

The principal element of our analytical approach is to define specific sets of genomic regulatory loci
WITHOUT ANY PRIOR KNOWLEDGE
of what genes (if any) they may (or may not) control.

This is in striking contrast with the approaches that being utilized in the prior art: to identify genes that are differentially regulated between different states and/or conditions, thus introducing the confounders and biases associated with the multiple hypothesis testing, overfitting, and random co-occurrence due to the very large excess of analyzed features (genes) over the relatively small numbers of samples (100K range or more of analyzed features versus just a few hundred analyzed samples at best). Attempts to address these issues using statistical approaches (corrections for multiple hypothesis testing) and experimental design improvements (multiple replications, validation in independent data sets and using independent analytical techniques) did not adequately resolve these problems.

Importantly, our analyses revealed that this approach identifies not only genes altered expression of which implicated in the broad spectrum of human physiological and pathological conditions. It also identifies genes defined by the independent studies as genetic loci comprising mutation signatures associated with development and progression of multiple common human disorders, including cancer, neurodevelopmental, neuropsychiatric, and neurodegenerative disorders, as well autoimmune and immuno-inflammatory diseases.

Supporting the concept that many major human disorders are driven by aberrant functions of primate-specific genomic regulatory networks with prominent unique-to-human (human-specific) components, this approach identifies genetic loci implicated in inter-individual genetic mosaicism (somatic mosaicism) of cells, tissues and organs in the human body.

Analytical Pipelines

Define panels of genomic regulatory loci

Human-specific genomic regulatory sequences (HSGRS)

Human stem cell-associated retroviral sequences (SCARS)

Identify genes regulated by HSGRS

Identify genes regulated by SCARS

Heat-Map-Guided (HMG) visualization of gene and protein expression profiles
using databases of distinct types of human cells, tissues, anatomical sites, organs, physiological states, pathological conditions, as well as regulatory, chemical, and environmental perturbations

Comparative gene set enrichment (CGSE) analyses of gene and protein expression profiles
using databases of distinct types of human cells, tissues, anatomical sites, organs, physiological states, pathological conditions, as well as regulatory, chemical, and environmental perturbations

Asses the statistical significance of observations

Formulate the concepts regarding the potential roles and impacts of genomic regulatory networks governed by HSGRS and SCARS in human development, physiology, and pathology

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Functional enhancers of naïve and primed hESC

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- **MERGE THE RESULTS OF THE ANALYSES TO EXPLORE THE CONTRIBUTION OF SCARS TO FUNCTIONS OF HUMAN-SPECIFIC GENOMIC REGULATORY NETWORKS IN HUMAN DEVELOPMENT AS WELL AS HEALTH AND DISEASE STATES**
- **IDENTIFY THE HIGH-VALUE GENETIC TARGETS FOR MECHANISTIC AND FUNCTIONAL STUDIES IN THE EXPERIMENTAL MODELS OF HUMAN NEUROGENESIS AND CORTICOGENESIS**

59K human-specific regulatory sequences

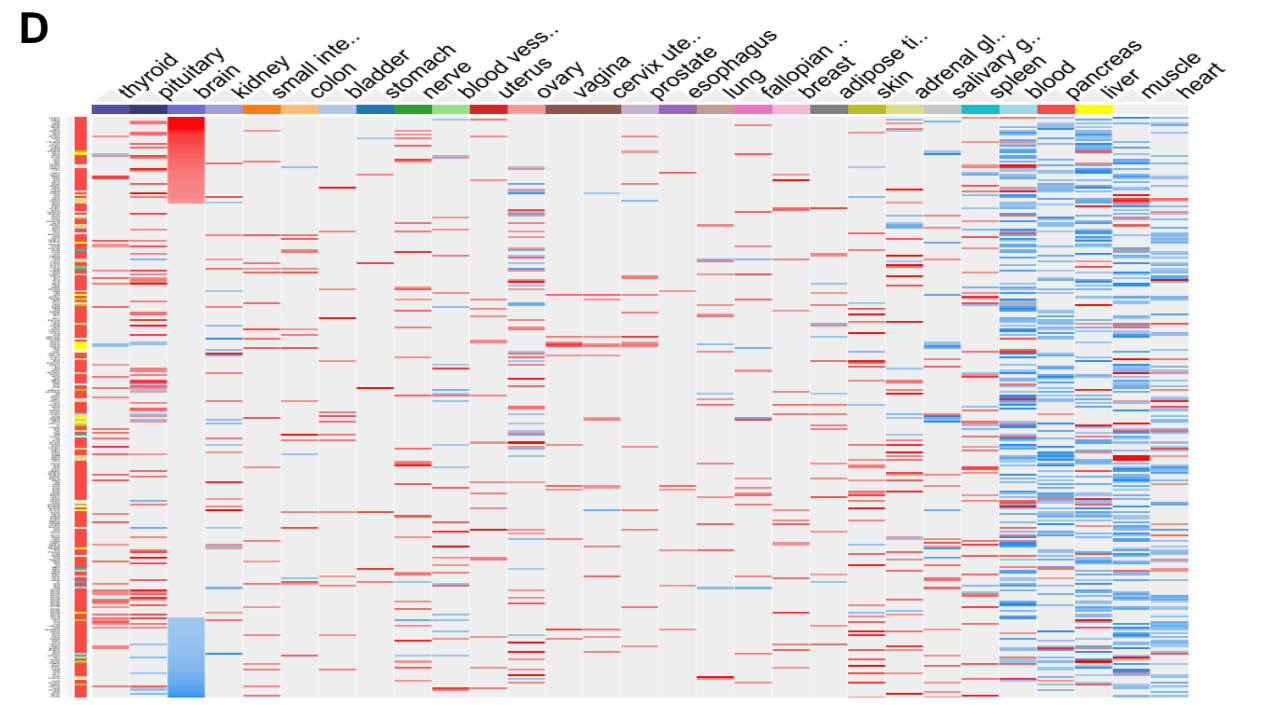
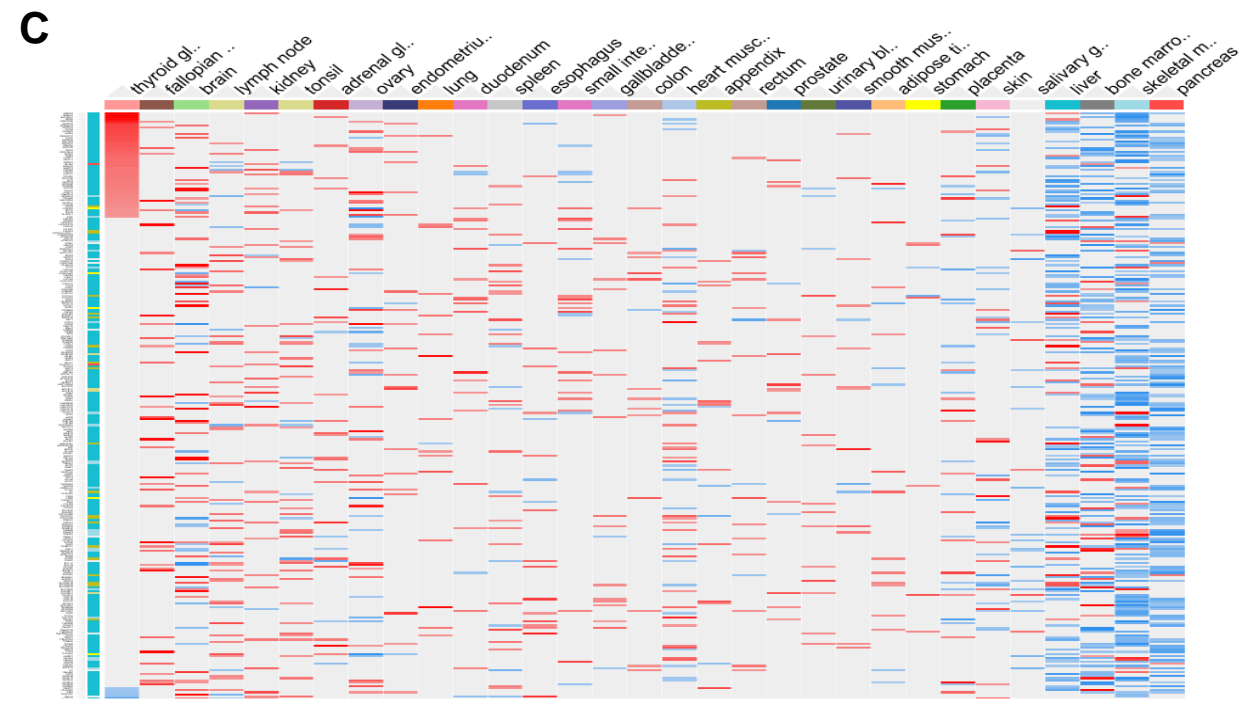
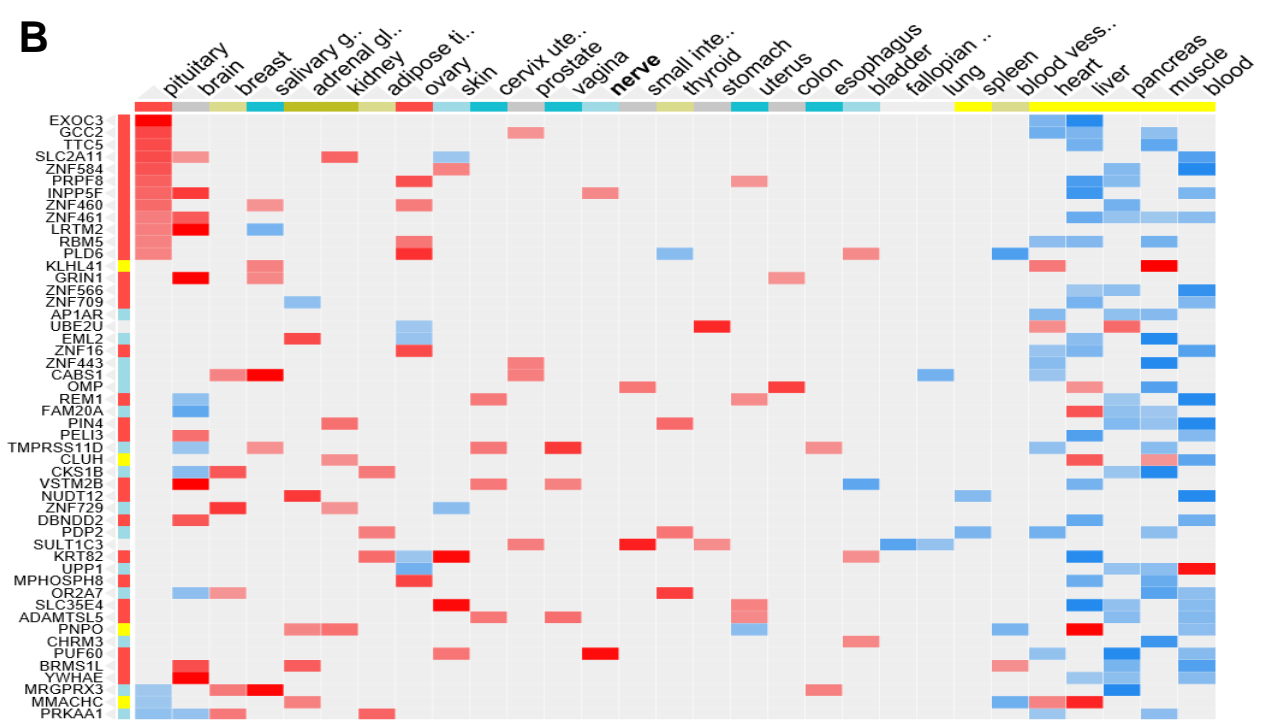
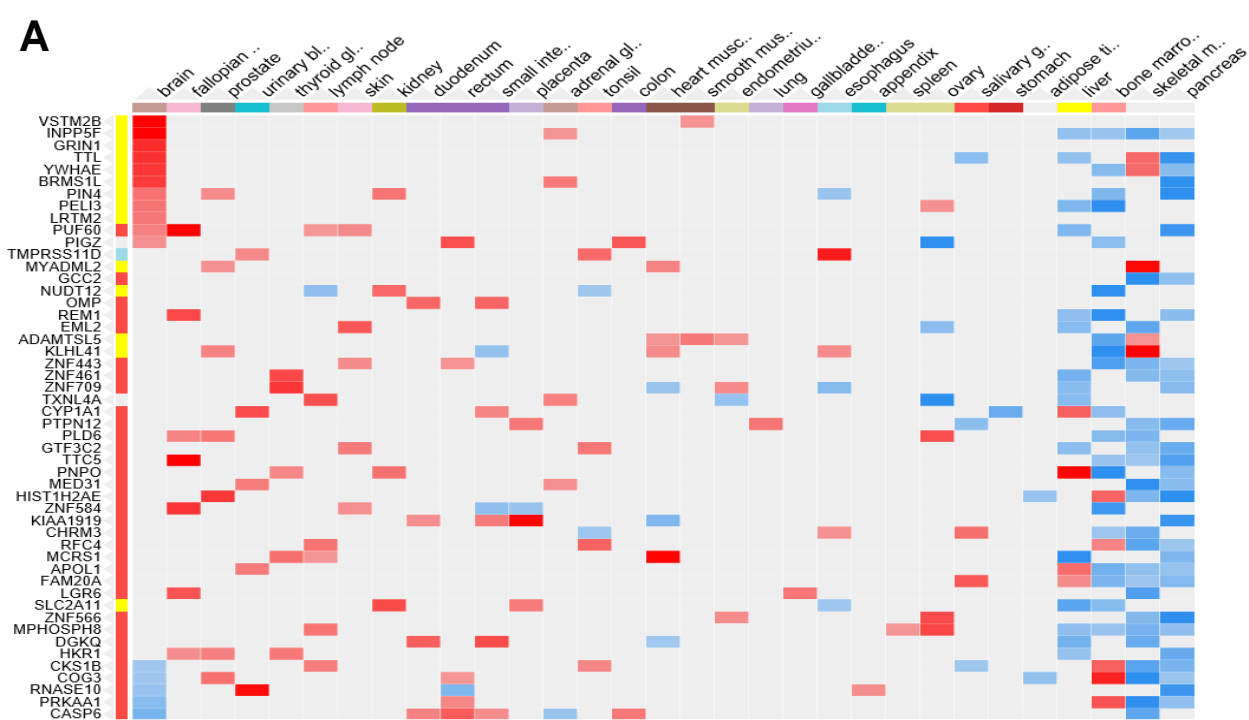
Analysis of genes representing the putative regulatory targets of human-specific regulatory sequences

Structurally, functionally, and evolutionary distinct families of human-specific regulatory sequences (HSRS) and associated putative regulatory target genes defined by the GREAT algorithm.

Classification category/Reference database	Number of records (hg19)	Associated genes
Fixed human-specific insertions.	11878	7979
Human-specific TE loci expressed in human dorsolateral prefrontal cortex	4637	4051
Set of duplicated regions in GRCh38 space	7599	6618
Fixed human-specific deletions	5883	5489
Human-specific STR expansions	4875	4844
hsTFBS	3803	1087
ace-DHS	3538	3445
FHSRR	4249	2810
Human-specific STR contractions	1279	973
hESC_FHSRR_DHS	1932	1458
DHS_FHSRR (non-hESC)	2118	552
HARs	2745	2281
haDHS	524	747
Human-biased CNCC enhances	1000	1439
Chimp-biased CNCC enhances	1000	1445
H3K4me3 peaks with human-specific enrichment in prefrontal neurons	410	578
Human-specific hESC functional enhancers	1619	1214
All HSRS	59089	13824

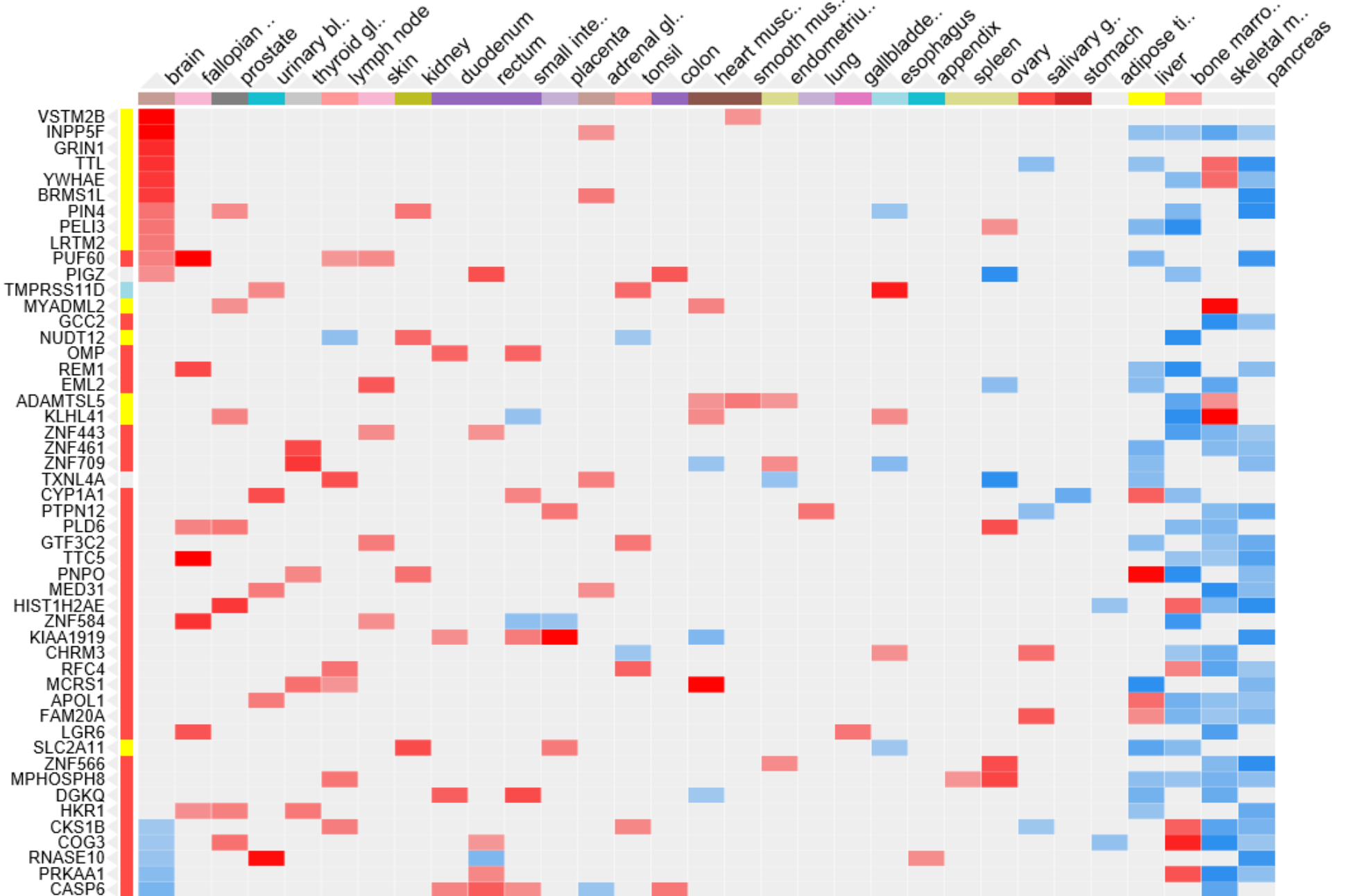
Legend: Definitions of structurally, functionally, and evolutionary distinct families of human-specific regulatory sequences (HSRS) can be found in Glinsky (2020);

Figure 1. Heat-Map-Guided Visualization (HMGV) analysis of expression profiles of mRNAs encoded by 90 genes associated with human-specific genomic regulatory loci derived from transcriptionally active in human dorsolateral prefrontal cortex (DLPFC) transposable elements (A; B; E) and mRNAs encoded by 326 genes associated with human-specific genomic regulatory loci derived from fixed human-specific insertions (C; D; F). Results of the HMGV analysis employing the following databases are shown: HPA Human Tissues' Gene Expression Profiles (A; C); GTEx Human Tissues' Gene Expression Profiles (B; D); BioGPS Human Cell Type and Tissue Gene Expression Profiles (E; F). In (A; B; C; E; F), tissues (columns) are sorted by the rank order function from left to right; genes (rows) are sorted by the cluster order function from top to bottom. In (D), both columns and rows are sorted by the cluster order function.



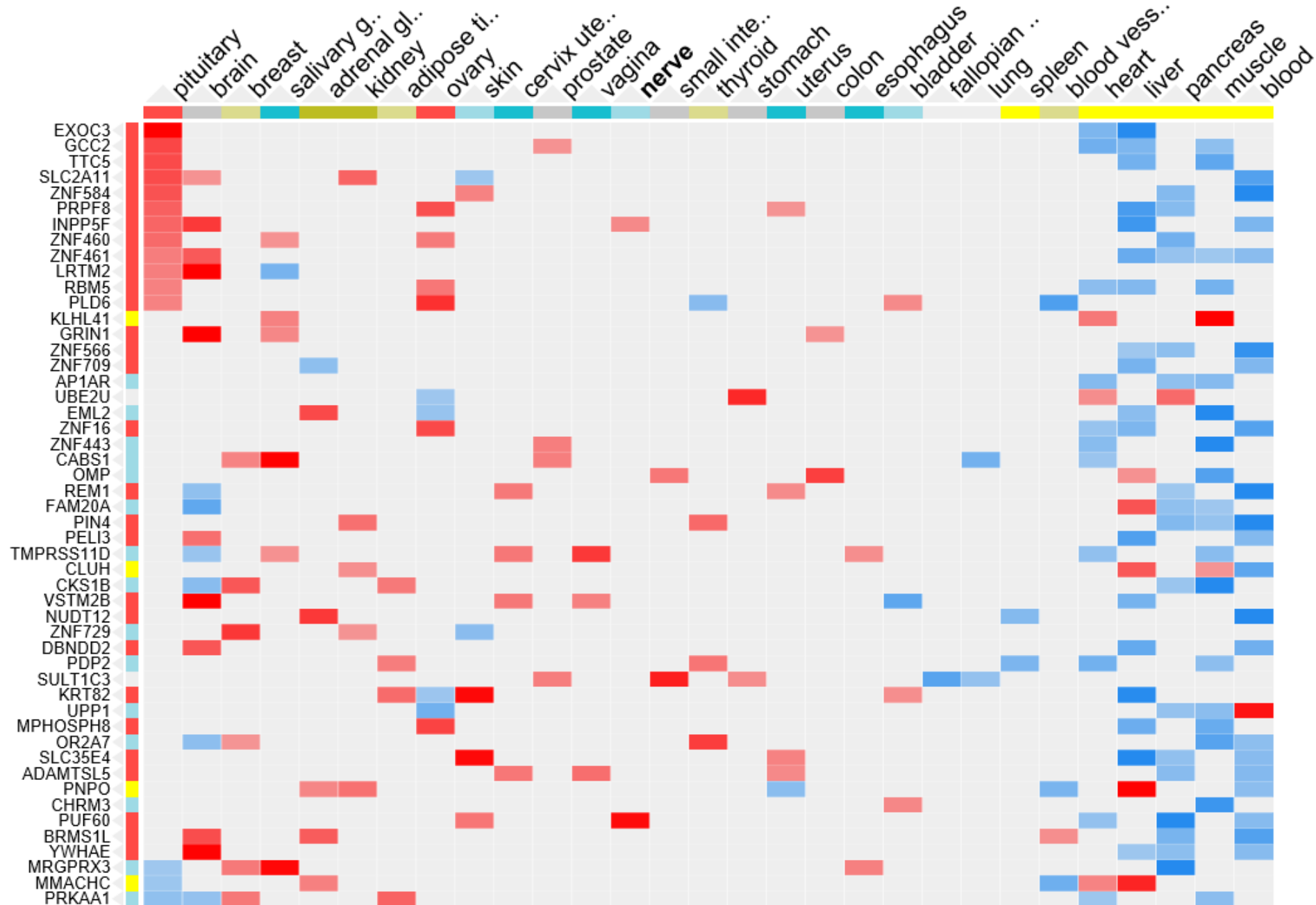
HeatMaps Visualization of expression profiles of mRNAs
encoded by 90 genes associated with human-specific genomic
regulatory loci derived from transcriptionally active in human
DLPFC transposable elements

HPA Human Tissues' Gene Expression Profiles



hsTE_DLPFC_1kb
90 genes
top 50 targets

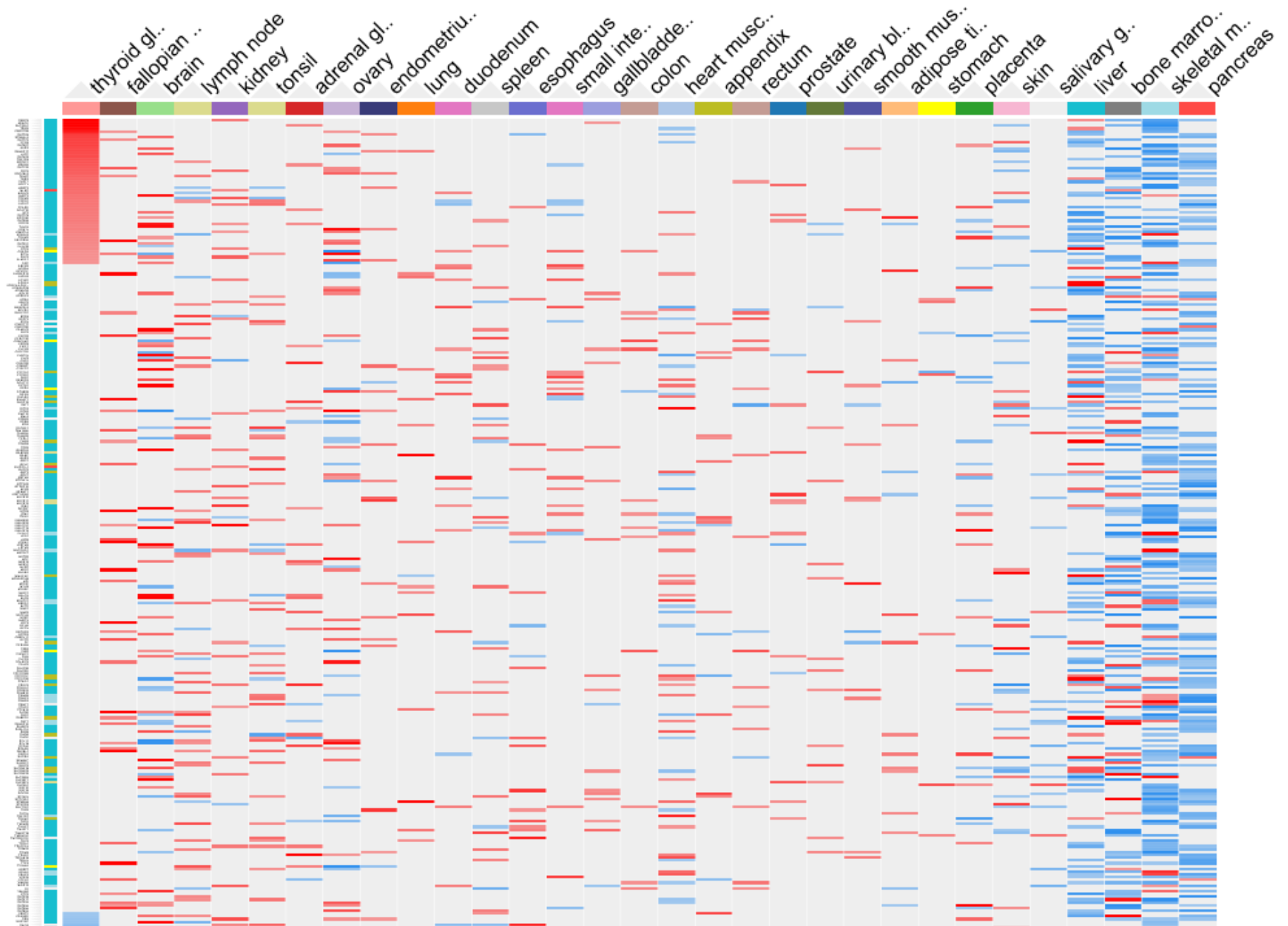
GTEx Human Tissues' Gene Expression Profiles



hsTE_DLPFC_1kb
 90 genes
 top 50 targets

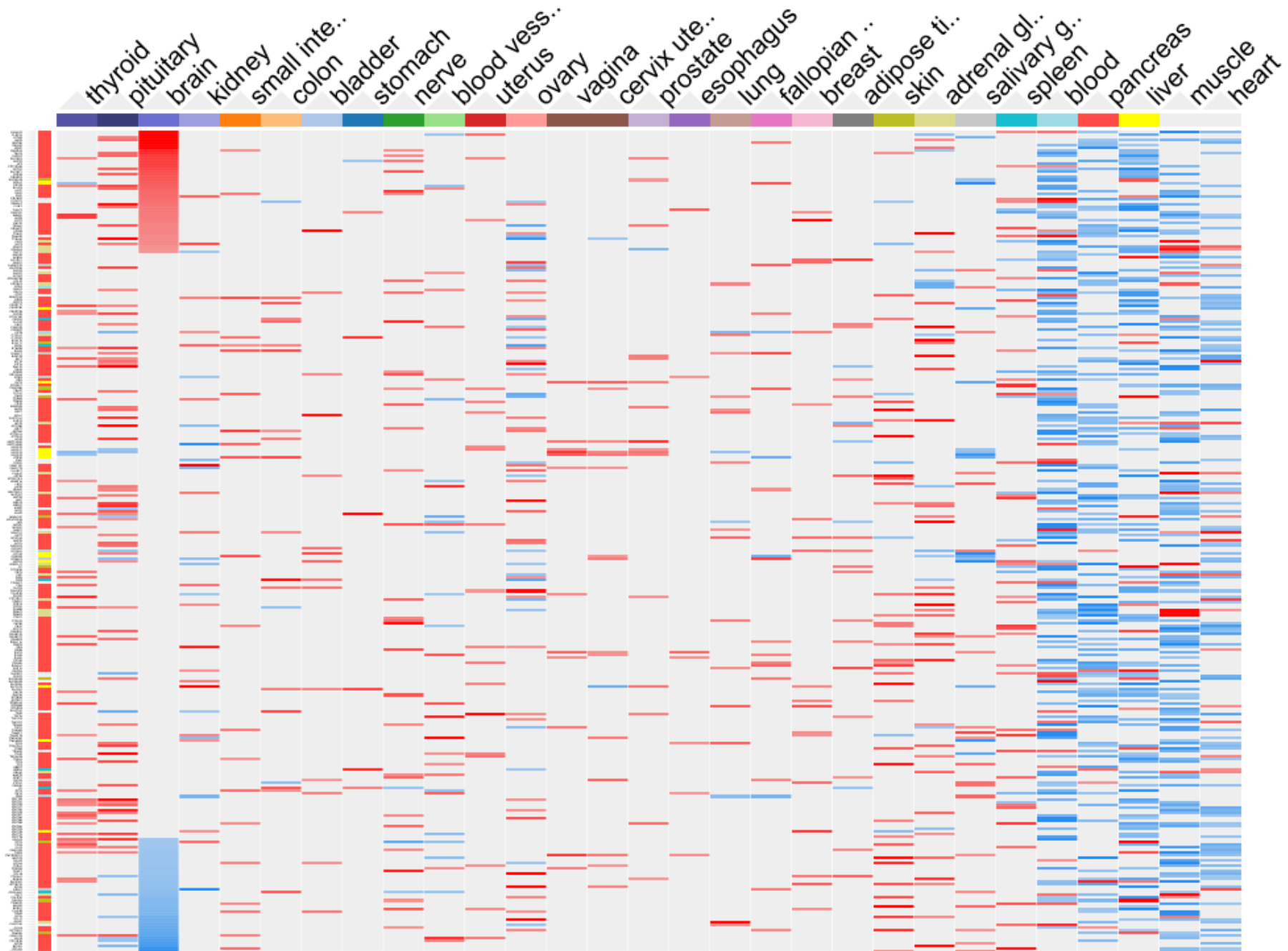
HeatMaps Visualization of expression profiles of mRNAs encoded by 326 genes associated with human-specific genomic regulatory loci derived from fixed human-specific insertions

HPA Human Tissues' Gene Expression Profiles

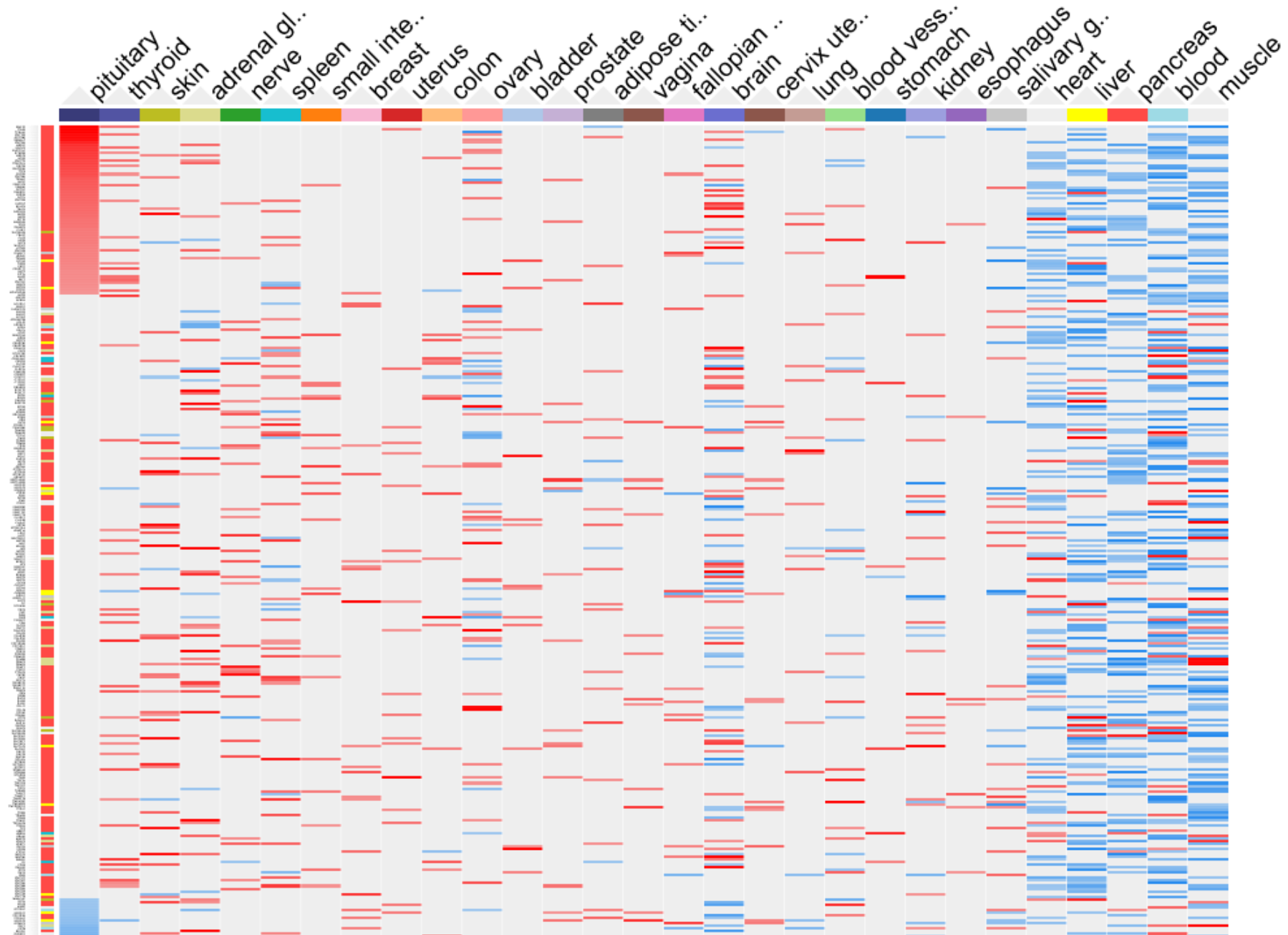


Fixed_hs_Insertions
1kb_326 genes
All targets

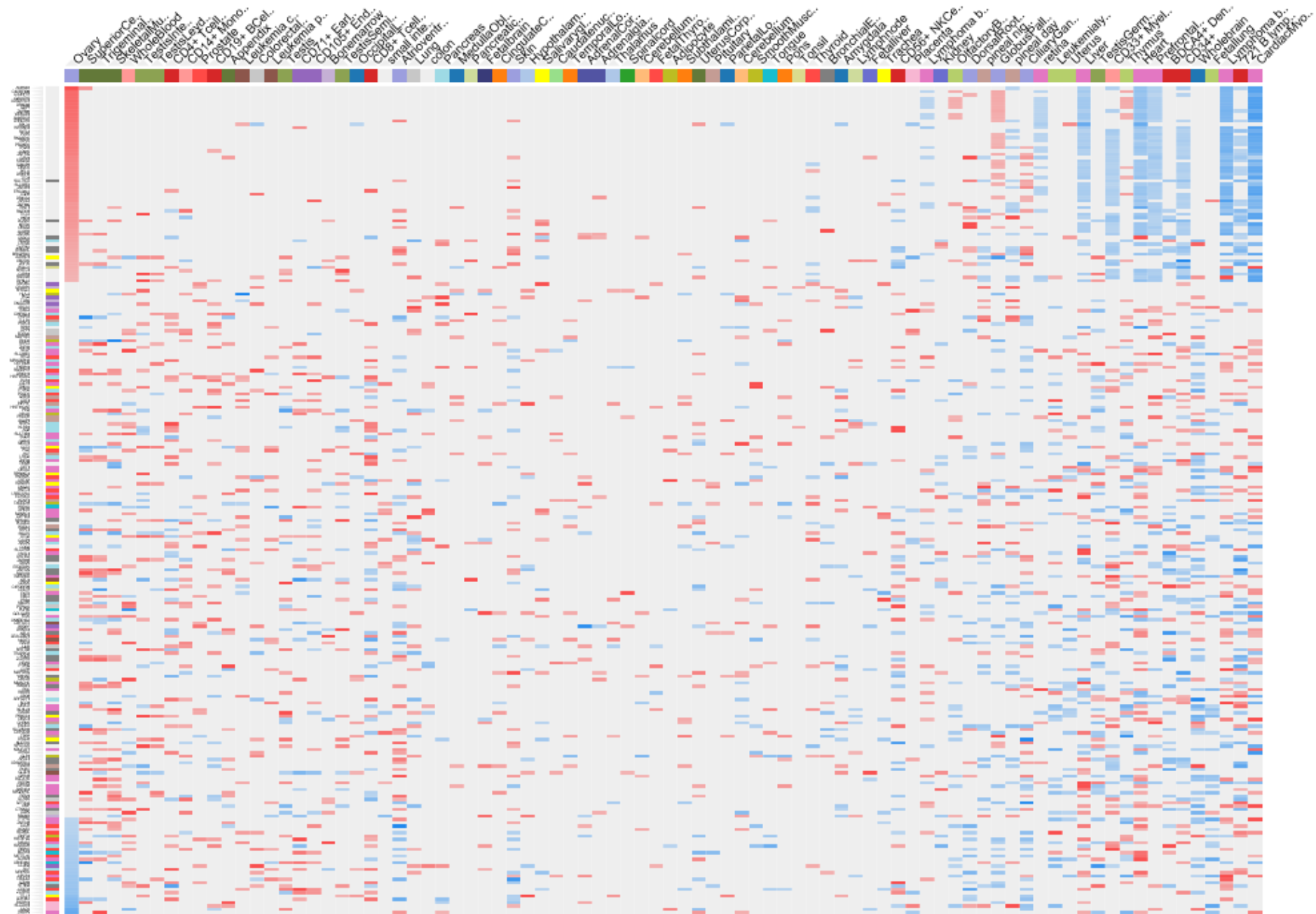
GTEx Human Tissues' Gene Expression Profiles



GTEx Human Tissues' Gene Expression Profiles



BioGPS Human Cell Type and Tissue Gene Expression Profiles



11,878 fixed human-specific insertions

11,878 fixed human-specific insertions

ARCHS4 Tissues 5986 genes

NEURONAL EPITHELIUM	p-value: 9.08E-13; Adjusted p-value: 4.9E-11
PREFRONTAL CORTEX	p-value: 9.08E-13; Adjusted p-value: 4.9E-11
SPINAL CORD	p-value: 9.78E-06; Adjusted p-value: 0.000264
SPINAL CORD (BULK)	p-value: 9.78E-06; Adjusted p-value: 0.000264
CINGULATE GYRUS	p-value: 4.1E-05; Adjusted p-value: 0.000886
CEREBELLUM	p-value: 0.000129; Adjusted p-value: 0.000129
FETAL BRAIN CORTEX	p-value: 0.00191; Adjusted p-value: 0.0294
HUMAN EMBRYO	p-value: 0.00257; Adjusted p-value: 0.0347
MOTOR NEURON	p-value: 0.006; Adjusted p-value: 0.0648
CEREBRAL CORTEX	p-value: 0.006; Adjusted p-value: 0.0648

ARCHS4 Tissues 7979 genes

PREFRONTAL CORTEX	p-value: 1.366E-47; Adjusted p-value: 1.475E-45
NEURONAL EPITHELIUM	p-value: 2.549E-37; Adjusted p-value: 1.377E-35
SPINAL CORD	p-value: 7.582E-24; Adjusted p-value: 2.047E-22
SPINAL CORD (BULK)	p-value: 7.582E-24; Adjusted p-value: 2.047E-22
CEREBELLUM	p-value: 1.084E-22; Adjusted p-value: 2.34E-21
CINGULATE GYRUS	p-value: 2.711E-20; Adjusted p-value: 4.879E-19
CEREBRAL CORTEX	p-value: 1.517E-14; Adjusted p-value: 2.341E-13
RENAL CORTEX	p-value: 1.61E-13; Adjusted p-value: 2.173E-12
MOTOR NEURON	p-value: 1.363E-11; Adjusted p-value: 1.636E-10
BRAIN (BULK)	p-value: 1.041E-09; Adjusted p-value: 1.124E-08

Jensen TISSUES 5986 genes

Hypothalamus	p-value: 4.717e-72; Adjusted p-value: 8.250e-69
Brain	p-value: 5.633e-42; Adjusted p-value: 3.794e-39
Adipose_tissue	p-value: 6.507e-42; Adjusted p-value: 3.794e-39
Heart	p-value: 1.215e-37; Adjusted p-value: 5.312e-35
Cerebral_cortex	p-value: 4.898e-37; Adjusted p-value: 1.713e-34
Lung	p-value: 2.896e-34; Adjusted p-value: 8.443e-32
Retina	p-value: 3.7298E-32; Adjusted p-value: 9.319E-30
Adrenal_gland	p-value: 1.047E-30; Adjusted p-value: 2.289E-28
Uterus	p-value: 1.304E-30; Adjusted p-value: 2.45E-28
Colon	p-value: 1.401E-30; Adjusted p-value: 2.45E-28

Jensen TISSUES 7979 genes

Hypothalamus	p-value: 4.198E-121; Adjusted p-value: 7.539E-118
Brain	p-value: 3.733E-74; Adjusted p-value: 3.352E-71
Cerebral_cortex	p-value: 8.295E-66; Adjusted p-value: 4.966E-63
Heart	p-value: 5.120E-58; Adjusted p-value: 2.299E-55
Lung	p-value: 7.403E-52; Adjusted p-value: 2.659E-49
Adipose_tissue	p-value: 2.312E-51; Adjusted p-value: 6.922E-49
Adrenal_gland	p-value: 8.1997E-51; Adjusted p-value: 2.104E-48
Gall_bladder	p-value: 1.1196E-48; Adjusted p-value: 2.513E-46
Colon	p-value: 2.1855E-48; Adjusted p-value: 4.361E-46
Kidney	p-value: 9.6324E-48; Adjusted p-value: 1.73E-45

11,878 fixed human-specific insertions

Jensen DISEASES 5986 genes

Carcinoma	p-value: 4.013e-91; Adjusted p-value: 6.915e-88
Kidney_cancer	p-value: 1.840e-46; Adjusted p-value: 1.585e-43
Skin_cancer	p-value: 8.144e-18; Adjusted p-value: 4.677e-15
Liver_cancer	p-value: 4.892e-14; Adjusted p-value: 2.107e-11
Acquired_metabolic_disease	p-value: 7.149e-14; Adjusted p-value: 2.464e-11
Melanoma	p-value: 3.929e-13; Adjusted p-value: 1.128e-10
Breast_cancer	p-value: 3.039E-09; Adjusted p-value: 7.48E-07
Pancreatic_cancer	p-value: 9.105E-09; Adjusted p-value: 1.961E-06
Type_2_diabetes_mellitus	p-value: 1.379E-08; Adjusted p-value: 2.64E-06
Lung_cancer	p-value: 4.953E-08; Adjusted p-value: 8.534E-06

Jensen DISEASES 7979 genes

Carcinoma	p-value: 1.590e-165; Adjusted p-value: 2.826e-162
Kidney_cancer	p-value: 1.353e-119; Adjusted p-value: 1.202e-116
Liver_cancer	p-value: 2.316e-49; Adjusted p-value: 1.372e-46
Skin_cancer	p-value: 1.909e-39; Adjusted p-value: 8.480e-37
Melanoma	p-value: 1.836e-35; Adjusted p-value: 6.527e-33
Acquired_metabolic_disease	p-value: 1.749e-31; Adjusted p-value: 5.180e-29
Breast_cancer	p-value: 1.948E-28; Adjusted p-value: 4.944E-26
Type_2_diabetes_mellitus	p-value: 1.811E-21; Adjusted p-value: 4.023E-19
Lung_cancer	p-value: 2.755E-18; Adjusted p-value: 5.439E-16
Pancreatic_cancer	p-value: 1.847E-16; Adjusted p-value: 3.283E-14

Disease Perturbations from GEO down 5986 genes

	P-value	Adjusted p-value
Bipolar Disorder C0005586 human GSE5389 sample 302	1.72813E-08	1.4499E-05
adrenoleukodystrophy DOID-10588 human GSE34309 sample 864	3.85054E-08	1.6153E-05
Primary open angle glaucoma C0339573 human GSE2705 sample 257	8.1833E-08	2.1431E-05
ulcerative colitis DOID-8577 human GSE6731 sample 759	1.02175E-07	2.1431E-05
schizophrenia DOID-5419 human GSE25673 sample 892	6.20235E-07	0.00010407
Breast Cancer C0006142 human GSE1378 sample 52	2.41773E-06	0.00032648
Crohn's disease DOID-8778 human GSE6731 sample 757	2.72388E-06	0.00032648
esophagus squamous cell carcinoma DOID-3748 human GSE63941 sample 659	6.02921E-06	0.00056206
idiopathic pulmonary fibrosis DOID-0050156 human GSE44723 sample 850	5.3956E-06	0.00056206
cardiomyopathy DOID-0050700 human GSE9128 sample 781	7.05452E-06	0.00059188

Disease Perturbations from GEO down 7979 genes

	P-value	Adjusted p-value
schizophrenia DOID-5419 human GSE25673 sample 892	2.88873E-25	2.42364E-22
Crohn's disease DOID-8778 human GSE6731 sample 757	1.49323E-17	6.2641E-15
Bipolar Disorder C0005586 human GSE5389 sample 302	2.47601E-17	6.92457E-15
adrenoleukodystrophy DOID-10588 human GSE34309 sample 864	7.75341E-17	1.62628E-14
ulcerative colitis DOID-8577 human GSE6731 sample 759	2.15119E-16	3.60969E-14
Primary open angle glaucoma C0339573 human GSE2705 sample 257	1.43072E-14	1.66042E-12
Ulcerative Colitis C0009324 human GSE6731 sample 249	1.58324E-14	1.66042E-12
Idiopathic pulmonary fibrosis DOID-0050156 human GSE44723 sample 850	1.56279E-14	1.66042E-12
Breast Cancer C0006142 human GSE1378 sample 52	7.77714E-13	7.25002E-11
Crohn's disease DOID-8778 human GSE6731 sample 758	1.8143E-12	1.5222E-10

11,878 fixed human-specific insertions

Disease Perturbations from GEO up 5986 genes

	P-value	Adjusted p-value
cardiomyopathy DOID-0050700 human GSE9128 sample 780	1.87765E-06	0.001446337
schizophrenia DOID-5419 human GSE25673 sample 891	3.44776E-06	0.001446337
Spinal Muscular Atrophy C0026847 mouse GSE10599 sample 235	8.69631E-06	0.001889865
idiopathic pulmonary fibrosis DOID-0050156 human GSE44723 sample 851	9.01008E-06	0.001889865
Primary open angle glaucoma C0339573 human GSE2705 sample 257	3.83914E-05	0.006442083
idiopathic urticaria DOID-1555 human GSE57178 sample 815	7.75193E-05	0.009776282
Diamond-Blackfan anaemia DOID-1339 human GSE14335 sample 472	8.15661E-05	0.009776282
Neurofibromatosis DOID-8712 mouse GSE1482 sample 667	0.000103508	0.010653102
schizophrenia DOID-5419 human GSE25673 sample 892	0.000132556	0.010653102
hepatitis C DOID-1883 human GSE20948 sample 598	0.000139671	0.010653102

ENCODE and ChEA Consensus TFs from ChIP-X 5986 genes

	P-value	Adjusted p-value
AR_CHEA	3.841E-28	3.9946E-26
SUZ12_CHEA	1.09013E-22	5.66869E-21
SMAD4_CHEA	3.08881E-18	1.07079E-16
REST_CHEA	1.13969E-16	2.9632E-15
NFE2L2_CHEA	9.57565E-14	1.99174E-12
TP63_CHEA	3.48038E-10	6.03266E-09
STAT3_CHEA	2.40449E-08	3.57239E-07
GATA1_CHEA	1.41526E-07	1.48683E-06
GATA2_CHEA	8.89814E-08	1.15676E-06
SALL4_CHEA	1.42965E-07	1.48683E-06

Disease Perturbations from GEO up 7979 genes

	P-value	Adjusted p-value
schizophrenia DOID-5419 human GSE25673 sample 891	1.8927E-19	1.58798E-16
Spinal Muscular Atrophy C0026847 mouse GSE10599 sample 235	5.92303E-18	2.48471E-15
schizophrenia DOID-5419 human GSE25673 sample 892	1.75756E-14	4.9153E-12
idiopathic pulmonary fibrosis DOID-0050156 human GSE44723 sample 850	5.0913E-13	1.0679E-10
morbid obesity DOID-11981 human GSE48964 sample 583	3.15E-11	5.28055E-09
idiopathic pulmonary fibrosis DOID-0050156 human GSE44723 sample 851	6.54233E-11	9.14836E-09
Primary open angle glaucoma C0339573 human GSE2705 sample 257	8.55002E-10	1.02478E-07
cardiomyopathy DOID-0050700 human GSE9128 sample 780	3.23739E-09	3.39521E-07
adrenoleukodystrophy DOID-10588 human GSE34309 sample 864	4.13498E-09	3.85472E-07
Cardiomyopathy, Dilated C0007193 human GSE3585 sample 198	6.70904E-09	5.62888E-07

ENCODE and ChEA Consensus TFs from ChIP-X 7979 genes

	P-value	Adjusted p-value
AR_CHEA	4.679E-85	4.866E-83
SUZ12_CHEA	1.084E-80	5.635E-79
SMAD4_CHEA	2.824E-61	9.789E-60
NFE2L2_CHEA	1.509E-60	3.923E-59
REST_CHEA	7.5683E-41	1.5742E-39
SOX2_CHEA	8.5218E-26	1.4771E-24
TRIM28_CHEA	2.2682E-22	3.3699E-21
SALL4_CHEA	1.8672E-19	2.4274E-18
GATA1_CHEA	6.2282E-19	7.1970E-18
TP63_CHEA	1.3467E-17	1.2732E-16

11,878 fixed human-specific insertions

ESCAPE 5986 genes

P-value Adjusted p-value

hESC_H3K27me3_20682450	5.89784E-14	1.79884E-11
mESC_H3K27me3_17603471	1.78285E-13	2.71885E-11
mESC_H3K9me3_19884255	1.91796E-09	1.94993E-07
CHiP_SUZ12-18974828	3.79545E-08	2.89403E-06
mMEF_K27me3_17603471	1.44703E-07	8.82688E-06
CHiP_MTF2-20144788	8.63335E-07	4.38862E-05
CHiP_SUZ12-18692474	1.42594E-06	6.21303E-05
CHiP_EZH2-18974828	1.21833E-05	0.000464488
mNPC_K27me3_17603471	5.49037E-05	0.001674563
CHiP_SUZ12-18555785	5.27687E-05	0.001674563

ESCAPE 7979 genes

P-value Adjusted p-value

mESC_H3K27me3_17603471	4.44942E-51	1.37932E-48
CHiP_SUZ12-18974828	2.39781E-38	3.71661E-36
hESC_H3K27me3_20682450	2.69286E-37	2.78263E-35
CHiP_SUZ12-18692474	3.02417E-32	2.34373E-30
CHiP_MTF2-20144788	5.57499E-32	3.4565E-30
CHiP_EZH2-18974828	1.0298E-31	5.32065E-30
mESC_H3K9me3_19884255	4.45745E-27	1.97402E-25
CHiP_JARID2-20064375	4.72639E-26	1.83148E-24
CHiP_RNF2-22325148	4.92907E-25	1.69779E-23
CHiP_RNF2-18974828	3.7711E-24	1.16904E-22

GTEx Tissue Sample Gene Expression Profiles up 5986 genes

P-value Adjusted p-value

GTEx-T2IS-0011-R5A-SM-32QP4_brain_female_20-29_years	7.39447E-21	2.15771E-17
GTEx-QDT8-0011-R10A-SM-32PKG_brain_female_30-39_years	5.34561E-20	7.79924E-17
GTEx-OIZI-0008-SM-2XCFCF_skin_male_40-49_years	6.25612E-19	6.08512E-16
GTEx-SNMC-1526-SM-2XCFCN_blood vessel_male_20-29_years	1.49898E-16	1.0935E-13
GTEx-QDT8-0011-R2A-SM-32PKQ_brain_female_30-39_years	6.57826E-16	3.83907E-13
GTEx-OIZI-0526-SM-2XCEG_blood vessel_male_40-49_years	1.52296E-15	7.40664E-13
GTEx-QMR6-0011-R8A-SM-32PKJ_brain_male_50-59_years	2.96092E-15	1.23428E-12
GTEx-NPJ8-0011-R8a-SM-2HMLG_brain_male_40-49_years	4.29977E-15	1.56834E-12
GTEx-SN8G-0526-SM-32PLE_blood vessel_female_50-59_years	5.27021E-14	1.70872E-11
GTEx-PVOW-0011-R5A-SM-32PL7_brain_male_40-49_years	7.63633E-14	2.22828E-11

GTEx Tissue Sample Gene Expression Profiles up 7979 genes

P-value Adjusted p-value

GTEx-SNMC-1526-SM-2XCFCN_blood vessel_male_20-29_years	1.00794E-41	2.94116E-38
GTEx-T2IS-0011-R5A-SM-32QP4_brain_female_20-29_years	1.1941E-40	1.74219E-37
GTEx-QDT8-0011-R10A-SM-32PKG_brain_female_30-39_years	1.21874E-39	1.18543E-36
GTEx-OIZI-0008-SM-2XCFCF_skin_male_40-49_years	3.80382E-34	2.77489E-31
GTEx-PVOW-0011-R5A-SM-32PL7_brain_male_40-49_years	7.93741E-31	4.63227E-28
GTEx-PVOW-0011-R3A-SM-32PKX_brain_male_40-49_years	6.28057E-30	3.05445E-27
GTEx-OIZI-0526-SM-2XCEG_blood vessel_male_40-49_years	8.54238E-30	3.56095E-27
GTEx-QDT8-0011-R2A-SM-32PKQ_brain_female_30-39_years	9.93569E-29	3.62404E-26
GTEx-TMMY-0626-SM-33HBD_blood vessel_female_40-49_years	1.71193E-28	5.55046E-26
GTEx-NPJ8-0011-R8a-SM-2HMLG_brain_male_40-49_years	1.91286E-28	5.58173E-26

11,878 fixed human-specific insertions

GTEX Tissue Sample Gene Expression Profiles down 7979 genes

	P-value	Adjusted p-value
GTEX-X638-0005-SM-47JX6_blood_female_70-79_years	1.04434E-63	3.04737E-60
GTEX-XQ3S-0006-SM-4BOQ4_blood_male_20-29_years	3.90159E-53	5.69242E-50
GTEX-XMK1-0005-SM-4B665_blood_male_40-49_years	1.28663E-47	1.25146E-44
GTEX-XMD3-0006-SM-4AT5X_blood_female_50-59_years	3.40004E-45	2.48033E-42
GTEX-NPJ7-0006-SM-3GACR_blood_female_60-69_years	3.02613E-42	1.76605E-39
GTEX-XLM4-0005-SM-4AT4P_blood_male_60-69_years	1.75487E-38	8.53453E-36
GTEX-WCDI-0005-SM-3NB2M_blood_male_50-59_years	7.88578E-38	3.28725E-35
GTEX-XGQ4-0004-SM-4AT5S_blood_male_50-59_years	5.61964E-37	2.04977E-34
GTEX-X15G-0005-SM-3NMDA_blood_female_50-59_years	5.55996E-36	1.80266E-33
GTEX-XUYS-0002-SM-47JXL_blood_male_50-59_years	9.02958E-35	2.3953E-32

GTEX Tissue Sample Gene Expression Profiles down 5986 genes

	P-value	Adjusted p-value
GTEX-XQ3S-0006-SM-4BOQ4_blood_male_20-29_years	4.60421E-31	1.34351E-27
GTEX-X638-0005-SM-47JX6_blood_female_70-79_years	2.69265E-30	3.92858E-27
GTEX-XMK1-0005-SM-4B665_blood_male_40-49_years	1.1337E-25	1.10271E-22
GTEX-XMD3-0006-SM-4AT5X_blood_female_50-59_years	5.63683E-22	4.11207E-19
GTEX-NPJ7-0006-SM-3GACR_blood_female_60-69_years	7.72356E-21	4.50747E-18
GTEX-VJYA-0005-SM-3P5ZD_blood_male_60-69_years	5.96893E-19	2.90289E-16
GTEX-X4XX-0005-SM-3NMCS_blood_male_60-69_years	8.15686E-19	3.40024E-16
GTEX-XOT4-0005-SM-4B64S_blood_female_60-69_years	4.54574E-18	1.65806E-15
GTEX-WCDI-0005-SM-3NB2M_blood_male_50-59_years	1.97267E-17	6.39584E-15
GTEX-X88G-0006-SM-47JX5_blood_male_30-39_years	4.52769E-17	1.32118E-14

**4,637 human-specific TE-encoded loci
expressed in human DLPFC**

4,637 human-specific TE-encoded loci expressed in human DLPFC

ARCHS4 Tissues 2323 genes

	P-value	Adjusted p-value
PREFRONTAL CORTEX	9.8570E-06	0.00107
CINGULATE GYRUS	0.000652	0.0295
NEURONAL EPITHELIUM	0.000820	0.0295
FETAL BRAIN CORTEX	0.004439	0.1199
CEREBELLUM		
BRAIN (BULK)		
PANCREATIC ISLET		
CEREBRAL CORTEX		
DENTATE GRANULE CELL		
DORSAL STRIATUM		

ARCHS4 Tissues 4051 genes

	P-value	Adjusted p-value
PREFRONTAL CORTEX	7.00452E-50	7.56488E-48
CINGULATE GYRUS	6.71991E-34	3.62875E-32
NEURONAL EPITHELIUM	8.68697E-32	3.12731E-30
CEREBELLUM	9.55784E-29	2.58062E-27
CEREBRAL CORTEX	3.61112E-21	7.80002E-20
SPINAL CORD	2.49377E-20	3.84754E-19
SPINAL CORD (BULK)	2.49377E-20	3.84754E-19
DORSAL STRIATUM	1.66653E-18	2.24982E-17
MOTOR NEURON	6.09151E-15	7.30981E-14
FETAL BRAIN CORTEX	9.14708E-15	9.87885E-14

Jensen TISSUES 2323 genes

	P-value	Adjusted p-value
Hypothalamus	6.32274E-21	8.46615E-18
Occipital_lobe	4.24993E-14	2.84533E-11
Brain	2.96068E-13	1.32145E-10
Parietal_lobe	6.54347E-12	2.19043E-09
Adipose_tissue	1.12994E-11	3.02598E-09
Frontal_lobe	2.30117E-11	5.13545E-09
Cerebral_cortex	1.36673E-10	2.61436E-08
Thyroid_gland	2.08353E-10	3.48731E-08
Lymph_node	2.7611E-10	4.1079E-08
Ovary	3.53351E-10	4.73136E-08

Jensen TISSUES 4051 genes

	P-value	Adjusted p-value
Hypothalamus	1.42728E-50	2.3279E-47
Brain	1.03046E-34	8.40342E-32
Cerebral_cortex	2.82988E-25	1.53851E-22
Frontal_lobe	6.36197E-23	2.59409E-20
Occipital_lobe	1.39104E-20	4.53756E-18
Heart	4.28896E-18	1.16588E-15
Ovary	6.32042E-17	1.28858E-14
Cerebellum	5.93135E-17	1.28858E-14
Parietal_lobe	2.07605E-16	3.76226E-14
Gall_bladder	1.09267E-15	1.78215E-13

4,637 human-specific TE-encoded loci expressed in human DLPFC

Jensen DISEASES 2323 genes

Jensen DISEASES 4051 genes

	P-value	Adjusted p-value
Carcinoma	1.7817E-14	2.295E-11
Kidney_cancer	1.324E-05	0.0085
Takayasu's_arteritis	0.00191	0.615
Liver_cancer	0.00475	0.615
Essential_hypertension	0.00481	0.615
Major_depressive_disorder	0.00572	0.615
Aicardi-Goutieres_syndrome	0.00548	0.615
Sclerocornea	0.00503	0.615
Schizophrenia	0.00311	0.615
Melanoma	0.00360	0.615

	P-value	Adjusted p-value
Carcinoma	1.28507E-65	2.0317E-62
Kidney_cancer	9.99971E-54	7.90477E-51
Liver_cancer	7.45524E-35	3.92891E-32
Melanoma	1.10876E-26	4.38237E-24
Skin_cancer	2.69121E-26	8.50962E-24
Breast_cancer	8.01818E-17	2.11279E-14
Acquired_metabolic_disease	1.65991E-14	3.74902E-12
Lung_cancer	2.30061E-14	4.54659E-12
Endometrial_cancer	9.04405E-12	1.58874E-09
Attention_deficit_hyperactivity_disorder	4.39818E-11	6.95353E-09

Disease Perturbations from GEO down 2323 genes

Disease Perturbations from GEO down 4051 genes

	P-value
dilated cardiomyopathy DOID-12930 human GSE42955 sample 678	0.00151
idiopathic pulmonary fibrosis DOID-0050156 human GSE24206 sample 869	0.00452
Bipolar Disorder C0005586 human GSE5389 sample 302	0.00559
autism spectrum disorder DOID-0060041 human GSE62632 sample 1037	0.00941
peripartum cardiomyopathy DOID-9997 human GSE42955 sample 817	0.01179
Acne C0702166 human GSE10432 sample 297	0.01285
cardiomyopathy DOID-0050700 human GSE9128 sample 781	0.01552
idiopathic pulmonary fibrosis DOID-0050156 human GSE24206 sample 872	0.01657
Down Syndrome C0013080 human GSE5390 sample 277	0.01875
Down syndrome DOID-14250 human GSE16677 sample 1064	0.01994

	P-value	Adjusted p-value
Bipolar Disorder C0005586 human GSE5389 sample 302	2.14561E-10	1.80017E-07
autism spectrum disorder DOID-0060041 human GSE62632 sample 1037	8.52294E-10	3.57537E-07
schizophrenia DOID-5419 human GSE25673 sample 892	3.04969E-09	8.52897E-07
idiopathic pulmonary fibrosis DOID-0050156 human GSE44723 sample 850	3.50643E-08	6.19354E-06
Crohn's disease DOID-8778 human GSE6731 sample 757	3.69102E-08	6.19354E-06
schizophrenia DOID-5419 human GSE25673 sample 891	1.45048E-07	2.02826E-05
ulcerative colitis DOID-8577 human GSE6731 sample 759	3.44284E-07	4.12649E-05
Crohn's disease DOID-8778 human GSE6731 sample 758	2.50335E-06	0.000262539
Alzheimer's disease DOID-10652 human GSE36980 sample 520	7.29851E-06	0.000663404
Nephroblastoma C0027708 human GSE2712 sample 418	8.00156E-06	0.000663404

4,637 human-specific TE-encoded loci expressed in human DLPFC

Disease Perturbations from GEO up 2323 genes

	P-value
Spinal Muscular Atrophy C0026847 mouse GSE10599 sample 235	0.00102
oligodendroglioma DOID-3181 human GSE15824 sample 858	0.00129
Hypoxia C0242184 human GSE4483 sample 440	0.00265
familial combined hyperlipidemia DOID-13809 human GSE11393 sample 773	0.00343
Familial combined hyperlipidaemia C0020474 human GSE11393 sample 267	0.00477
Acute Lung Injury C0242488 human GSE10474 sample 168	0.00495
astrocytoma DOID-3069 human GSE15824 sample 861	0.00581
oligodendroglioma DOID-3181 human GSE15824 sample 859	0.00725
fragile X syndrome DOID-14261 human GSE7329 sample 809	0.00738
Huntington's disease DOID-12858 human GSE1751 sample 795	0.00813

ENCODE and ChEA Consensus TFs from CHIP-X 2323 genes

	P-value	Adjusted p-value
AR_CHEA	2.35459E-06	0.000245
UBTF_ENCODE	8.26486E-05	0.004298
NFE2L2_CHEA	0.00051872	0.017982
CREB1_CHEA	0.000838763	0.021808
SMAD4_CHEA	0.00141901	0.029515
REST_CHEA	0.001930935	0.033470
PPARD_CHEA	0.005210793	0.077417
E2F1_CHEA	0.007865845	0.102256
TCF3_ENCODE	0.00921029	0.106430
BHLHE40_ENCODE	0.016264119	0.154696

Disease Perturbations from GEO up 4051 genes

	P-value	Adjusted p-value
Spinal Muscular Atrophy C0026847 mouse GSE10599 sample 235	1.30588E-20	1.09564E-17
adrenoleukodystrophy DOID-10588 human GSE34309 sample 864	1.45115E-11	6.08757E-09
morbid obesity DOID-11981 human GSE48964 sample 583	4.21321E-08	1.17829E-05
schizophrenia DOID-5419 human GSE25673 sample 892	7.40939E-08	1.55412E-05
schizophrenia DOID-5419 human GSE25673 sample 891	1.09191E-07	1.83222E-05
idiopathic pulmonary fibrosis DOID-0050156 human GSE44723 sample 851	2.54256E-07	3.55534E-05
idiopathic pulmonary fibrosis DOID-0050156 human GSE44723 sample 850	4.62071E-07	5.53825E-05
adrenoleukodystrophy DOID-10588 human GSE34308 sample 709	7.11277E-07	7.45952E-05
smoldering myeloma DOID-9551 human GSE47552 sample 562	8.43186E-06	0.000786037
melanoma DOID-1909 human GSE6887 sample 948	1.68944E-05	0.001417438

ENCODE and ChEA Consensus TFs from CHIP-X 4051 genes

	P-value	Adjusted p-value
AR_CHEA	1.64997E-45	1.71596E-43
SMAD4_CHEA	8.28178E-35	4.30653E-33
NFE2L2_CHEA	1.04996E-28	3.63986E-27
SUZ12_CHEA	1.75619E-28	4.56609E-27
REST_CHEA	7.0257E-19	1.46134E-17
TRIM28_CHEA	1.70459E-09	2.95463E-08
STAT3_CHEA	1.13034E-07	1.47386E-06
GATA1_CHEA	1.13374E-07	1.47386E-06
TCF3_CHEA	3.29897E-07	3.81215E-06
UBTF_ENCODE	8.55045E-07	8.89247E-06

4,637 human-specific TE-encoded loci expressed in human DLPFC

GTEx Tissue Sample Gene Expression Profiles up 2323 genes

P-value Adjusted p-value

GTEx-QDT8-0011-R10A-SM-32PKG_brain_female_30-39_years	7.1111E-12	2.07502E-08
GTEx-PVOW-0011-R3A-SM-32PKX_brain_male_40-49_years	9.49921E-10	9.23956E-07
GTEx-QVJO-0011-R10A-SM-2S1QJ_brain_female_60-69_years	7.11092E-10	9.23956E-07
GTEx-WVLH-0011-R10A-SM-3MJFM_brain_male_50-59_years	2.68224E-09	1.79484E-06
GTEx-T2IS-0011-R5A-SM-32QP4_brain_female_20-29_years	3.69056E-09	1.79484E-06
GTEx-QMR6-0011-R10A-SM-32PKO_brain_male_50-59_years	3.36753E-09	1.79484E-06
GTEx-QMR6-0011-R8A-SM-32PKJ_brain_male_50-59_years	2.36488E-08	9.85818E-06
GTEx-WL46-0011-R10A-SM-3MJFQ_brain_male_50-59_years	8.65217E-08	3.15588E-05
GTEx-N7MS-0011-R3a-SM-33HC6_brain_male_60-69_years	1.69843E-07	5.50669E-05
GTEx-NL3H-0011-R3a-SM-2I3GL_brain_male_60-69_years	2.34416E-07	6.84027E-05

GTEx Tissue Sample Gene Expression Profiles up 4051 genes

P-value Adjusted p-value

GTEx-QDT8-0011-R10A-SM-32PKG_brain_female_30-39_years	4.63841E-42	1.35349E-38
GTEx-PVOW-0011-R3A-SM-32PKX_brain_male_40-49_years	4.30355E-36	6.27888E-33
GTEx-T2IS-0011-R5A-SM-32QP4_brain_female_20-29_years	4.51564E-33	4.39221E-30
GTEx-PVOW-0011-R5A-SM-32PL7_brain_male_40-49_years	2.56809E-31	1.87342E-28
GTEx-QMR6-0011-R10A-SM-32PKO_brain_male_50-59_years	1.2302E-30	7.17944E-28
GTEx-QMR6-0011-R8A-SM-32PKJ_brain_male_50-59_years	2.1578E-28	1.04941E-25
GTEx-N7MS-0011-R3a-SM-33HC6_brain_male_60-69_years	1.01871E-26	4.24658E-24
GTEx-WVLH-0011-R10A-SM-3MJFM_brain_male_50-59_years	1.71691E-26	6.26244E-24
GTEx-QVJO-0011-R10A-SM-2S1QJ_brain_female_60-69_years	4.26311E-26	1.3822E-23
GTEx-PVOW-2526-SM-2XCF7_brain_male_40-49_years	5.24644E-26	1.53091E-23

GTEx Tissue Sample Gene Expression Profiles down 2323 genes

P-value Adjusted p-value

GTEx-XQ3S-0006-SM-4BOQ4_blood_male_20-29_years	1.57265E-10	4.58427E-07
GTEx-XOT4-0005-SM-4B64S_blood_female_60-69_years	7.49373E-09	1.09221E-05
GTEx-WHSE-0006-SM-3NMBW_blood_male_20-29_years	1.18638E-08	1.15277E-05
GTEx-WZTO-0006-SM-3NM9T_blood_male_40-49_years	2.0799E-07	0.000152
GTEx-U3ZG-0326-SM-47JXN_muscle_male_50-59_years	5.52956E-07	0.000288
GTEx-SJXC-0005-SM-2XCE7_blood_male_60-69_years	5.91986E-07	0.000288
GTEx-X638-0005-SM-47JX6_blood_female_70-79_years	1.2182E-06	0.000503
GTEx-PVOW-0006-SM-3NMB8_blood_male_40-49_years	1.37946E-06	0.000503
GTEx-NPJ7-0006-SM-3GACR_blood_female_60-69_years	1.6573E-06	0.000537
GTEx-T6MN-0005-SM-32PLJ_blood_male_50-59_years	2.45953E-06	0.000717

GTEx Tissue Sample Gene Expression Profiles down 4051 genes

P-value Adjusted p-value

GTEx-X638-0005-SM-47JX6_blood_female_70-79_years	4.65379E-32	1.35798E-28
GTEx-XQ3S-0006-SM-4BOQ4_blood_male_20-29_years	8.89061E-30	1.29714E-26
GTEx-XMK1-0005-SM-4B665_blood_male_40-49_years	9.61904E-26	9.35612E-23
GTEx-NPJ7-0006-SM-3GACR_blood_female_60-69_years	4.2827E-24	3.12423E-21
GTEx-X88G-0006-SM-47JX5_blood_male_30-39_years	1.85727E-22	1.0839E-19
GTEx-UJHI-0006-SM-3DB8H_blood_female_50-59_years	1.04583E-21	5.08622E-19
GTEx-XLM4-0005-SM-4AT4P_blood_male_60-69_years	1.99878E-21	8.33207E-19
GTEx-XMD3-0006-SM-4AT5X_blood_female_50-59_years	1.7854E-20	6.51223E-18
GTEx-WCDI-0005-SM-3NB2M_blood_male_50-59_years	3.80474E-20	1.23358E-17
GTEx-OIZF-0006-SM-2I5GQ_blood_male_60-69_years	4.35816E-20	1.27171E-17

4,637 human-specific TE-encoded loci expressed in human DLPFC

Tissue Protein Expression from Human Proteome Map 4051 genes

adult frontal cortex

p-value: 0.00006954

Adjusted p-value: 0.002086

fetal brain

p-value: 0.001310;

Adjusted p-value: 0.01966

adult retina

b cells

adult pancreas

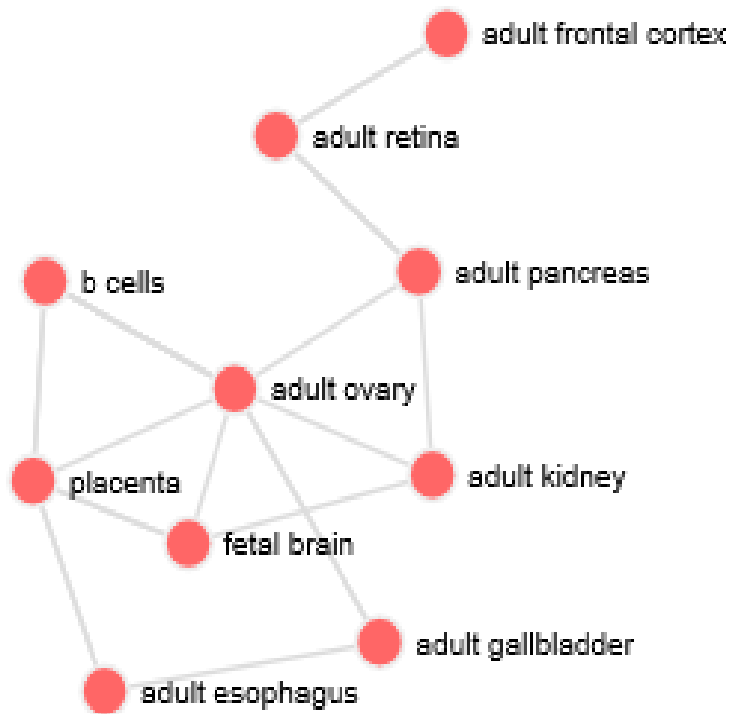
placenta

adult kidney

adult esophagus

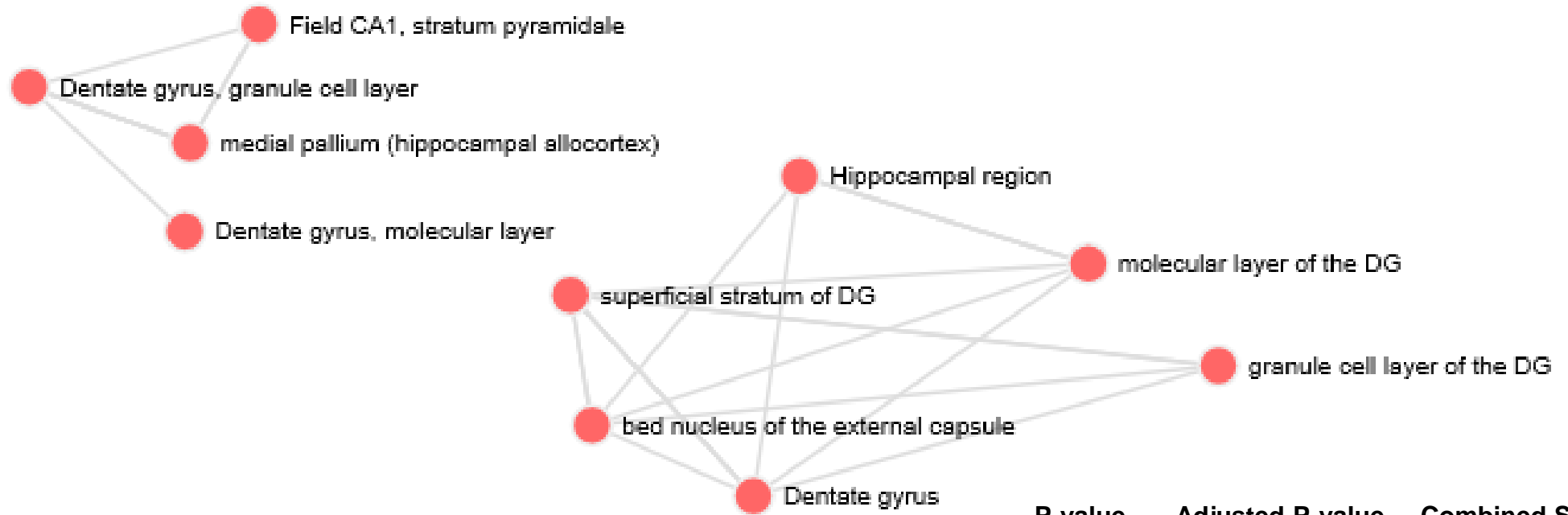
adult gallbladder

adult ovary



4,637 human-specific TE-encoded loci expressed in human DLPFC

Allen Brain Atlas up 4051 genes

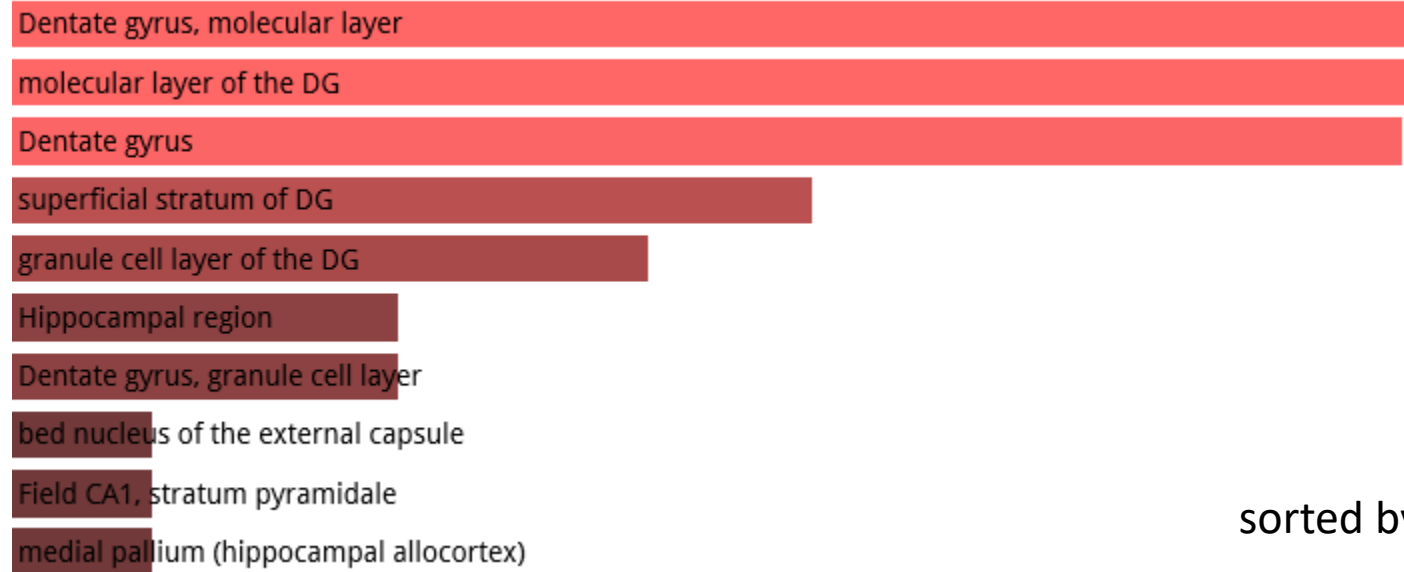


	P-value	Adjusted P-value	Combined Score
Dentate gyrus	2.06994E-09	1.5124E-06	36.78
Dentate gyrus, molecular layer	1.91586E-09	1.5124E-06	35.25
granule cell layer of the DG	1.83231E-08	8.0328E-06	33.31
molecular layer of the DG	1.91586E-09	1.5124E-06	32.40
Hippocampal region	3.77717E-08	1.1828E-05	31.40
Dentate gyrus, granule cell layer	3.77717E-08	1.1828E-05	31.15
bed nucleus of the external capsule	7.67266E-08	1.6818E-05	31.02
Field CA1, stratum pyramidale	7.67266E-08	1.6818E-05	30.60
medial pallium (hippocampal allocortex)	7.67266E-08	1.6818E-05	29.96
superficial stratum of DG	1.13777E-08	6.235E-06	29.70

sorted by combined score ranking

4,637 human-specific TE-encoded loci expressed in human DLPFC

Allen Brain Atlas up 4051 genes



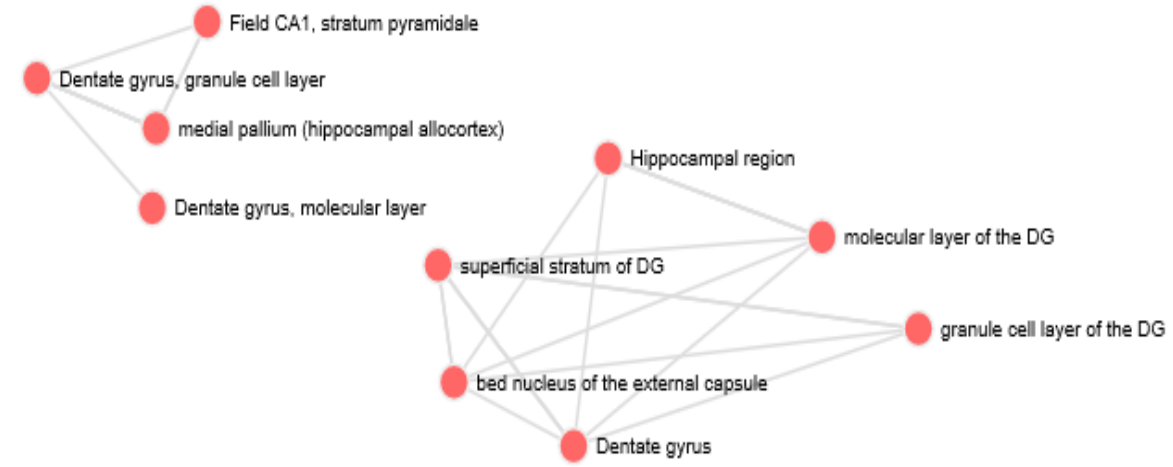
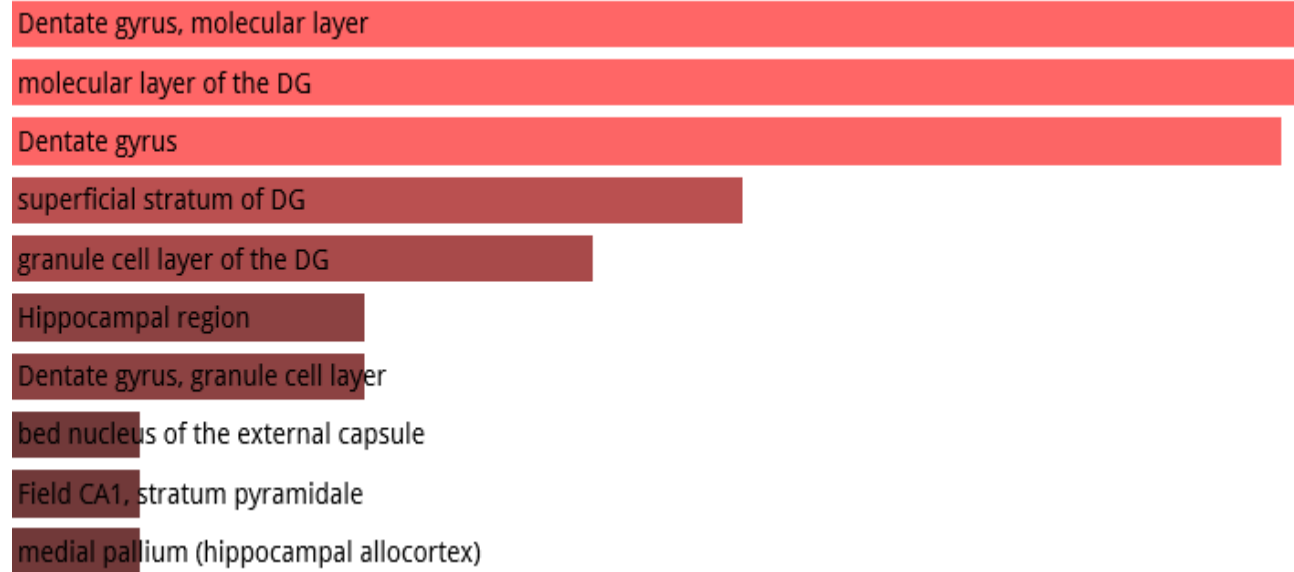
sorted by p-value ranking

TOP 15 CATEGORIES

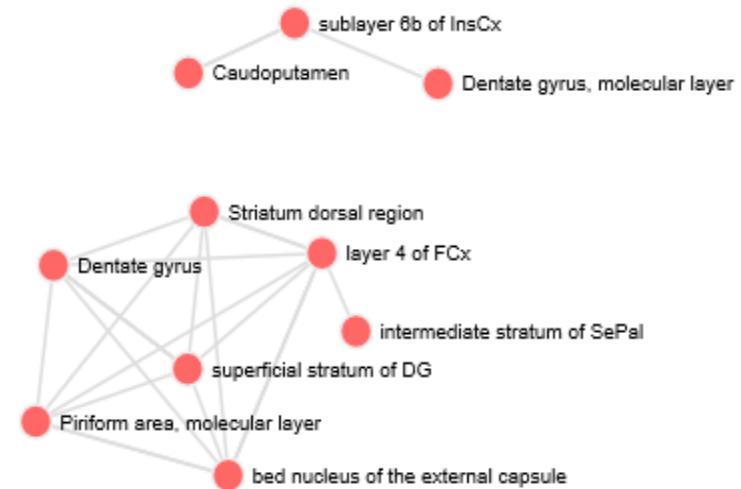
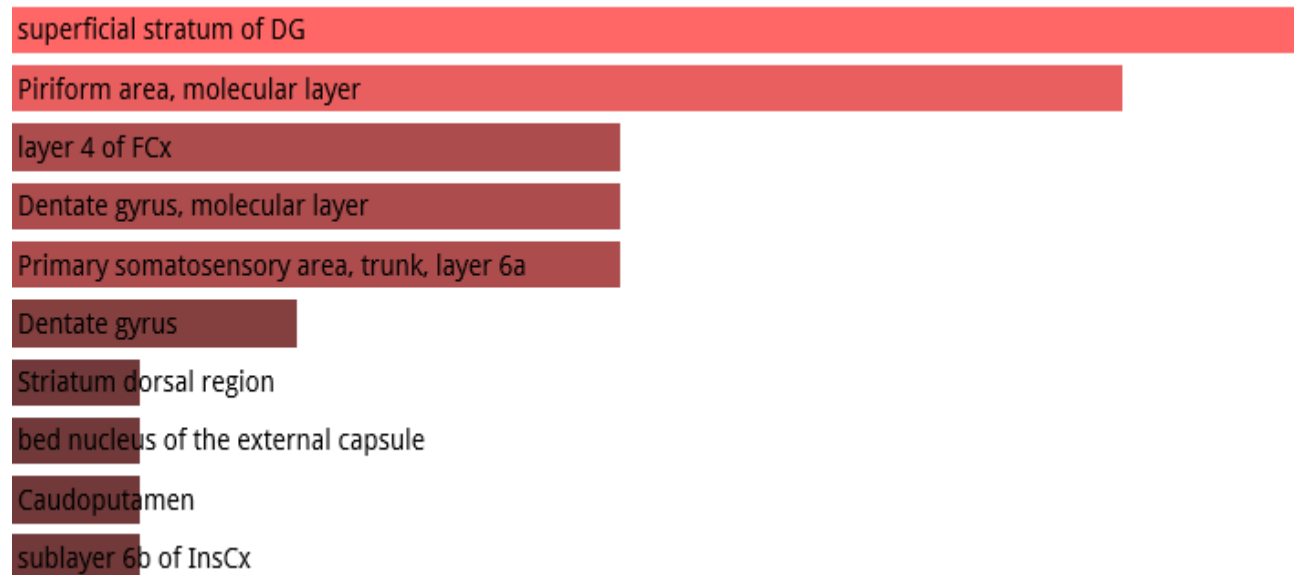
Term	Overlap	P-value	Adjusted P-value
Dentate gyrus	110/320	2.07E-09	1.51E-06
Dentate gyrus, molecular layer	105/301	1.92E-09	1.51E-06
molecular layer of the DG	105/301	1.92E-09	1.51E-06
superficial stratum of DG	179/601	1.14E-08	6.24E-06
granule cell layer of the DG	102/301	1.83E-08	8.03E-06
Hippocampal region	101/301	3.78E-08	1.18E-05
Dentate gyrus, granule cell layer	101/301	3.78E-08	1.18E-05
bed nucleus of the external capsule	100/301	7.67E-08	1.68E-05
Field CA1, stratum pyramidale	100/301	7.67E-08	1.68E-05
medial pallium (hippocampal allocortex)	100/301	7.67E-08	1.68E-05
mantle zone of DG	174/599	1.22E-07	2.4E-05
Subiculum, dorsal part, molecular layer	99/301	1.54E-07	2.4E-05
hilus of the DG	99/301	1.54E-07	2.4E-05
Dentate gyrus, polymorph layer	99/301	1.54E-07	2.4E-05
hippocampus (cortex Ammonis)	98/301	3.03E-07	2.88E-05

4,637 human-specific TE-encoded loci expressed in human DLPFC

Allen Brain Atlas up 4051 genes



Allen Brain Atlas up 2323 genes



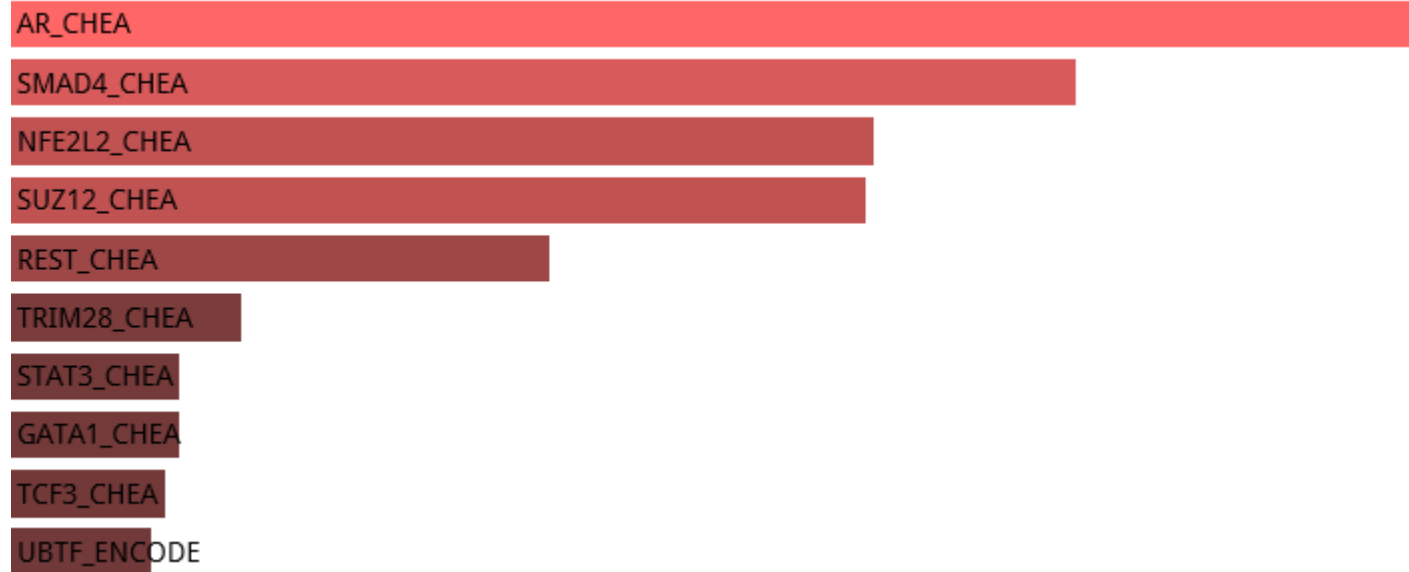
**Comparisons of putative regulatory target genes
associated with**

11,878 fixed human-specific insertions

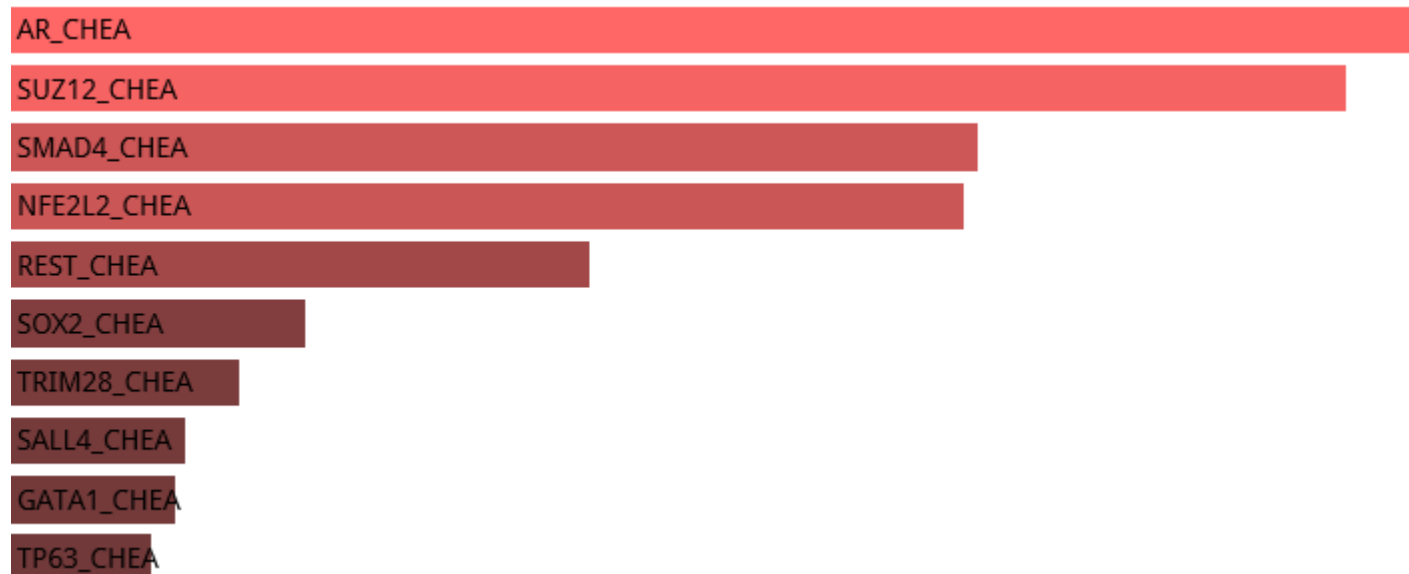
and

**4,637 human-specific TE - encoded loci expressed in
human dorsolateral prefrontal cortex (DLPFC)**

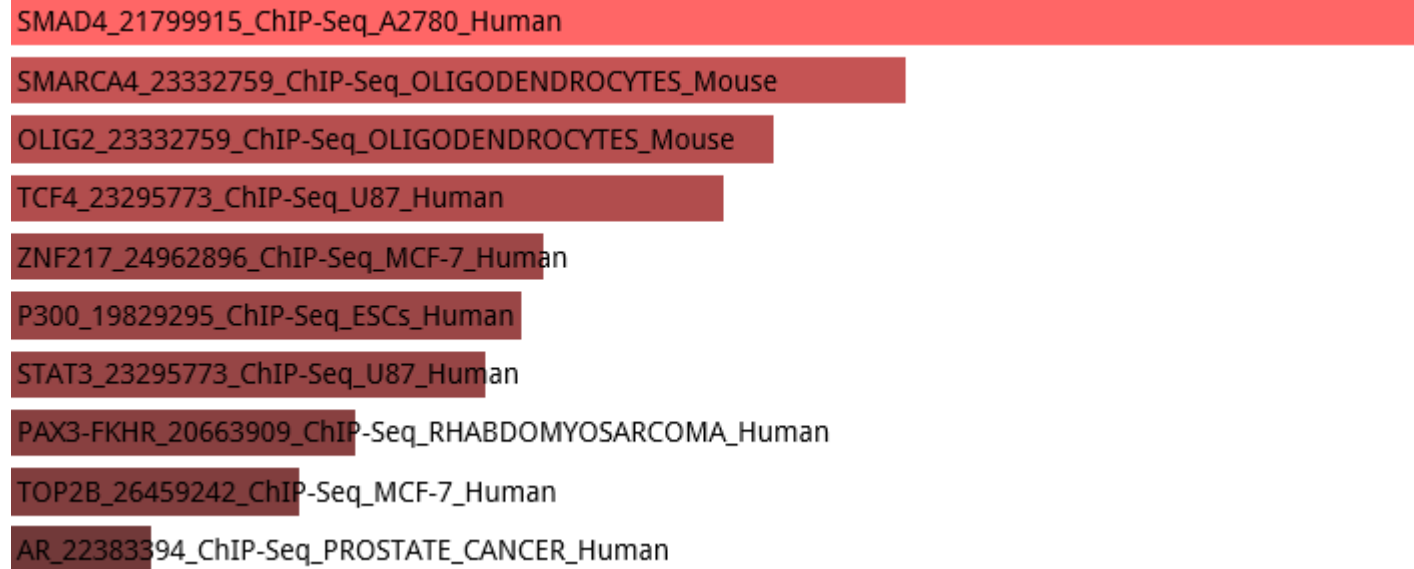
4,637 human-specific TE-encoded loci expressed in human DLPFC
ENCODE and ChEA Consensus TFs from ChIP-X 4051 genes



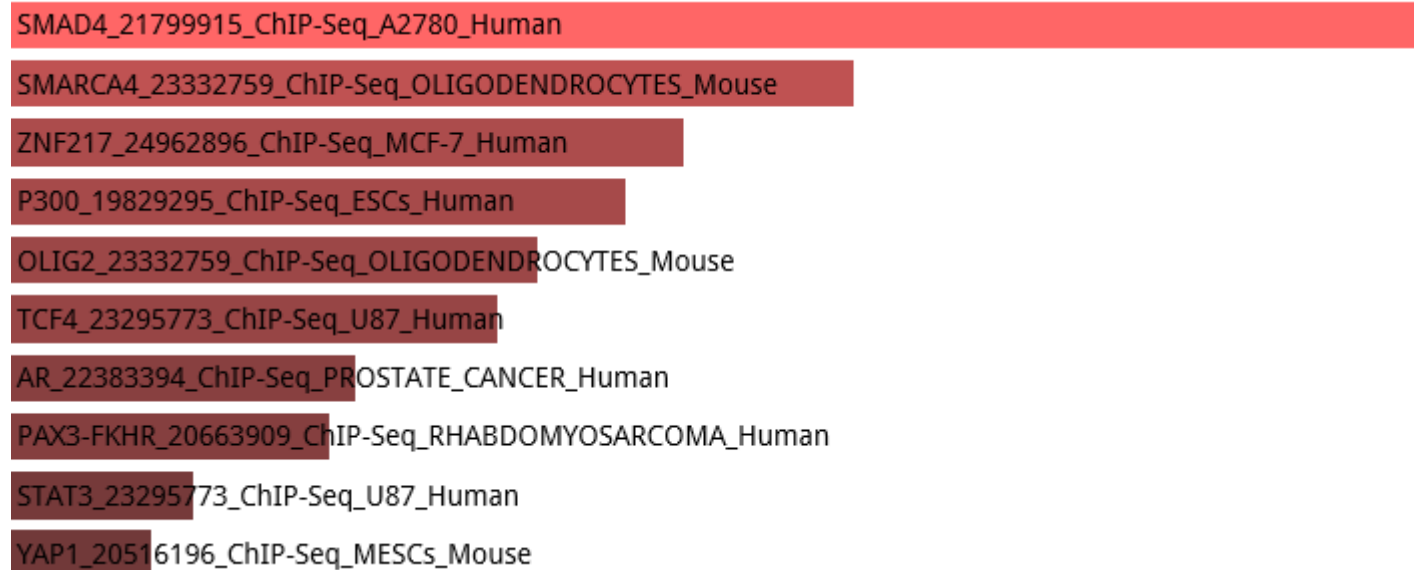
11,878 fixed human-specific insertions
ENCODE and ChEA Consensus TFs from ChIP-X 7979 genes



4,637 human-specific TE-encoded loci expressed in human DLPFC
ChEA 2016 4051 genes



11,878 fixed human-specific insertions
ChEA 2016 7979 genes



4,637 human-specific TE-encoded loci expressed in human DLPFC

TF Perturbations Followed by Expression 4051 genes

BCL11B_KO_MOUSE_GSE9330_CREEDSID_GENE_28_UP
SOX5_OE_FETALCORTEX_HUMAN_GSE89057_RNASEQ_UP
DMRTA2_OE_MOUSE_GSE25179_CREEDSID_GENE_2204_DOWN
REST_INHIBITION_HUMAN_GSE40695_CREEDSID_GENE_1710_UP
EOMES_KO_MOUSE_GSE43387_CREEDSID_GENE_2147_DOWN
ZEB2_SHRNA_H1_HUMAN_GSE69618_DAY6_RNASEQ_DOWN
NRL_DEFICIENCY_MOUSE_GSE4051_CREEDSID_GENE_263_DOWN
NFKB1_ACTIVATION_HUMAN_GSE20736_CREEDSID_GENE_2522_DOWN
MECP2_KO_MOUSE_GSE8720_CREEDSID_GENE_2418_UP
MYC_KD_HUMAN_GSE54872_CREEDSID_GENE_1307_UP

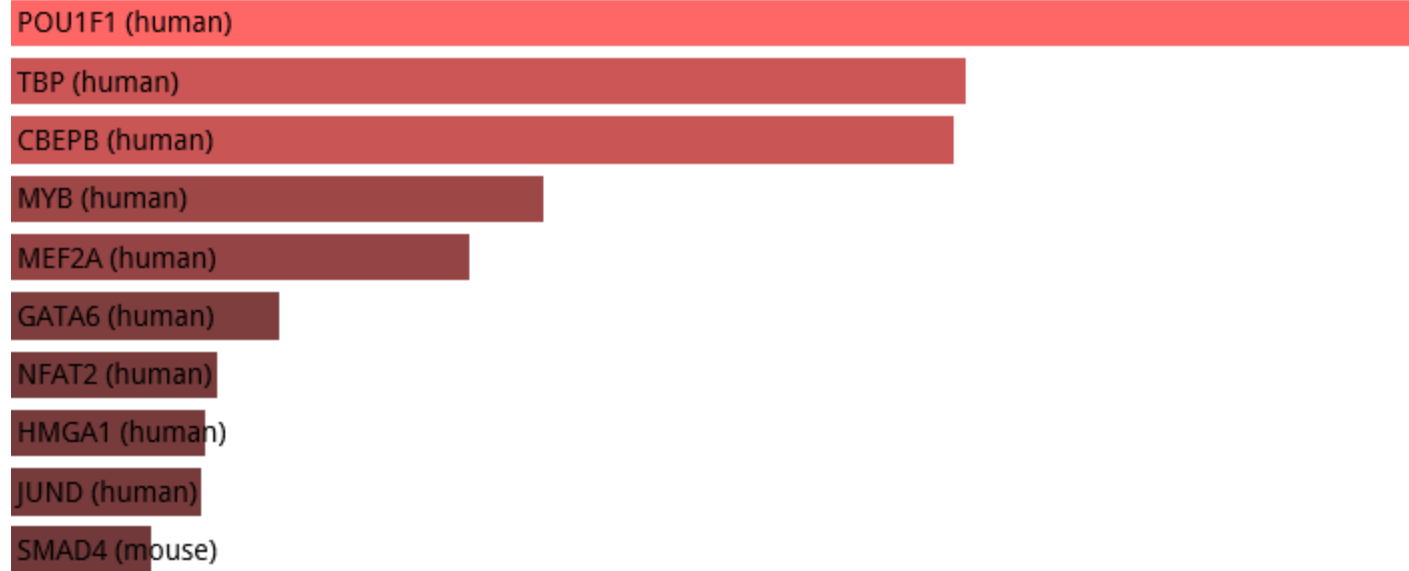
11,878 fixed human-specific insertions

TF Perturbations Followed by Expression 7979 genes

BCL11B_KO_MOUSE_GSE9330_CREEDSID_GENE_28_UP
SOX5_OE_FETALCORTEX_HUMAN_GSE89057_RNASEQ_UP
POU5F1_KD_HUMAN_GSE21135_CREEDSID_GENE_1329_UP
POU4F1_KO_MOUSE_GSE19997_CREEDSID_GENE_2947_UP
GATA6_OE_HESC_HUMAN_GSE69322_KSRMEDIA_RNASEQ_DOWN
DMRTA2_OE_MOUSE_GSE25179_CREEDSID_GENE_2204_DOWN
ZEB2_SHRNA_H1_HUMAN_GSE69618_DAY6_RNASEQ_DOWN
PITX2_KO_786O_HUMAN_GSE67844_RNASEQ_UP
NFKB1_ACTIVATION_HUMAN_GSE20736_CREEDSID_GENE_2522_DOWN
REST_INHIBITION_HUMAN_GSE40695_CREEDSID_GENE_1710_UP

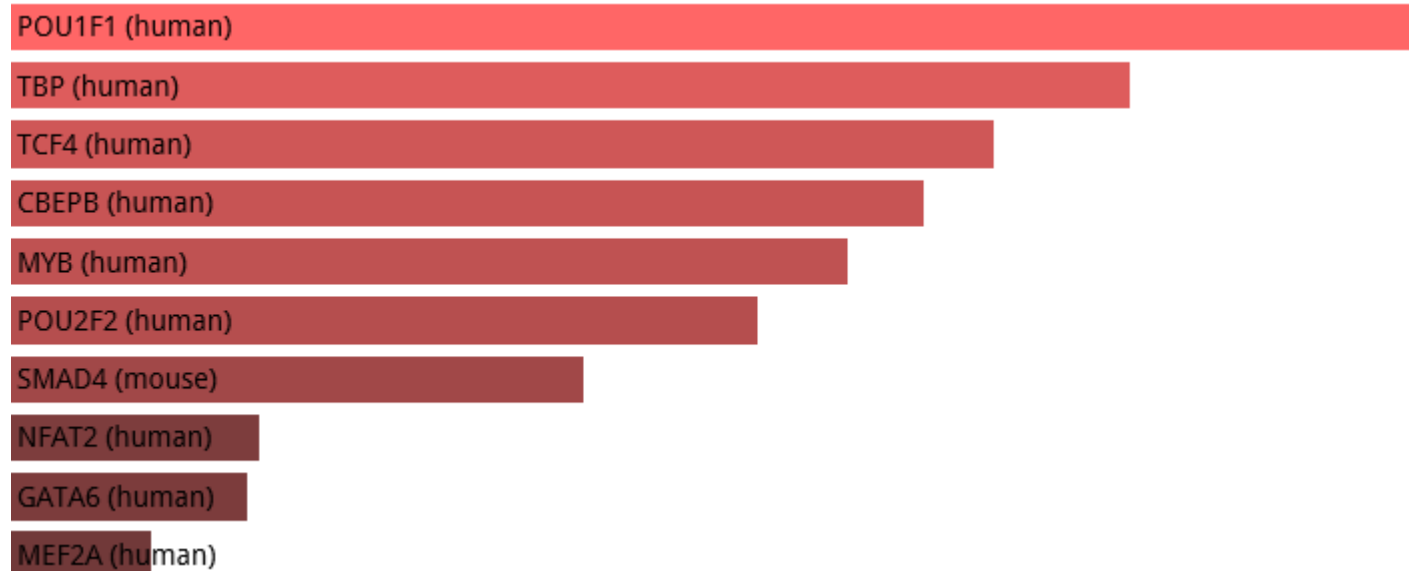
4,637 human-specific TE-encoded loci expressed in human DLPFC

TRANSFAC and JASPAR PWMs 4051 genes



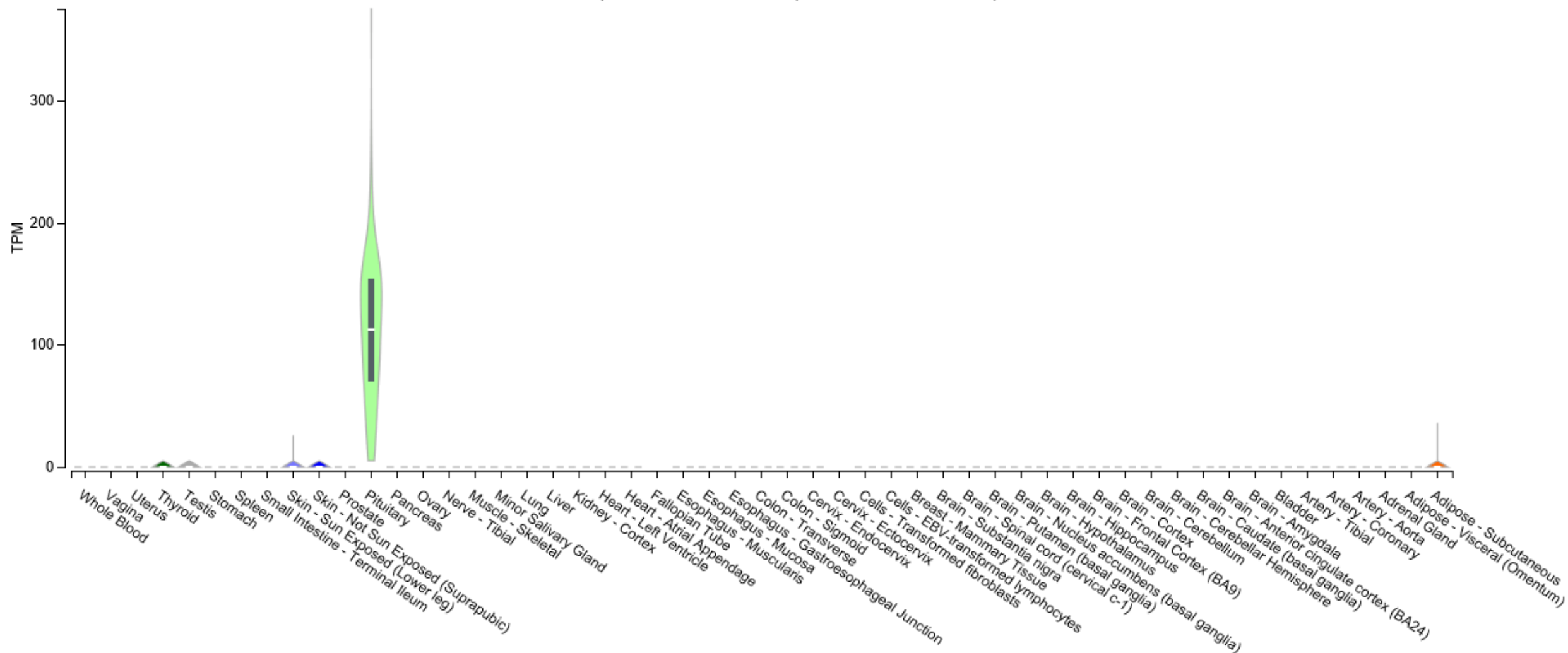
11,878 fixed human-specific insertions

TRANSFAC and JASPAR PWMs 7979 genes



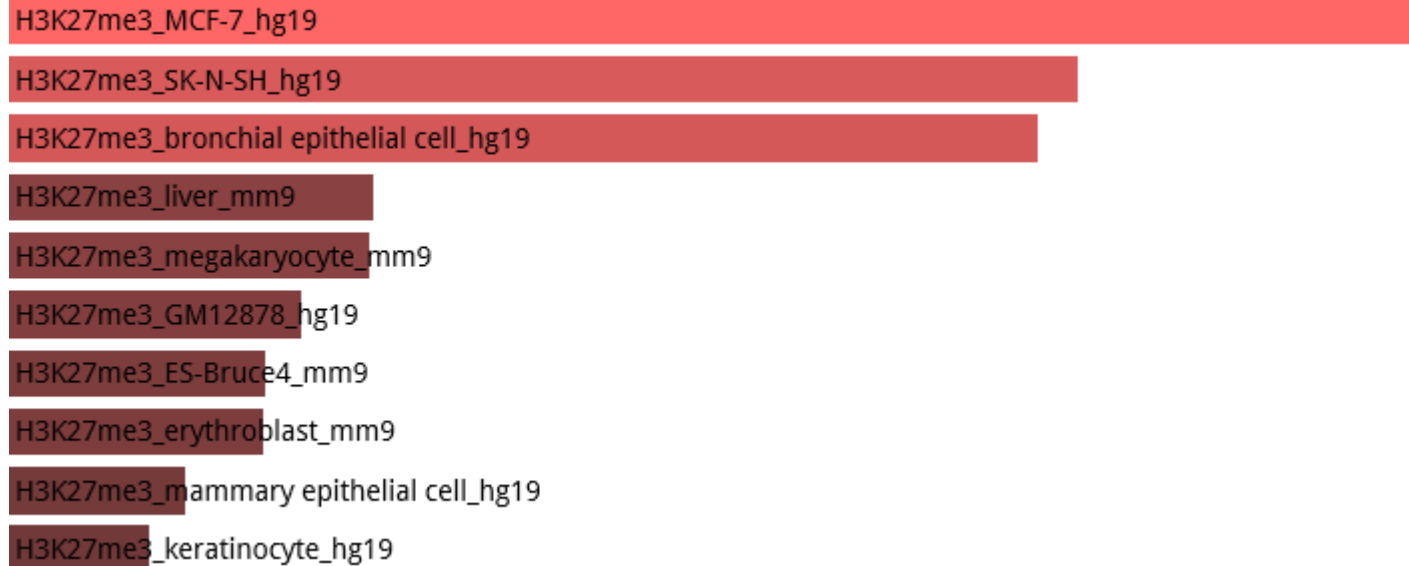
RNA-Seq Expression Data from GTEx (53 Tissues, 570 Donors)

Gene expression for POU1F1 (ENSG00000064835.6)



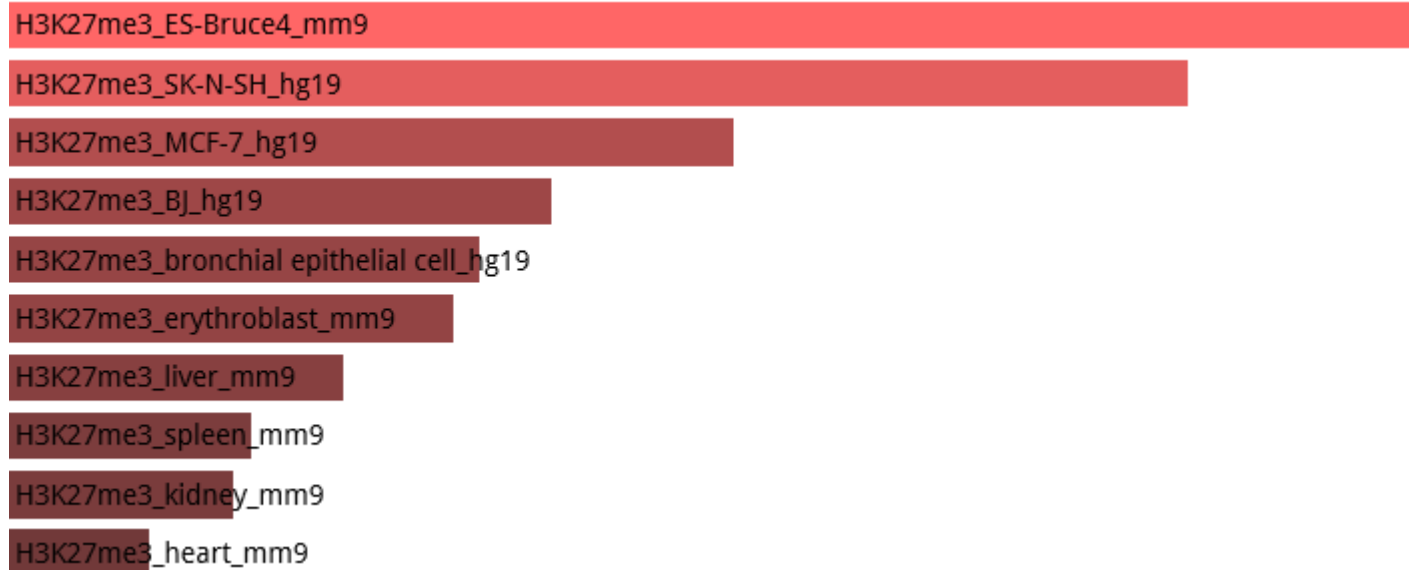
4,637 human-specific TE-encoded loci expressed in human DLPFC

ENCODE Histone Modifications 2015 4051 genes



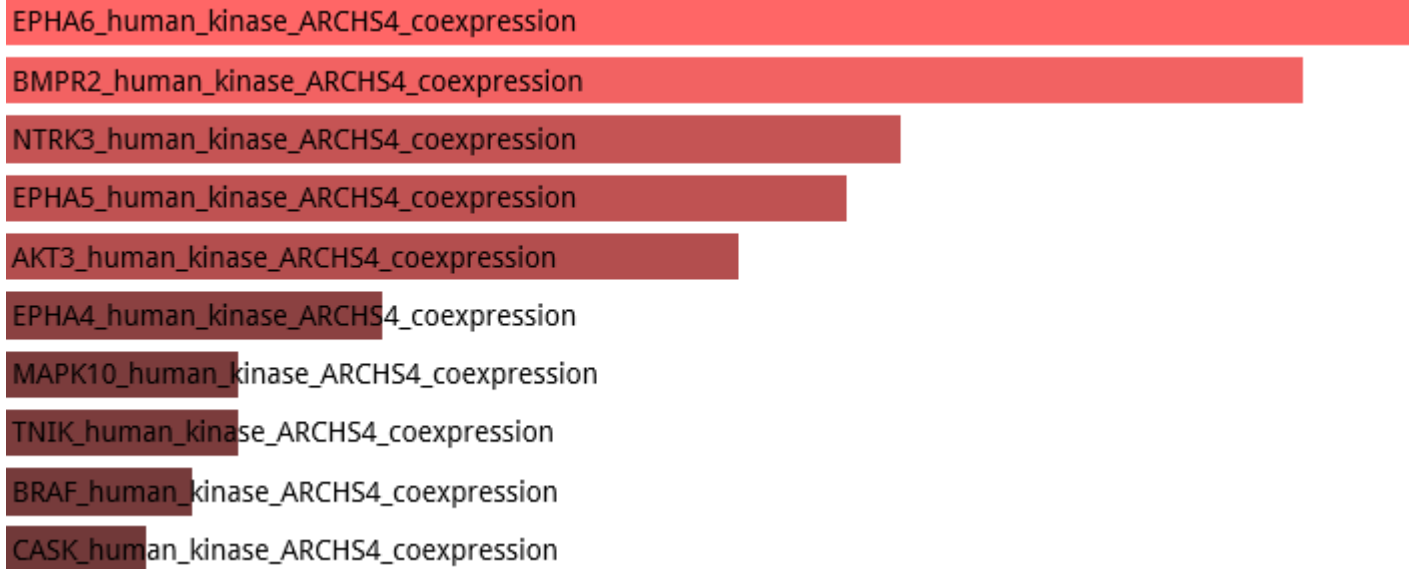
11,878 fixed human-specific insertions

ENCODE Histone Modifications 2015 7979 genes genes



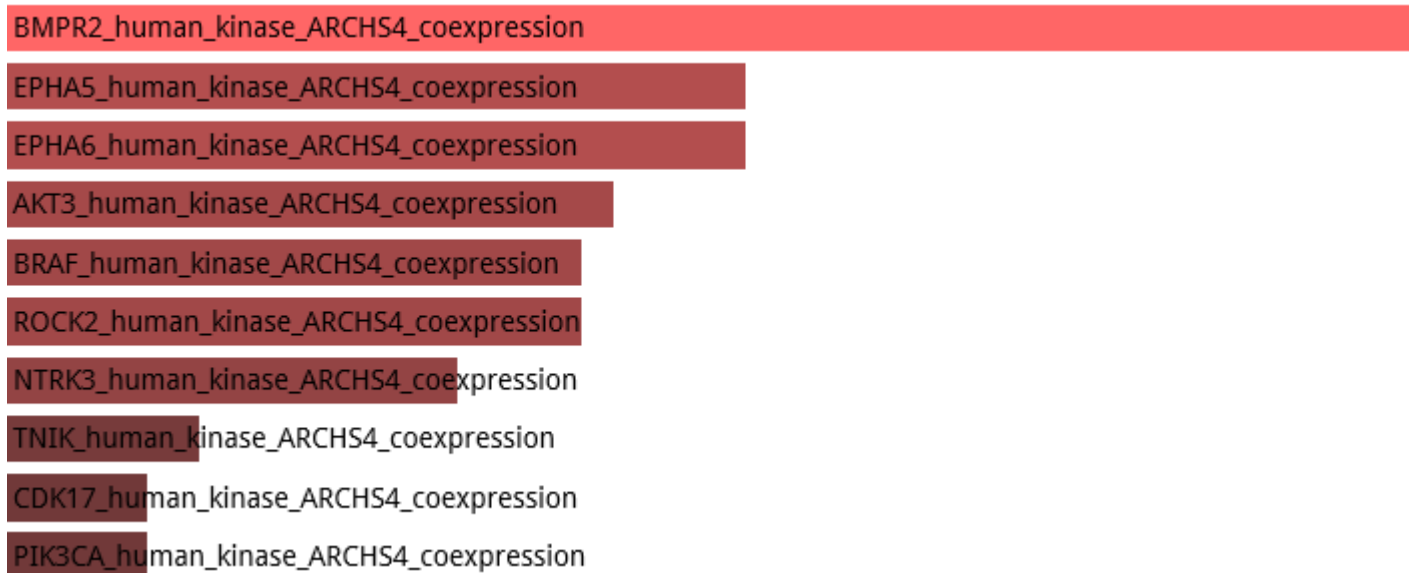
4,637 human-specific TE-encoded loci expressed in human DLPFC

ARCHS4 Kinases Co-expression 4051 genes



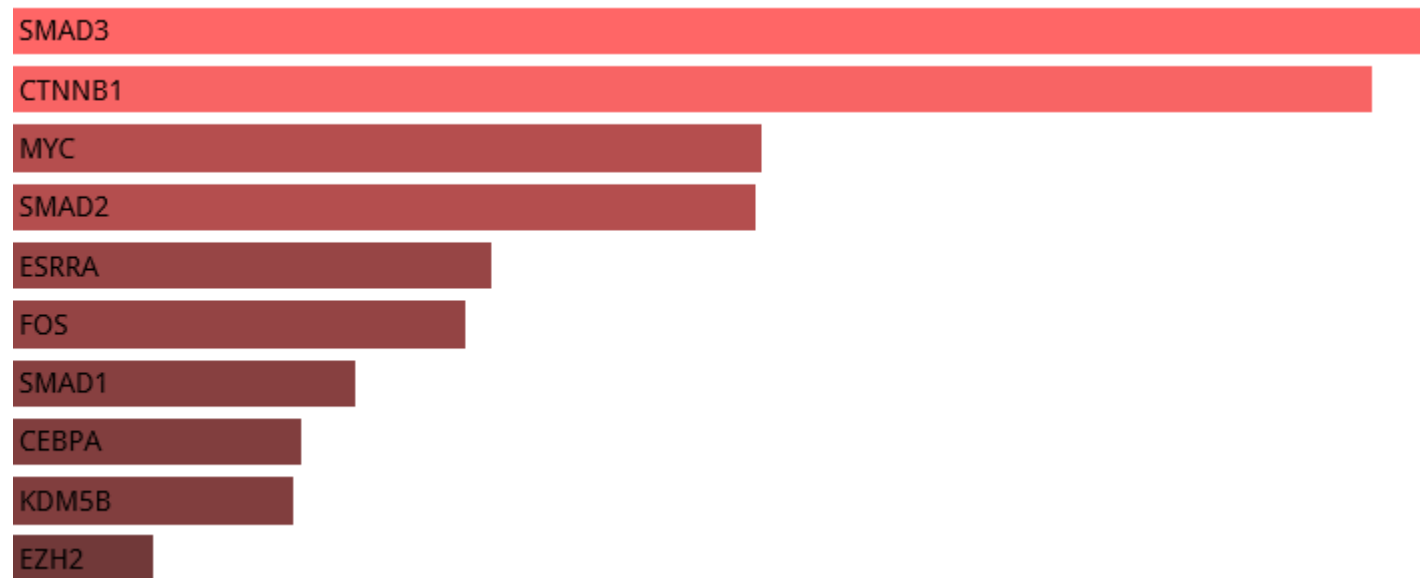
11,878 fixed human-specific insertions

ARCHS4 Kinases Co-expression 7979 genes genes



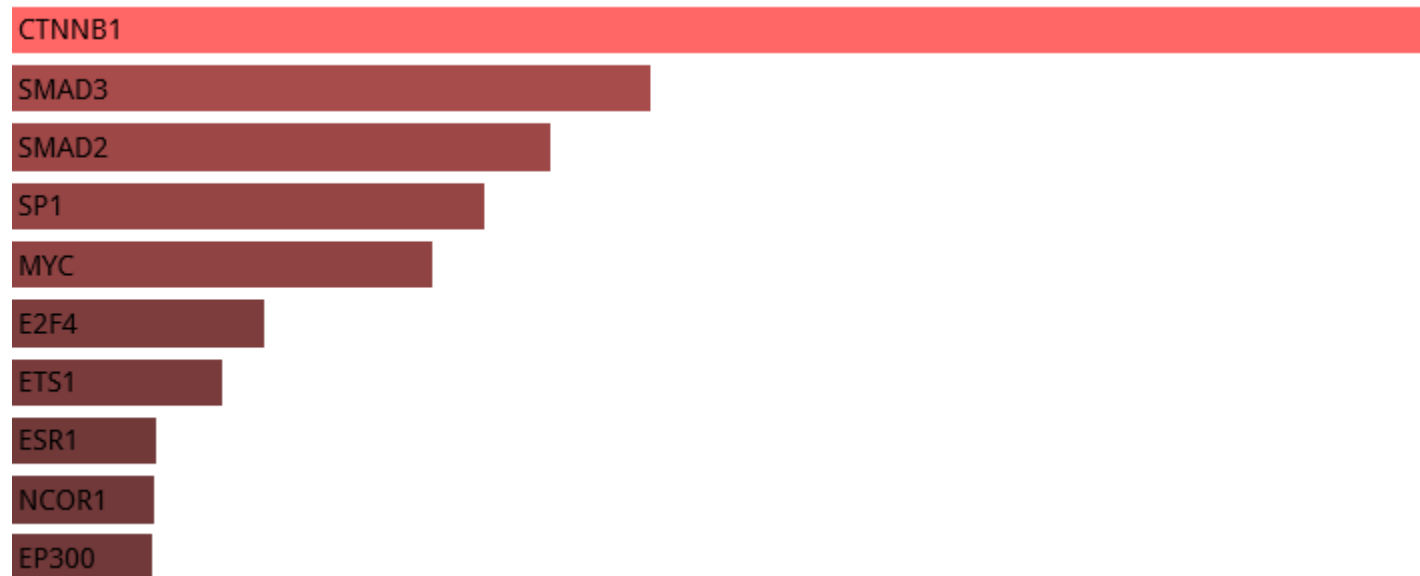
4,637 human-specific TE-encoded loci expressed in human DLPFC

Transcription Factor PPIs 4051 genes



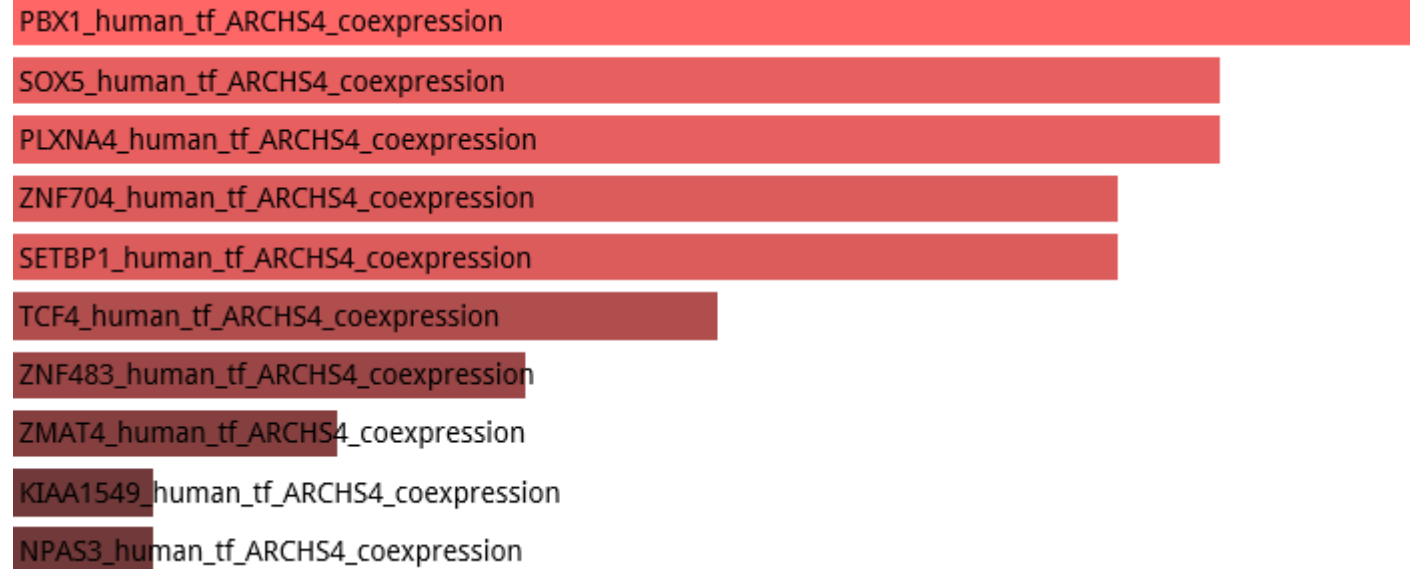
11,878 fixed human-specific insertions

Transcription Factor PPIs 7979 genes



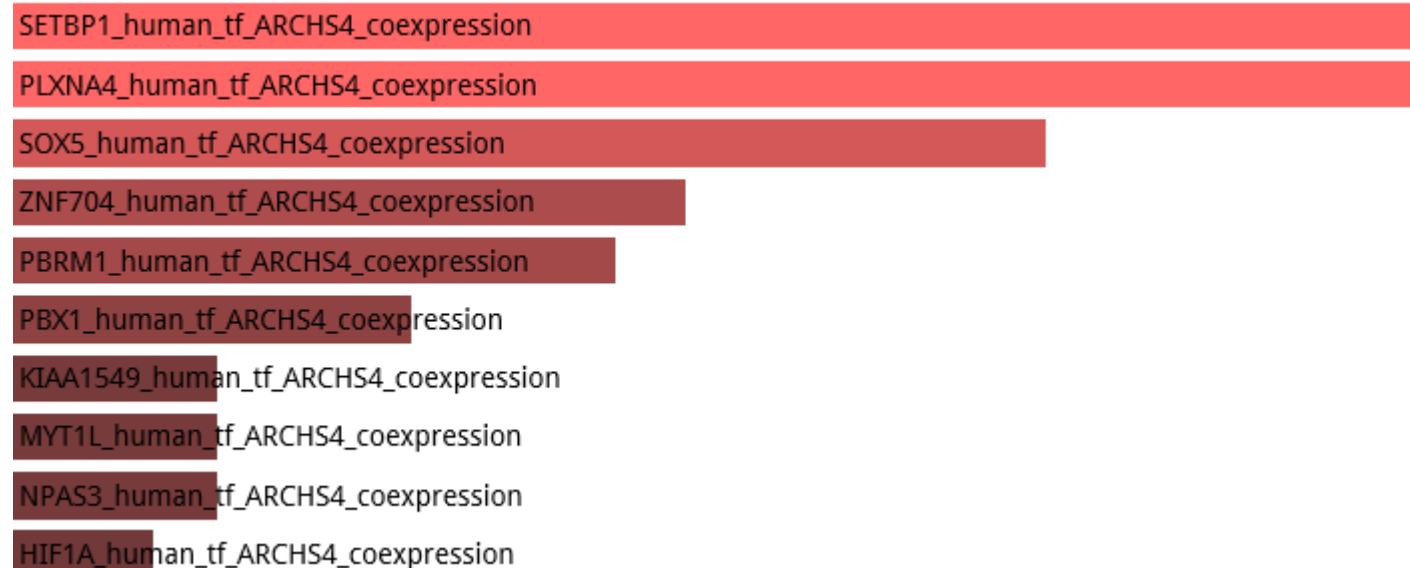
4,637 human-specific TE-encoded loci expressed in human DLPFC

ARCHS4 TFs Co-expression 4051 genes



11,878 fixed human-specific insertions

ARCHS4 TFs Co-expression 7979 genes



4,637 human-specific TE-encoded loci expressed in human DLPFC

ESCAPE 4051 genes

JARID2-20075857_UP	9.91998E-22	2.95615E-19
CHiP_SUZ12-18974828	1.24005E-14	1.84767E-12
mESC_H3K27me3_17603471	3.60039E-13	3.57639E-11
CHiP_SUZ12-18692474	2.53887E-11	1.89146E-09
CHiP_SUZ12-18555785	1.53774E-10	9.16493E-09
CHiP_JARID2-20064375	2.44213E-10	1.21292E-08
CHiP_EZH2-18974828	6.03372E-10	2.56864E-08
CHiP_MTF2-20144788	2.38371E-09	8.87931E-08
hESC_H3K27me3_20682450	2.0445E-08	6.76958E-07
CHiP_RNF2-22325148	8.34666E-08	2.4873E-06

11,878 fixed human-specific insertions

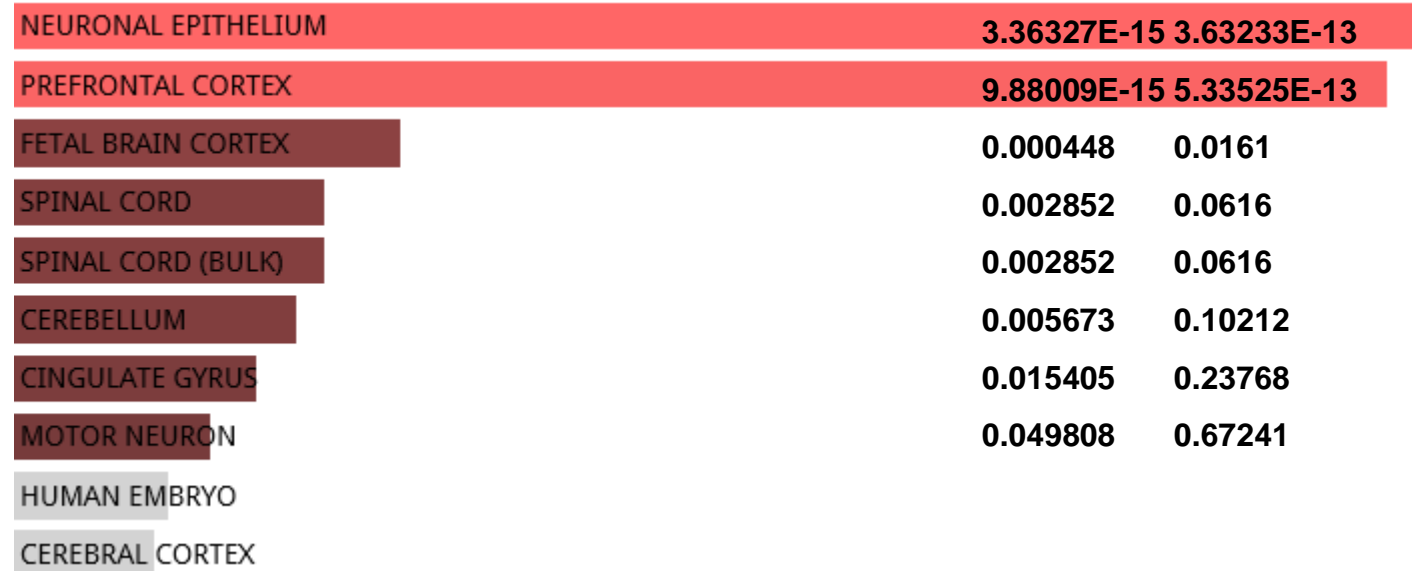
ESCAPE 7979 genes

mESC_H3K27me3_17603471	4.44942E-51	1.37932E-48
CHiP_SUZ12-18974828	2.39781E-38	3.71661E-36
hESC_H3K27me3_20682450	2.69286E-37	2.78263E-35
CHiP_SUZ12-18692474	3.02417E-32	2.34373E-30
CHiP_MTF2-20144788	5.57499E-32	3.4565E-30
CHiP_EZH2-18974828	1.0298E-31	5.32065E-30
mESC_H3K9me3_19884255	4.45745E-27	1.97402E-25
CHiP_JARID2-20064375	4.72639E-26	1.83148E-24
CHiP_RNF2-22325148	4.92907E-25	1.69779E-23
CHiP_RNF2-18974828	3.7711E-24	1.16904E-22

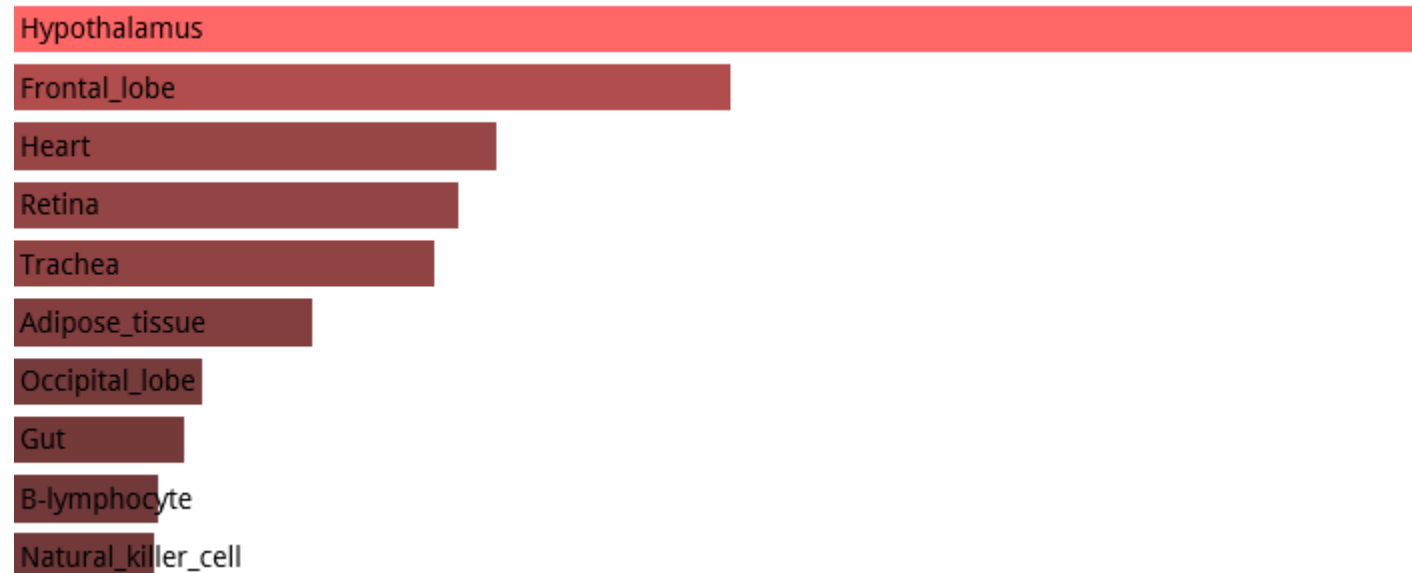
7,599 duplicated regions in the human genome

7,599 duplicated regions in the human genome

ARCHS4 Tissues 6618 genes

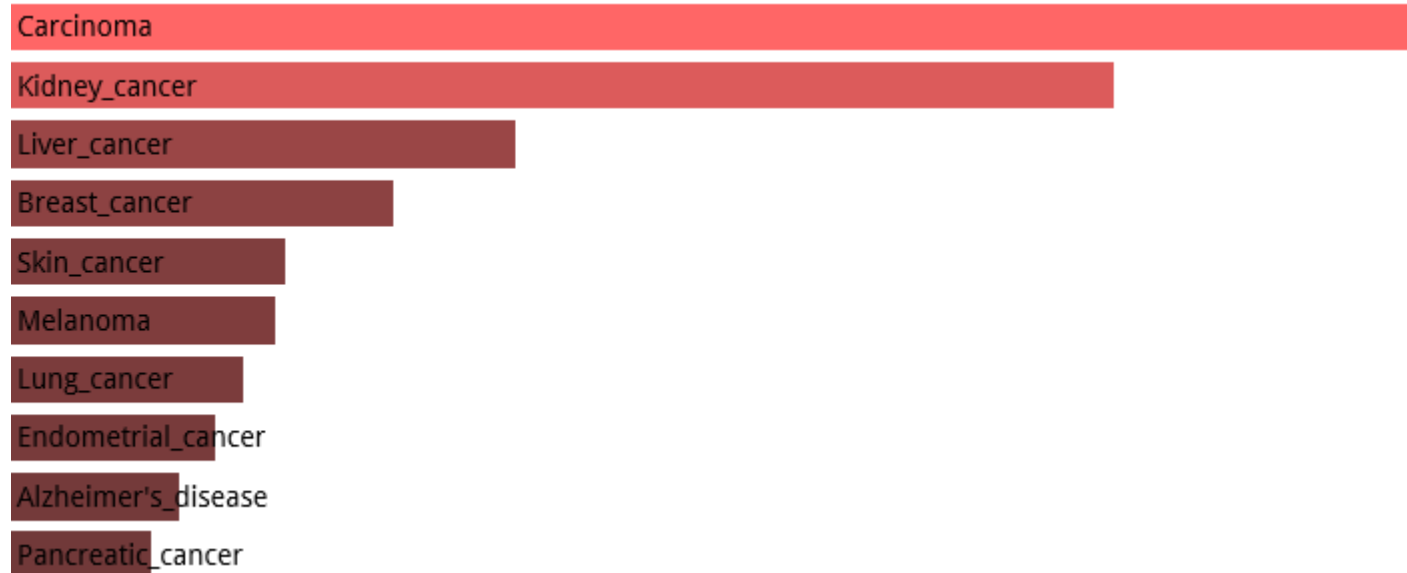


Jensen TISSUES 6618 genes

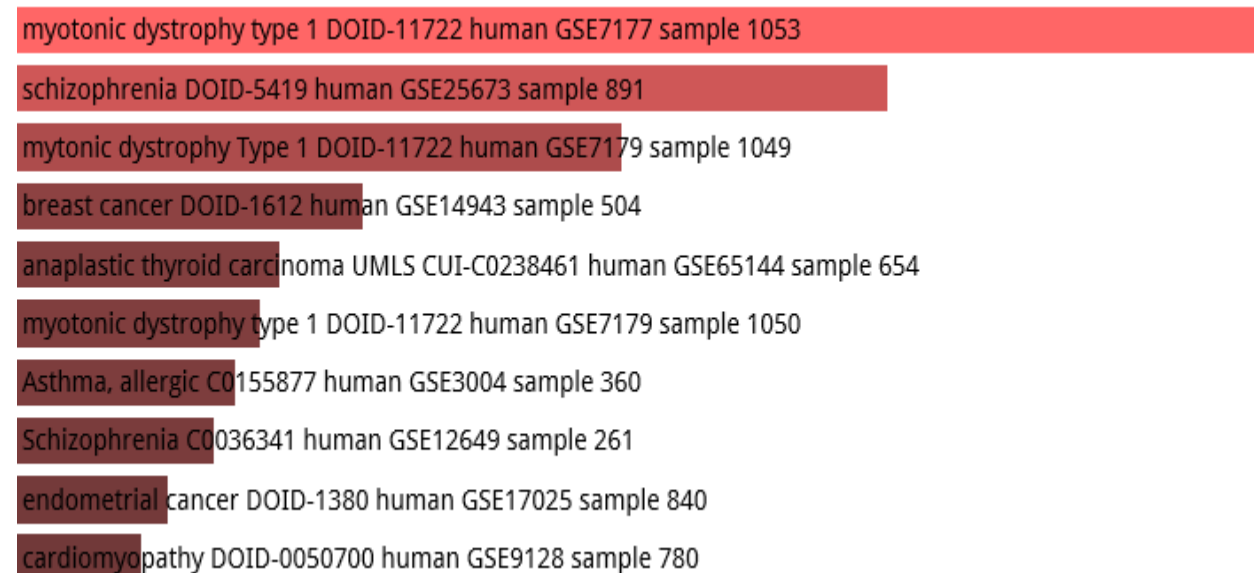


7,599 duplicated regions in the human genome

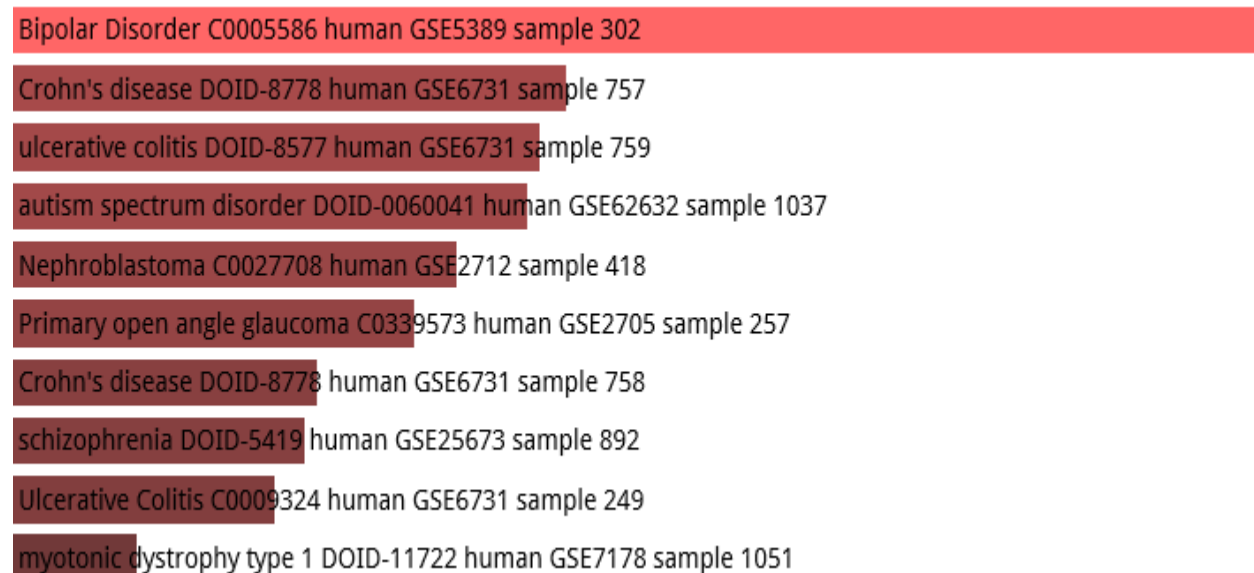
Jensen DISEASES 6618 genes



Disease Perturbations from GEO up



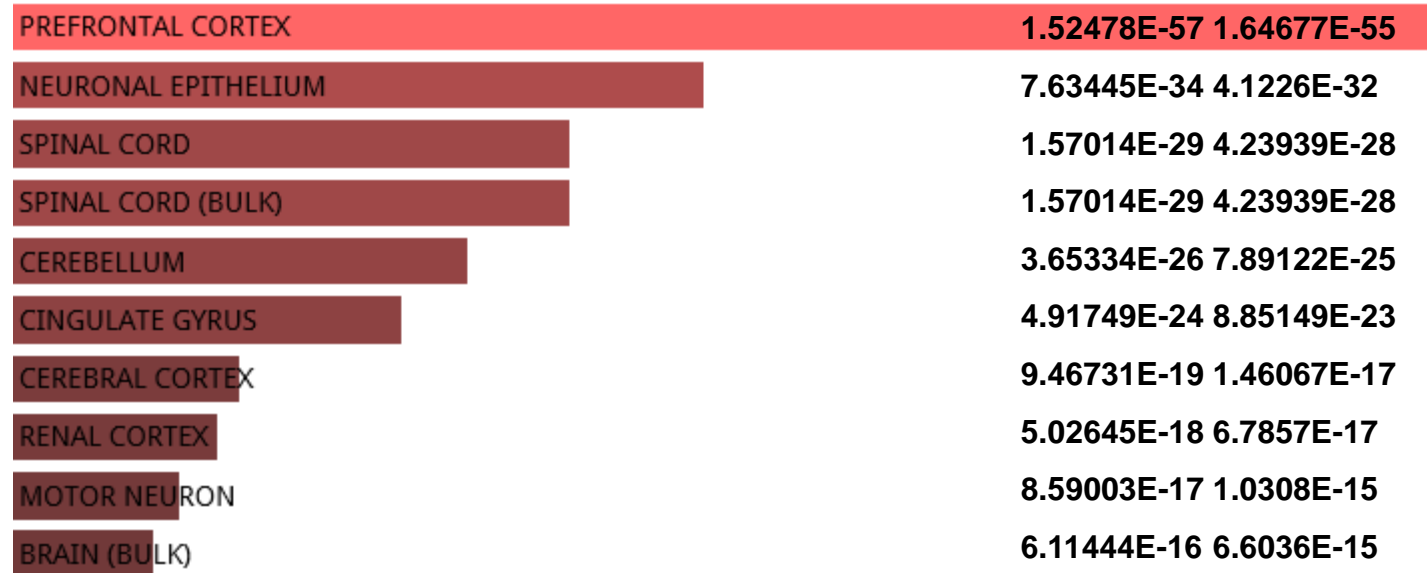
Disease Perturbations from GEO down



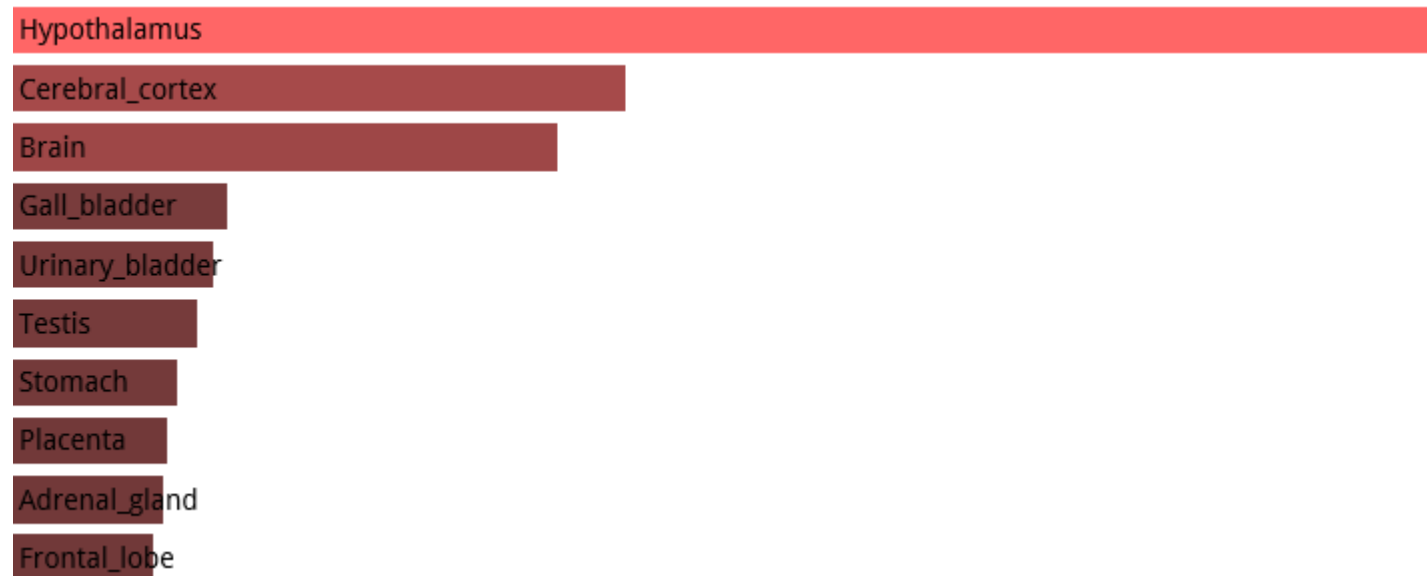
5,883 fixed human-specific deletions

5,883 fixed human-specific deletions

ARCHS4 Tissues 5489 genes



Jensen TISSUES 5489 genes



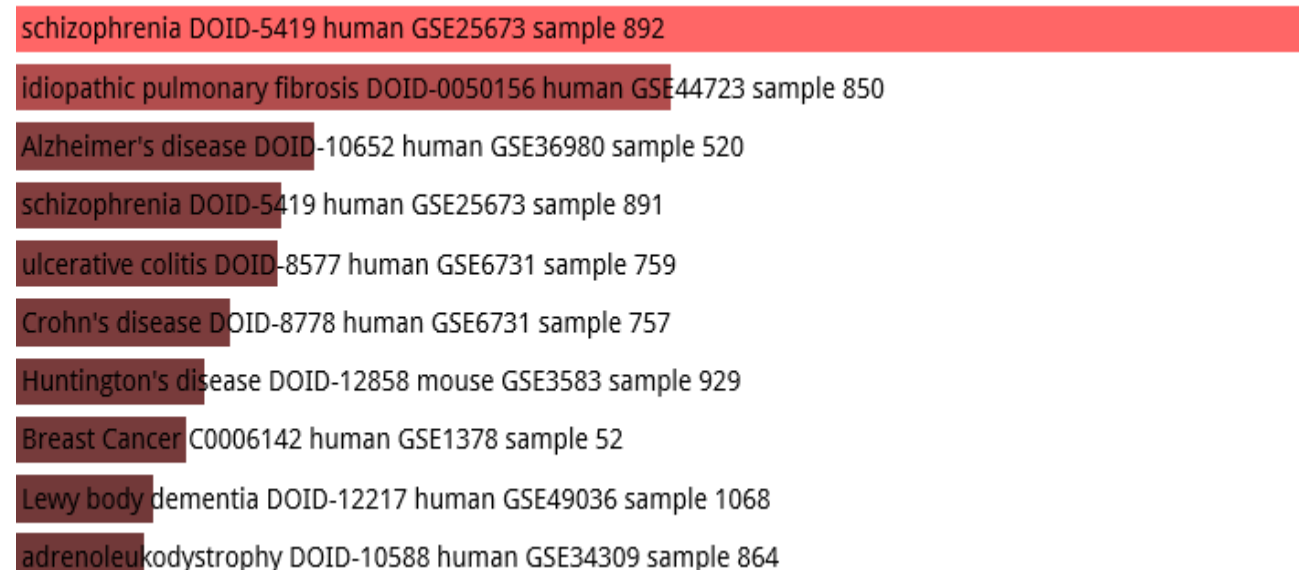
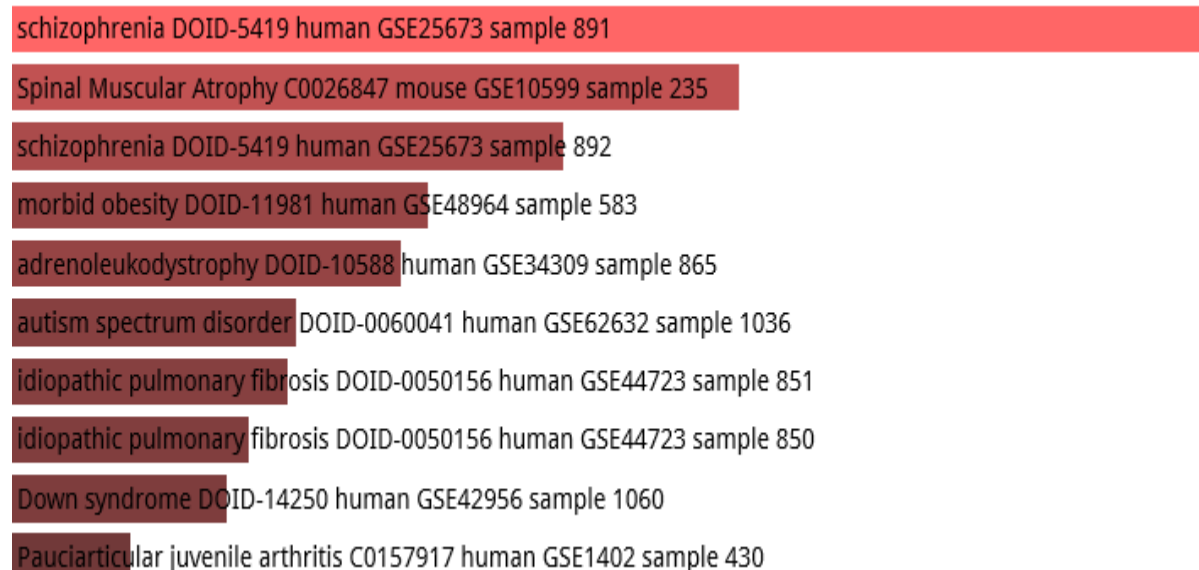
5,883 fixed human-specific deletions

Jensen DISEASES 5489 genes



Disease Perturbations from GEO up

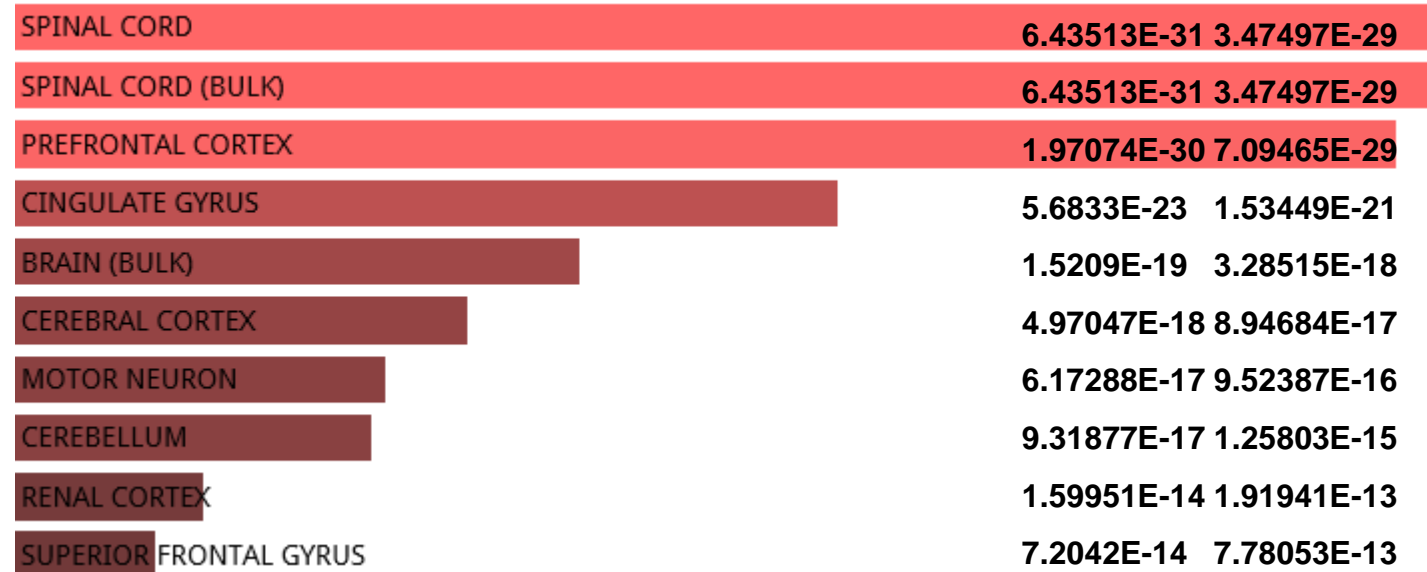
Disease Perturbations from GEO down



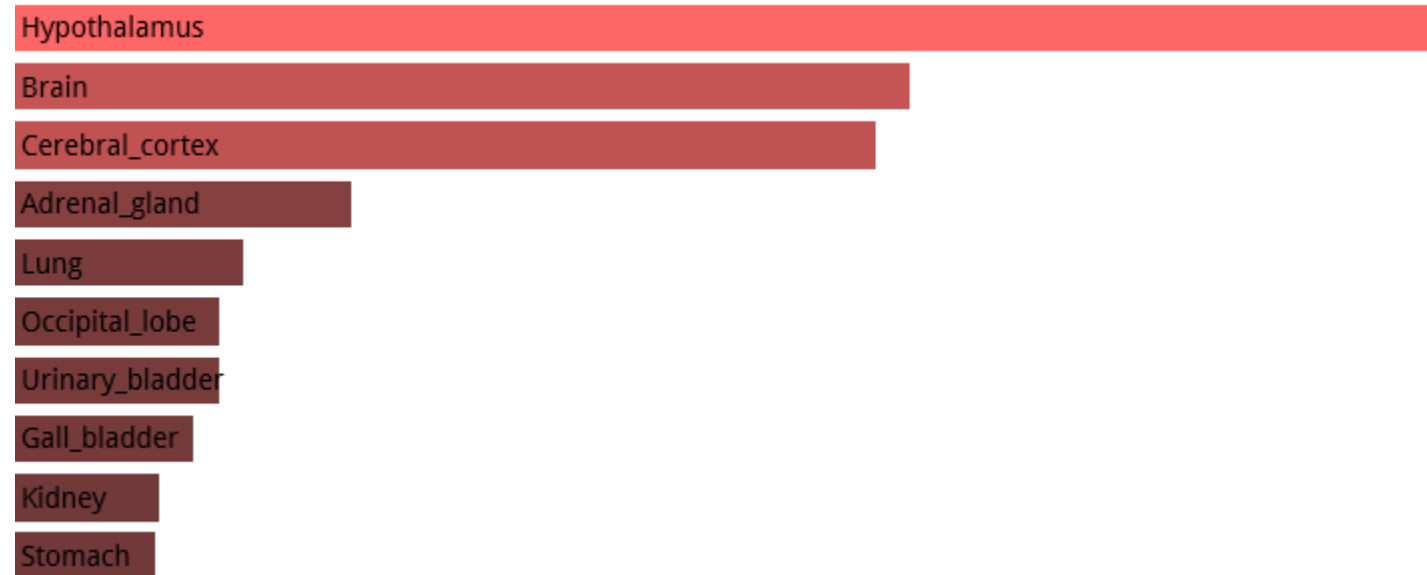
4,875 human-specific STR expansions

4,875 human-specific STR expansions

ARCHS4 Tissues 4844 genes

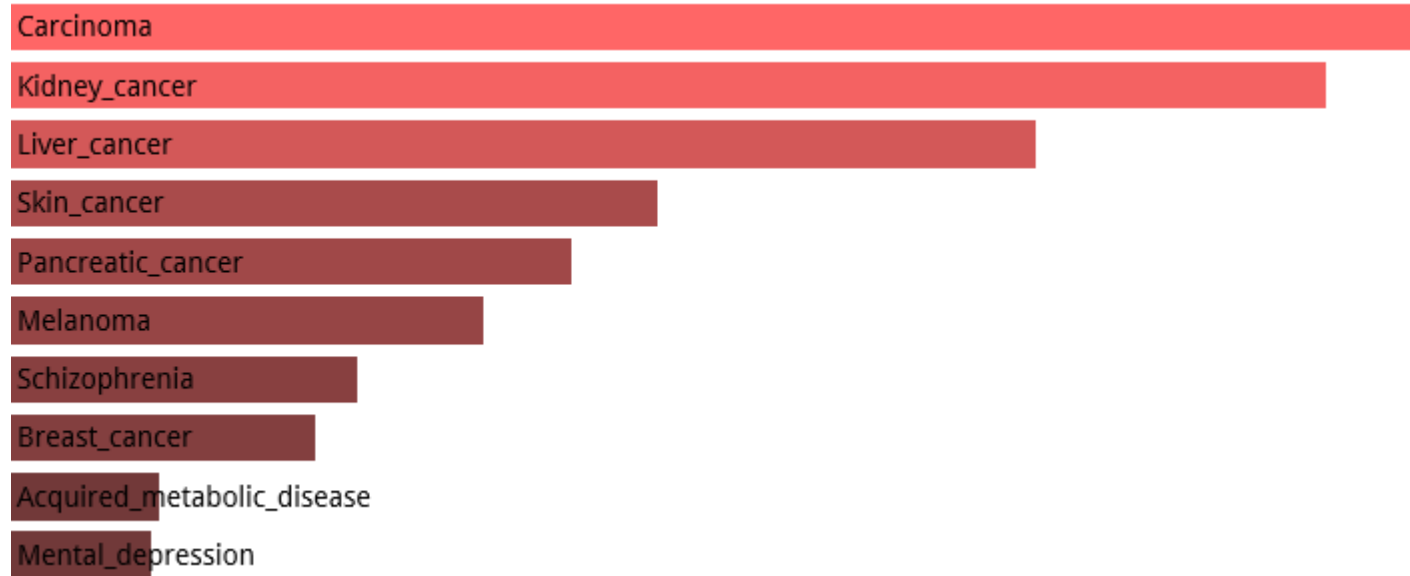


Jensen TISSUES 4844 genes



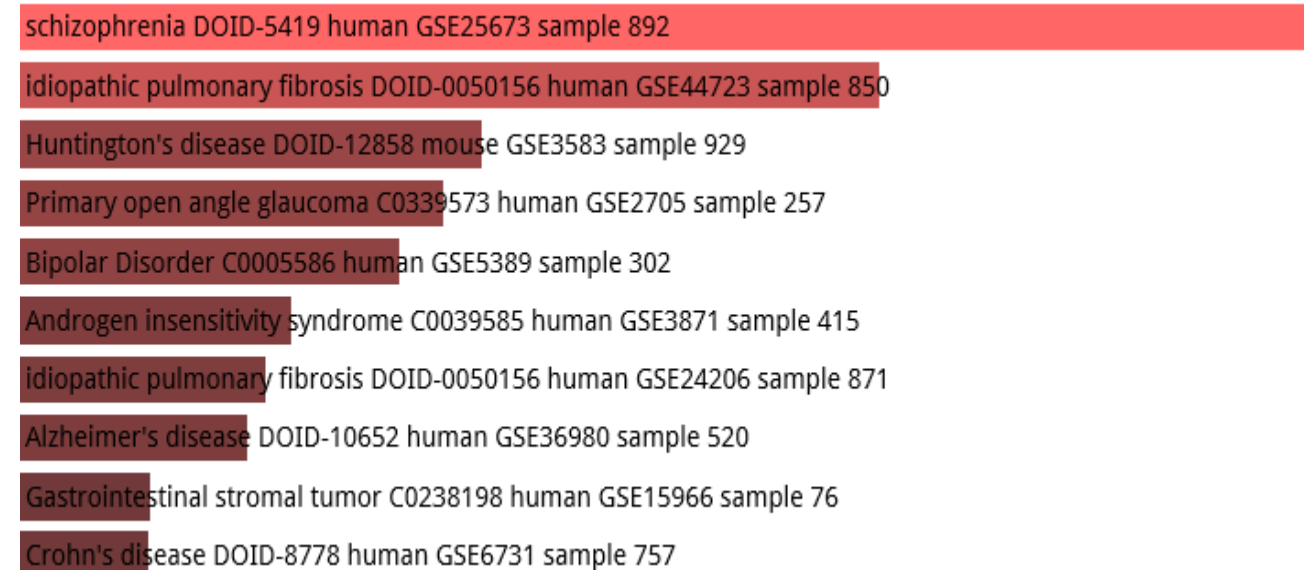
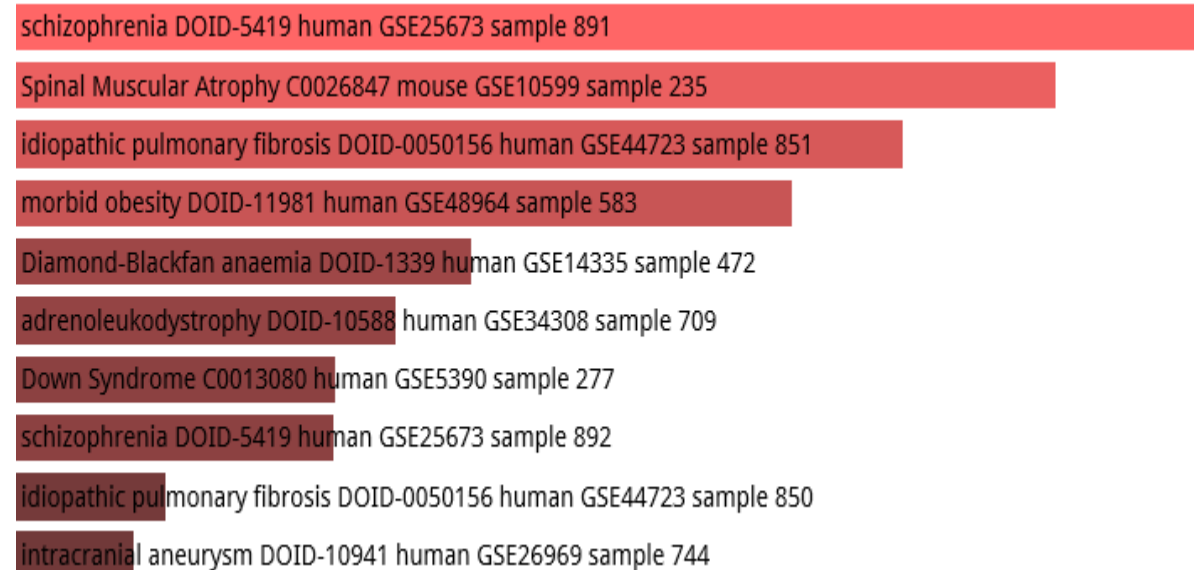
4,875 human-specific STR expansions

Jensen DISEASES 4844 genes



Disease Perturbations from GEO up

Disease Perturbations from GEO down



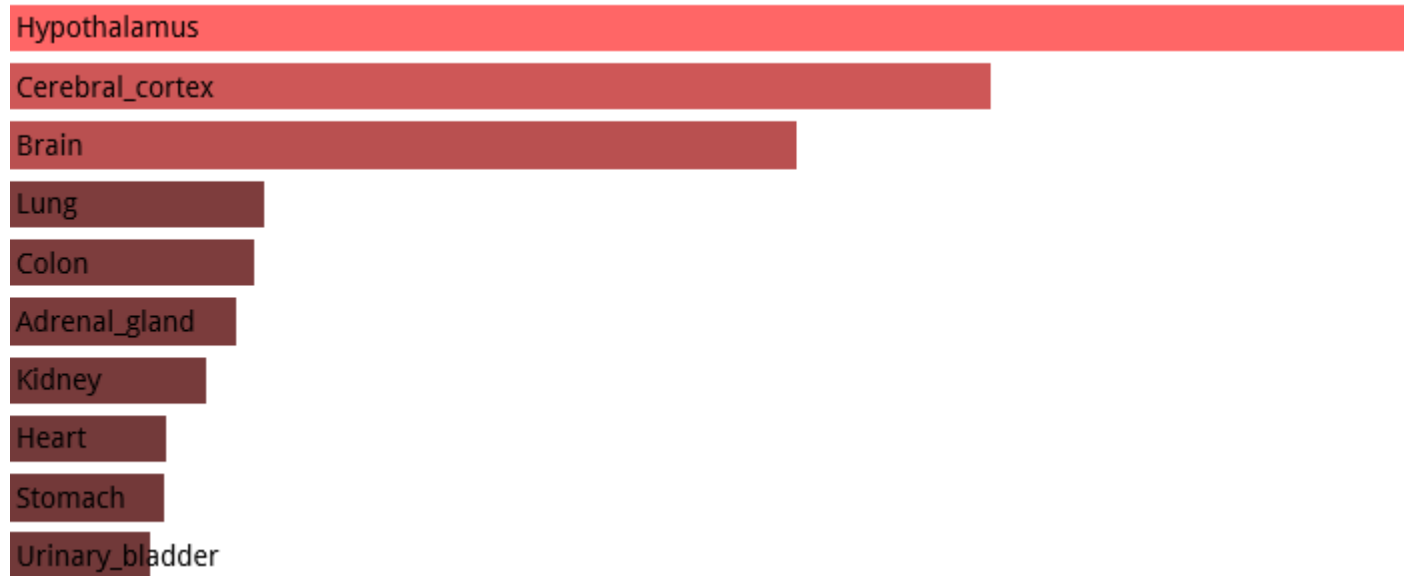
3,538 human-specific ace-DHS

3,538 human-specific ace-DHS

ARCHS4 Tissues 3445 genes



Jensen TISSUES 3445 genes

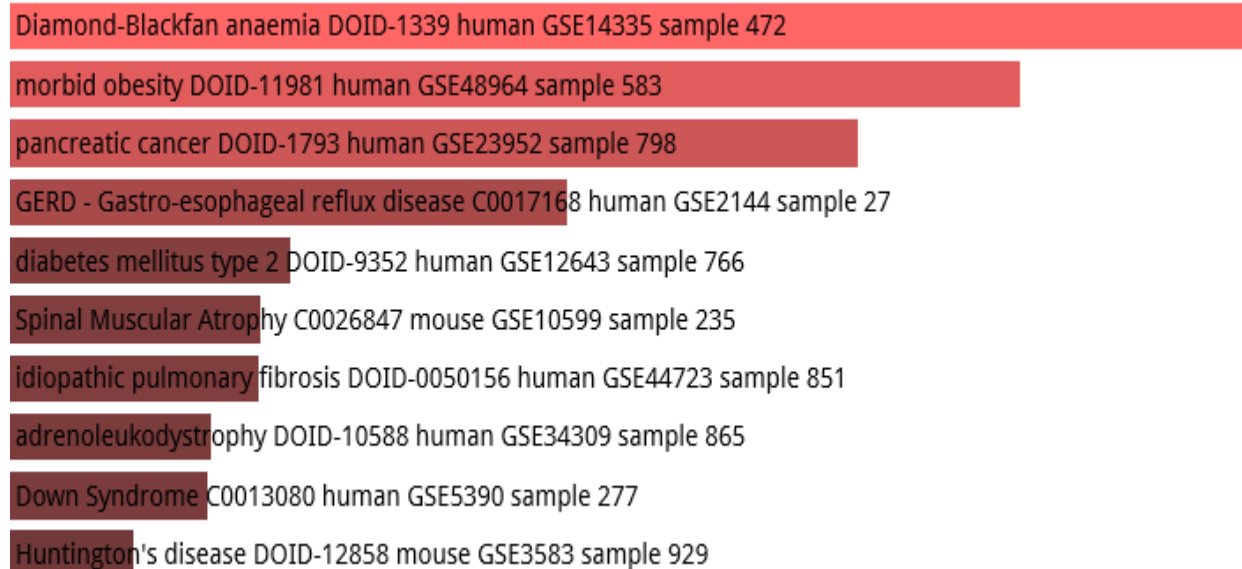


3,538 human-specific ace-DHS

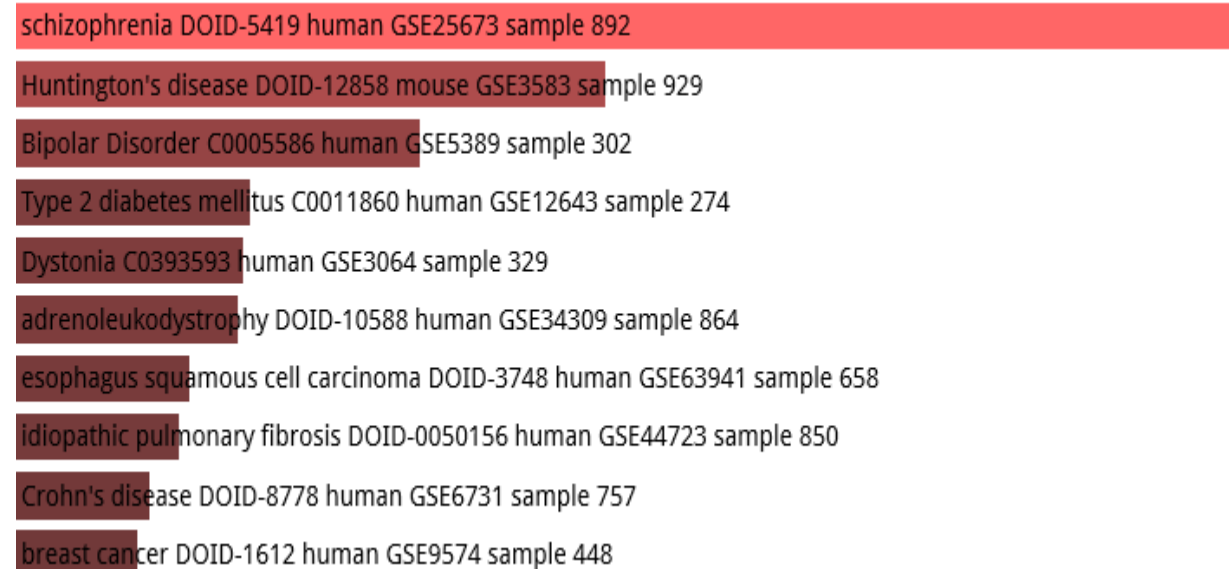
Jensen DISEASES 3445 genes



Disease Perturbations from GEO up

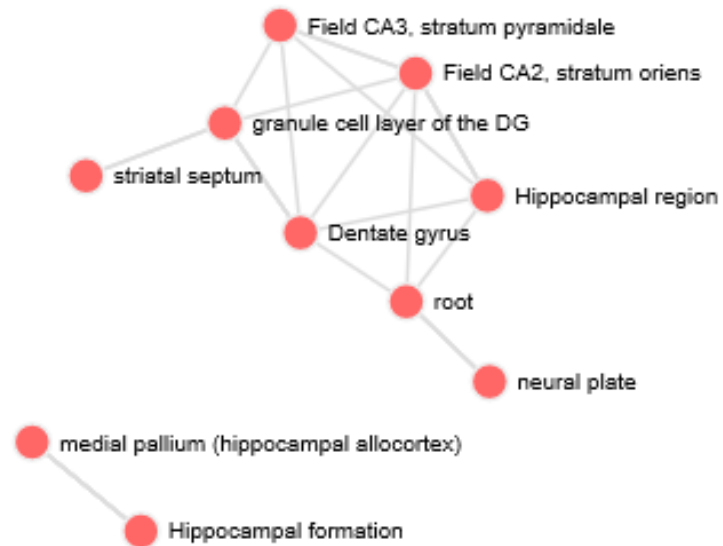
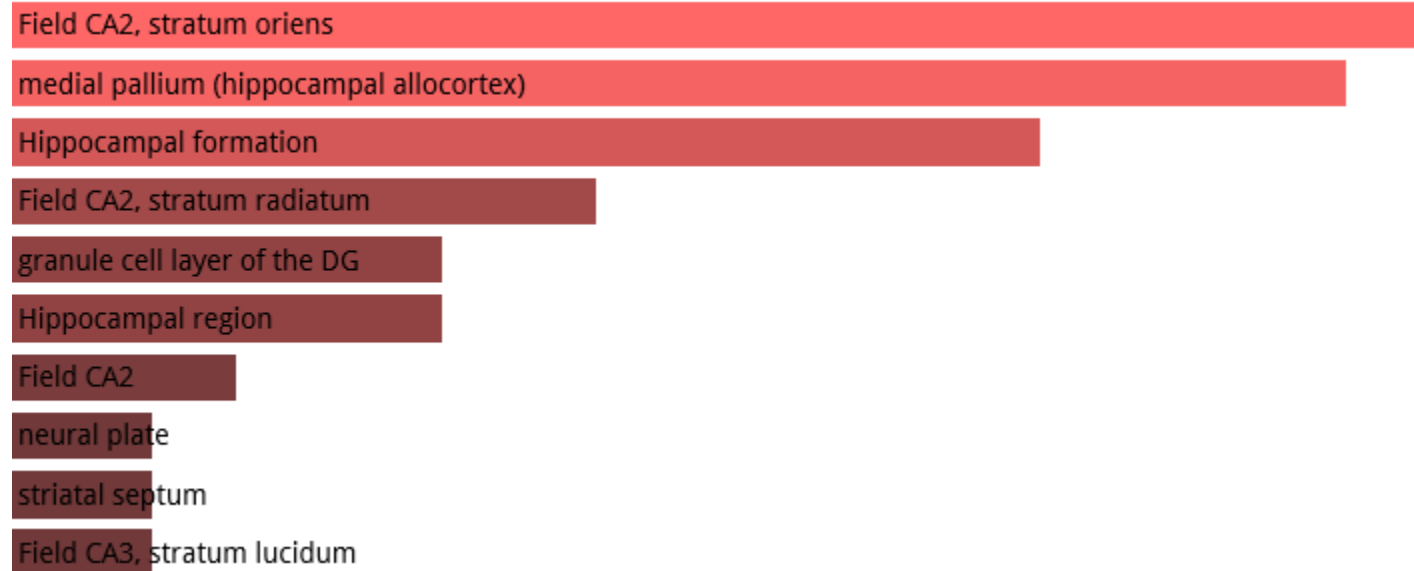


Disease Perturbations from GEO down



3,538 human-specific ace-DHS

Allen Brain Atlas up 3445 genes



3,803 human-specific TFBS in hESC

3,803 human-specific TFBS in hESC

ARCHS4 Tissues 1087 genes

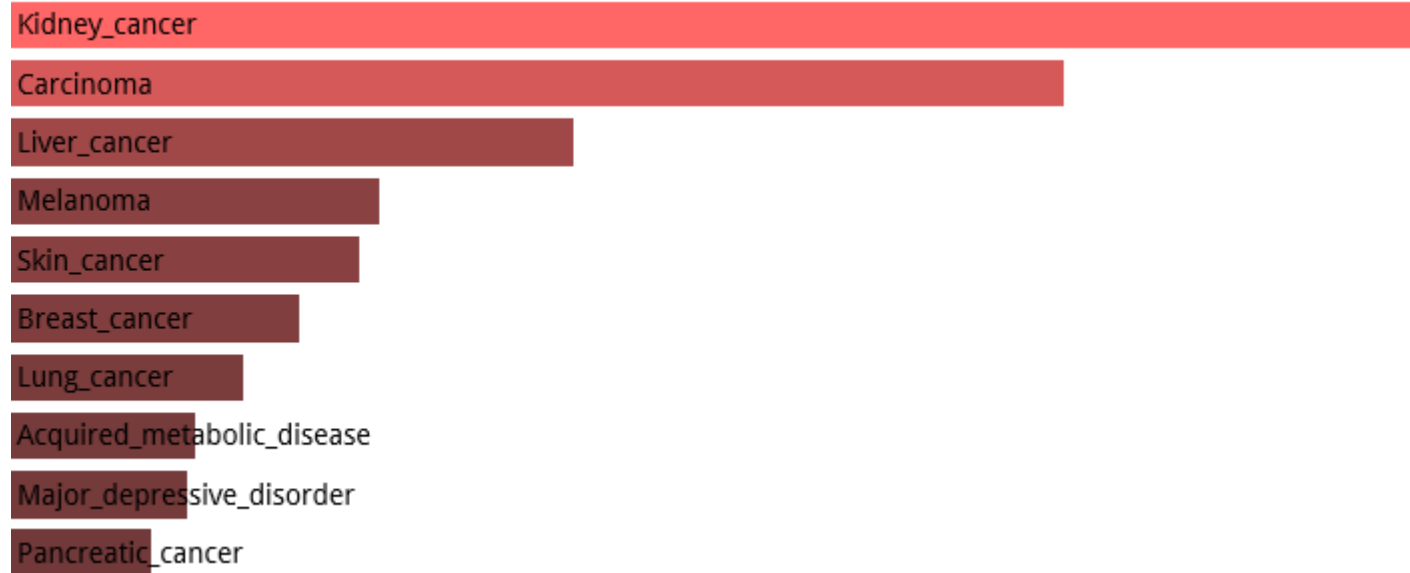
PREFRONTAL CORTEX	8.51152E-18	9.19244E-16
SPINAL CORD	3.57976E-17	1.28871E-15
SPINAL CORD (BULK)	3.57976E-17	1.28871E-15
CEREBELLUM	2.94817E-16	6.36805E-15
CINGULATE GYRUS	2.94817E-16	6.36805E-15
MOTOR NEURON	2.62138E-12	4.71848E-11
NEURONAL EPITHELIUM	8.6747E-12	1.33838E-10
SENSORY NEURON	2.7969E-11	3.35628E-10
CEREBRAL CORTEX	2.7969E-11	3.35628E-10
DORSAL STRIATUM	2.68742E-10	2.63856E-09

Jensen TISSUES 1087 genes

Hypothalamus		
Adult		
Ganglion		
Cerebellar_Purkinje_cell		
Neural_stem_cell		
Neural_tube		
Lumbar_spine		
dg1		
nt2d1		
Ec		

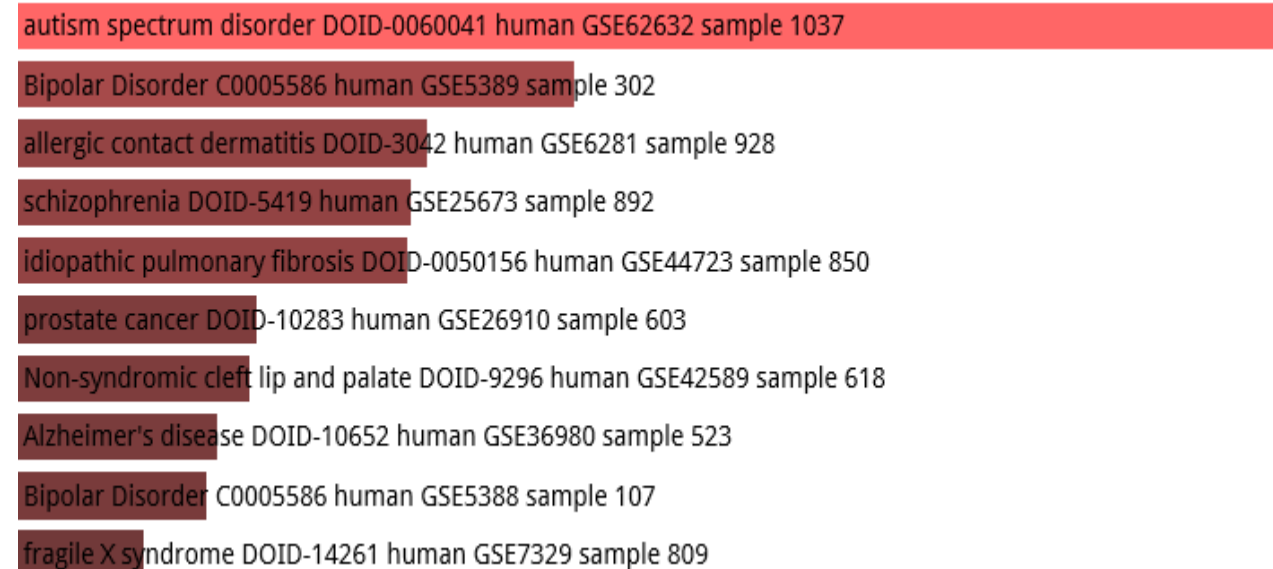
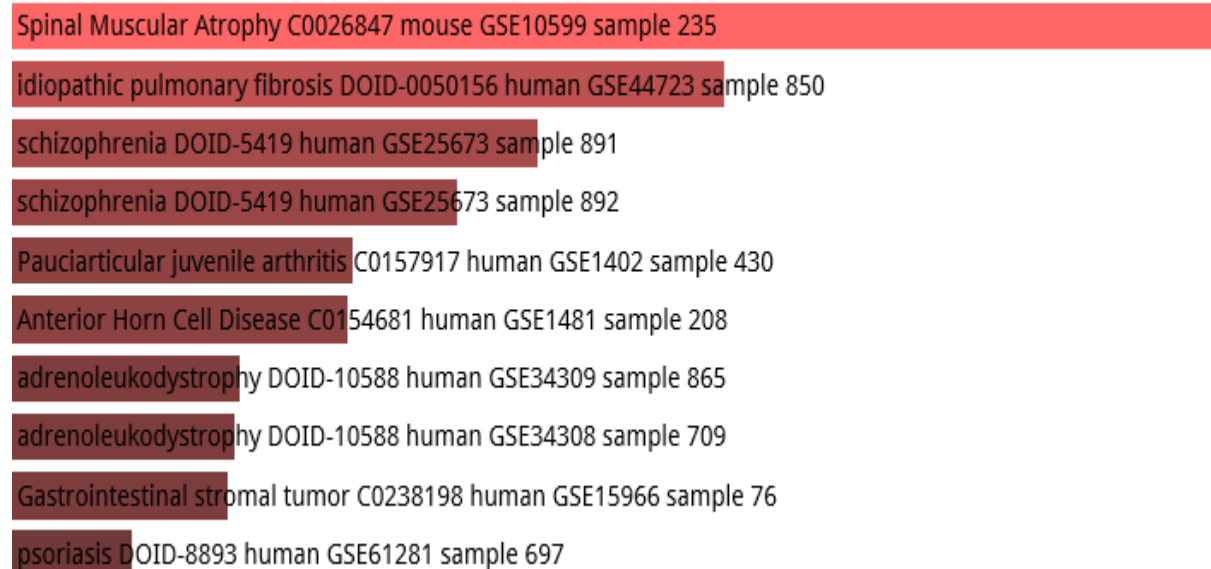
3,803 human-specific TFBS in hESC

Jensen DISEASES 1087 genes



Disease Perturbations from GEO up

Disease Perturbations from GEO down



4,249 fixed human-specific regulatory loci

4,249 fixed human-specific regulatory loci

ARCHS4 Tissues 2810 genes

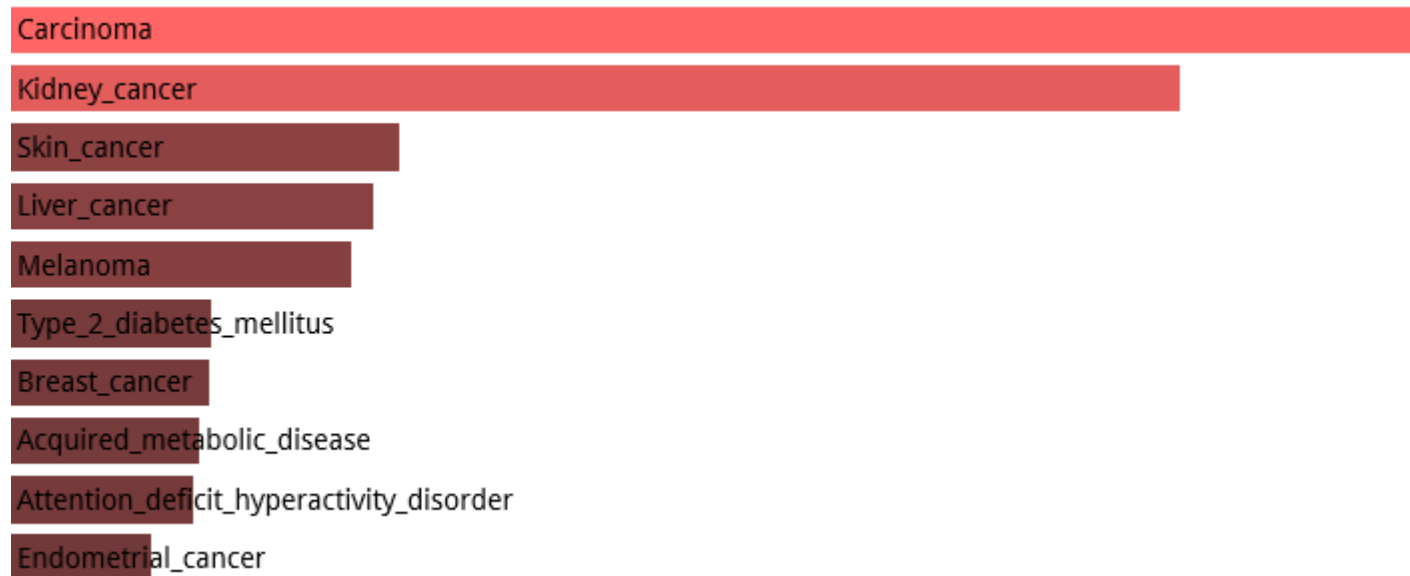
PREFRONTAL CORTEX	3.47228E-19	3.75007E-17
NEURONAL EPITHELIUM	3.67273E-13	1.98328E-11
SPINAL CORD	7.7021E-11	2.07957E-09
SPINAL CORD (BULK)	7.7021E-11	2.07957E-09
MOTOR NEURON	2.44617E-10	5.28373E-09
CEREBELLUM	5.19957E-10	9.35923E-09
CINGULATE GYRUS	2.2585E-09	3.48455E-08
SENSORY NEURON	2.54348E-07	3.43369E-06
RENAL CORTEX	8.70052E-07	1.04406E-05
FETAL BRAIN CORTEX	2.11278E-06	2.2818E-05

Jensen TISSUES 2810 genes

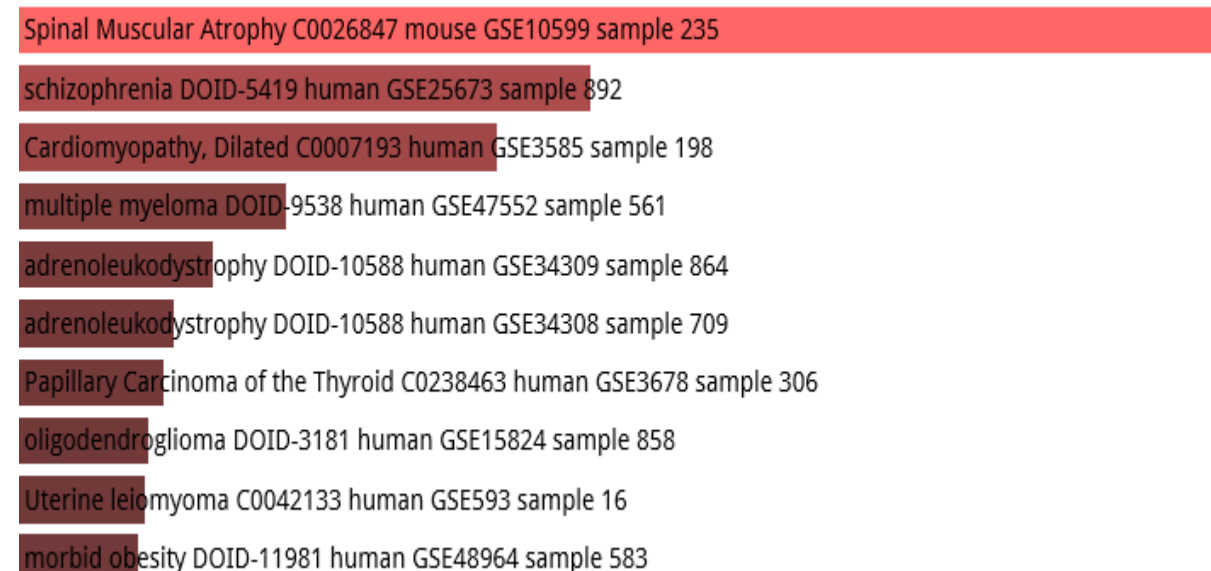
Hypothalamus		
Brain		
Cerebral_cortex		
Testis		
Colon		
Ovary		
Cardiac_muscle		
Urinary_bladder		
Gall_bladder		
Heart		

4,249 fixed human-specific regulatory loci

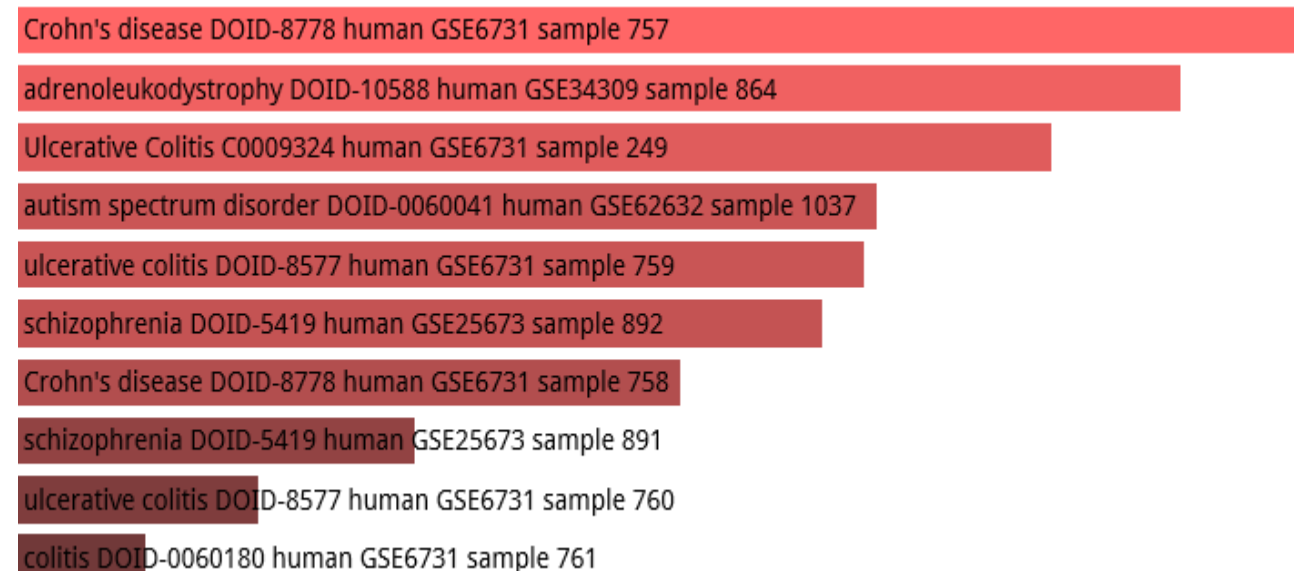
Jensen DISEASES 2810 genes



Disease Perturbations from GEO up



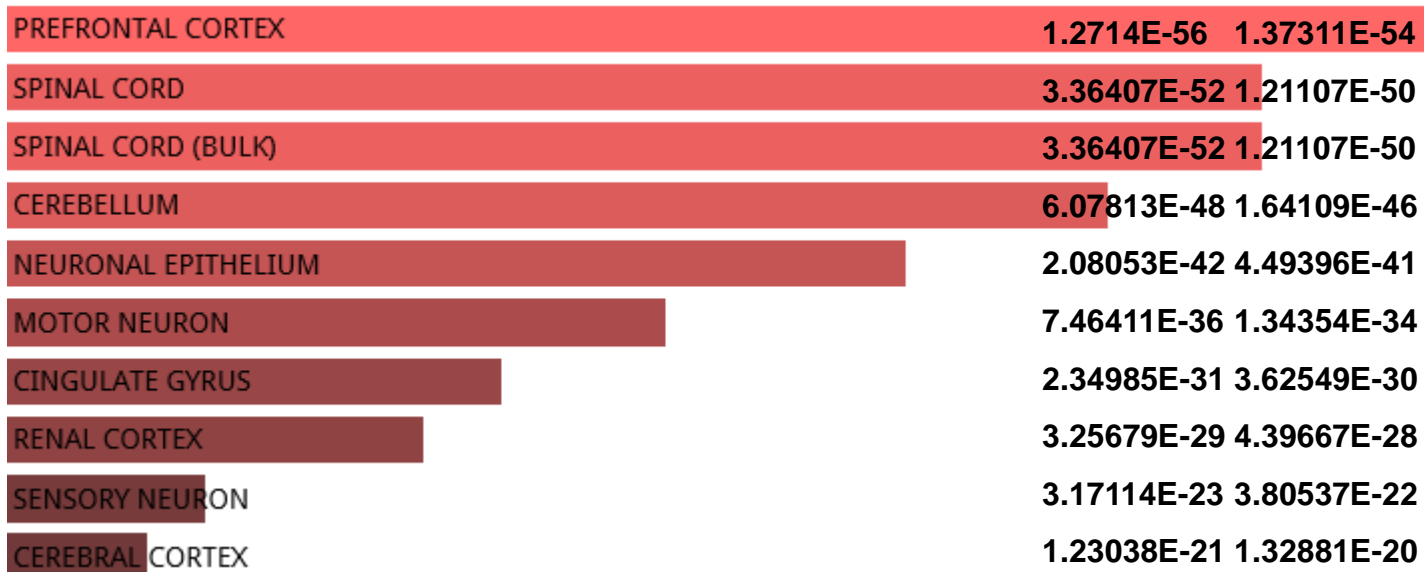
Disease Perturbations from GEO down



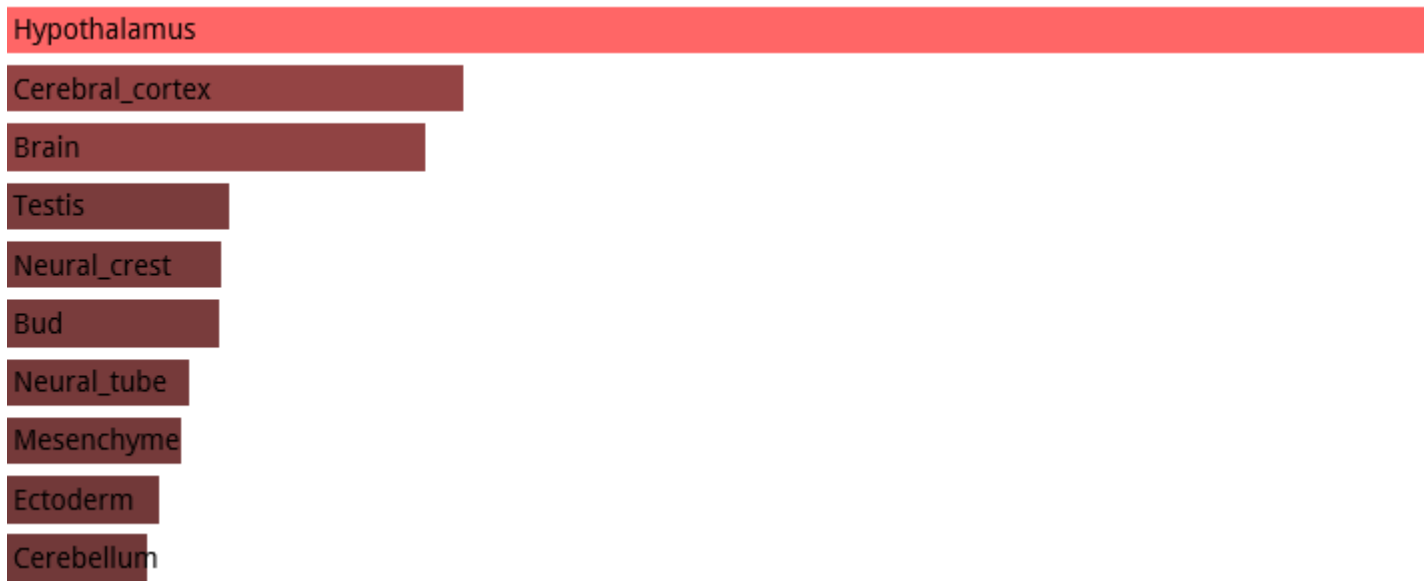
2,745 human accelerated regions

2,745 human accelerated regions

ARCHS4 Tissues 2281 genes

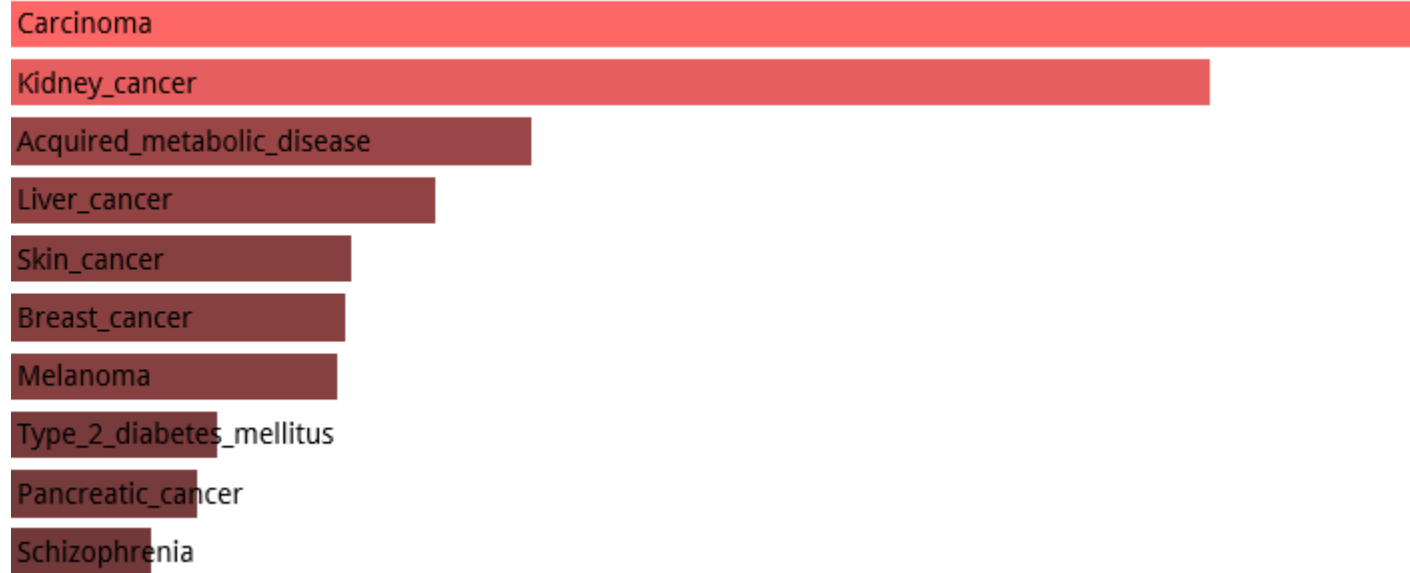


Jensen TISSUES 2281 genes

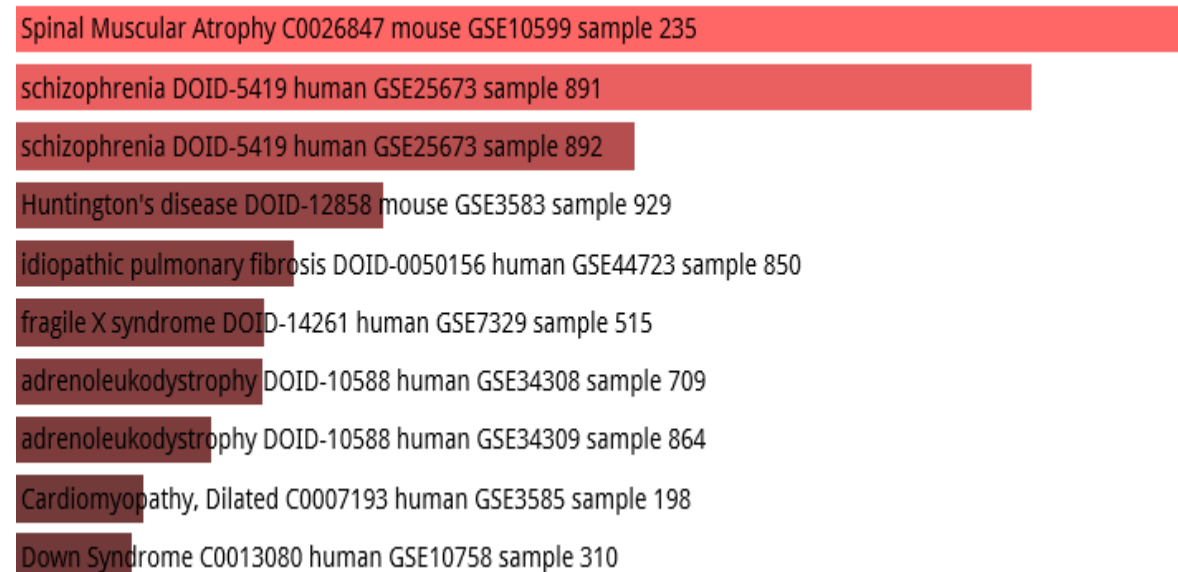


2,745 human accelerated regions

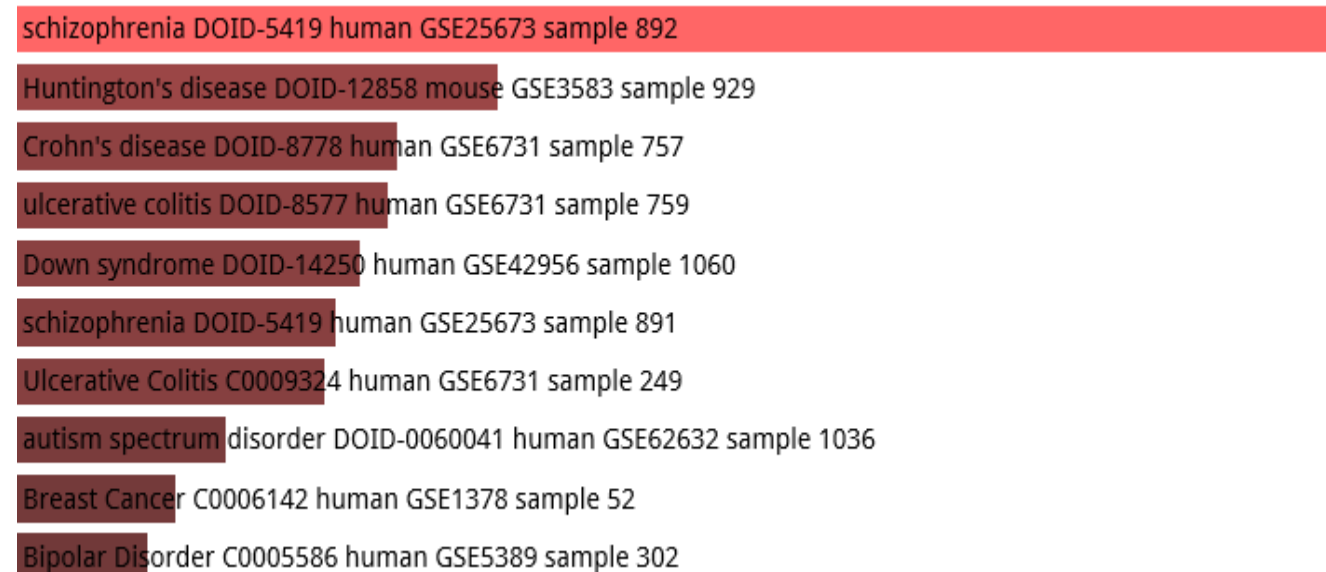
Jensen DISEASES 2281 genes



Disease Perturbations from GEO up



Disease Perturbations from GEO down



**1,932 fixed human-specific regulatory regions
(hESC DHS)**

1,932 fixed human-specific regulatory regions (hESC DHS)

ARCHS4 Tissues 1458 genes

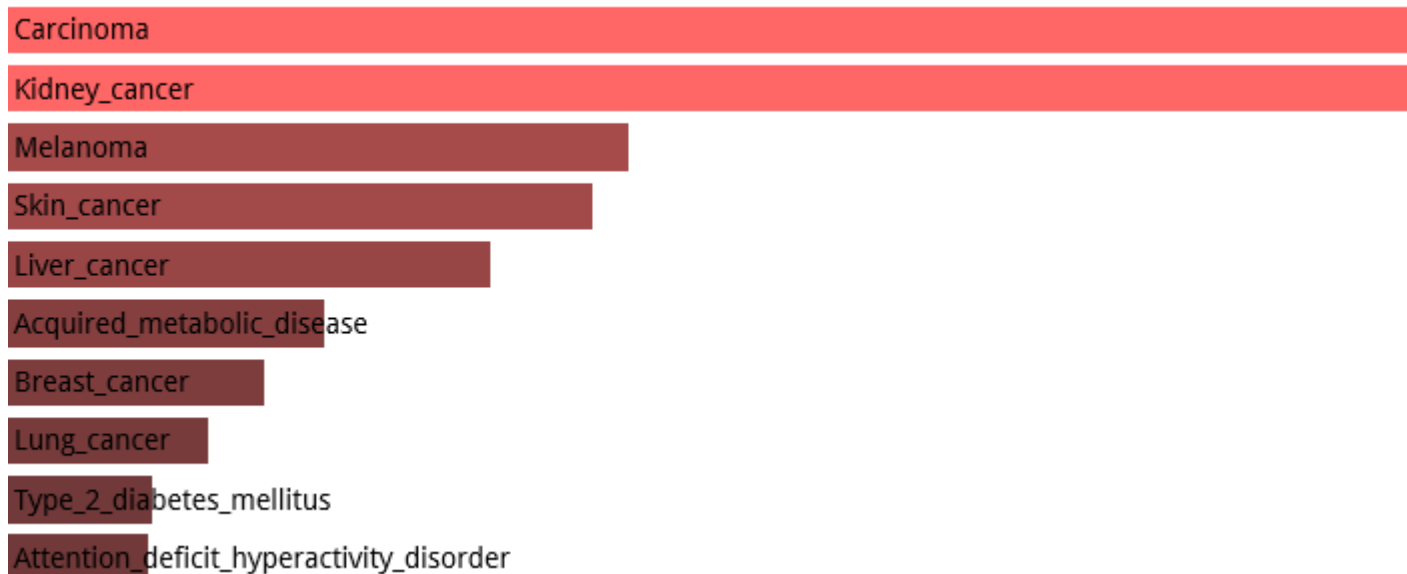
PREFRONTAL CORTEX	5.87167E-35	6.34141E-33
CINGULATE GYRUS	2.10798E-29	1.13831E-27
CEREBELLUM	1.07388E-28	3.86597E-27
SPINAL CORD	2.40716E-28	5.19946E-27
SPINAL CORD (BULK)	2.40716E-28	5.19946E-27
MOTOR NEURON	5.19032E-23	9.34258E-22
DORSAL STRIATUM	3.04713E-20	4.70128E-19
SENSORY NEURON	1.19152E-19	1.60855E-18
CEREBRAL CORTEX	4.5693E-19	5.48316E-18
DENTATE GRANULE CELL	6.33607E-18	6.84295E-17

Jensen TISSUES 1458 genes

Hypothalamus		
Cerebral_cortex		
Brain		
Ganglion		
Neural_crest		
Mesenchyme		
Adult		
Ear		
Mesenchymal_stem_cell		
3T3-L1_cell		

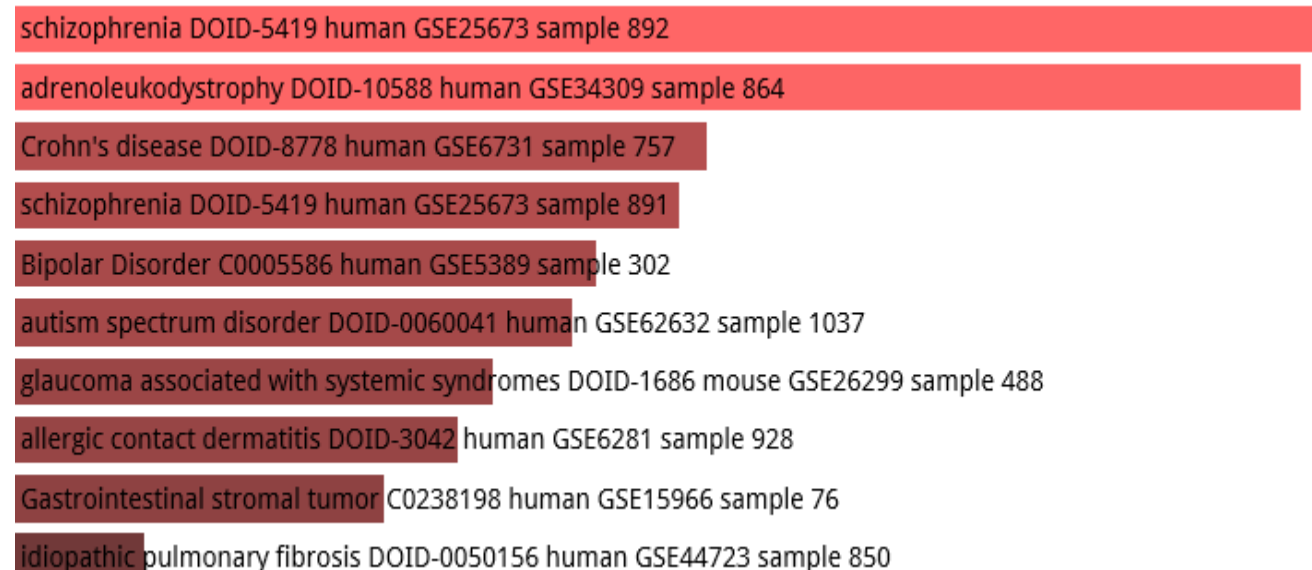
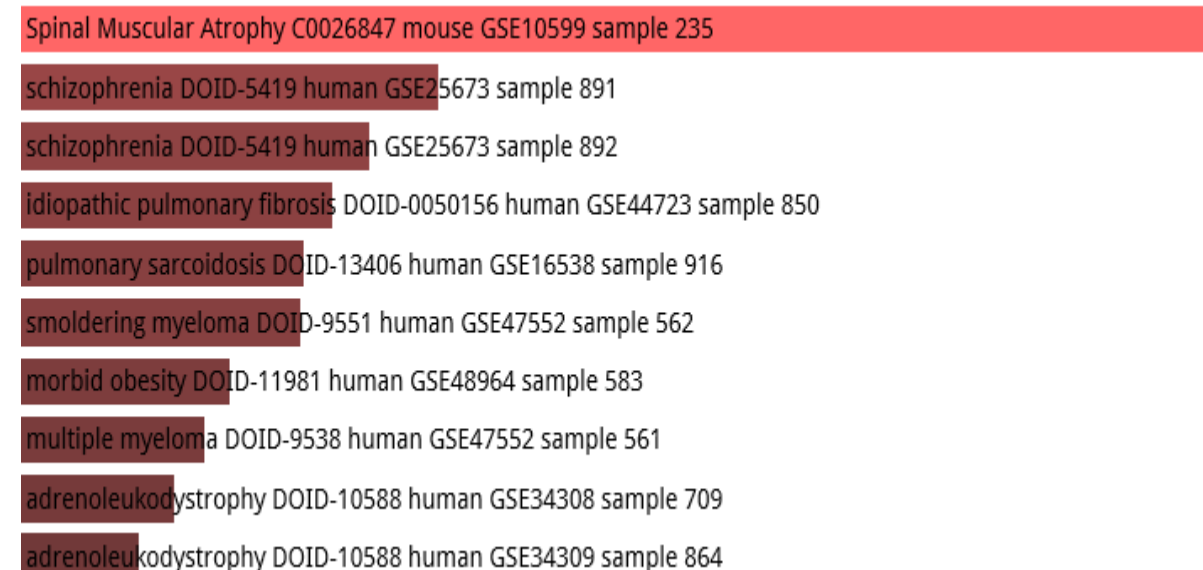
1,932 fixed human-specific regulatory regions (hESC DHS)

Jensen DISEASES 1458 genes



Disease Perturbations from GEO up

Disease Perturbations from GEO down



**1,000 human-biased cranial neural
crest cells (CNCCs) enhancers**

1,000 human-biased cranial neural crest cells (CNCCs) enhancers

ARCHS4 Tissues 1439 genes

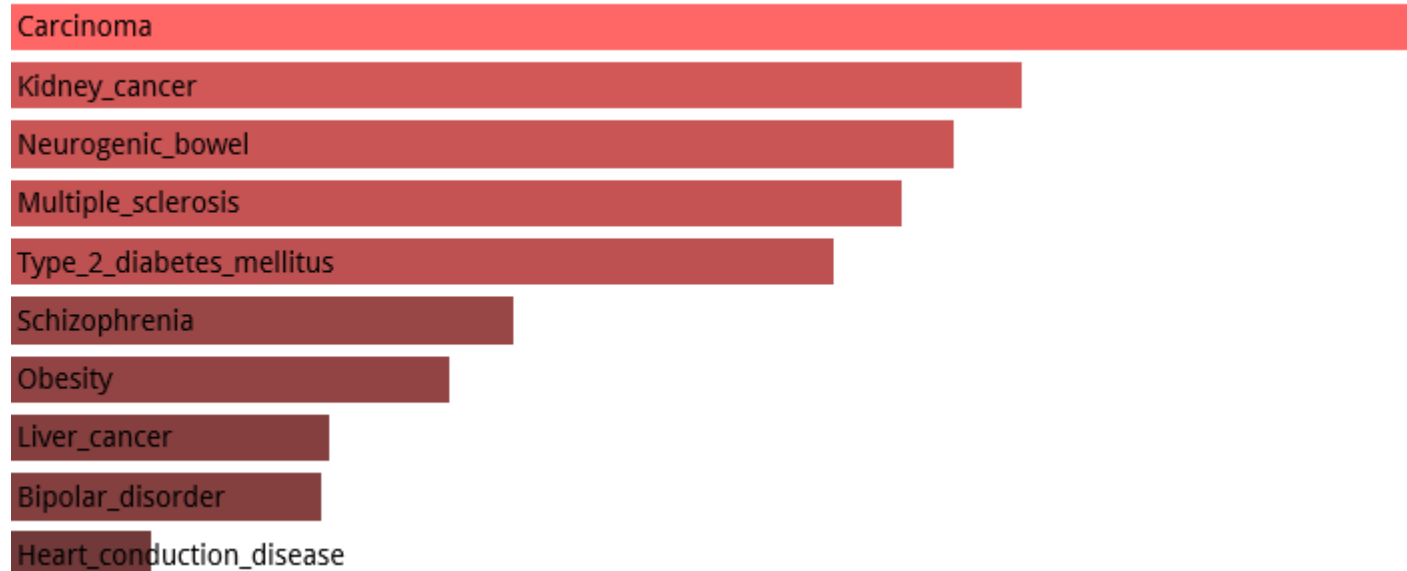
PREFRONTAL CORTEX	5.00308E-22	5.40332E-20
SPINAL CORD	7.24755E-18	2.60912E-16
SPINAL CORD (BULK)	7.24755E-18	2.60912E-16
CEREBELLUM	1.1175E-15	3.01725E-14
CINGULATE GYRUS	2.20974E-14	4.77304E-13
CEREBRAL CORTEX	3.95272E-14	7.11489E-13
MOTOR NEURON	1.24537E-13	1.92143E-12
RENAL CORTEX	6.6998E-13	9.04473E-12
DORSAL STRIATUM	3.43946E-12	4.12736E-11
NEURONAL EPITHELIUM	3.79083E-09	4.0941E-08

Jensen TISSUES 1439 genes

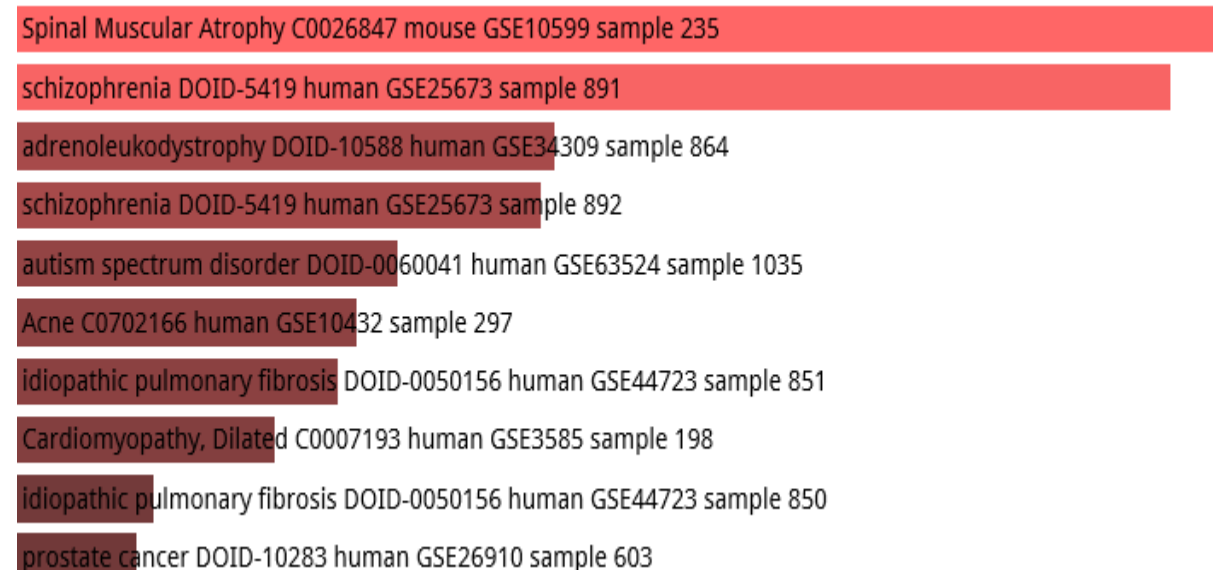
Hypothalamus		
Brain		
Cerebral_cortex		
Uterus		
Esophagus		
Adrenal_gland		
Frontal_lobe		
Urinary_bladder		
Heart		
Cerebellum		

1,000 human-biased cranial neural crest cells (CNCCs) enhancers

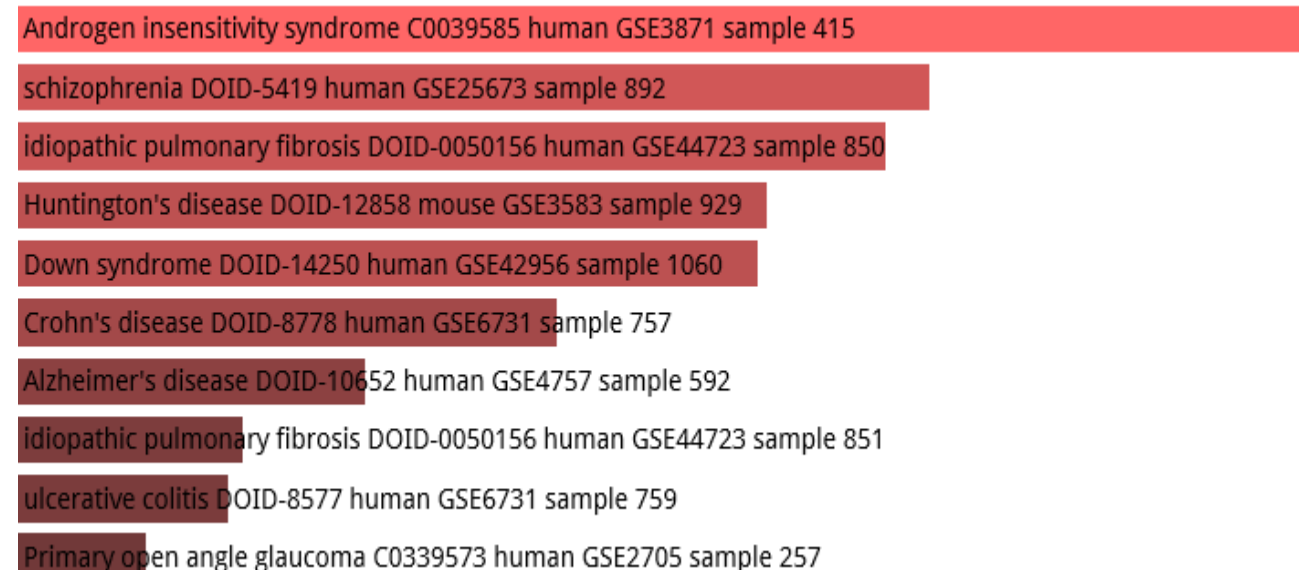
Jensen DISEASES 1439 genes



Disease Perturbations from GEO up



Disease Perturbations from GEO down



**1,000 chimp-biased cranial neural
crest cells (CNCCs) enhancers**

1,000 chimp-biased cranial neural crest cells (CNCCs) enhancers

ARCHS4 Tissues 1445 genes

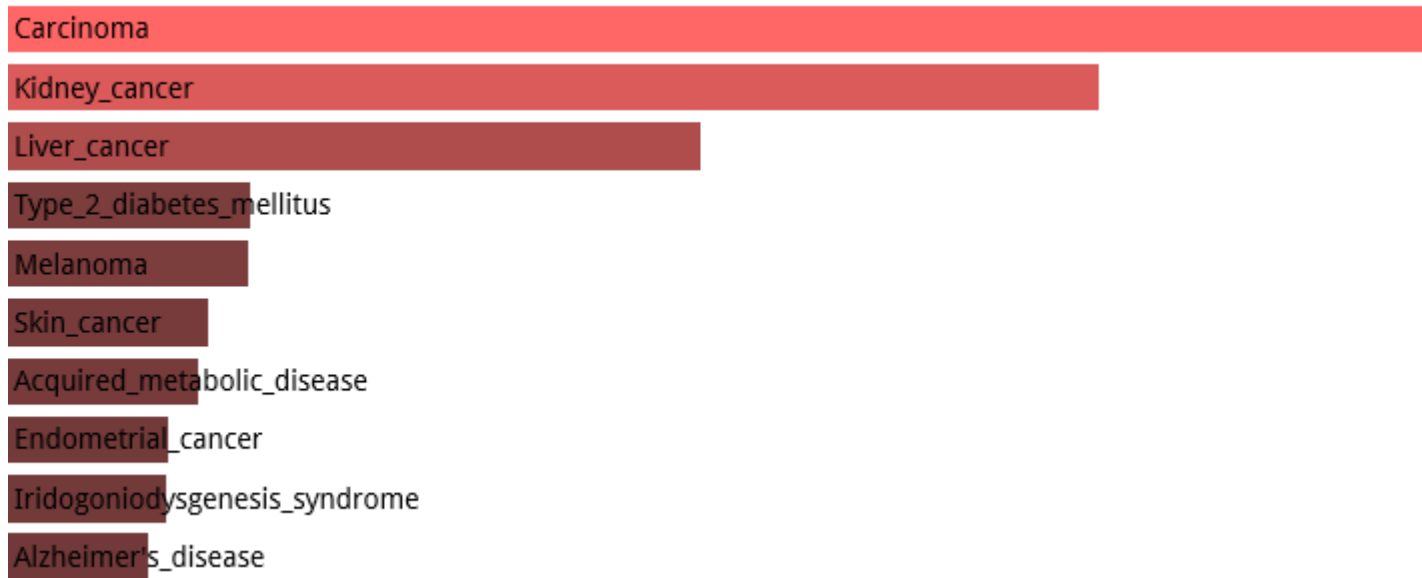
SPINAL CORD	1.84547E-12	9.96554E-11
SPINAL CORD (BULK)	1.84547E-12	9.96554E-11
PREFRONTAL CORTEX	5.4055E-12	1.94598E-10
RENAL CORTEX	7.24092E-11	1.95505E-09
SMALL INTESTINE (BULK TISSUE)	2.19188E-09	4.73446E-08
CEREBELLUM	3.4921E-09	5.38781E-08
CINGULATE GYRUS	3.4921E-09	5.38781E-08
NEURONAL EPITHELIUM	2.1315E-08	2.87753E-07
ADIPOSE (BULK TISSUE)	1.80906E-07	1.95378E-06
LUNG (BULK TISSUE)	1.80906E-07	1.95378E-06

Jensen TISSUES 1445 genes

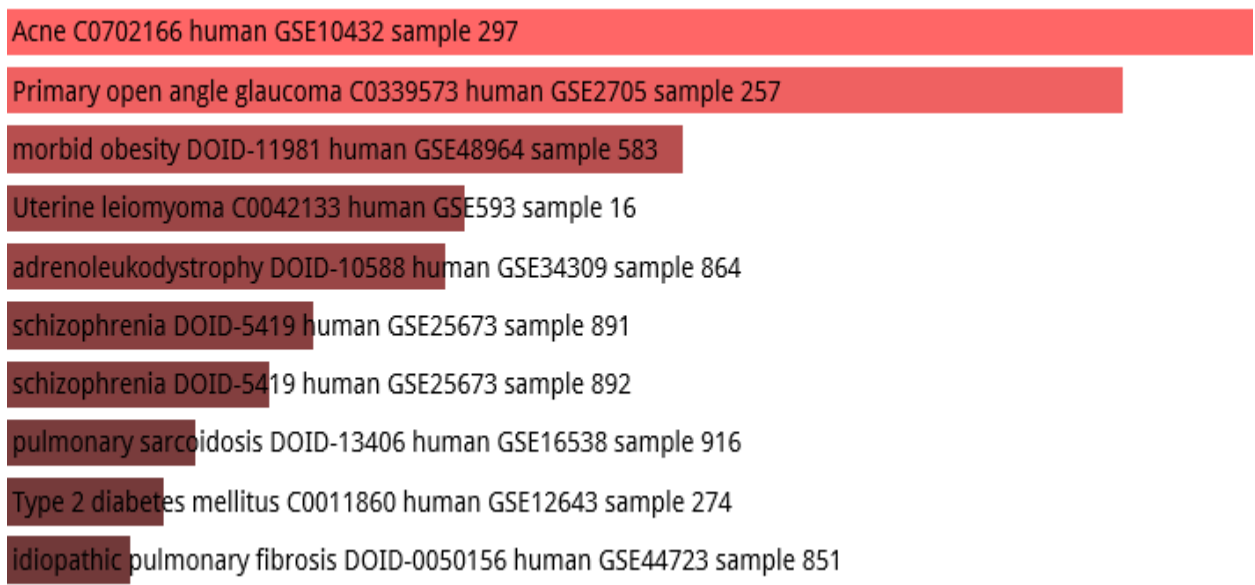
Hypothalamus
Brain
Cerebral_cortex
Uterus
Heart
Prostate_gland
Ovary
Placenta
Esophagus
Spleen

1,000 chimp-biased cranial neural crest cells (CNCCs) enhancers

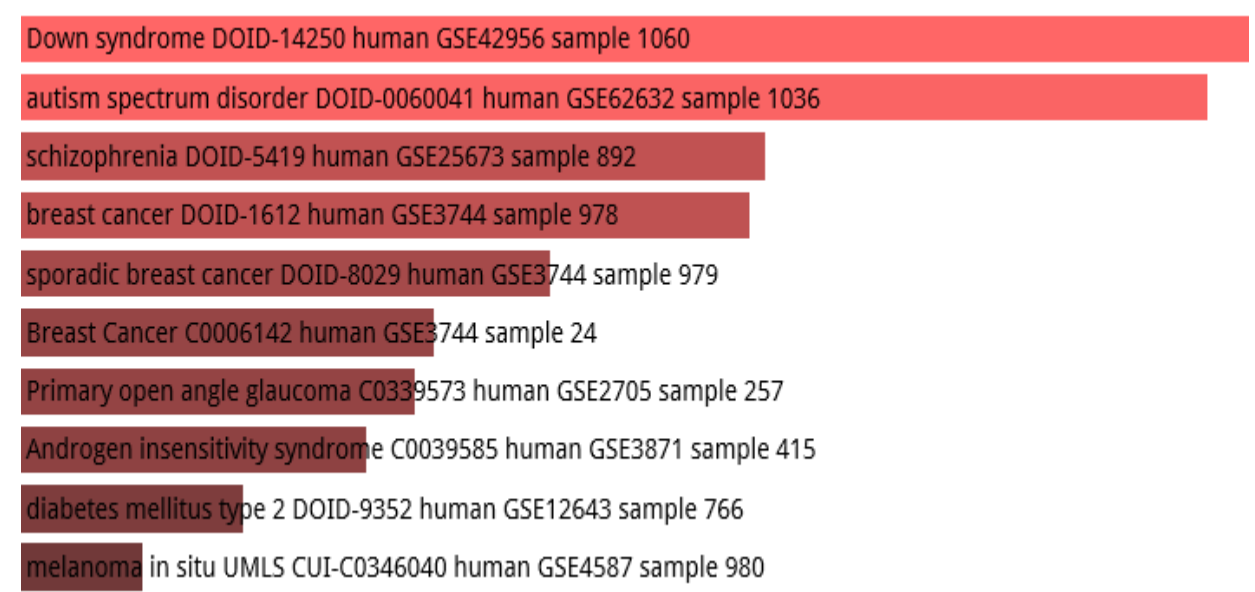
Jensen DISEASES 1445 genes



Disease Perturbations from GEO up



Disease Perturbations from GEO down



**1,619 human-specific functional
hESC enhancers**

1,619 human-specific functional hESC enhancers

ARCHS4 Tissues 1214 genes

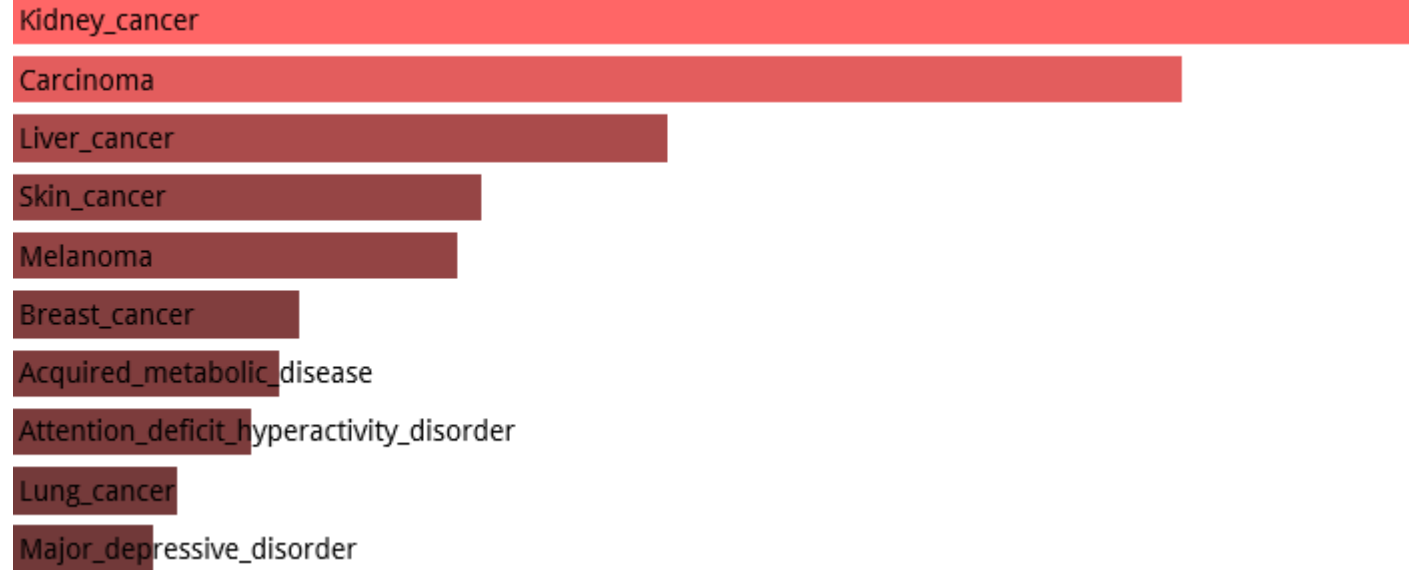
PREFRONTAL CORTEX	5.14081E-14	5.55208E-12
CEREBELLUM	5.87258E-09	3.17119E-07
NEURONAL EPITHELIUM	9.56422E-09	3.44312E-07
FETAL BRAIN CORTEX	1.56576E-07	4.22754E-06
CINGULATE GYRUS	8.90068E-07	1.92255E-05
CEREBRAL CORTEX	4.56907E-06	8.22432E-05
MOTOR NEURON	9.96038E-06	0.0001536
SPINAL CORD	2.11583E-05	0.0002539
SPINAL CORD (BULK)	2.11583E-05	0.0002539
BRAIN (BULK)	8.82976E-05	0.0009536

Jensen TISSUES 1214 genes

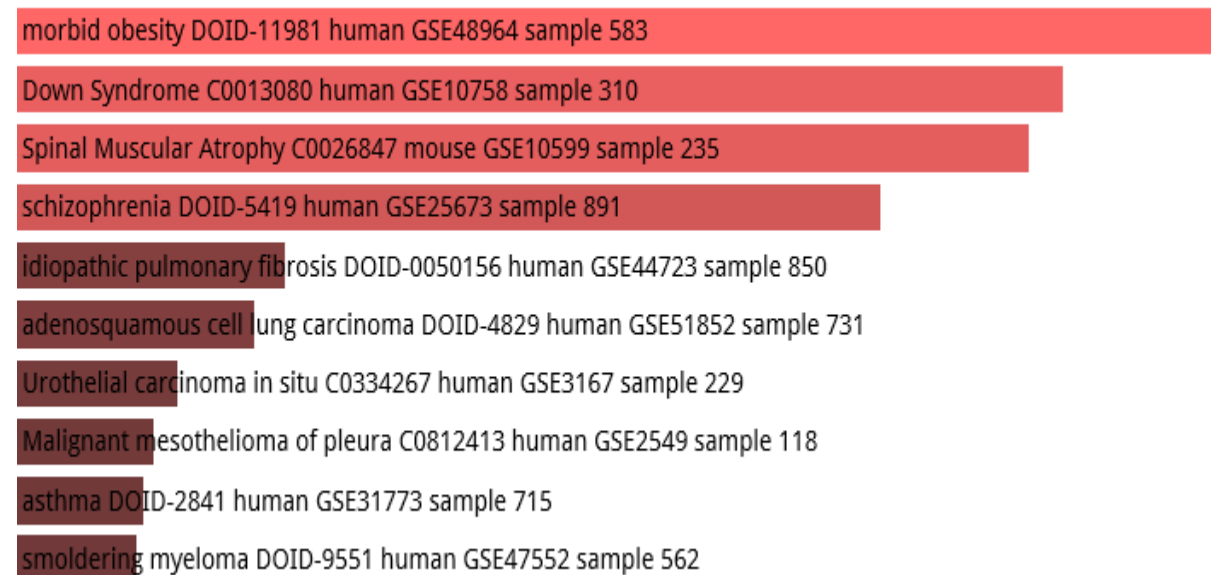
Hypothalamus		
Brain		
Heart		
Occipital_lobe		
Frontal_lobe		
Saliva		
Parietal_lobe		
Cerebral_cortex		
JEG-3_cell		
Temporal_lobe		

1,619 human-specific functional hESC enhancers

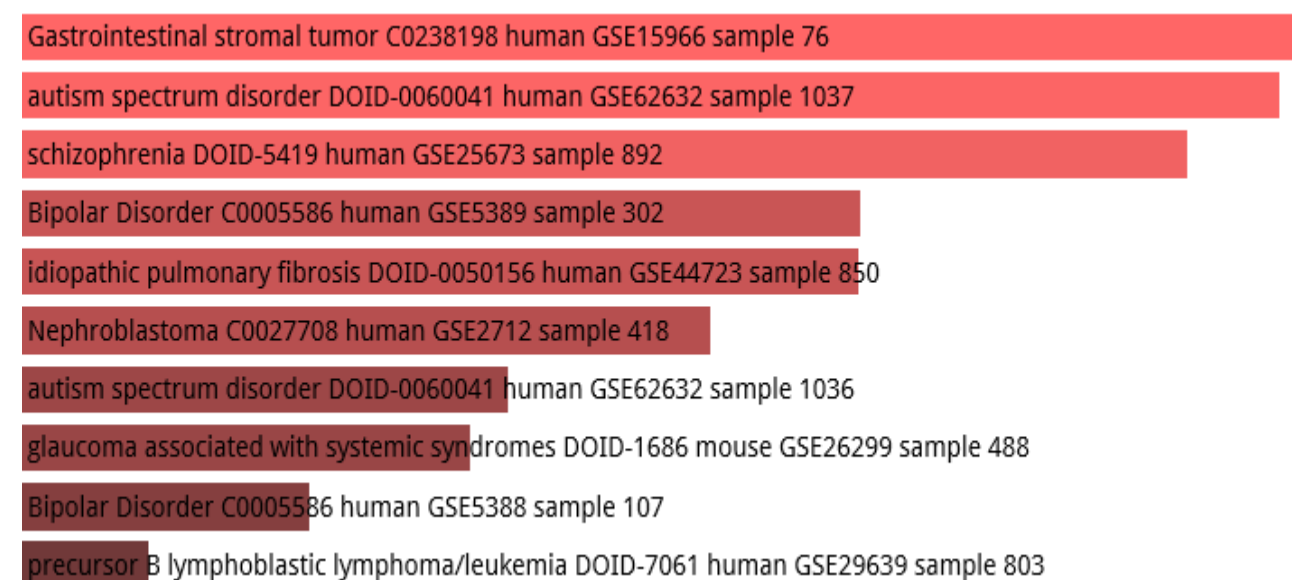
Jensen DISEASES 1214 genes



Disease Perturbations from GEO up 1214 genes

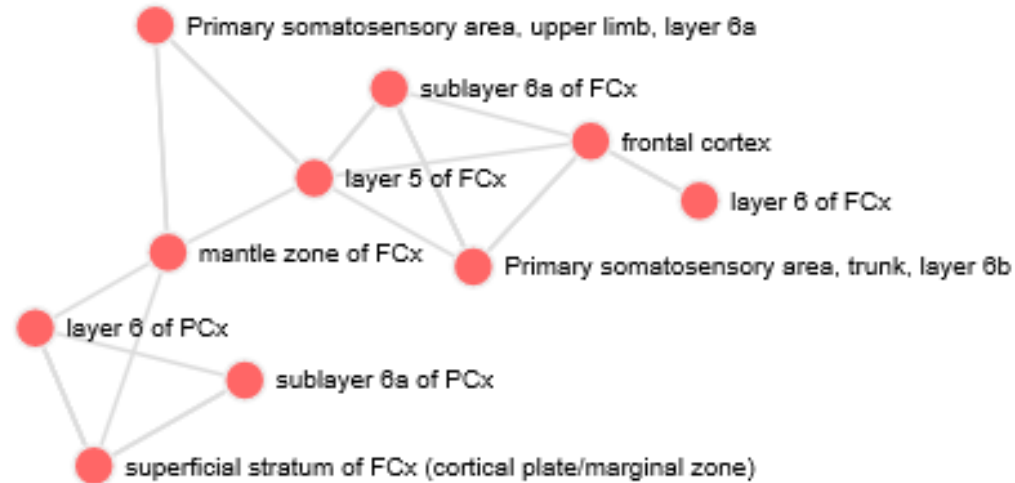
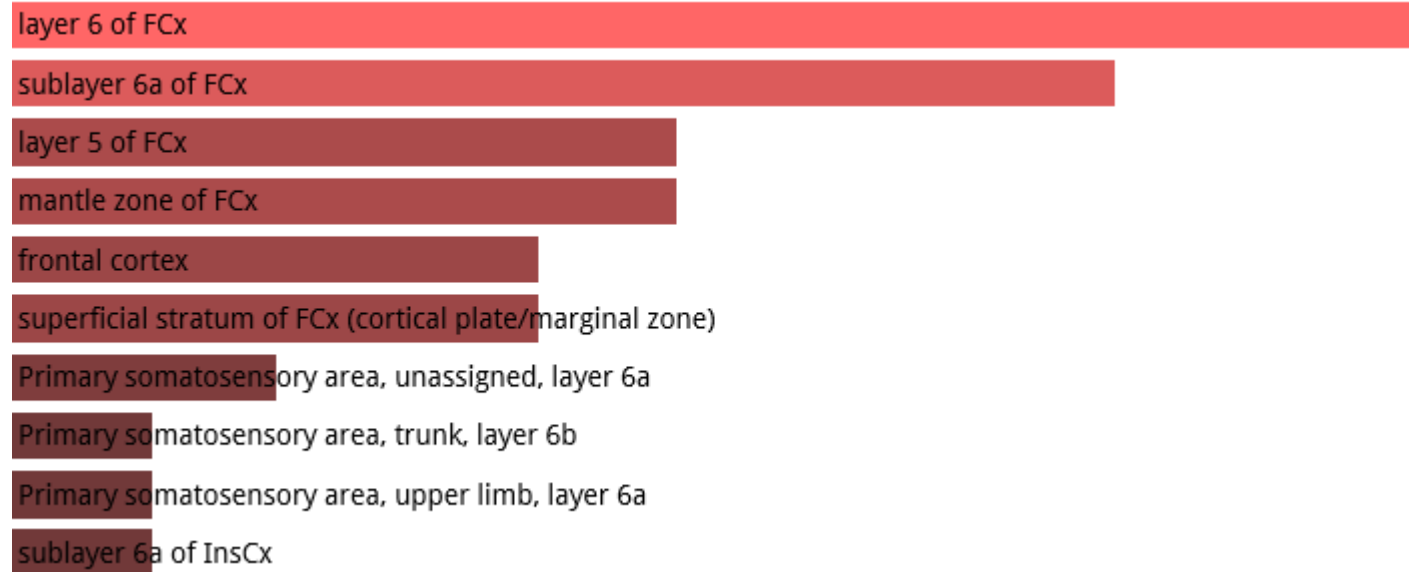


Disease Perturbations from GEO down 1214 genes



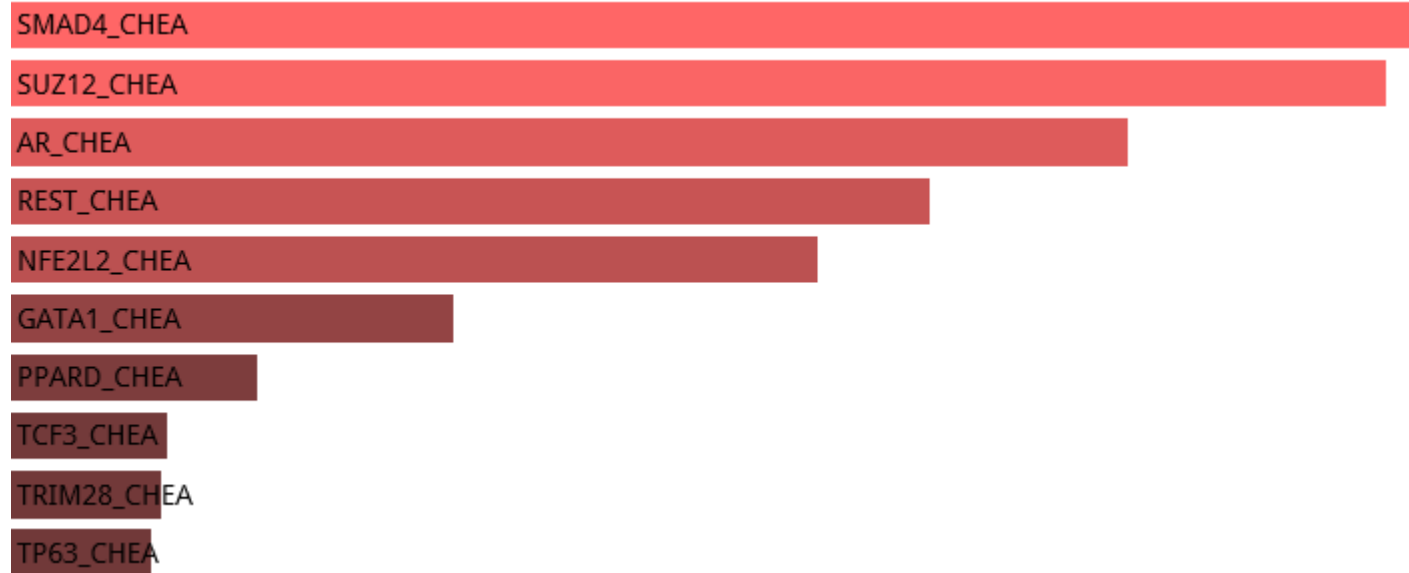
1,619 human-specific functional hESC enhancers

Allen Brain Atlas up 1214 genes

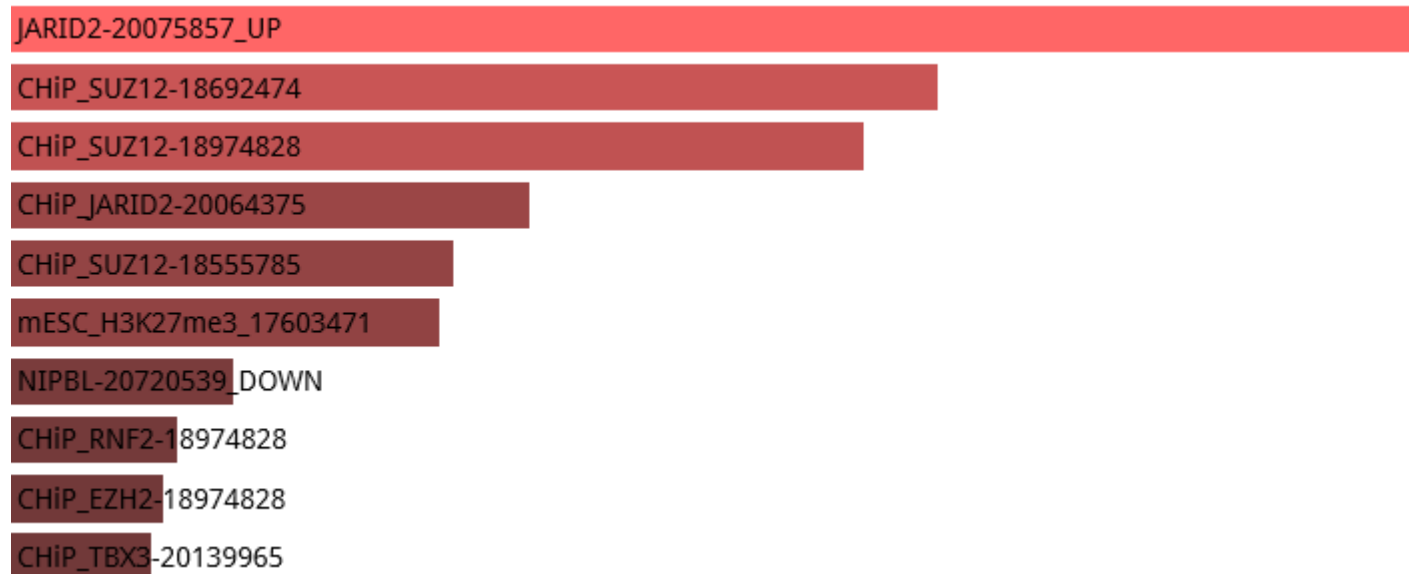


1,619 human-specific functional hESC enhancers

ENCODE and ChEA Consensus TFs from ChIP-X 1214 genes



ESCAPE 1214 genes



1,619 human-specific functional hESC enhancers

GTEX Tissue Sample Gene Expression Profiles up 1214 genes

GTEX-QDT8-0011-R10A-SM-32PKG_brain_female_30-39_years

GTEX-QMR6-0011-R10A-SM-32PKO_brain_male_50-59_years

GTEX-QDT8-0011-R2A-SM-32PKQ_brain_female_30-39_years

GTEX-VJYA-1726-SM-3NMDQ_nerve_male_60-69_years

GTEX-POMQ-2026-SM-2S1OD_nerve_female_20-29_years

GTEX-QMR6-0011-R8A-SM-32PKJ_brain_male_50-59_years

GTEX-PVOW-0011-R3A-SM-32PKX_brain_male_40-49_years

GTEX-X585-0008-SM-46MU4_skin_male_50-59_years

GTEX-S7SE-0011-R10A-SM-2XCDF_brain_male_50-59_years

GTEX-XBEC-0008-SM-4AT3X_skin_male_50-59_years

GTEX Tissue Sample Gene Expression Profiles down 1214 genes

GTEX-SN8G-0001-SM-3NM8L_blood_female_50-59_years

GTEX-XXEK-0004-SM-4BRWO_blood_male_50-59_years

GTEX-TMZS-0001-SM-3P61Q_blood_male_60-69_years

GTEX-XUYS-0002-SM-47JXL_blood_male_50-59_years

GTEX-WFON-0001-SM-3P61W_blood_male_40-49_years

GTEX-XYKS-0002-SM-4BRWN_blood_female_60-69_years

GTEX-XUJ4-0004-SM-4BOQE_blood_female_60-69_years

GTEX-WHPG-0004-SM-3NMDO_blood_male_50-59_years

GTEX-T6MO-0003-SM-3NMAG_blood_female_40-49_years

GTEX-WFJO-0002-SM-3P61X_blood_male_30-39_years

524 human-specific DHS

524 human-specific DHS

ARCHS4 Tissues 747 genes

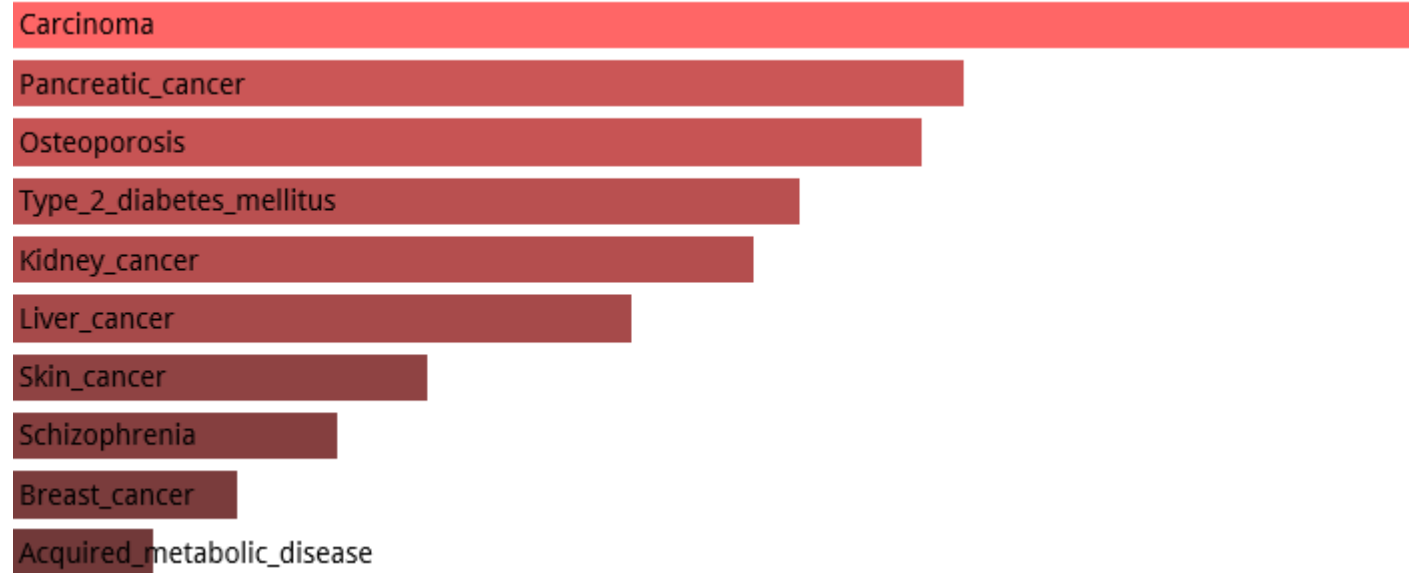
PREFRONTAL CORTEX	1.84669E-23	1.99442E-21
CEREBELLUM	2.96622E-17	1.60176E-15
MIDBRAIN	6.7695E-17	2.43702E-15
SPINAL CORD	3.4404E-16	7.43127E-15
SPINAL CORD (BULK)	3.4404E-16	7.43127E-15
CEREBRAL CORTEX	2.94376E-12	3.97408E-11
CINGULATE GYRUS	2.94376E-12	3.97408E-11
NEURONAL EPITHELIUM	2.94376E-12	3.97408E-11
MOTOR NEURON	1.18181E-11	1.41817E-10
RENAL CORTEX	1.71559E-10	1.85284E-09

Jensen TISSUES 747 genes

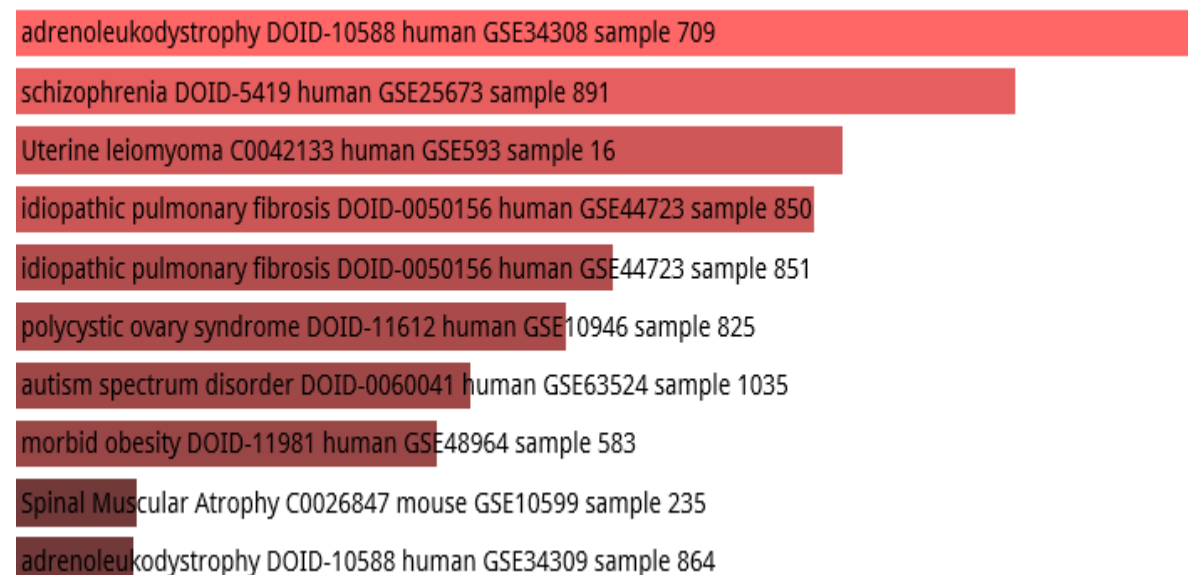
Hypothalamus
Brain
Cerebral_cortex
Neural_crest
Testis
Gall_bladder
Parietal_lobe
Frontal_lobe
Occipital_lobe
Urinary_bladder

524 human-specific DHS

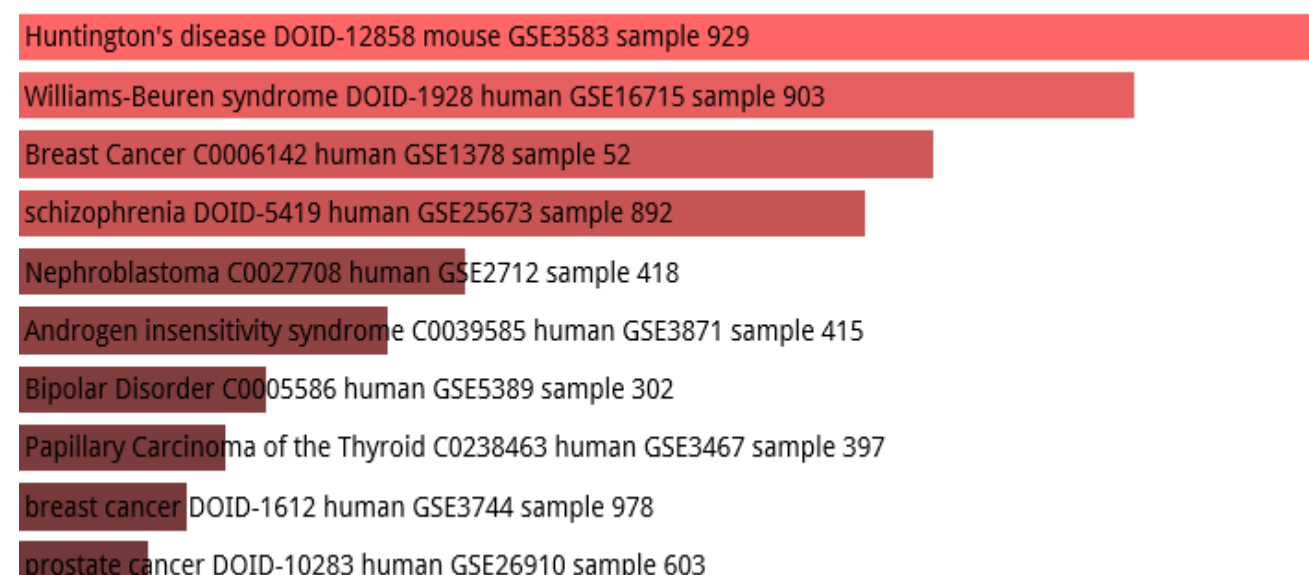
Jensen DISEASES 747 genes



Disease Perturbations from GEO up 747 genes



Disease Perturbations from GEO down 747 genes



1,279 human-specific STR contractions

ARCHS4 Tissues 973 genes

SUPERIOR FRONTAL GYRUS	1.4292E-06	0.000154
SPINAL CORD	2.86362E-05	0.001031
SPINAL CORD (BULK)	2.86362E-05	0.001031
BRAIN (BULK)	9.20916E-05	0.002486
SUBCUTANEOUS ADIPOSE TISSUE	0.000192908	0.004167
RENAL CORTEX	0.000275908	0.004257
ADIPOSE (BULK TISSUE)	0.000275908	0.004257
BREAST (BULK TISSUE)	0.000391501	0.005285
MOTOR NEURON	0.010836889	0.130043
LUNG (BULK TISSUE)	0.013964132	0.150813

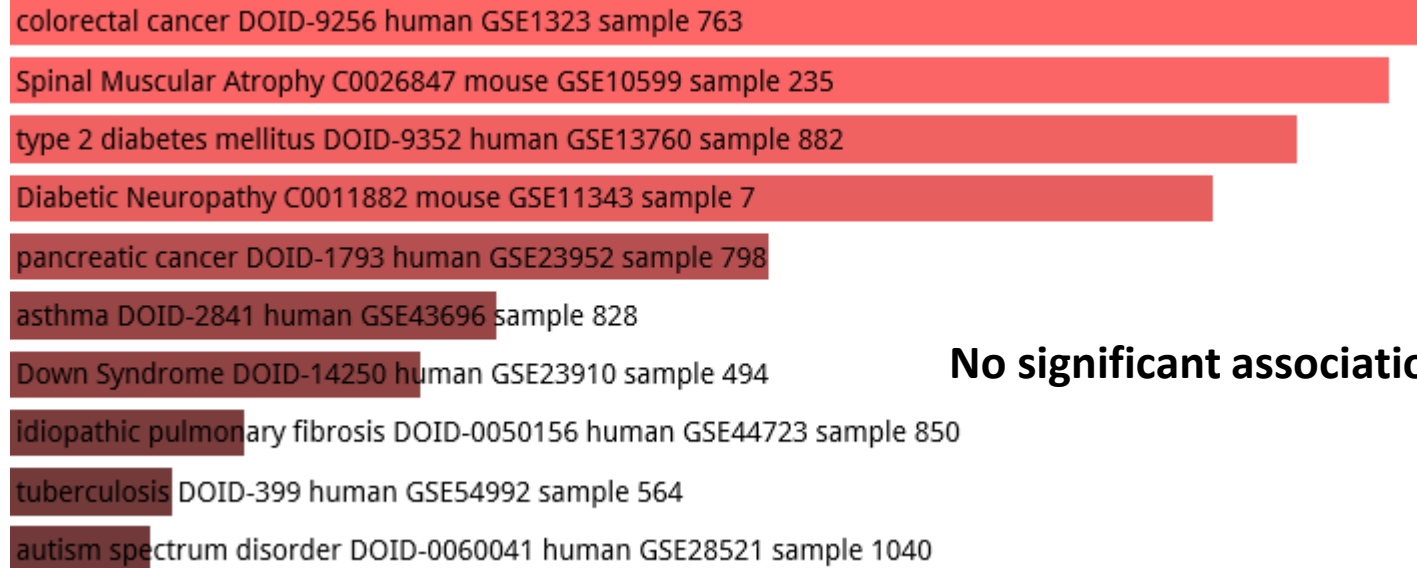
Jensen TISSUES 973 genes

Brain
Eye
Retinoblastoma_cell
ac2
Occipital_lobe
Germinal_epithelium
Barrett's_esophagus
Somite
Helam
Sputum

Jensen DISEASES 973 genes

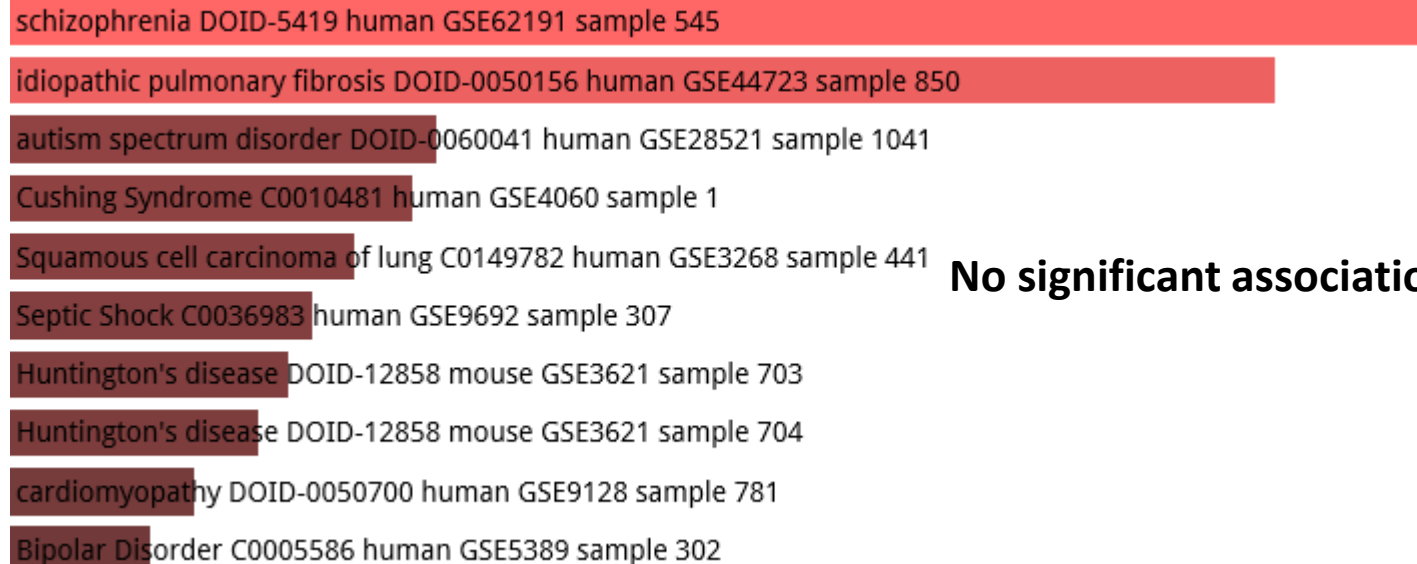
Miller-Dieker_lissencephaly_syndrome
Wolf-Hirschhorn_syndrome
Chondrodysplasia_punctata
Chromosome_1p36_deletion_syndrome
Hemolytic_anemia
Mixed_connective_tissue_disease
Large_intestine_cancer
relapsing-remitting_multiple_sclerosis
Pancreatic_cancer
Beckwith-Wiedemann_syndrome

Disease Perturbations from GEO up 973 genes



No significant associations

Disease Perturbations from GEO down 973 genes



No significant associations

**2,118 fixed human-specific regulatory regions
(non-hESC DHS)**

ARCHS4 Tissues 552 genes

PREFRONTAL CORTEX	4.6456E-05	0.00167
SPINAL CORD	4.6456E-05	0.00167
SPINAL CORD (BULK)	2.79873E-05	0.00167
MOTOR NEURON	0.000311	0.00840
CINGULATE GYRUS	0.003676	0.06616
BRAIN (BULK)	0.003676	0.06616
NEURONAL EPITHELIUM	0.005300	0.08178
CEREBRAL CORTEX	0.007545	0.09054
SUPERIOR FRONTAL GYRUS	0.007545	0.09054
DORSAL STRIATUM	0.027142	0.29314

**410 H3K4me3 sites with human-specific
enrichment in prefrontal cortex neurons**

ARCHS4 Tissues 578 genes

SUPERIOR FRONTAL GYRUS	0.000933	0.10074
NEURONAL EPITHELIUM	0.008682	0.31255
FETAL BRAIN	0.006177	0.31255
CEREBRAL CORTEX	0.016511	0.40212
MOTOR NEURON	0.022340	0.40212
BRAIN (BULK)	0.022340	0.40212
CEREBELLUM	0.039369	0.47242
SPINAL CORD	0.039369	0.47242
SPINAL CORD (BULK)	0.039369	0.47242
FIBROBLAST		

No significant associations

42,847 human genes not linked by the GREAT algorithm with HSRS were randomly split into 21 control gene sets of various sizes ranging from 2,847 to 6,847 genes and subjected to the GSEA

**GSEA OF TWENTY-ONE CONTROL GENE SETS NOT
ASSOCIATED WITH HUMAN-SPECIFIC GENOMIC
REGULATORY LOCI**

Random Set 1

ASTROCYTE

KIDNEY (BULK TISSUE)

GASTRIC TISSUE (BULK)

SPINAL CORD (BULK)

SMALL INTESTINE (BULK TISSUE)

OVARY (BULK TISSUE)

SPINAL CORD

PLACENTA (BULK)

SKIN (BULK TISSUE)

HUMAN EMBRYO

ARCHS4 Tissues random 6000 genes

NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

NO DATA AVAILABLE

Random Set 1

Jensen DISEASES random 6000 genes

Reticular_dysgenesis
Amyotrophic_lateral_sclerosis_type_8
Succinic_semi-aldehyde_dehydrogenase_deficiency
Rhizomelic_chondrodysplasia_punctata
Biotinidase_deficiency
Intrahepatic_cholestasis
Conversion_disorder
Retinal_ischemia
Hypersensitivity_reaction_type_II_disease
Pseudohermaphroditism

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes

pancreatic ductal adenocarcinoma DOID-3498 mouse GSE53659 sample 699
juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 772
amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679
juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 771
nemaline myopathy DOID-3191 mouse GSE3384 sample 971
nemaline myopathy DOID-3191 mouse GSE3384 sample 971
Nemaline Myopathy C0206157 mouse GSE3384 sample 317
lupus erythematosus DOID-8857 human GSE30153 sample 739
Duchenne muscular dystrophy (DMD) C0013264 mouse GSE1008 sample 298
idiopathic pulmonary fibrosis DOID-0050156 human GSE24206 sample 872

NO SIGNIFICANT ENRICHMENT

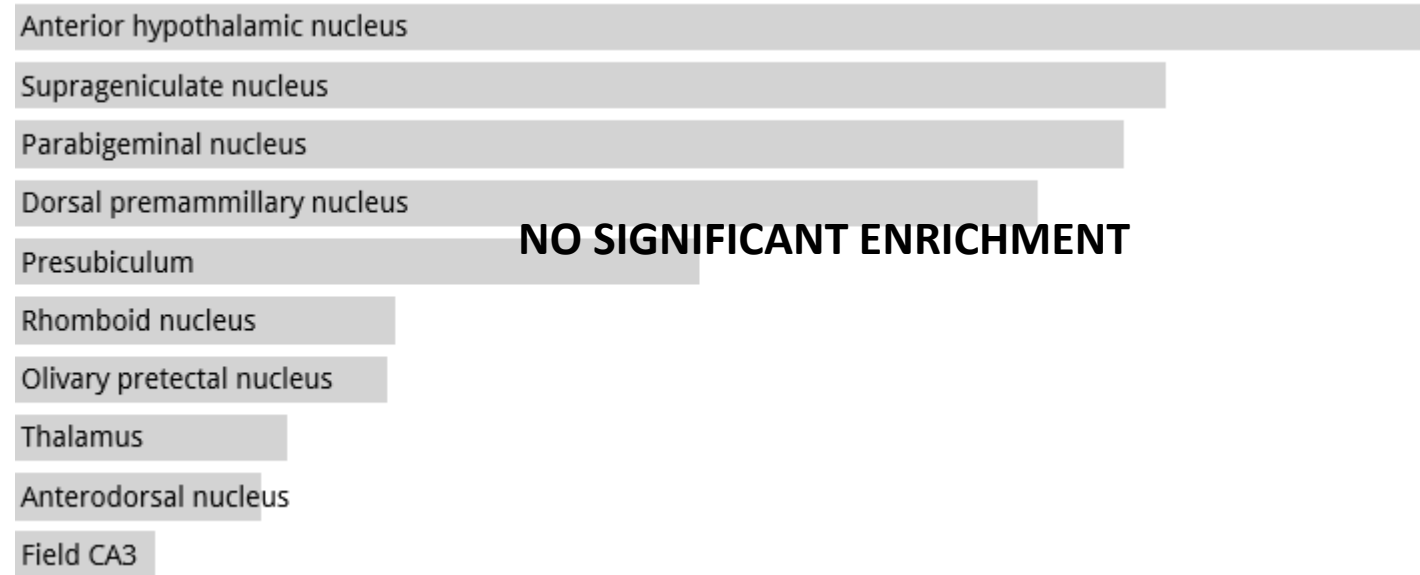
Disease Perturbations from GEO down random 6000 genes

pulmonary tuberculosis DOID-2957 mouse GSE48027 sample 831
polycystic ovary syndrome DOID-11612 human GSE48301 sample 558
Nicotine addiction C0028043 human GSE11208 sample 325
autism spectrum disorder DOID-0060041 human GSE62632 sample 1037
mental retardation DOID-1059 human GSE375 sample 1047
Diabetic Neuropathy C0011882 mouse GSE11343 sample 7
Alcohol poisoning C0392620 rat GSE3311 sample 288
asthma DOID-2841 human GSE43696 sample 830
systemic lupus erythematosus DOID-9074 human GSE55447 sample 1075
Thymic Carcinoma C0205969 mouse GSE2501 sample 344

NO SIGNIFICANT ENRICHMENT

Random Set 1

Allen Brain Atlas up random 6000 genes



Random Set 2

ARCHS4 Tissues random 6000 genes

GASTRIC TISSUE (BULK)

SKIN (BULK TISSUE)

OMENTUM

SMALL INTESTINE (BULK TISSUE)

CINGULATE GYRUS

COLON (BULK TISSUE)

NEURONAL EPITHELIUM

PREFRONTAL CORTEX

PANCREATIC ISLET

MYOBLAST

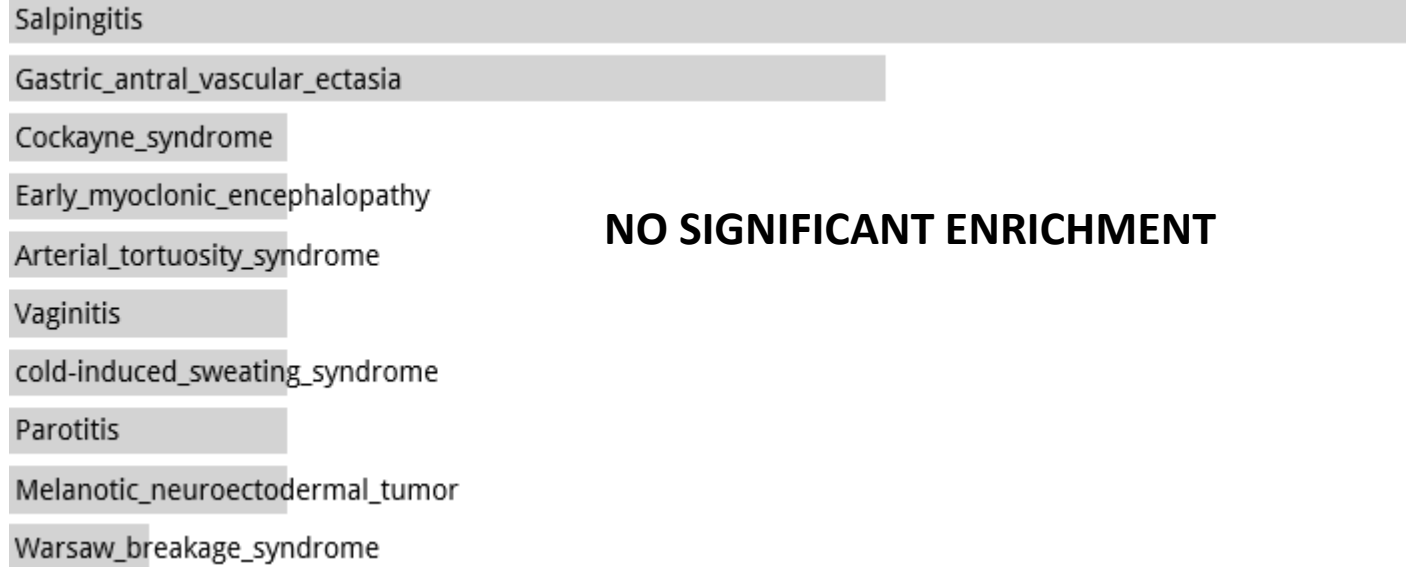
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

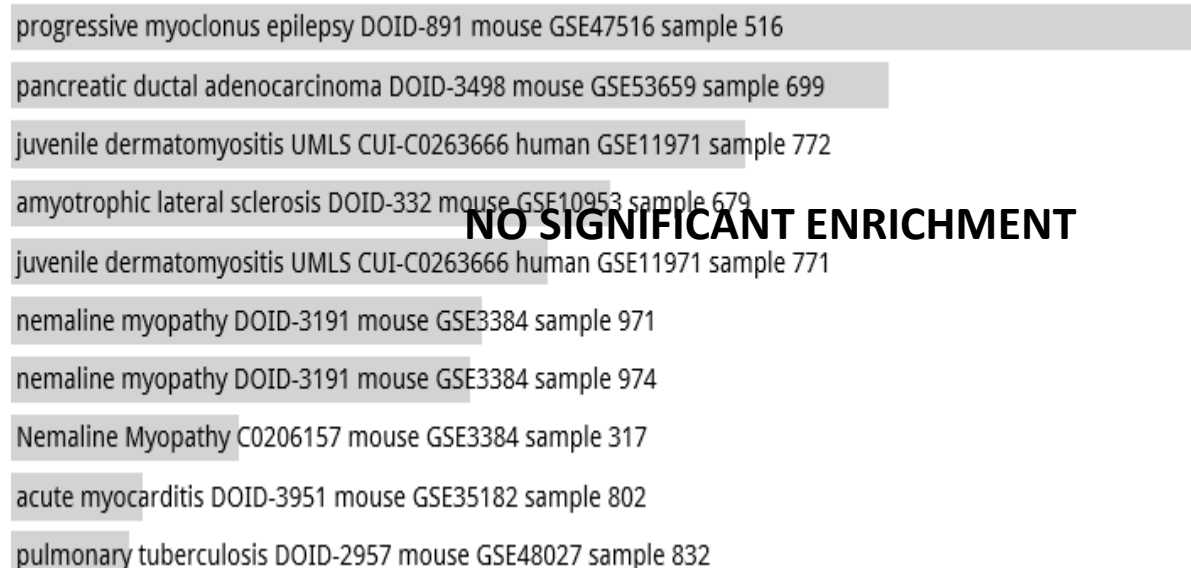
NO DATA AVAILABLE

Random Set 2

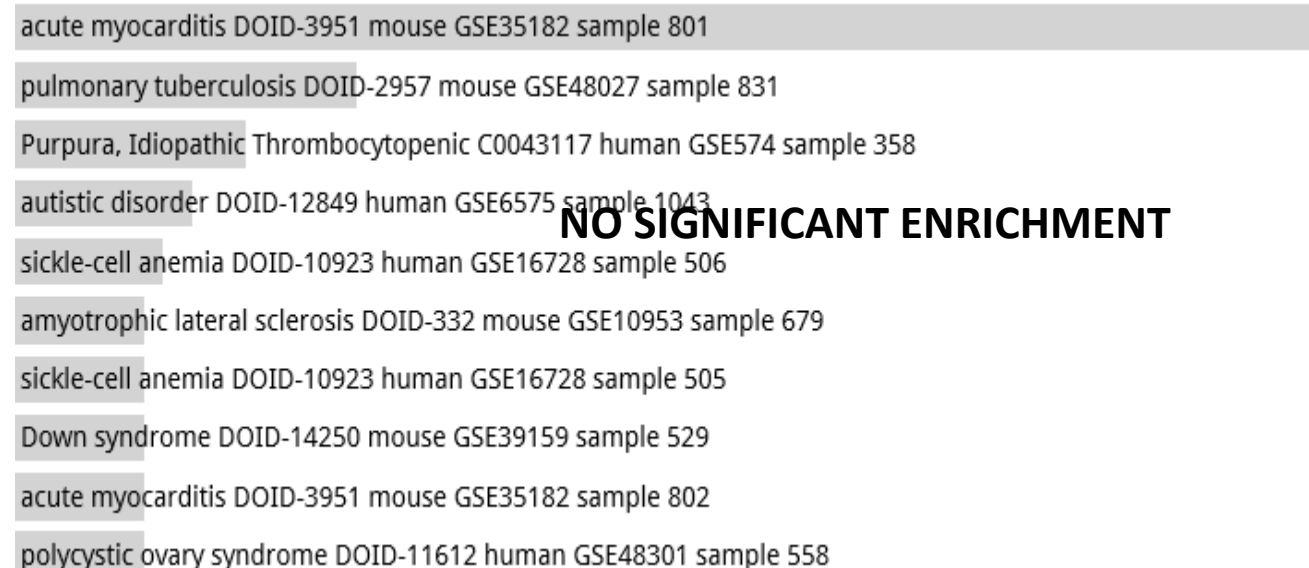
Jensen DISEASES random 6000 genes



Disease Perturbations from GEO up random 6000 genes

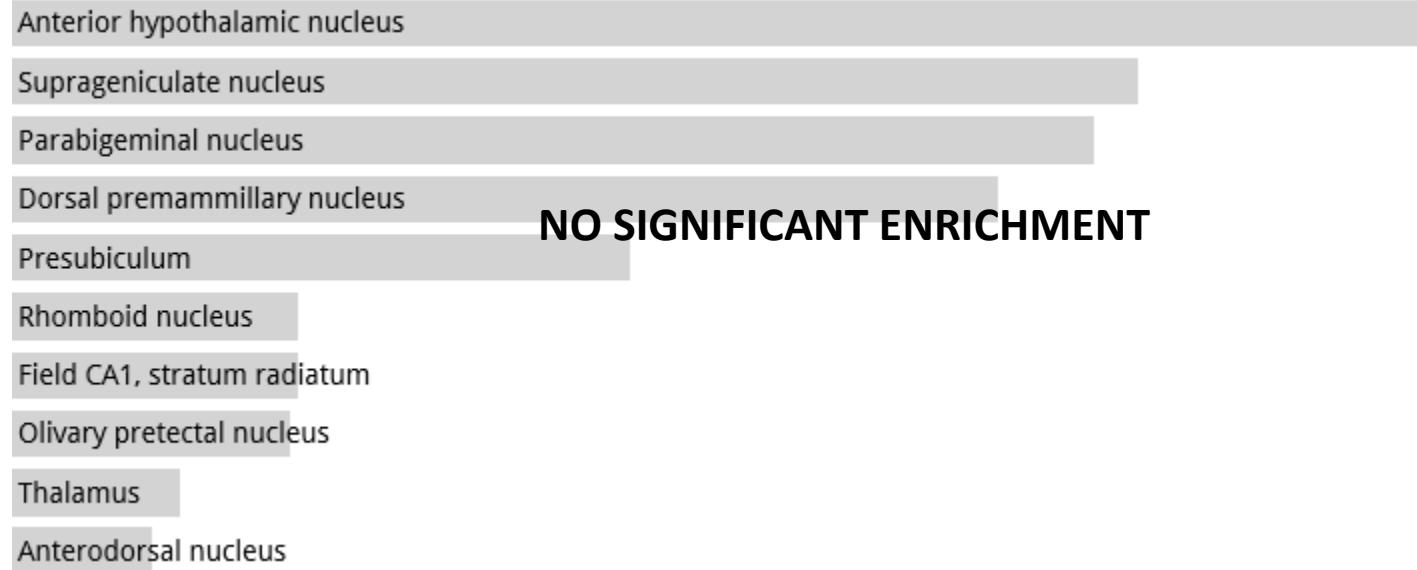


Disease Perturbations from GEO down random 6000 genes



Random Set 2

Allen Brain Atlas up random 6000 genes



Random Set 3

ARCHS4 Tissues random 6000 genes

COLONIC MUCOSA

VENTRICLE

PREFRONTAL CORTEX

BRAIN (BULK)

SUPERIOR FRONTAL GYRUS

NEURONAL EPITHELIUM

ADIPOSE (BULK TISSUE)

HUMAN ZYGOTE

RENAL CORTEX

MYOBLAST

NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

NO DATA AVAILABLE

Random Set 3

Jensen DISEASES random 6000 genes

Hereditary_mucosal_leukokeratosis

Echolalia

Dumping_syndrome

X-linked_endothelial_corneal_dystrophy

IMAGe_syndrome

Clouston_syndrome

Oculodentodigital_dysplasia

Irritant_dermatitis

Brain_glioma

Pachyonychia_congenita

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes

Dental cavity, complex C0399396 human GSE1629 sample 175

Alzheimer's disease DOID-10652 human GSE36980 sample 519

actinic keratosis DOID-8866 human GSE2503 sample 628

Actinic keratosis C0022602 human GSE2503 sample 350

acute myeloid leukemia DOID-9119 human GSE9476 sample 782

ulcerative colitis DOID-8577 human GSE9452 sample 924

type 2 diabetes mellitus DOID-9352 human GSE23343 sample 895

acute myeloid leukemia DOID-9119 human GSE9476 sample 783

polycystic ovary syndrome DOID-11612 human GSE48301 sample 558

Epilepsy C0014544 human GSE7486 sample 417

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 6000 genes

multiple myeloma DOID-9538 human GSE6691 sample 787

acute myocarditis DOID-3951 mouse GSE35182 sample 801

Smoldering multiple myeloma C1531608 human GSE5900 sample 404

autistic disorder DOID-12849 human GSE6575 sample 1043

autistic disorder DOID-12849 human GSE6575 sample 1042

pulmonary tuberculosis DOID-2957 mouse GSE48027 sample 831

mental retardation DOID-1059 human GSE6575 sample 1044

Down syndrome DOID-14250 mouse GSE39159 sample 529

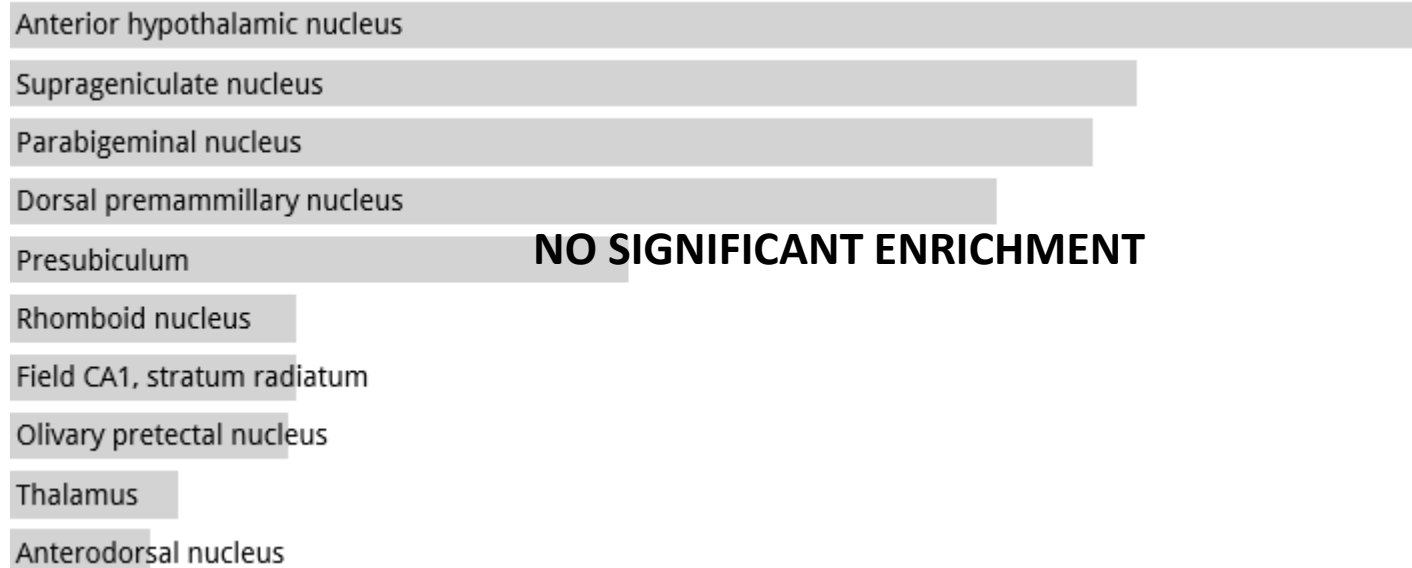
Septic Shock C0036983 human GSE9692 sample 307

West Nile fever DOID-2366 human GSE30719 sample 874

NO SIGNIFICANT ENRICHMENT

Random Set 3

Allen Brain Atlas up random 6000 genes



Random Set 4

GASTRIC TISSUE (BULK)

PREFRONTAL CORTEX

COLON (BULK TISSUE)

LUNG (BULK TISSUE)

OMENTUM

NEURONAL EPITHELIUM

LIVER (BULK TISSUE)

ILEUM (BULK)

FETAL BRAIN CORTEX

SUPERIOR FRONTAL GYRUS

ARCHS4 Tissues random 6000 genes

NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

NO DATA AVAILABLE

Random Set 4

Jensen DISEASES random 6000 genes

Urinary_schistosomiasis

Hermaphroditism

Zellweger_syndrome

Latex_allergy

Scimitar_syndrome

Conversion_disorder

Gastric_lymphoma

Juvenile_polyposis_syndrome

Invasive_lobular_carcinoma

Acute_promyelocytic_leukemia

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes

pancreatic ductal adenocarcinoma DOID-3498 mouse GSE53659 sample 699

juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 772

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679

juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 771

Cardiac Hypertrophy C1383860 rat GSE1055 sample 354

nemaline myopathy DOID-3191 mouse GSE3384 sample 974

nemaline myopathy DOID-3191 mouse GSE3384 sample 971

Nemaline Myopathy C0206157 mouse GSE3384 sample 317

Down syndrome DOID-14250 human GSE20910 sample 1063

idiopathic pulmonary fibrosis DOID-0050156 human GSE24206 sample 872

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 6000 genes

pulmonary tuberculosis DOID-2957 mouse GSE48027 sample 831

acute myocarditis DOID-3951 mouse GSE35182 sample 801

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679

nemaline myopathy DOID-3191 mouse GSE3384 sample 971

Purpura, Idiopathic Thrombocytopenic C0043117 human GSE574 sample 358

juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 772

West Nile fever DOID-2366 human GSE30719 sample 874

Nemaline Myopathy C0206157 mouse GSE3384 sample 317

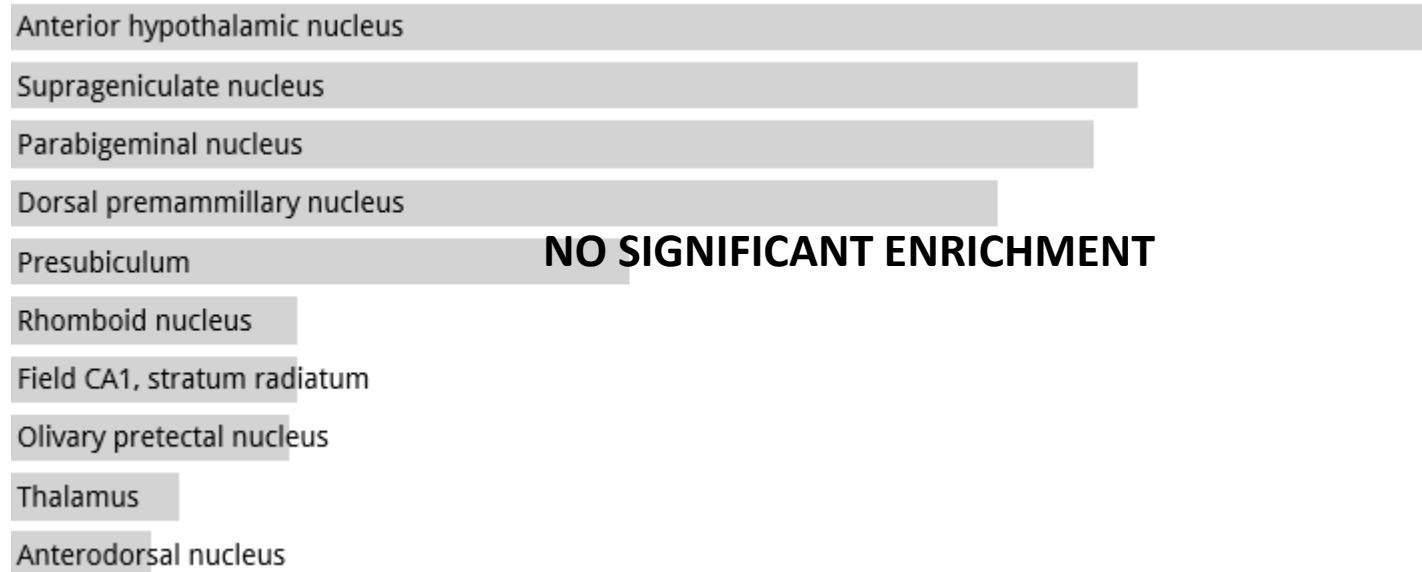
juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 771

acute myocardial infarction DOID-9408 mouse GSE775 sample 1000

NO SIGNIFICANT ENRICHMENT

Random Set 4

Allen Brain Atlas up random 6000 genes



Random Set 5

ARCHS4 Tissues random 6000 genes

SMALL INTESTINE (BULK TISSUE)

VASCULAR SMOOTH MUSCLE

SKIN (BULK TISSUE)

THYROID (BULK TISSUE)

STROMAL CELL

RESPIRATORY SMOOTH MUSCLE

RENAL CORTEX

SKELETAL MUSCLE (BULK TISSUE)

REGULATORY T CELLS

PANCREATIC ISLET

NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

NO DATA AVAILABLE

Random Set 5

Jensen DISEASES random 6000 genes

Enthesopathy

Gyrate_atrophy

Vitelliform_macular_dystrophy

Opisthorchiasis

Amelogenesis_imperfecta

3-methylglutaconic_aciduria

Mastitis

Cone_dystrophy

Hydrocephalus

Fundus_dystrophy

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes

nemaline myopathy DOID-3191 mouse GSE3384 sample 974

Nemaline Myopathy C0206157 mouse GSE3384 sample 317

Depression DOID-1595 mouse GSE26025 sample 463

autistic disorder DOID-12849 human GSE6575 sample 1043

Retinal damage C0235272 rat GSE1001 sample 312 **NO SIGNIFICANT ENRICHMENT**

Retinitis Pigmentosa C0035334 mouse GSE128 sample 33

Alzheimer's disease DOID-10652 human GSE36980 sample 523

Alcohol poisoning C0392620 rat GSE3311 sample 288

non-systemic juvenile idiopathic arthritis (sJIA) (subgroup-RF+ polyarthritis) DOID-848 human GSE21521 sample

colitis DOID-0060180 mouse GSE34874 sample 800

Disease Perturbations from GEO down random 6000 genes

polycystic ovary syndrome DOID-11612 human GSE48301 sample 558

glaucoma associated with systemic syndromes DOID-1686 mouse GSE26299 sample 491

MS (Multiple Sclerosis) C0026769 mouse GSE842 sample 381

breast adenocarcinoma DOID-3458 human GSE61304 sample 1071

Muscular Dystrophy C0026850 mouse GSE21521 sample 1043 **NO SIGNIFICANT ENRICHMENT**

Wolfram syndrome UMLS CUI-C0043207 mouse GSE15293 sample 695

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 681

Huntington's disease DOID-12858 mouse GSE3248 sample 723

Duchenne muscular dystrophy (DMD) C0013264 mouse GSE466 sample 328

Emery-Dreifuss muscular dystrophy, autosomal recessive form DOID-11726 mouse GSE8000 sample 677

Random Set 5

Allen Brain Atlas up random 6000 genes

Presubiculum

Endopiriform nucleus, dorsal part

dorsal endopiriform nucleus

Copula pyramidis

bed nucleus of the external capsule

mantle zone of r7Co

Agranular insular area, posterior part, layer 6a

Entorhinal area, medial part, dorsal zone, layer 5

corticoid layer of TuStr

Infralimbic area, layer 6b

NO SIGNIFICANT ENRICHMENT

Random Set 6

ARCHS4 Tissues random 6000 genes

NO DATA AVAILABLE

Jensen TISSUES random 6000 genes

NO DATA AVAILABLE

Random Set 6

Jensen DISEASES random 6000 genes

NO DATA AVAILABLE

Disease Perturbations from GEO up random 6000 genes

NO DATA AVAILABLE

Disease Perturbations from GEO down random 6000 genes

Systemic lupus erythematosus (SLE) C0024141 human GSE12374 sample 123

NO SIGNIFICANT ENRICHMENT

Random Set 6

Allen Brain Atlas up random 6000 genes

NO DATA AVAILABLE

Random Set 7

ARCHS4 Tissues random 6847 genes

LYMPHOCYTE

OMENTUM

GASTRIC TISSUE (BULK)

PREFRONTAL CORTEX

SPINAL CORD (BULK)

SPINAL CORD

LUNG (BULK TISSUE)

CARDIAC MUSCLE FIBER

BREAST (BULK TISSUE)

NEURONAL EPITHELIUM

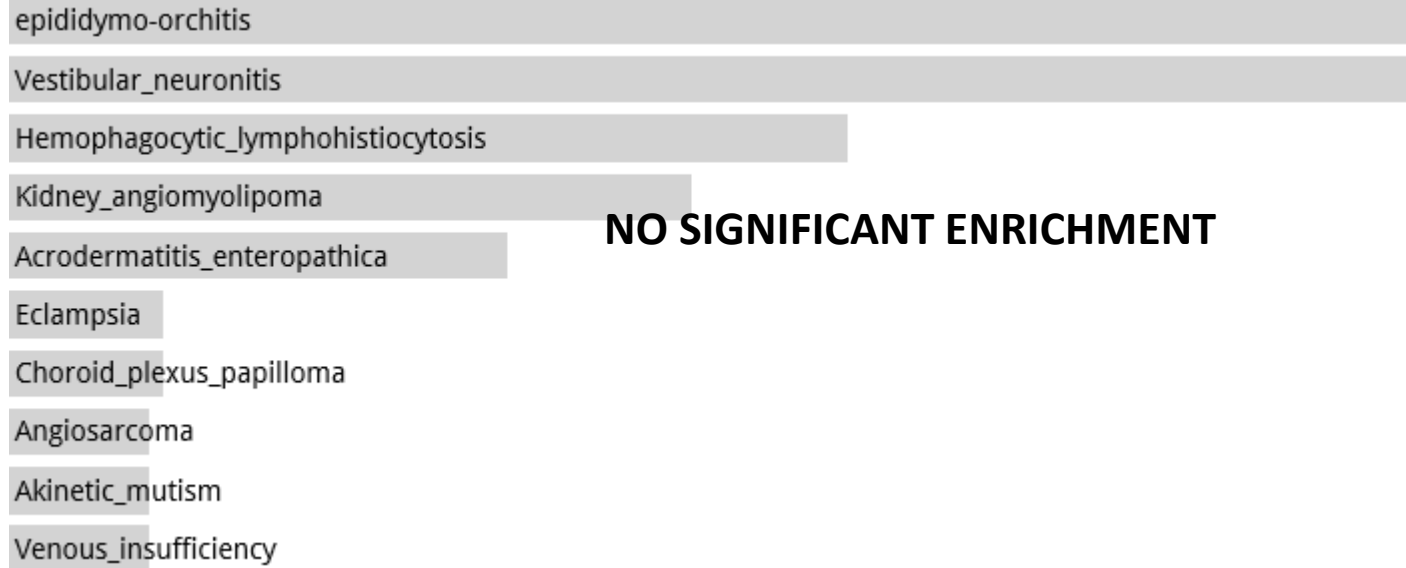
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

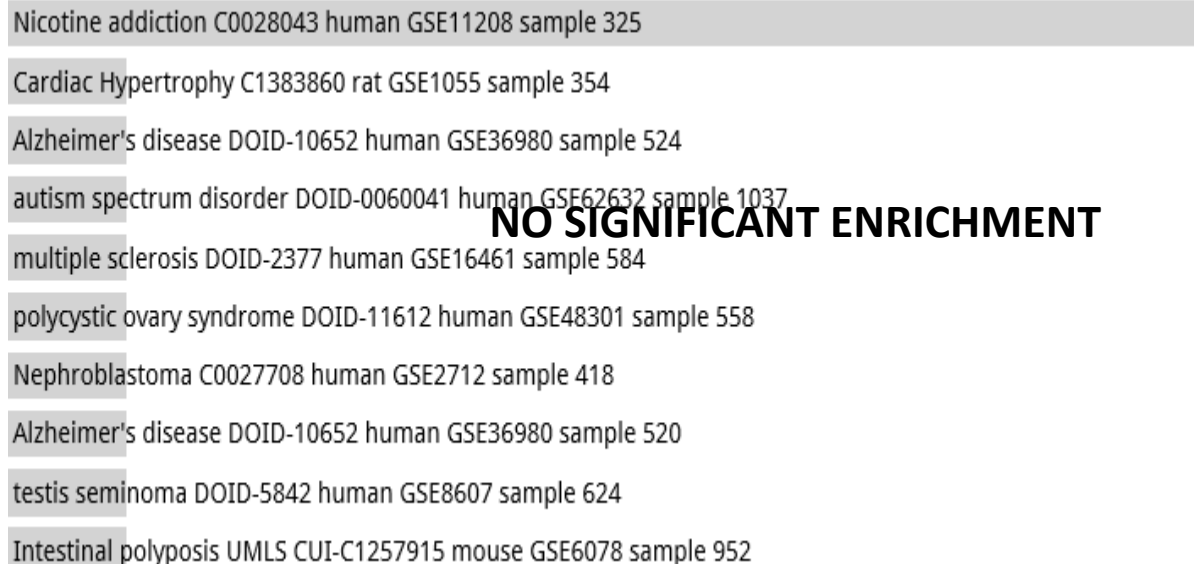
NO DATA AVAILABLE

Random Set 7

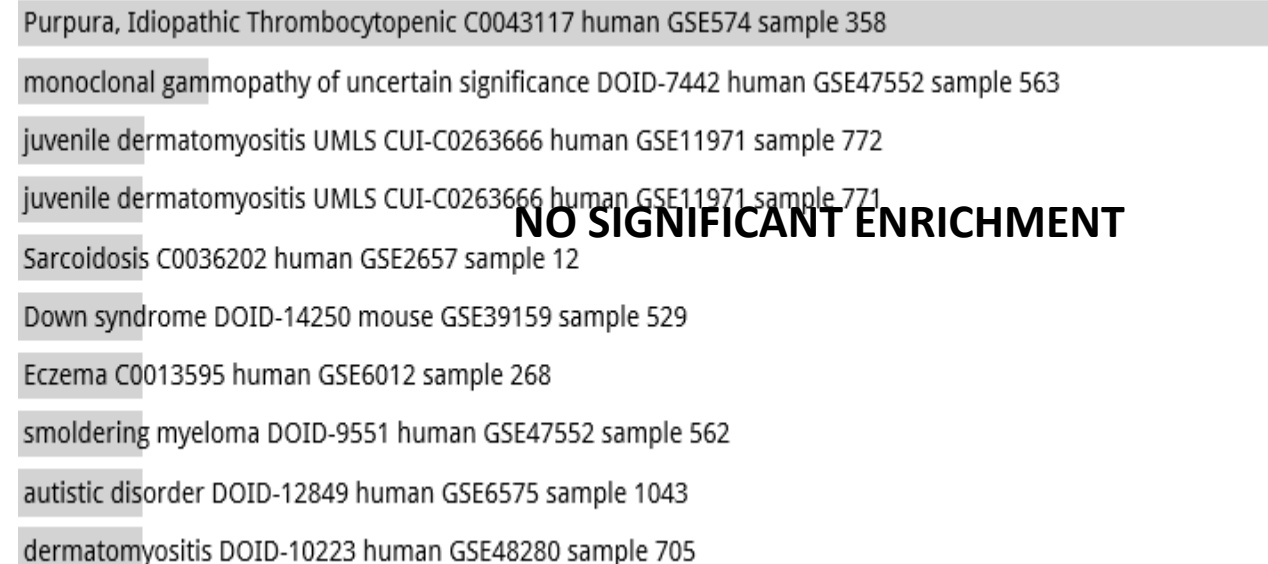
Jensen DISEASES random 6847 genes



Disease Perturbations from GEO up random 6000 genes

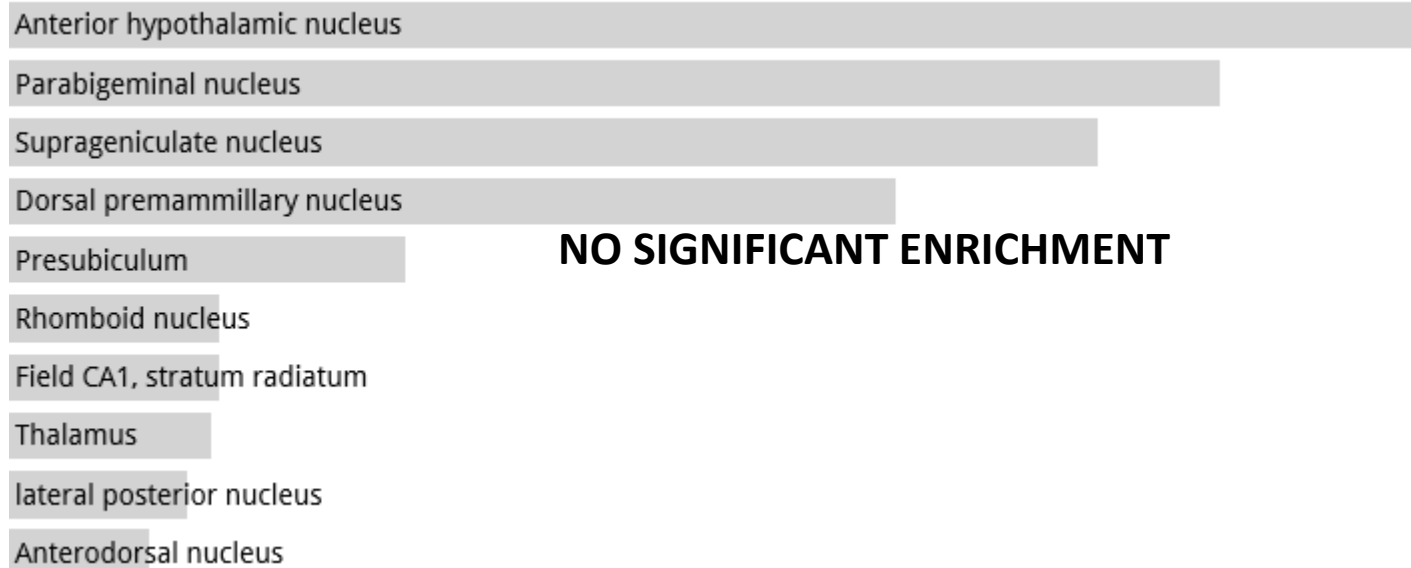


Disease Perturbations from GEO down random 6000 genes



Random Set 7

Allen Brain Atlas up random 6847 genes



Random Set 8

ARCHS4 Tissues random 6001 genes

GASTRIC TISSUE (BULK)

SKIN (BULK TISSUE)

RENAL CORTEX

KIDNEY (BULK TISSUE)

PREFRONTAL CORTEX

PANCREATIC ISLET

SPINAL CORD (BULK)

SMALL INTESTINE (BULK TISSUE)

COLON (BULK TISSUE)

SPINAL CORD

NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6001 genes

NO DATA AVAILABLE

Random Set 8

Jensen DISEASES random 6001 genes

Early_myoclonic_encephalopathy

Salpingitis

Vaginitis

cold-induced_sweating_syndrome

Parotitis

Carnitine_palmitoyltransferase_II_deficiency **NO SIGNIFICANT ENRICHMENT**

Poliomyelitis

Factor_XI_deficiency

cytochrome-c_oxidase_deficiency_disease

Ascariasis

Disease Perturbations from GEO up random 6000 genes

pancreatic ductal adenocarcinoma DOID-3498 mouse GSE53659 sample 699

Schistosomiasis C0036323 mouse GSE19525 sample 439

juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 772

juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 771

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679

pancreatic ductal adenocarcinoma DOID-3498 human GSE15471 sample 604

nemaline myopathy DOID-3191 mouse GSE3384 sample 974

asthma DOID-2841 human GSE31773 sample 714

glaucoma associated with systemic syndromes DOID-1686 mouse GSE26299 sample 488

Congestive heart disease C0018802 mouse GSE2236 sample 258

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 6000 genes

acute myocarditis DOID-3951 mouse GSE35182 sample 801

pulmonary tuberculosis DOID-2957 mouse GSE48027 sample 831

autistic disorder DOID-12849 human GSE6575 sample 1043

Purpura, Idiopathic Thrombocytopenic C0043117 human GSE574 sample 358

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679

Nicotine addiction C0028043 human GSE11208 sample 325

sickle-cell anemia DOID-10923 human GSE16728 sample 505

polycystic ovary syndrome DOID-11612 human GSE48301 sample 558

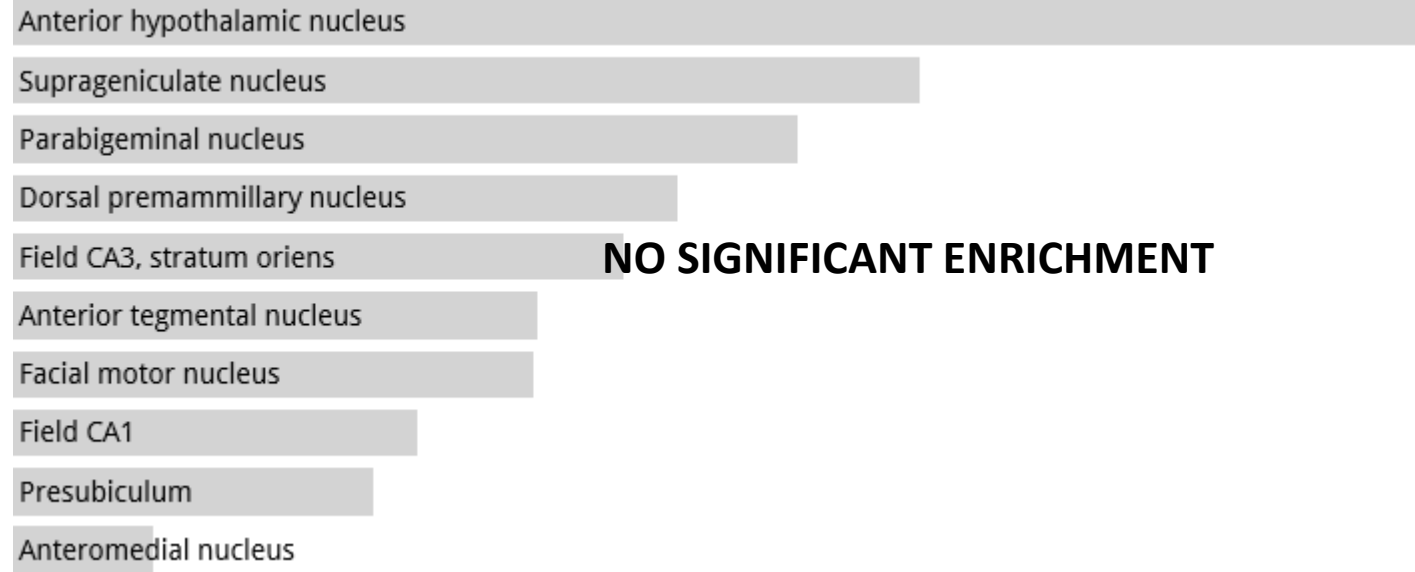
autism spectrum disorder DOID-0060041 human GSE62632 sample 1037

Cardiac Hypertrophy C1383860 rat GSE1055 sample 354

NO SIGNIFICANT ENRICHMENT

Random Set 8

Allen Brain Atlas up random 6001 genes



Random Set 9

ARCHS4 Tissues random 6000 genes

COLONIC MUCOSA

PREFRONTAL CORTEX

VENTRICLE

ASTROCYTE

MYOBLAST

BRAIN (BULK)

FETAL BRAIN CORTEX

NEURONAL EPITHELIUM

ATRIUM

ADIPOSE (BULK TISSUE)

NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6000 genes

NO DATA AVAILABLE

Random Set 9

Jensen DISEASES random 6000 genes

Hereditary_mucosal_leukokeratosis

Dumping_syndrome

X-linked_endothelial_corneal_dystrophy

Clouston_syndrome

Enlarged_vestibular_aqueduct

Oculodentodigital_dysplasia

Porphyria_cutanea_tarda

MEDNIK_syndrome

Granulomatous_amebic_encephalitis

Pachyonychia_congenita

NO SIGNIFICANT ENRICHMENT

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes

Dental cavity, complex C0399396 human GSE1629 sample 175

actinic keratosis DOID-8866 human GSE2503 sample 628

Actinic keratosis C0022602 human GSE2503 sample 350

acute myeloid leukemia DOID-9119 human GSE9476 sample 782

Epilepsy C0014544 human GSE7486 sample 417

epidermolysis bullosa simplex DOID-4644 human GSE28315 sample 711

lupus erythematosus DOID-8857 human GSE30153 sample 739

cardiomyopathy DOID-0050700 human GSE9128 sample 781

type 2 diabetes mellitus DOID-9352 human GSE23343 sample 895

autism spectrum disorder DOID-0060041 human GSE25507 sample 1032

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 6000 genes

multiple myeloma DOID-9538 human GSE6691 sample 787

Smoldering multiple myeloma C1531608 human GSE5900 sample 404

autistic disorder DOID-12849 human GSE6575 sample 1043

acute myocarditis DOID-3951 mouse GSE35182 sample 801

pulmonary tuberculosis DOID-2957 mouse GSE48027 sample 831

autistic disorder DOID-12849 human GSE6575 sample 1042

mental retardation DOID-1059 human GSE6575 sample 1044

Septic Shock C0036983 human GSE9692 sample 307

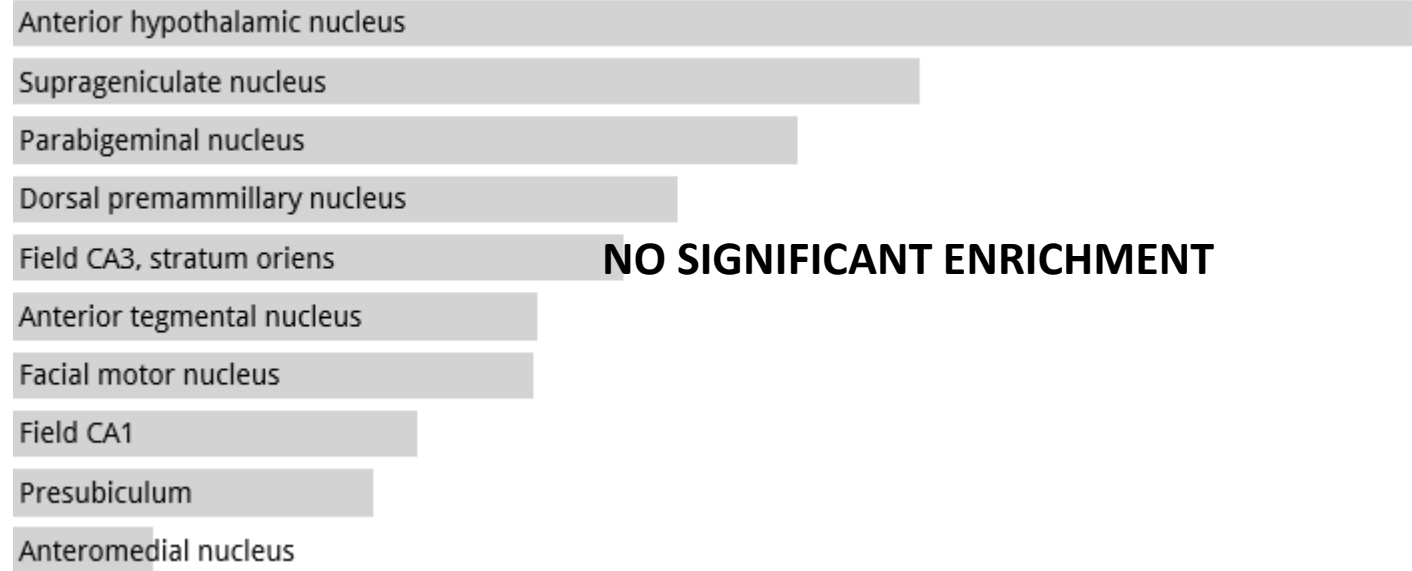
West Nile fever DOID-2366 human GSE30719 sample 874

Waldenstrom Macroglobulinemia UMLS CUI-C0024419 human GSE6691 sample 785

NO SIGNIFICANT ENRICHMENT

Random Set 9

Allen Brain Atlas up random 6000 genes



Random Set 10

ARCHS4 Tissues random 6000 genes

GASTRIC TISSUE (BULK)

PREFRONTAL CORTEX

OMENTUM

COLON (BULK TISSUE)

LUNG (BULK TISSUE)

NEURONAL EPITHELIUM

LIVER (BULK TISSUE)

FETAL BRAIN CORTEX

RENAL CORTEX

AMNIOTIC FLUID

NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6000 genes

NO DATA AVAILABLE

Random Set 10

Jensen DISEASES random 6000 genes

Laryngeal_squamous_cell_carcinoma

Urinary_schistosomiasis

Ebola_hemorrhagic_fever

Hermaphroditism

Irritant_dermatitis

Zellweger_syndrome

Aortic_valve_insufficiency

Scimitar_syndrome

Gastric_lymphoma

Multiple_chemical_sensitivity

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes

Alzheimer's disease DOID-10652 human GSE36980 sample 519

polycystic ovary syndrome DOID-11612 human GSE48301 sample 558

Cardiac Hypertrophy C1383860 rat GSE1055 sample 354

ulcerative colitis DOID-8577 human GSE37283 sample 594

pancreatic ductal adenocarcinoma DOID-3498 mouse GSE53659 sample 699

juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 772

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679

juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 771

nemaline myopathy DOID-3191 mouse GSE3384 sample 974

nemaline myopathy DOID-3191 mouse GSE3384 sample 971

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 6000 genes

pulmonary tuberculosis DOID-2957 mouse GSE48027 sample 831

acute myocarditis DOID-3951 mouse GSE35182 sample 801

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679

nemaline myopathy DOID-3191 mouse GSE3384 sample 971

Down syndrome DOID-14250 mouse GSE39159 sample 529

Cardiomyopathy, Dilated C0007193 human GSE3586 sample 323

Purpura, Idiopathic Thrombocytopenic C0043117 human GSE574 sample 358

acute myocardial infarction DOID-9408 mouse GSE775 sample 1000

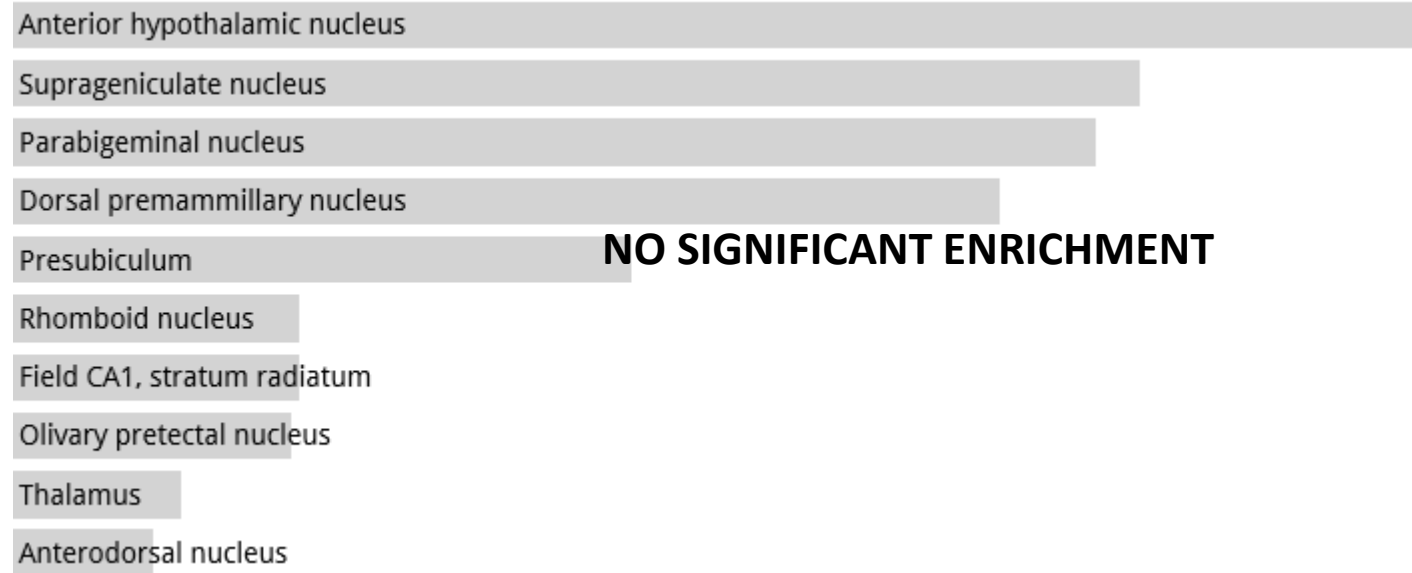
papillary thyroid carcinoma DOID-3969 human GSE54958 sample 652

juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 772

NO SIGNIFICANT ENRICHMENT

Random Set 10

Allen Brain Atlas up random 6000 genes



Random Set 11

ARCHS4 Tissues random 4000 genes

OMENTUM

VENTRICLE

SKIN (BULK TISSUE)

WHARTONS JELLY

VASCULAR SMOOTH MUSCLE

OVARY (BULK TISSUE)

CEREBELLUM

CHONDROCYTE

THYROID (BULK TISSUE)

SPINAL CORD (BULK)

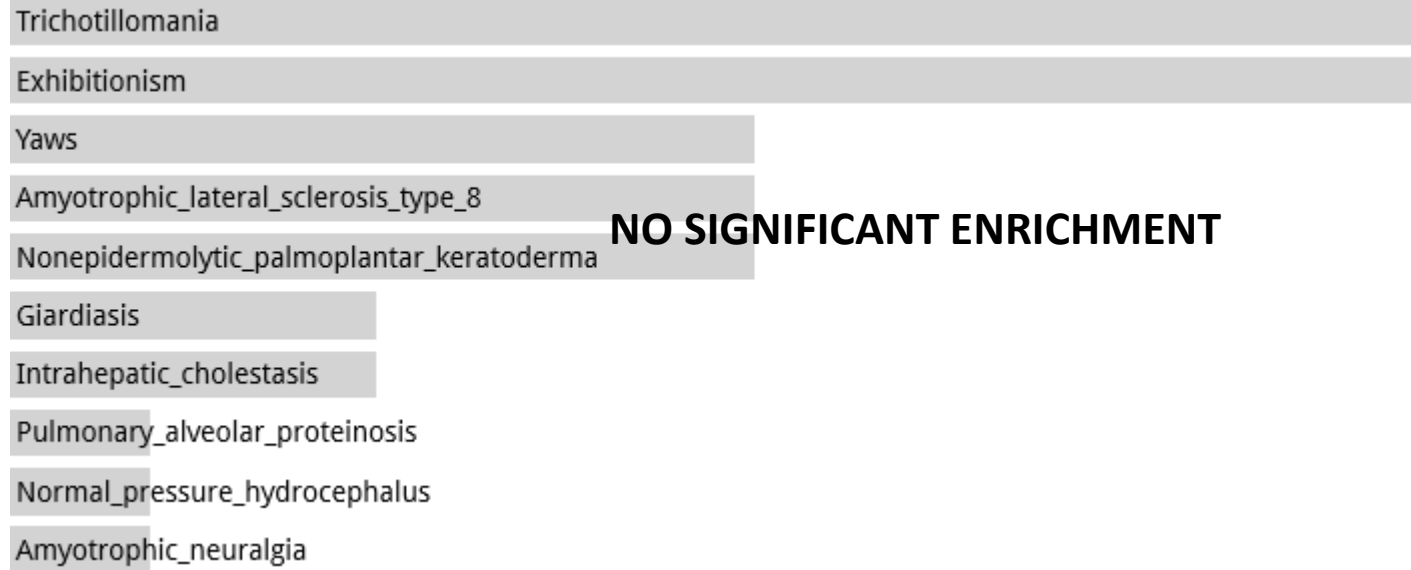
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

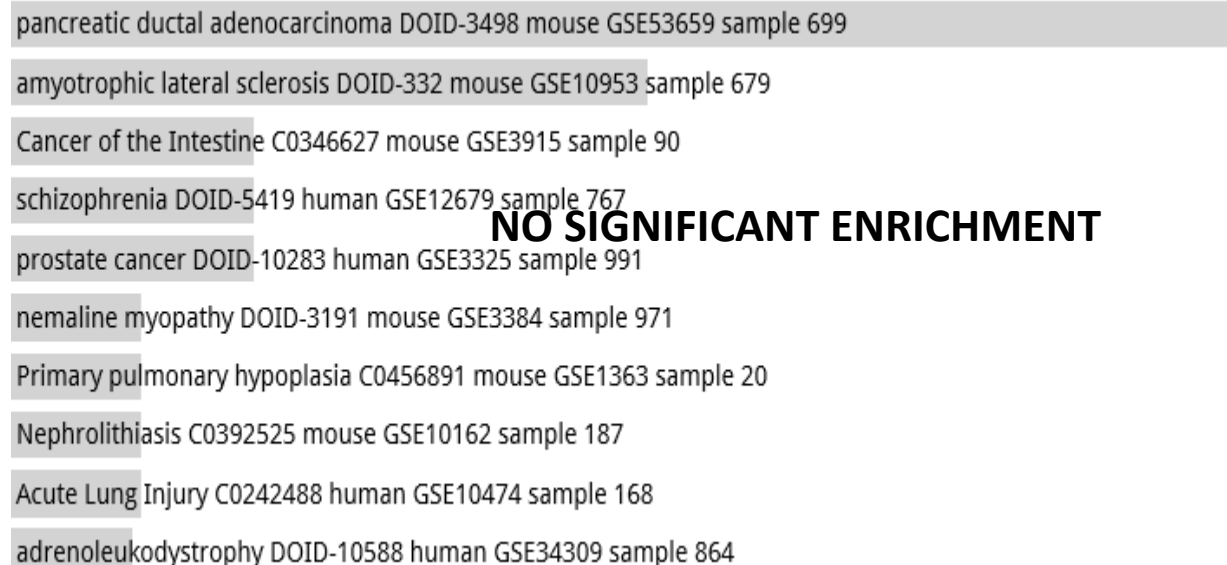
NO DATA AVAILABLE

Random Set 11

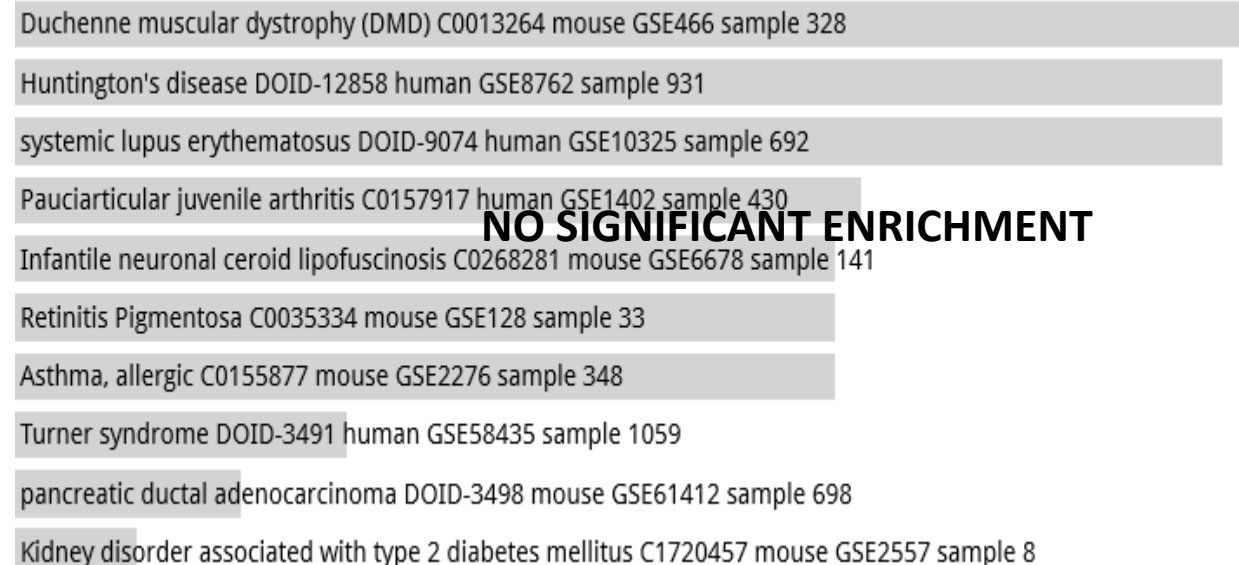
Jensen DISEASES random 4000 genes



Disease Perturbations from GEO up random 4000 genes

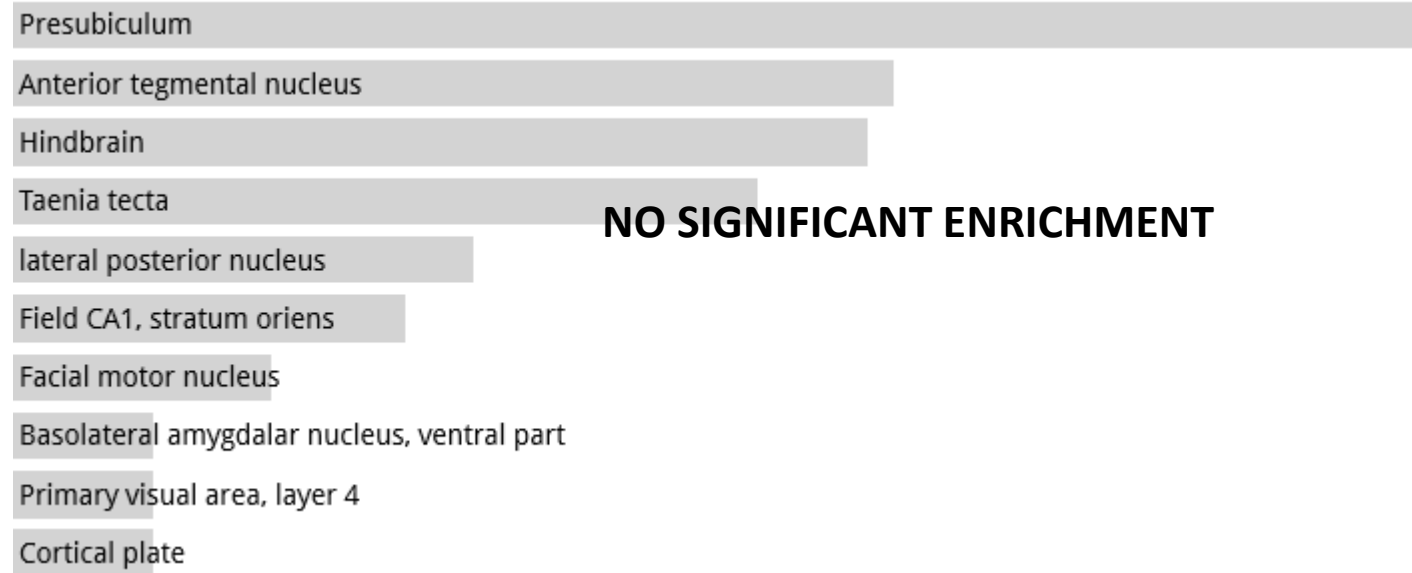


Disease Perturbations from GEO down random 4000 genes



Random Set 11

Allen Brain Atlas up random 4000 genes



Random Set 12

ARCHS4 Tissues random 4000 genes

GASTRIC TISSUE (BULK)

RENAL CORTEX

SKIN (BULK TISSUE)

SMALL INTESTINE (BULK TISSUE)

PANCREATIC ISLET

PREFRONTAL CORTEX

KIDNEY (BULK TISSUE)

SPINAL CORD (BULK)

PLACENTA (BULK)

SPINAL CORD

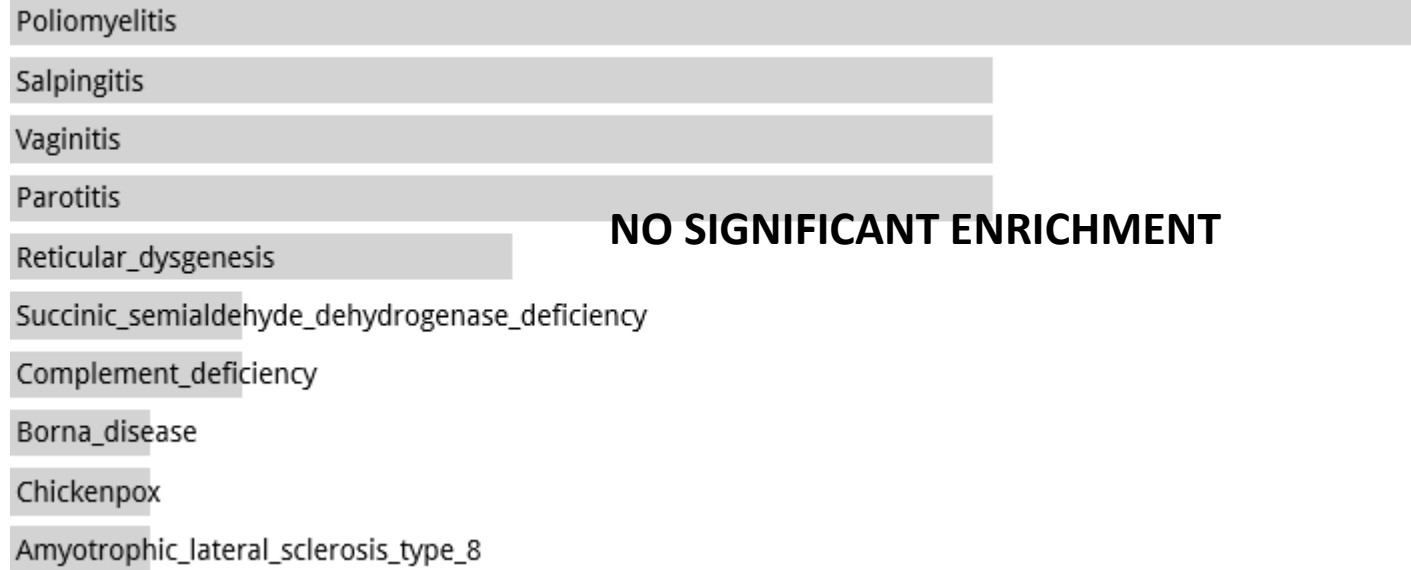
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

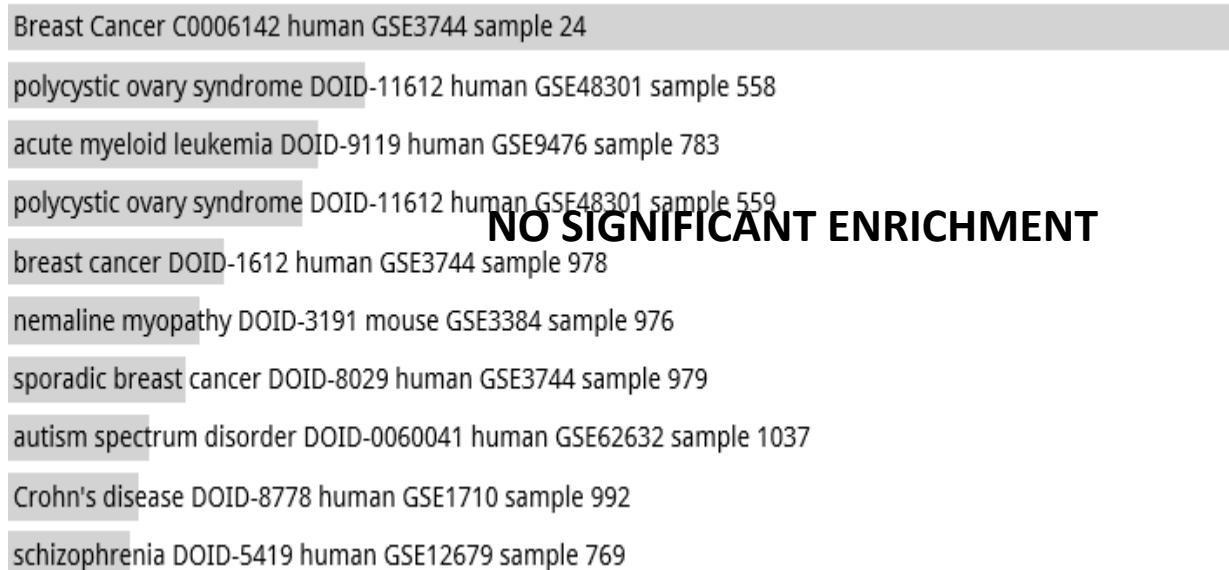
NO DATA AVAILABLE

Random Set 12

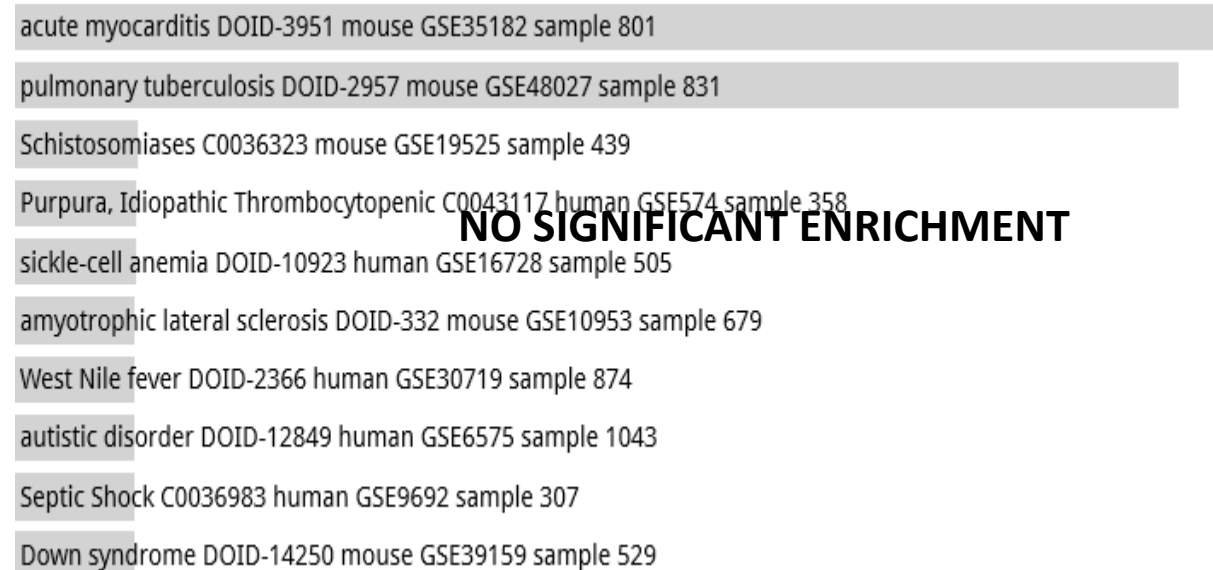
Jensen DISEASES random 4000 genes



Disease Perturbations from GEO up random 4000 genes



Disease Perturbations from GEO down random 4000 genes



Random Set 12

Allen Brain Atlas up random 4000 genes

dorsal juxtacommissural pretectal nucleus

periventricular stratum of JcPL

JcPL part of the periaqueductal gray

periventricular stratum of PcPL

mantle zone of JcPD

PcPL part of the periaqueductal gray

intermediate stratum of JcPD

dorsal part of JcP

CoPV part of the periaqueductal gray

periventricular stratum of CoPV

NO SIGNIFICANT ENRICHMENT

Random Set 13

ARCHS4 Tissues random 4000 genes

COLON (BULK TISSUE)

ILEUM (BULK)

SMALL INTESTINE (BULK TISSUE)

GASTRIC TISSUE (BULK)

OMENTUM

NEURONAL EPITHELIUM

SKIN (BULK TISSUE)

CEREBELLUM

PREFRONTAL CORTEX

MYOBLAST

NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

Random Set 13

Jensen DISEASES random 4000 genes

cytochrome-c_oxidase_deficiency_disease

Cockayne_syndrome

Arterial_tortuosity_syndrome

cold-induced_sweating_syndrome

NO SIGNIFICANT ENRICHMENT

Esotropia

Xeroderma_pigmentosum

Favism

Placenta_praevia

Carnitine_palmitoyltransferase_II_deficiency

Patent_foramen_ovale

Disease Perturbations from GEO up random 4000 genes

autism spectrum disorder DOID-0060041 human GSE62632 sample 1037

Severe acute respiratory syndrome (SARS) C1175175 human GSE1739 sample 165

Nicotine addiction C0028043 human GSE11208 sample 325

ulcerative colitis DOID-8577 human GSE37283 sample 594

NO SIGNIFICANT ENRICHMENT

COPD - Chronic obstructive pulmonary disease C0024117 human GSE3320 sample 255

asthma DOID-2841 human GSE43696 sample 827

Idiopathic fibrosing alveolitis C0085786 human GSE10921 sample 272

Cancer of the testis C0153594 human GSE1818 sample 246

bipolar disorder DOID-3312 human GSE5392 sample 573

Ulcerative Colitis C0009324 human GSE1710 sample 264

Disease Perturbations from GEO down random 4000 genes

autistic disorder DOID-12849 human GSE6575 sample 1043

Purpura, Idiopathic Thrombocytopenic C0043117 human GSE574 sample 358

pulmonary tuberculosis DOID-2957 mouse GSE48027 sample 831

sickle-cell anemia DOID-10923 human GSE16728 sample 506

NO SIGNIFICANT ENRICHMENT

acute myocarditis DOID-3951 mouse GSE35182 sample 802

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679

Septic Shock C0036983 human GSE9692 sample 307

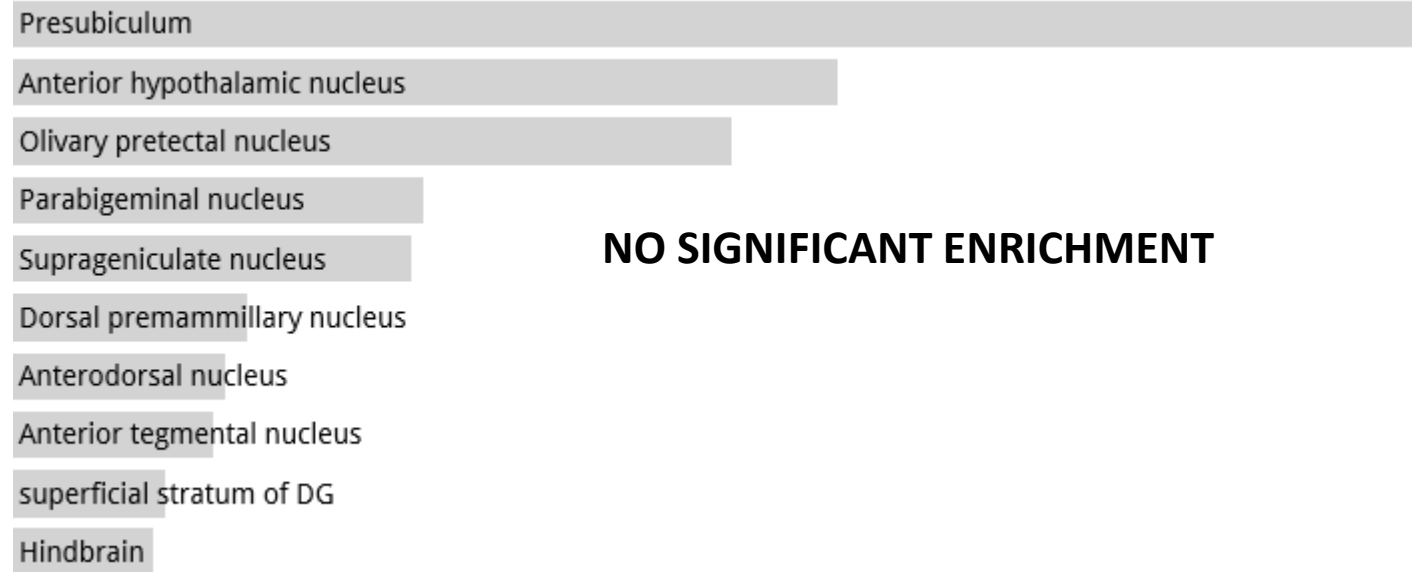
Down syndrome DOID-14250 mouse GSE39159 sample 529

juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 772

amyotrophic lateral sclerosis DOID-332 human GSE833 sample 1007

Random Set 13

Allen Brain Atlas up random 4000 genes



Random Set 14

ARCHS4 Tissues random 4000 genes

COLONIC MUCOSA

p-value: 0.0048; Adjusted p-value: 0.5188

CD19+ B CELLS

BLYMPHOCYTE

ILEUM (BULK)

NO SIGNIFICANT ENRICHMENT

COLON (BULK TISSUE)

PLASMA CELL

GASTRIC TISSUE (BULK)

BLASTOCYST

PERIPHERAL BLOOD

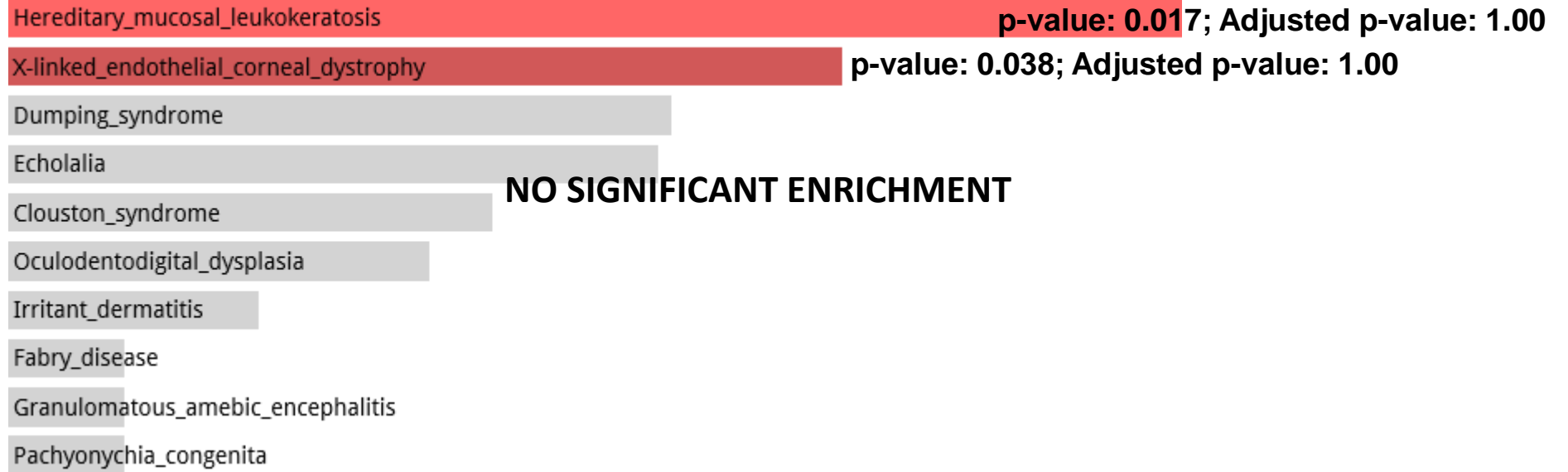
SPLEEN (BULK TISSUE)

Jensen TISSUES random 4000 genes

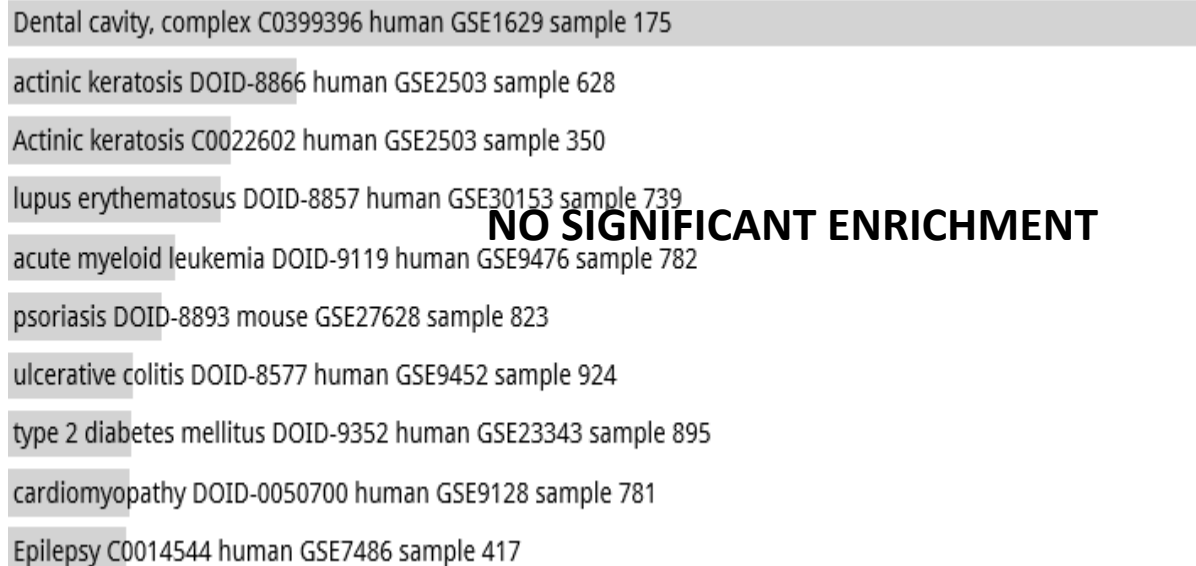
NO DATA AVAILABLE

Random Set 14

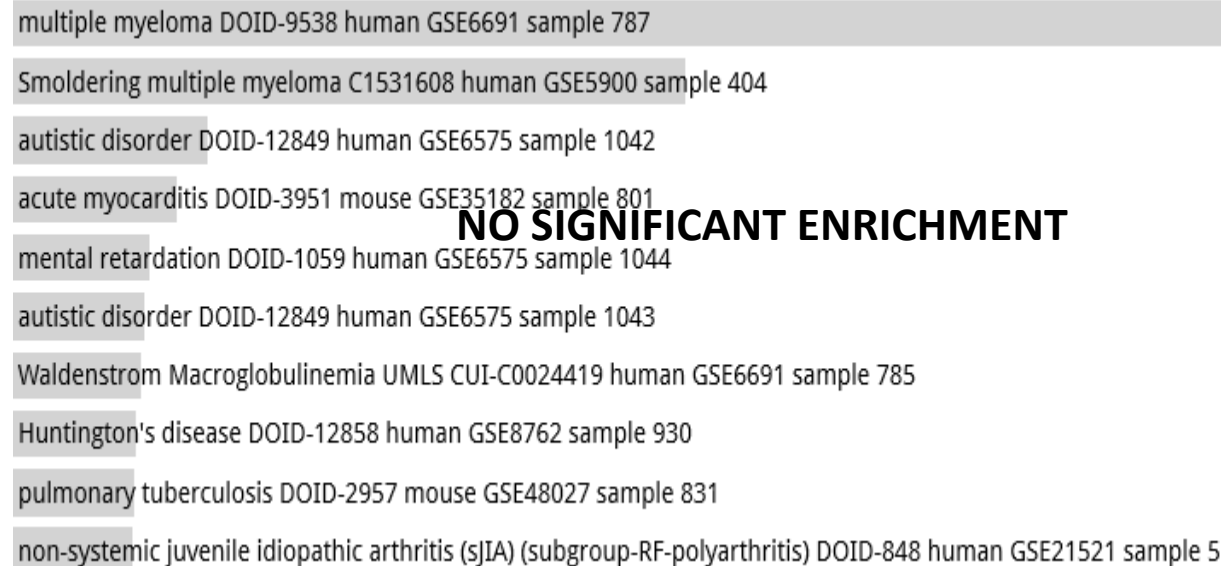
Jensen DISEASES random 4000 genes



Disease Perturbations from GEO up random 4000 genes



Disease Perturbations from GEO down random 4000 genes



Random Set 14

Allen Brain Atlas up random 4000 genes

periventricular stratum of the VAP

Nucleus of the solitary tract, lateral part

Lateral amygdalar nucleus

intermediate stratum of r4Ve

r9 part of the vestibular column

NO SIGNIFICANT ENRICHMENT

Lateral reticular nucleus, parvicellular part

mantle zone of r9Ve

r4 part of magnocellular medial vestibular nucleus

periventricular stratum of r8Tr

superficial stratum of r7Lim

Random Set 15

COLON (BULK TISSUE)

GASTRIC TISSUE (BULK)

PREFRONTAL CORTEX

ILEUM (BULK)

LUNG (BULK TISSUE)

NEURONAL EPITHELIUM

OMENTUM

LIVER (BULK TISSUE)

AMNIOTIC FLUID

RENAL CORTEX

ARCHS4 Tissues random 4000 genes

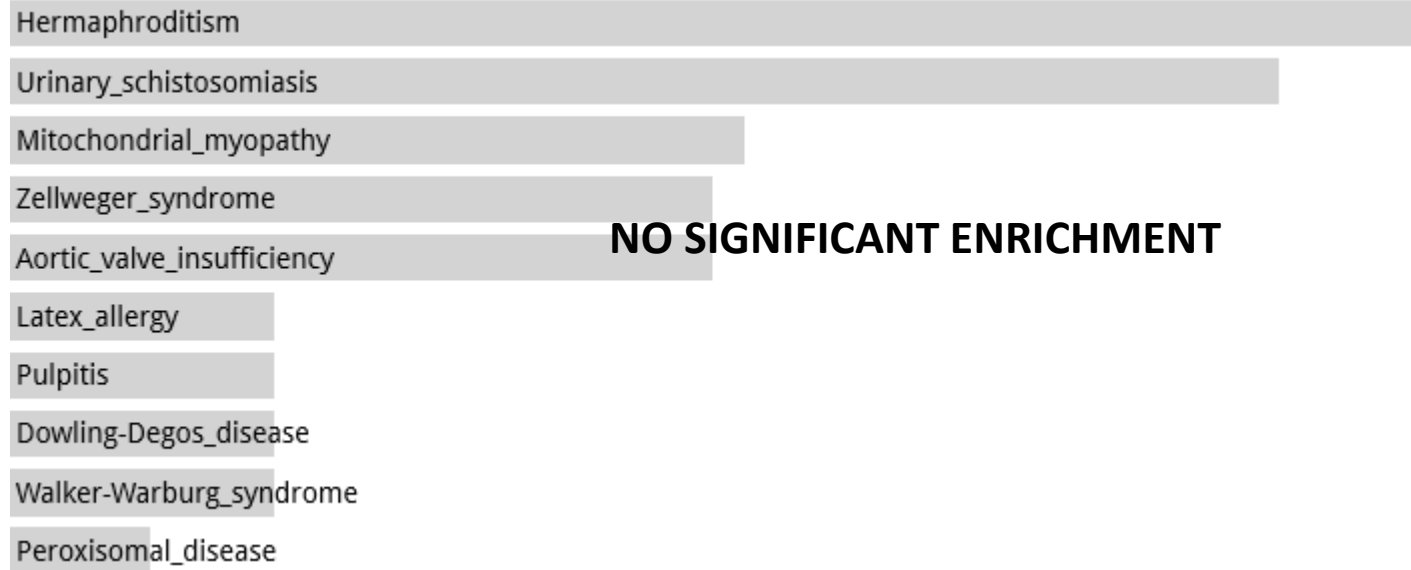
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

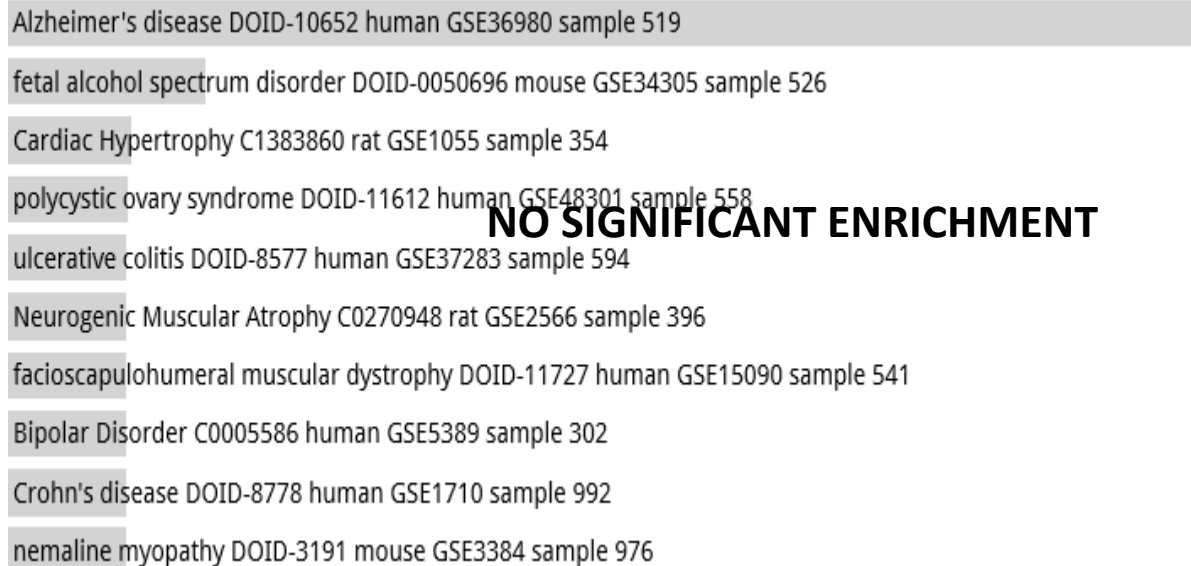
NO DATA AVAILABLE

Random Set 15

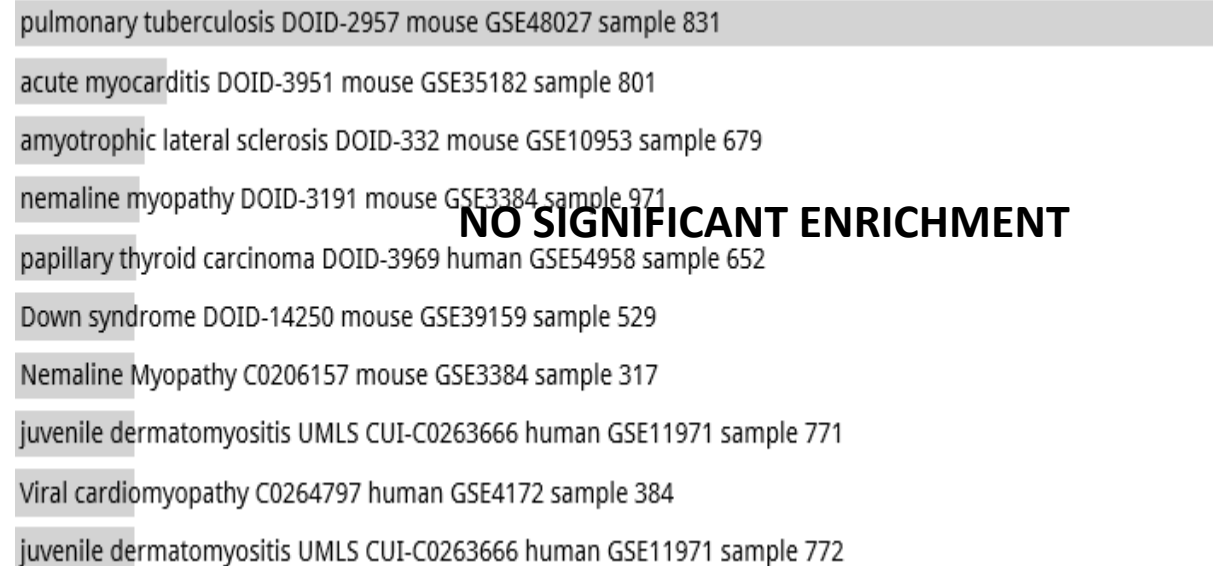
Jensen DISEASES random 4000 genes



Disease Perturbations from GEO up random 4000 genes



Disease Perturbations from GEO down random 4000 genes



Random Set 15

Allen Brain Atlas up random 4000 genes

rhombomere 8

retropontine reticular area

medullary hindbrain (medulla)

Visceral area

colliculus superior

mantle zone of SC

superficial gray layer of SC

Retrosplenial area, dorsal part, layer 1

Paragigantocellular reticular nucleus, dorsal part

Gustatory areas

NO SIGNIFICANT ENRICHMENT

Random Set 16

ARCHS4 Tissues random 4000 genes

PREFRONTAL CORTEX

SPINAL CORD (BULK)

SUPERIOR FRONTAL GYRUS

OMENTUM

SPINAL CORD

LUNG (BULK TISSUE)

NEURONAL EPITHELIUM

FETAL BRAIN CORTEX

ASTROCYTE

BRAIN (BULK)

NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

Random Set 16

Jensen DISEASES random 4000 genes

Scimitar_syndrome

Western_equine_encephalitis

Opitz-GBBB_syndrome

Conversion_disorder

Gastric_lymphoma

Cutaneous_porphyria

Leiomyomatosis

Epididymitis

DNA_ligase_IV_deficiency

Juvenile_polyposis_syndrome

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 4000 genes

pancreatic ductal adenocarcinoma DOID-3498 mouse GSE53659 sample 699

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679

autism spectrum disorder DOID-0060041 human GSE62632 sample 1037

RA (rheumatoid arthritis) C0003873 human GSE3592 sample 183

schizophrenia DOID-5419 human GSE27383 sample 548

Cancer of the Intestine C0346627 mouse GSE3915 sample 90

schizophrenia DOID-5419 human GSE12679 sample 767

Duchenne muscular dystrophy (DMD) C0013264 mouse GSE1472 sample 62

swine influenza DOID-0050211 human GSE48466 sample 498

psoriasis DOID-8893 human GSE26952 sample 982

NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 4000 genes

pulmonary tuberculosis DOID-2957 mouse GSE48027 sample 831

Purpura, Idiopathic Thrombocytopenic C0043117 human GSE574 sample 358

West Nile fever DOID-2366 human GSE30719 sample 874

amyotrophic lateral sclerosis DOID-332 mouse GSE10953 sample 679

Down syndrome DOID-14250 mouse GSE39159 sample 529

pancreatic ductal adenocarcinoma DOID-3498 mouse GSE53659 sample 699

Cardiomyopathy, Dilated C0007193 human GSE3586 sample 323

nemaline myopathy DOID-3191 mouse GSE3384 sample 971

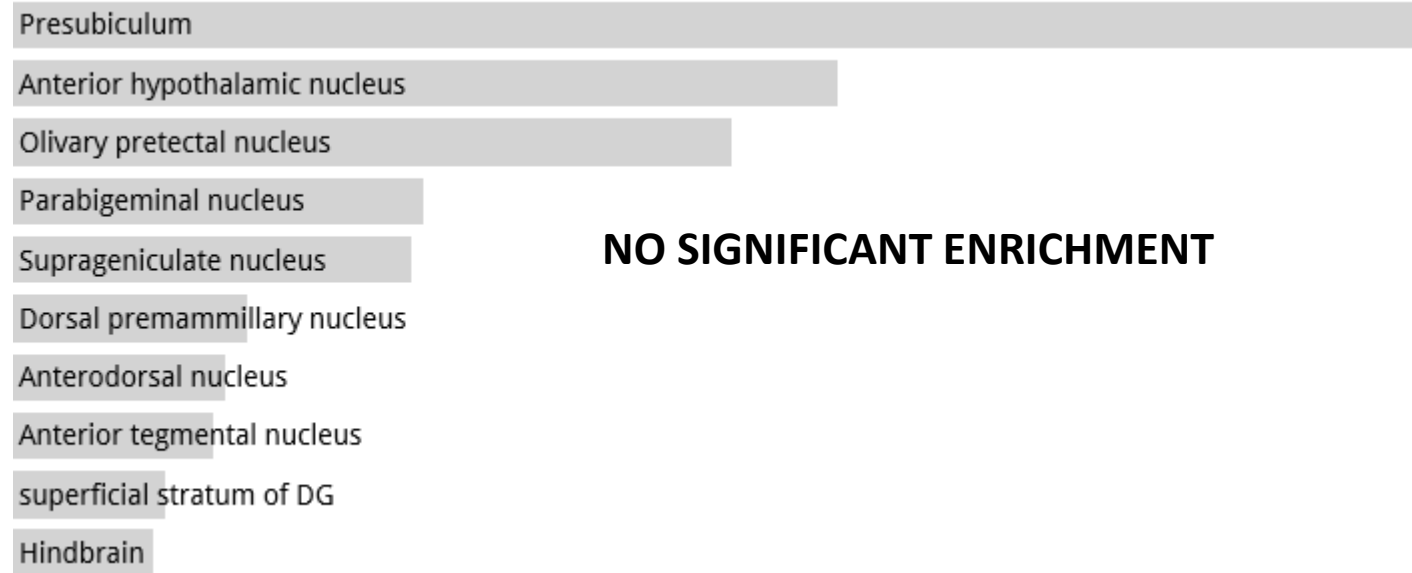
juvenile dermatomyositis UMLS CUI-C0263666 human GSE11971 sample 772

Schistosomiasis C0036323 mouse GSE19525 sample 439

NO SIGNIFICANT ENRICHMENT

Random Set 16

Allen Brain Atlas up random 4000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 17

ARCHS4 Tissues random 4000 genes

SMALL INTESTINE (BULK TISSUE)

VASCULAR SMOOTH MUSCLE

SKIN (BULK TISSUE)

THYROID (BULK TISSUE)

STROMAL CELL

RESPIRATORY SMOOTH MUSCLE

RENAL CORTEX

SKELETAL MUSCLE (BULK TISSUE)

REGULATORY T CELLS

PANCREATIC ISLET

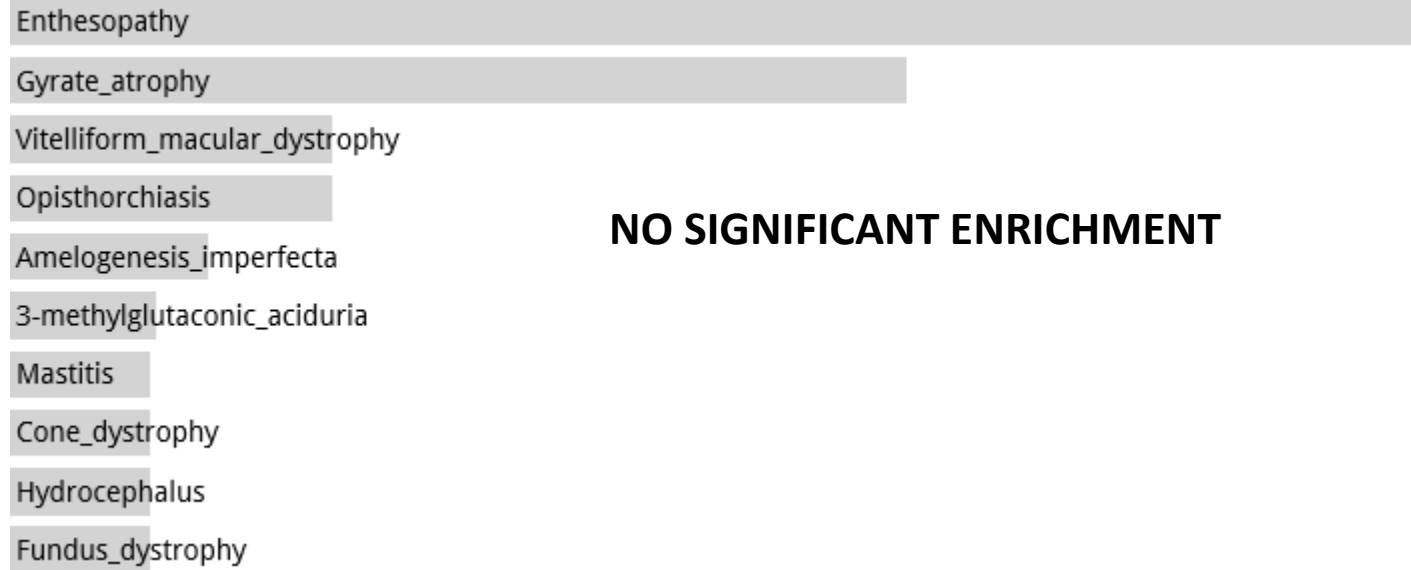
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

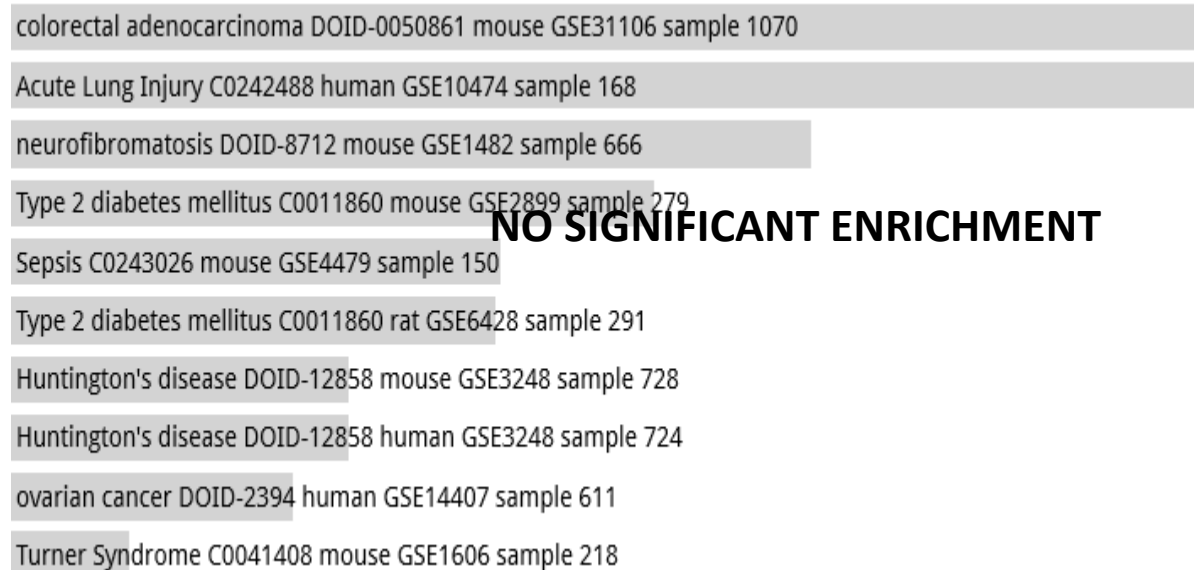
NO DATA AVAILABLE

Random Set 17

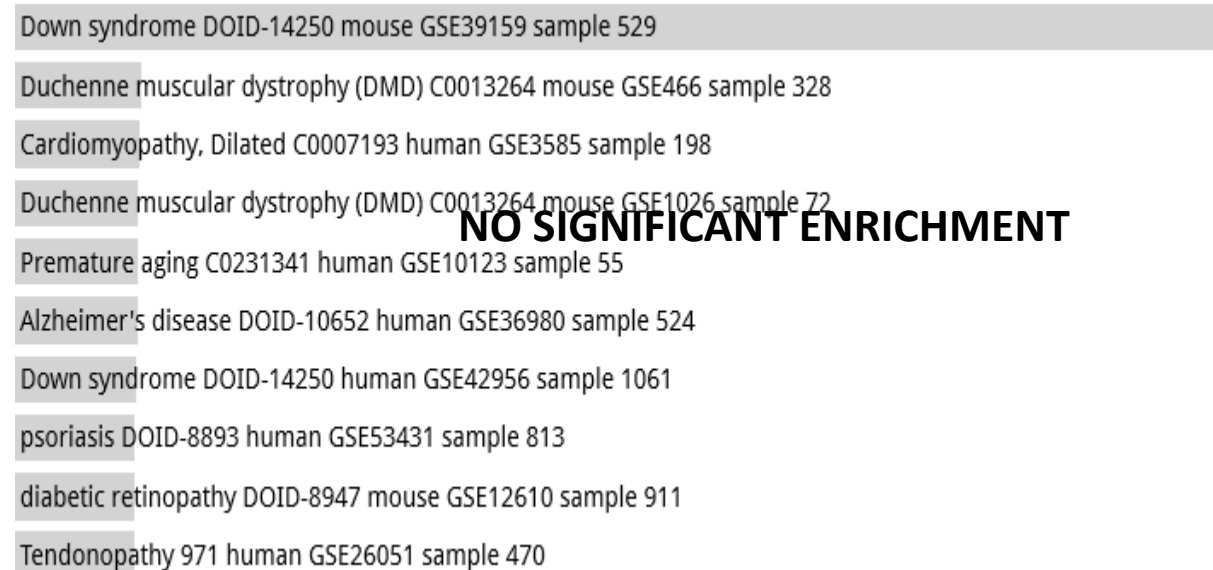
Jensen DISEASES random 4000 genes



Disease Perturbations from GEO up random 4000 genes



Disease Perturbations from GEO down random 4000 genes



Random Set 17

Allen Brain Atlas up random 4000 genes

Presubiculum

Endopiriform nucleus, dorsal part

dorsal endopiriform nucleus

Copula pyramidis

bed nucleus of the external capsule

mantle zone of r7Co

Agranular insular area, posterior part, layer 6a

Entorhinal area, medial part, dorsal zone, layer 5

corticoid layer of TuStr

Infralimbic area, layer 6b

NO SIGNIFICANT ENRICHMENT

Random Set 18

ARCHS4 Tissues random 4000 genes

NO DATA AVAILABLE

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

Random Set 18

Jensen DISEASES random 4000 genes

NO DATA AVAILABLE

Disease Perturbations from GEO up random 4000 genes

Disease Perturbations from GEO down random 4000 genes

NO DATA AVAILABLE

NO DATA AVAILABLE

Random Set 18

Allen Brain Atlas up random 4000 genes

NO DATA AVAILABLE

Random Set 19

ARCHS4 Tissues random 4000 genes

NO DATA AVIALABLE

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

Random Set 19

Jensen DISEASES random 4000 genes

NO DATA AVAILABLE

Disease Perturbations from GEO up random 4000 genes

Disease Perturbations from GEO down random 4000 genes

NO DATA AVAILABLE

NO DATA AVAILABLE

Random Set 19

Allen Brain Atlas up random 4000 genes

NO DATA AVAILABLE

Random Set 20

ARCHS4 Tissues random 4000 genes

LYMPHOCYTE

BLASTOCYST

BLOOD DENDRITIC CELLS

BONE MARROW (BULK TISSUE)

SPINAL CORD (BULK)

GASTRIC TISSUE (BULK)

SPINAL CORD

MOTOR NEURON

PREFRONTAL CORTEX

NEURONAL EPITHELIUM

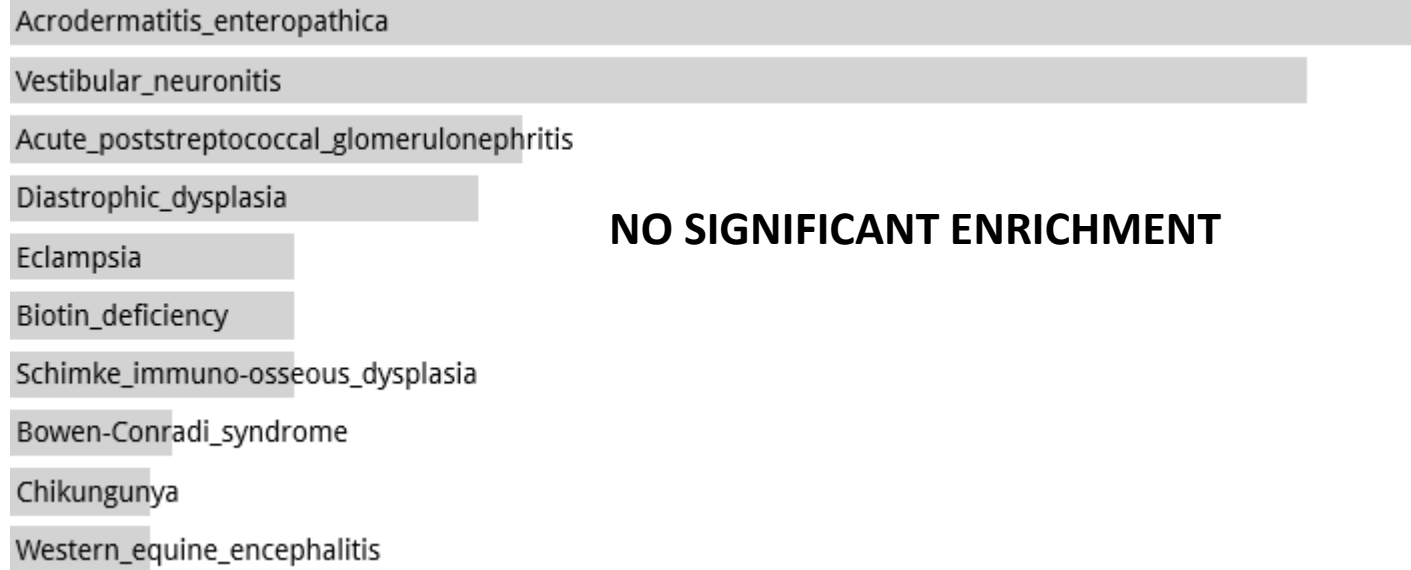
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

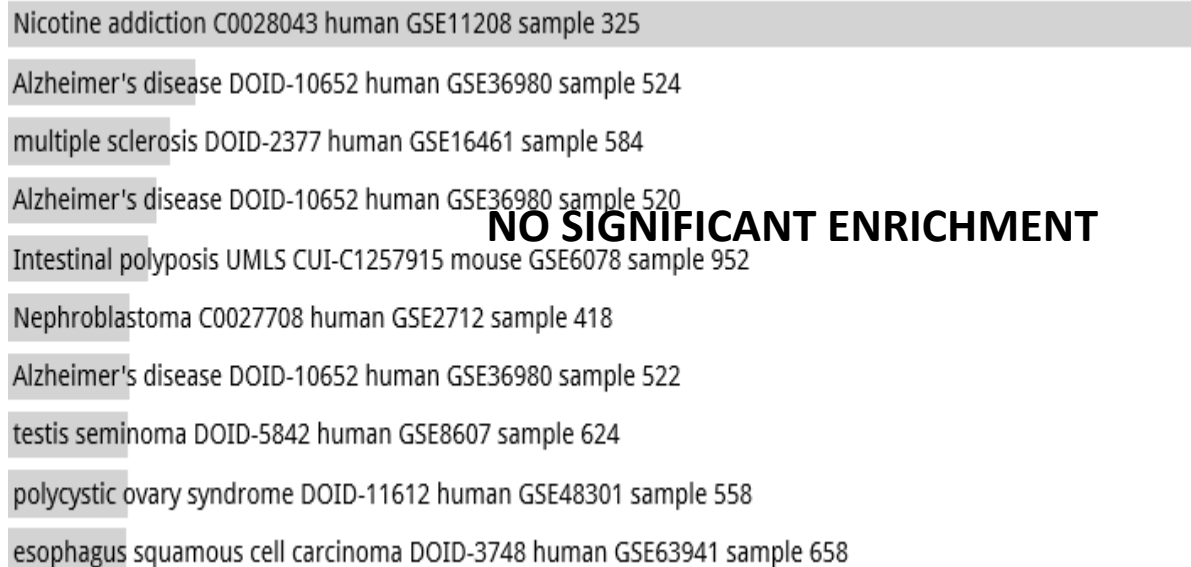
NO DATA AVAILABLE

Random Set 20

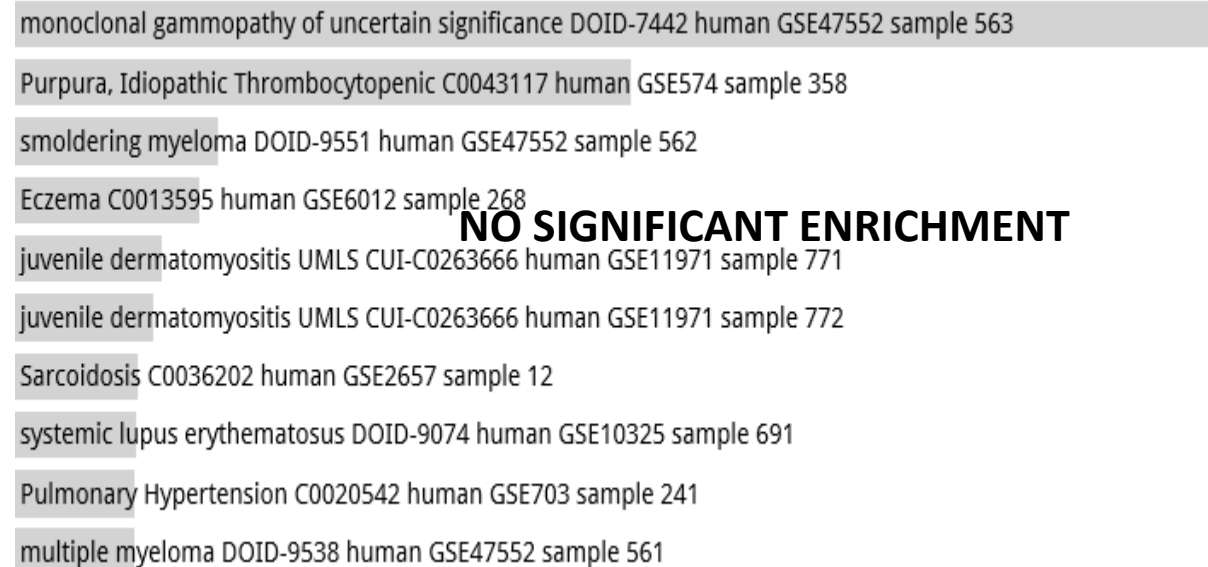
Jensen DISEASES random 4000 genes



Disease Perturbations from GEO up random 4000 genes

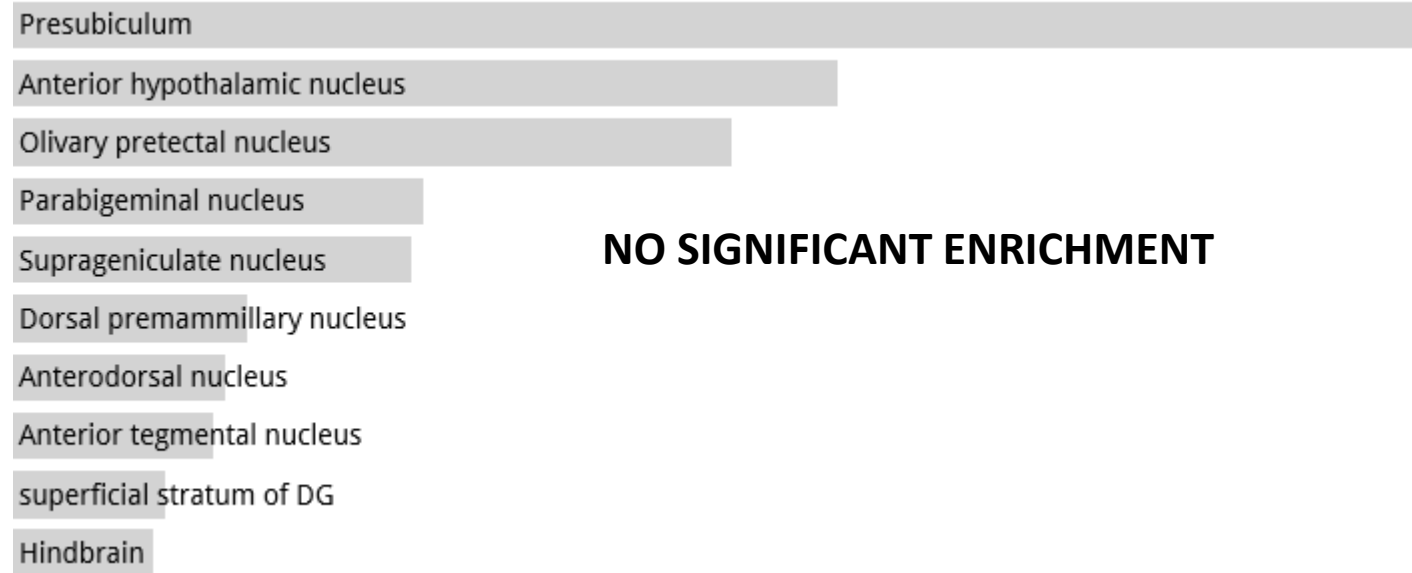


Disease Perturbations from GEO down random 4000 genes



Random Set 20

Allen Brain Atlas up random 4000 genes

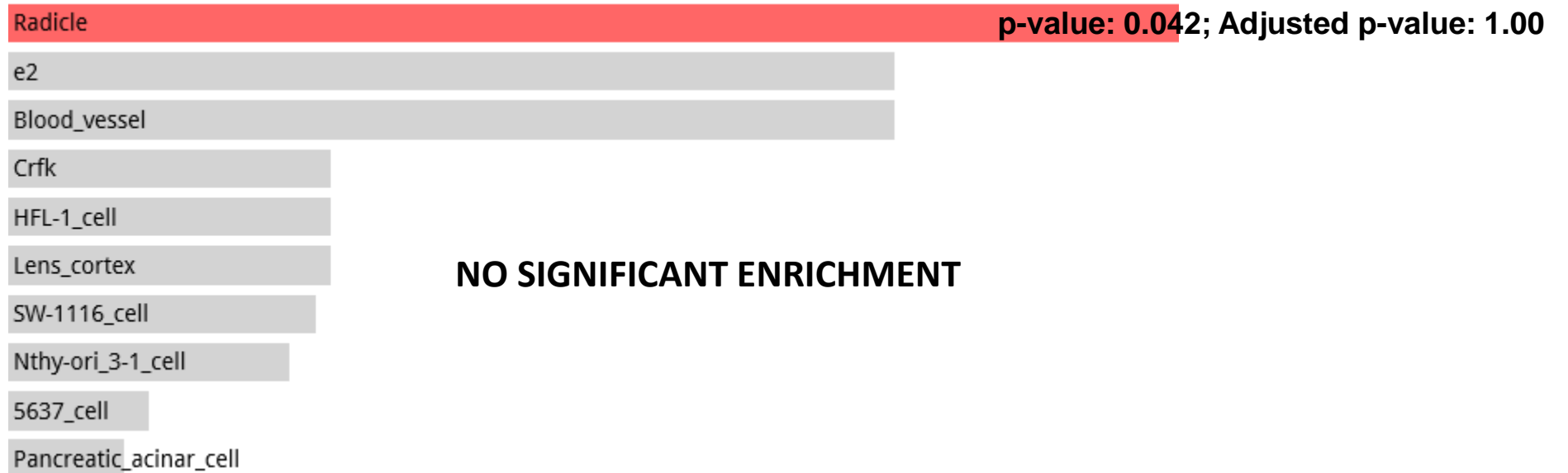


Random Set 21

ARCHS4 Tissues random 2847 genes



Jensen TISSUES random 2847 genes

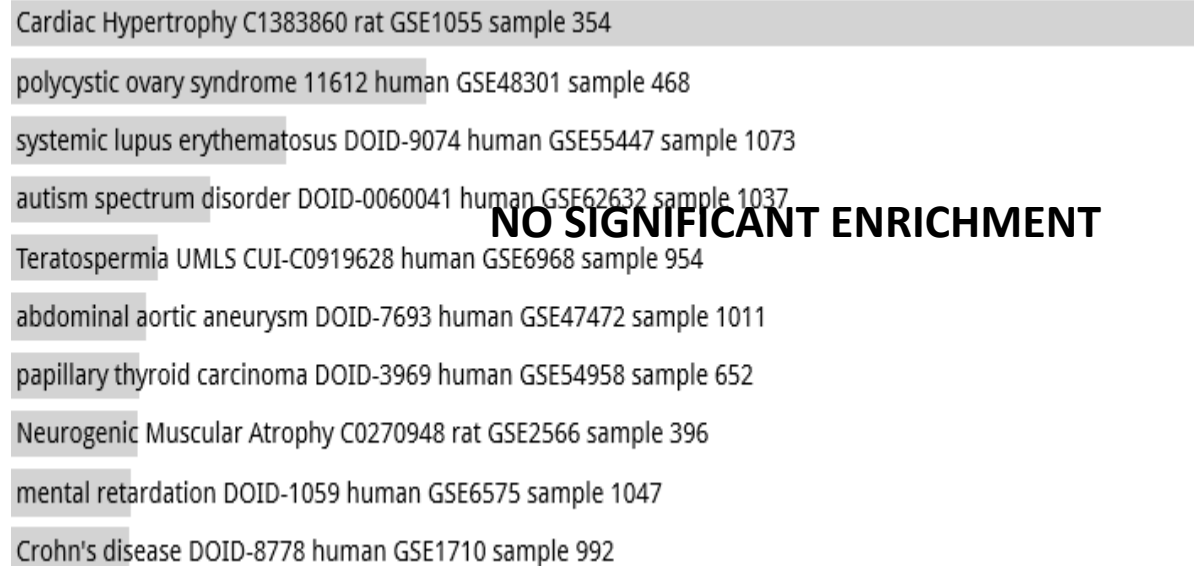


Random Set 21

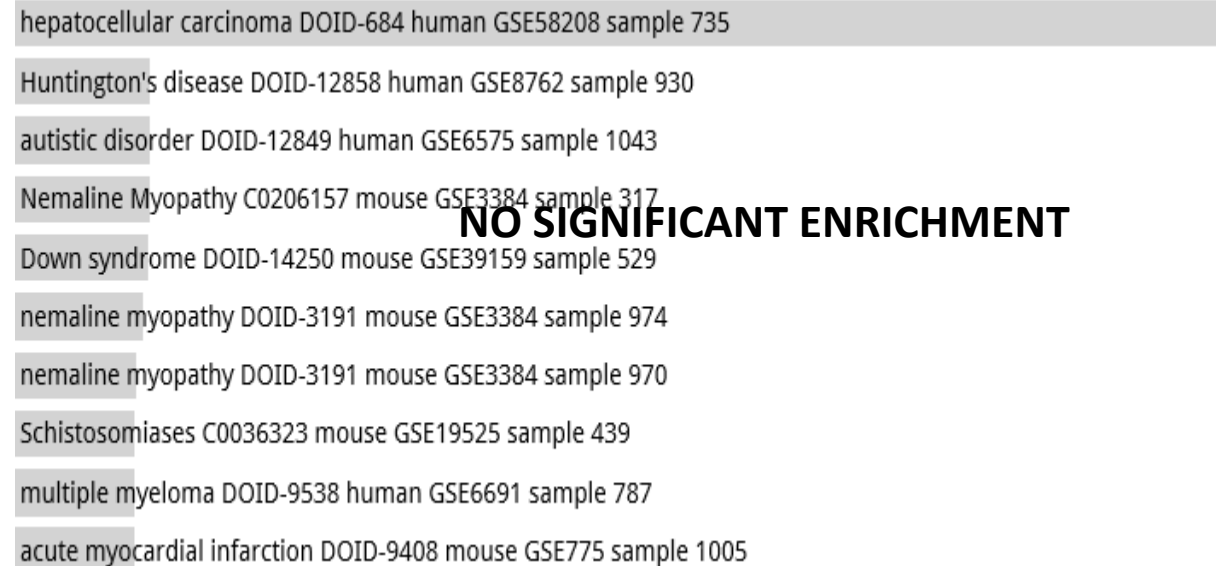
Jensen DISEASES random 2847 genes



Disease Perturbations from GEO up random 2847 genes

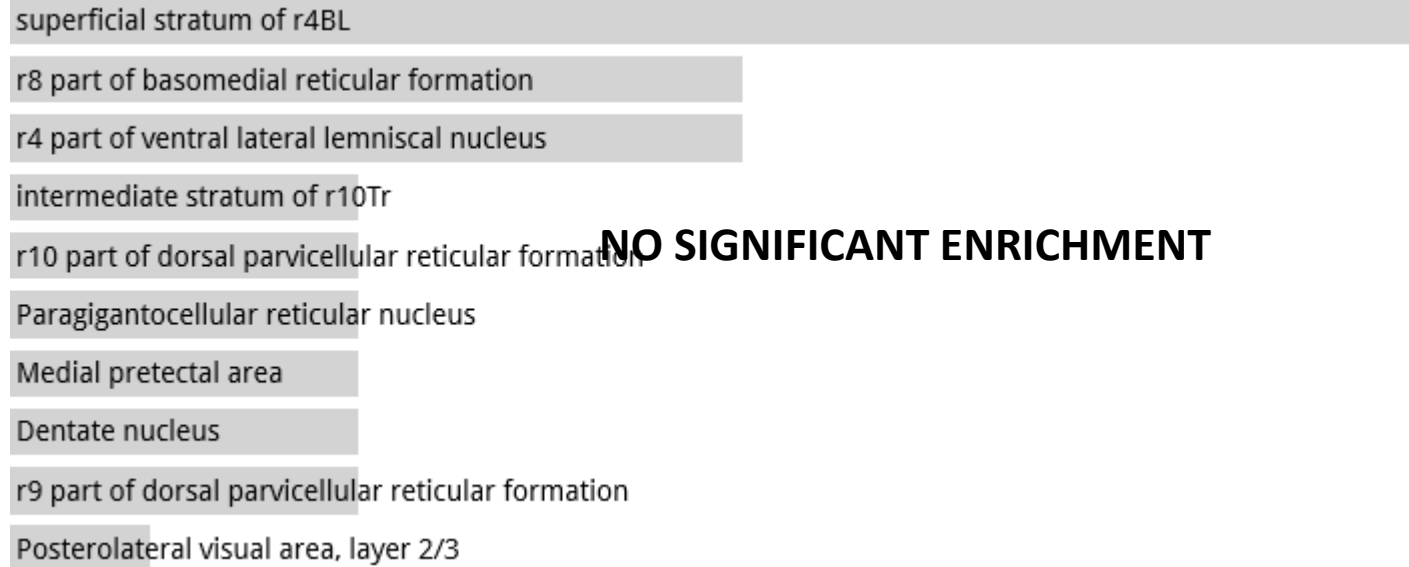


Disease Perturbations from GEO down random 2847 genes

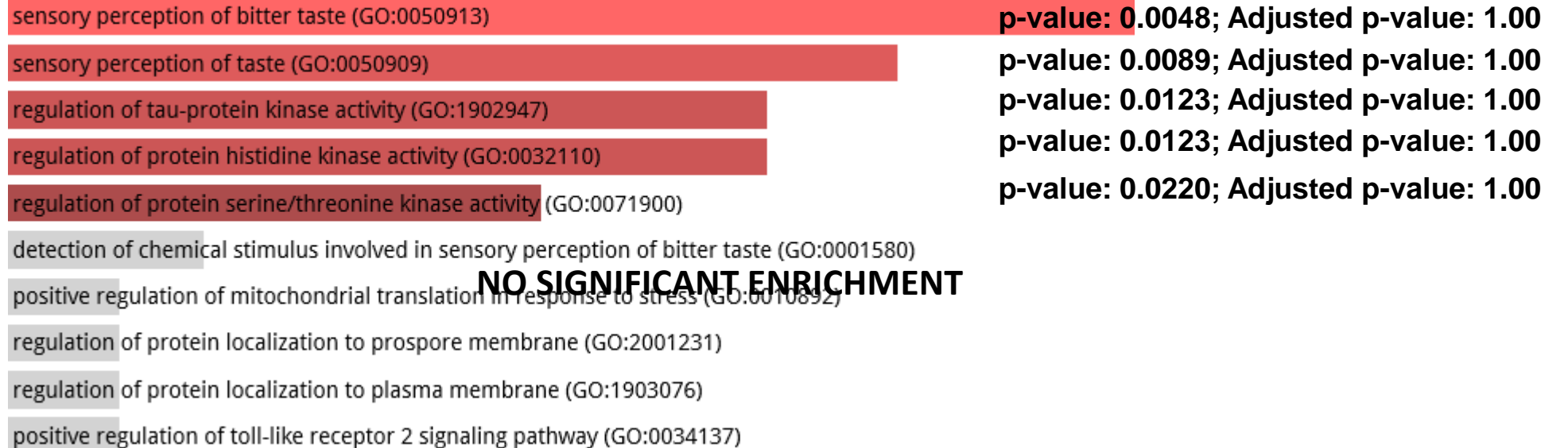


Random Set 21

Allen Brain Atlas up random 2847 genes

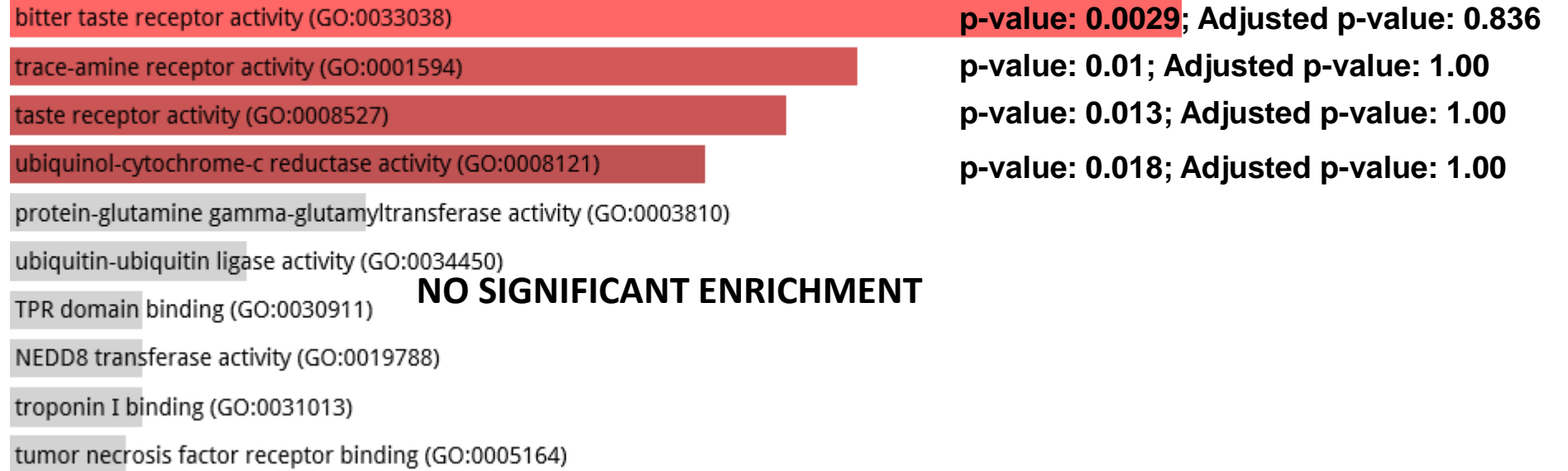


GO Biological Process 2017b 2847 genes

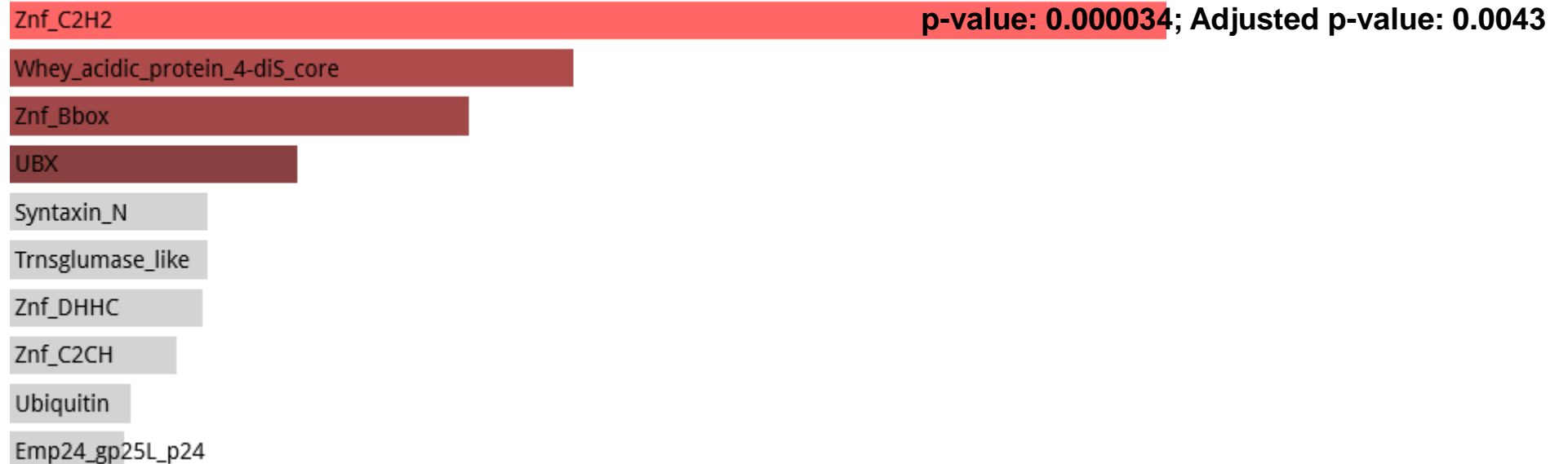


Random Set 21

GO Molecular Function 2017 2847 genes



Pfam InterPro Domains 2847 genes



Random Set 21

GWAS Catalog 2019 2847 genes

Alcoholic chronic pancreatitis	p-value: 0.00049; Adjusted p-value: 0.31
Plasminogen activator inhibitor type 1 levels (PAI-1)	
Total bilirubin levels in HIV-1 infection	
Cerebellum growth	
Response to simvastatin treatment (PCSK9 protein level change)	
NO SIGNIFICANT ENRICHMENT	
Pancreatic ductal adenocarcinoma	
Hepcidin/transferrin saturation ratio	
Renal function-related traits (sCR)	
Proteinuria and chronic kidney disease	
Coffee consumption (cups per day)	

GO Biological Process 2018 2847 genes

bitter taste receptor activity (GO:0033038)	p-value: 0.00047; Adjusted p-value: 0.1939
taste receptor activity (GO:0008527)	p-value: 0.0034; Adjusted p-value: 0.7151
trace-amine receptor activity (GO:0001594)	p-value: 0.018; Adjusted p-value: 1.00
oxidoreductase activity, acting on diphenols and related substances as donors, cytochrome as acceptor (GO:0004711)	p-value: 0.028; Adjusted p-value: 1.00
ubiquinol-cytochrome-c reductase activity (GO:0008121)	p-value: 0.028; Adjusted p-value: 1.00
NO SIGNIFICANT ENRICHMENT	
uridylyltransferase activity (GO:0070569)	
protein-glutamine gamma-glutamyltransferase activity (GO:0003810)	
ubiquitin-ubiquitin ligase activity (GO:0034450)	
MHC protein binding (GO:0042287)	
small protein activating enzyme activity (GO:0008641)	

Random Set 21

CORUM 2847 genes

Class C Vps complex (VPS11, VPS18, VPS16) (human)	p-value: 0.0029; Adjusted p-value: 0.2781
Class C Vps complex (VPS11, VPS18, STX7) (human)	p-value: 0.0029; Adjusted p-value: 0.2781
Class C VPS/HOPS complex (human)	p-value: 0.0048; Adjusted p-value: 0.3109
TBP-TAF complex (human)	p-value: 0.01; Adjusted p-value: 0.3975
TIF-IB complex (mouse)	p-value: 0.01; Adjusted p-value: 0.3975
TFIID complex (human)	
TFIID complex, B-cell specific (human)	
TRAF2-TRADD complex (human)	
TSC1-TSC2 complex (human)	
TFTC complex (TATA-binding protein-free TAF-II-containing complex) (human)	

NO SIGNIFICANT ENRICHMENT

CORUM database of protein complexes identified by mass spectrometry

