source files for download and local analysis. Older versions available: [v3](#), [v2](#), [v1](#).

[Build Query](#) [Set Options](#) [Documentation](#)

Use one of the three methods below to enter a set of variants. If an r^2 threshold is specified (see the Set Options tab), results for each variant will be shown in a separate table along with other variants in LD. If r^2 is set to NA, only queried variants will be shown, together in one table.

Query (comma-delimited list of rsIDs
OR a single region as chrN:start-end):
or, upload a text file (one refSNP ID per line):

 1

no file selected

Query SNP: **rs16892766** and variants with $r^2 \geq 0.8$

chr	pos (hg38)	LD (r ²)	LD (D)	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNase	Proteins bound	Motifs changed	NHGRI/EBI GWAS hits	GRASP QTL hits	Selected eQTL hits	GENCODE genes	dbSNP func annot
8	116618444	1	1	rs16892766	A	C	0.12	0.08	0.00	0.09		FAT	STRM, LNG, GI	7 tissues	FOXA1,GR	Rhox11	2 hits	1 hit	3 hits	24kb 3' of EIF3H	
8	116618773	0.97	1	rs200235517	CG	C	0.47	0.10	0.04	0.09			7 tissues			lrf				23kb 3' of EIF3H	
8	116618774	0.97	1	rs58147231	GA	G	0.47	0.10	0.04	0.09			7 tissues			lrf			1 hit	23kb 3' of EIF3H	
8	116623363	0.89	0.97	rs16888589	A	G	0.12	0.08	0.00	0.09		ESDR	8 tissues			Ik-1,STAT			3 hits	19kb 3' of EIF3H	

<https://pubs.broadinstitute.org/mammals/haploreg/haploreg.php>

Ward and Kellis, Nucleic Acids Research 40-D930, 2011

ENCODE/Roadmap Data From RegulomeDB



RegulomeDB

Enter dbSNP IDs, 0-based coordinates, BED files, VCF files, GFF3 files (hg19).

rs16892766

Submit

RegulomeDB

The search has evaluated 1 input line(s) and found

Summary of SNP analysis

Show 1 entries

Coordinate (0-based)	dbSNP ID	Regulome DB Score
chr8:117630682	rs16892766	2b

Showing 1 to 1 of 1 entries

Download BED GFF Full Output

A project of the Center for Genomics and Personalized Medicine at

Protein Binding

Method	Location	Bound Protein	Cell Type	Additional Info	Reference
ChIP-seq	chr8:117630539..117630739	FOXA1	ECC-1	DMSO_0.02pct	ENCODE
ChIP-seq	chr8:117630626..117630842	NR3C1	ECC-1	DEX_100nM	ENCODE

Chromatin structure

Method	Location	Cell Type	Additional Info	Reference
DNase-seq	chr8:117630480..117630690	Rptec		ENCODE
DNase-seq	chr8:117630480..117630730	Nrlf		ENCODE
DNase-seq	chr8:117630500..117630710	Nha		ENCODE
DNase-seq	chr8:117630500..117630770	Hah		ENCODE
DNase-seq	chr8:117630510..117630704	Aosmc	Serum	ENCODE
DNase-seq	chr8:117630520..117630790	Hvmlf		ENCODE
DNase-seq	chr8:117630625..117631002	Hrb		ENCODE
FAIRE	chr8:117630519..117630761	Medulo		ENCODE

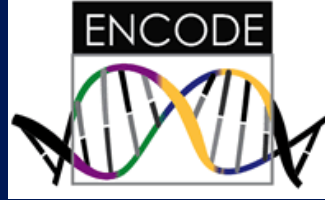
Histone modifications

Method	Location	Histone Mark	Cell Type	Additional Info	Reference
ChIP-seq	chr8:110578383..117647033	H3k09me3	Dnd41		ENCODE
ChIP-seq	chr8:116009496..120997897	H2az	Hepg2		ENCODE
ChIP-seq	chr8:117409399..118413945	H4k20me1	Hmec		ENCODE
ChIP-seq	chr8:117555446..118475451	H4k20me1	Nrlf		ENCODE
ChIP-seq	chr8:117384699..117650386	H2az	Dnd41		ENCODE

<http://www.regulomedb.org>

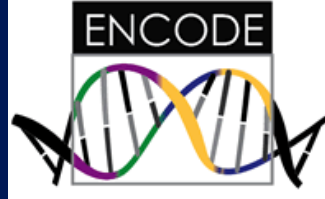
Cherry, Snyder, Genome Research 22-1790,2012

Accessing SCREEN from ENCODE portal



The screenshot shows the ENCODE portal's 'Encyclopedia' menu with 'Visualize (SCREEN)' selected. Below the menu is a diagram titled 'DE: Encyclopedia of DNA Elements'. The diagram illustrates the relationship between various genomic features and the data used to identify them. At the top, a DNA double helix is shown with 'Hypersensitive Sites' (orange), 'CH₃' (blue) and 'CH₃CO' (yellow) methyl groups, and 'RNA polymerase' (green) bound to it. Below the DNA, a series of blue boxes represent different data types: '5C ChIA-PET Hi-C', 'DNase-seq FAIRE-seq ATAC-seq', 'ChIP-seq', 'WGBS RRBS methyl array', 'Computational predictions', 'RNA-seq', and 'CLIP-seq RIP-seq'. Arrows point from these boxes to a genomic track at the bottom, which is divided into 'Long-range regulatory elements (enhancers, repressors/silencers, insulators)', 'Promoters', 'Genes', and 'Transcripts'. The ENCODE logo is in the bottom left, and a citation is at the bottom center: 'Based on an image by Darryl Leja (NHGRI), Ian Dunham (EBI), Michael Pazin (NHGRI)'.

SCREEN



SCREEN: Search Candidate cis-Regulatory Elements by ENCODE

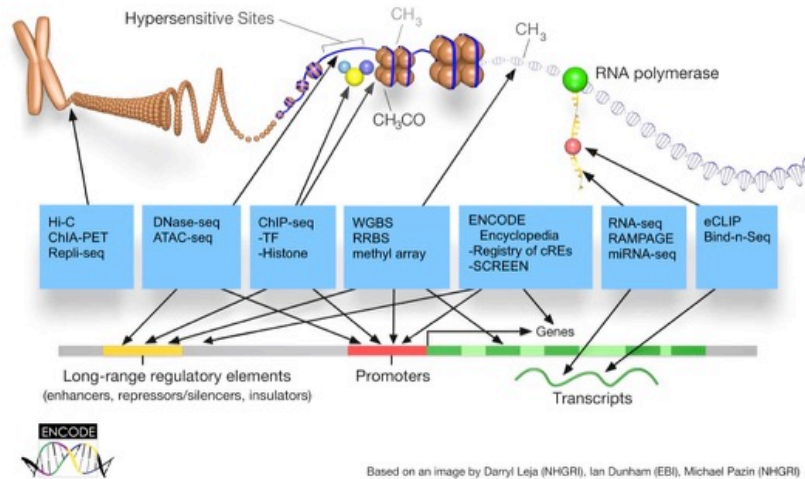
Overview

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Based on an image by Darryl Leja (NHGRI), Ian Dunham (EBI), Michael Pazin (NHGRI)

SCREEN is a web interface for searching and visualizing the Registry of candidate cis-Regulatory Elements (ccREs) derived from [ENCODE data](#). The Registry contains 1.31M human ccREs in hg19 and 0.43M mouse ccREs in mm10, with orthologous ccREs cross-referenced. SCREEN presents the data that support biochemical activities of the ccREs and the expression of nearby genes in specific cell and tissue types.

You may launch SCREEN using the search box below or browse a curated list of SNPs from the NHGRI-EBI Genome Wide Association Study (GWAS) catalog to annotate genetic variants using ccREs.

[Browse GWAS](#)

Enter a gene name or alias, a SNP rsID, a ccRE accession, or a genomic region in the form chr:start-end. You may also enter a cell type name to filter results.

Examples: "K562 chr11:5226493-5403124", "SOX4", "rs4846913", "EH37E0204974"

Search Human
(hg19)

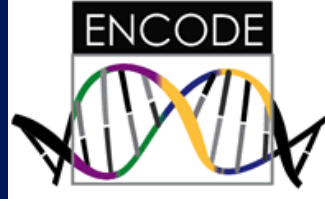
Search Mouse
(mm10)

© 2017 Weng Lab @ UMass Med, ENCODE Data Analysis Center

Zhiping Weng, Jill Moore, Michael Purcaro, Henry Pratt

<http://screen.encodeproject.org>

SCREEN



SCREEN: Search Candidate cis-Regulatory Elements by ENCODE

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How SCREEN was constructed

How to use SCREEN

Download sets of SCREEN elements

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ChIA-PET Repli-seq ATAC-seq CHIP-seq -TF -Histone WGBS RRBS methyl array ENCODE Encyclopedia -Registry of cREs -SCREEN RAMPAGE miRNA-seq Bind-n-Seq

Long-range regulatory elements (enhancers, repressors/silencers, insulators) Promoters Transcripts

Based on an image by Darryl Leja (NHGRI), Ian Dunham (EBI), Michael Pazin (NHGRI)

chr11:5226493-5403124

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[Search Human \(hg19\)](#) [Search Mouse \(mm10\)](#)

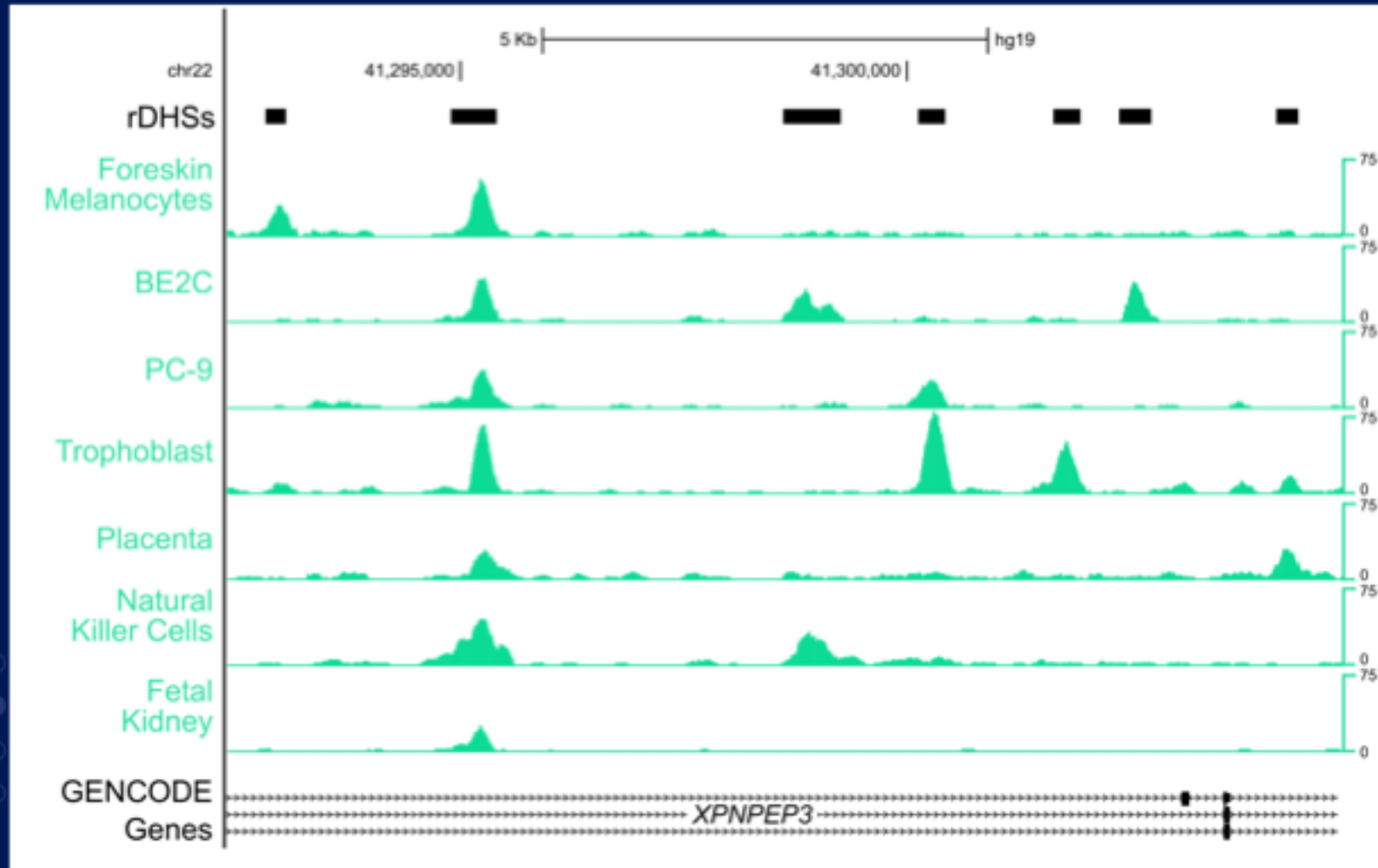
© 2017 Weng Lab @ UMass Med, ENCODE Data Analysis Center



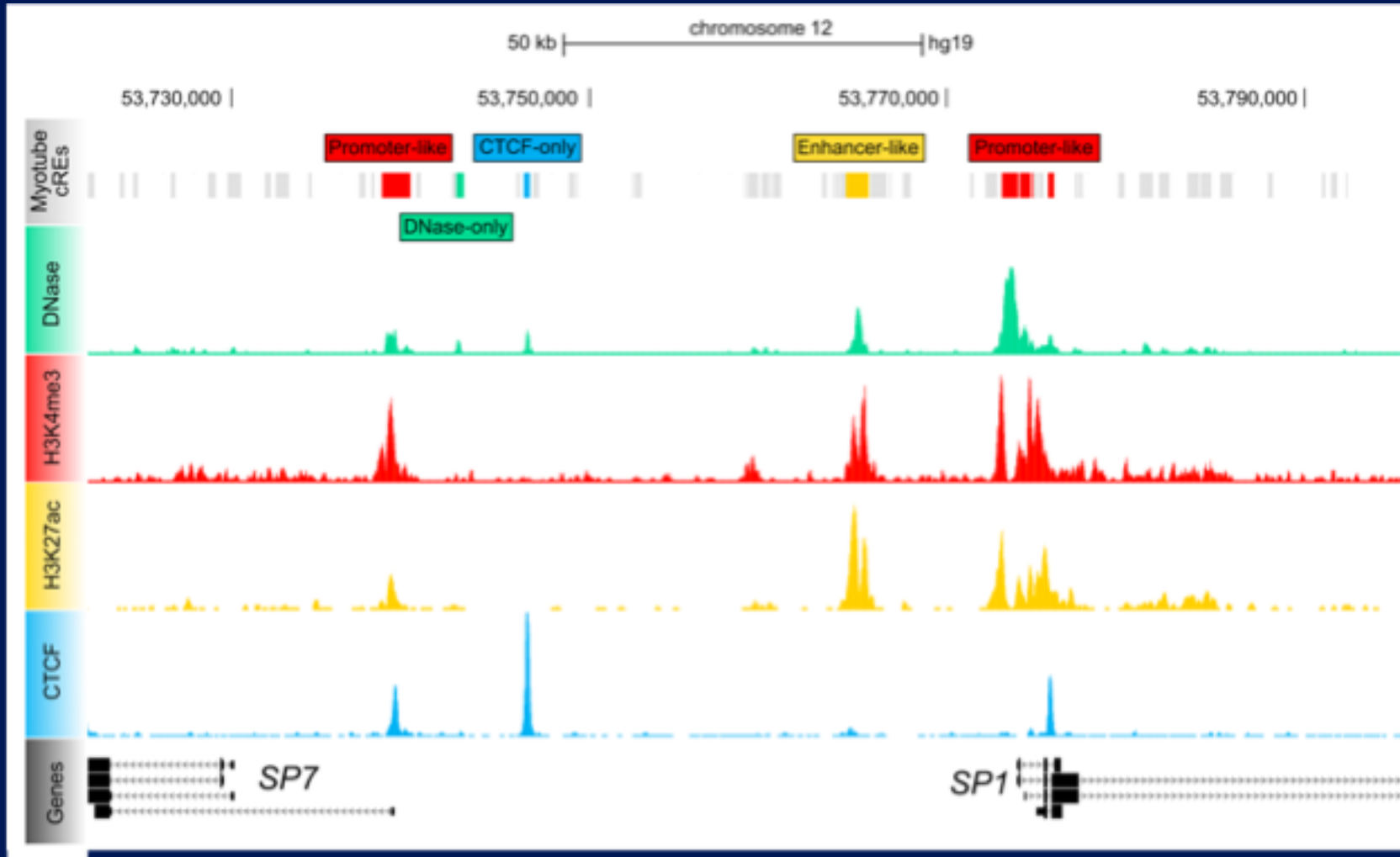
ENCODE Registry of Candidate cis-Regulatory Elements (ccREs)

- Genomic regions that may function as regulatory elements including promoters, enhancers, and boundary elements
- Data from hundreds of cell and tissue types
- Registries in both human and mouse
- Data from ENCODE, Roadmap Epigenomics, and FANTOM

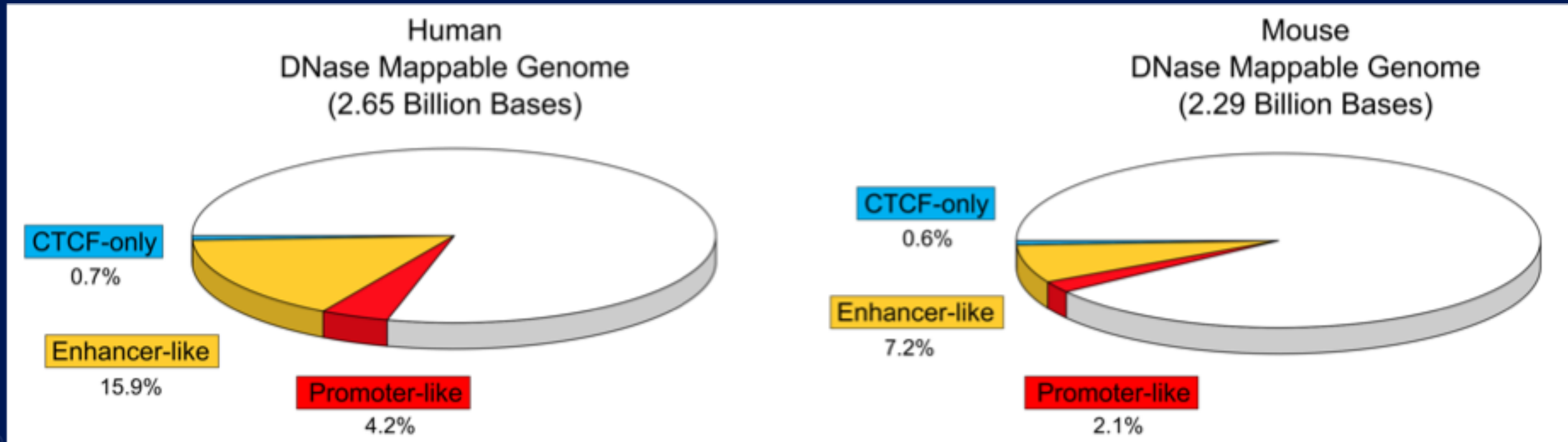
Index of Candidate Regulatory Elements Based on DNase From 100's of Cell Types



Annotating Elements with Histone Modification And CTCF Data



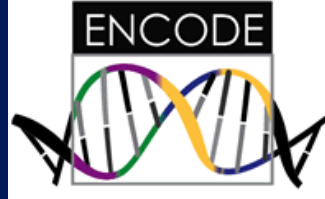
Human and mouse candidate regulatory elements



About 1.3 million

About 400k

SCREEN Queries



SCREEN: Search Candidate cis-Regulatory Elements by ENCODE

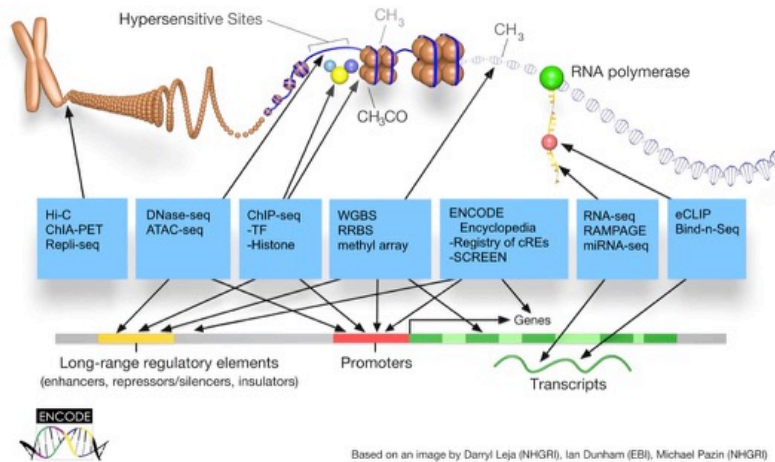
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Search Human
(hg19)

Search Mouse
(mm10)

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SCREEN: ccRE Table



SCREEN hg19 chr11:5226493-5403124 Search

Biosamples i

TSV Search:

cell type	tissue
<input type="radio"/> A172	brain
<input type="radio"/> A549	lung
<input type="radio"/> A549 treated with dexamethasone	lung
<input type="radio"/> A549 treated with ethanol	lung
<input type="radio"/> A673	muscle
<input type="radio"/> ACC112	salivary glands
<input type="radio"/> adipocyte	adipose
<input type="radio"/> adipose derived mesenchymal stem cell in vitro differentiated cells	stem cell
<input type="radio"/> adrenal gland female adult (51 years)	adrenal
<input type="radio"/> adrenal gland female fetal (108 days)	adrenal

Total: 622 « < 1 2 3 ... 63 > »

ccRE Search Results Bed Upload

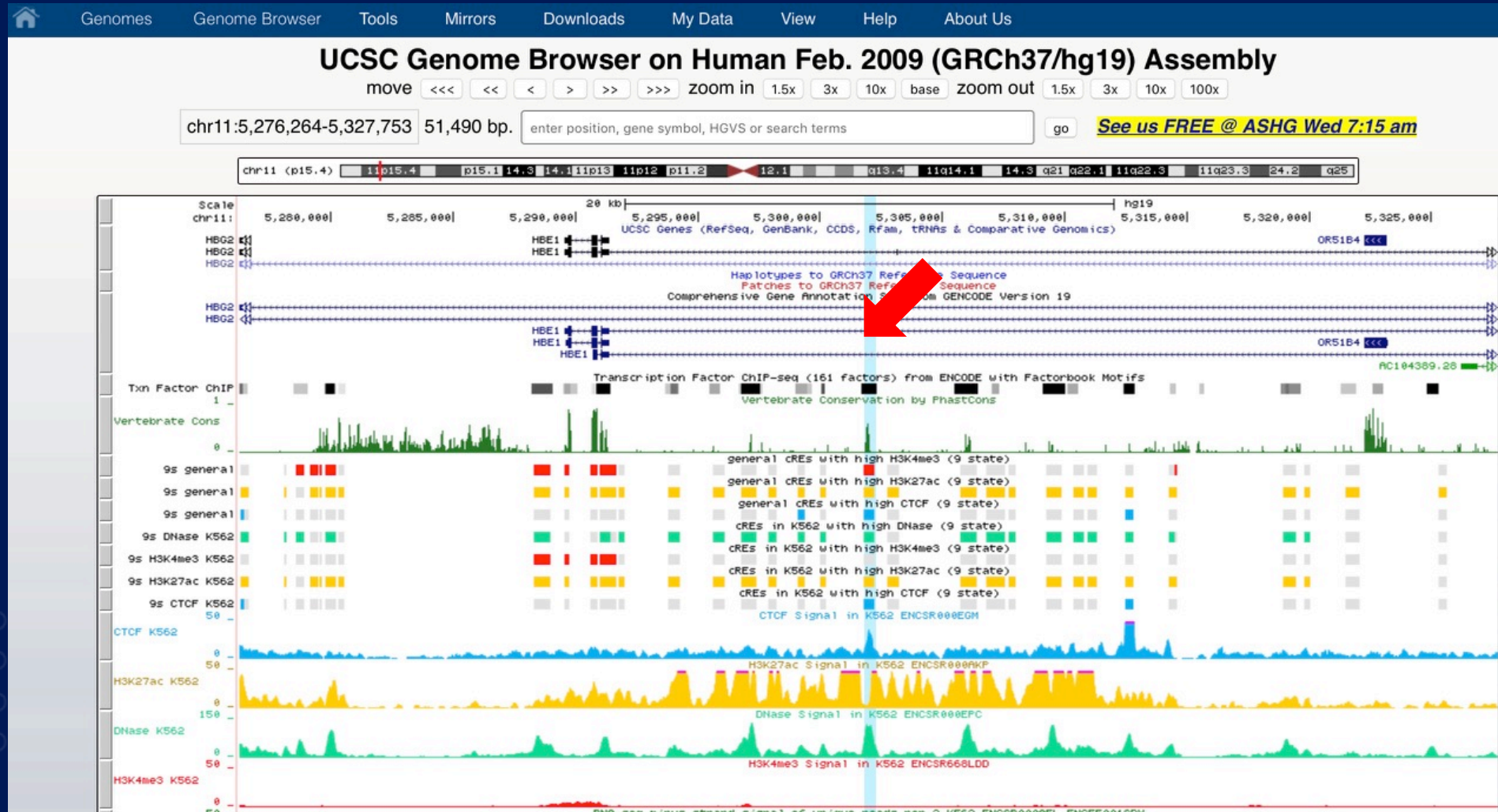
Candidate cis-Regulatory Elements (ccREs) that meet your search criteria are listed in the table below.

- Click a ccRE accession to view details about the ccRE, including top tissues, nearby genomic features, etc.
- Click a gene ID to view the expression profile of the gene.

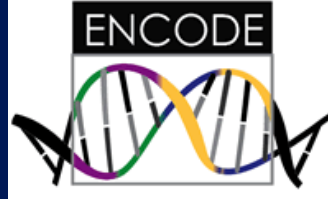
Search:

accession	DNase Z	H3K4me3 Z	H3K27ac Z	CTCF Z	chr	start	length	nearest genes: protein-coding / all	cart	genome browsers
<input type="radio"/> EH37E1090133 P ■ ■ ■	5.33	2.42	1.63	1.37	chr11	5,247,589	1,032	pc: HBB, HBD, HBG1 all: HBB, CoTC_ribozyme, HBD		UCSC
<input type="radio"/> EH37E0204932 ★ D ■ ■ ■	4.20	1.91	3.59	2.19	chr11	5,301,786	446	pc: HBE1, OR51B4, HBG2 all: HBE1, OR51B4, HBG2		UCSC
<input type="radio"/> EH37E0204974 ★ D ■ ■ ■	3.73	1.26	2.28	5.28	chr11	5,401,971	367	pc: OR51M1, OR51J1, OR51B6 all: OR51M1, OR51J1, OR51B6		UCSC
<input type="radio"/> EH37E0204969 ★ D ■ ■ ■	3.53	2.17	4.64	1.56	chr11	5,388,661	199	pc: OR51B6, OR51M1, OR51B5 all: OR51B6, OR51M1, OR51B5		UCSC
<input type="radio"/> EH37E0204963 ★ D ■ ■ ■	3.53	1.93	3.72	1.32	chr11	5,385,367	492	pc: OR51B6, OR51B5, OR51M1 all: OR51B6, OR51B5, OR51M1		UCSC
<input type="radio"/> EH37E0204970 ★ D ■ ■ ■	3.46	2.04	5.03	1.20	chr11	5,388,866	595	pc: OR51B6, OR51M1, OR51B5 all: OR51B6, OR51M1, OR51B5		UCSC
<input type="radio"/> EH37E0204972 ★ D ■ ■ ■	3.30	1.45	1.82	5.57	chr11	5,391,140	230	pc: OR51B6, OR51M1, OR51B5 all: OR51B6, OR51M1, OR51B5		UCSC
<input type="radio"/> EH37E0204928 ★ D ■ ■ ■	3.17	1.36	3.47	1.53	chr11	5,296,800	608	pc: HBE1, HBG2, HBG1 all: HBE1, HBG2, HBG1		UCSC

UCSC Browser View of SCREEN Tracks



SCREEN: ccRE Table



SCREEN hg19 chr11:5226493-5403124 Search

Biosamples *i* ccRE Search Results Bed Upload

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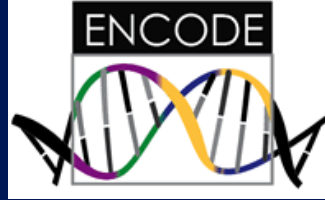
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ccRE Details



SCREEN hg19 chr11:5226493-5403124

ccRE Search Results Bed Upload ccRE Details

EH37504932 chr11:5,301,786-5,302,232 ★ D

Top Tissues Nearby Genomic Features TF and His-mod Intersection FANTOM Intersection Associated Gene Expression Associated RAMPAGE Signal Orthologous ccREs in mm10 Signal Profile Linked Genes

H3K4me3 Z-scores **H3K27ac Z-scores**

TSV Search:

cell type	H3K4me3 and DNase	H3K4me3 only
A549 treated with ethanol	--	1.91
common myeloid progenitor CD34+ male adult (42 years)	2.71	1.37
myotube	2.05	1.36
mucosa of stomach male adult (59 years)	--	1.21
common myeloid progenitor cultured	--	1.17
iPS-18 cell	--	1.04
OCI-LY3	--	1.02
foreskin fibroblast male newborn	2.13	0.99
Panc1	2.42	0.93
H7-hESC	1.66	0.92

Total: 210

H3K27ac Z-scores

TSV Search:

cell type	H3K27ac and DNase	H3K27ac only
K562	3.48	3.59
common myeloid progenitor CD34+ female	3.50	3.36
HCT116	3.27	2.72
fibroblast of lung male fetal (12 weeks)	3.30	2.65
fibroblast of lung (11 years and 45 years)	--	2.57
bipolar spindle neuron derived from induced pluripotent stem cell	2.69	2.50
astrocyte	2.81	2.35
T helper memory cells from peripheral blood 2	--	2.25
endothelial cell of umbilical vein newborn	--	2.22
PC-9	2.22	2.20

Total: 136

CTCF Z-scores **DNase Z-scores**

Biosamples

TSV Search:

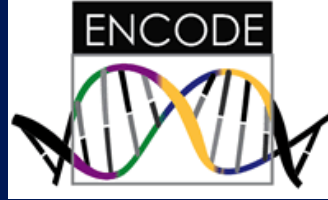
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adipocyte	adipose
adipose derived mesenchymal stem cell in vitro differentiated cells	stem cell
adrenal gland female adult (51 years)	adrenal
adrenal gland female fetal (108 days)	adrenal

Total: 622

Chromosome

chr11

Views Within ccRE Details Tab



SCREEN hg19 chr11:5226493-5403124 Search

ccRE Search Results Bed Upload ccRE Details

EH37E0204932 chr11:5,301,786-5,302,232 ★ D

Top Tissues Nearby Genomic Features TF and His-mod Intersection FANTOM Intersection Associated Gene Expression Associated RAMPAGE Signal Orthologous ccREs in mm10 Signal Profile **Linked Genes**

H3K4me3 Z-scores

TSV Search:

cell type	H3K4me3 and DNase	H3K4me3 only
A549 treated with ethanol	--	1.91
common myeloid progenitor CD34+ male adult (42 years)	2.71	1.37
myotube	2.05	1.36
mucosa of stomach male adult (59 years)	--	1.21
common myeloid progenitor cultured	--	1.17
iPS-18 cell	--	1.04
OCI-LY3	--	1.02
foreskin fibroblast male newborn	2.13	0.99
Panc1	2.42	0.93
H7-hESC	1.66	0.92

Total: 210

H3K27ac Z-scores

TSV Search:

cell type	H3K27ac and DNase	H3K27ac only
K562	3.48	3.59
common myeloid progenitor CD34+ female	3.50	3.36
HCT116	3.27	2.72
fibroblast of lung male fetal (12 weeks)	3.30	2.65
fibroblast of lung (11 years and 45 years)	--	2.57
bipolar spindle neuron derived from induced pluripotent stem cell	2.69	2.50
astrocyte	2.81	2.35
T helper memory cells from peripheral blood 2	--	2.25
endothelial cell of umbilical vein newborn	--	2.22
PC-9	2.22	2.20

Total: 136

CTCF Z-scores

DNase Z-scores

Biosamples

TSV Search:

cell type	tissue
A172	brain
A549	lung
A549 treated with dexamethasone	lung
A549 treated with ethanol	lung
A673	muscle
ACC112	salivary glands
adipocyte	adipose
adipose derived mesenchymal stem cell in vitro differentiated cells	stem cell
adrenal gland female adult (51 years)	adrenal
adrenal gland female fetal (108 days)	adrenal

Total: 622

Chromosome

chr11

Nearby Genomic Features

Biosamples ⓘ

TSV Search:

cell type	tissue
<input type="radio"/> A172	brain
<input type="radio"/> A549	lung
<input type="radio"/> A549 treated with dexamethasone	lung
<input type="radio"/> A549 treated with ethanol	lung
<input type="radio"/> A673	muscle
<input type="radio"/> ACC112	salivary glands
<input type="radio"/> adipocyte	adipose
<input type="radio"/> adipose derived mesenchymal stem cell in vitro differentiated cells	stem cell
<input type="radio"/> adrenal gland female adult (51 years)	adrenal
<input type="radio"/> adrenal gland female fetal (108 days)	adrenal

Total: 622

Chromosome: chr11

ccRE Search Results | Bed Upload | ccRE Details

EH37E0204933 chr11:5,301,786-5,302,232 ★ D

Top Tissues | **Nearby Genomic Features** | TAD and His-mod Intersection | FANTOM Intersection | Associated Gene Expression | Associated RAMPAGE Signal | Orthologous ccREs in mm10 | Signal Profile

Nearby Genes ⓘ

Search:

symbol	distance
HBE1	10,398
OR51B4	20,994
HBG2	25,745
HBG1	30,664
CTD-2643I7.1	35,009
OR51B2	43,350

Total: 6

Nearby ccRES ⓘ

Search:

accession	distance
EH37E0204933	961
EH37E0204931	1,753
EH37E0204930	2,700
EH37E0204929	3,898
EH37E0204934	3,940
EH37E0204928	4,824
EH37E0204935	5,033
EH37E0204936	5,752
EH37E0204927	6,116
EH37E0204937	7,450

Total: 519

Nearby SNPs ⓘ

TSV Search:

accession	distance
rs57169181	37
rs143052503	58
rs7119428	152
rs9736333	170
rs12292063	174
rs10837757	188
rs60240093	237
rs61388411	335
rs3919413	637
rs4288782	642

Total: 11

Genes within TAD ⓘ


Search:

symbol

Other ccRES within TAD and <100 kb ⓘ

Search:

accession distance



22

Transcription Factor And Histone Modifications



SCREEN hg19 chr11:5226493-5403124 Search

ccRE Search Results Bed Upload ccRE Details

EH37E0204932 chr11:5,301,781-5,302,232 ★ D

Top Tissues Nearby Genomic Features TF and His-mod Intersection FANTOM Intersection Associated Gene Expression Associated RAMPAGE Signal Orthologous ccREs in mm10 Linked Genes

TFs that bind this ccRE Search:

factor	# experiments that support TF binding	# experiments in total
JUND	9	13
MAFK	7	9
JUN	7	8
MYC	6	11
FOS	6	6
POLR2A	6	81
NFE2L2	4	4
TCF12	4	8
SMARCA4	4	4
MNT	3	7

Total: 150

Histone Marks at this ccRE Search:

mark	# of experiments that support histone modification	# experiments in total
H3K4me1	52	248
H3K27ac	24	213
H3K4me2	13	40
H2AFZ	6	43
H3K4me3	3	316
H3K27me3	3	245
H3K9ac	3	93
H3K36me3	2	251
H4K91ac	1	6
H2BK20ac	1	3

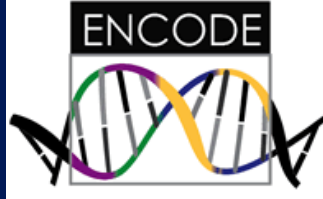
Total: 17

Biosamples Search:

cell type	tissue
A172	brain
A549	lung
A549 treated with dexamethasone	lung
A549 treated with ethanol	lung
A673	muscle
ACC112	salivary glands
adipocyte	adipose
adipose derived mesenchymal stem cell in vitro differentiated cells	stem cell
adrenal gland female adult (51 years)	adrenal
adrenal gland female fetal (108 days)	adrenal

Total: 622

Link Out To Factorbook TF Resource



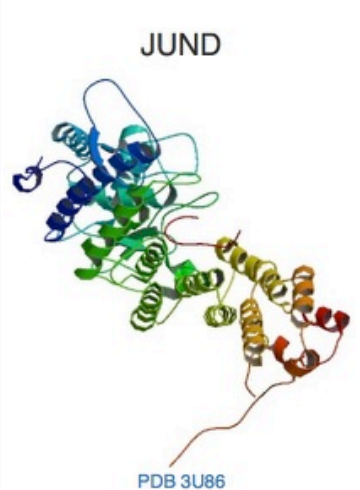
Factorbook **human** mouse

JUND

Function [Histone Profiles](#) [Motif Enrichment](#) [Histone Heatmaps](#) [TF Heatmaps](#) [Nucleosome Profiles](#)

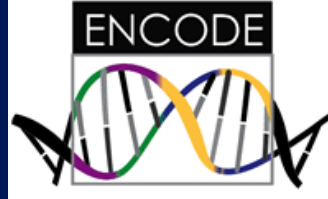
The protein encoded by this intronless gene is a member of the JUN family, and a functional component of the AP1 transcription factor complex. This protein has been proposed to protect cells from p53-dependent senescence and apoptosis. Alternative translation initiation site usage results in the production of different isoforms (PMID:12105216).
— RefSeq, Nov 2013

Transcription factor JunD is a protein that in humans is encoded by the JUND gene.
— wikipedia



PDB	3U86
ENCODE	experiments
Ensemble	search
Entrez	3727
GO	search
Gene Card	search
HGNC	search
RefSeq	search
UCSC	browse
UniProt	search
Wikipedia	JUND

Gene Expression



Biosamples ⓘ

TSV Search:

cell type	tissue
<input type="radio"/> A172	brain
<input type="radio"/> A549	lung
<input type="radio"/> A549 treated with dexamethasone	lung
<input type="radio"/> A549 treated with ethanol	lung
<input type="radio"/> A673	muscle
<input type="radio"/> ACC112	salivary glands
<input type="radio"/> adipocyte	adipose
<input type="radio"/> adipose derived mesenchymal stem cell in vitro differentiated cells	stem cell
<input type="radio"/> adrenal gland female adult (51 years)	adrenal
<input type="radio"/> adrenal gland female fetal (108 days)	adrenal

Total: 622

« < 1 2 3 ... 63 > »

Chromosome

chr11

Coordinates: chr11:5226493-5403124 ⓘ

ccRE Bed ccRE
Search Results Upload Details

EH37E0204932 chr11:5,301,786-5,302,232 ★ D

Top Nearby TF and His-mod FANTOM Associated Associated Orthologous ccREs Signal Linked
Tissues Genomic Features Intersection Intersection Gene Expression RAMPAGE Signal in mm10 Profile Genes

HBE1 Gene Expression Profile by RNA-seq ⓘ

Group by

Experiment Tissue **Tissue Max**

Biosample Types

TSV

cell line

in vitro differentiated cells

induced pluripotent stem cell line

primary cell

stem cell

tissue

Cellular Compartments

TSV

cell

chromatin

cytosol

membrane

nucleolus

nucleoplasm

nucleus

TPM/FPKM

TPM

FPKM

Scale

Linear

Log2

Replicates

Ind. Avg.

Tissue of origin

Download figure

Tissue	Expression Value	ENCSR ID	Cell Line / Condition
blood	11.51	ENCSR000CPH	K562
uterus	3.83	ENCSR000BYB	Ishikawa
lung	2.2	ENCSR000COO	AG04450
embryo	2.14	ENCSR500UOD	mesodermal cell
brain	2.02	ENCSR000CPN	SK-N-SH
connective tissue	0.52	ENCSR000CPM	fibroblast of lung
liver	0	ENCSR329MHM	HepG2
epithelium	0	ENCSR859HWB	hepatocyte
muscle	0	ENCSR000COY	skeletal muscle myoblast
blood vessel	0	ENCSR000COZ	endothelial cell of umbilical vein
heart	0	ENCSR294NDO	cardiac muscle cell
pancreas	0	ENCSR000BYM	Panc1
mammary gland	0	ENCSR000BYE	T47D

Orthologous ccRE

SCREEN hg19

Biosamples i

TSV Search:

cell type	tissue
<input type="radio"/> A172	brain
<input type="radio"/> A549	lung
<input type="radio"/> A549 treated with dexamethasone	lung
<input type="radio"/> A549 treated with ethanol	lung
<input type="radio"/> A673	muscle
<input type="radio"/> ACC112	salivary glands
<input type="radio"/> adipocyte	adipose

ccRE Search Results | Bed Upload | ccRE Details

EH37E0204932 chr11:5,301,786-5,302,232 ★ D

[Top Tissues](#)
[Nearby Genomic Features](#)
[TF and His-mod Intersection](#)
[FANTOM Intersection](#)
[Associated Gene Expression](#)
[Associated RAMPAGE Signal](#)
[Orthologous ccREs in mm10](#)
[Signal Profile](#)
[Linked Genes](#)

Search:

accession	chromosome	start	end	overlap
EM10E0298925	chr11	5,301,812	5,302,217	405 bp

Total: 1

Orthologous ccRE

SCREEN mm10

Accessions

TSV Search:

Assay

EM10E0298925

Total: 1

Biosamples ⓘ

TSV Search:

cell type	tissue
<input type="radio"/> 129.DLCR liver male embryo (14.5 days)	liver
<input type="radio"/> 129 E14TG2a.4	ESC
<input type="radio"/> 129 ES-E14	ESC
<input type="radio"/> 129 G1E	blood
<input type="radio"/> 129 liver male embryo (14.5 days)	liver
<input type="radio"/> 129 ZHBTc4-mESC treated with doxycycline hyclate	ESC

ccRE Bed

Candidate cis-Regulatory Elements (ccREs) that meet your search criteria are listed in the table below.

- Click a ccRE accession to view details about the ccRE, including top tissues, nearby genomic features, etc.
- Click a gene ID to view the expression profile of the gene.
- Click the Δ following a gene ID to explore the differential expression of the gene between two cell types.

Search:

accession ⓘ	DNase Z	H3K4me3 Z	H3K27ac Z	CTCF Z	chr	start	length	nearest genes: protein-coding / all ⓘ	cart	genome browsers
<input type="radio"/> EM10E0298925 ★ D ■ ■ ■	4.21	2.65	4.81	2.86	chr7	103,860,917	398	pc: Hbb-y Δ , Hbb-bh1 Δ , Hbb-bh2 Δ all: Hbb-y Δ , Hbb-bh1 Δ , Hbb-bh2 Δ	<input type="button" value="cart"/>	<input type="button" value="UCSC"/>

Add all to cart Clear cart Download bed Download JSON found 1 results

■ High H3K4me3
■ P/D Proximal/Distal to a Transcription Start Site

■ High H3K27ac

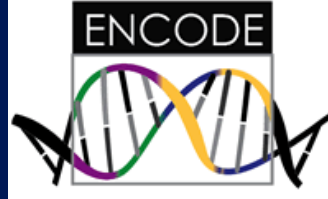
■ High CTCF
★ High DNase and High H3K4me3, H3K27ac, or CTCF in the same cell type

■ High DNase

Z-score < 1.64

No data

Linked Genes



SCREEN hg19 chr11:5226493-5403124 Search

ccRE Search Results Bed Upload ccRE Details

EH37E0204932 chr11:5,301,786-5,302,232 ★ D

Top Tissues Nearby Genomic Features TF and His-mod Intersection FANTOM Intersection Associated Gene Expression Associated RAMPAGE Signal Orthologous ccREs in mm10 Signal Profile **Linked Genes**

Linked Genes Search:

gene	biosample	supporting exp	based on
HBG1	K562	H3K4me3_ChIA-PET	
HBG2	K562	H3K4me3_ChIA-PET	
HBBP1	K562	POLR2A_ChIA-PET	
CTD-264317.1	K562	POLR2A_ChIA-PET	
HBE1	K562	POLR2A_ChIA-PET	
OR51B4	K562	POLR2A_ChIA-PET	
CoTC_ribozyme	K562	POLR2A_ChIA-PET	
HBB	K562	POLR2A_ChIA-PET	
HBG1	K562	POLR2A_ChIA-PET	

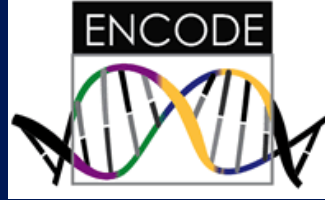
Total: 9

Biosamples: TSV Search: []

cell type	tissue
<input type="radio"/> A172	brain
<input type="radio"/> A549	lung
<input type="radio"/> A549 treated with dexamethasone	lung
<input type="radio"/> A549 treated with ethanol	lung
<input type="radio"/> A673	muscle
<input type="radio"/> ACC112	salivary glands
<input type="radio"/> adipocyte	adipose
<input type="radio"/> adipose derived mesenchymal stem cell in vitro differentiated cells	stem cell
<input type="radio"/> adrenal gland female adult (51 years)	adrenal
<input type="radio"/> adrenal gland female fetal (108 days)	adrenal

Total: 622

Downloading SCREEN ccRE



SCREEN: Search candidate cis-Regulatory Elements by ENCODE

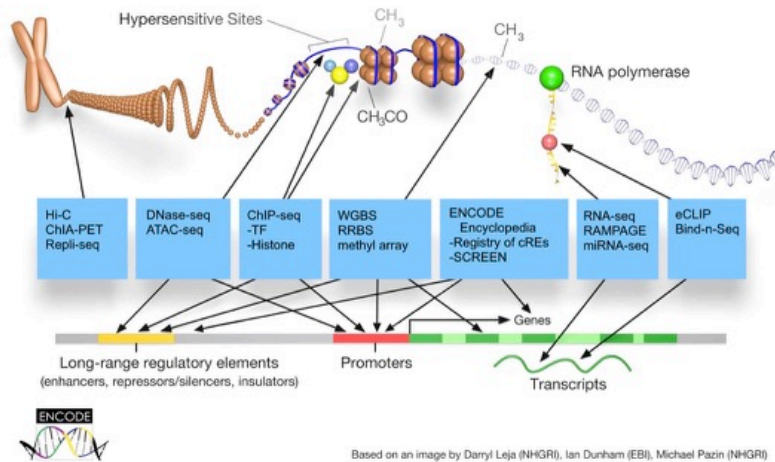
Overview

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Based on an image by Darryl Leja (NHGRI), Ian Dunham (EBI), Michael Pazin (NHGRI)

SCREEN is a web interface for searching and visualizing the Registry of candidate cis-Regulatory Elements (ccREs) derived from ENCODE data. The Registry contains 1.31M human ccREs in hg19 and 0.43M mouse ccREs in mm10, with orthologous ccREs cross-referenced. SCREEN presents the data that support biochemical activities of the ccREs and the expression of nearby genes in specific cell and tissue types.

You may launch SCREEN using the search box below or browse a curated list of SNPs from the NHGRI-EBI Genome Wide Association Study (GWAS) catalog to annotate genetic variants using ccREs.

[Browse GWAS](#)

Enter a gene name or alias, a SNP rsID, a ccRE accession, or a genomic region in the form chr:start-end. You may also enter a cell type name to filter results.

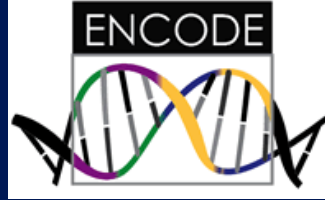
Examples: "K562 chr11:5226493-5403124", "SOX4", "rs4846913", "EH37E0204974"

Search Human
(hg19)

Search Mouse
(mm10)

© 2017 Weng Lab @ UMass Med, ENCODE Data Analysis Center

Downloading SCREEN ccRE



SCREEN: Search Candidate cis-Regulatory Elements by ENCODE

Overview About Tutorials Downloads Versions

Cell type-agnostic

Search:

Assembly	5 group	9 state high H3K27ac	9 state high H3K4me3	9 state high CTCF
hg19				
mm10				

Total: 2

Cell type-specific

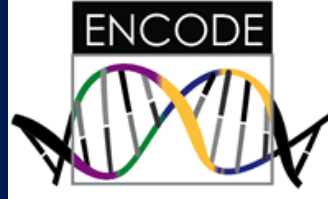
Search:


Assembly	Tissue	Biosample	5 group	9 state high DNase	9 state high H3K27ac	9 state high H3K4me3	9 state high CTCF
hg19	umbilical cord	umbilical cord male fetal (76 days)			-	-	-
hg19	thymus	thymus male fetal (108 days)			-	-	-
hg19	spinal cord	spinal cord female fetal (113 days)			-	-	-
hg19	thymus	thymus female fetal (105 days)			-	-	-
hg19	muscle	muscle of leg male fetal (96 days)			-	-	-
hg19	placenta	placenta female fetal (85 days)			-	-	-
hg19	ESC	H9 stably expressing fusion protein		-			-
hg19	thyroid	thyroid gland male adult (54 years)					-
hg19	blood	natural killer cell female adult (34 years)			-	-	-
hg19	thyroid	thyroid gland male adult (37 years)				-	-

Total: 756

« < 1 2 3 ... 76 > »

SCREEN: BED Upload



SCREEN hg19 

Biosamples ⓘ

TSV Search:

cell type	tissue
<input type="radio"/> A172	brain
<input type="radio"/> A549	lung
<input type="radio"/> A549 treated with dexamethasone	lung
<input type="radio"/> A549 treated with ethanol	lung
<input type="radio"/> A673	muscle
<input type="radio"/> ACC112	salivary glands
<input type="radio"/> adipocyte	adipose
<input type="radio"/> adipose derived mesenchymal stem cell in vitro differentiated cells	stem cell
<input type="radio"/> adrenal gland female adult (51 years)	adrenal
<input type="radio"/> adrenal gland female fetal (108 days)	adrenal

Total: ...

ccRE Search Results **Bed Upload**

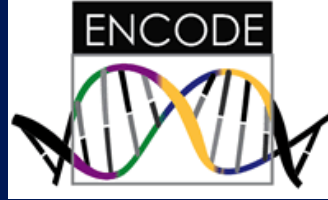
Candidate cis-Regulatory Elements (ccREs) that meet your search criteria are listed in the table below.

- Click a ccRE accession to view details about the ccRE, including top tissues, nearby genomic features, etc.
- Click a gene ID to view the expression profile of the gene.

Search:

accession ⓘ	DNase Z	H3K4me3 Z	H3K27ac Z	CTCF Z	chr	start	length	nearest genes: protein-coding / all	cart	genome browsers
<input type="radio"/> EH37E1090133 P ■ ■ ■	5.33	2.42	1.63	1.37	chr11	5,247,589	1,032	pc: HBB, HBD, HBG1 all: HBB, CoTC_ribozyme, HBD	<input type="button" value="🛒 +"/>	<input type="button" value="UCSC"/>
<input type="radio"/> EH37E0204932 ★ D ■ ■ ■ ■	4.20	1.91	3.59	2.19	chr11	5,301,786	446	pc: HBE1, OR51B4, HBG2 all: HBE1, OR51B4, HBG2	<input type="button" value="🛒 +"/>	<input type="button" value="UCSC"/>
<input type="radio"/> EH37E0204974 ★ D ■ ■ ■ ■	3.73	1.26	2.28	5.28	chr11	5,401,971	367	pc: OR51M1, OR51J1, OR51B6 all: OR51M1, OR51J1, OR51B6	<input type="button" value="🛒 +"/>	<input type="button" value="UCSC"/>
<input type="radio"/> EH37E0204969 ★ D ■ ■ ■ ■	3.53	2.17	4.64	1.56	chr11	5,388,661	199	pc: OR51B6, OR51M1, OR51B5 all: OR51B6, OR51M1, OR51B5	<input type="button" value="🛒 +"/>	<input type="button" value="UCSC"/>
<input type="radio"/> EH37E0204963 ★ D ■ ■ ■ ■	3.53	1.93	3.72	1.32	chr11	5,385,367	492	pc: OR51B6, OR51B5, OR51M1 all: OR51B6, OR51B5, OR51M1	<input type="button" value="🛒 +"/>	<input type="button" value="UCSC"/>
<input type="radio"/> EH37E0204970 ★ D ■ ■ ■ ■	3.46	2.04	5.03	1.20	chr11	5,388,866	595	pc: OR51B6, OR51M1, OR51B5 all: OR51B6, OR51M1, OR51B5	<input type="button" value="🛒 +"/>	<input type="button" value="UCSC"/>
<input type="radio"/> EH37E0204972 ★ D ■ ■ ■ ■	3.30	1.45	1.82	5.57	chr11	5,391,140	230	pc: OR51B6, OR51M1, OR51B5 all: OR51B6, OR51M1, OR51B5	<input type="button" value="🛒 +"/>	<input type="button" value="UCSC"/>
<input type="radio"/> EH37E0204928 ★ D ■ ■ ■ ■	3.17	1.36	3.47	1.53	chr11	5,296,800	608	pc: HBE1, HBG2, HBG1 all: HBE1, HBG2, HBG1	<input type="button" value="🛒 +"/>	<input type="button" value="UCSC"/>

SCREEN: BED Upload



SCREEN hg19

Biosamples ⓘ

TSV Search:

cell type	tissue	
<input type="radio"/> naive thymus-derived CD4-positive alpha-beta T cell female adult (35 years)	blood	
<input type="radio"/> natural killer cell female adult (34 years)	blood	
<input type="radio"/> natural killer cell male adult (37 years)	blood	
<input type="radio"/> regulatory T cell male adult (28 years)	blood	
<input type="radio"/> T-cell	blood	
<input type="radio"/> T-cell male adult (21 years)	blood	
<input type="radio"/> T-cell male adult (36 years)	blood	
<input type="radio"/> T-cell male adult (37 years)	blood	

ccRE **Bed Upload** **ccRE Details**

Search Results

ccRE intersection

Upload bed files here to be automatically intersected with all available ccREs.

Drop bed files here, or click to select bed files to upload.

Beds

	A	B	C	D
1	chr5	131950000	132000000	
2				
3				
4				

SCREEN: BED Upload



SCREEN hg19

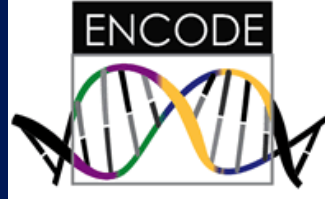
ccREs in Cart [Bed Upload](#)


- Click a ccRE accession to view details about the ccRE, including top tissues, nearby genomic features, etc.
- Click a gene ID to view the expression profile of the gene.

Search:


	accession	DNase Z	H3K4me3 Z	H3K27ac Z	CTCF Z	chr	start	length	experimental evidence	nearest genes: protein-coding / all	cart	genome browsers
<input type="radio"/>	EH37E1252294 ★ P	4.02	3.20	3.95	2.01	chr5	131,998,755	729	--	pc: IL13, IL4, KIF3A all: AC004041.2, IL13, IL4		UCSC
<input type="radio"/>	EH37E0801987 ★ D	3.94	1.85	2.87	1.14	chr5	131,971,535	533	--	pc: RAD50, IL13, IL4 all: AC004041.2, RAD50, IL13		UCSC
<input type="radio"/>	EH37E1252295 ★ P	3.70	3.53	4.06	1.41	chr5	131,999,675	497	--	pc: IL13, IL4, KIF3A all: AC004041.2, IL13, IL4		UCSC
<input type="radio"/>	EH37E0801994 ★ D	3.21	0.90	1.81	1.19	chr5	131,987,488	724	--	pc: IL13, IL4, RAD50 all: AC004041.2, IL13, IL4		UCSC
<input type="radio"/>	EH37E0801983 D	2.99	1.48	1.71	1.39	chr5	131,963,165	497	--	pc: RAD50, IL13, IL4 all: RAD50, AC004041.2, IL13		UCSC
<input type="radio"/>	EH37E0801997 ★ D	2.98	1.69	3.17	2.84	chr5	131,996,786	874	--	pc: IL13, IL4, KIF3A all: AC004041.2, IL13, IL4		UCSC
<input type="radio"/>	EH37E0801998 ★ D	2.95	1.53	3.18	1.52	chr5	131,997,662	259	--	pc: IL13, IL4, KIF3A all: AC004041.2, IL13, IL4		UCSC
<input type="radio"/>	EH37E1252292 ★ P	2.95	2.09	3.65	1.26	chr5	131,975,395	436	--	pc: IL13, RAD50, IL4 all: AC004041.2, IL13, RAD50		UCSC
<input type="radio"/>	EH37E1252293 ★ P	2.87	3.76	1.53	2.14	chr5	131,991,277	1,443	--	pc: IL13, IL4, RAD50 all: AC004041.2, IL13, RAD50		UCSC





NHGRI ENCODE Tutorials





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Research Institute

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These tutorials were prepared for biologists using human and/or mouse genetic data to study disease, gene regulation, and basic biology. They explain what data are available, what they mean, how they can be displayed and downloaded and how they can be used in genetic research on human disease.

- [Navigating the ENCODE Encyclopedia: Exploring Candidate Regulatory Elements, Linked Genes and Genetic Variation with SCREEN, October 2017, ASHG](#) **New**
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- [2016 Society of Toxicology Annual Meeting Workshop](#)
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