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)

Identify genes regulated by HSGRS

Human stem cell-associated retroviral sequences (SCARS)

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

Analytical Pipelines

Define panels of genomic regulatory loci

Functional enhancers of naïve and primed hESC

Human stem cell-associated retroviral sequences (SCARS)

Identify genes regulated by HSGRS

Identify genes regulated by SCARS

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Comparative gene set enrichment (CGSE) analyses of gene and protein expression profiles

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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

- **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 NEUROGENSIS AND CORTICOGENESIS**

59K human-specific regulatory sequences

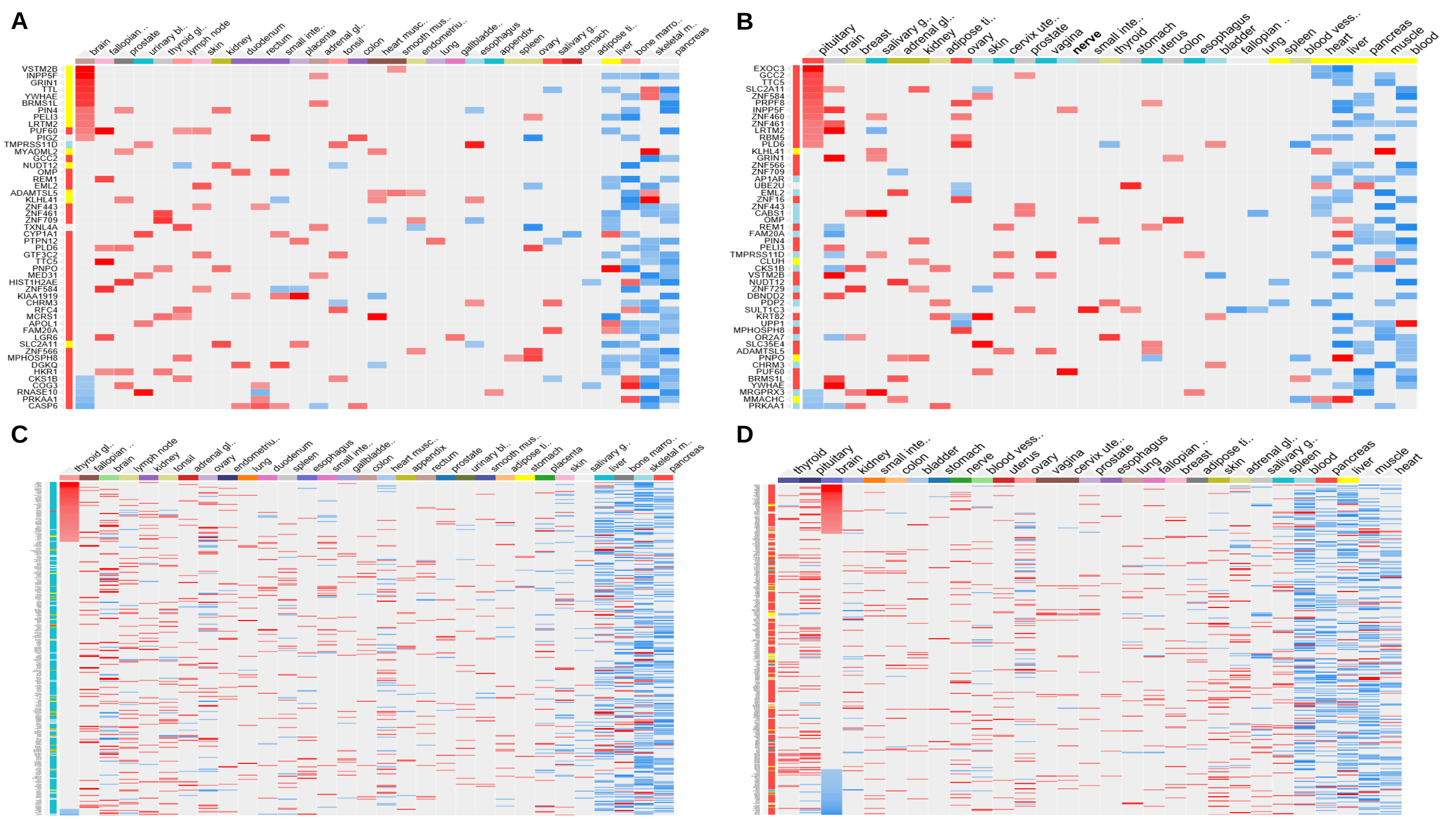
Analysis of genes representing the putative regulatory targets of human-specific regulatory sequences

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.


A
brain
fallopian ..
prostate
urinary bl..
thyroid gl..
lymph node
skin
kidney
duodenum
rectum
small inte..
placenta
adrenal gl..
tonsil
colon
heart musc..
smooth mus..
endometriu..
lung
gallbladde..
esophagus
appendix
spleen
ovary
salivary g..
stomach
adipose ti..
liver
bone marro..
skeletal m..
pancreas
VSTM2B
INPP5F
GRIN1
TTL
YWHAE
BRMS1L
PIN4
PELI3
LRTM2
PUF60
PIGZ
TMPRSS11D
MYADML2
GCC2
NUDT12
OMP
REM1
EML2
ADAMTSL5
KLHL41
ZNF443
ZNF461
ZNF709
TXNL4A
CYP1A1
PTPN12
PLD6
GTF3C2
TTC5
PNPO
MED31
HIST1H2AE
ZNF584
KIAA1919
CHRM3
RFC4
MCRS1
APOL1
FAM20A
LGR6
SLC2A11
ZNF566
MPHOSPH8
DGKQ
HKR1
CKS1B
COG3
RNASE10
PRKAA1
CASP6
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pituitary
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vagina
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small inte..
thyroid
stomach
uterus
colon
esophagus
bladder
fallopian ..
lung
spleen
blood vess..
heart
liver
pancreas
muscle
blood
EXOC3
GCC2
TTC5
SLC2A11
ZNF584
PRPF8
INPP5F
ZNF460
ZNF461
LRTM2
RBM5
PLD6
KLHL41
GRIN1
ZNF566
ZNF709
AP1AR
UBE2U
EML2
ZNF16
ZNF443
CABS1
OMP
REM1
FAM20A
PIN4
PELI3
TMPRSS11D
CLUH
CKS1B
VSTM2B
NUDT12
ZNF729
DBNDD2
PDP2
SULT1C3
KRT82
UPP1
MPHOSPH8
OR2A7
SLC35E4
ADAMTSL5
PNPO
CHRM3
PUF60
BRMS1L
YWHAE
MRGPRX3
MMACHC
PRKAA1
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thyroid gl..
fallopian ..
brain
lymph node
kidney
tonsil
adrenal gl..
ovary
endometriu..
lung
duodenum
spleen
esophagus
small inte..
gallbladde..
colon
heart musc..
appendix
rectum
prostate
urinary bl..
smooth mus..
adipose ti..
stomach
placenta
skin
salivary g..
liver
bone marro..
skeletal m..
pancreas
D
thyroid
pituitary
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kidney
small inte..
colon
bladder
stomach
nerve
blood vess..
uterus
ovary
vagina
cervix ute..
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esophagus
lung
fallopian ..
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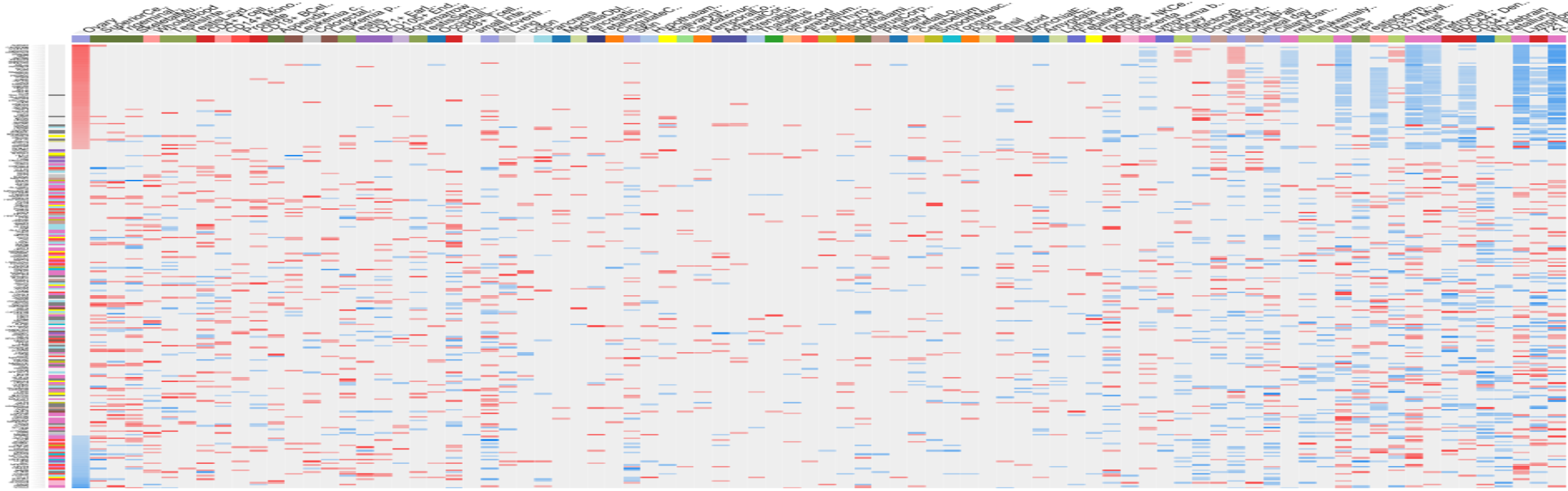
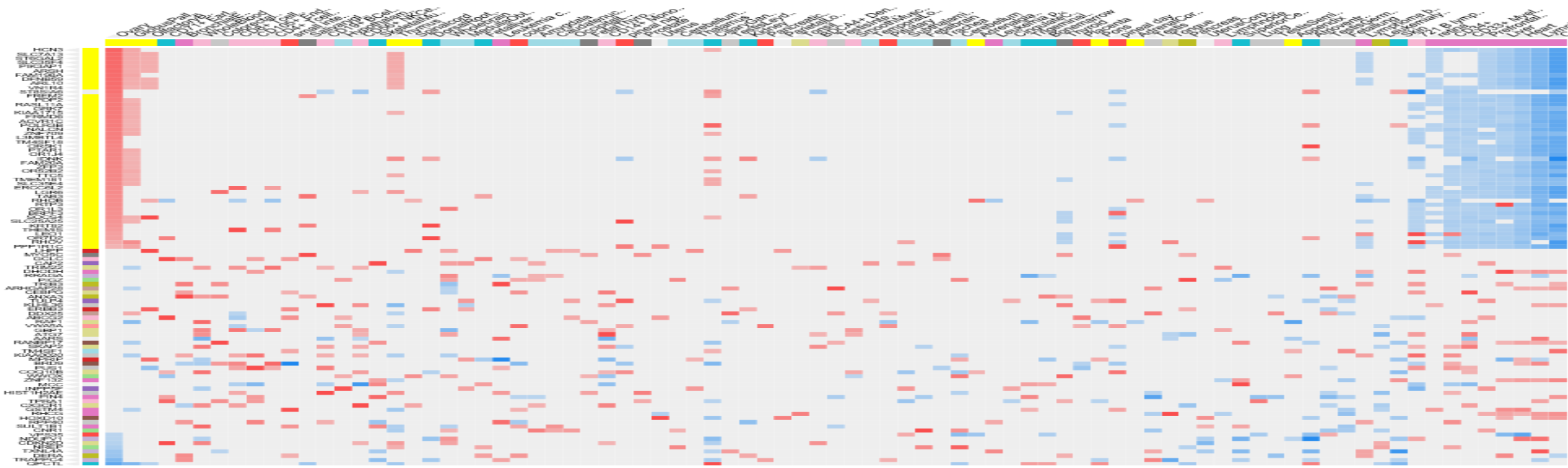
adipose ti..
skin
adrenal gl..
salivary g..
spleen
blood
pancreas
liver
muscle
heart

E

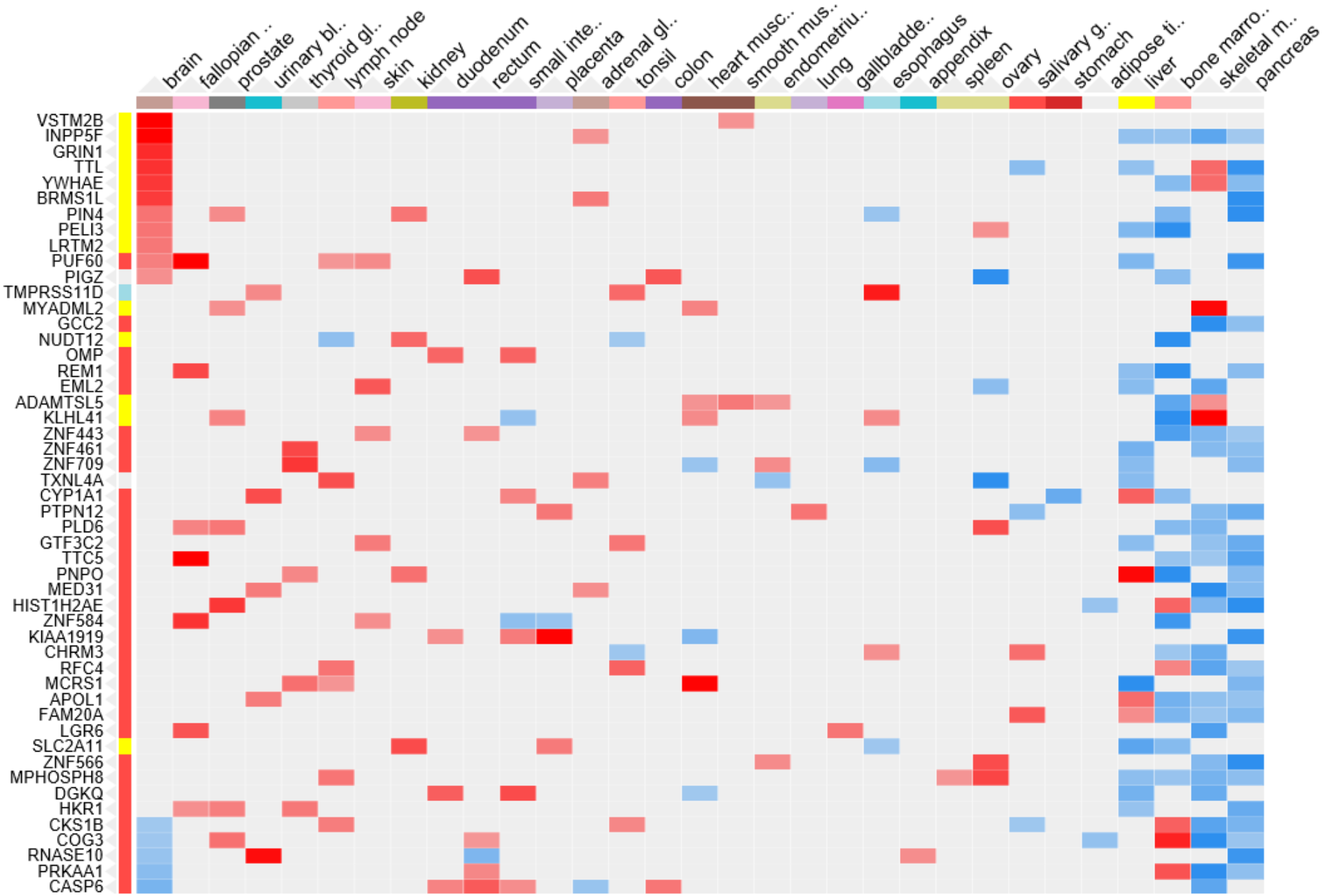


F



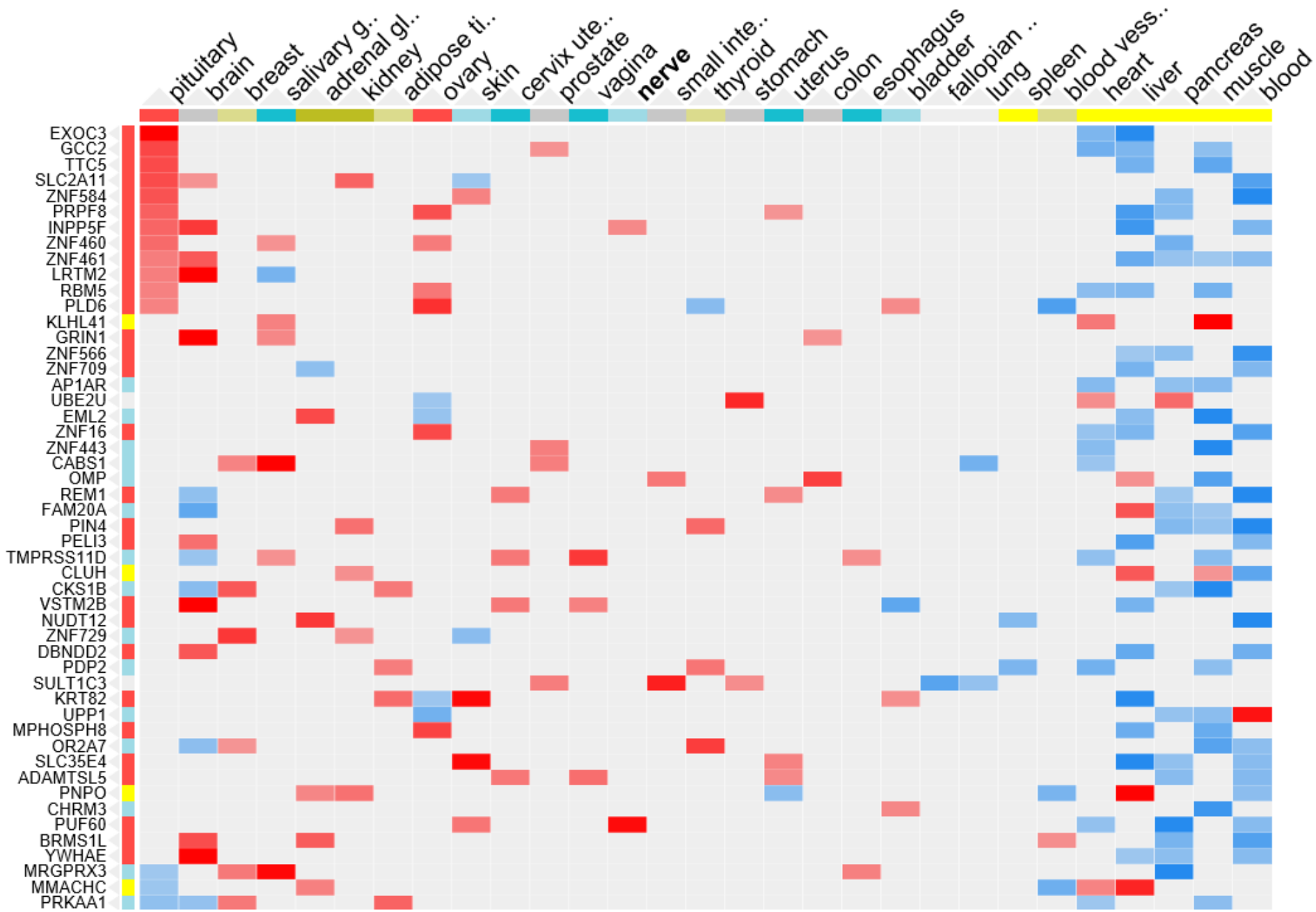
HeatMaps Visualization of expression profiles of mRNAs encoded by 90 genes associated with human-specific genomic regulatory loci derived from transcriptionally active in human DLPFC transposable elements

HPA Human Tissues' Gene Expression Profiles



hsTE_DLPFC_1kb
90 genes
top 50 targets

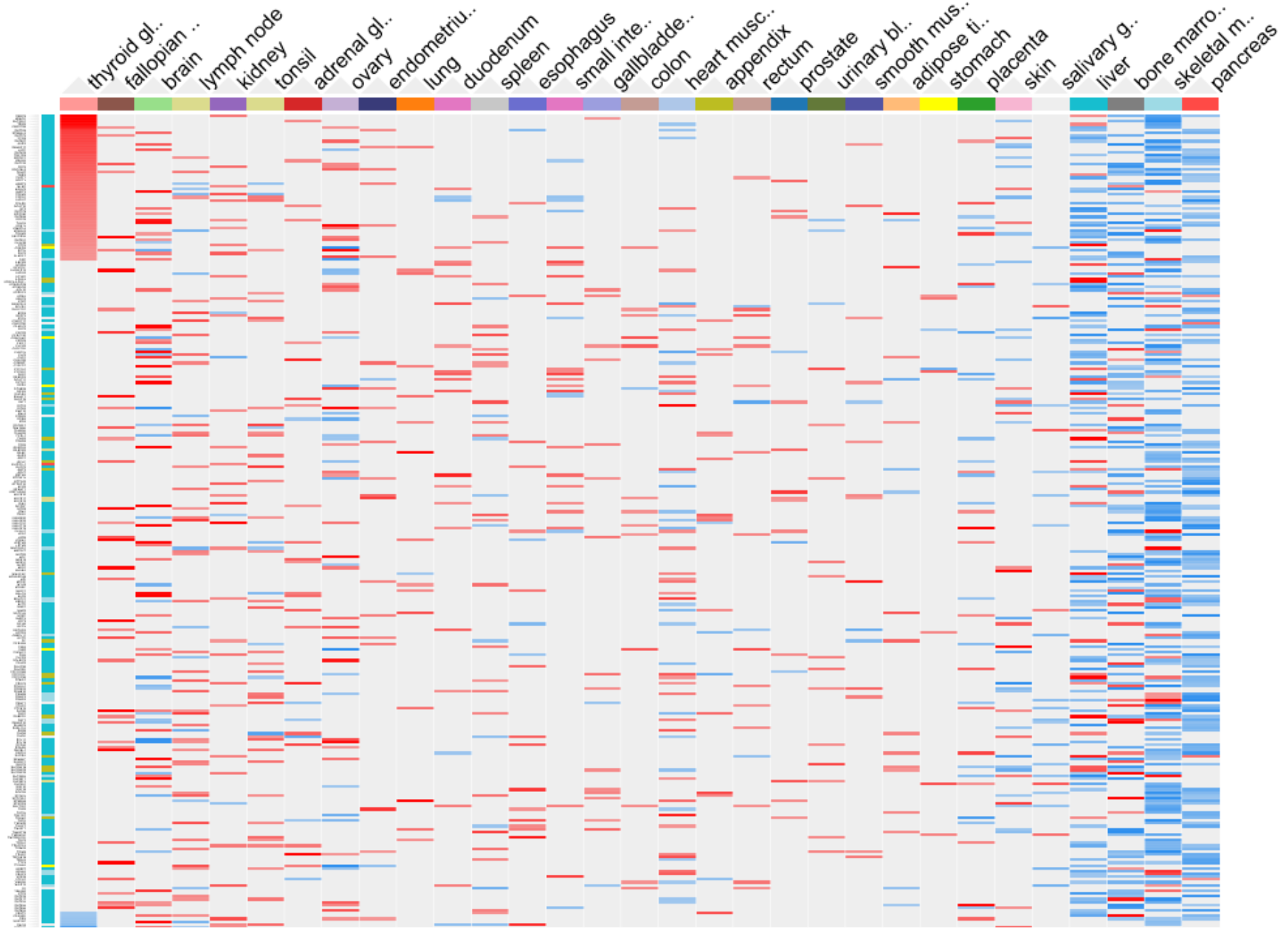
GTEx Human Tissues' Gene Expression Profiles



hsTE_DLPFC_1kb
90 genes
top 50 targets

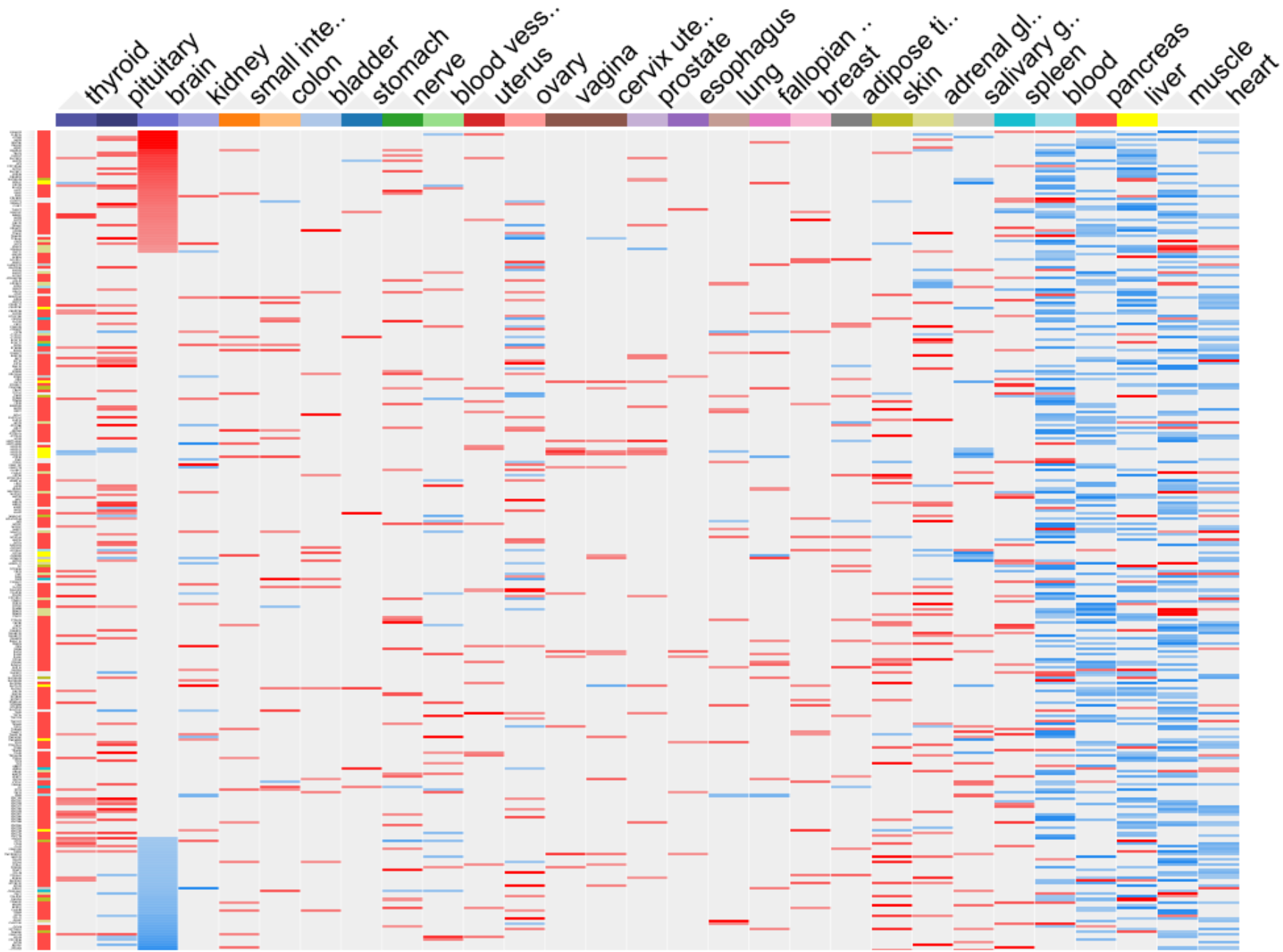
HeatMaps Visualization of expression profiles of mRNAs encoded by 326 genes associated with human-specific genomic regulatory loci derived from fixed human-specific insertions

HPA Human Tissues' Gene Expression Profiles



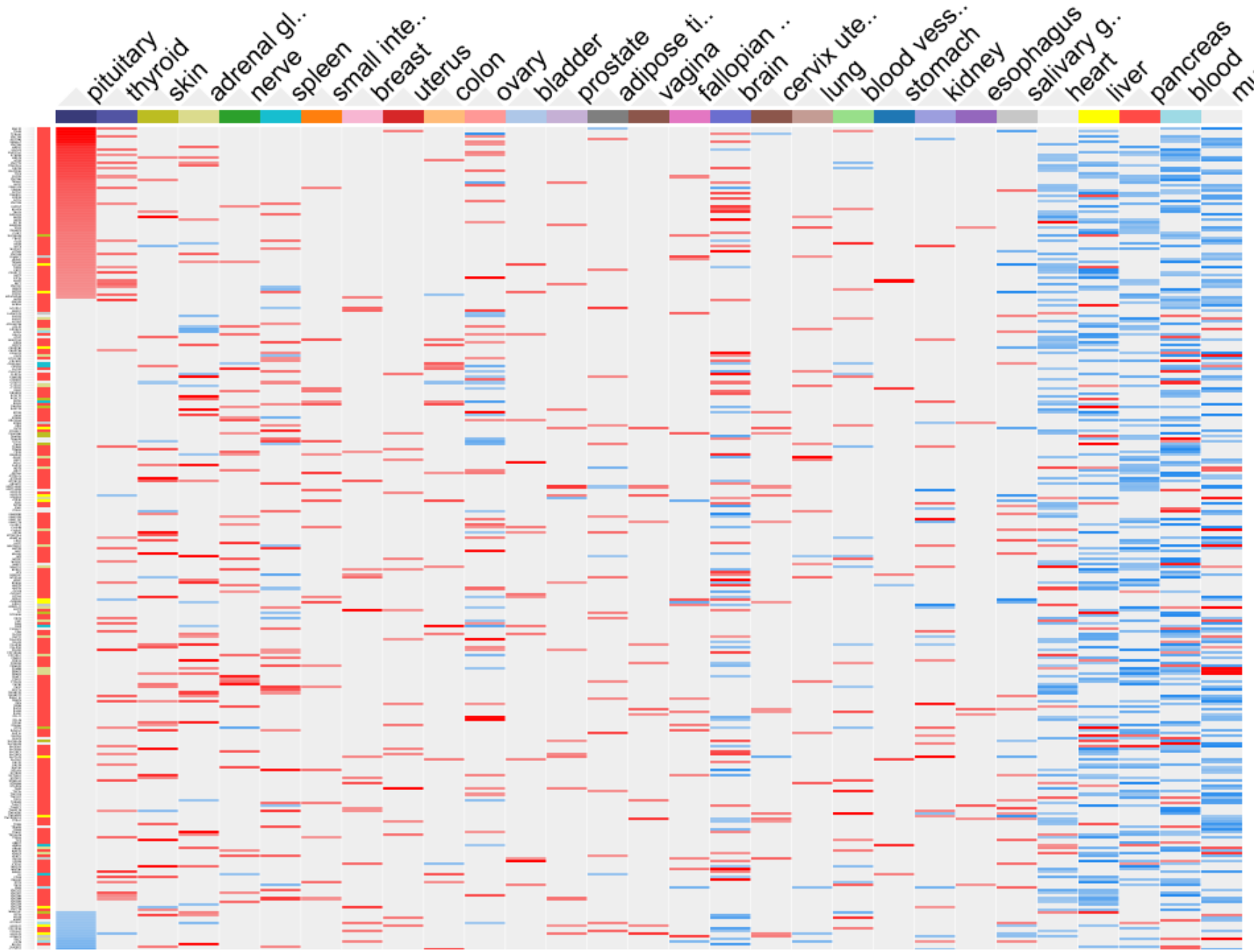
Fixed_hs_Insertions
1kb_326 genes
All targets

GTEx Human Tissues' Gene Expression Profiles



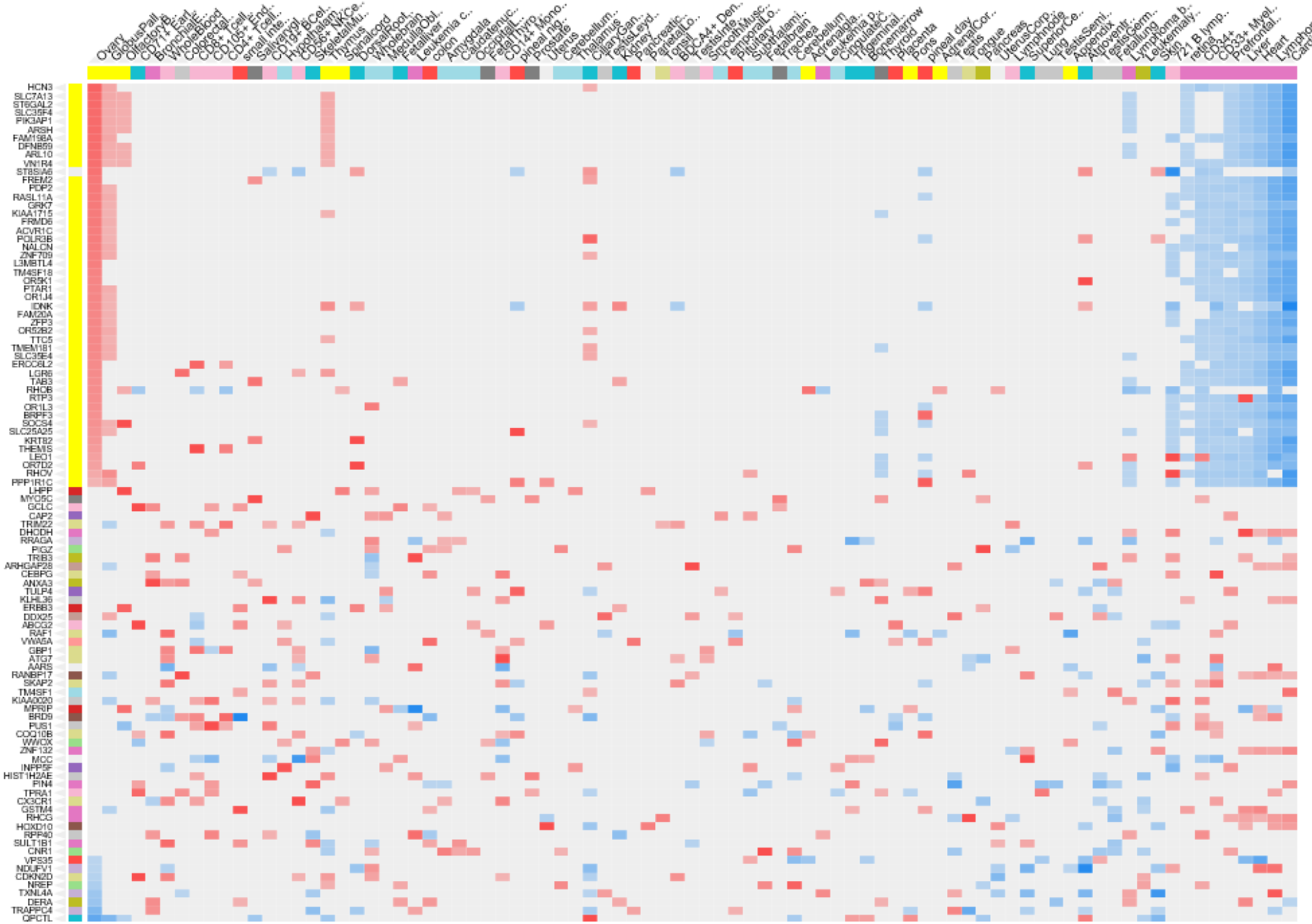
Fixed_hs_Insertions
1kb_326 genes
All targets

GTEx Human Tissues' Gene Expression Profiles



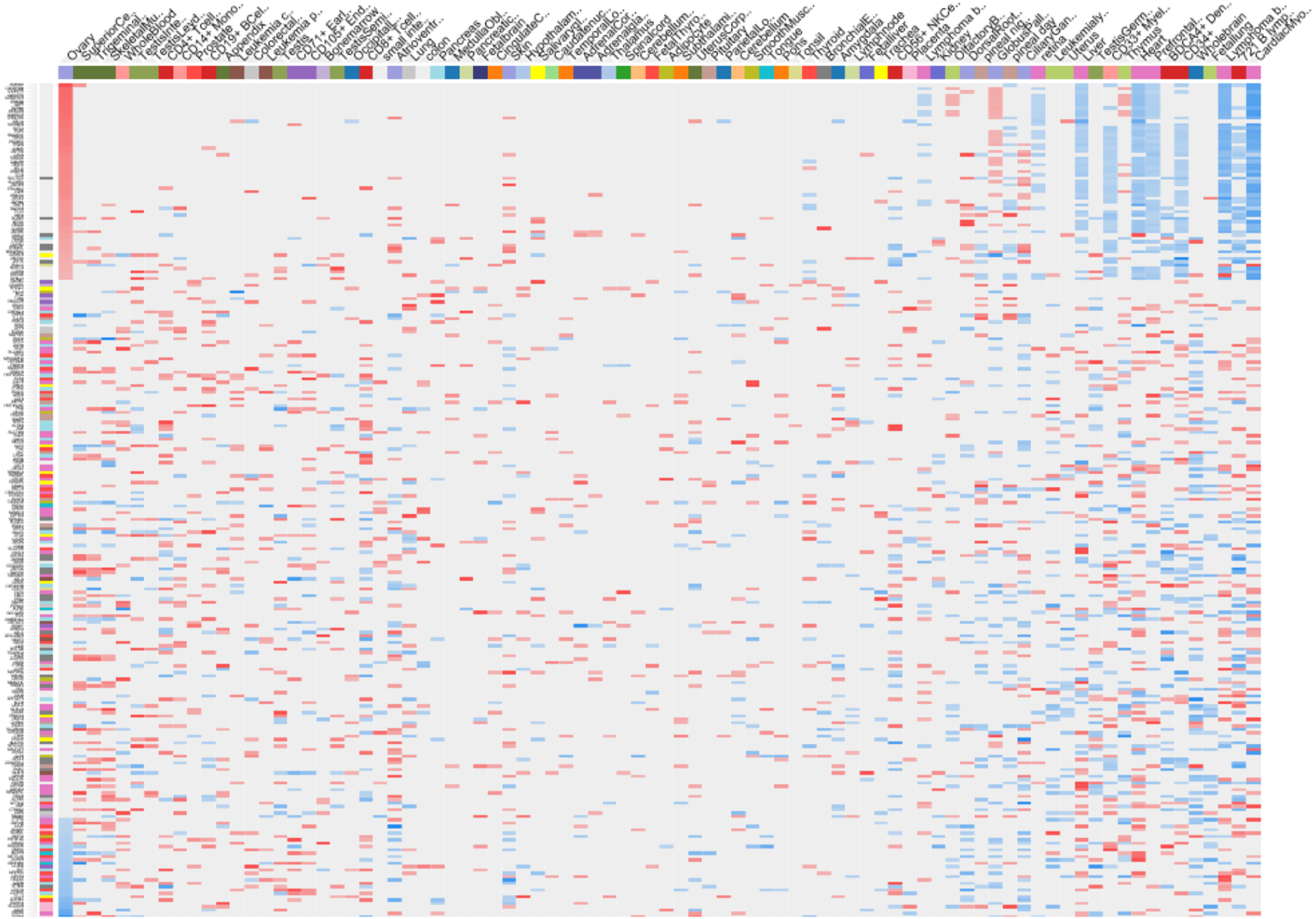
Fixed_hs_Insertions
1kb_326 genes
All targets

BioGPS Human Cell Type and Tissue Gene Expression Profiles



Fixed_hs_Insertions
1kb_326 genes
Top 100 targets

BioGPS Human Cell Type and Tissue Gene Expression Profiles



hsTE_DLPFC_10kb
336 genes
top 250 targets

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

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 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

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

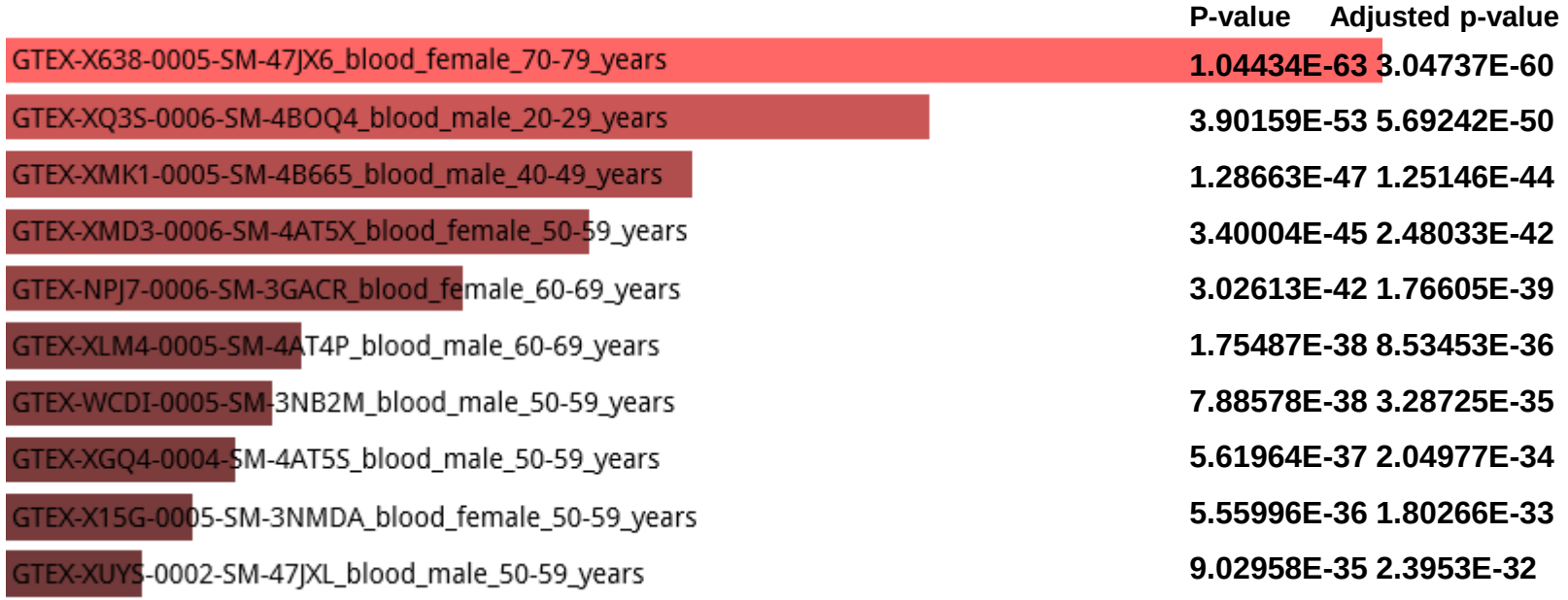
	P-value	Adjusted p-value
GTEX-T2IS-0011-R5A-SM-32QP4_brain_female_20-29_years	7.39447E-21	2.15771E-17
GTEX-QDT8-0011-R10A-SM-32PKG_brain_female_30-39_years	5.34561E-20	7.79924E-17
GTEX-OIZI-0008-SM-2XCFD_skin_male_40-49_years	6.25612E-19	6.08512E-16
GTEX-SNMC-1526-SM-2XCFN_blood vessel_male_20-29_years	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

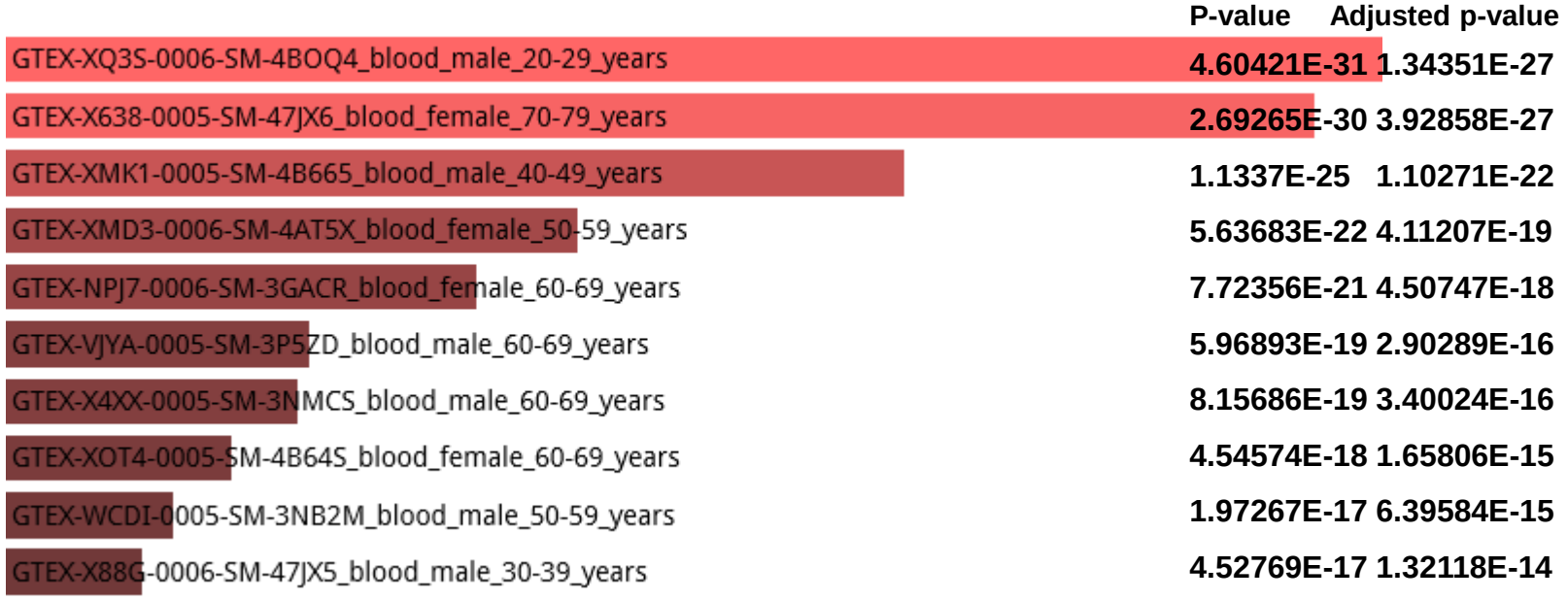
	P-value	Adjusted p-value
GTEX-SNMC-1526-SM-2XCFN_blood vessel_male_20-29_years	1.00794E-41	2.94116E-38
GTEX-T2IS-0011-R5A-SM-32QP4_brain_female_20-29_years	1.1941E-40	1.74219E-37
GTEX-QDT8-0011-R10A-SM-32PKG_brain_female_30-39_years	1.21874E-39	1.18543E-36
GTEX-OIZI-0008-SM-2XCFD_skin_male_40-49_years	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



GTEx Tissue Sample Gene Expression Profiles down 5986 genes



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

	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

Jensen DISEASES 4051 genes

	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

	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

Disease Perturbations from GEO down 4051 genes

	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

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 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

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

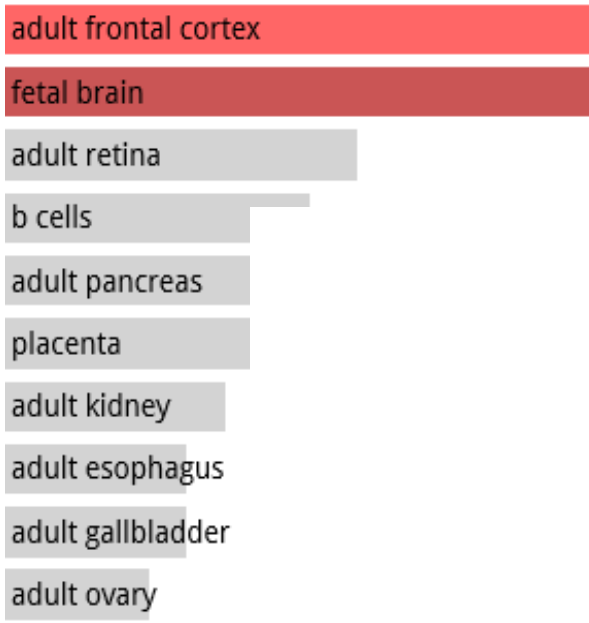
	P-value	Adjusted p-value
GTEX-XQ3S-0006-SM-4BOQ4_blood_male_20-29_years	1.57265E-10	4.58427E-07
GTEX-XOT4-0005-SM-4B64S_blood_female_60-69_years	7.49373E-09	1.09221E-05
GTEX-WHSE-0006-SM-3NMBW_blood_male_20-29_years	1.18638E-08	1.15277E-05
GTEX-WZTO-0006-SM-3NM9T_blood_male_40-49_years	2.0799E-07	0.000152
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

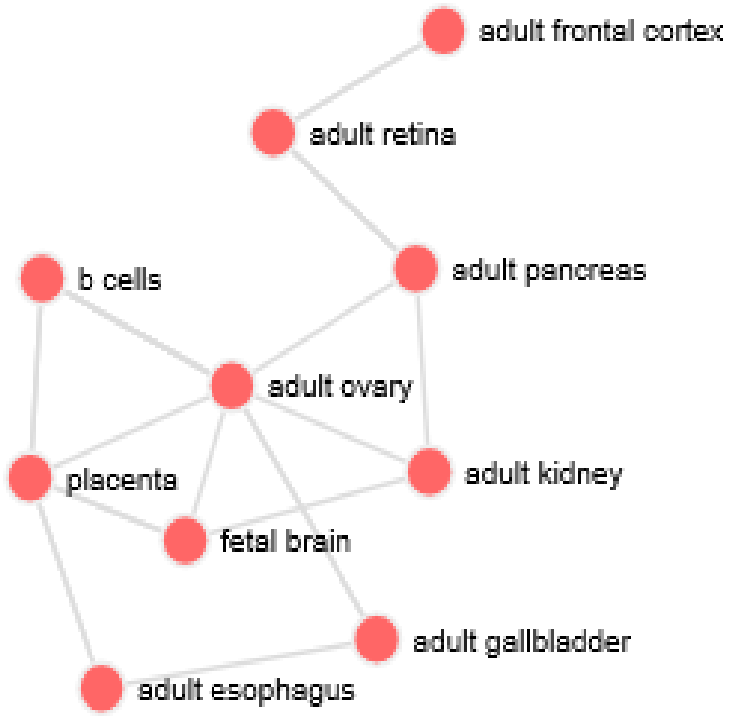
4,637 human-specific TE-encoded loci expressed in human DLPFC

Tissue Protein Expression from Human Proteome Map 4051 genes



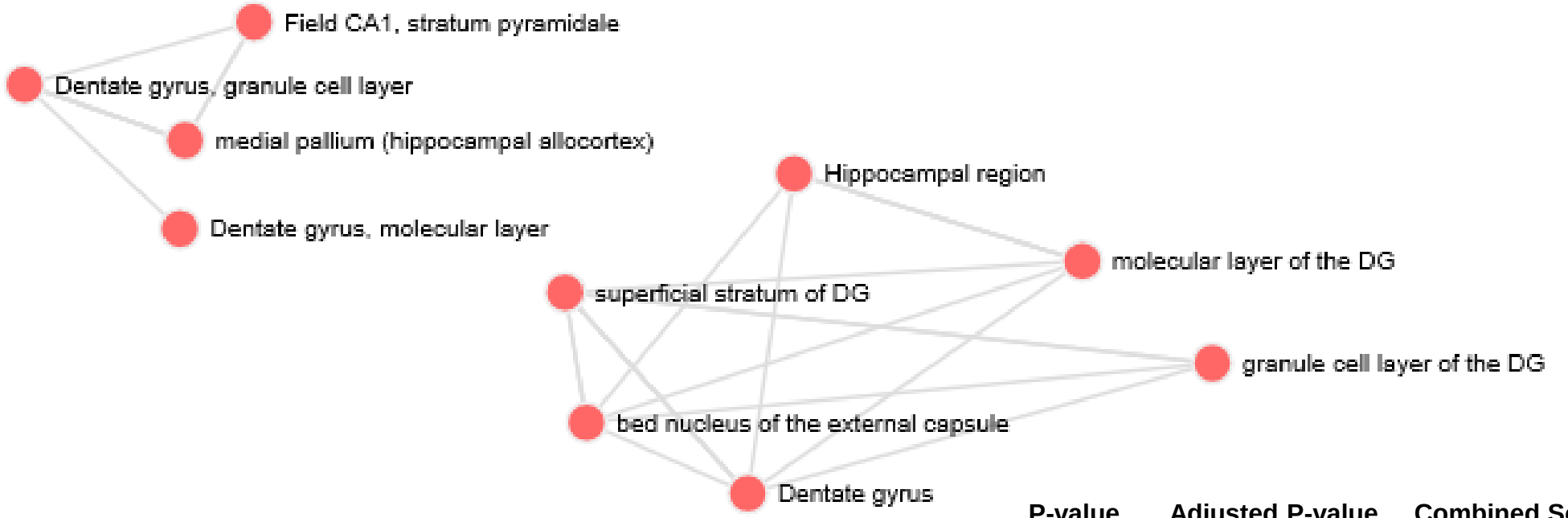
p-value: 0.00006954 **Adjusted p-value: 0.002086**

p-value: 0.001310; **Adjusted p-value: 0.01966**



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

Allen Brain Atlas up 4051 genes

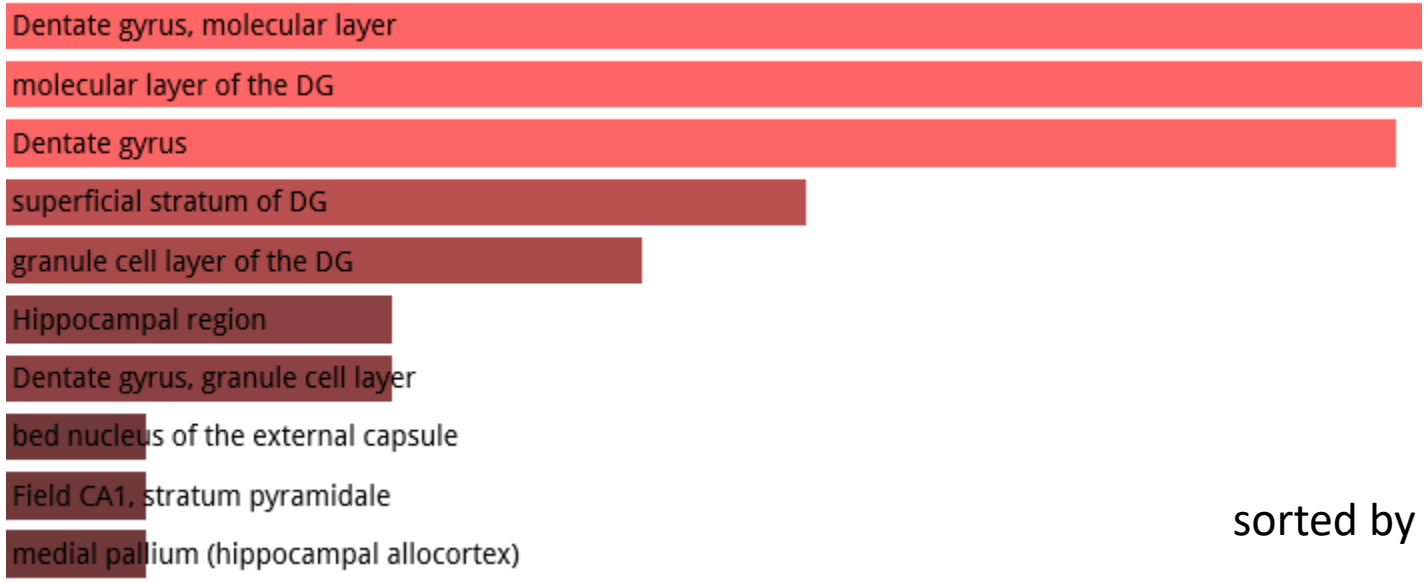


	P-value	Adjusted P-value	Combined Score
Dentate gyrus	2.06994E-09	1.5124E-06	36.78
Dentate gyrus, molecular layer	1.91586E-09	1.5124E-06	35.25
granule cell layer of the DG	1.83231E-08	8.0328E-06	33.31
molecular layer of the DG	1.91586E-09	1.5124E-06	32.40
Hippocampal region	3.77717E-08	1.1828E-05	31.40
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



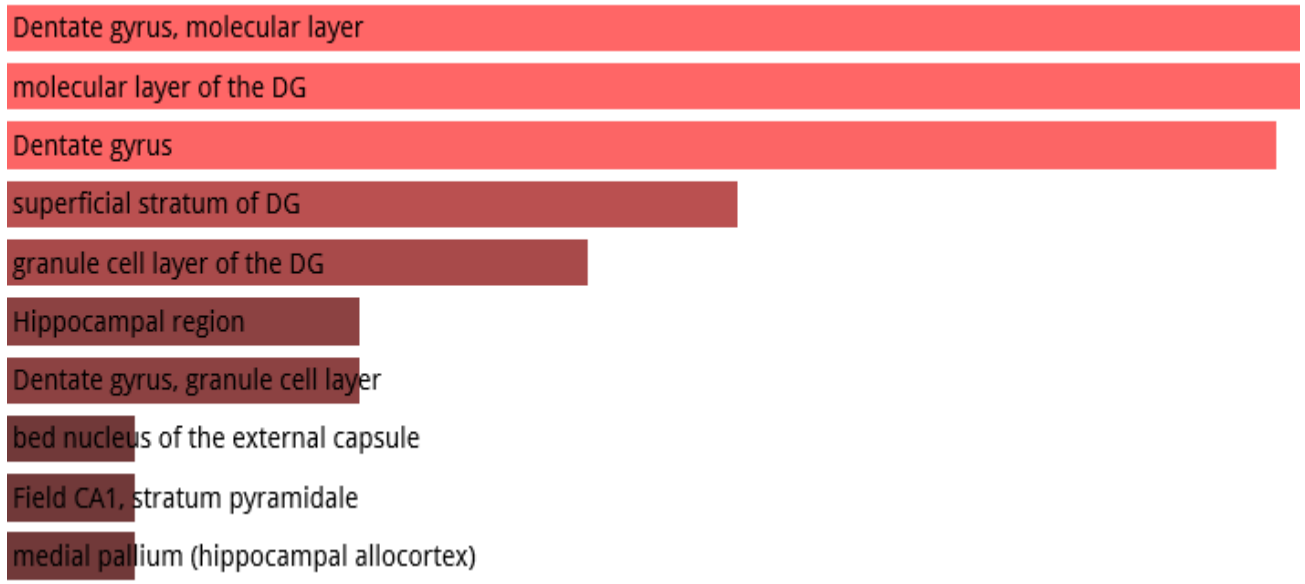
sorted by p-value ranking

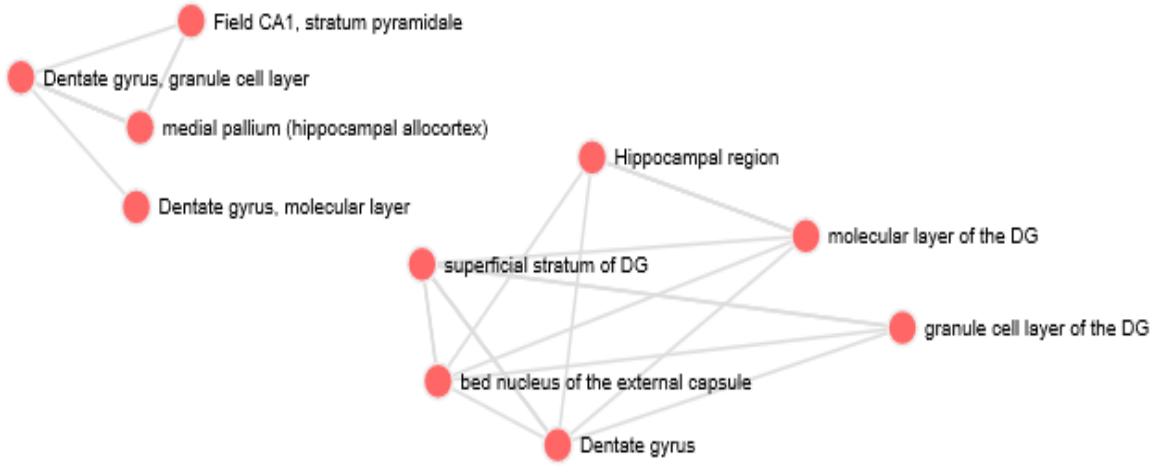
TOP 15 CATEGORIES

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

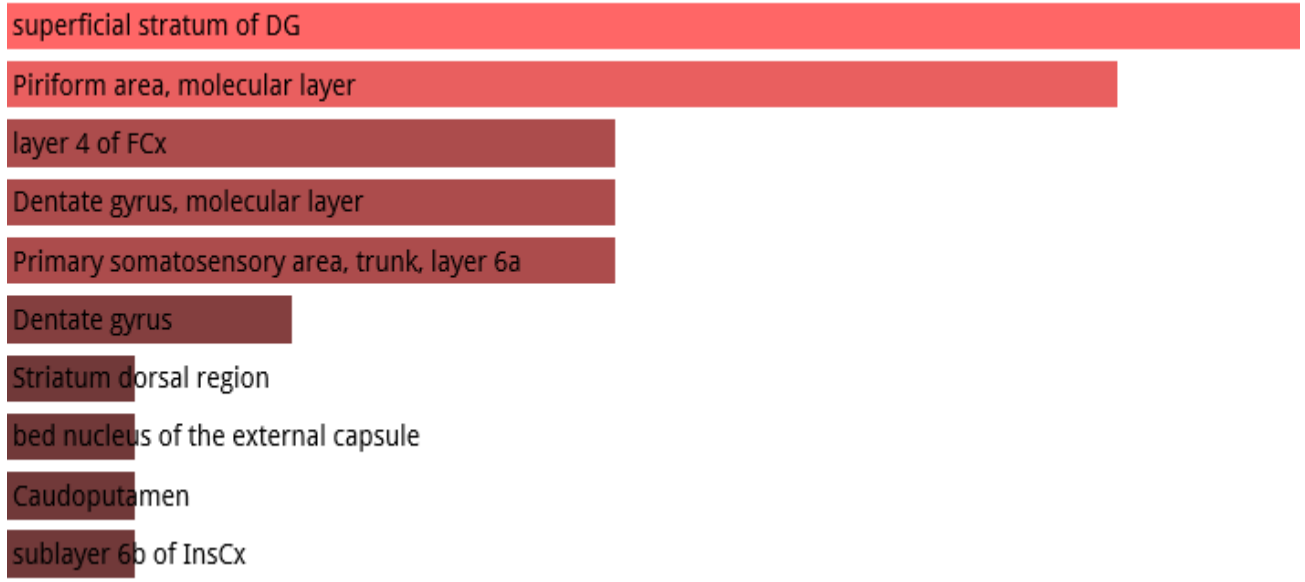
4,637 human-specific TE-encoded loci expressed in human DLPFC

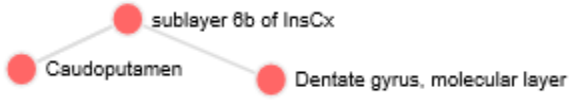
Allen Brain Atlas up 4051 genes

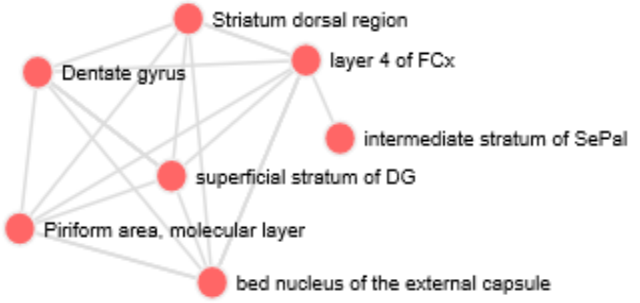




Allen Brain Atlas up 2323 genes







Comparisons of putative regulatory target genes associated with

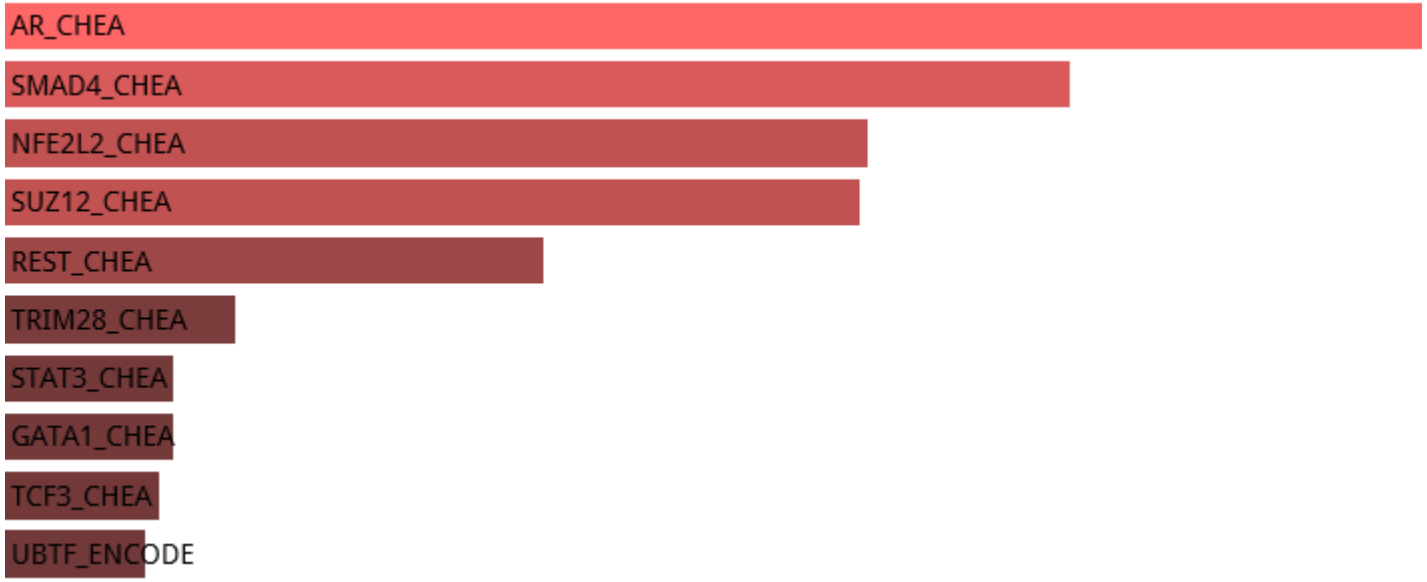
11,878 fixed human-specific insertions

and

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

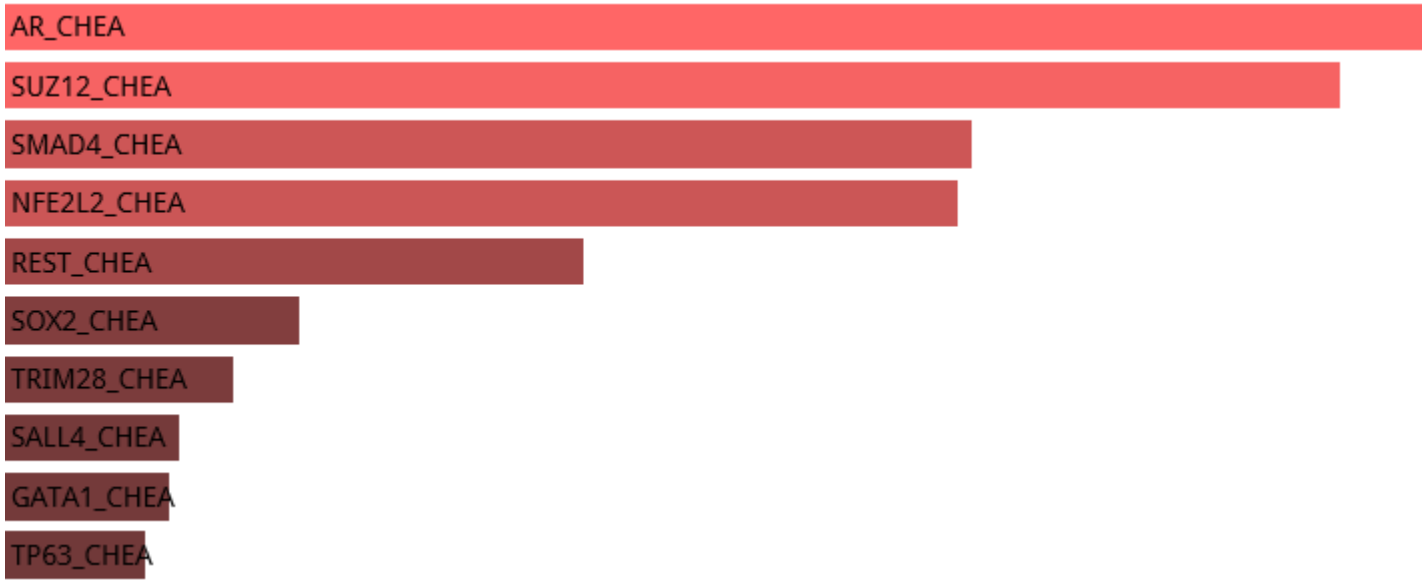
4,637 human-specific TE-encoded loci expressed in human DLPFC

ENCODE and ChEA Consensus TFs from ChIP-X 4051 genes



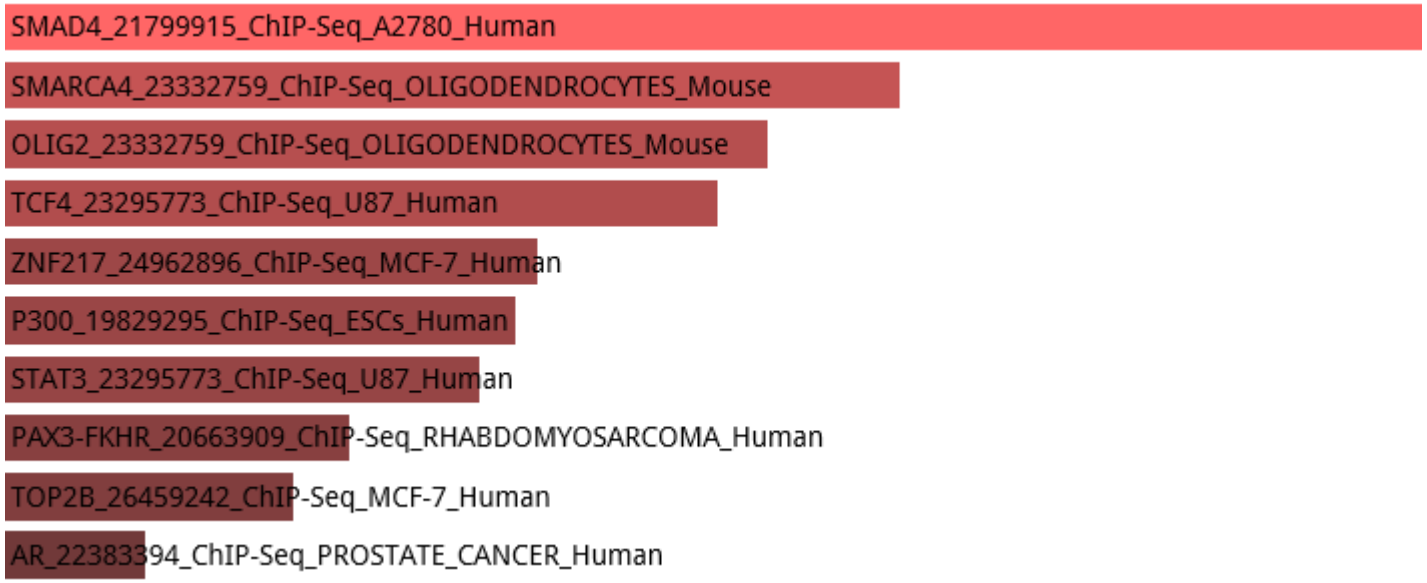
11,878 fixed human-specific insertions

ENCODE and ChEA Consensus TFs from ChIP-X 7979 genes



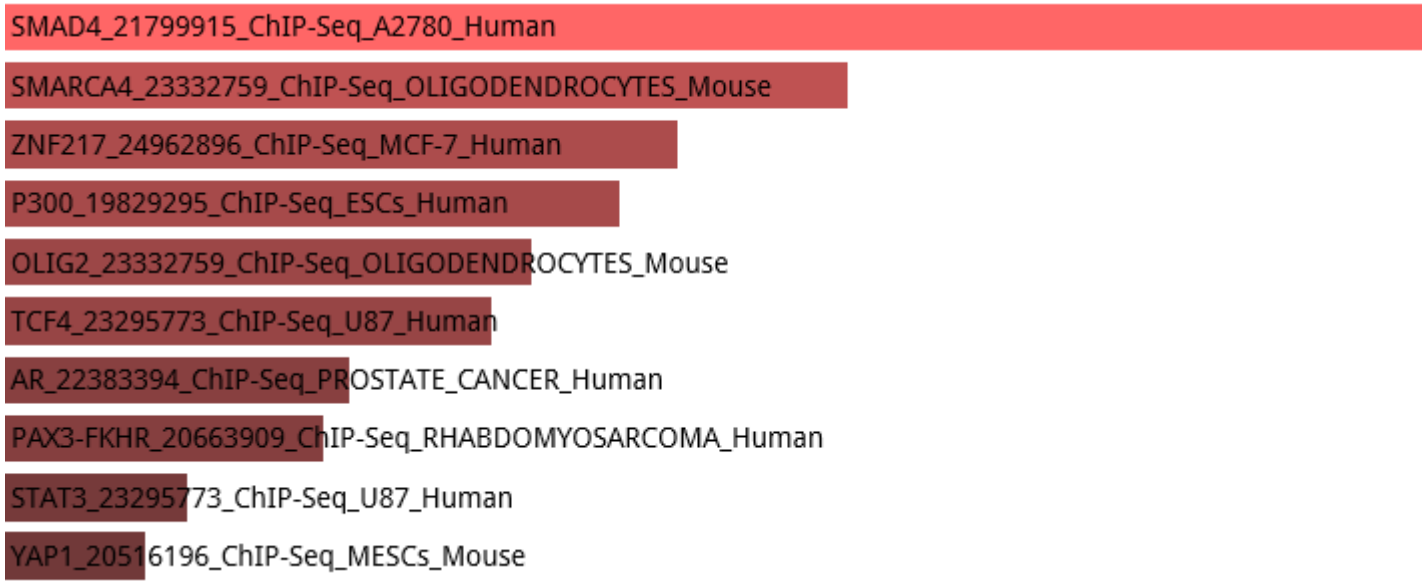
4,637 human-specific TE-encoded loci expressed in human DLPFC

ChEA 2016 4051 genes



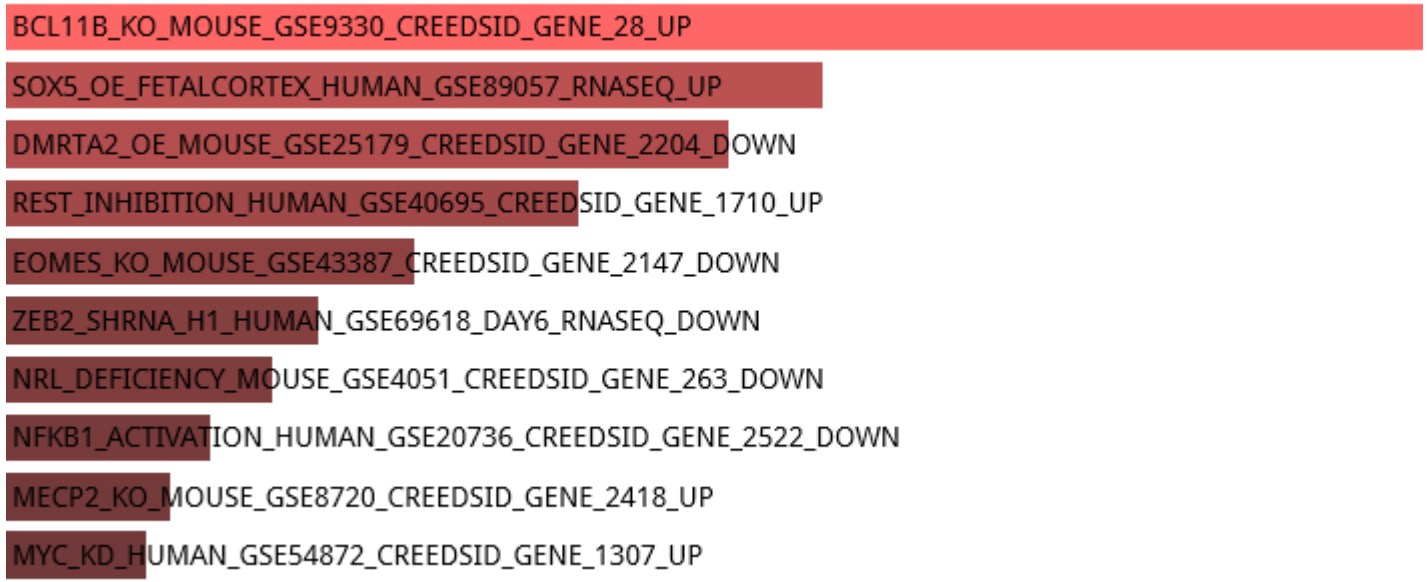
11,878 fixed human-specific insertions

ChEA 2016 7979 genes



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

TF Perturbations Followed by Expression 4051 genes



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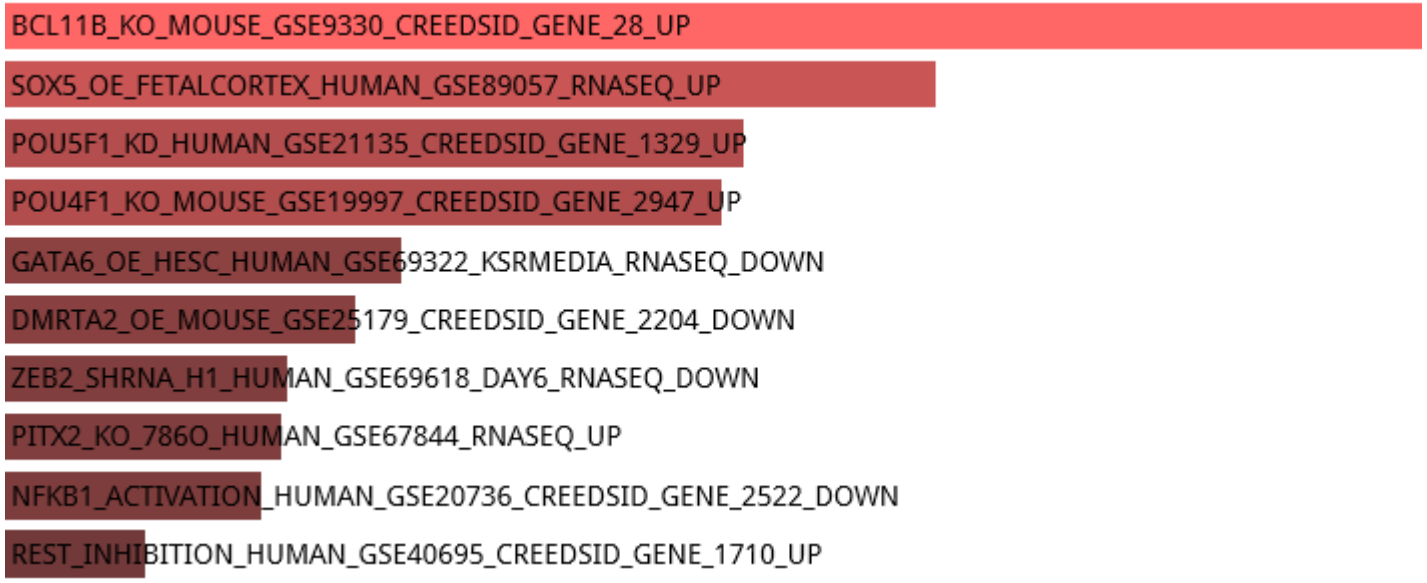
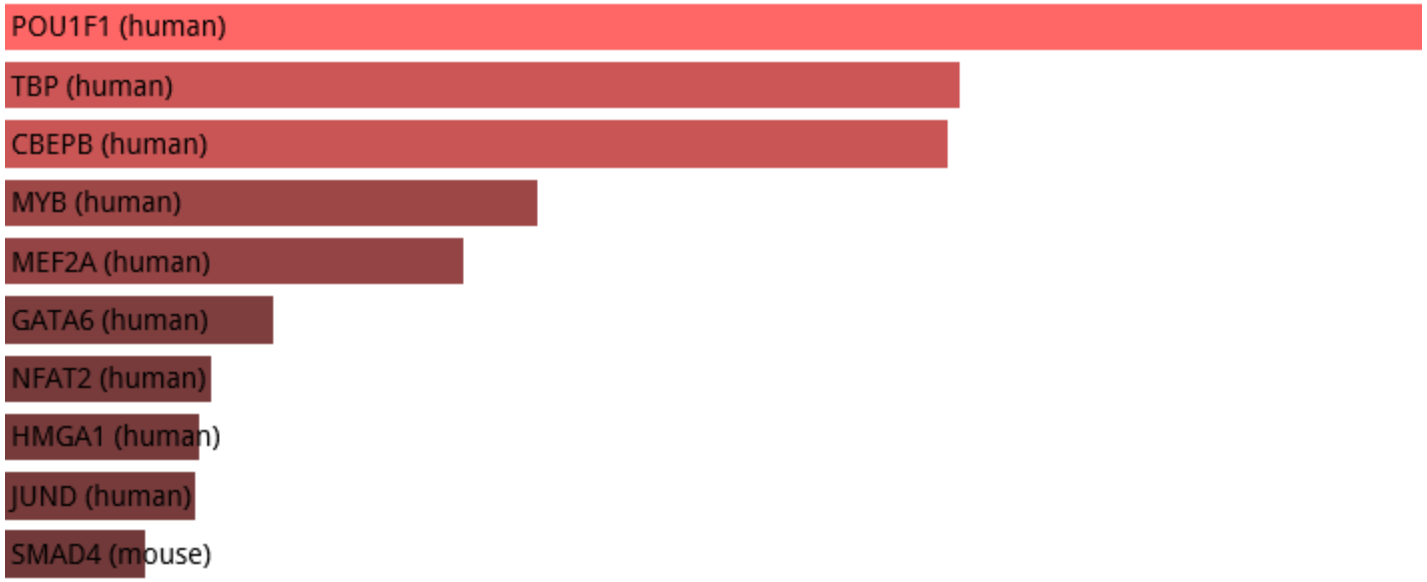
TF Perturbations Followed by Expression 7979 genes



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

TRANSFAC and JASPAR PWMs 4051 genes



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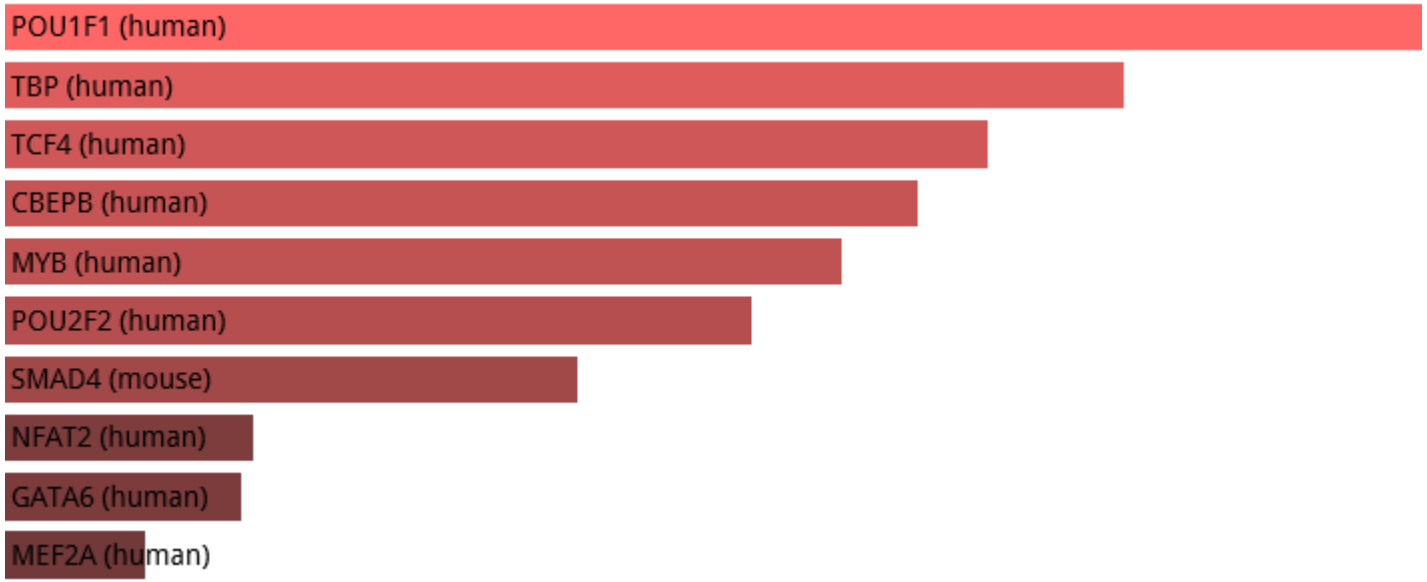
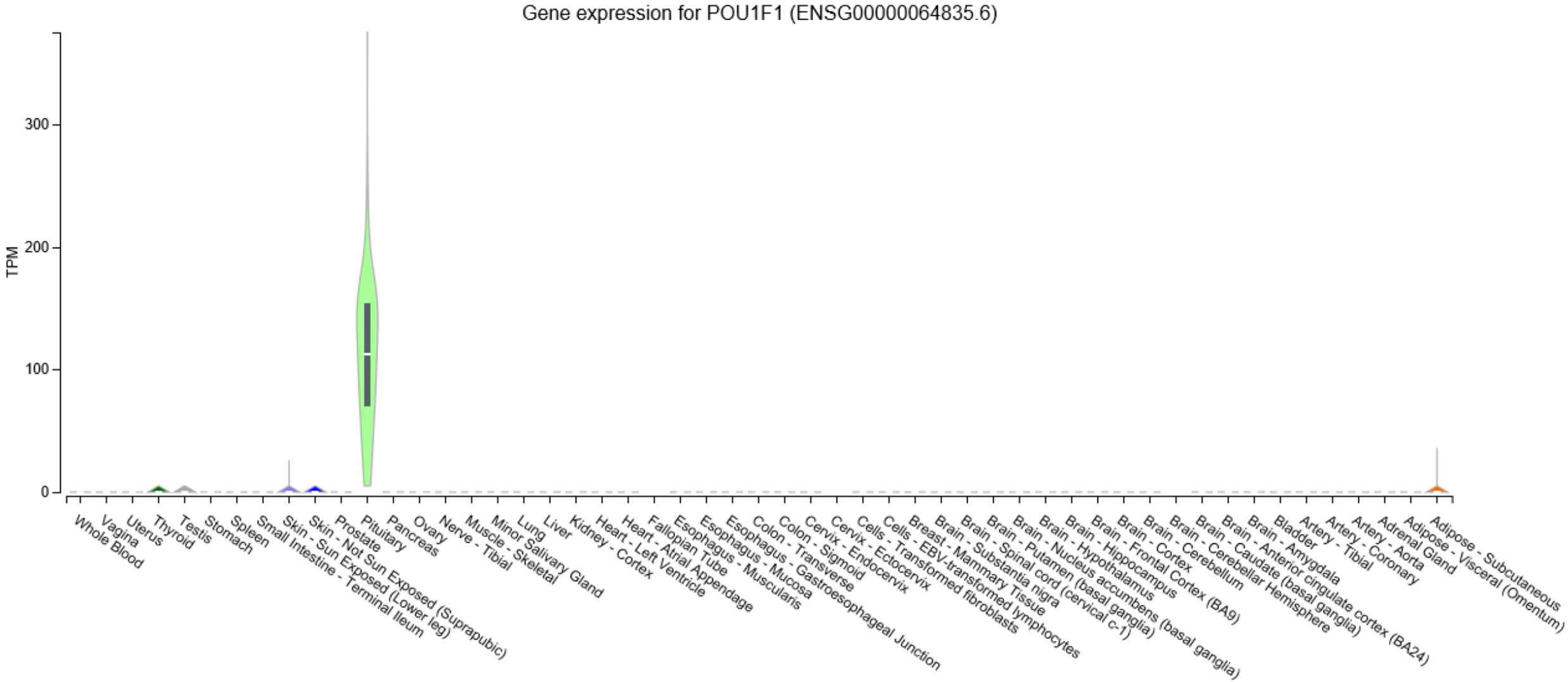
TRANSFAC and JASPAR PWMs 7979 genes



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



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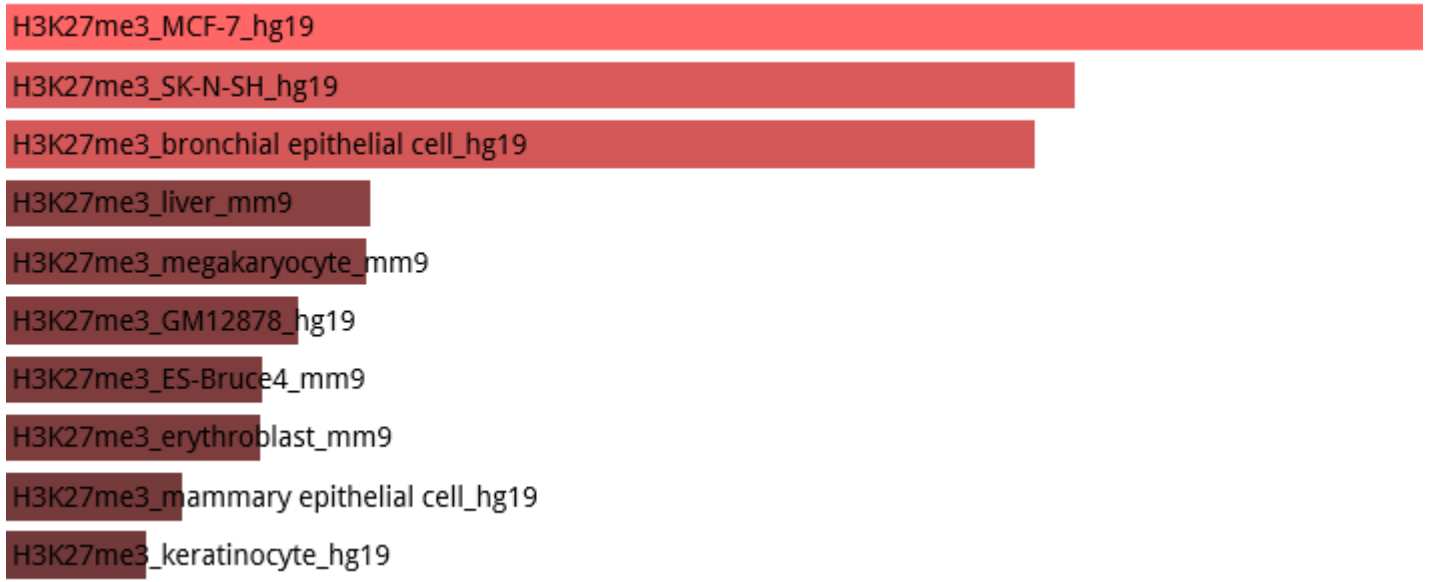
ENCODE Histone Modifications 2015 4051 genes



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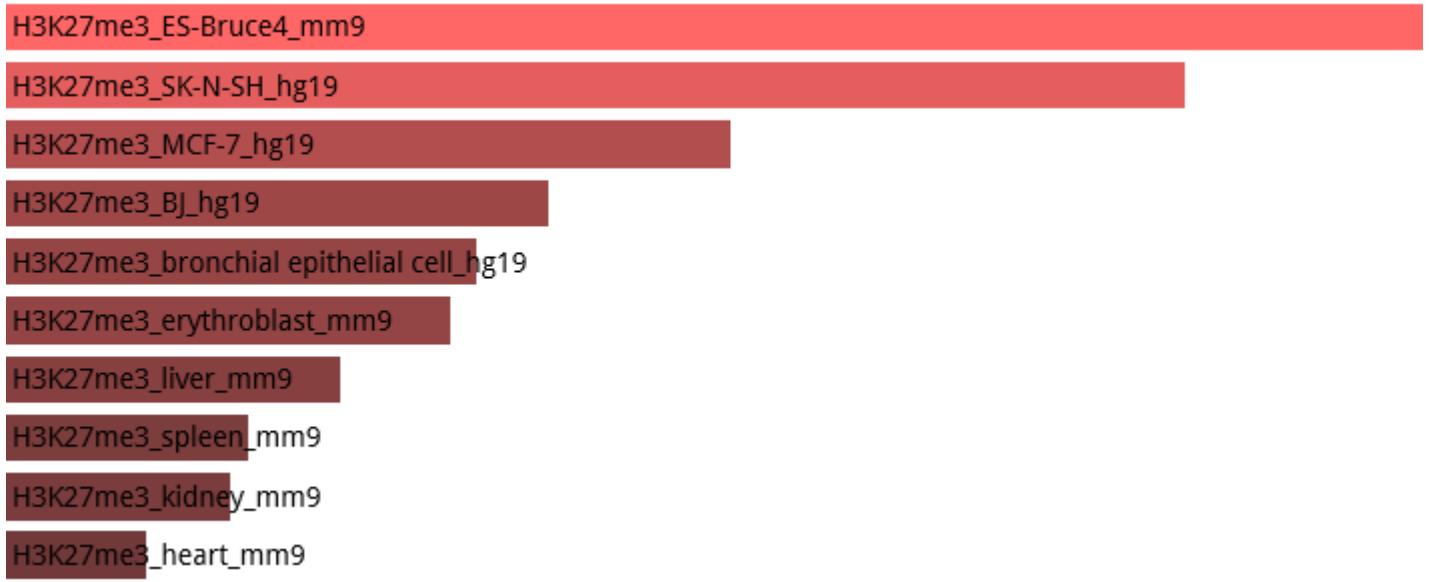
ENCODE Histone Modifications 2015 7979 genes genes



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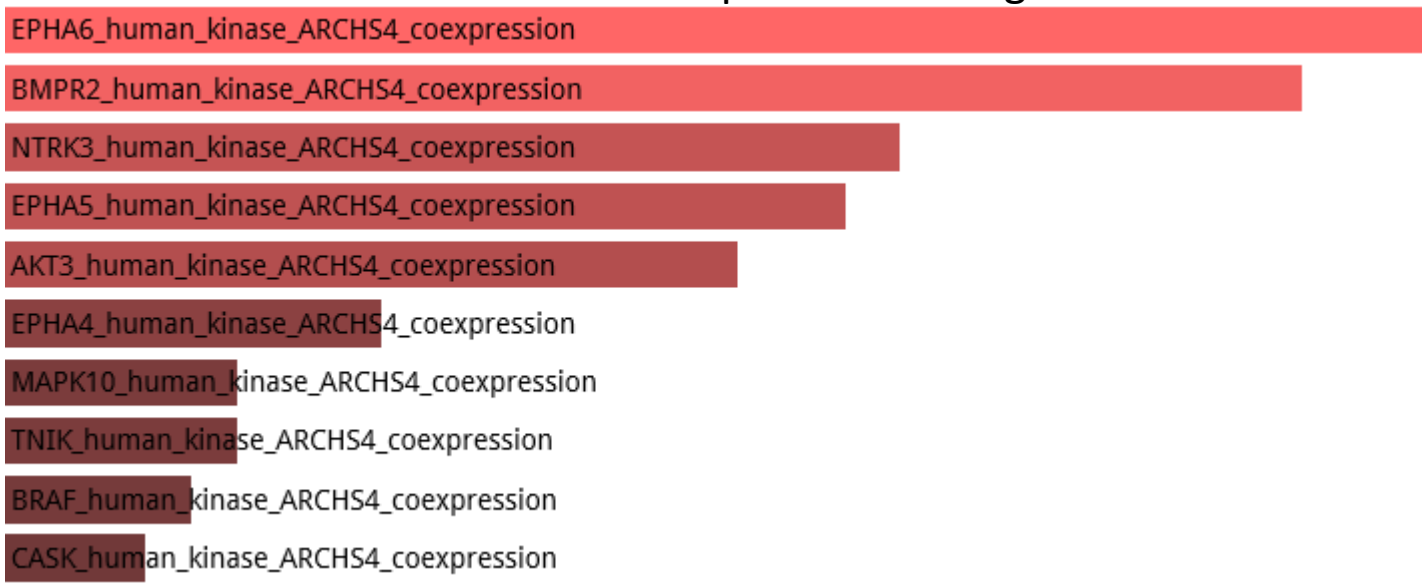
ARCHS4 Kinases Co-expression 4051 genes



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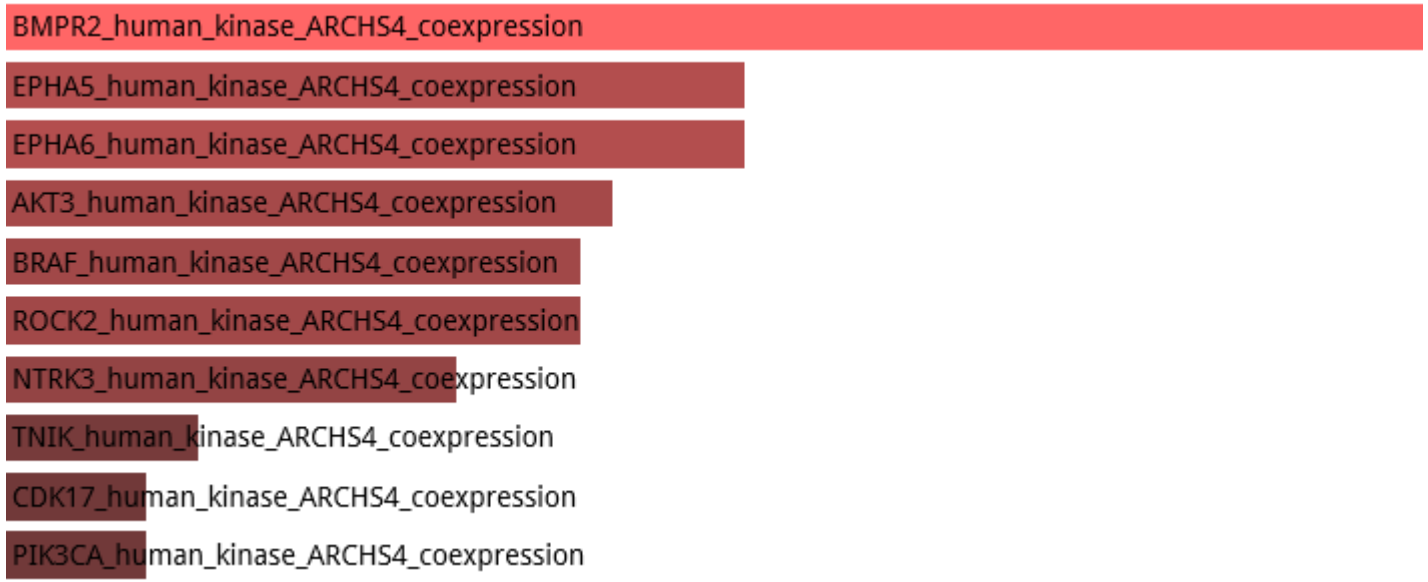

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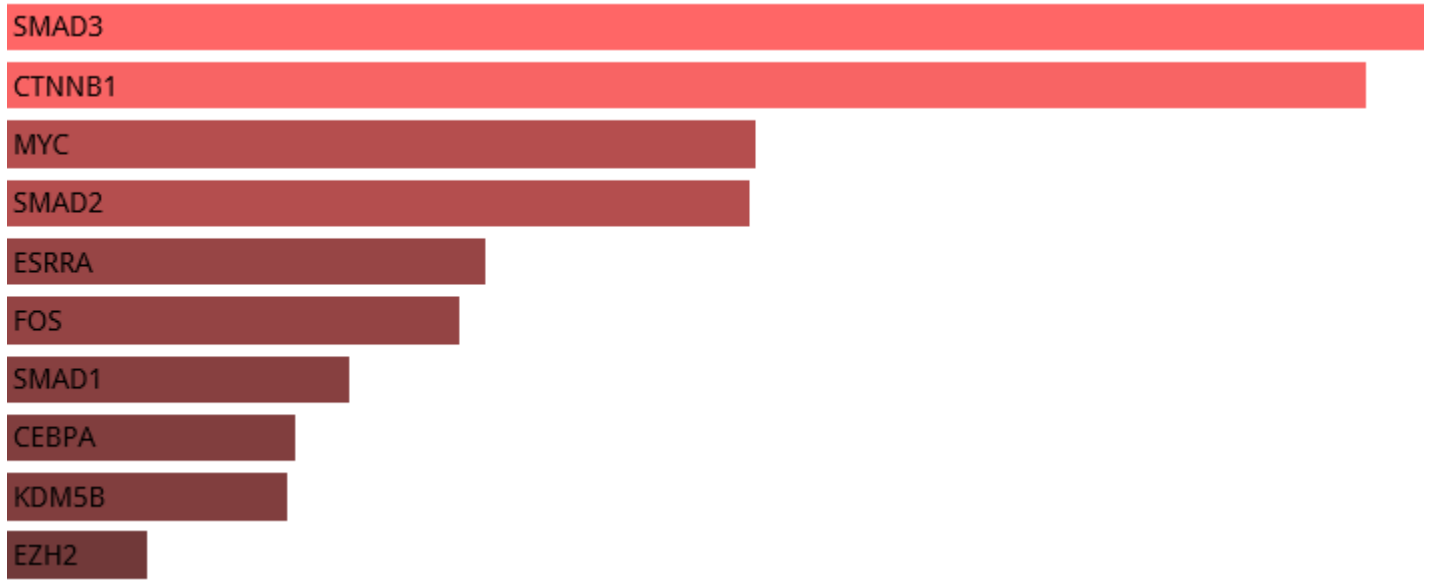
Transcription Factor PPIs 4051 genes



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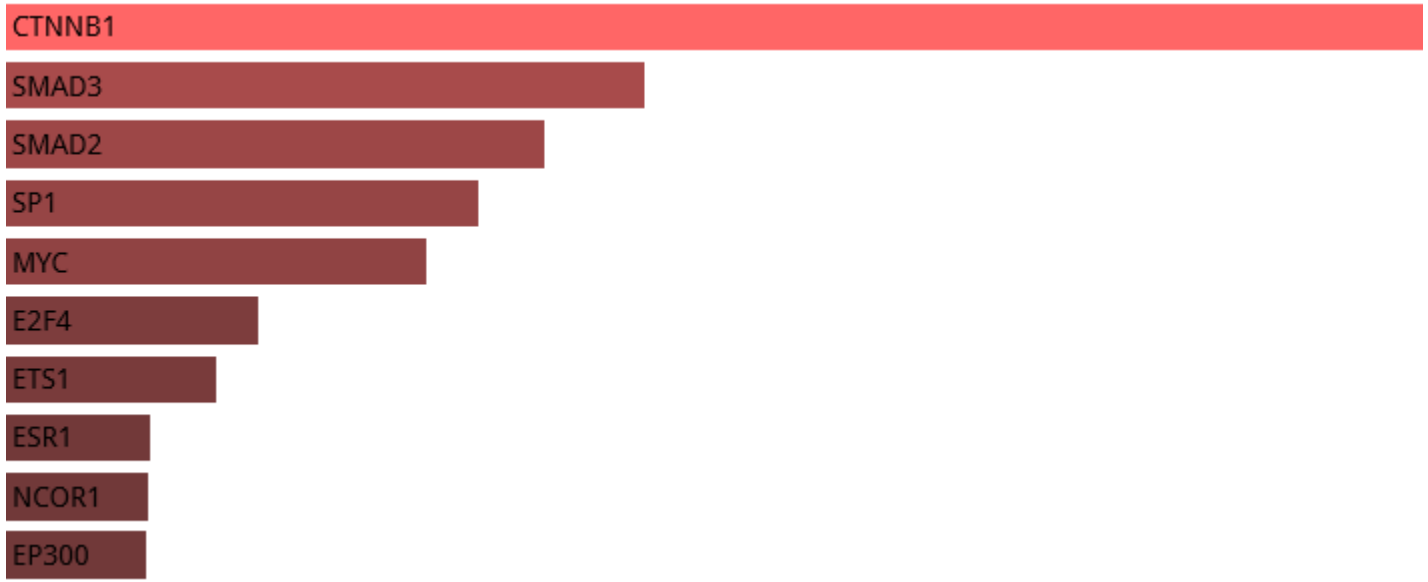
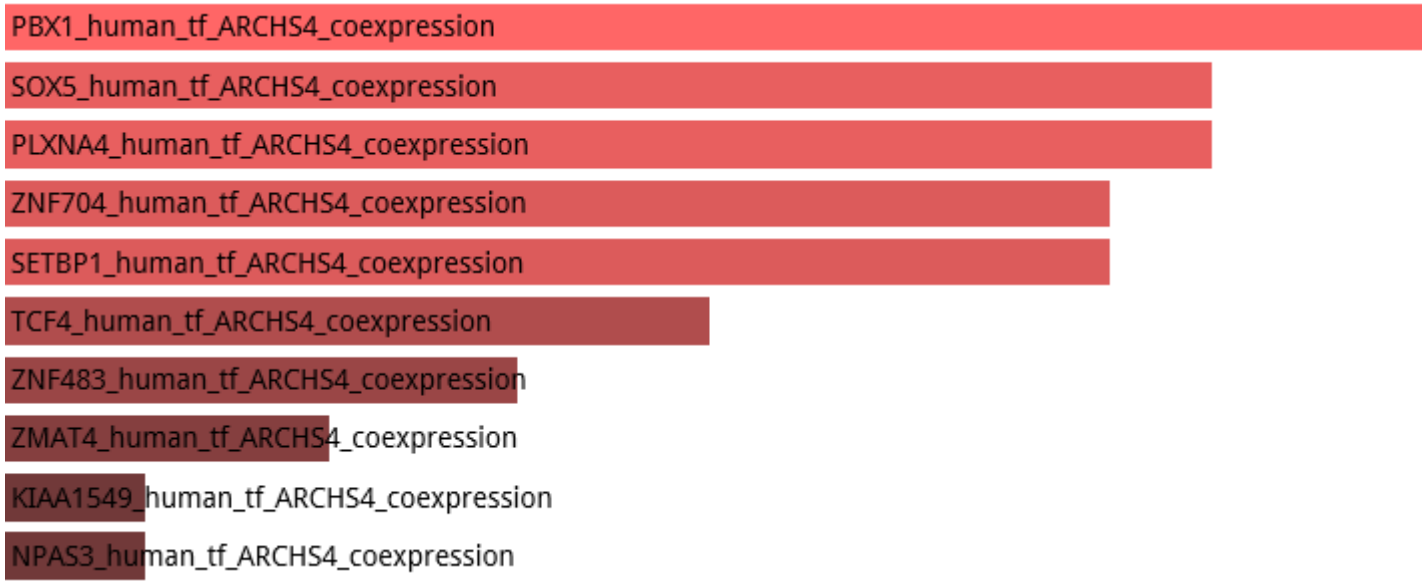
Transcription Factor PPIs 7979 genes



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

ARCHS4 TFs Co-expression 4051 genes



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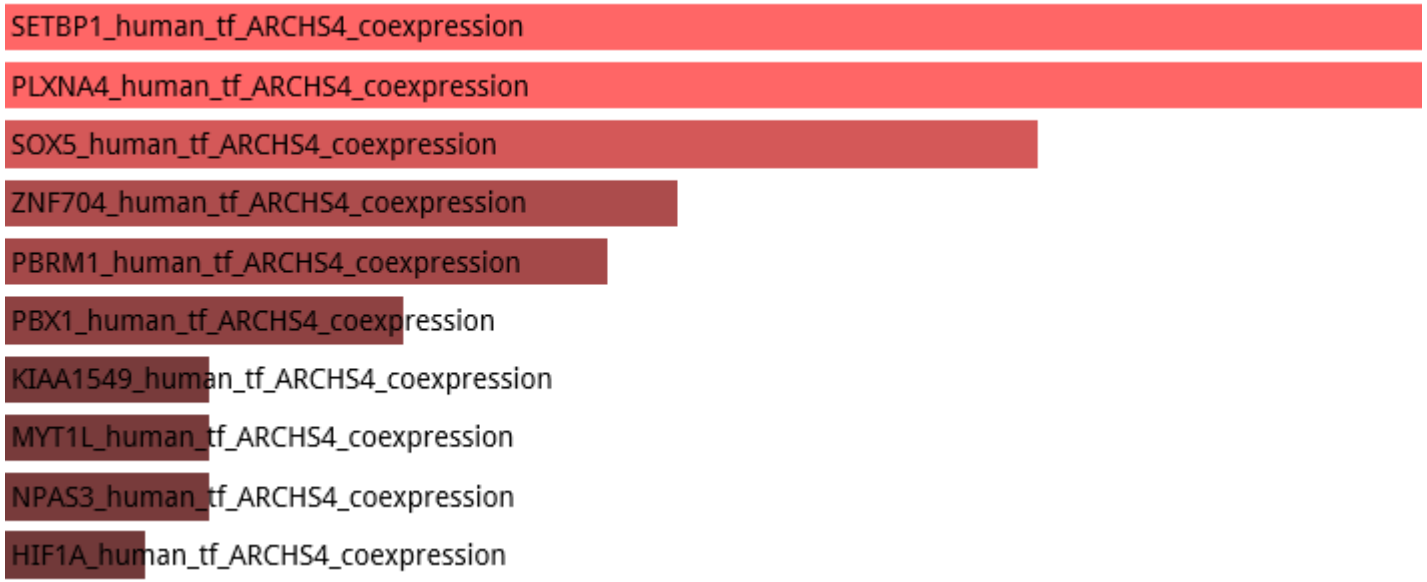
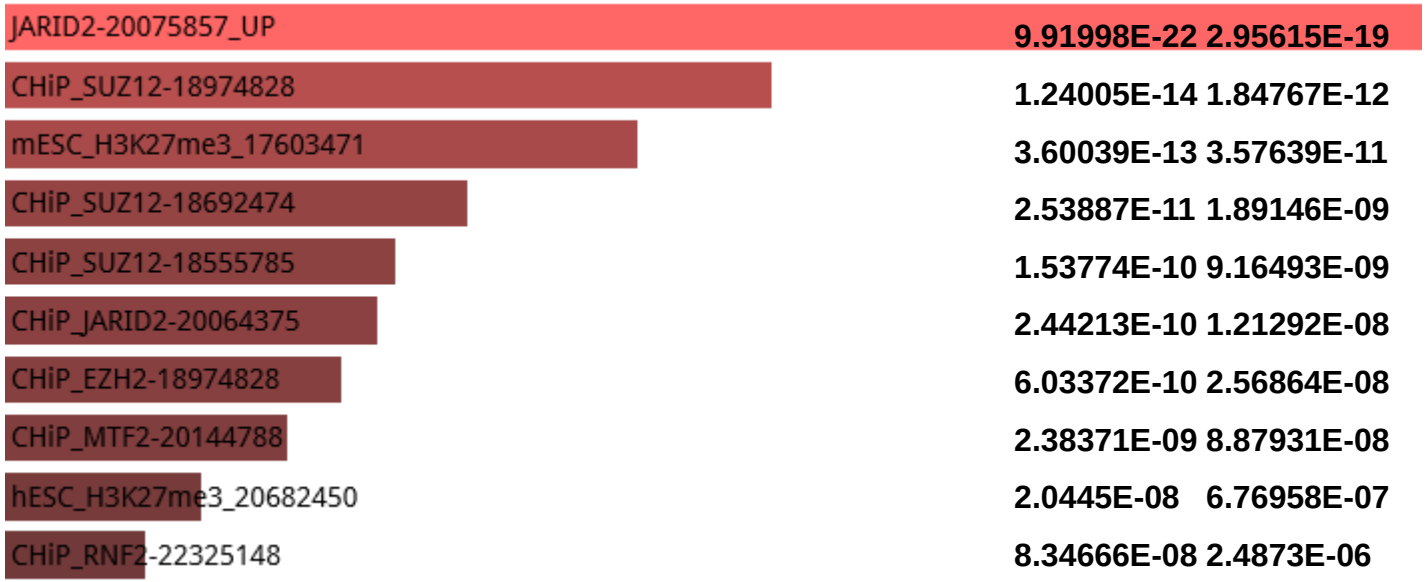
ARCHS4 TFs Co-expression 7979 genes



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

ESCAPE 4051 genes



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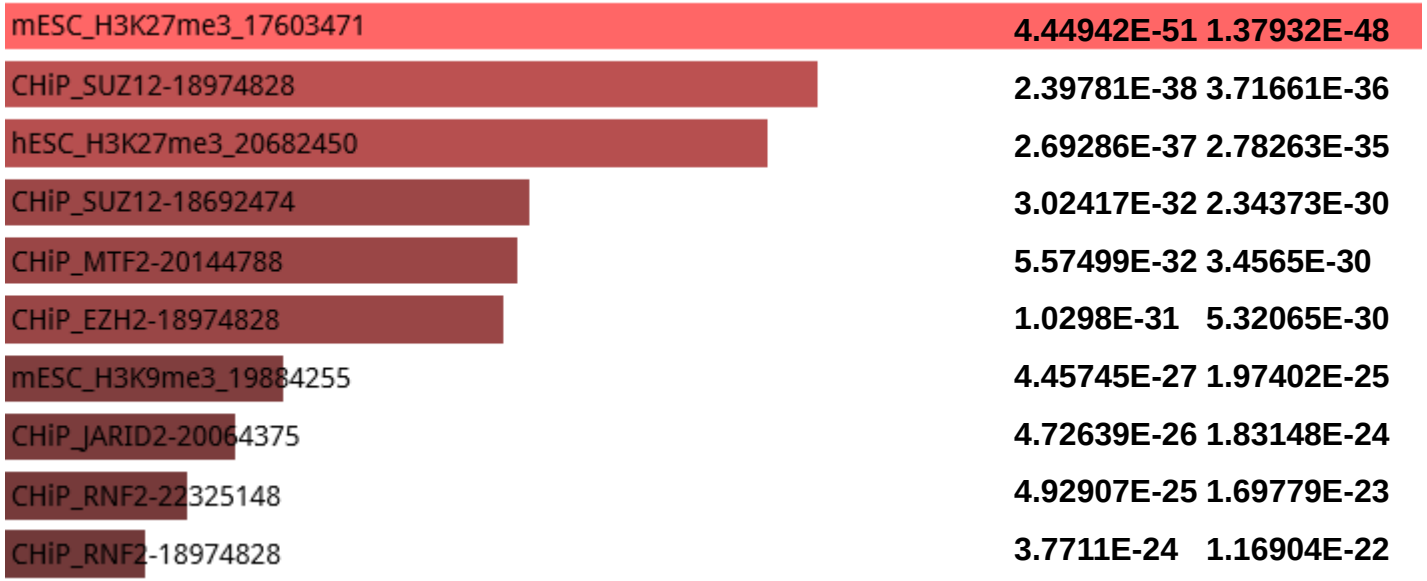

7,599 duplicated regions in the human genome

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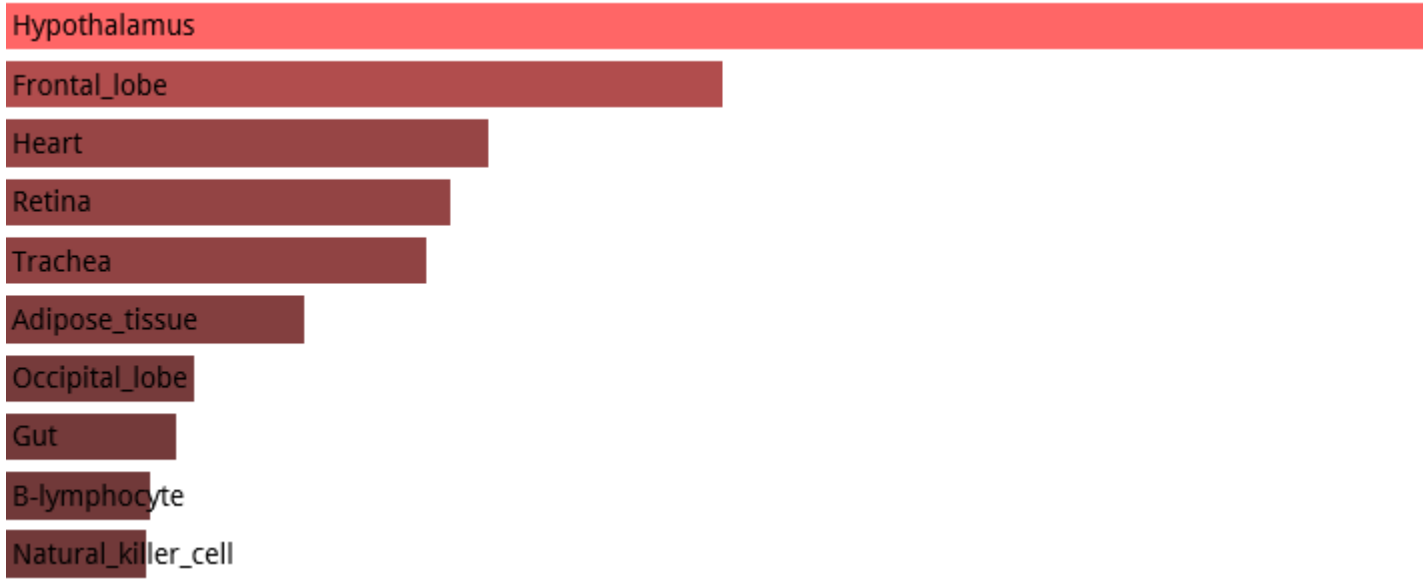
ARCHS4 Tissues 6618 genes

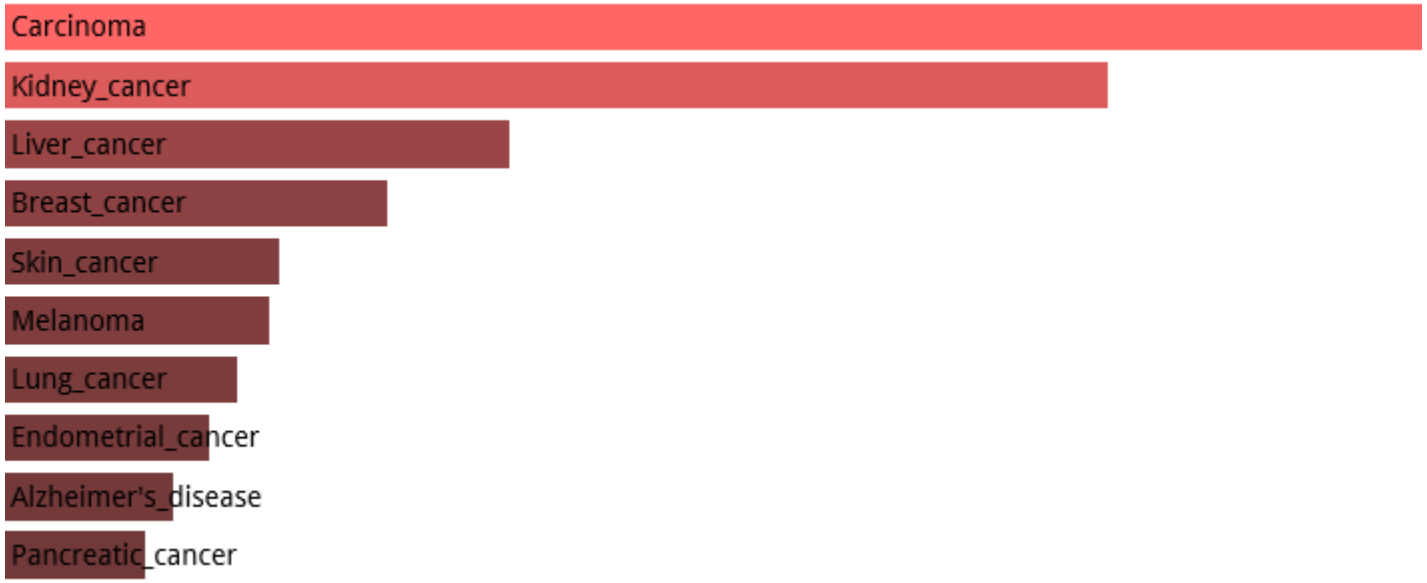
NEURONAL EPITHELIUM	**3.36327E-15**	**3.63233E-13**
PREFRONTAL CORTEX	**9.88009E-15**	**5.33525E-13**
FETAL BRAIN CORTEX	**0.000448**	**0.0161**
SPINAL CORD	**0.002852**	**0.0616**
SPINAL CORD (BULK)	**0.002852**	**0.0616**
CEREBELLUM	**0.005673**	**0.10212**
CINGULATE GYRUS	**0.015405**	**0.23768**
MOTOR NEURON	**0.049808**	**0.67241**
HUMAN EMBRYO		
CEREBRAL CORTEX		

Jensen TISSUES 6618 genes

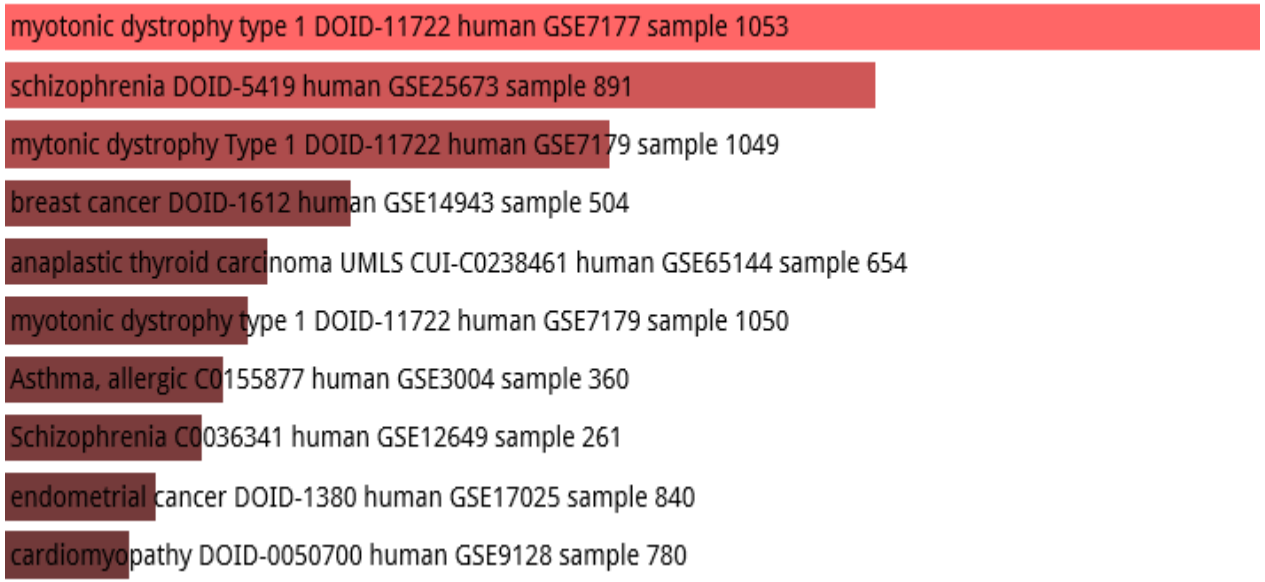


7,599 duplicated regions in the human genome

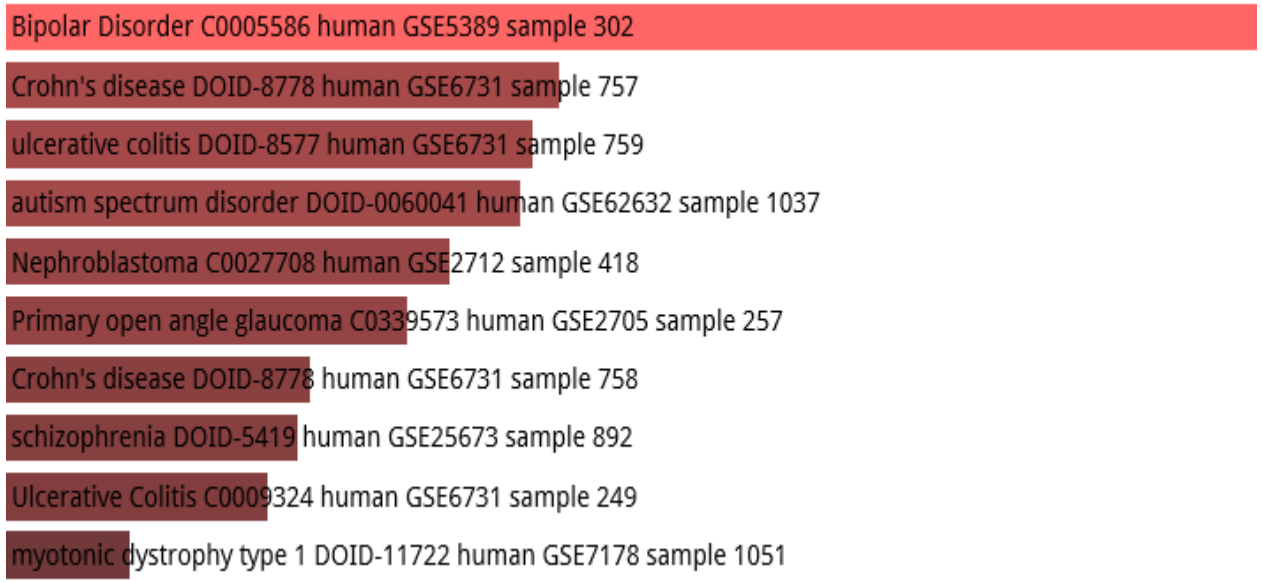
Jensen DISEASES 6618 genes



Disease Perturbations from GEO up



Disease Perturbations from GEO down



5,883 fixed human-specific deletions

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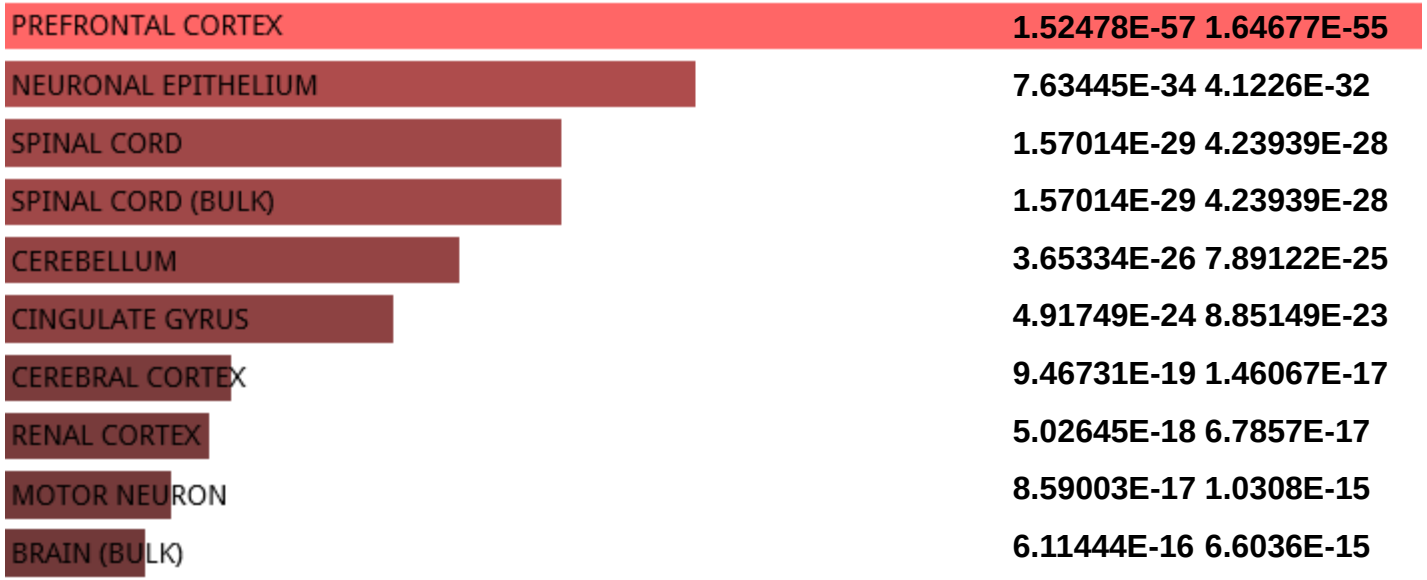
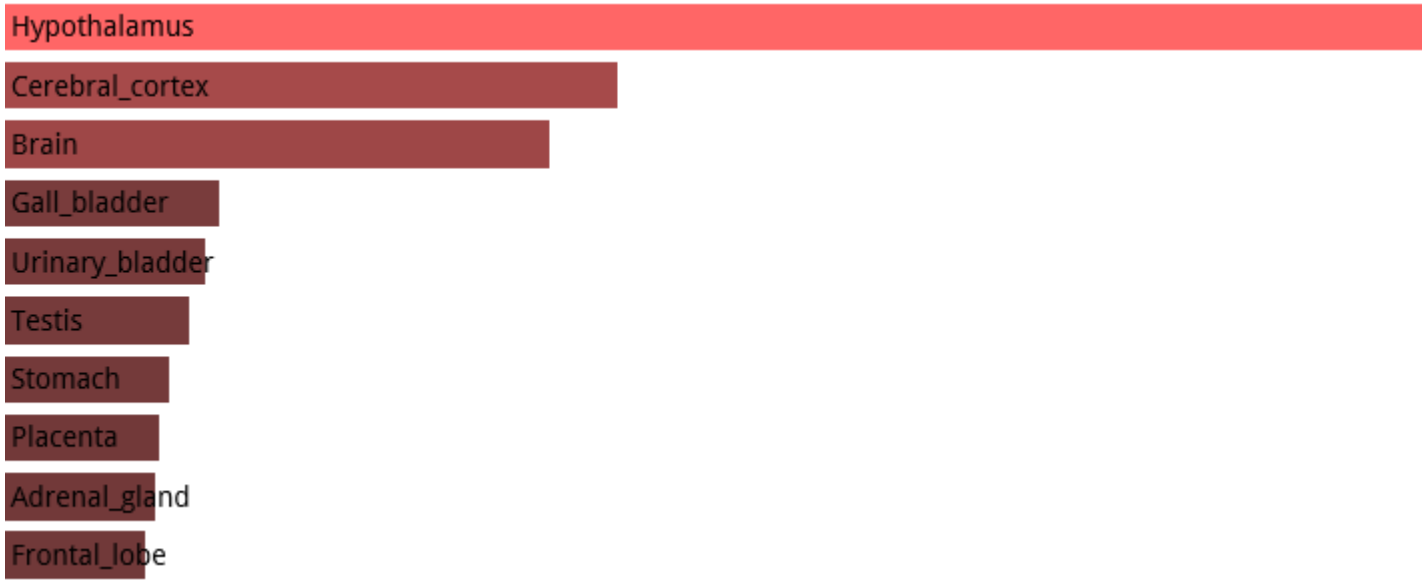
ARCHS4 Tissues 5489 genes



Jensen TISSUES 5489 genes



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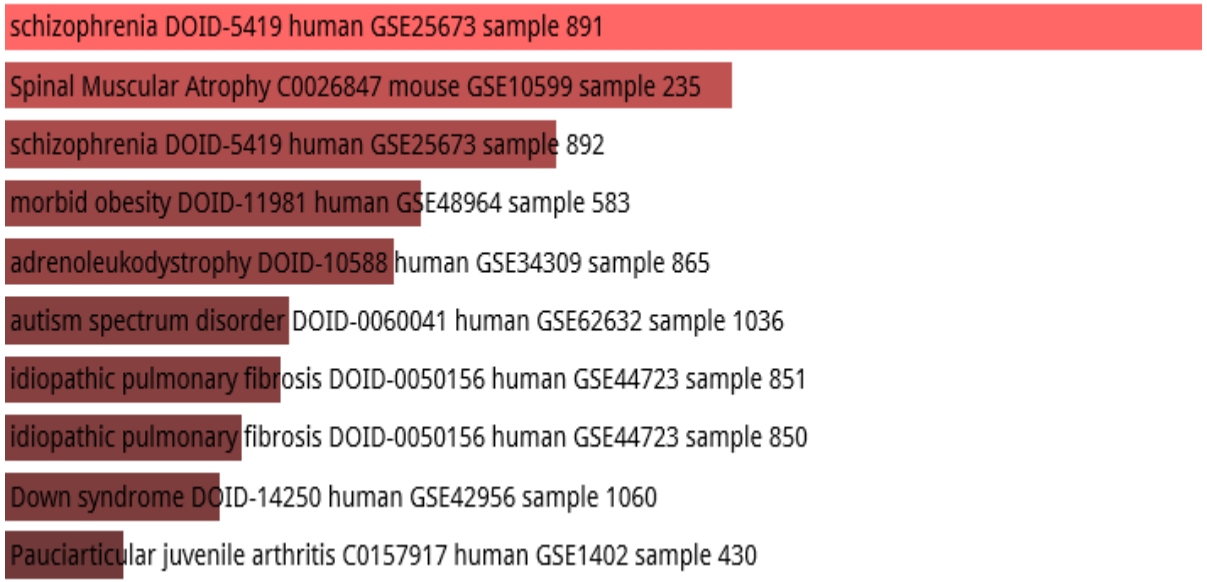
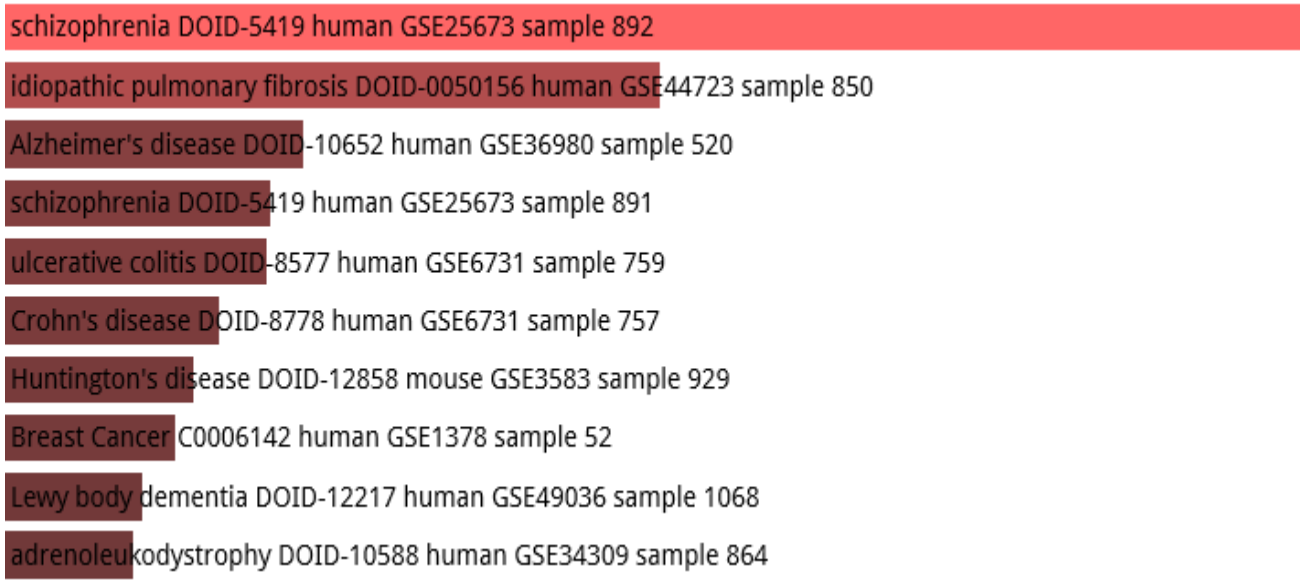


Disease Perturbations from GEO down



4,875 human-specific STR expansions

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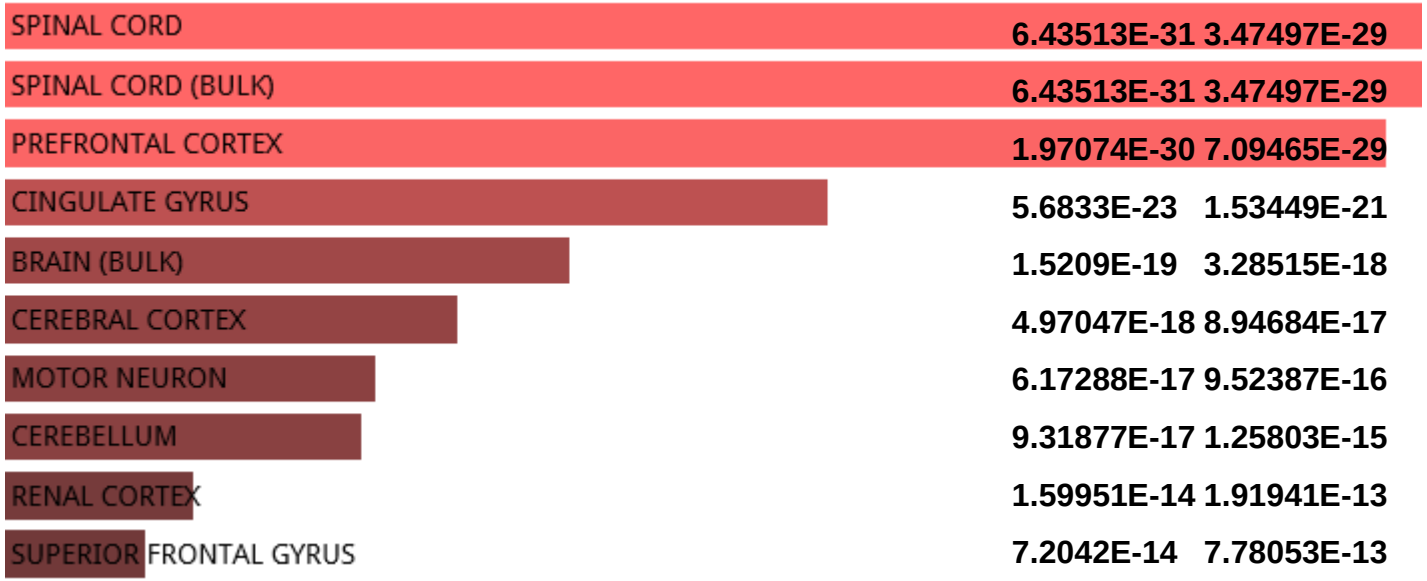
ARCHS4 Tissues 4844 genes



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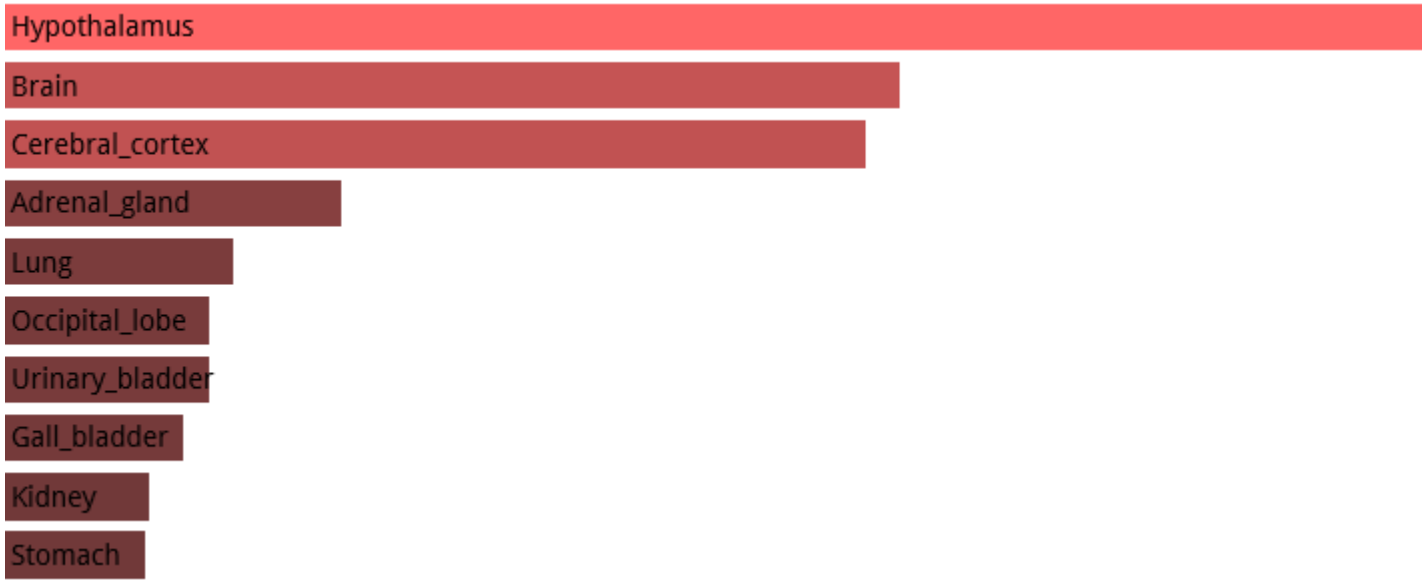
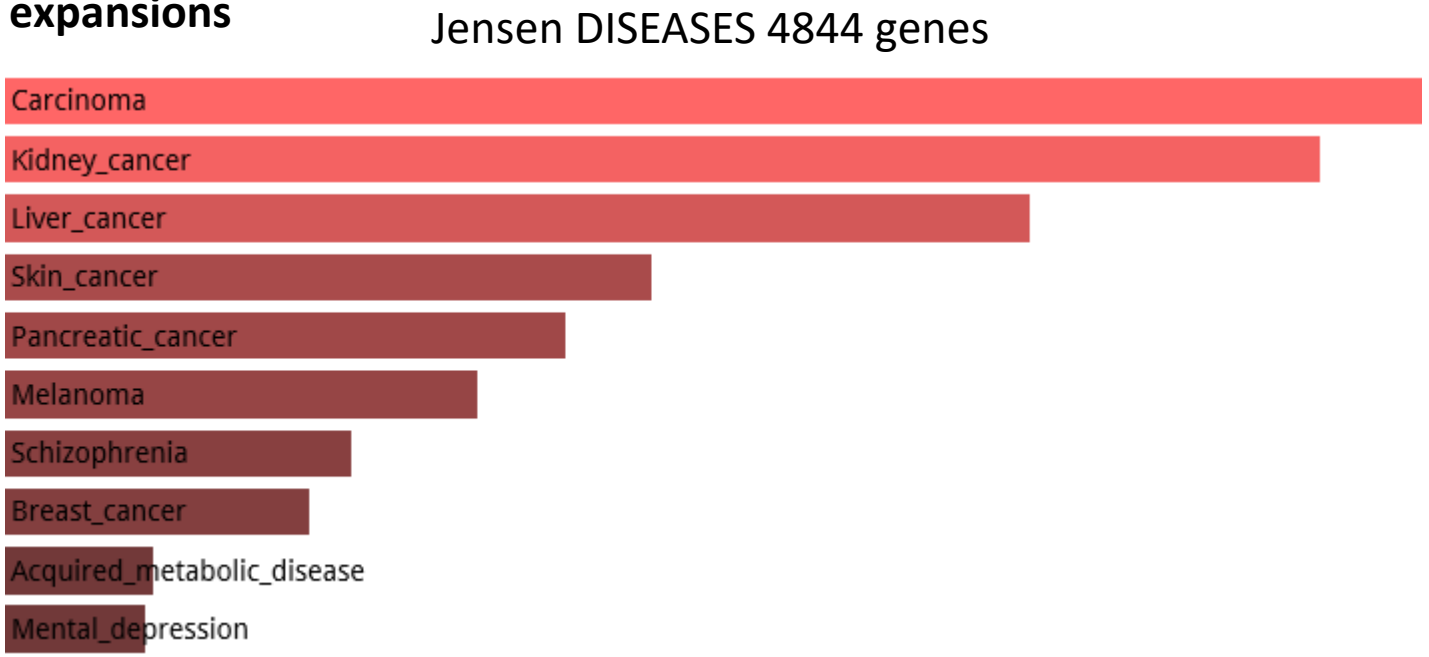
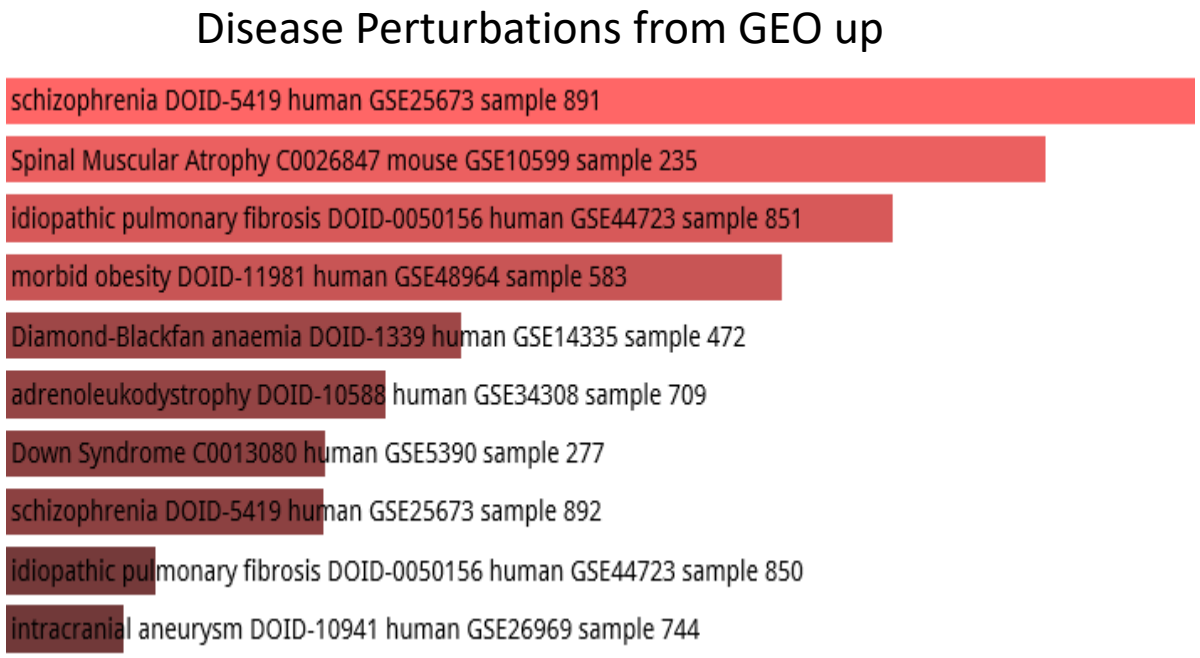
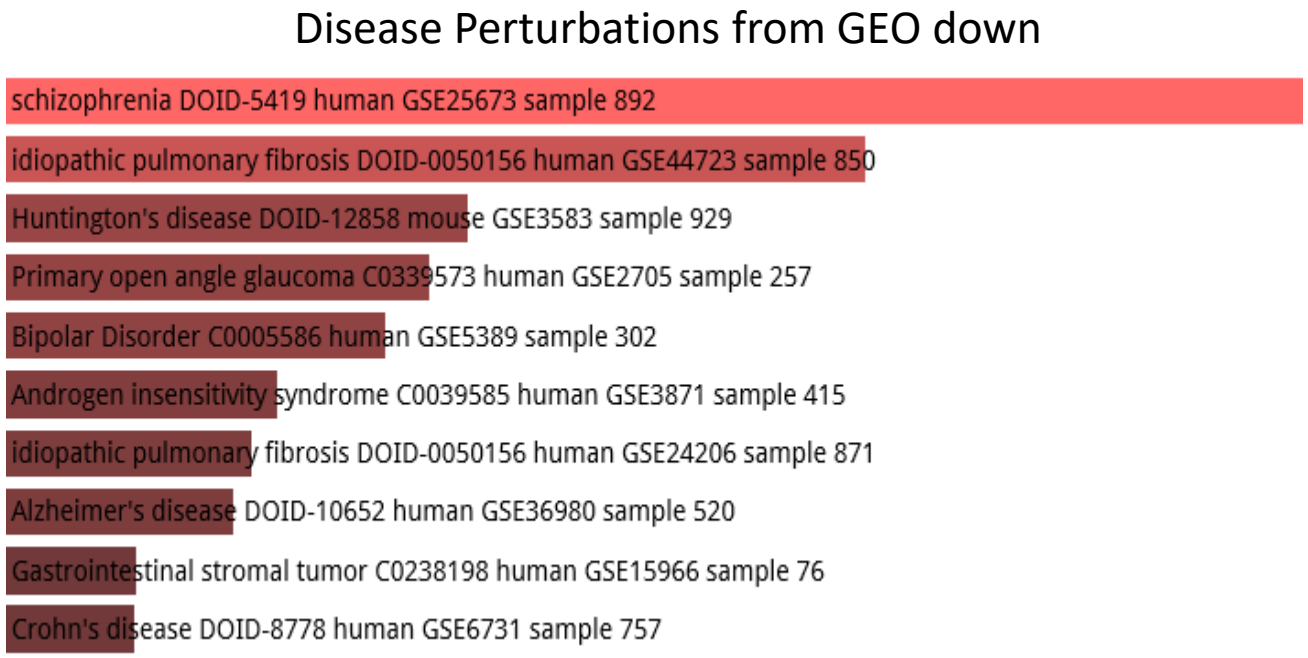


4,875 human-specific STR expansions







3,538 human-specific ace-DHS

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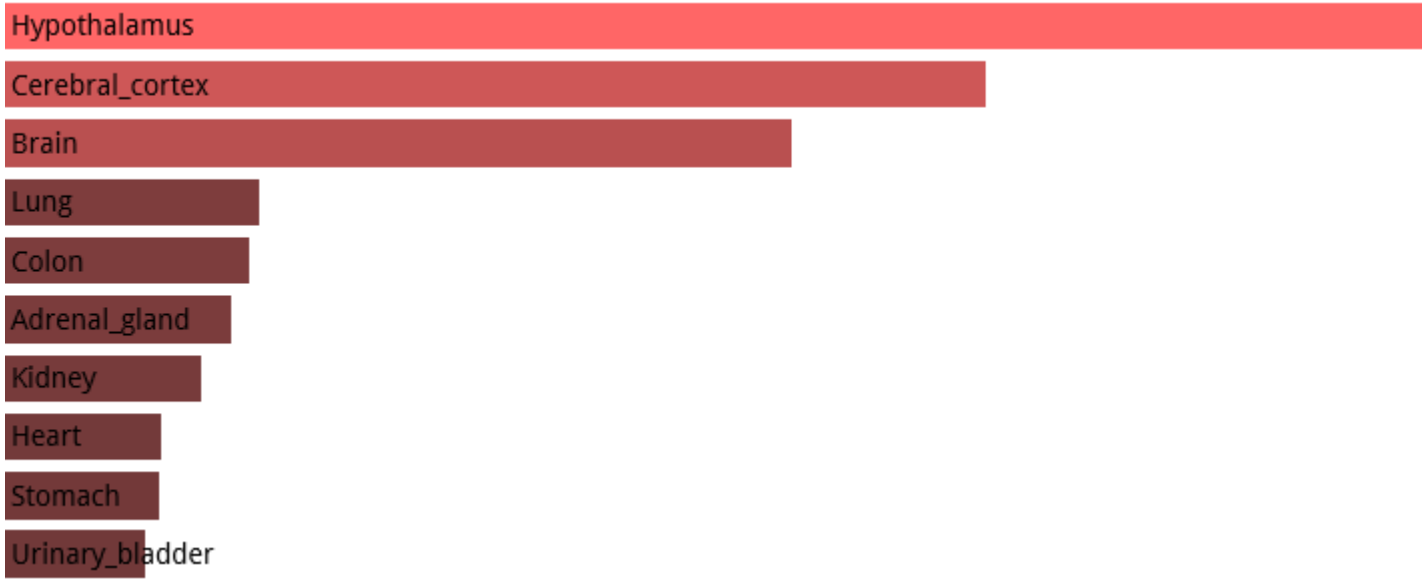
ARCHS4 Tissues 3445 genes

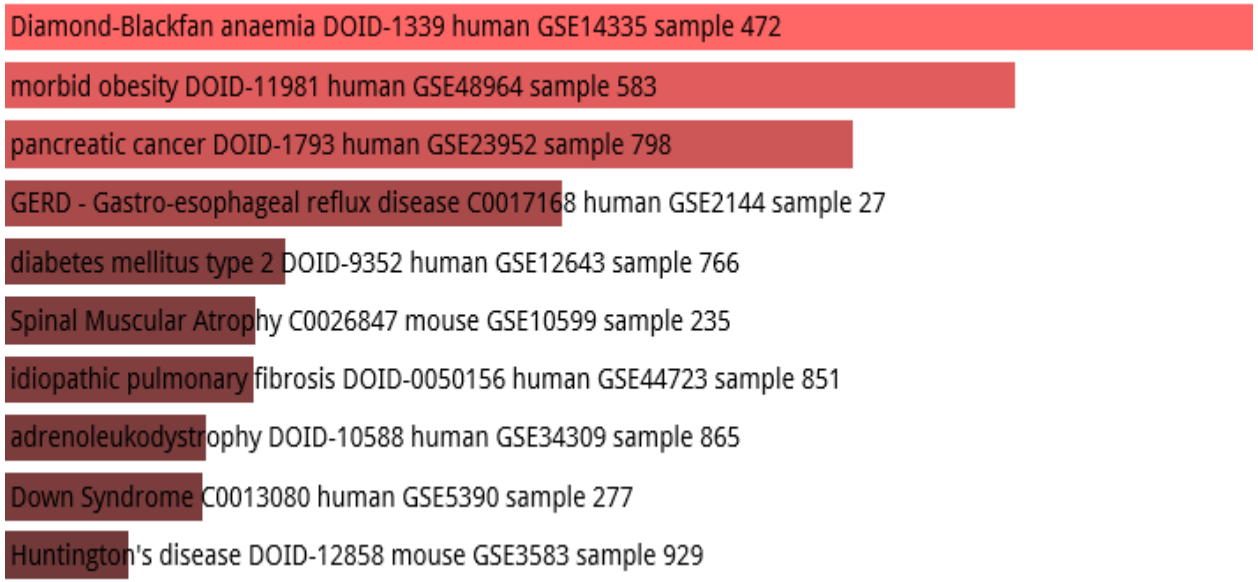
Jensen TISSUES 3445 genes



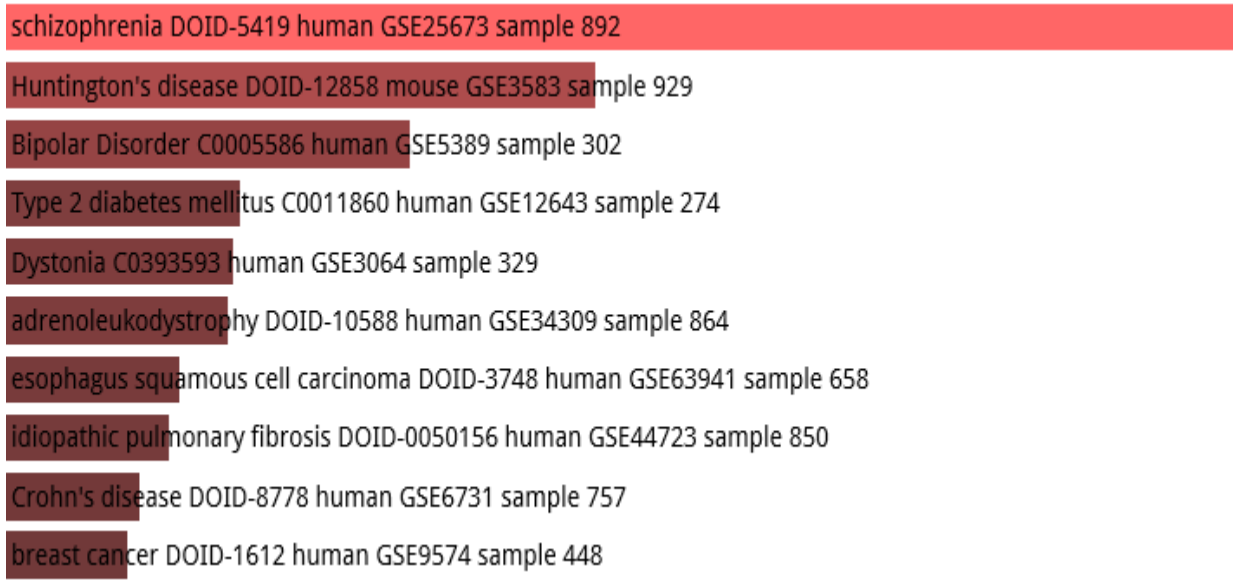
3,538 human-specific ace-DHS

Jensen DISEASES 3445 genes

Disease Perturbations from GEO up

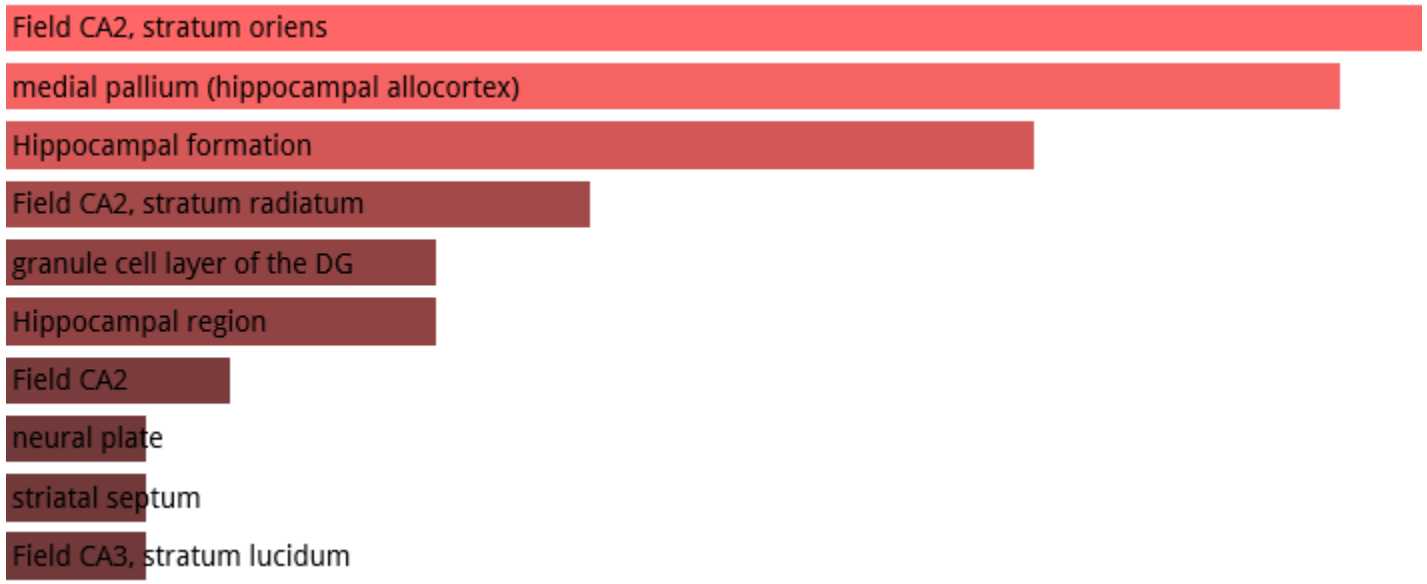


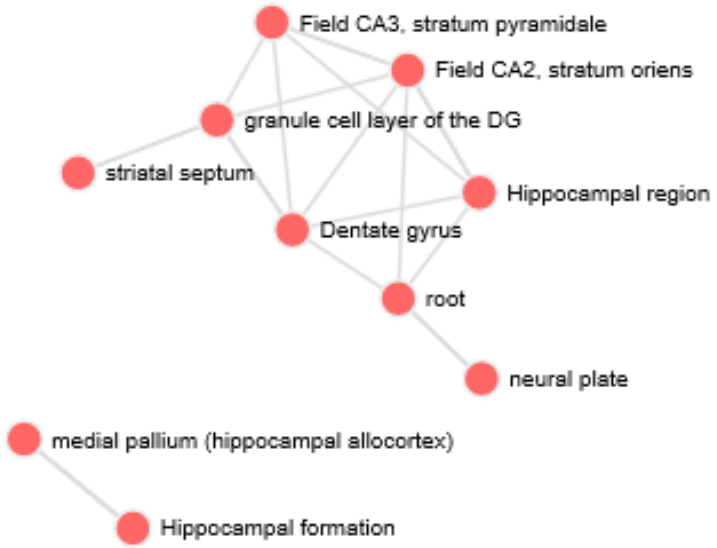
Disease Perturbations from GEO down



3,538 human-specific ace-DHS

Allen Brain Atlas up 3445 genes





3,803 human-specific TFBS in hESC

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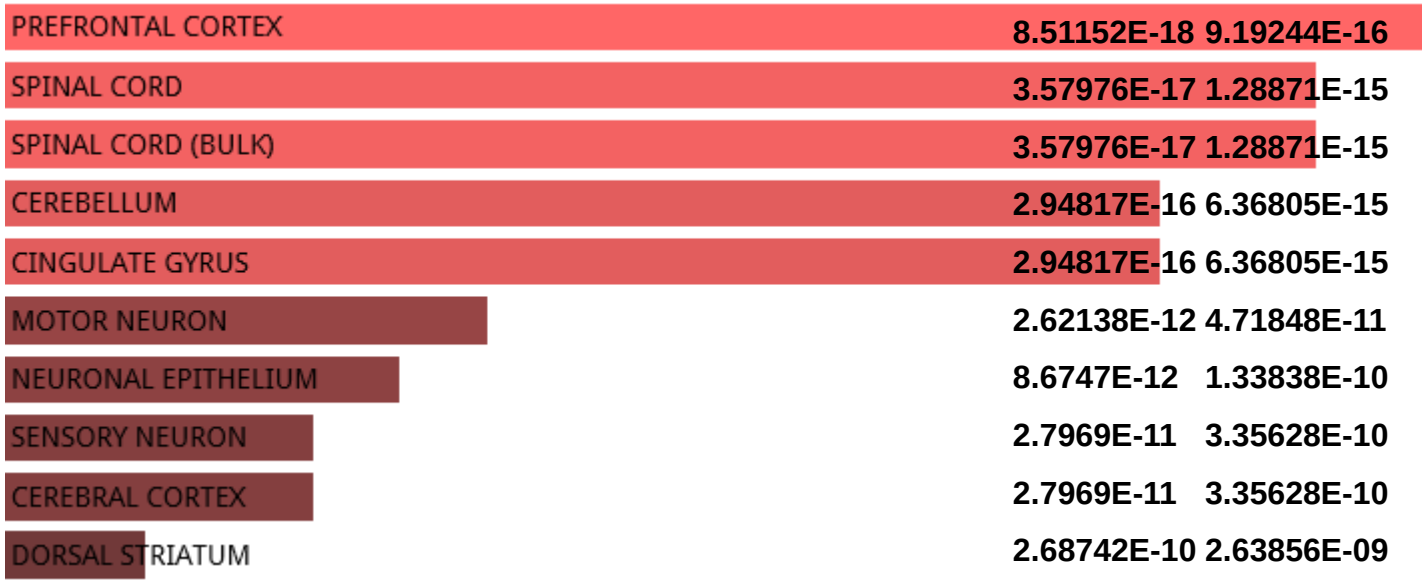
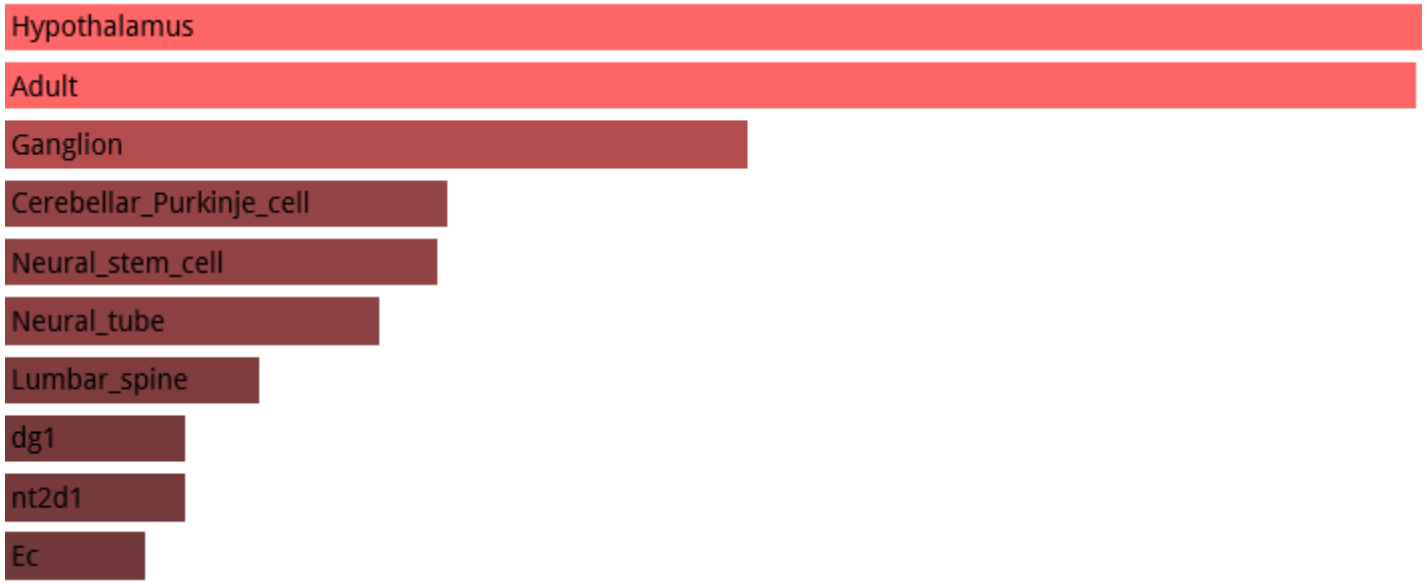
ARCHS4 Tissues 1087 genes



Jensen TISSUES 1087 genes



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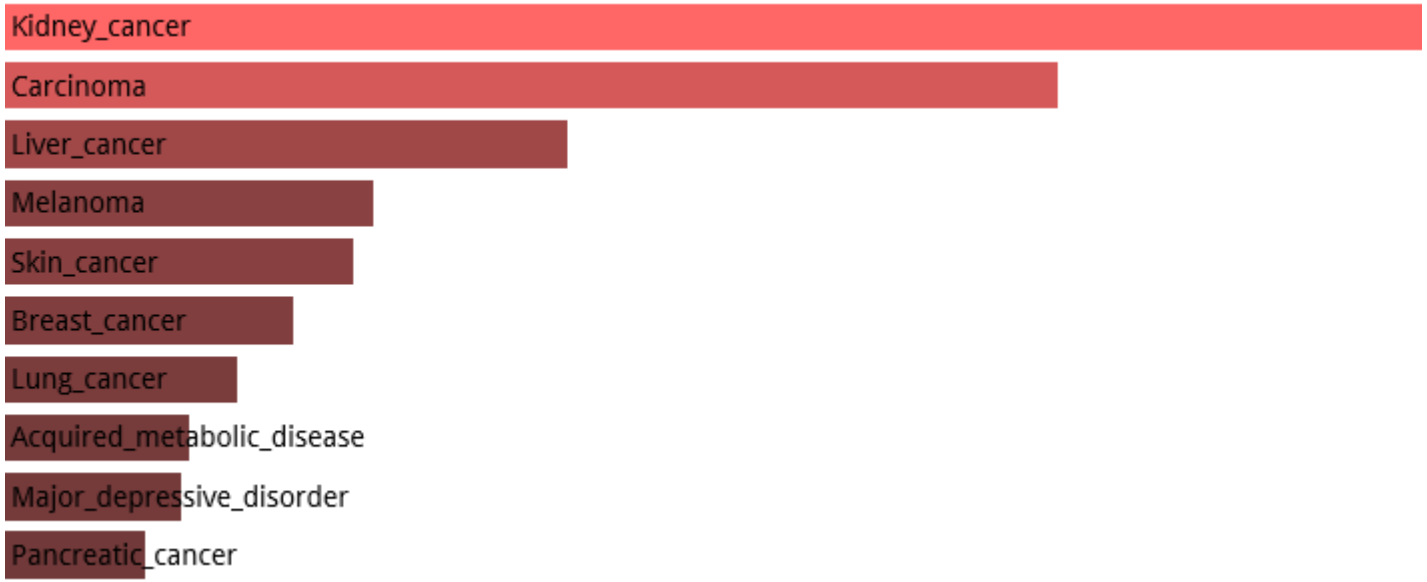
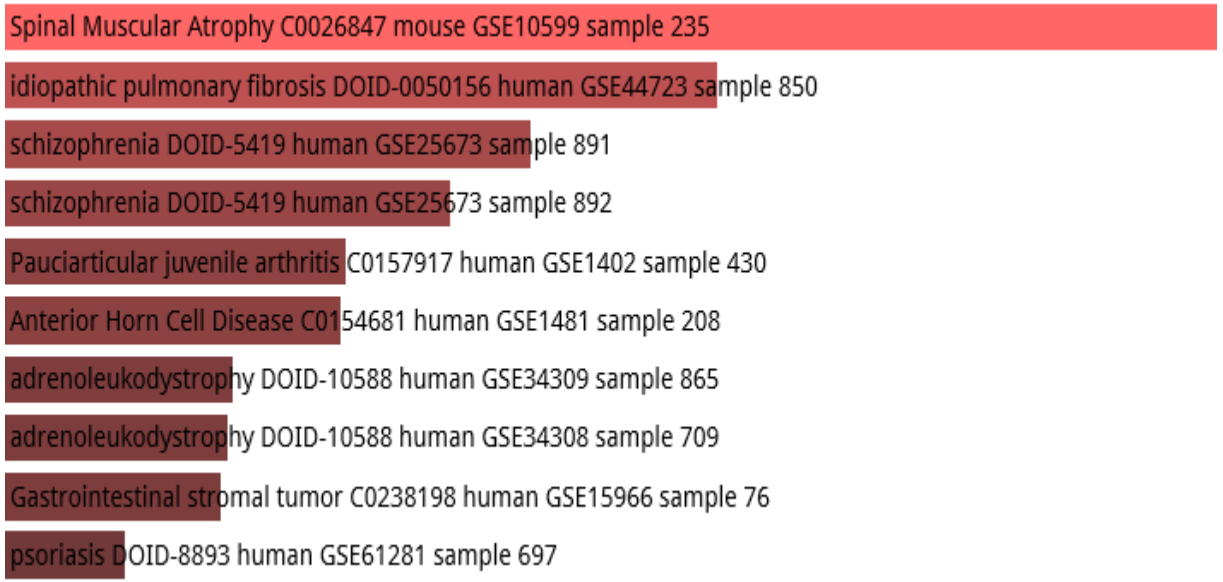
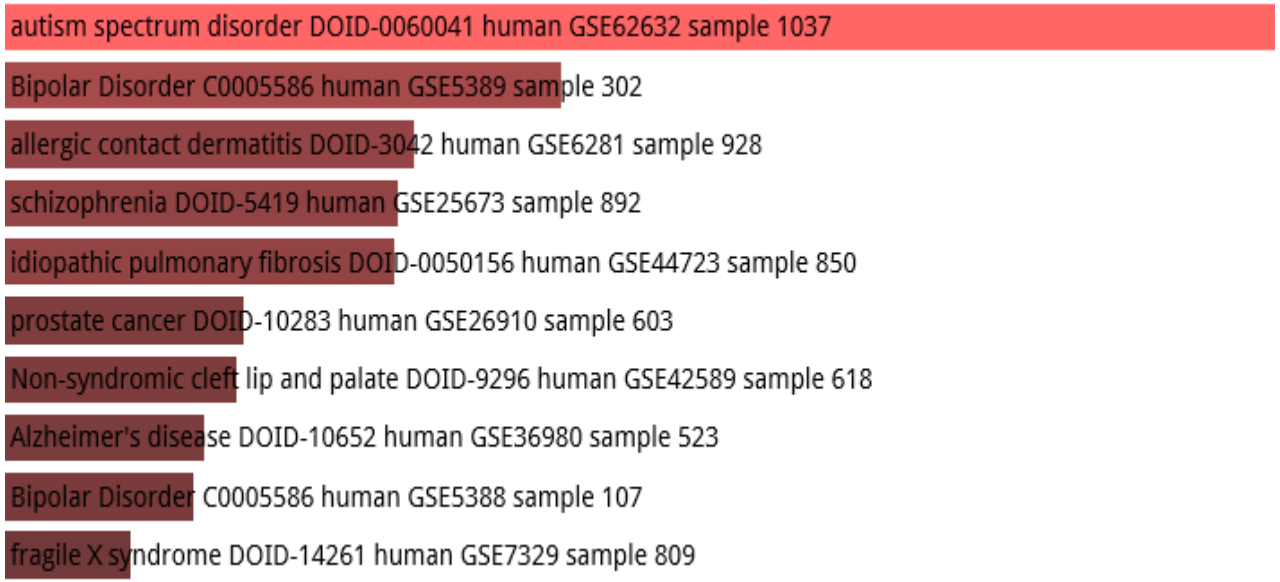


Disease Perturbations from GEO up



Disease Perturbations from GEO down



4,249 fixed human-specific regulatory loci

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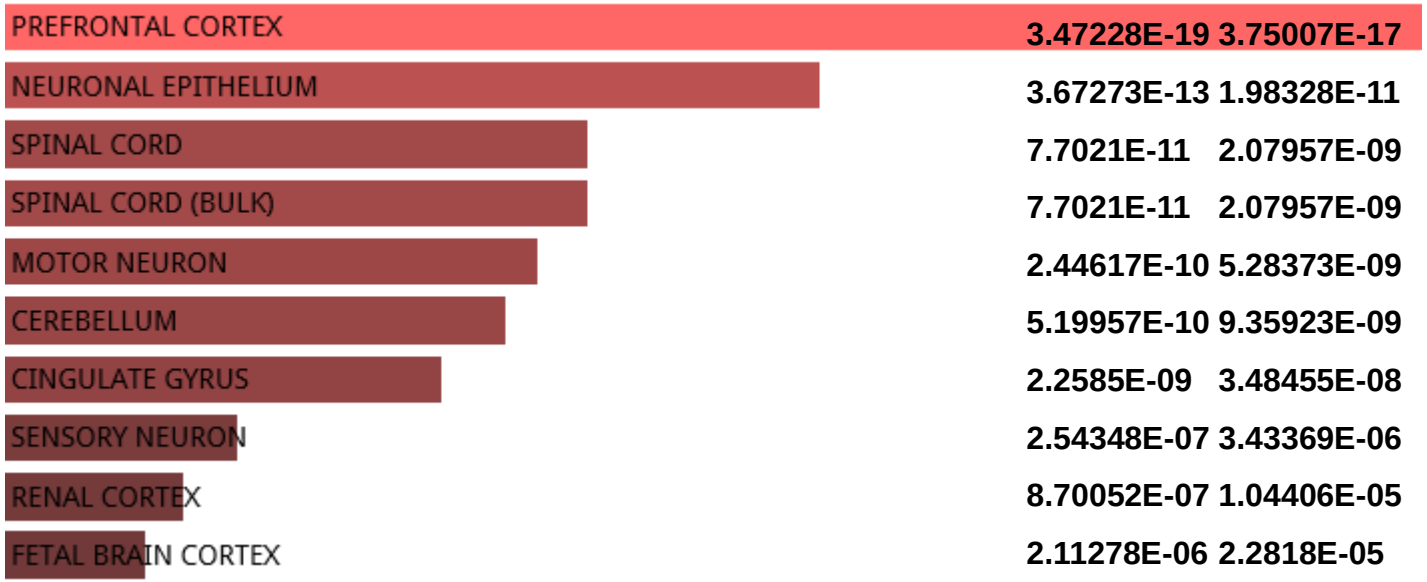
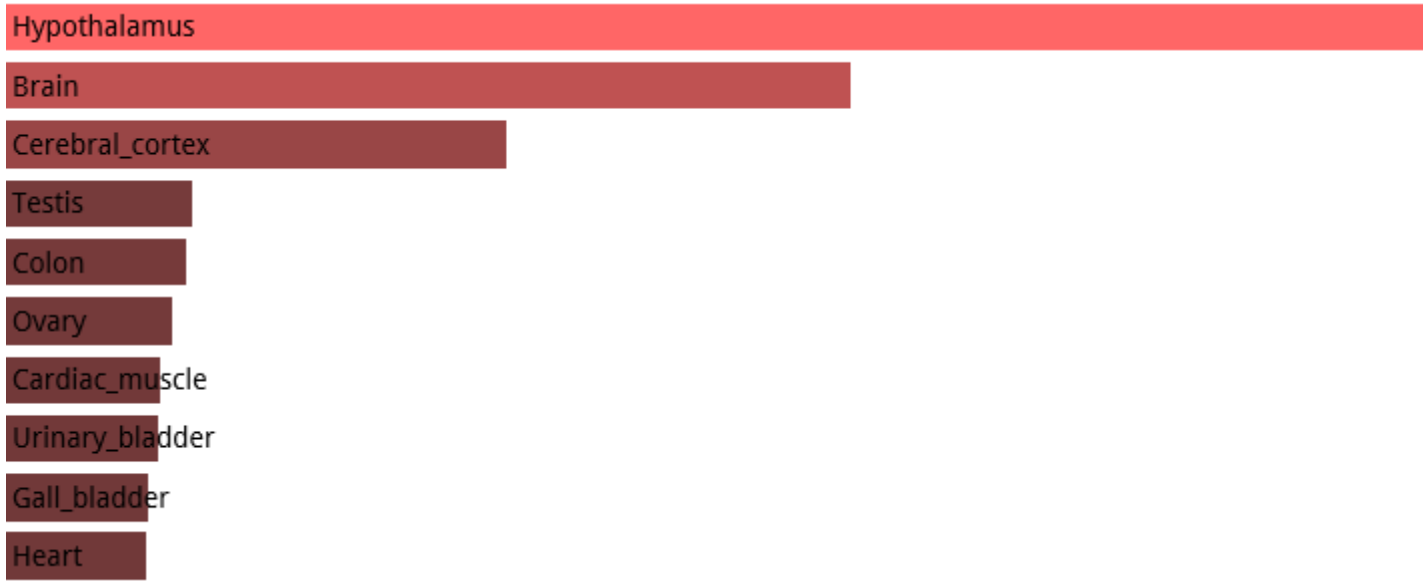
ARCHS4 Tissues 2810 genes

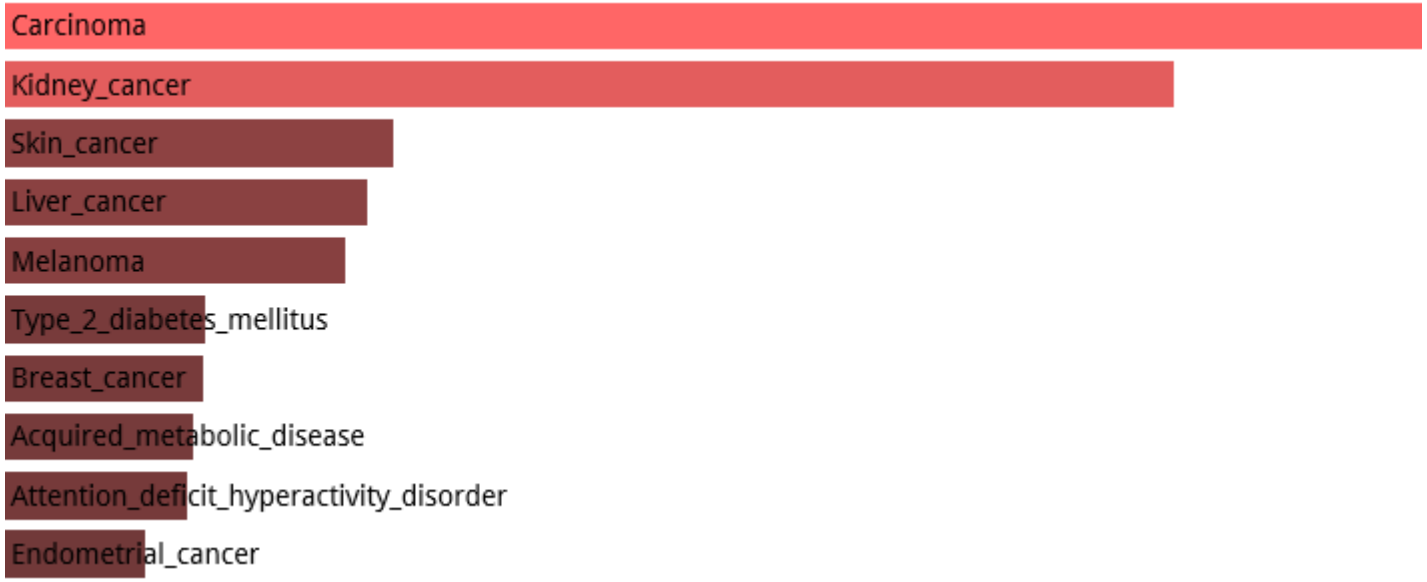


Jensen TISSUES 2810 genes

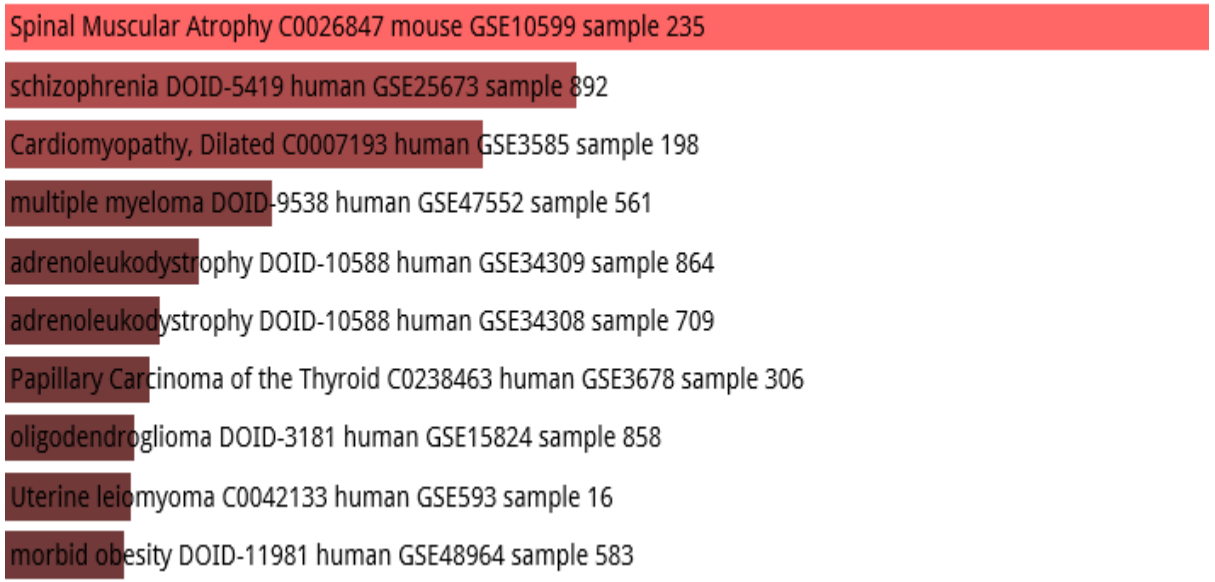


4,249 fixed human-specific regulatory loci

Jensen DISEASES 2810 genes



Disease Perturbations from GEO up

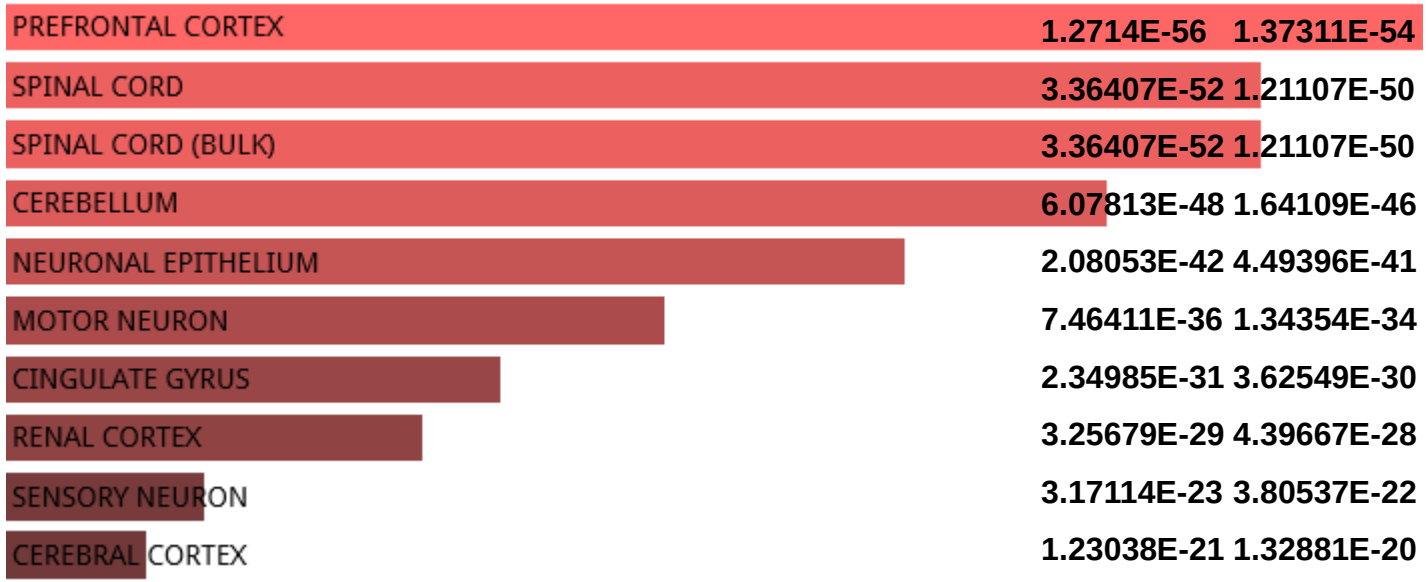


Disease Perturbations from GEO down

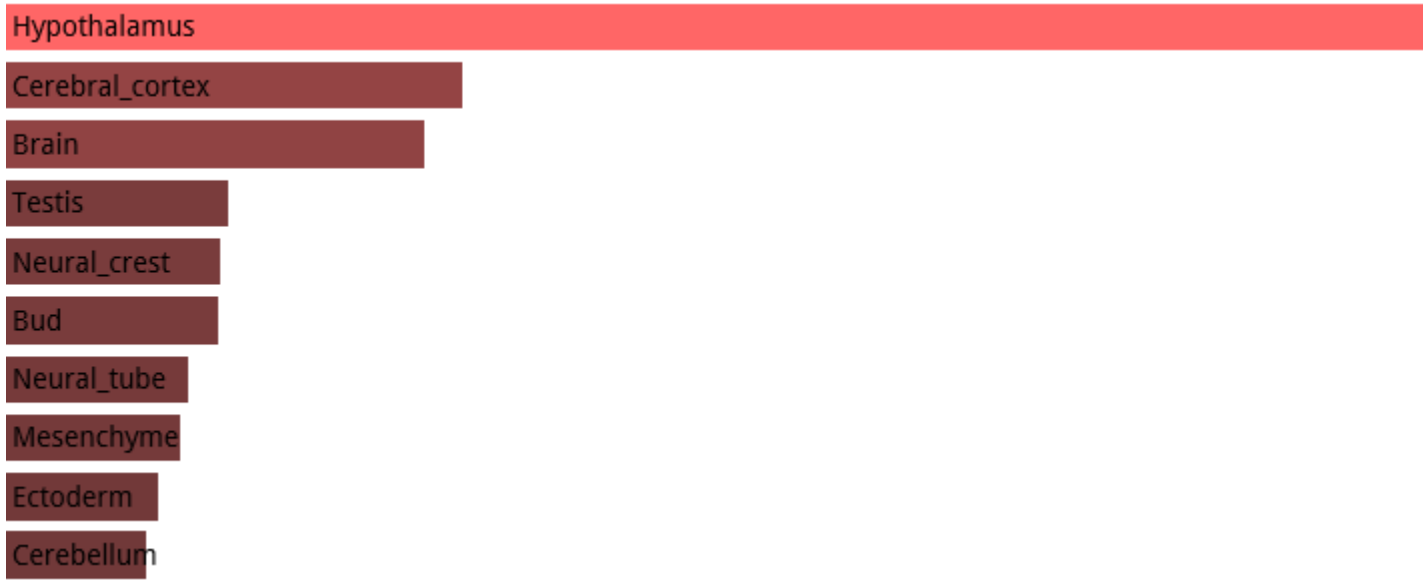
2,745 human accelerated regions

2,745 human accelerated regions

ARCHS4 Tissues 2281 genes

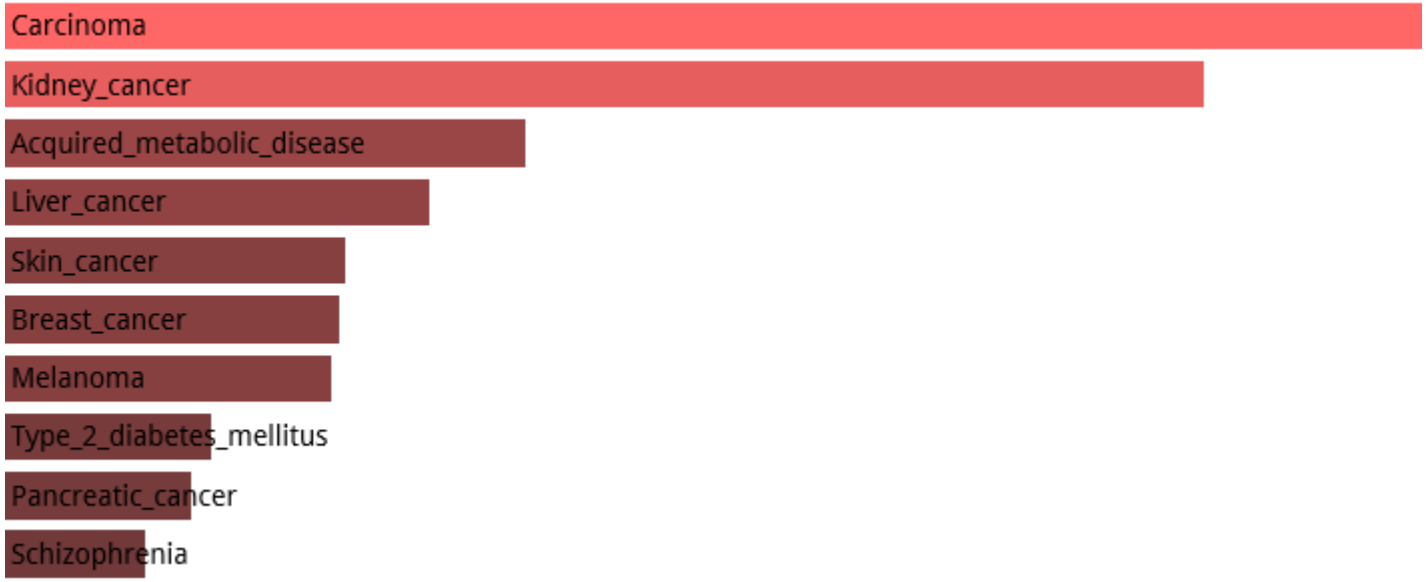


Jensen TISSUES 2281 genes

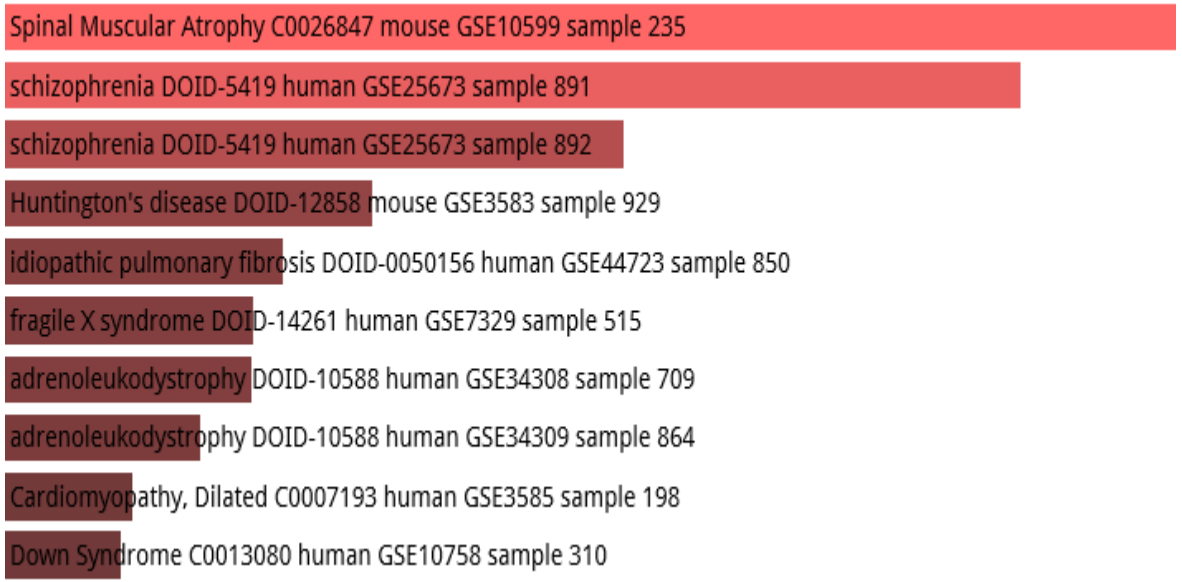


2,745 human accelerated regions

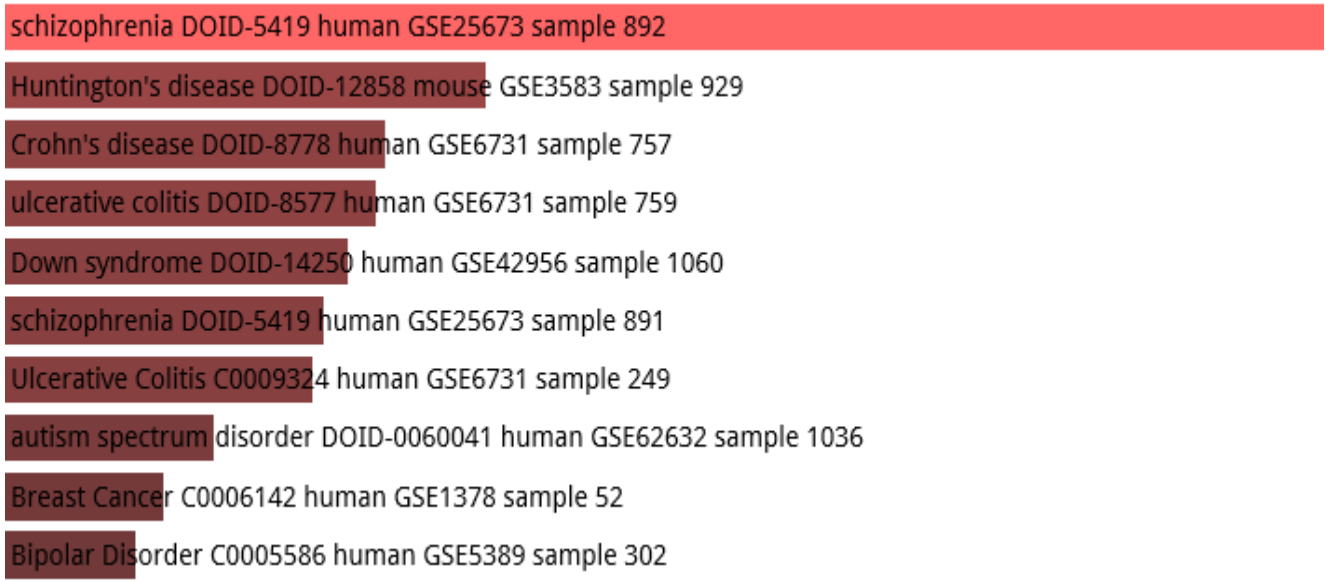
Jensen DISEASES 2281 genes



Disease Perturbations from GEO up



Disease Perturbations from GEO down



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

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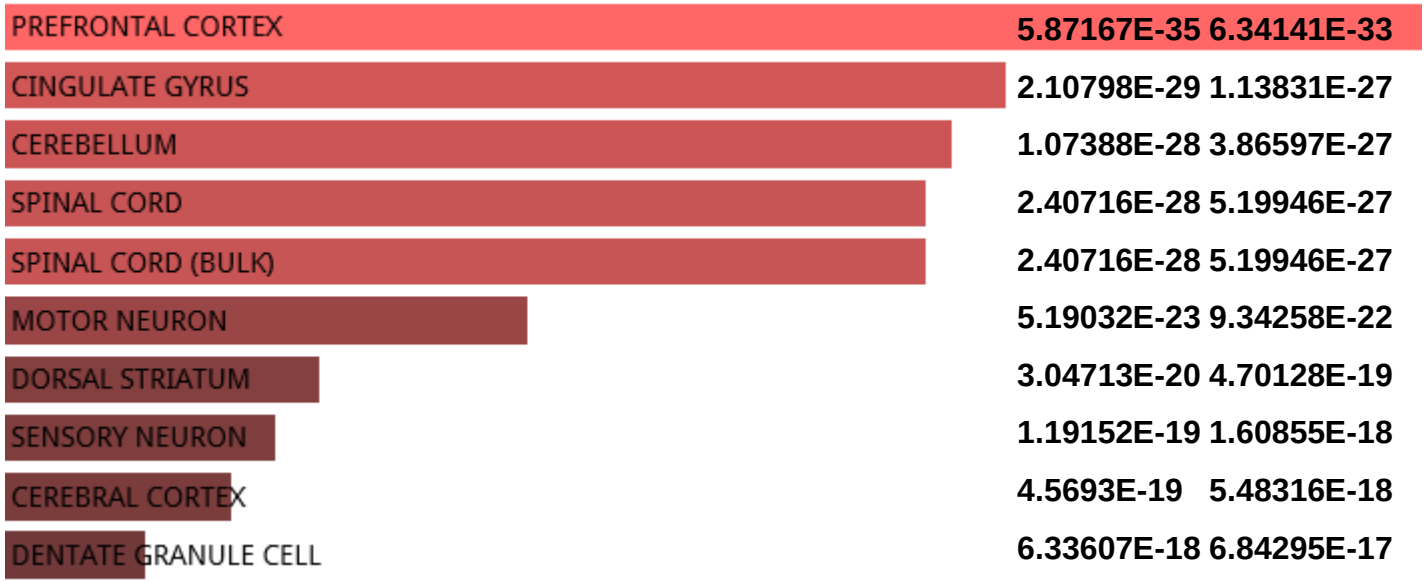
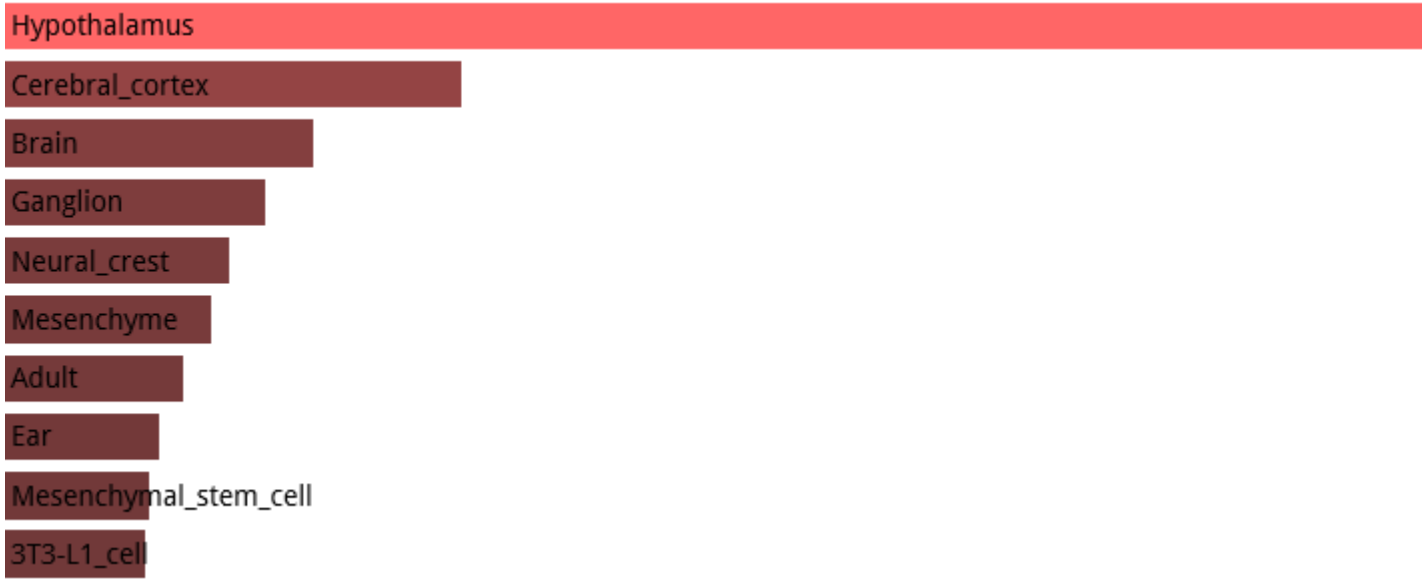
ARCHS4 Tissues 1458 genes

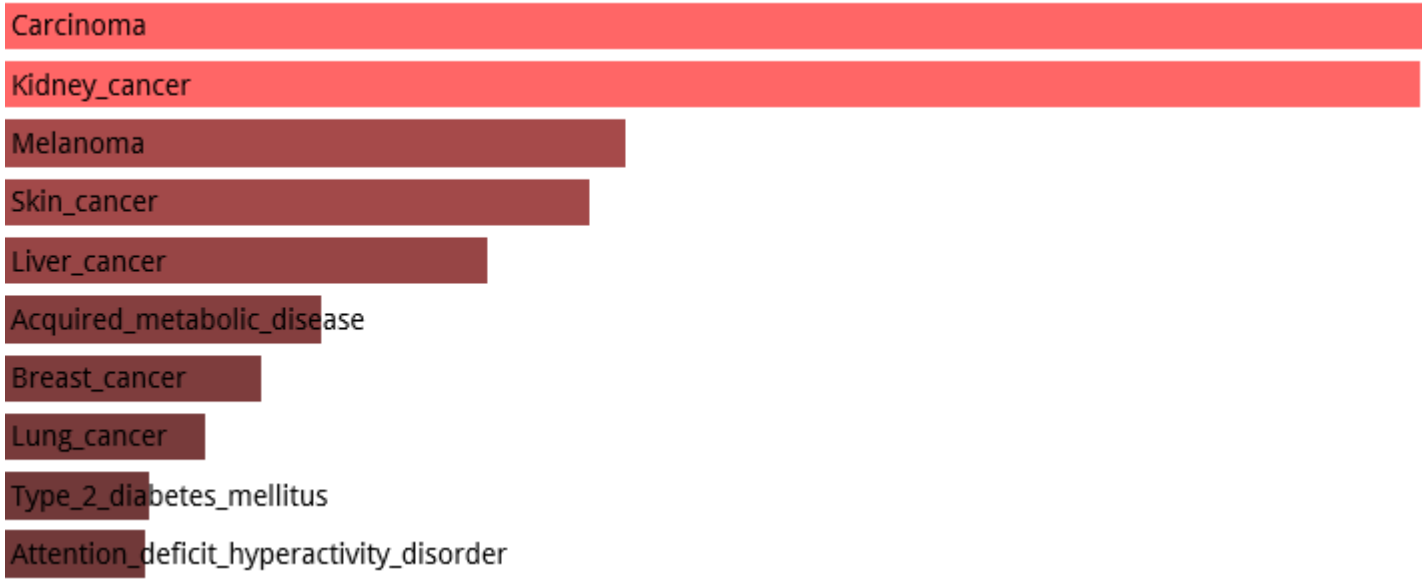


Jensen TISSUES 1458 genes

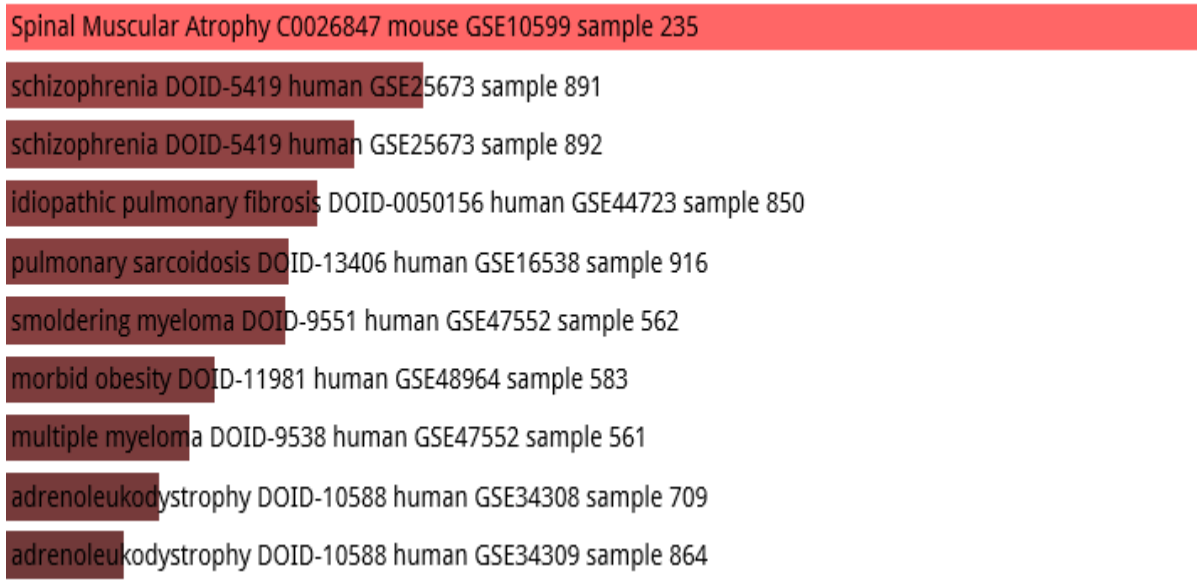


1,932 fixed human-specific regulatory regions (hESC DHS)

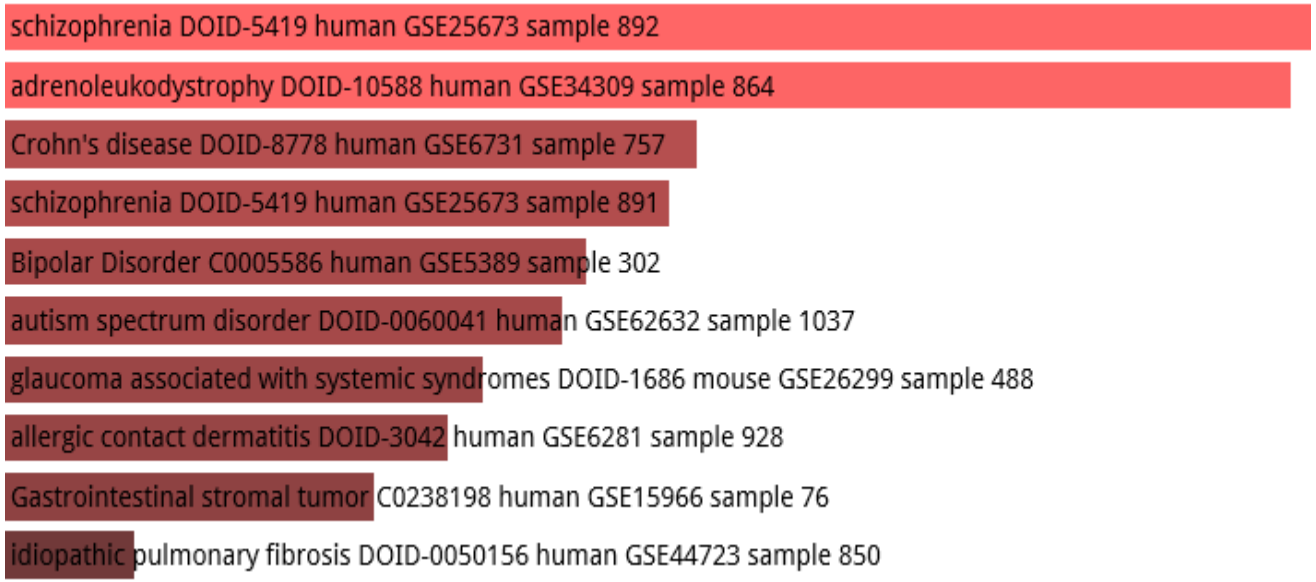
Jensen DISEASES 1458 genes



Disease Perturbations from GEO up



Disease Perturbations from GEO down



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

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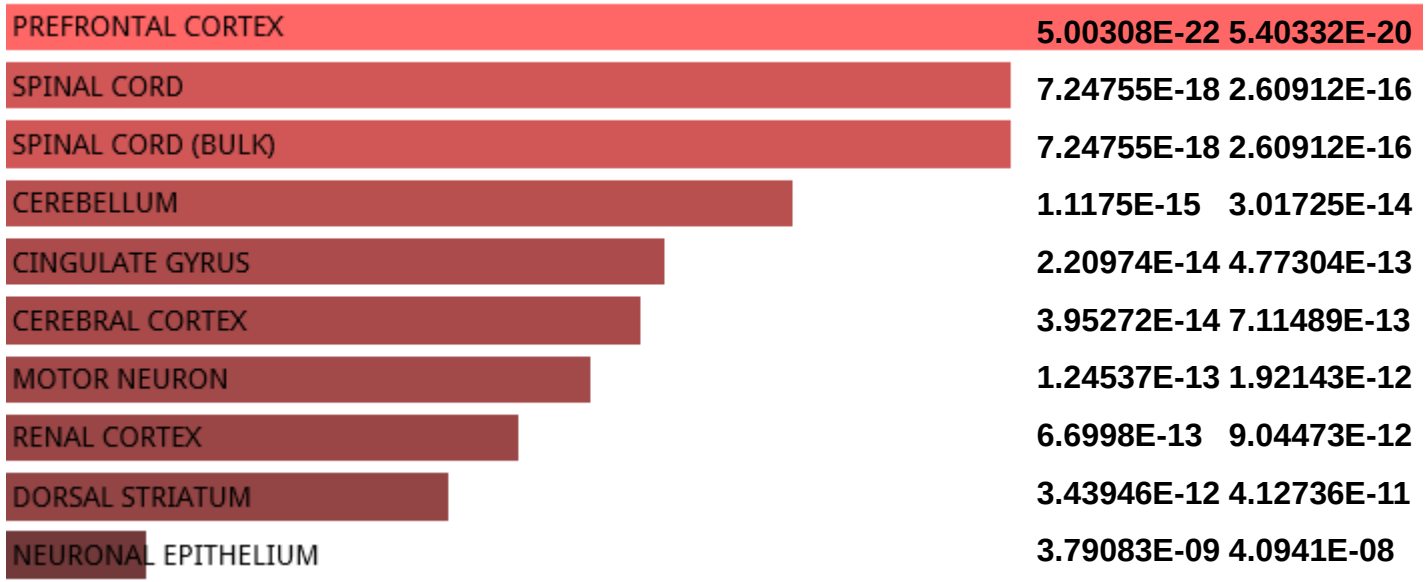
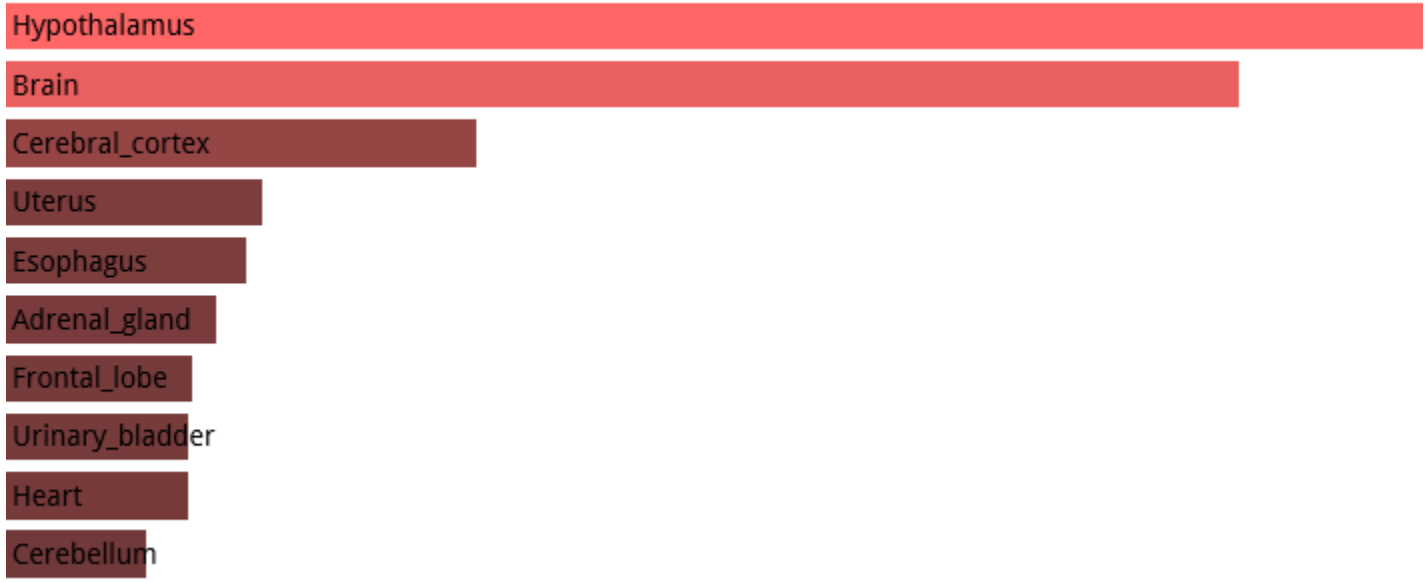
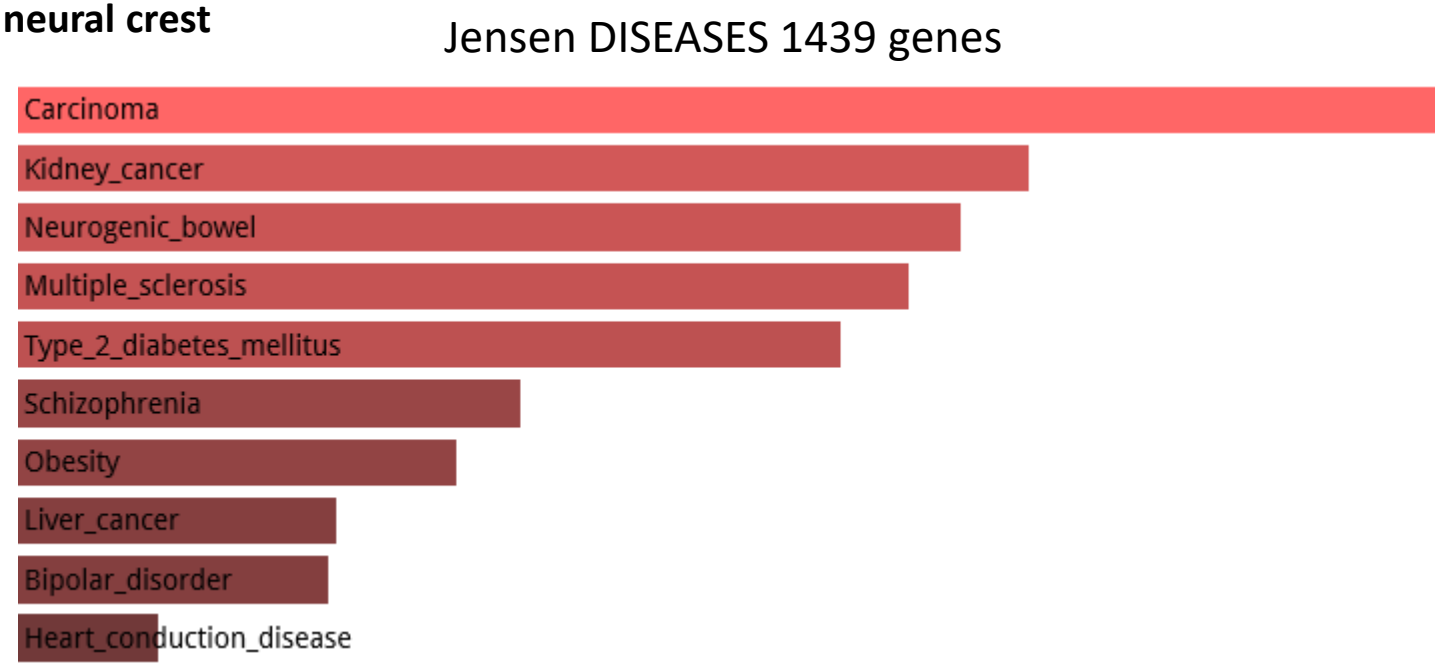
ARCHS4 Tissues 1439 genes

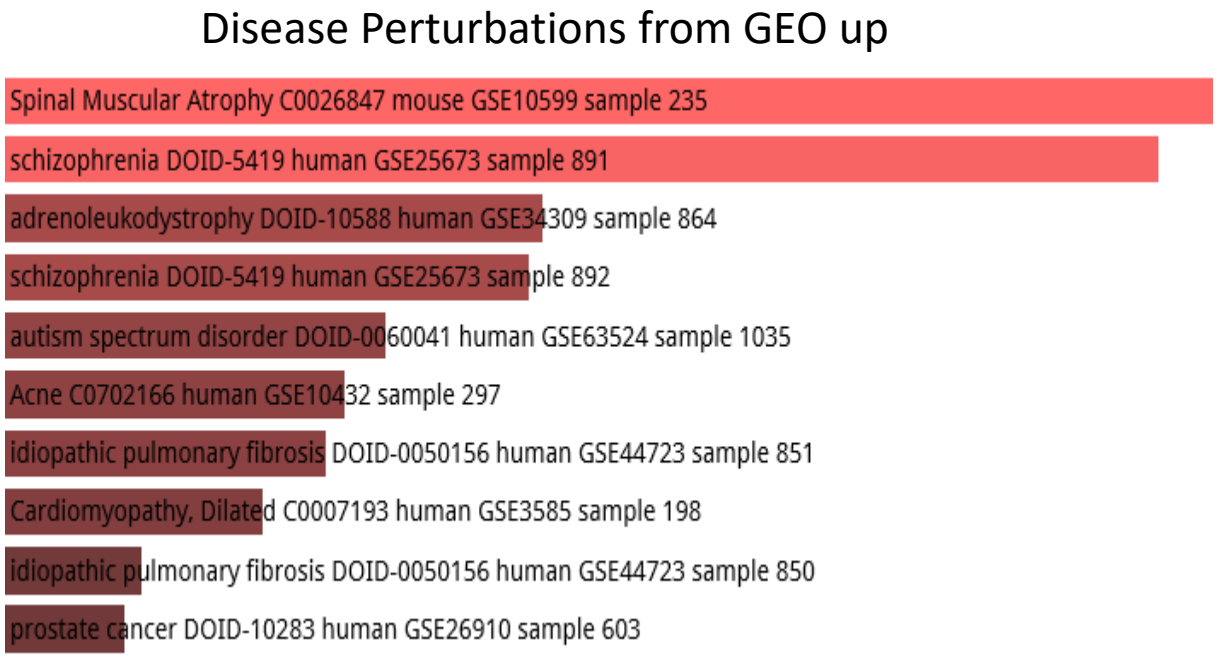


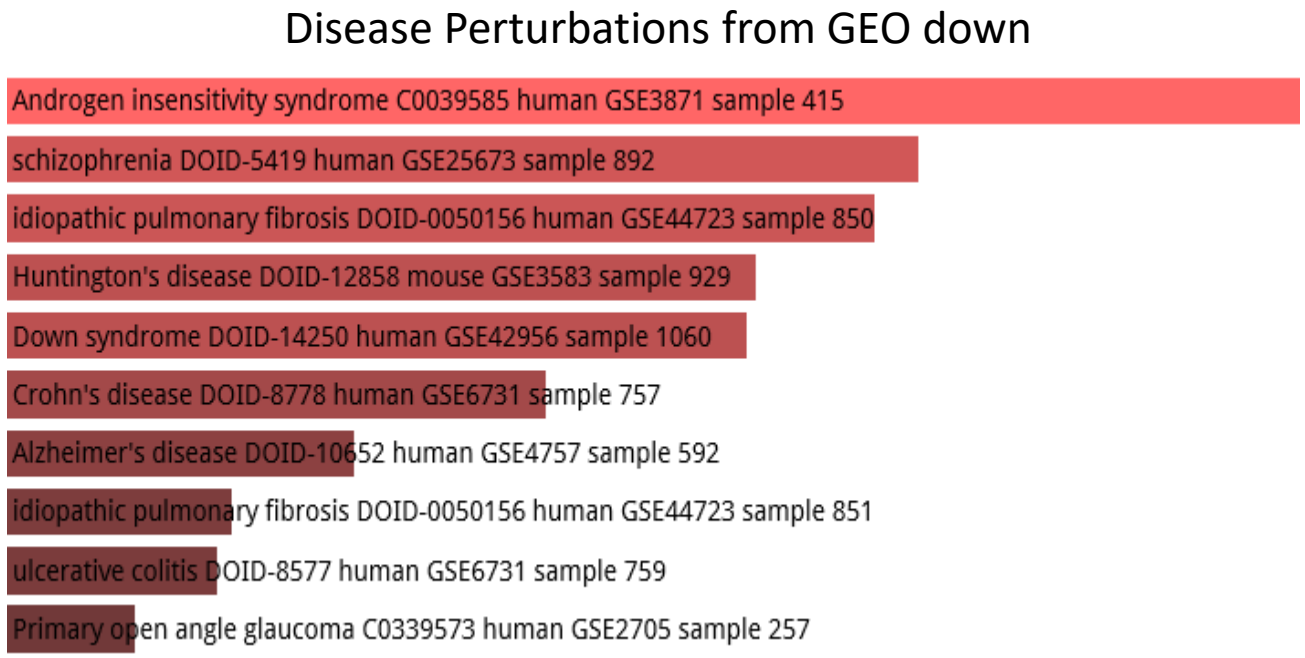
Jensen TISSUES 1439 genes



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



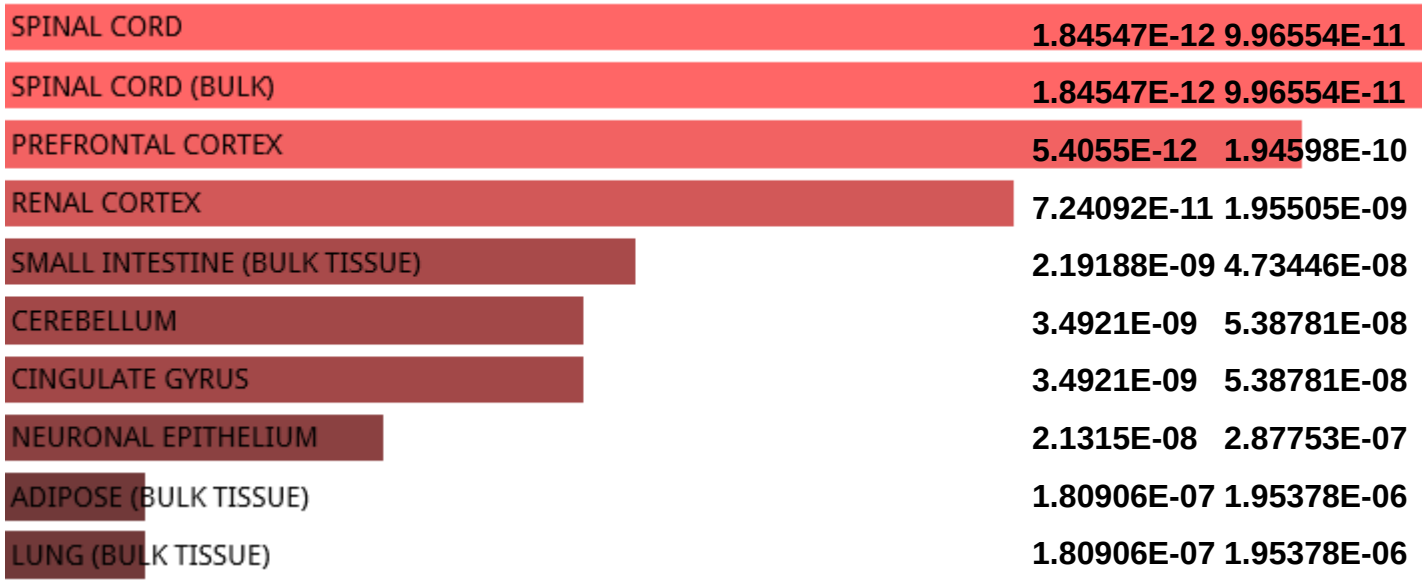




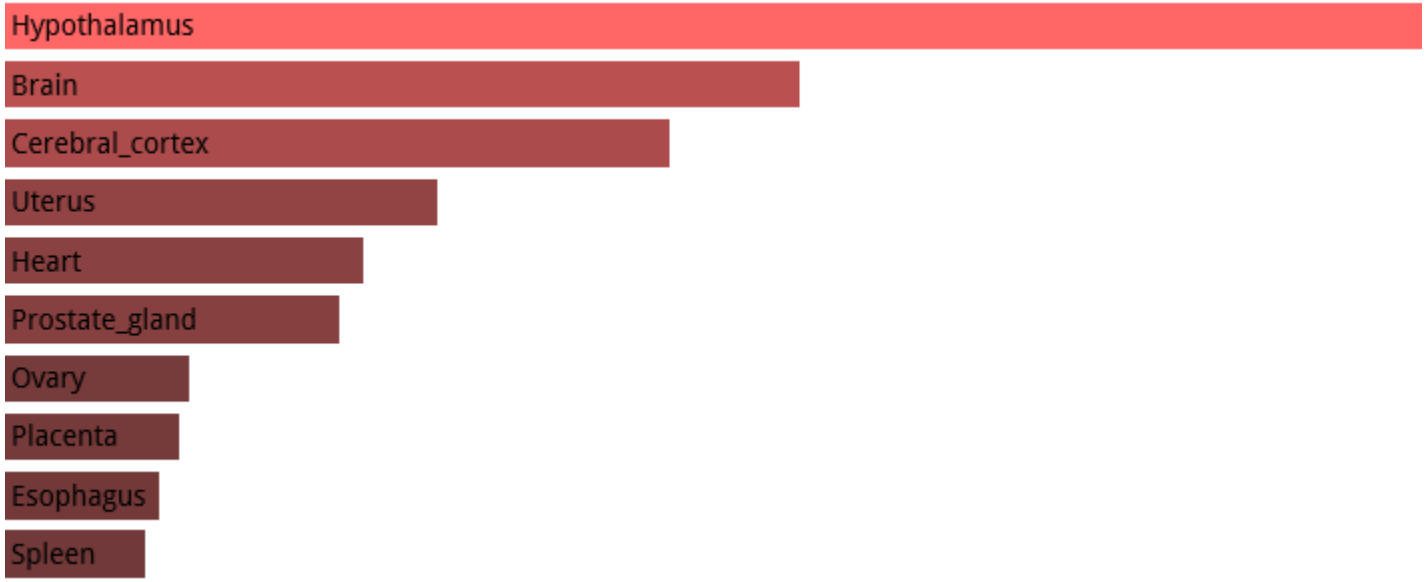
1,000 chimp-biased cranial neural crest cells (CNCCs) enhancers

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

ARCHS4 Tissues 1445 genes

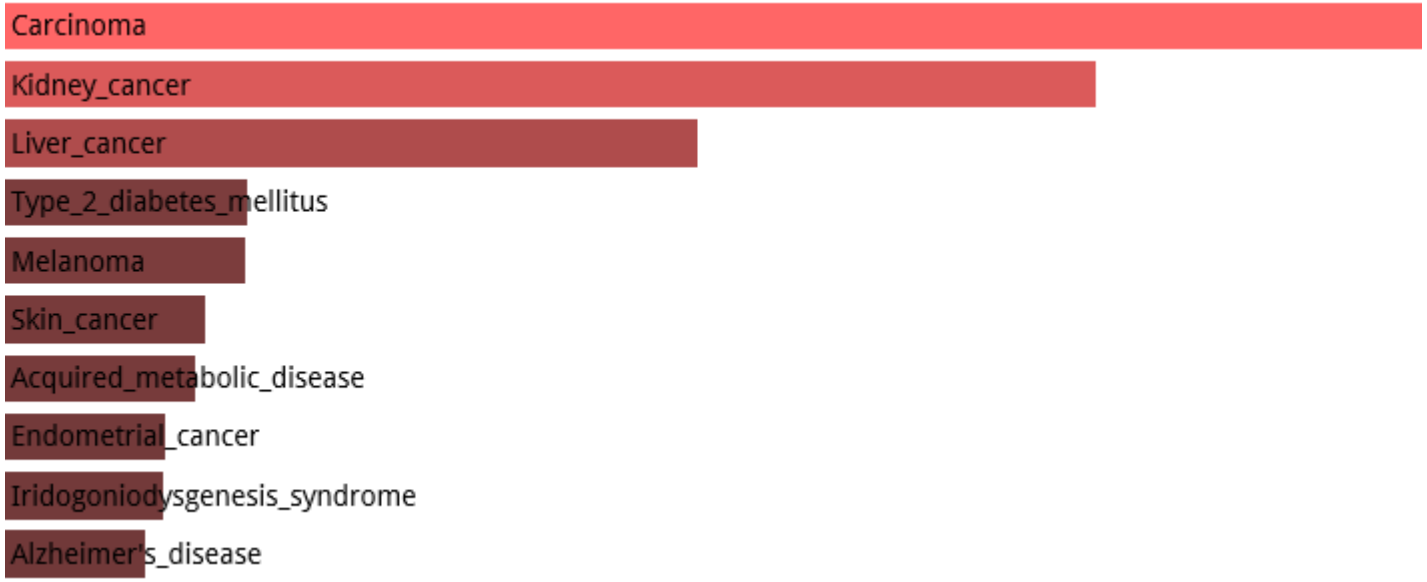


Jensen TISSUES 1445 genes

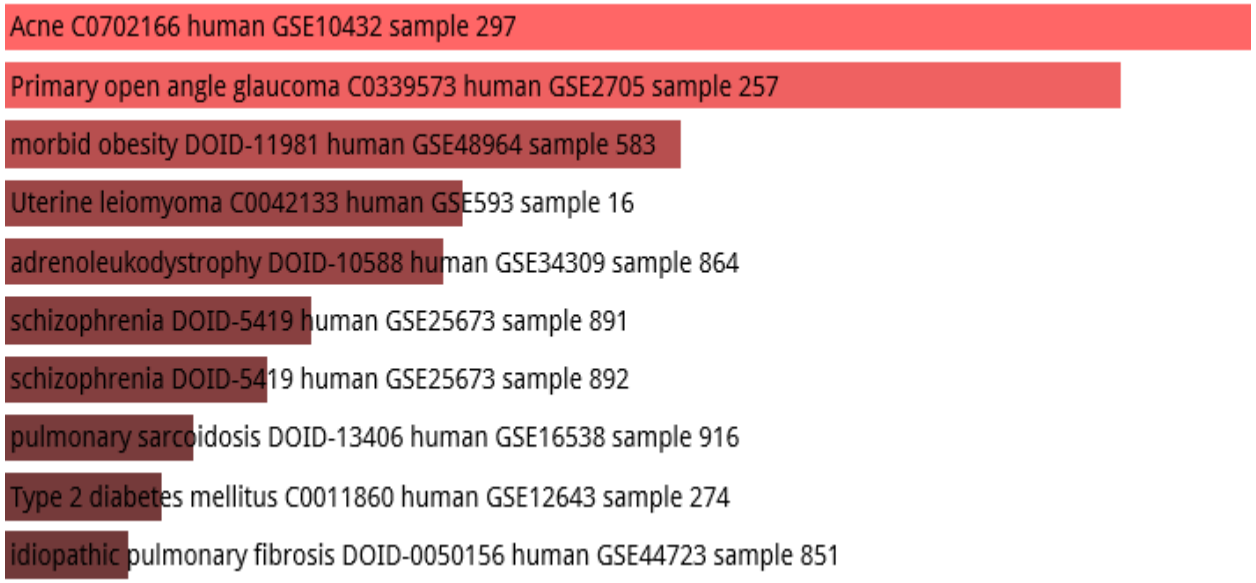


1,000 chimp-biased cranial neural crest cells (CNCCs) enhancers

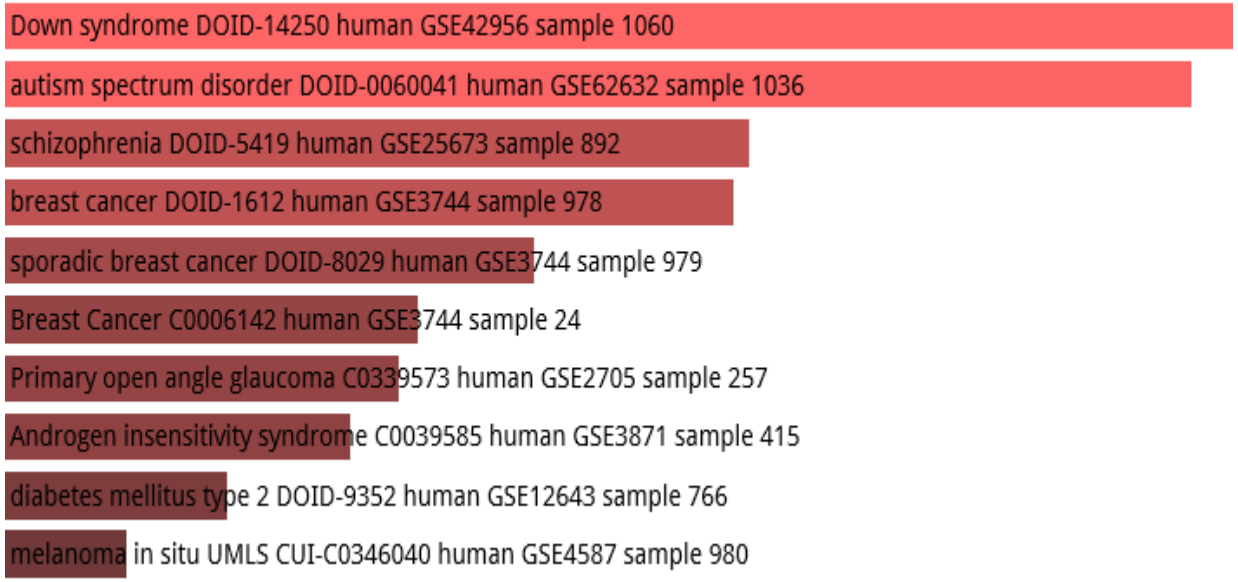
Jensen DISEASES 1445 genes



Disease Perturbations from GEO up



Disease Perturbations from GEO down



1,619 human-specific functional hESC enhancers

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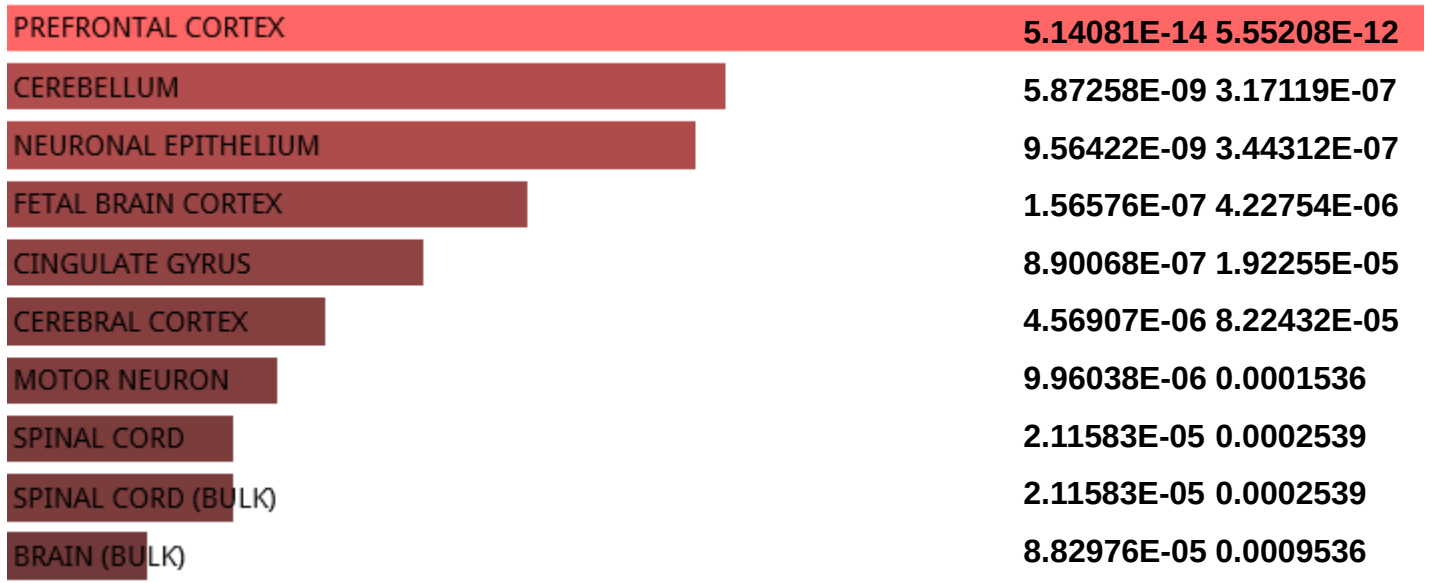
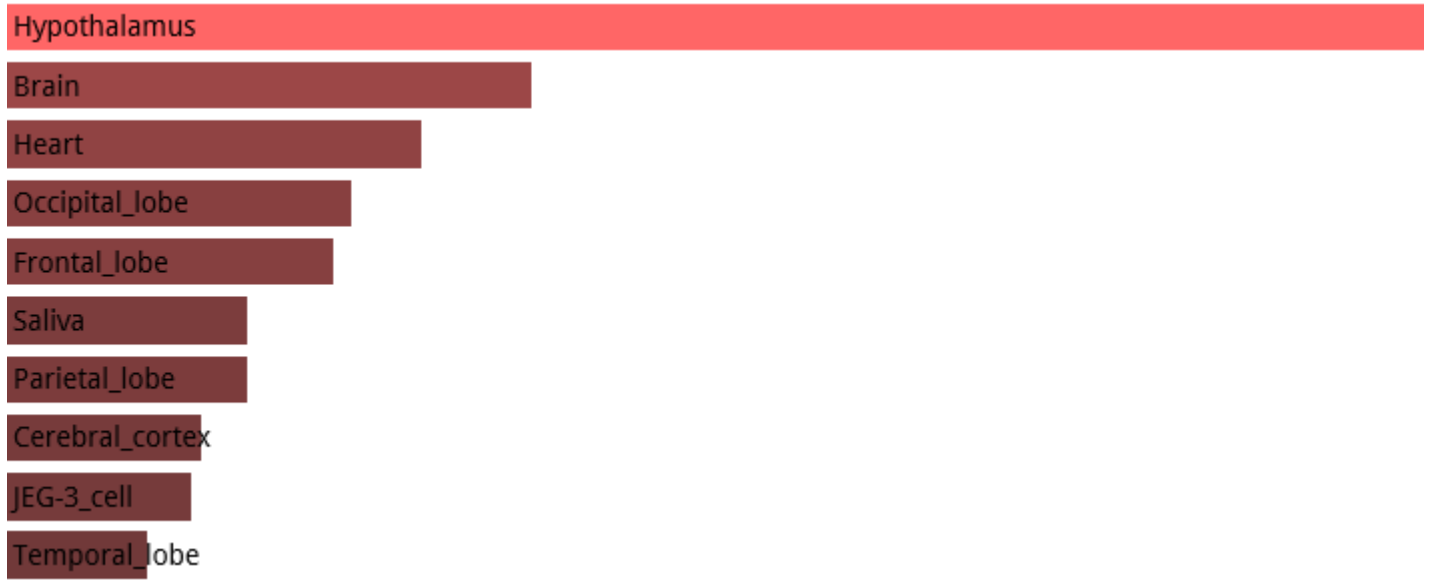
ARCHS4 Tissues 1214 genes

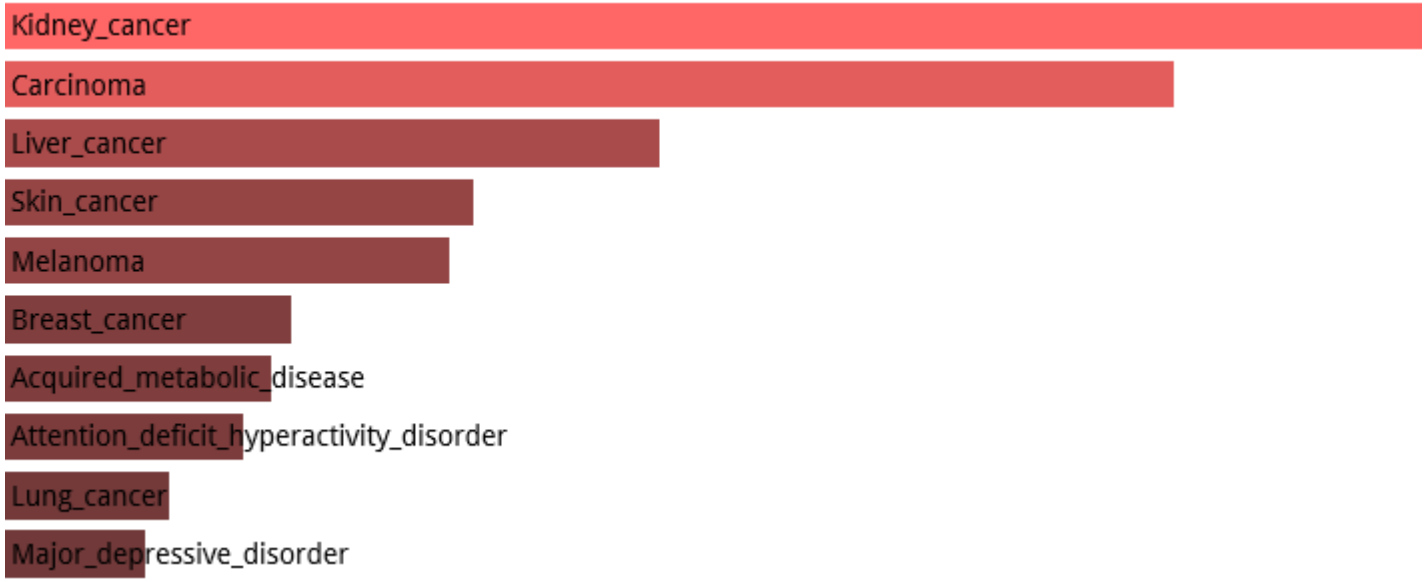


Jensen TISSUES 1214 genes

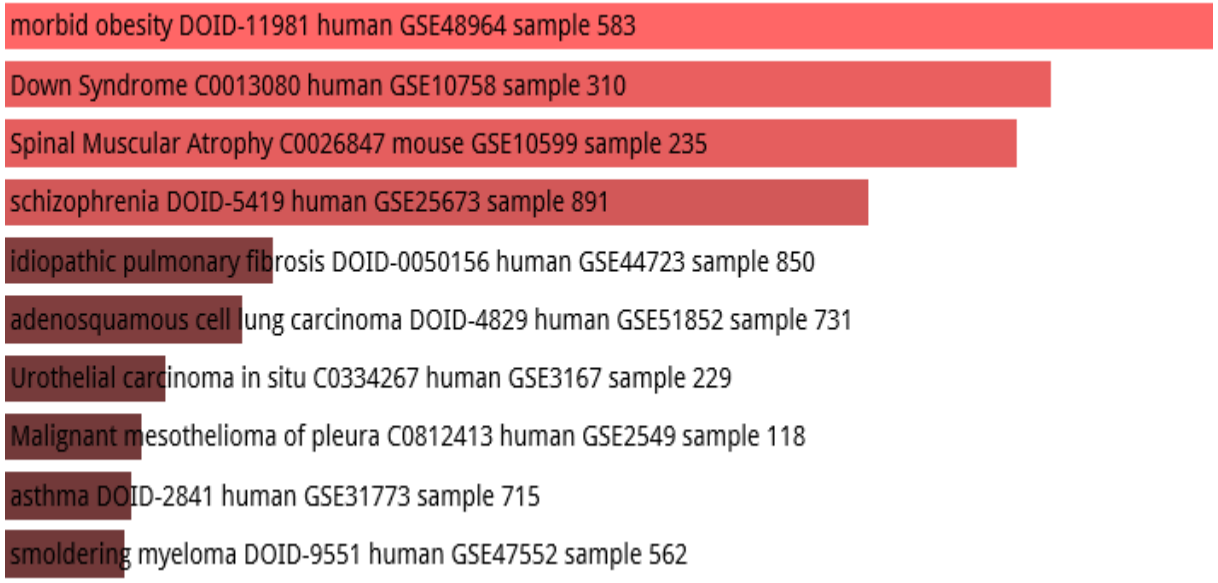


1,619 human-specific functional hESC enhancers

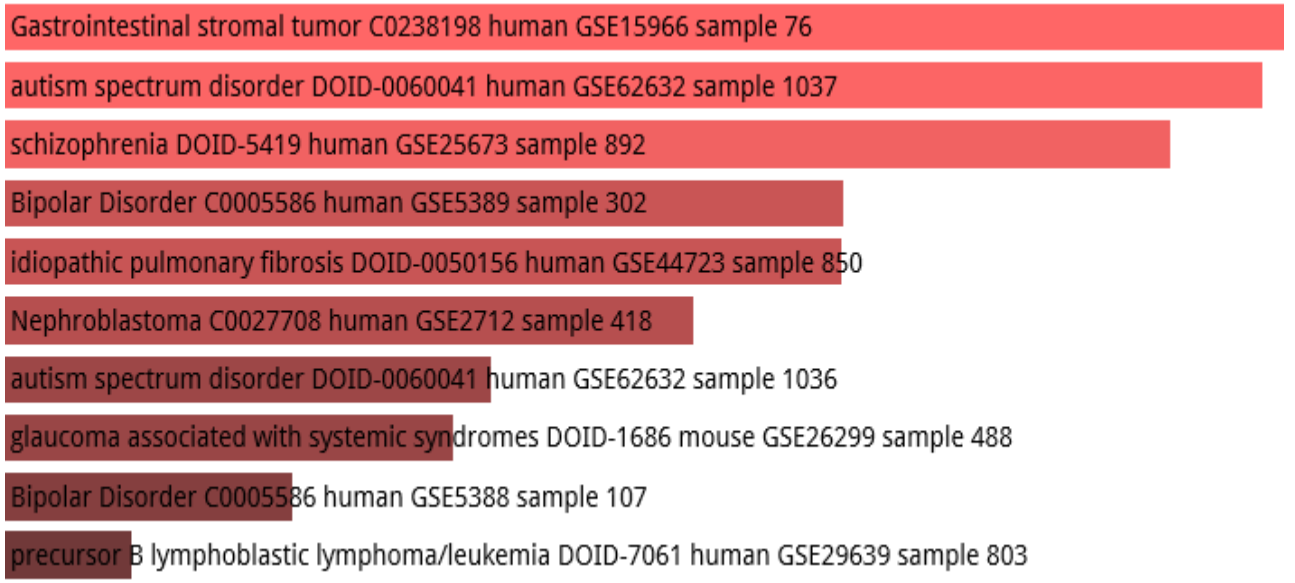
Jensen DISEASES 1214 genes



Disease Perturbations from GEO up 1214 genes

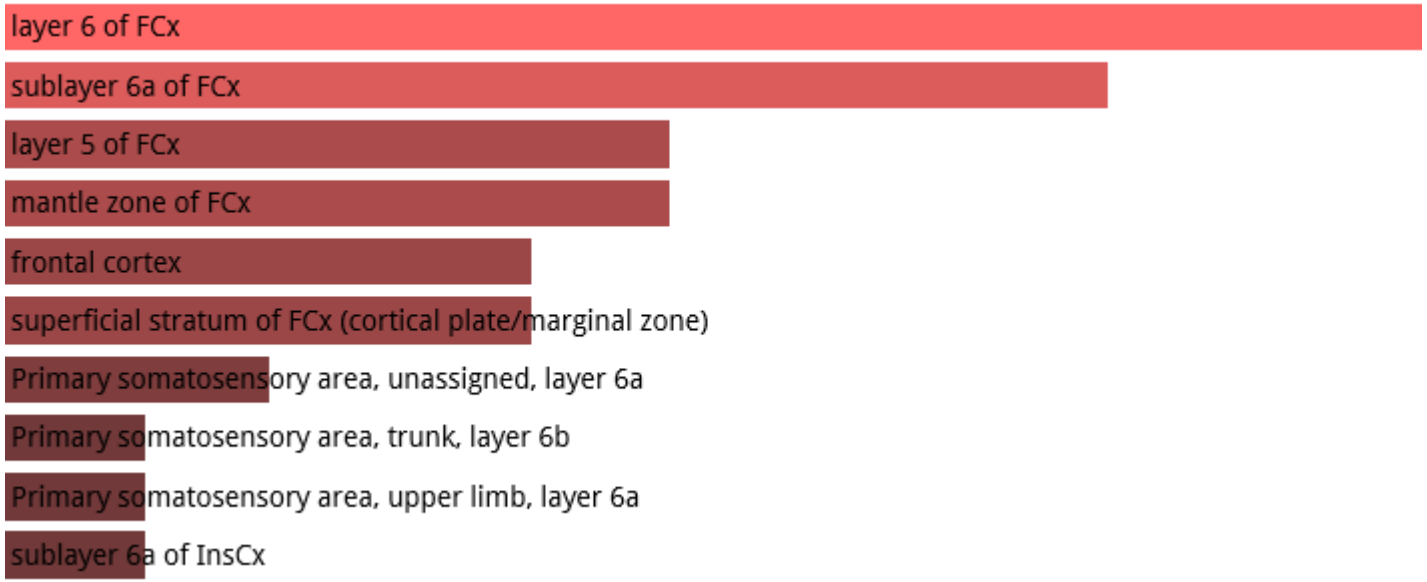


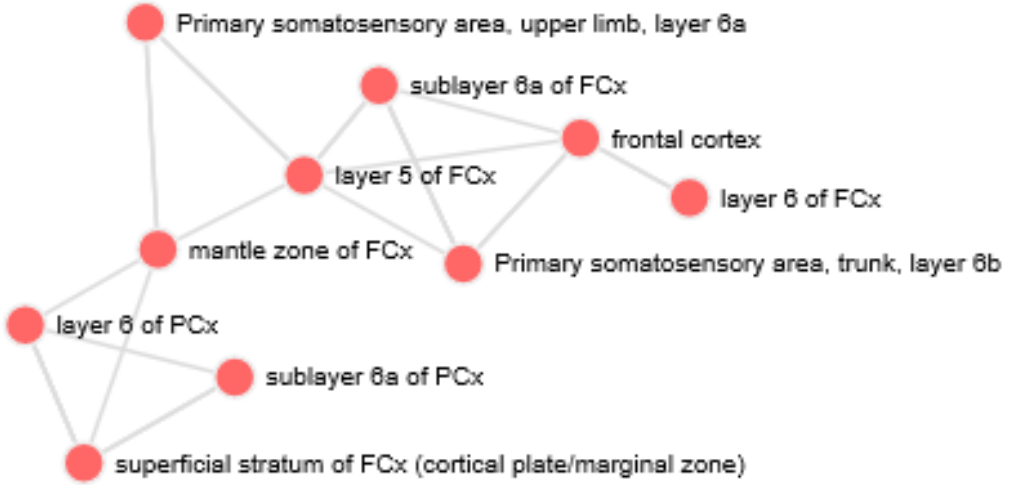
Disease Perturbations from GEO down 1214 genes



1,619 human-specific functional hESC enhancers

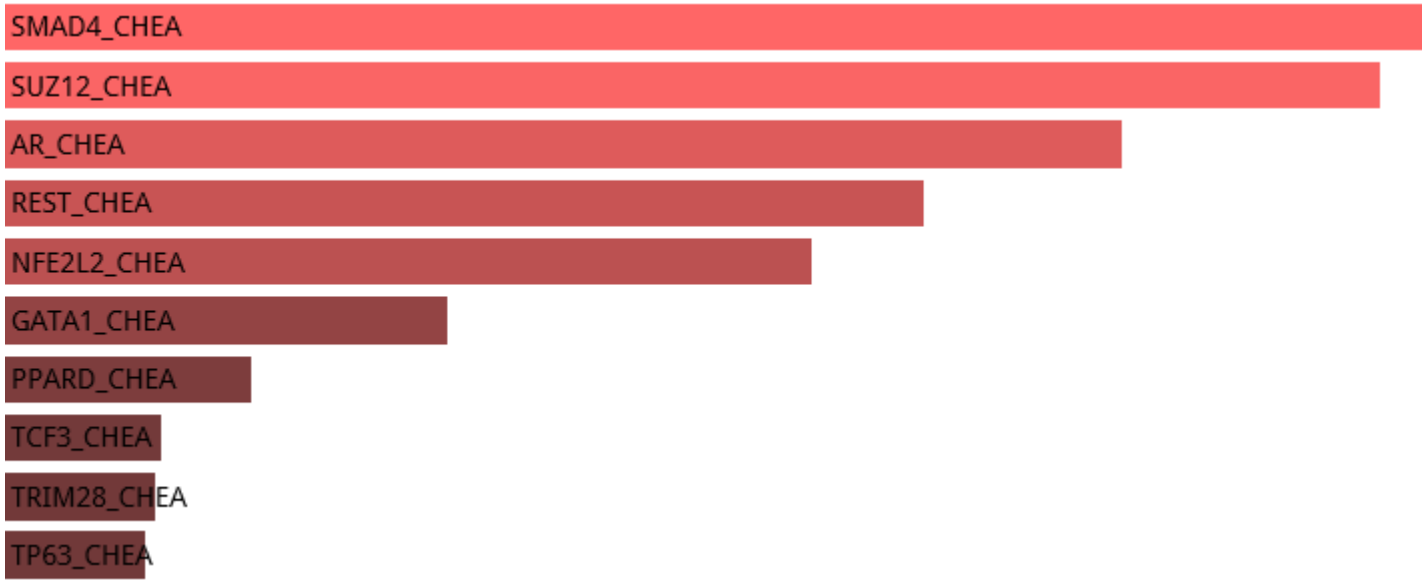
Allen Brain Atlas up 1214 genes



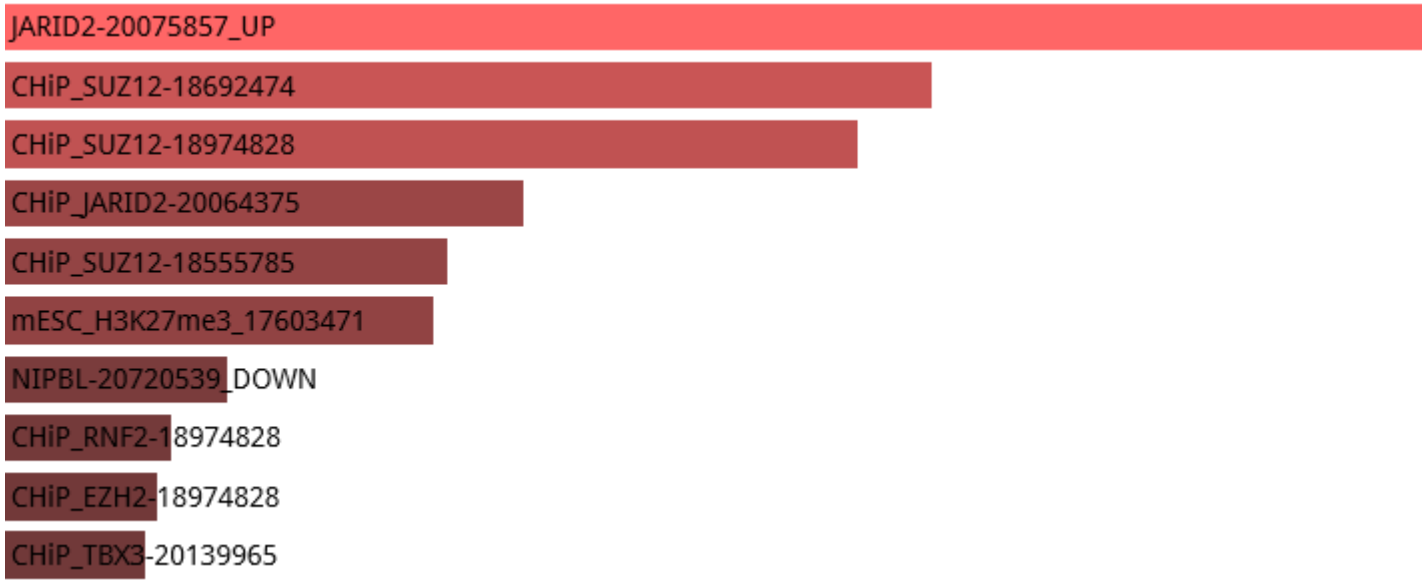


1,619 human-specific functional hESC enhancers

ENCODE and ChEA Consensus TFs from ChIP-X 1214 genes

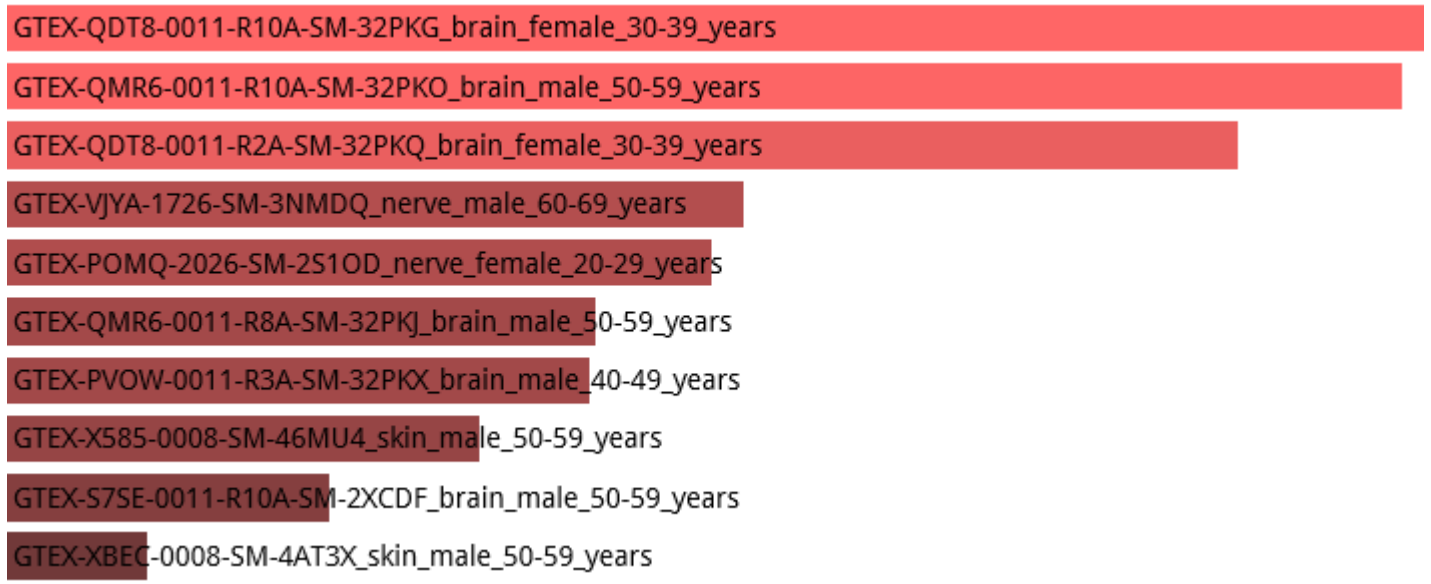


ESCAPE 1214 genes

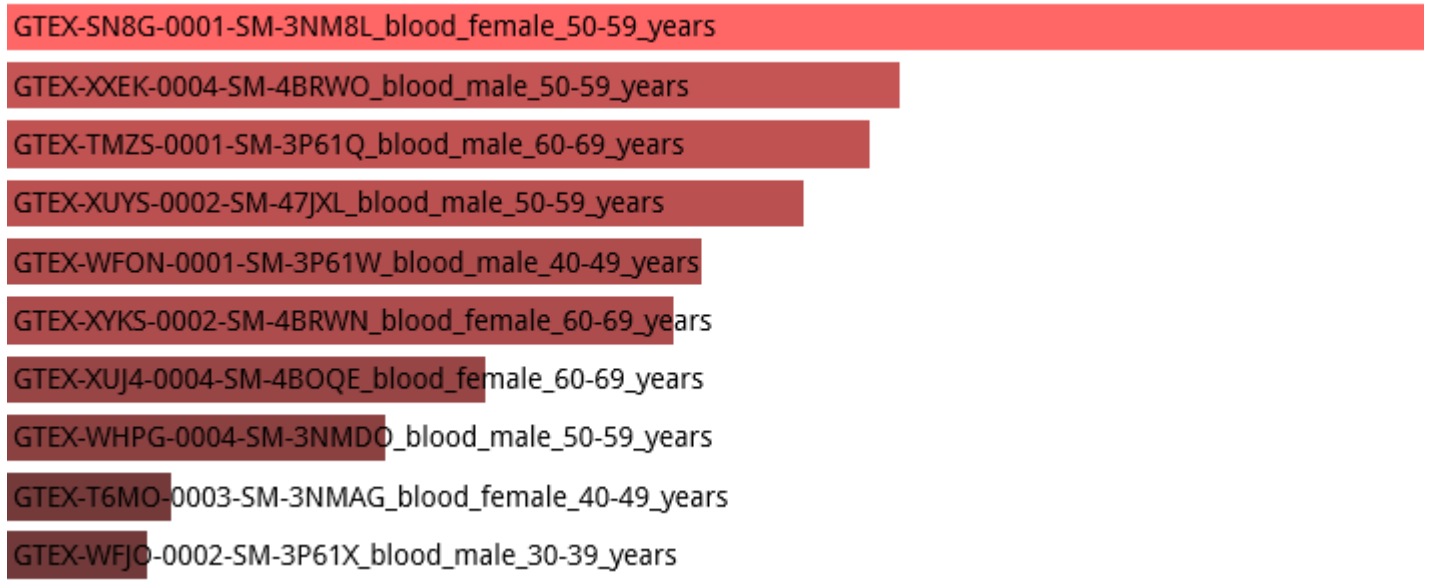


1,619 human-specific functional hESC enhancers

GTEx Tissue Sample Gene Expression Profiles up 1214 genes



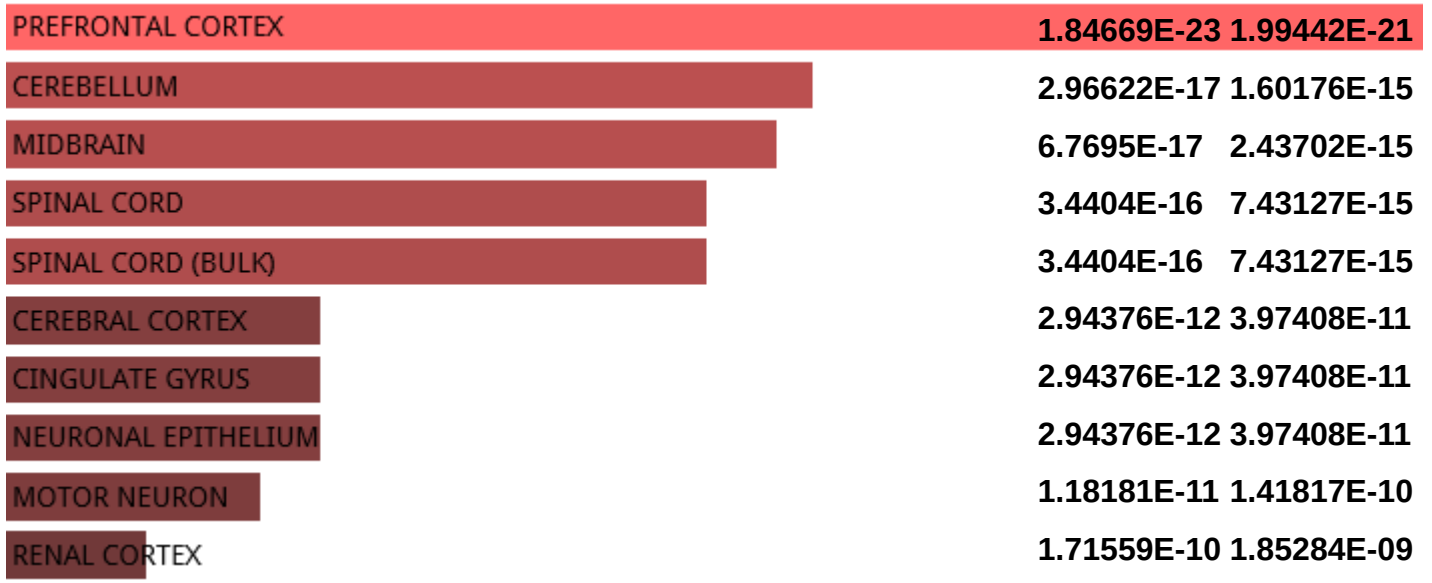
GTEx Tissue Sample Gene Expression Profiles down 1214 genes



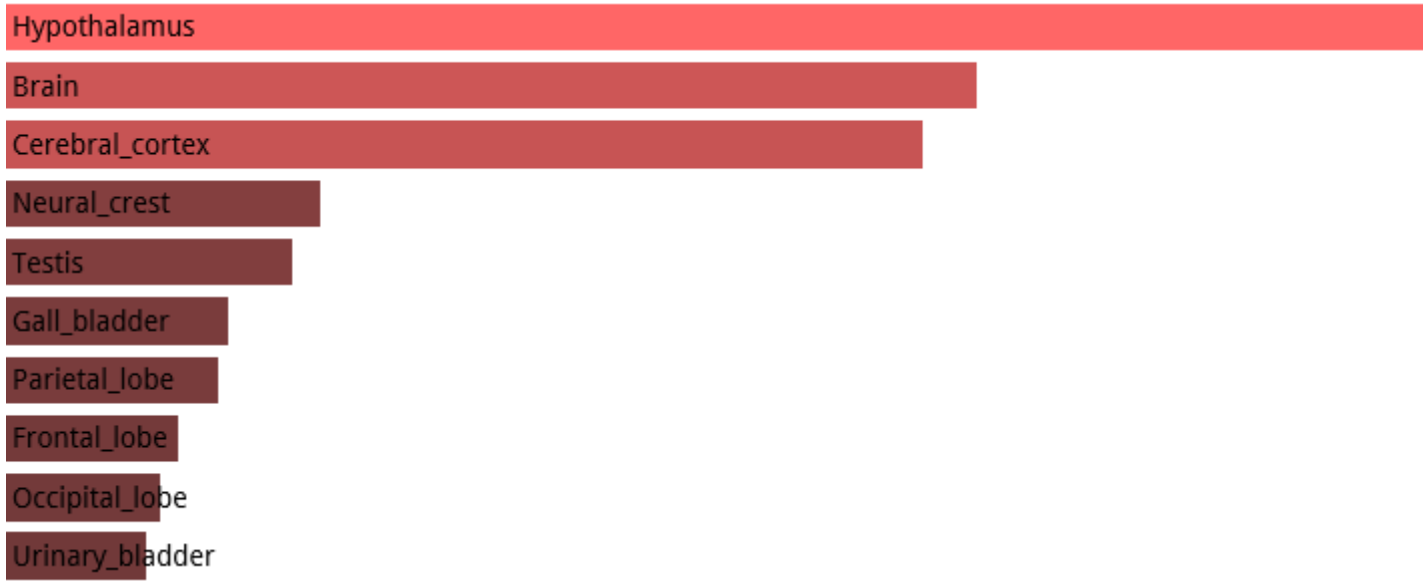
524 human-specific DHS

524 human-specific DHS

ARCHS4 Tissues 747 genes

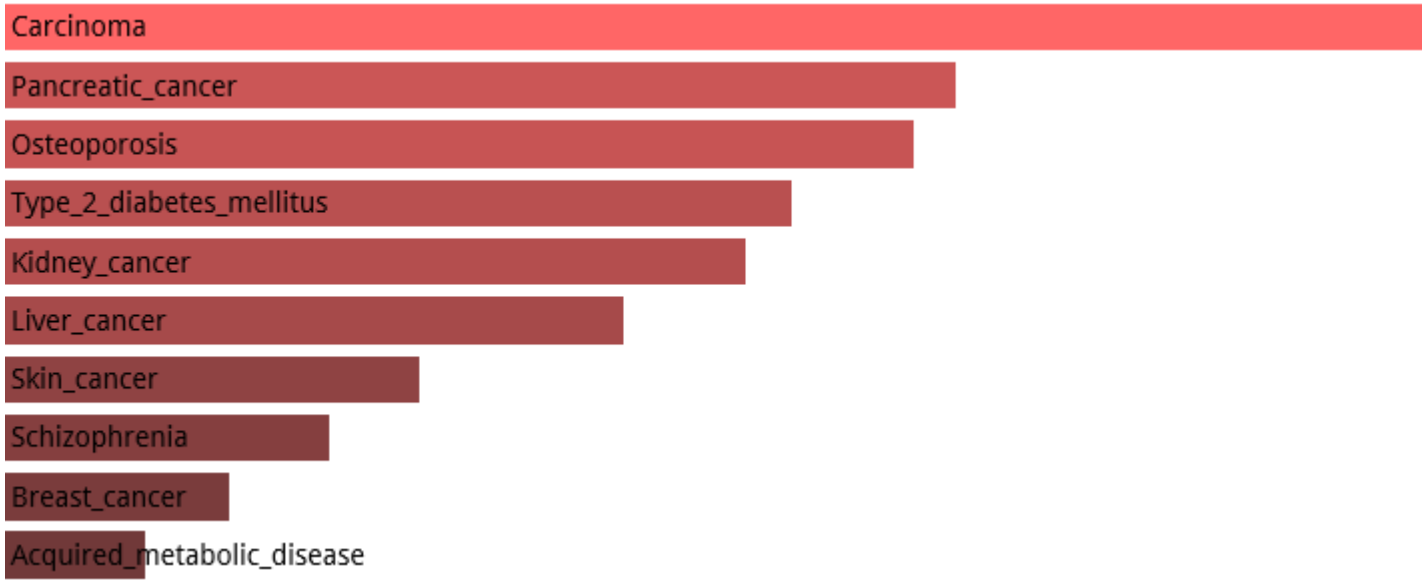


Jensen TISSUES 747 genes

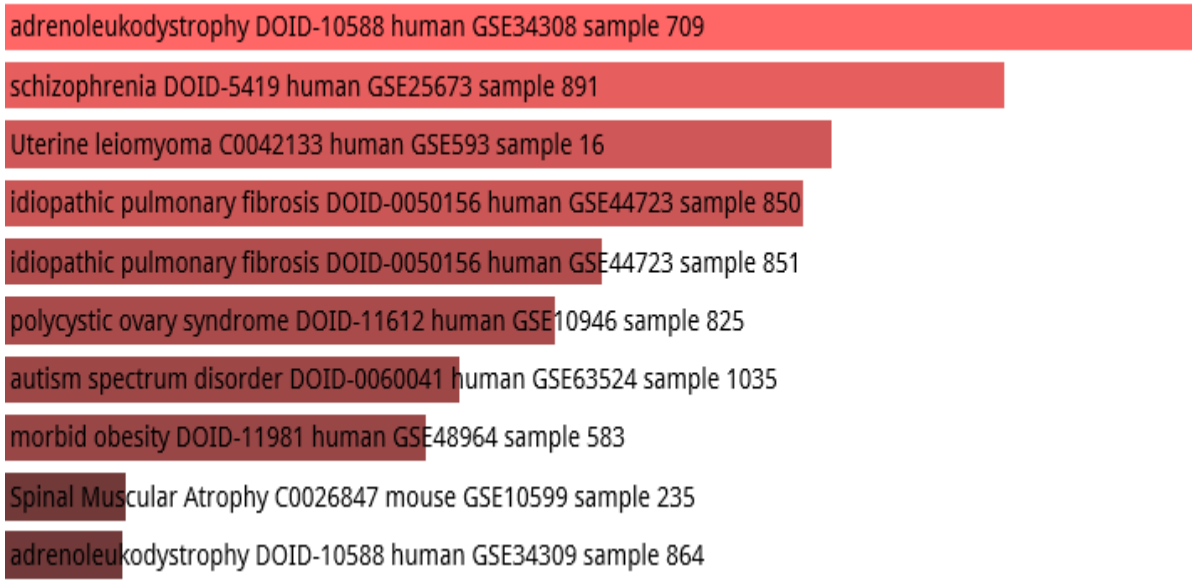


524 human-specific DHS

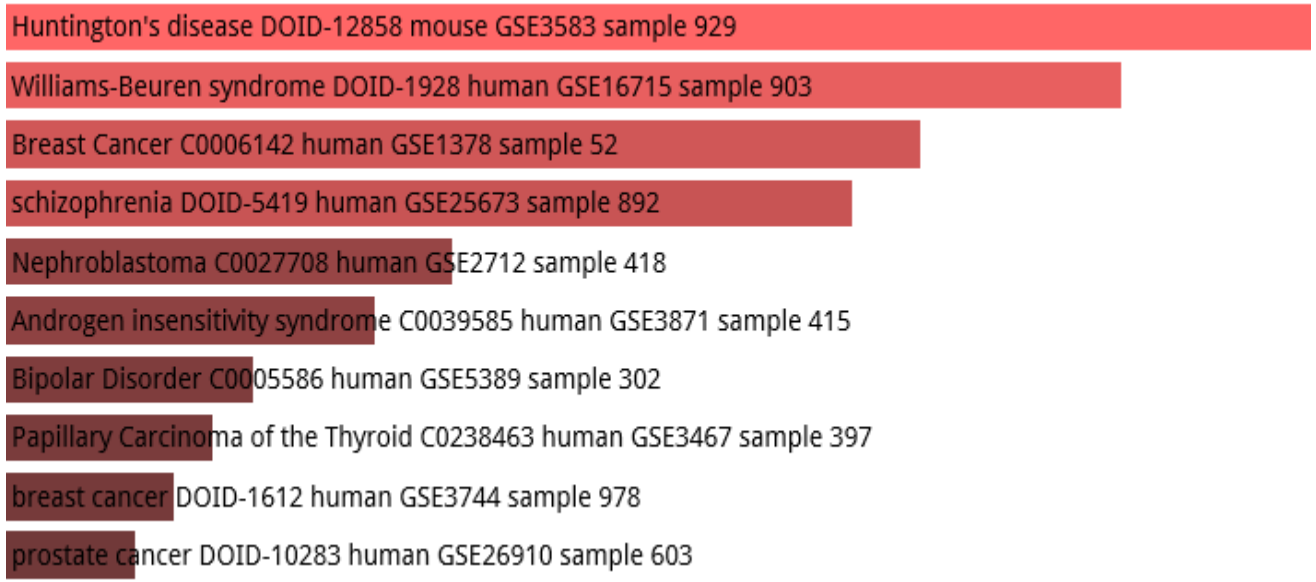
Jensen DISEASES 747 genes



Disease Perturbations from GEO up 747 genes



Disease Perturbations from GEO down 747 genes

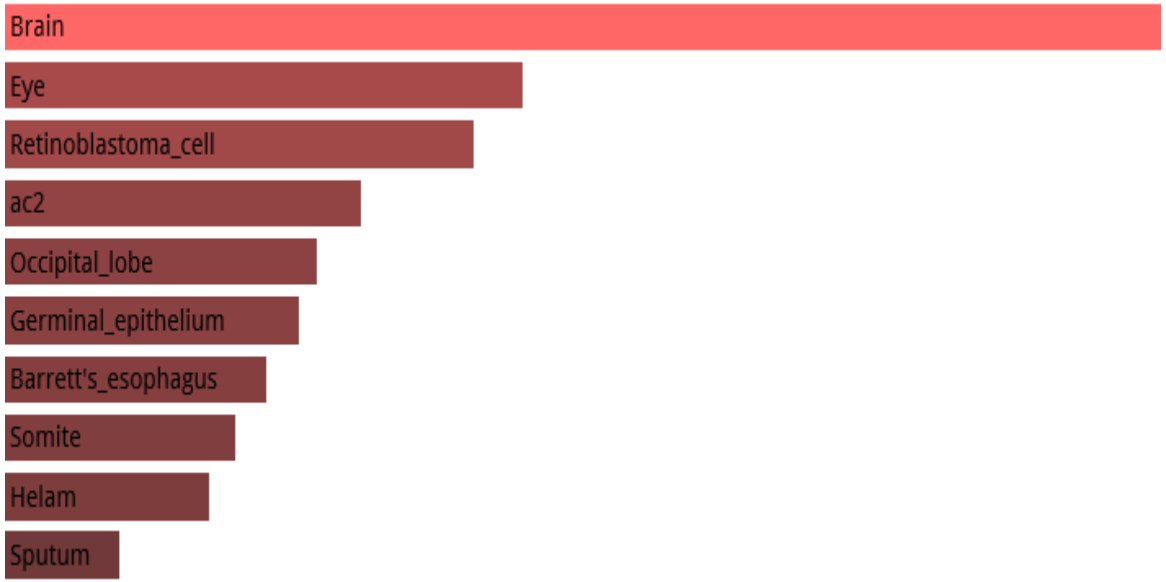


1,279 human-specific STR contractions

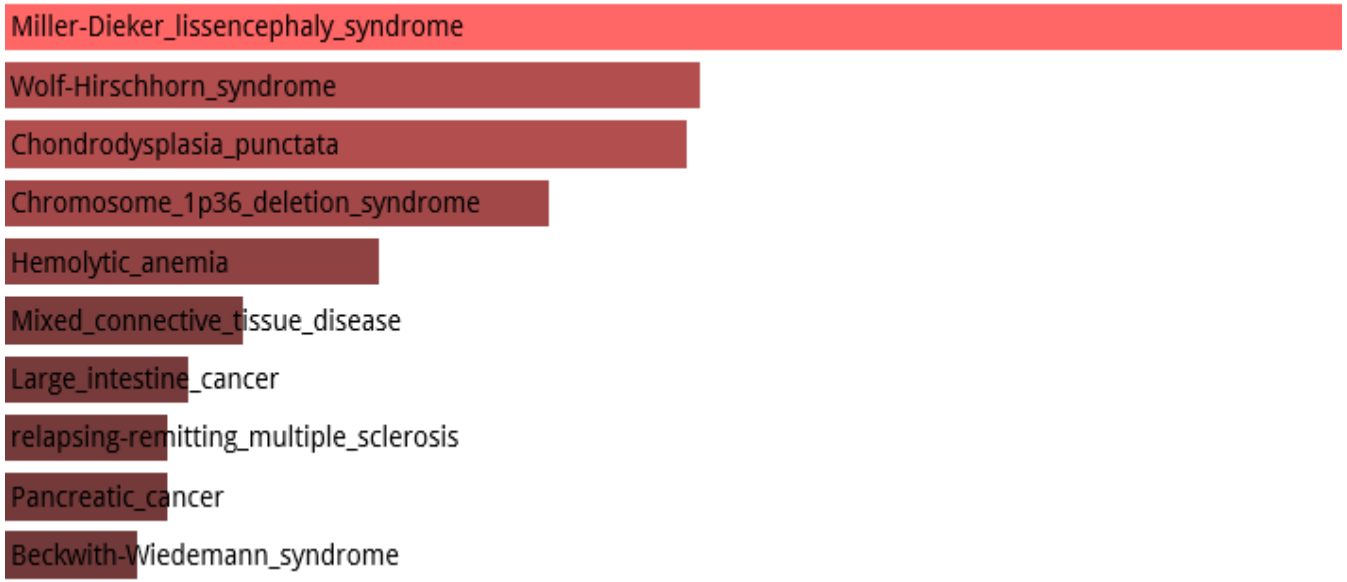
ARCHS4 Tissues 973 genes

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

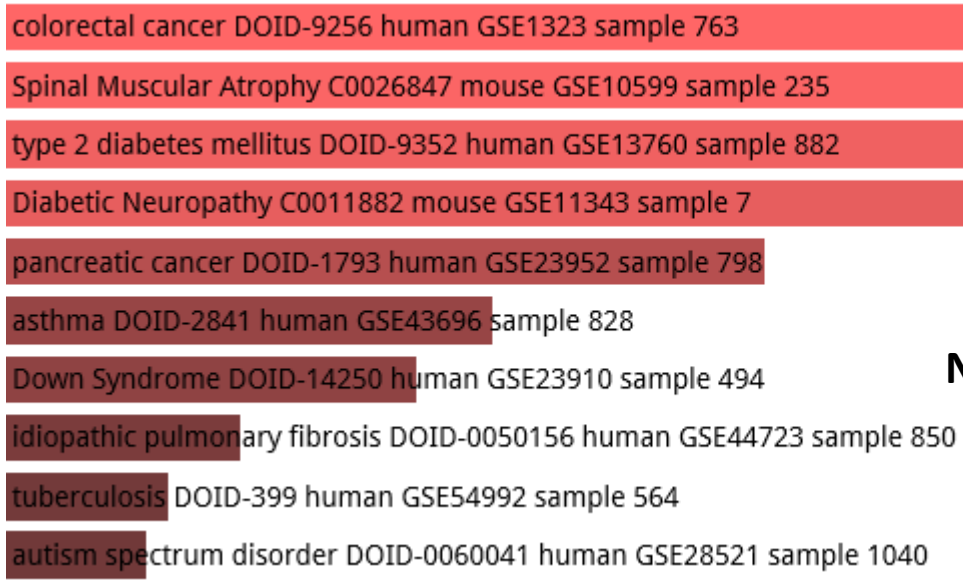
Jensen TISSUES 973 genes



Jensen DISEASES 973 genes

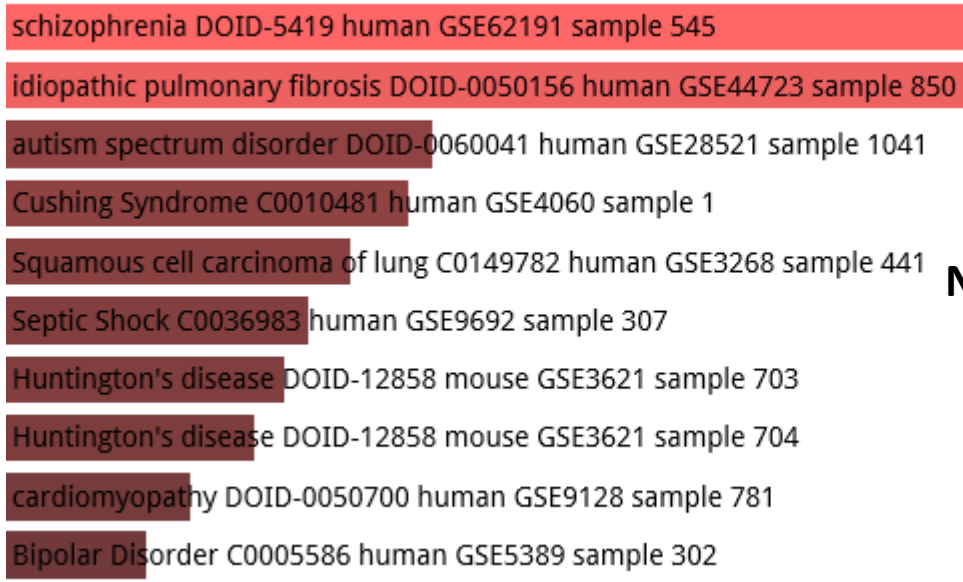


Disease Perturbations from GEO up 973 genes



No significant associations

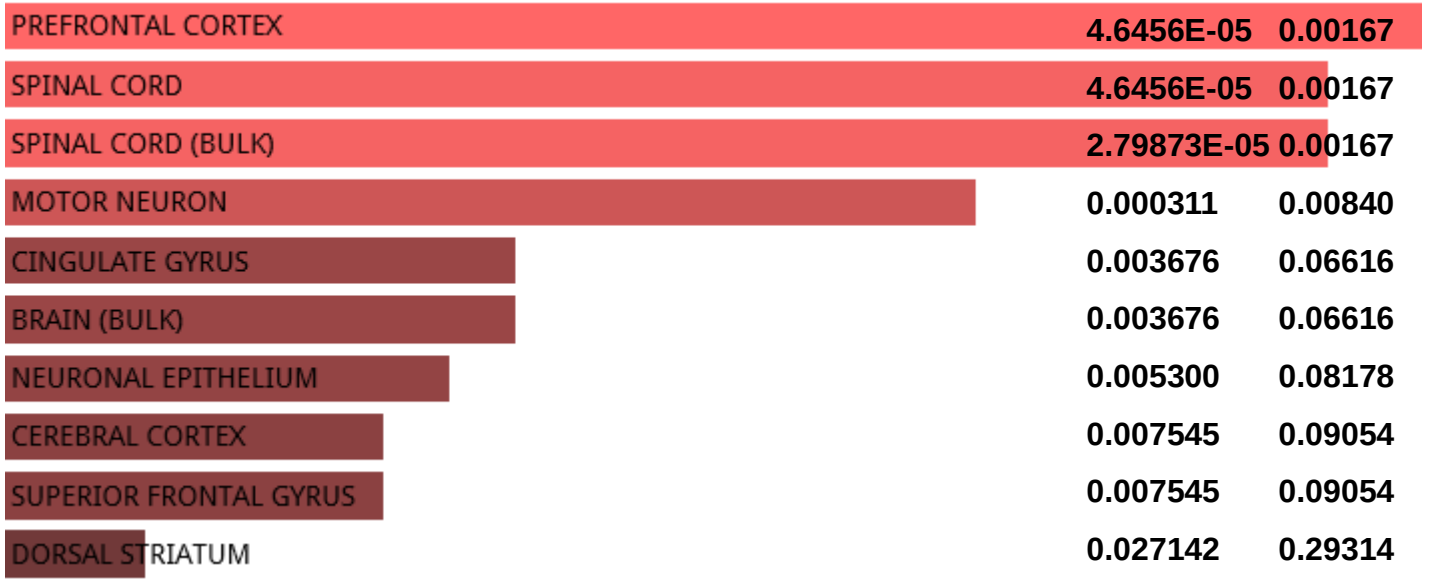
Disease Perturbations from GEO down 973 genes



No significant associations

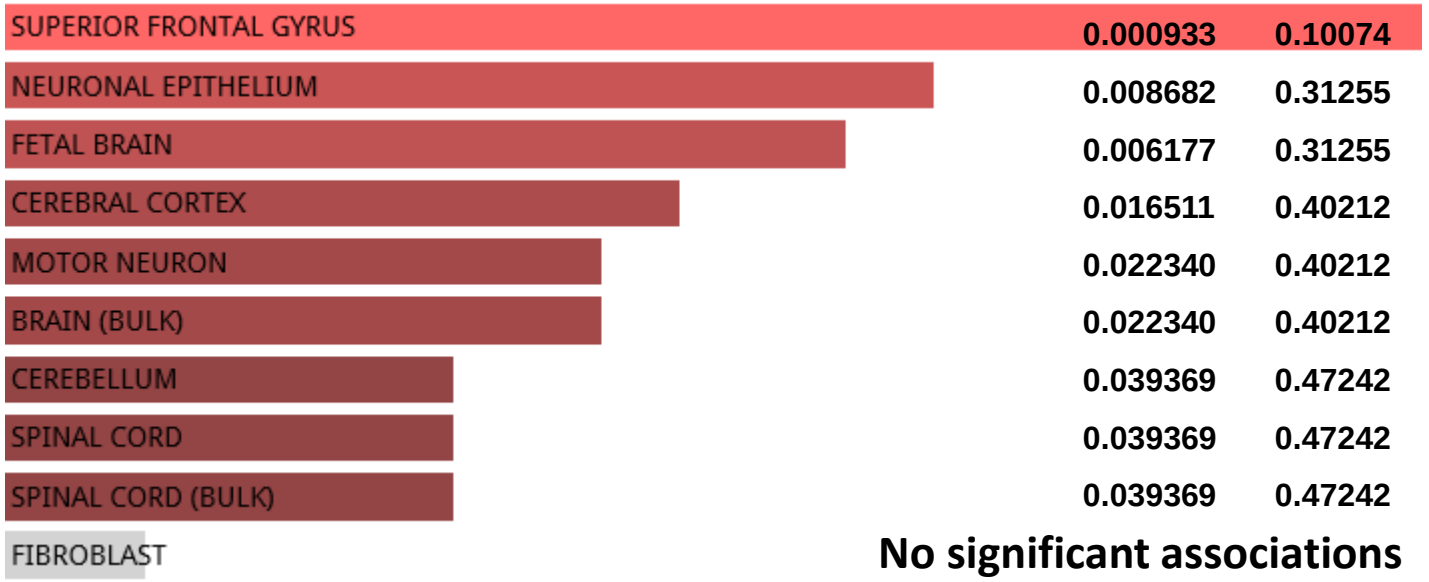
2,118 fixed human-specific regulatory regions (non-hESC DHS)

ARCHS4 Tissues 552 genes



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

ARCHS4 Tissues 578 genes



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

ARCHS4 Tissues random 6000 genes

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

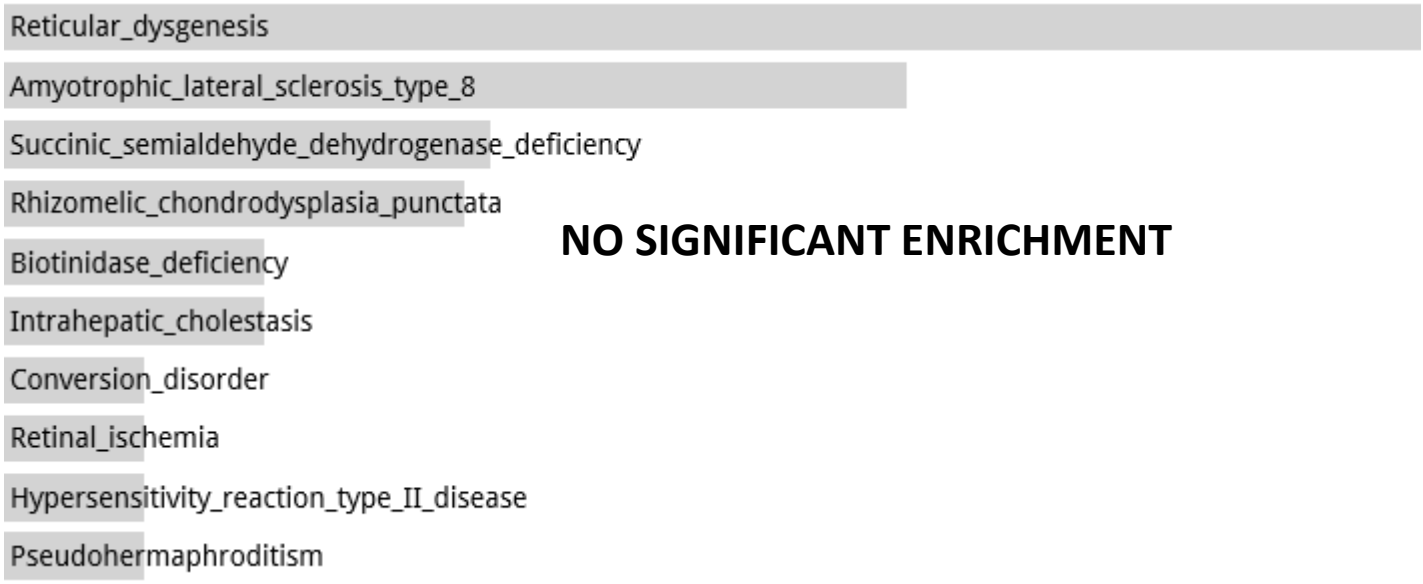
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

NO DATA AVAILABLE

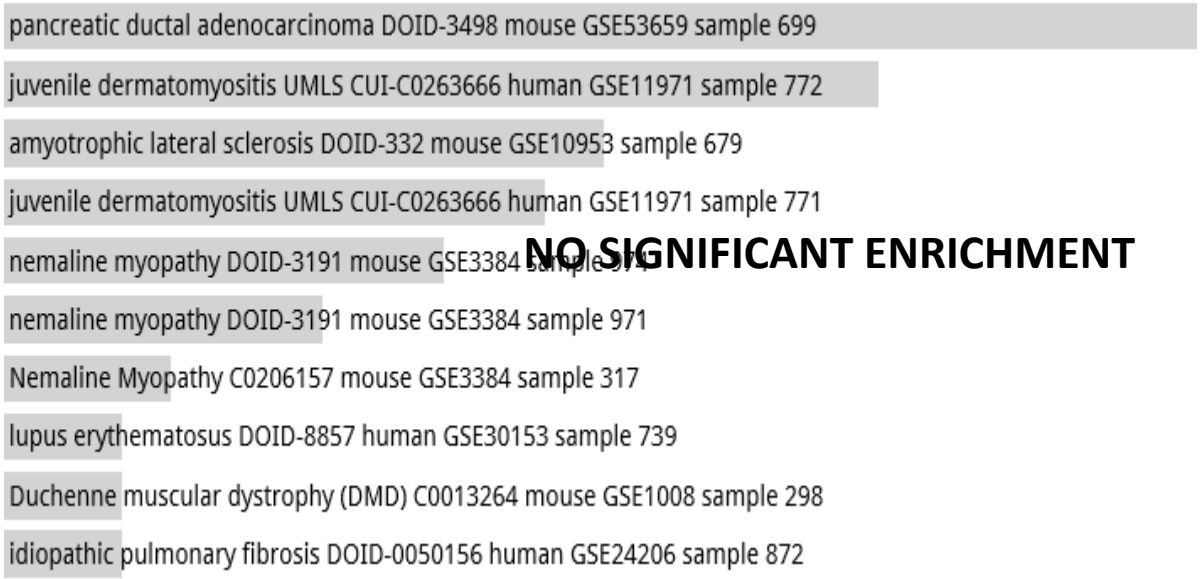
Random Set 1

Jensen DISEASES random 6000 genes



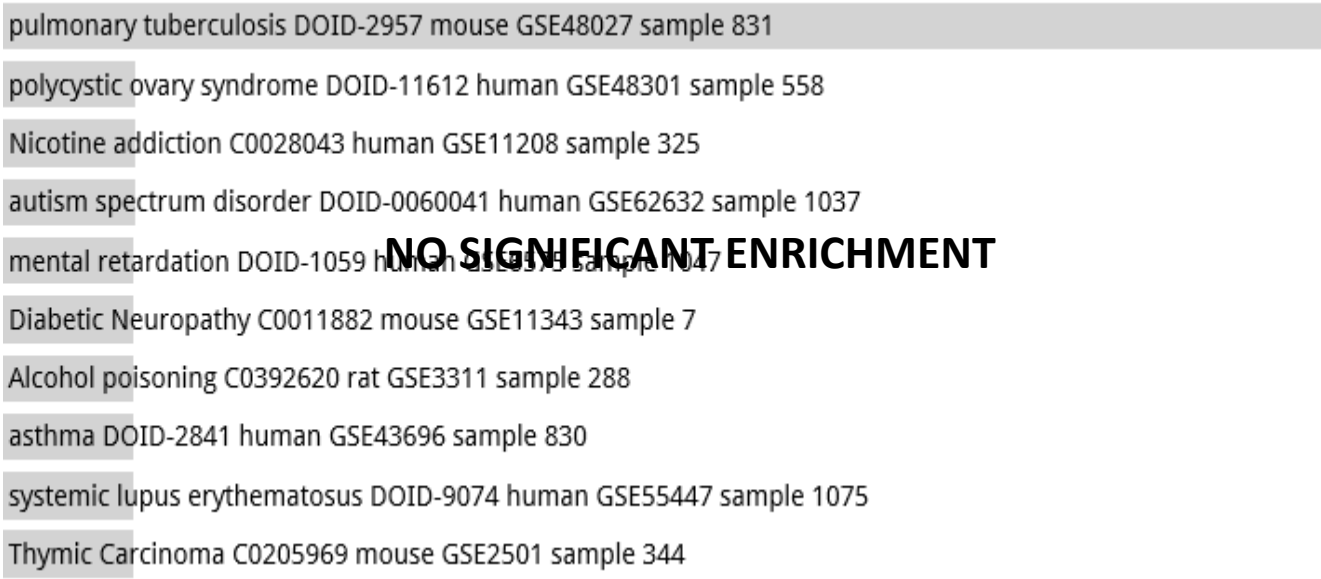
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes



NO SIGNIFICANT ENRICHMENT

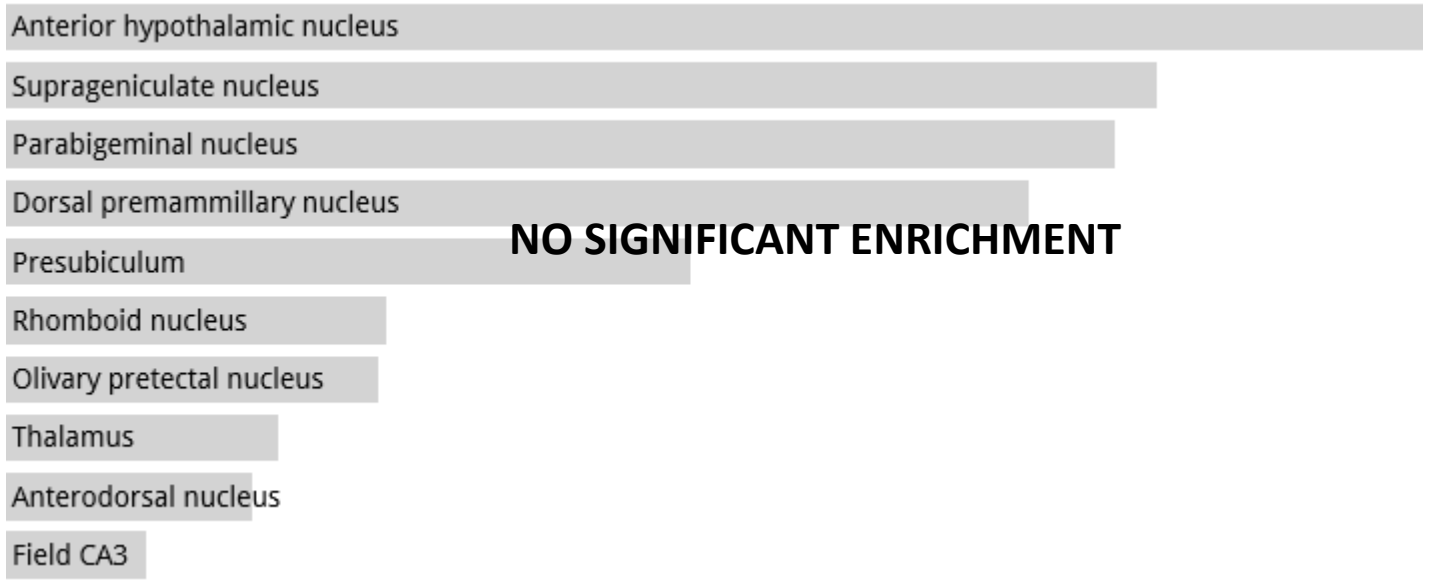
Disease Perturbations from GEO down random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 1

Allen Brain Atlas up random 6000 genes



NO SIGNIFICANT ENRICHMENT

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

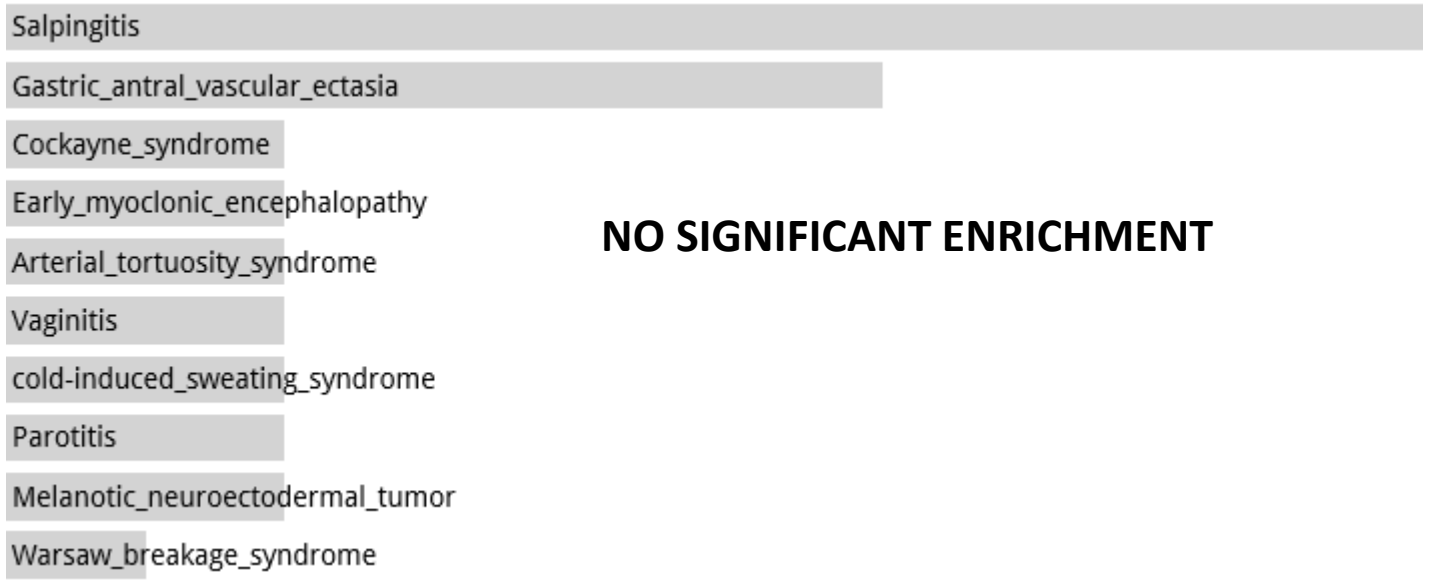
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

NO DATA AVAILABLE

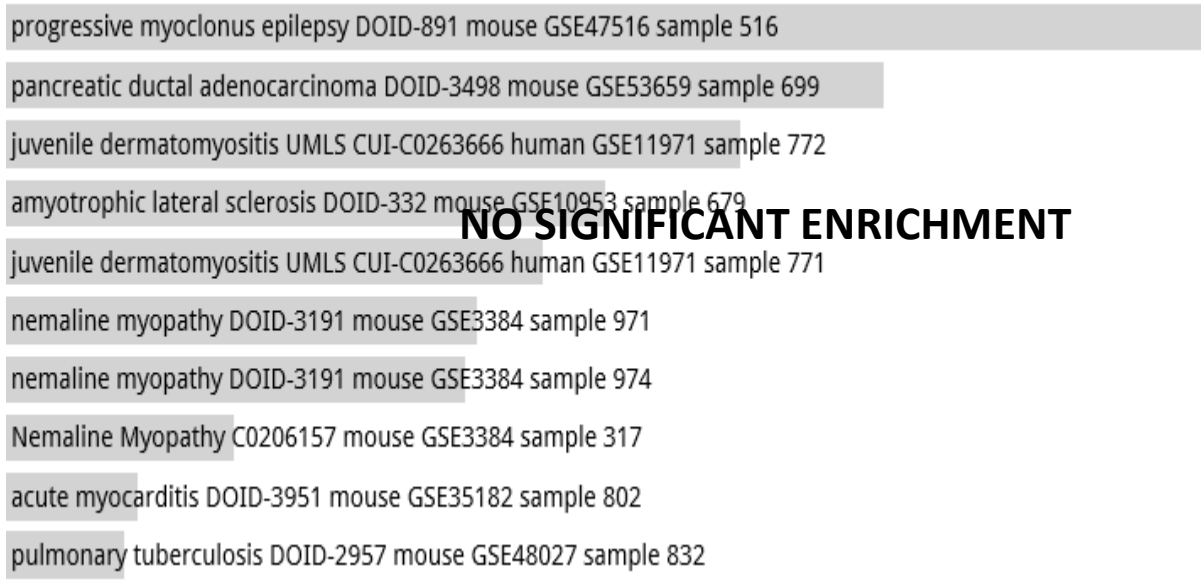
Random Set 2

Jensen DISEASES random 6000 genes



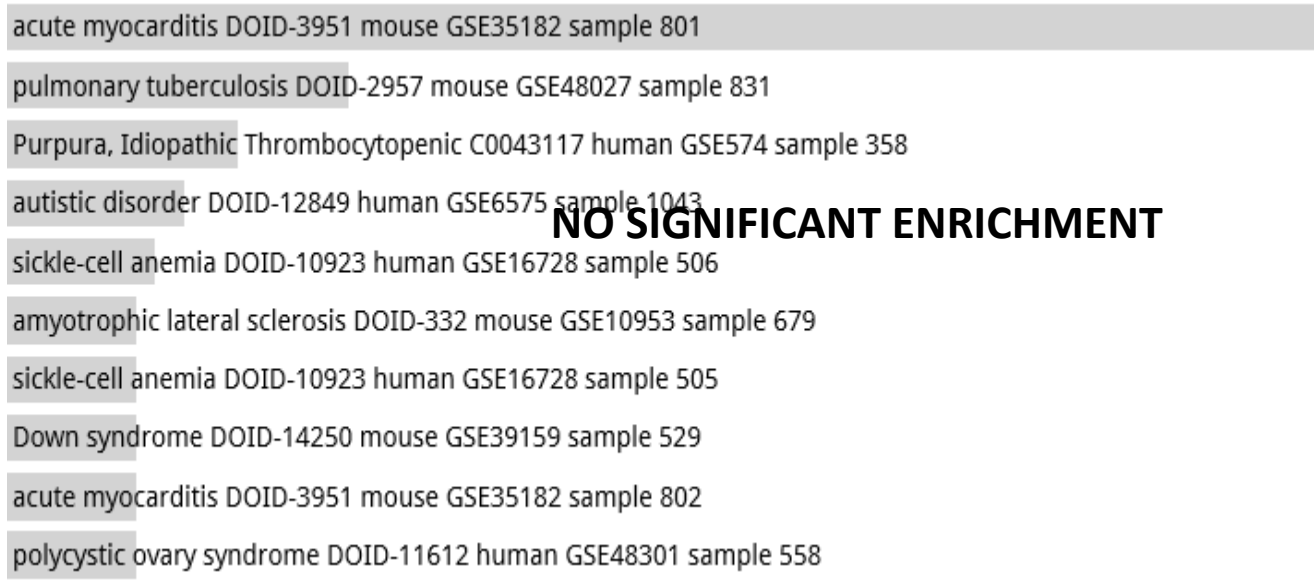
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes



NO SIGNIFICANT ENRICHMENT

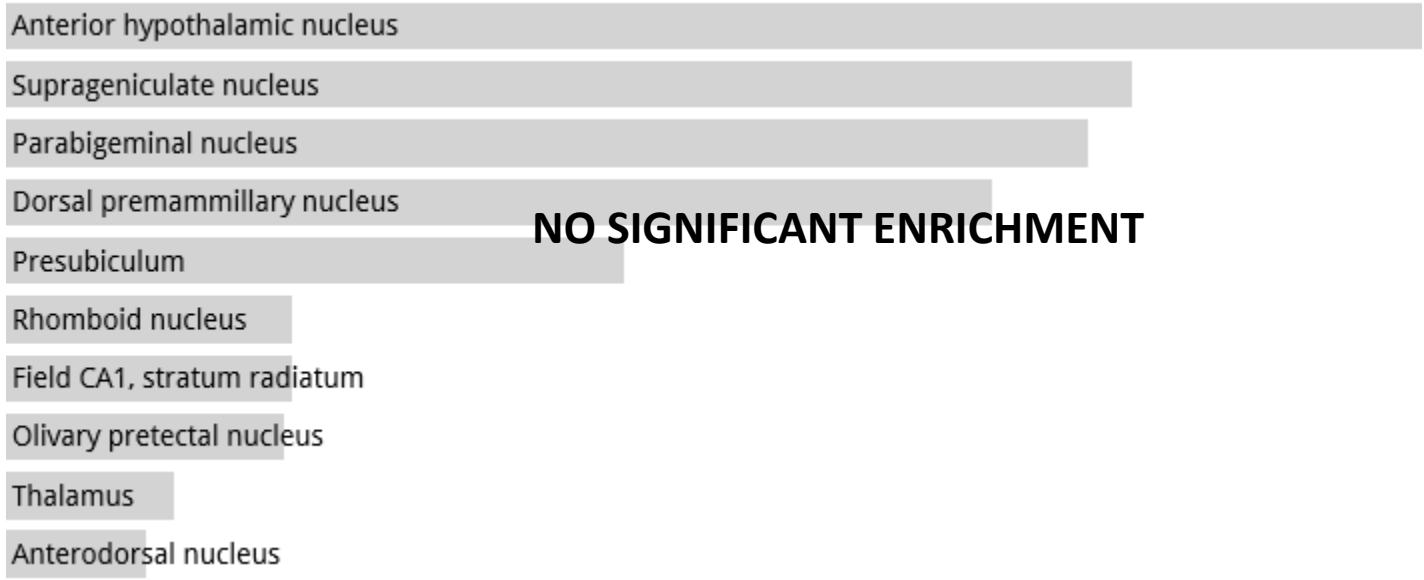
Disease Perturbations from GEO down random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 2

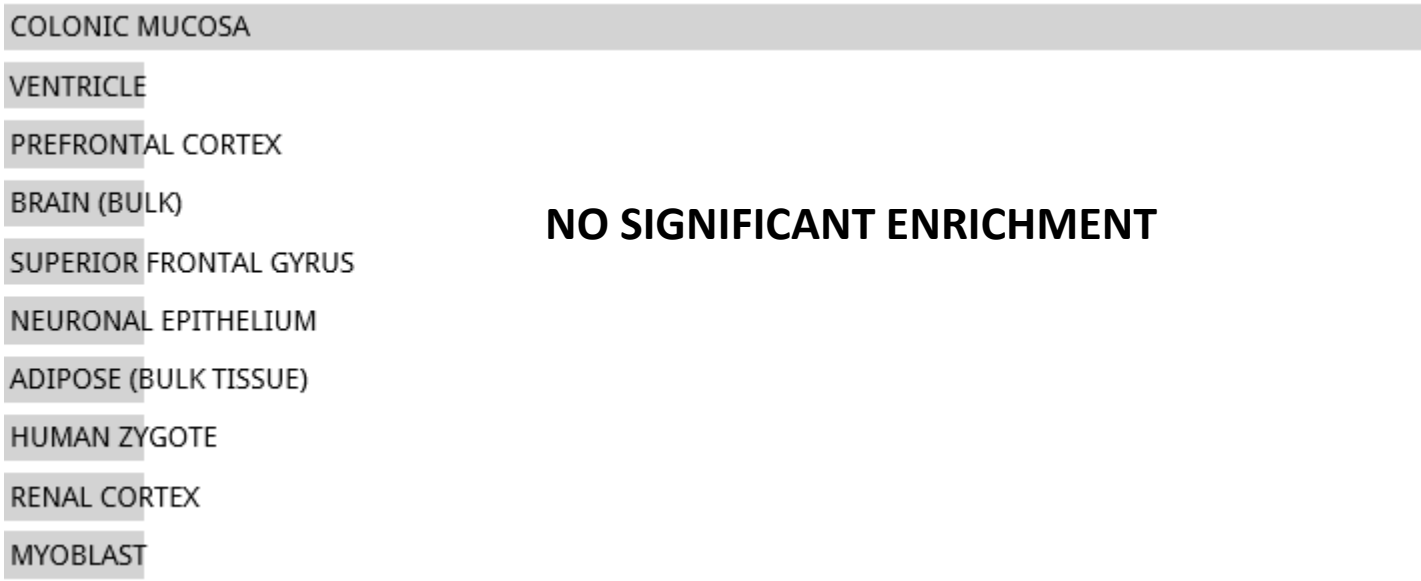
Allen Brain Atlas up random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 3

ARCHS4 Tissues random 6000 genes



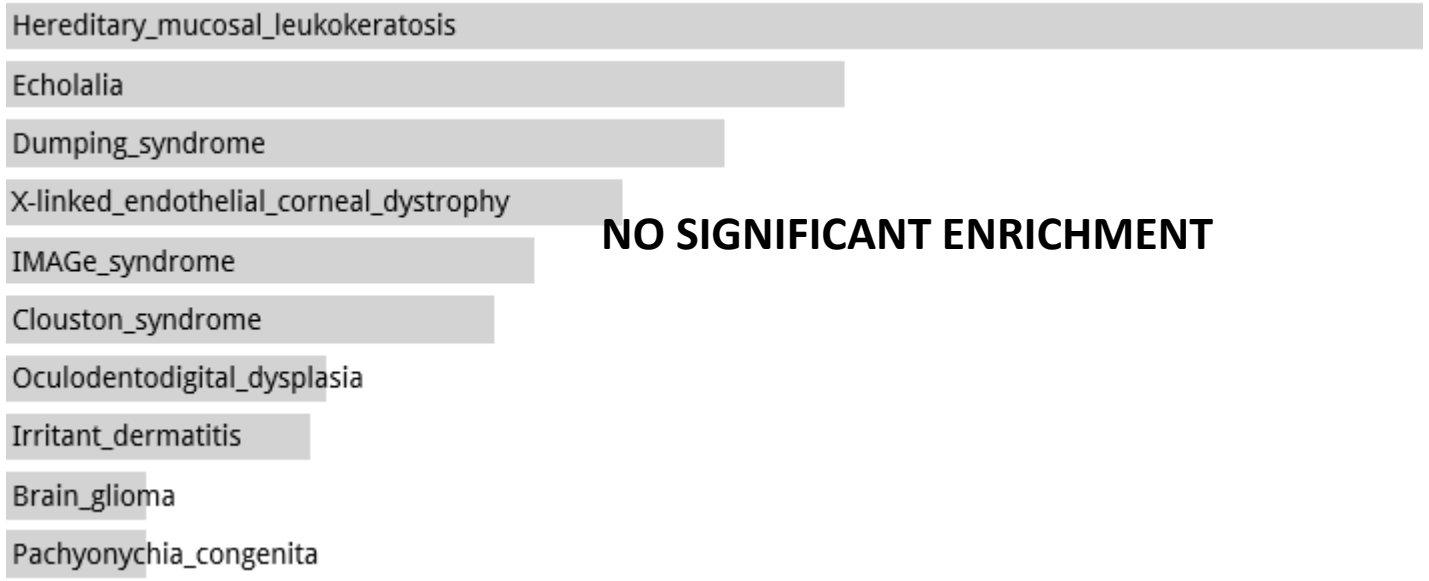
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

NO DATA AVAILABLE

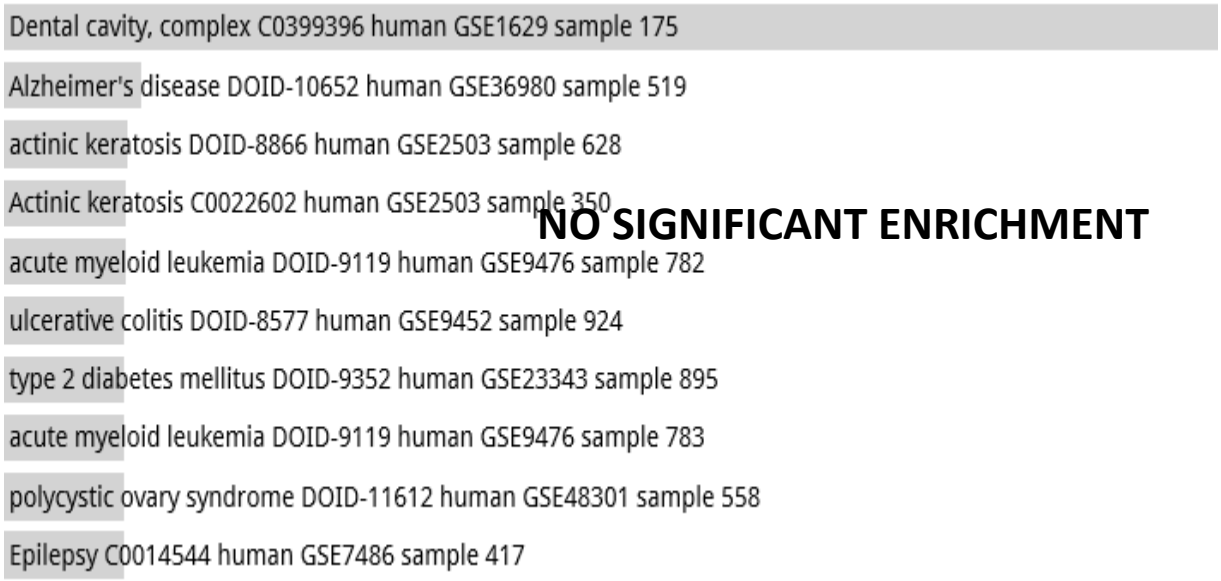
Random Set 3

Jensen DISEASES random 6000 genes



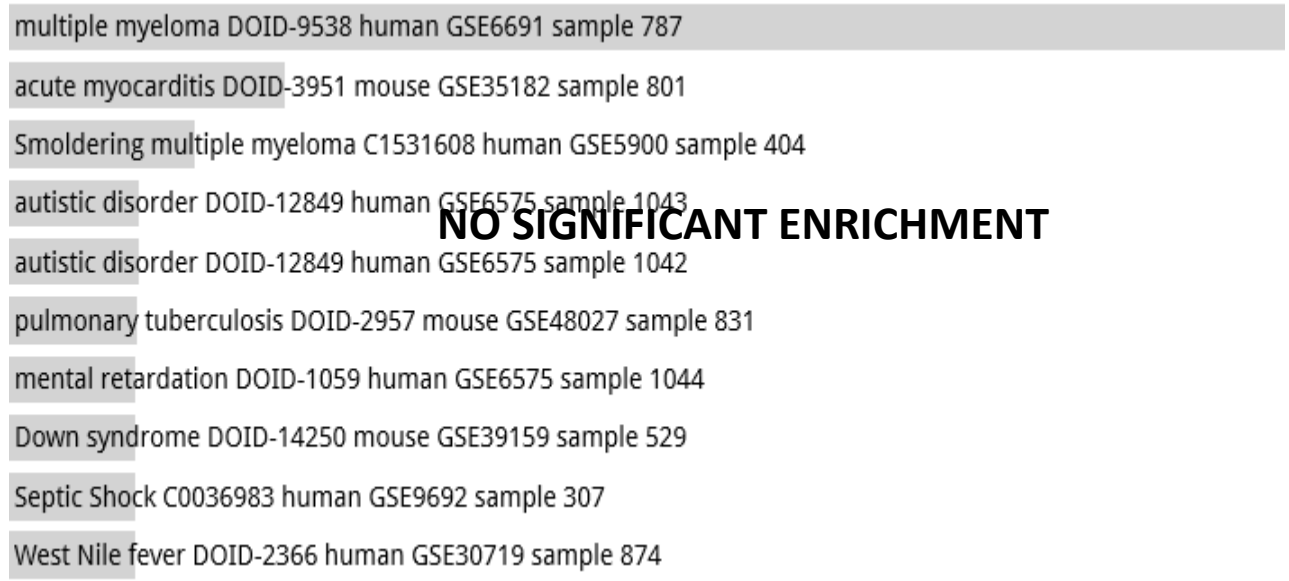
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes



NO SIGNIFICANT ENRICHMENT

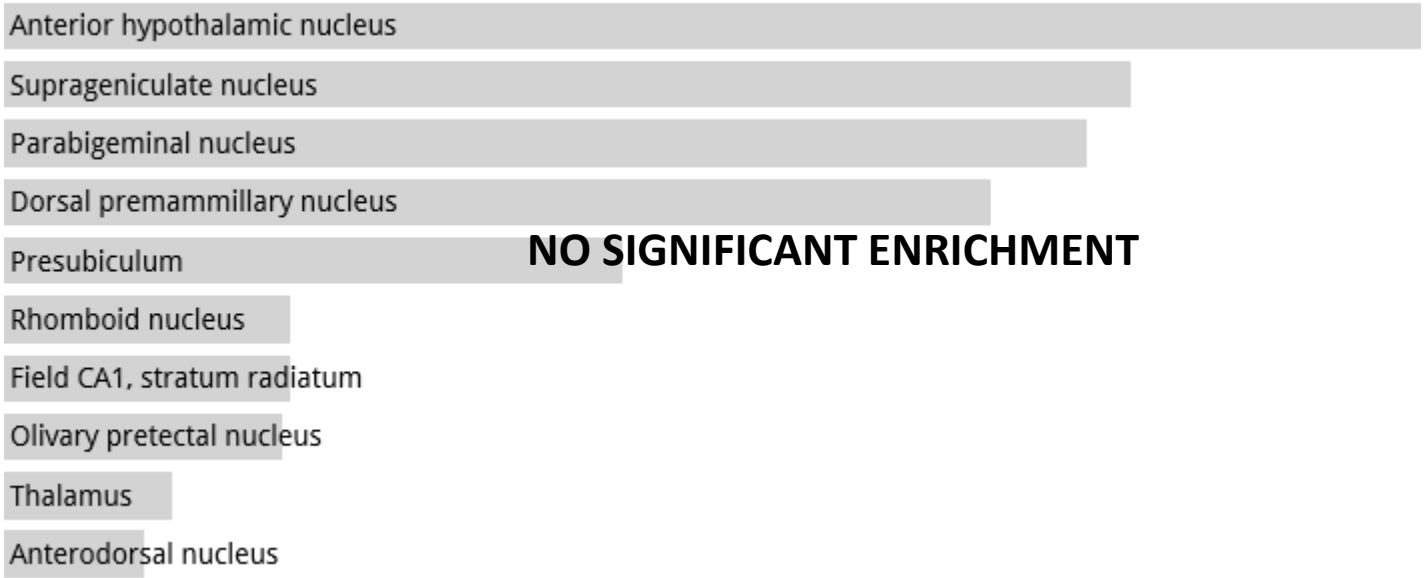
Disease Perturbations from GEO down random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 3

Allen Brain Atlas up random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 4

ARCHS4 Tissues random 6000 genes

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

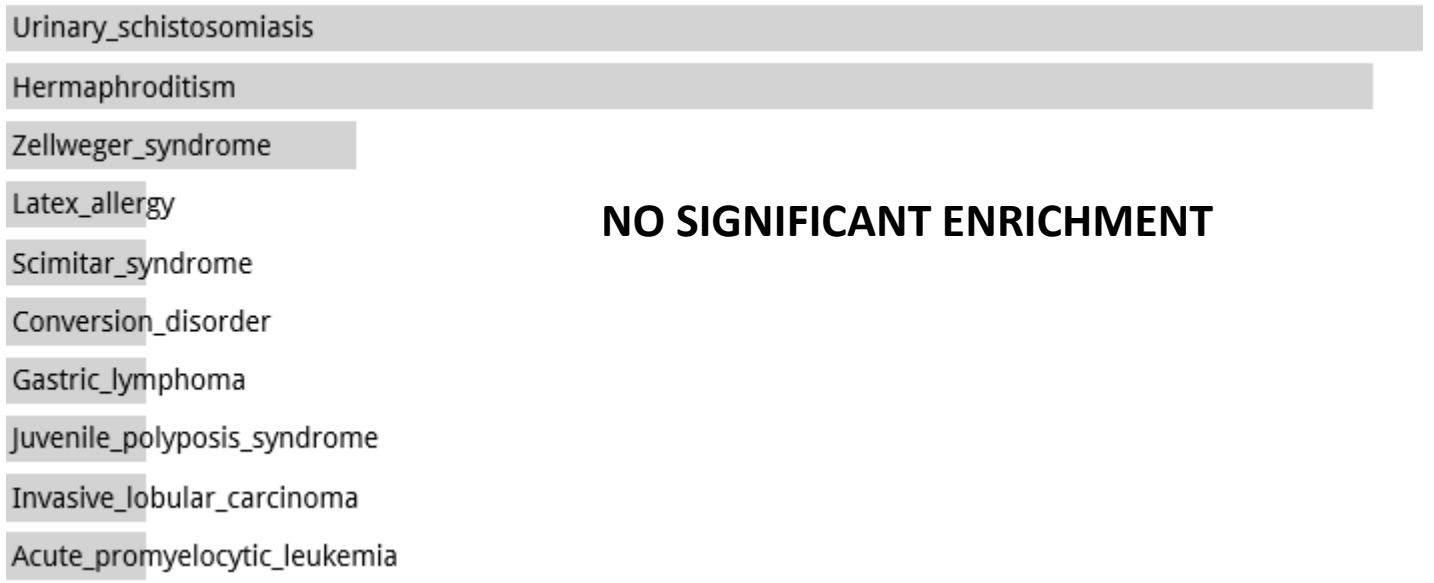
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

NO DATA AVAILABLE

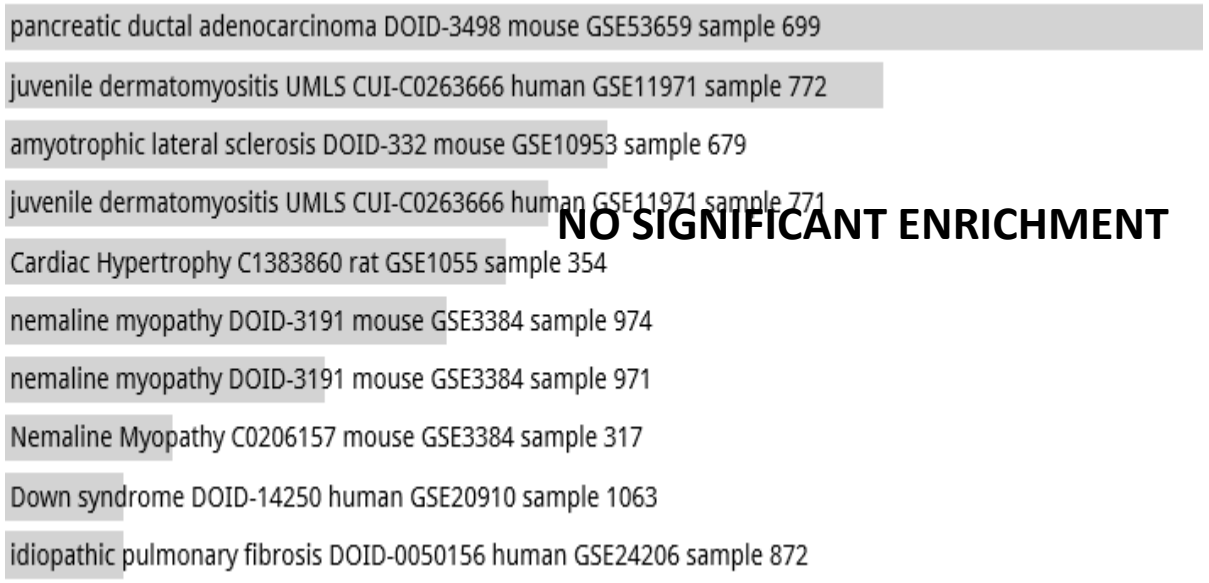
Random Set 4

Jensen DISEASES random 6000 genes



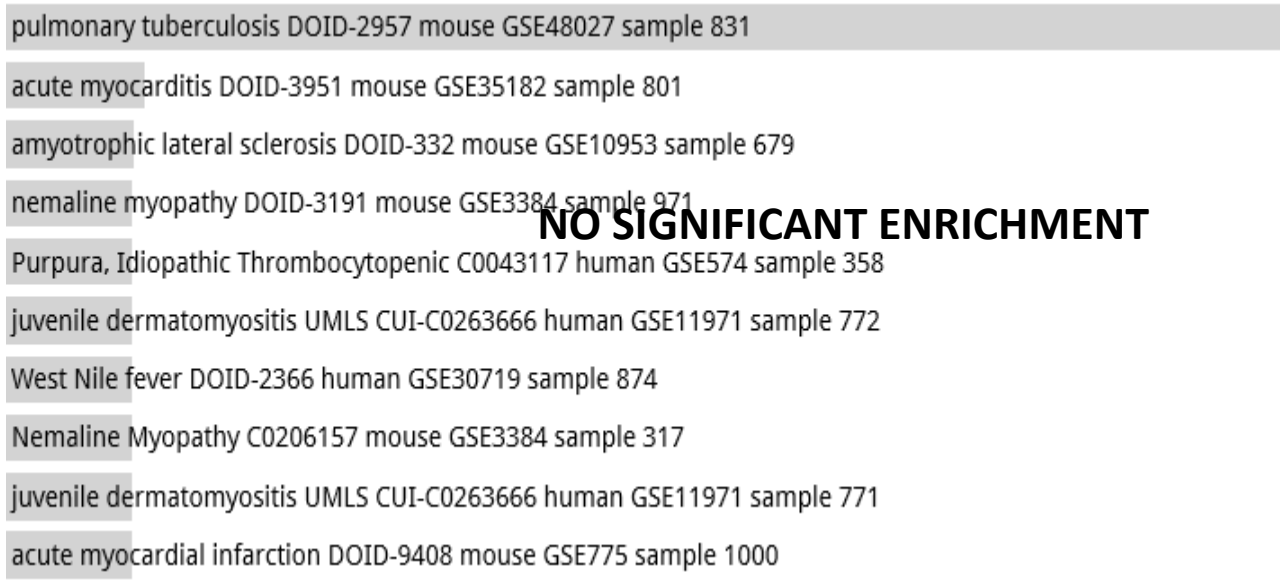
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes



NO SIGNIFICANT ENRICHMENT

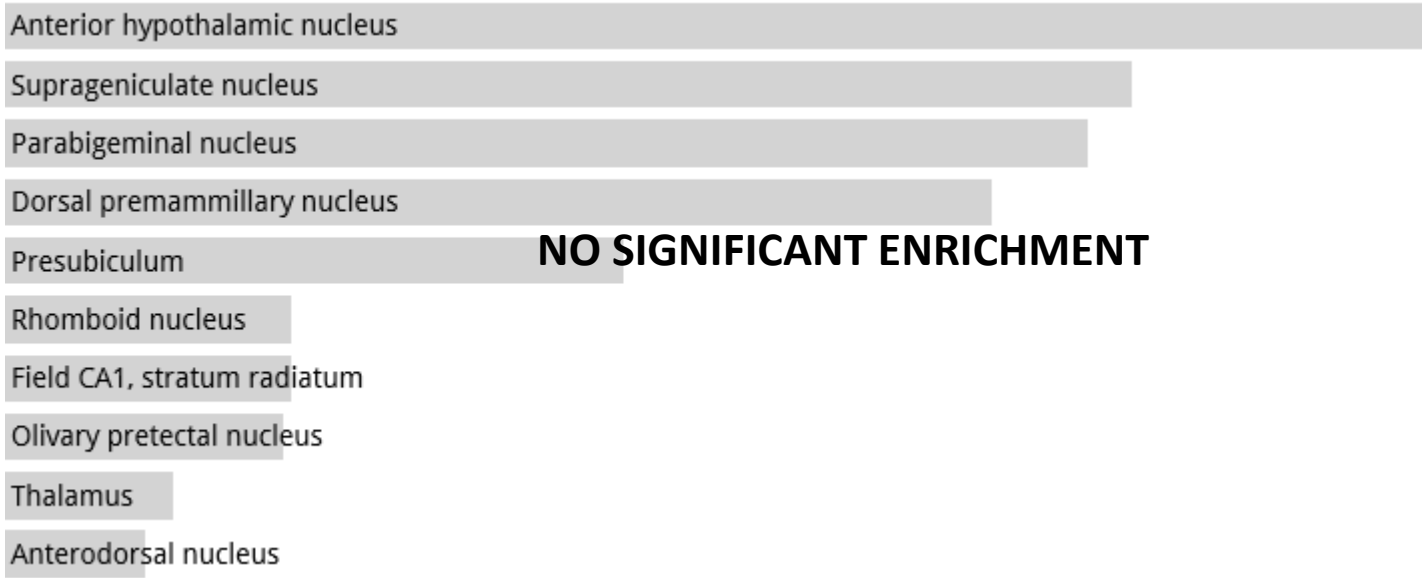
Disease Perturbations from GEO down random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 4

Allen Brain Atlas up random 6000 genes



NO SIGNIFICANT ENRICHMENT

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

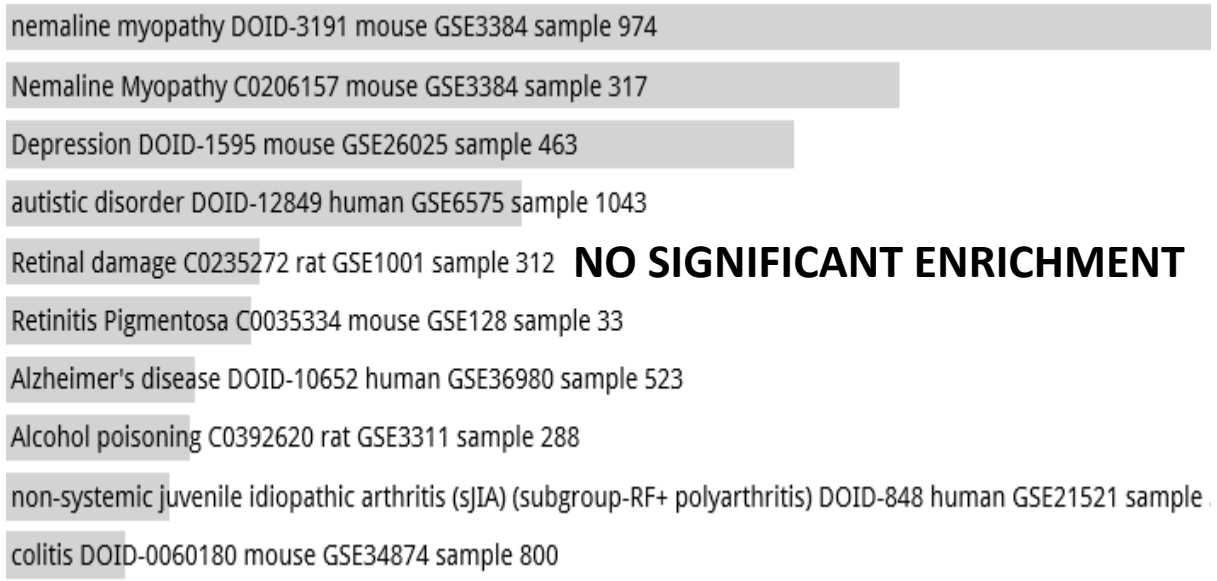
NO DATA AVAILABLE

Random Set 5

Jensen DISEASES random 6000 genes

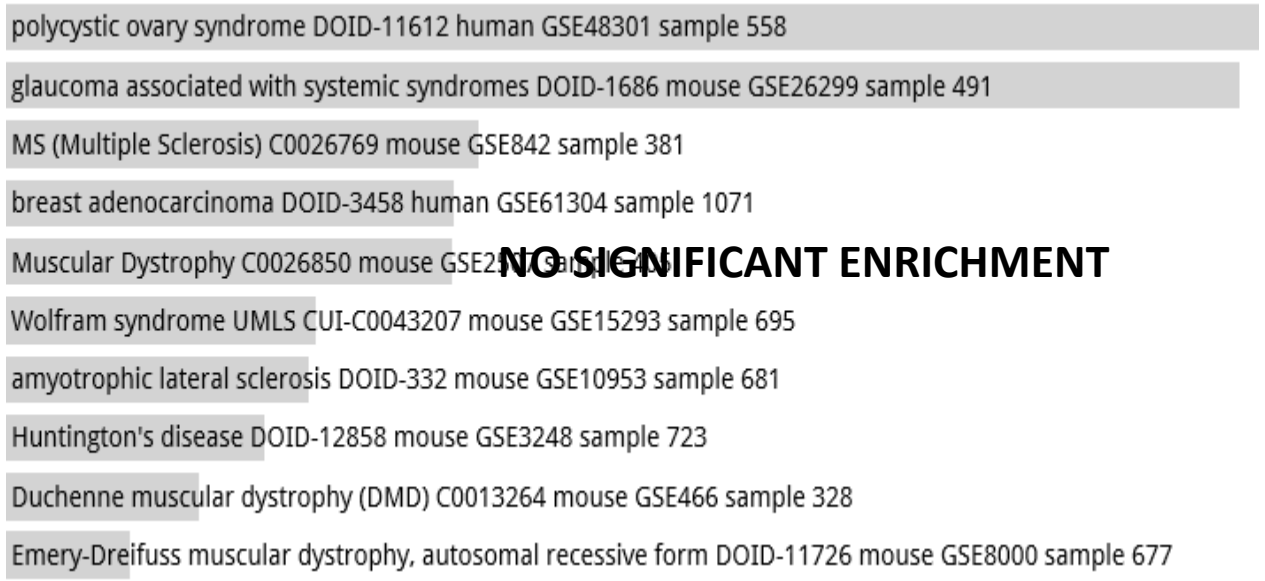
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes



NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 6000 genes



NO SIGNIFICANT ENRICHMENT

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

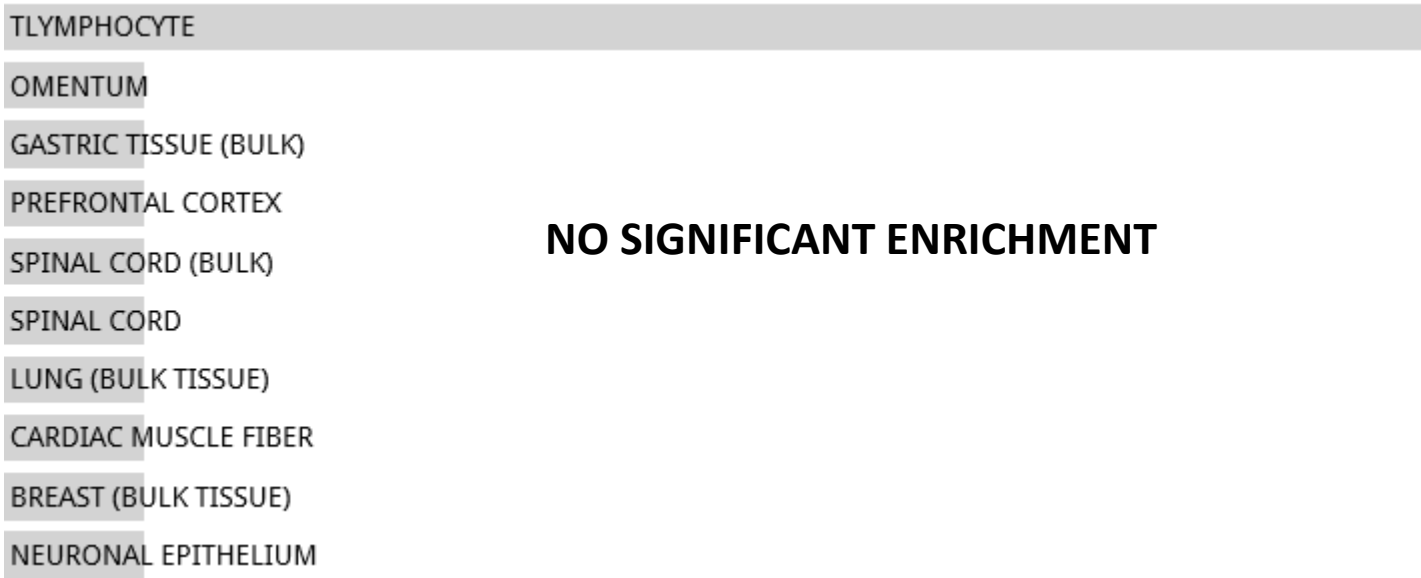
Random Set 6

Allen Brain Atlas up random 6000 genes

NO DATA AVAILABLE

Random Set 7

ARCHS4 Tissues random 6847 genes



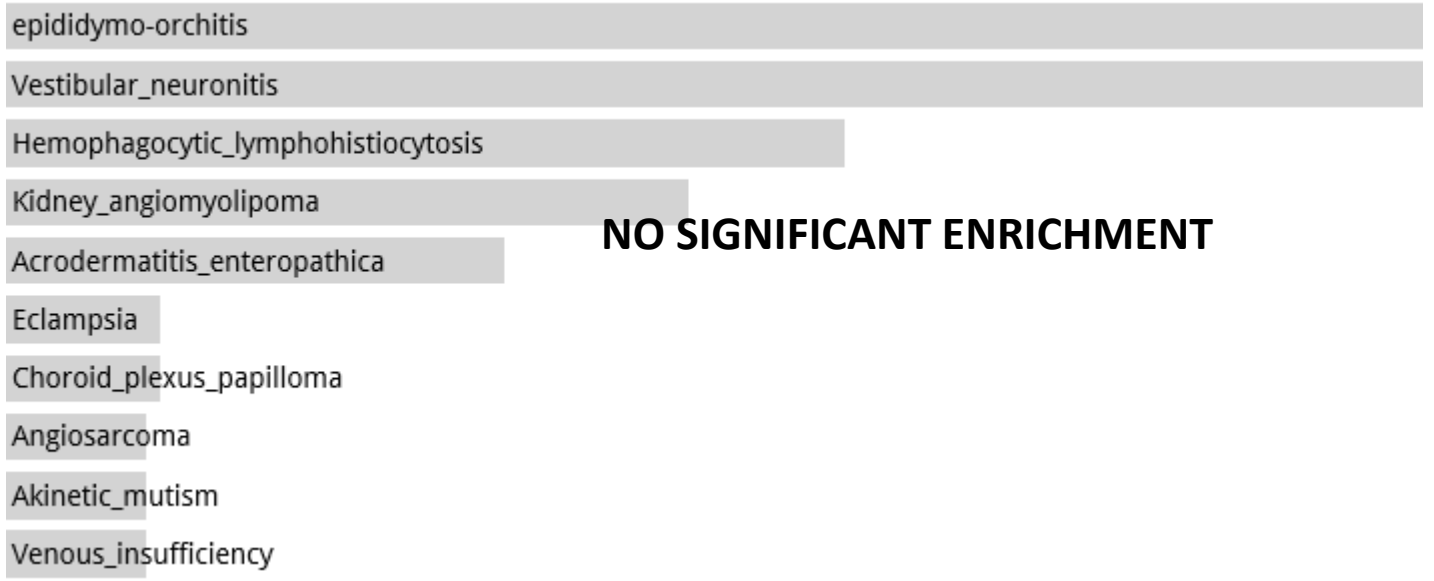
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6847 genes

NO DATA AVAILABLE

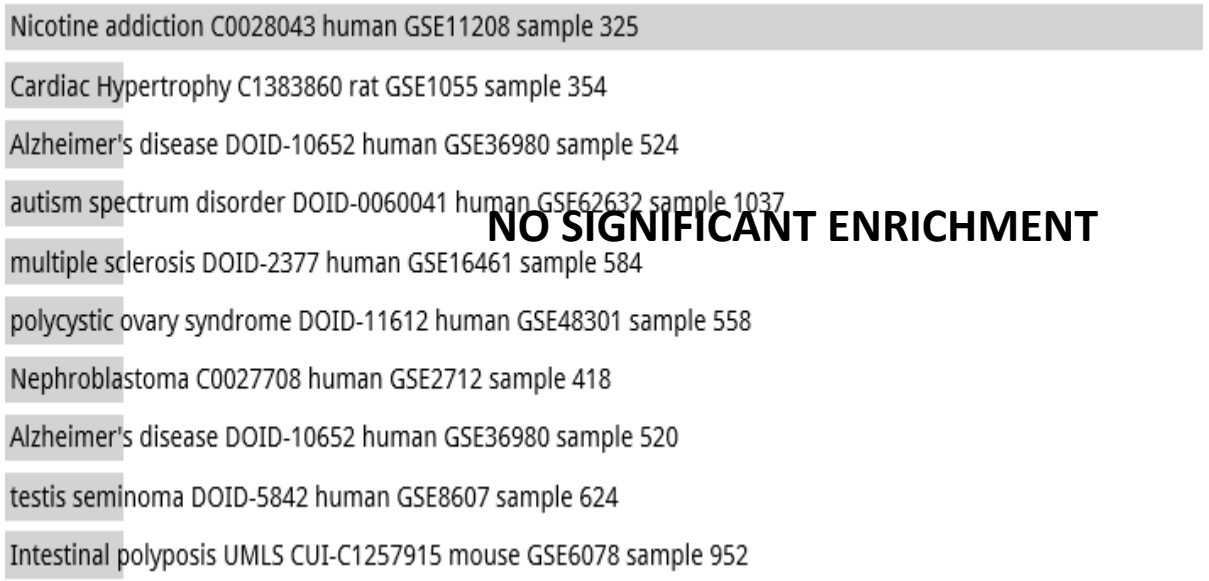
Random Set 7

Jensen DISEASES random 6847 genes



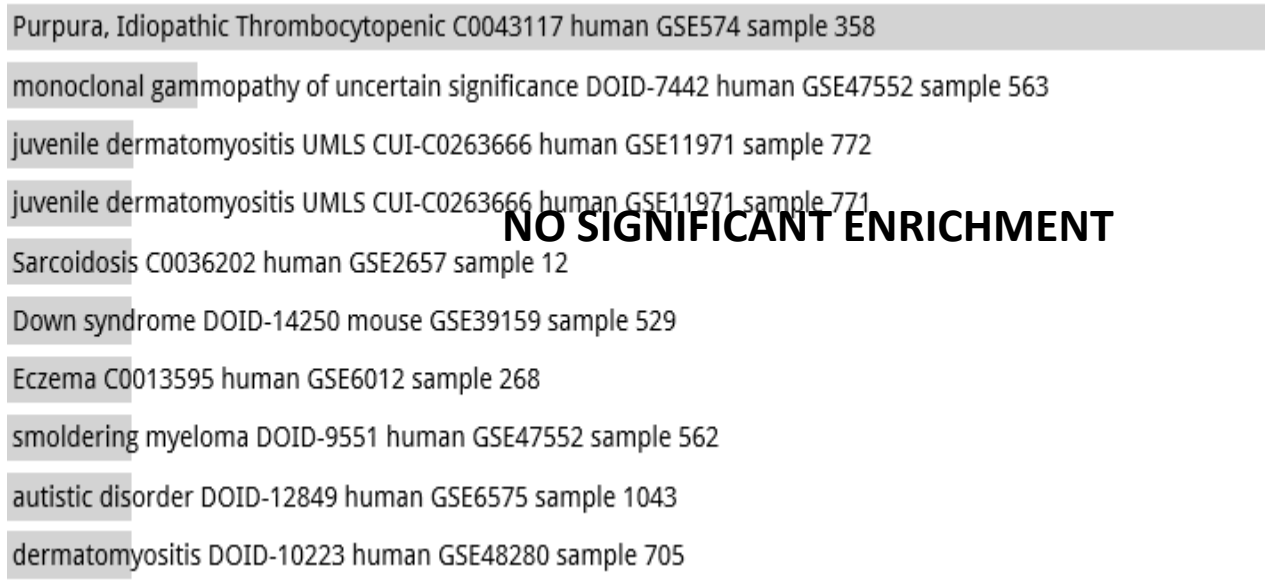
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes



NO SIGNIFICANT ENRICHMENT

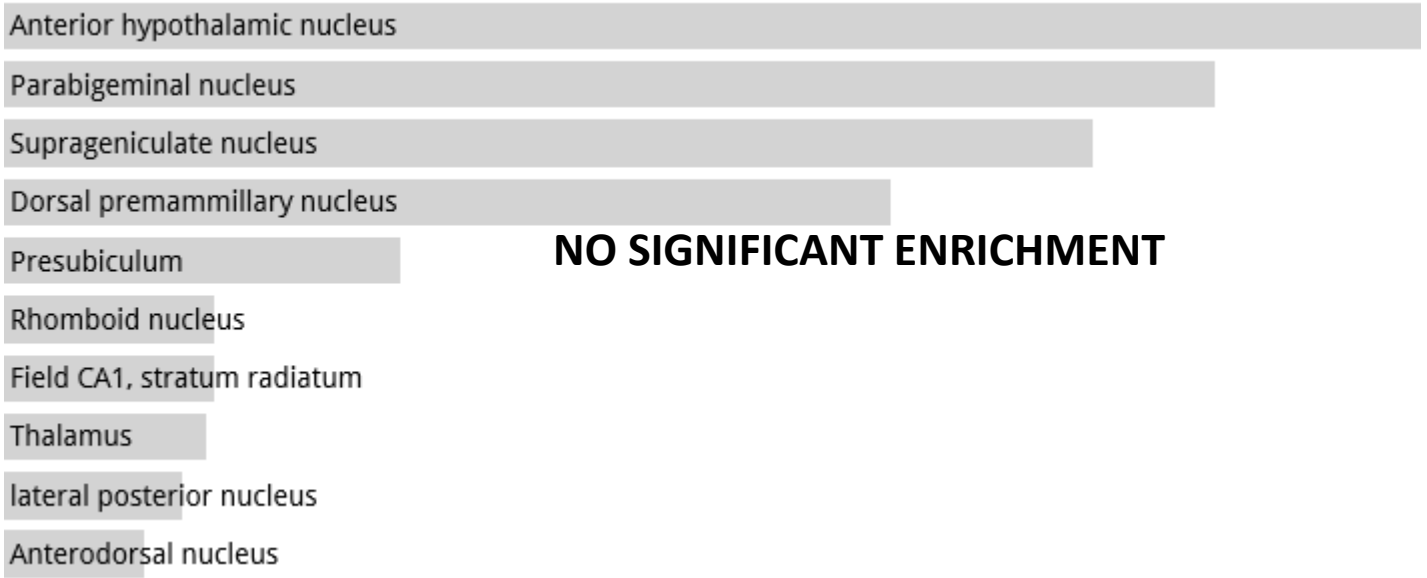
Disease Perturbations from GEO down random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 7

Allen Brain Atlas up random 6847 genes



NO SIGNIFICANT ENRICHMENT

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

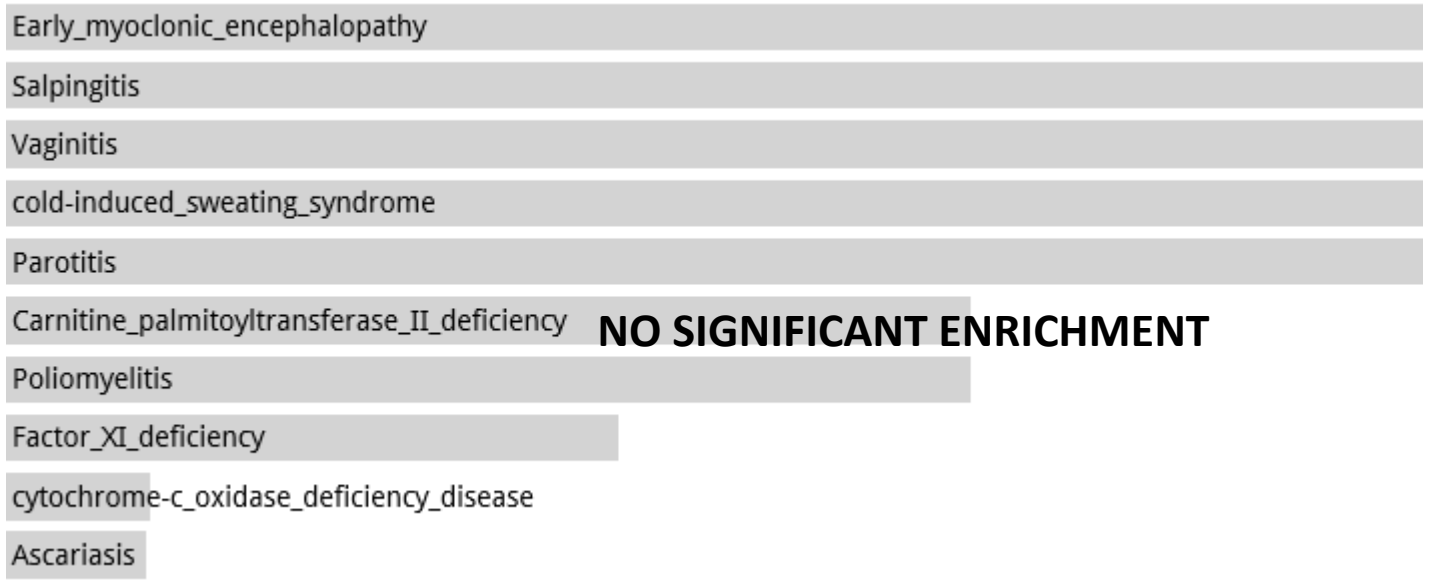
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6001 genes

NO DATA AVAILABLE

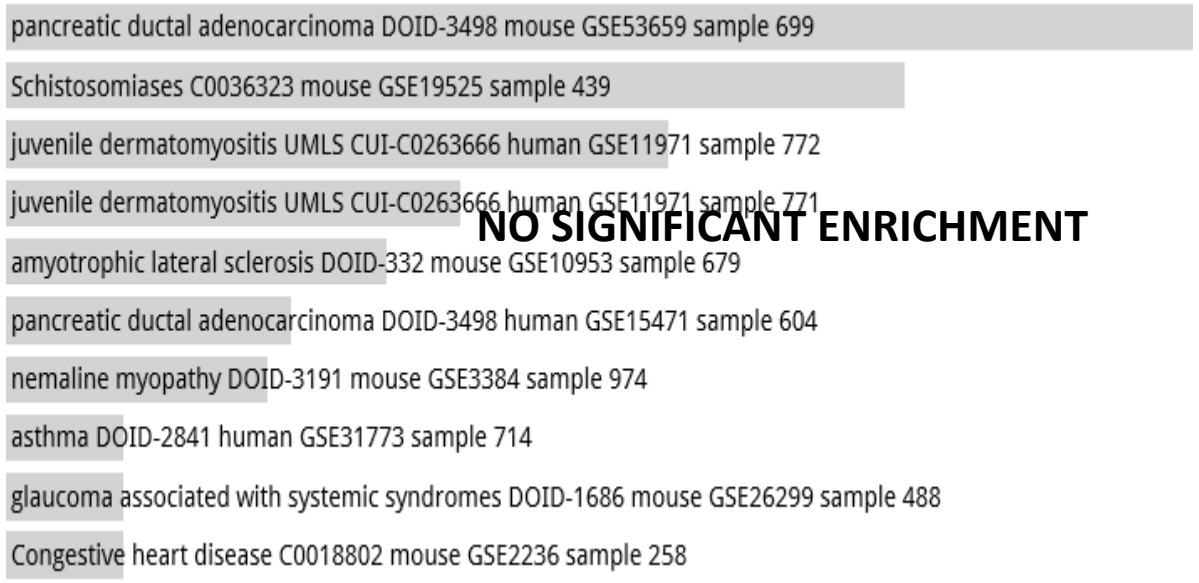
Random Set 8

Jensen DISEASES random 6001 genes



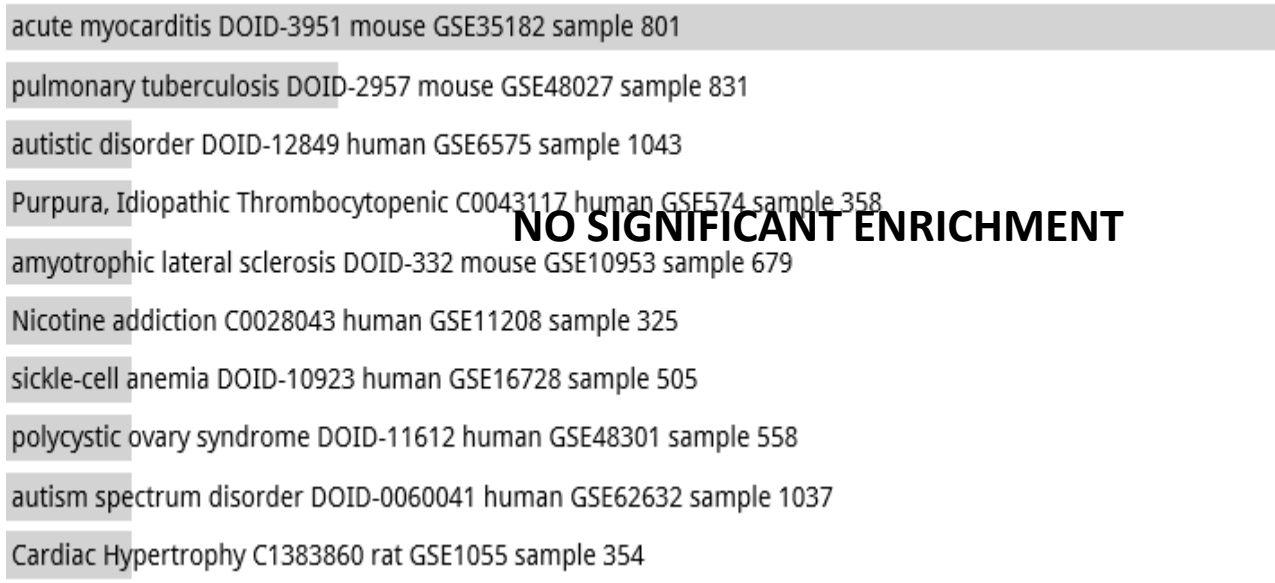
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes



NO SIGNIFICANT ENRICHMENT

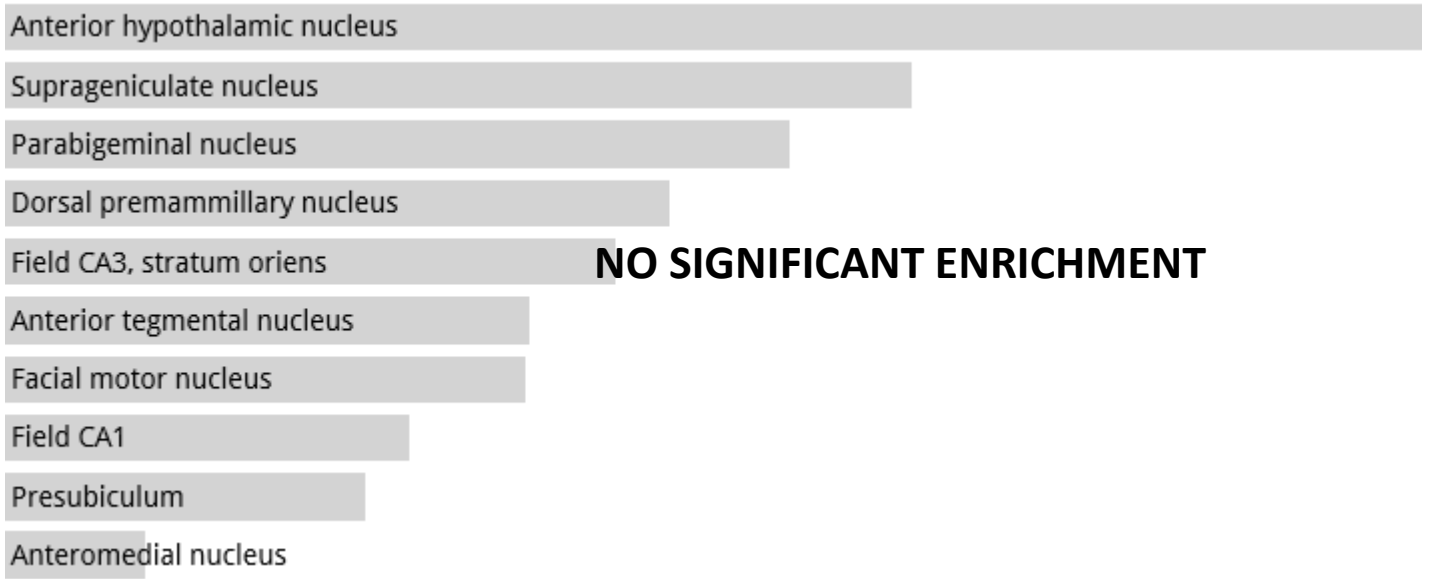
Disease Perturbations from GEO down random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 8

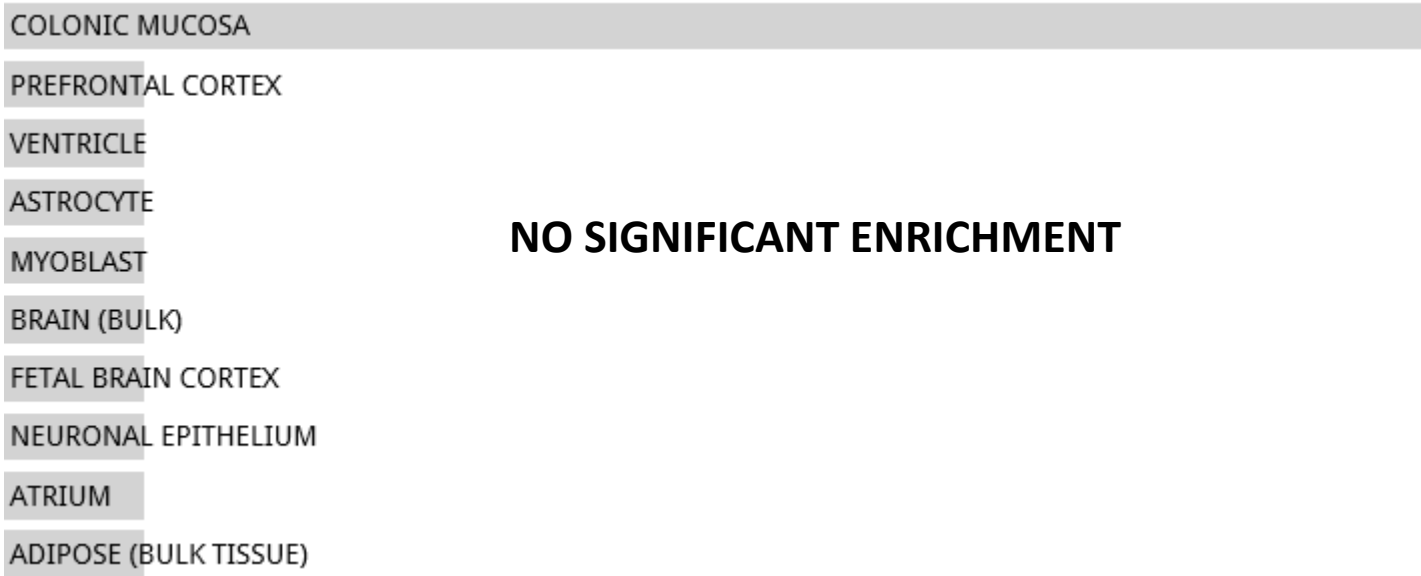
Allen Brain Atlas up random 6001 genes



NO SIGNIFICANT ENRICHMENT

Random Set 9

ARCHS4 Tissues random 6000 genes



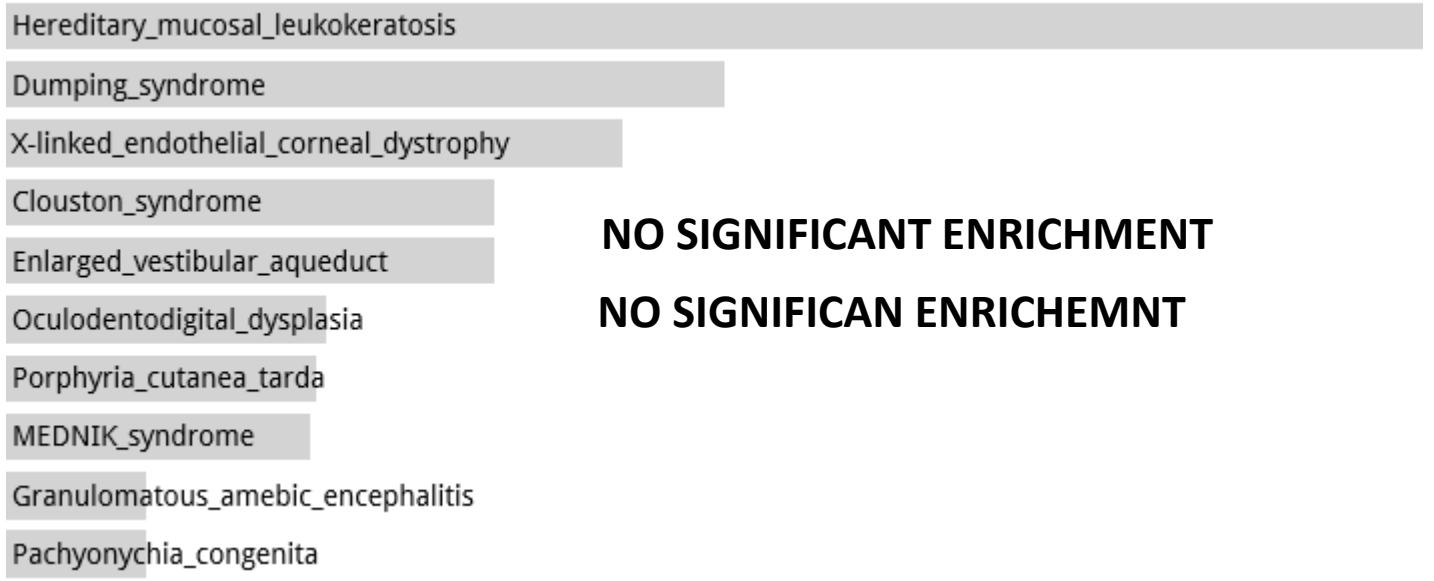
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6000 genes

NO DATA AVAILABLE

Random Set 9

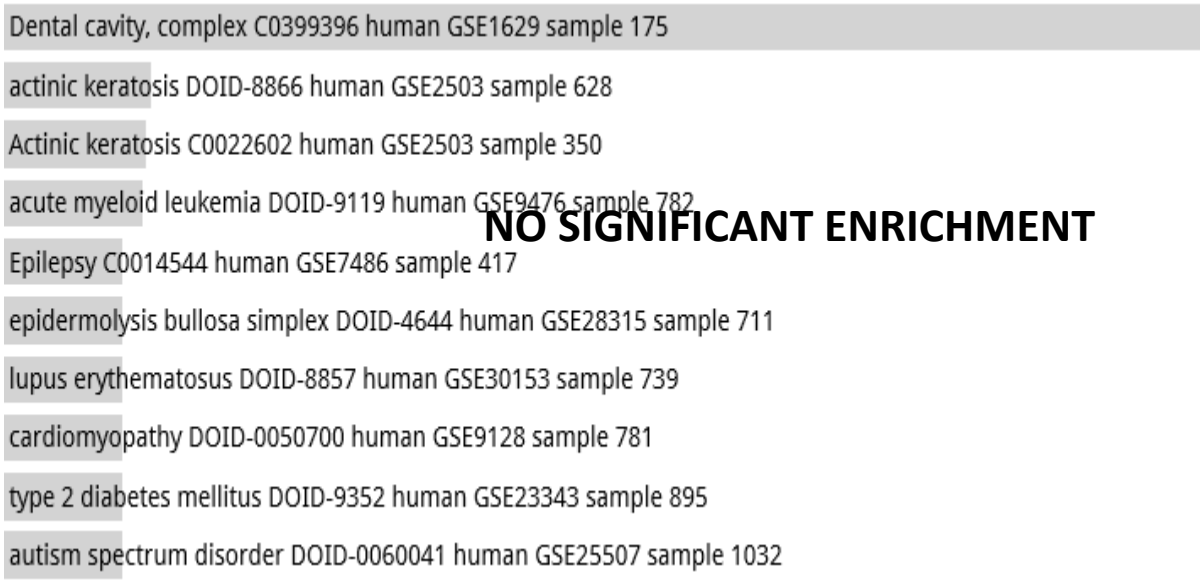
Jensen DISEASES random 6000 genes



NO SIGNIFICANT ENRICHMENT

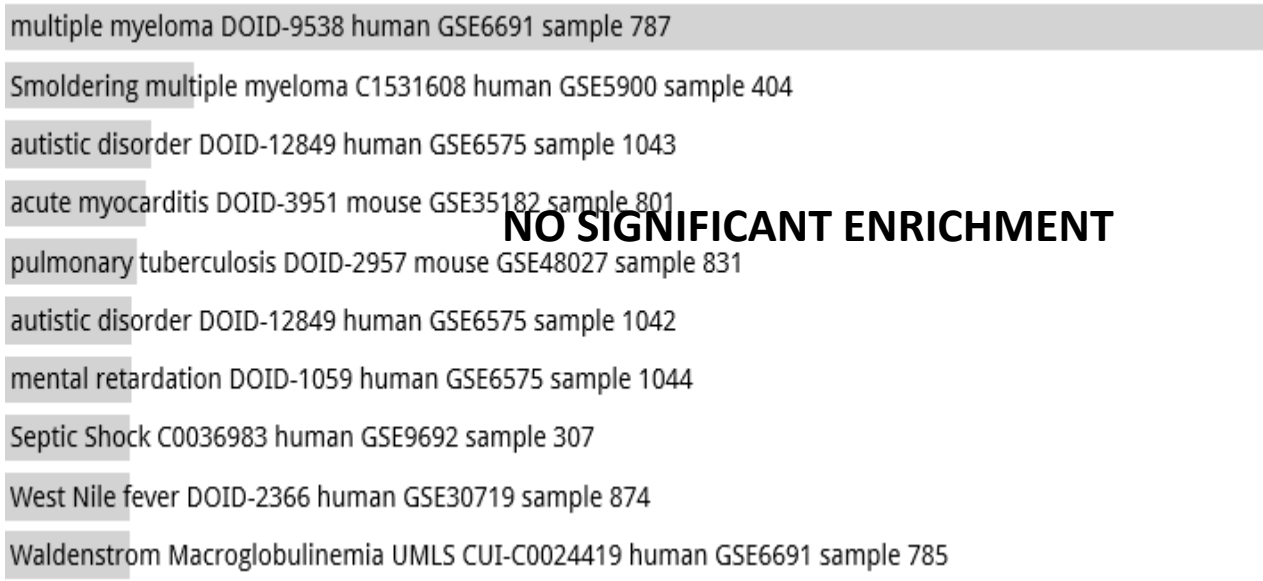
NO SIGNIFICAN ENRICHEMNT

Disease Perturbations from GEO up random 6000 genes



NO SIGNIFICANT ENRICHMENT

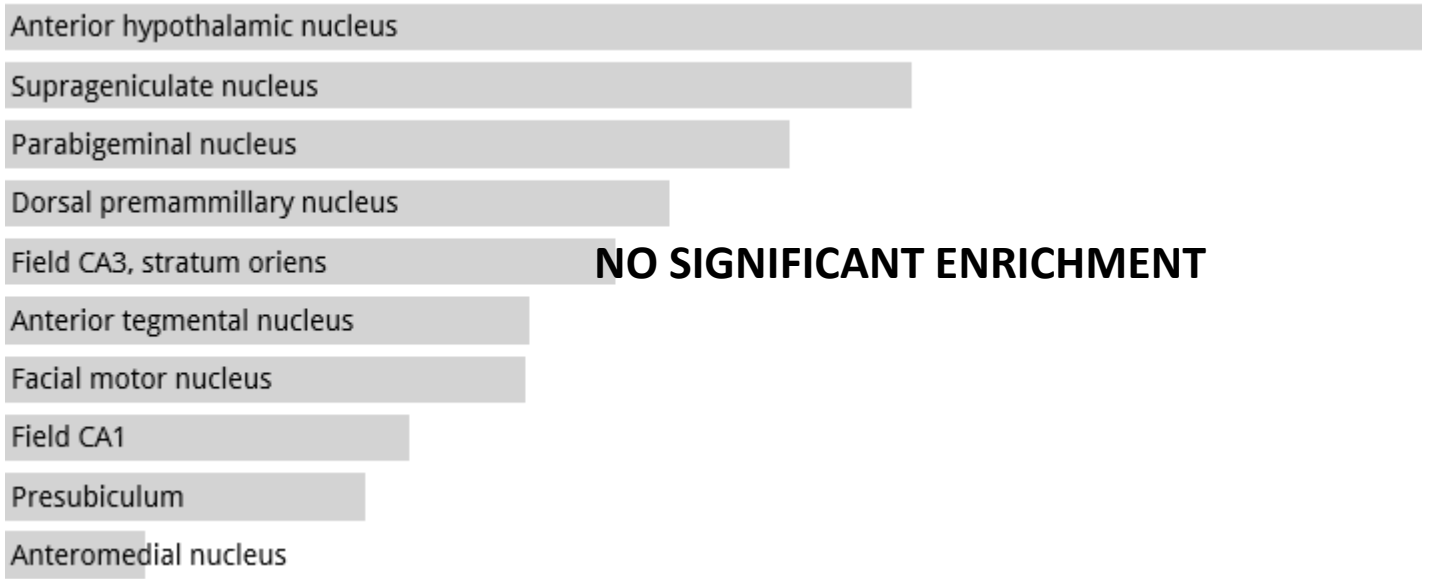
Disease Perturbations from GEO down random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 9

Allen Brain Atlas up random 6000 genes



NO SIGNIFICANT ENRICHMENT

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

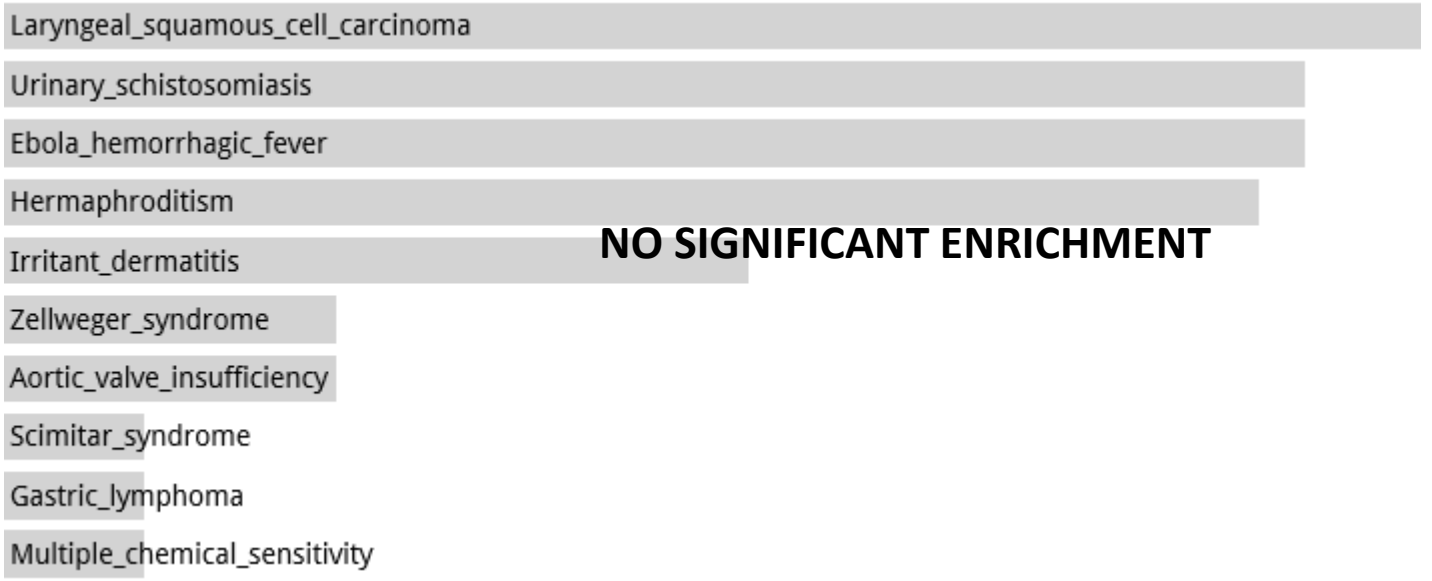
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 6000 genes

NO DATA AVAILABLE

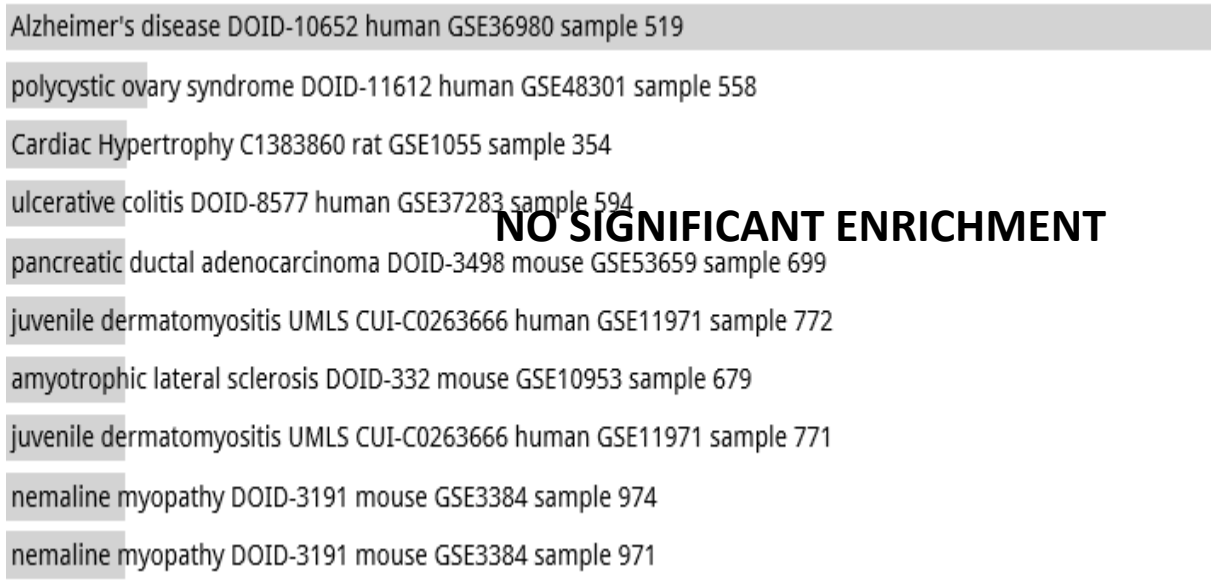
Random Set 10

Jensen DISEASES random 6000 genes



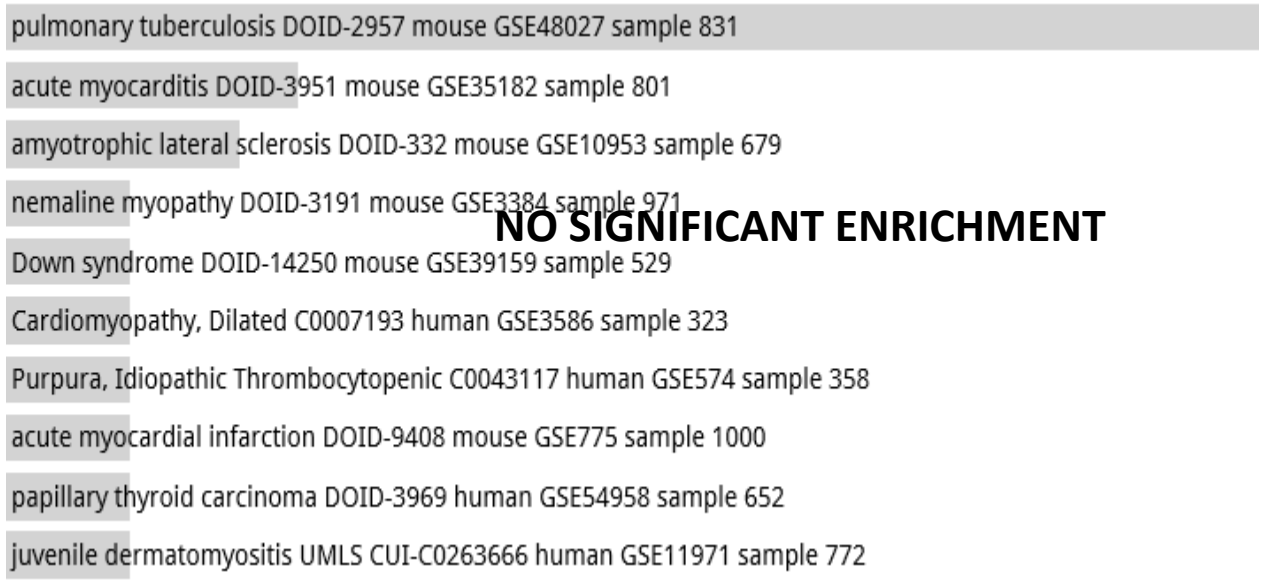
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 6000 genes



NO SIGNIFICANT ENRICHMENT

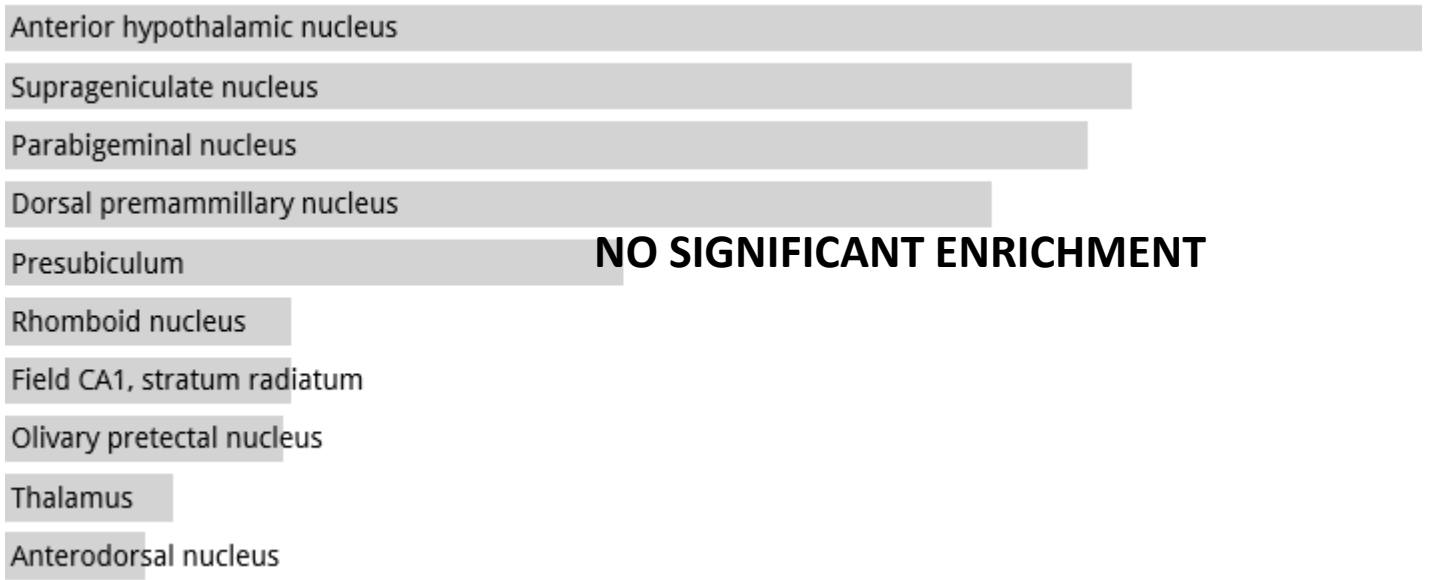
Disease Perturbations from GEO down random 6000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 10

Allen Brain Atlas up random 6000 genes



NO SIGNIFICANT ENRICHMENT

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)

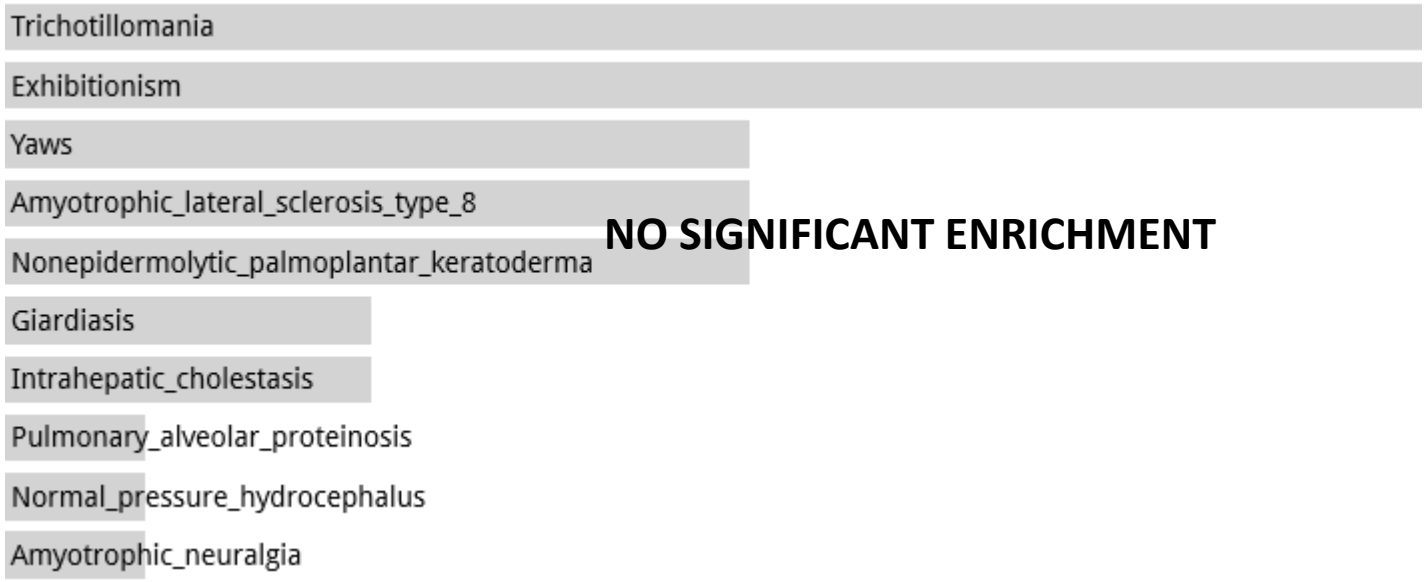
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

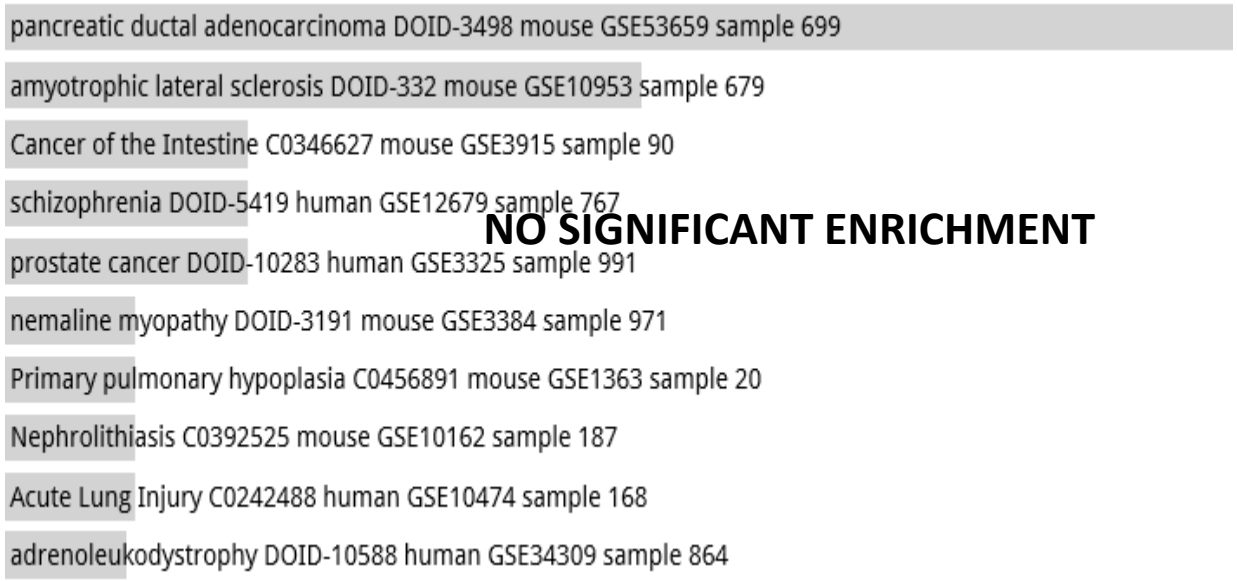
Random Set 11

Jensen DISEASES random 4000 genes



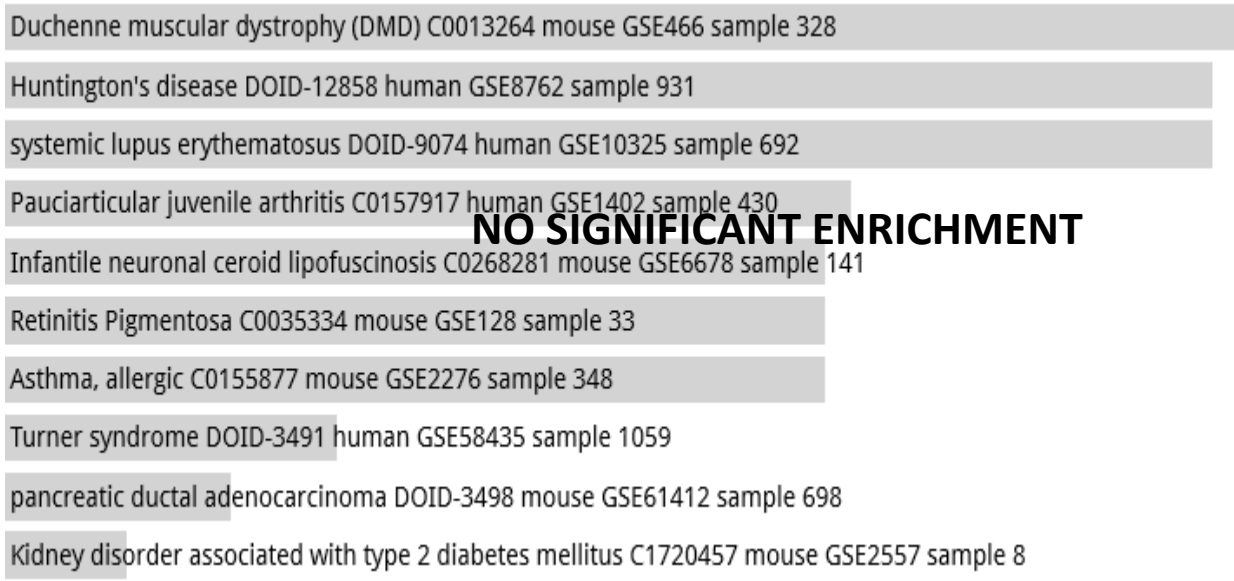
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 4000 genes



NO SIGNIFICANT ENRICHMENT

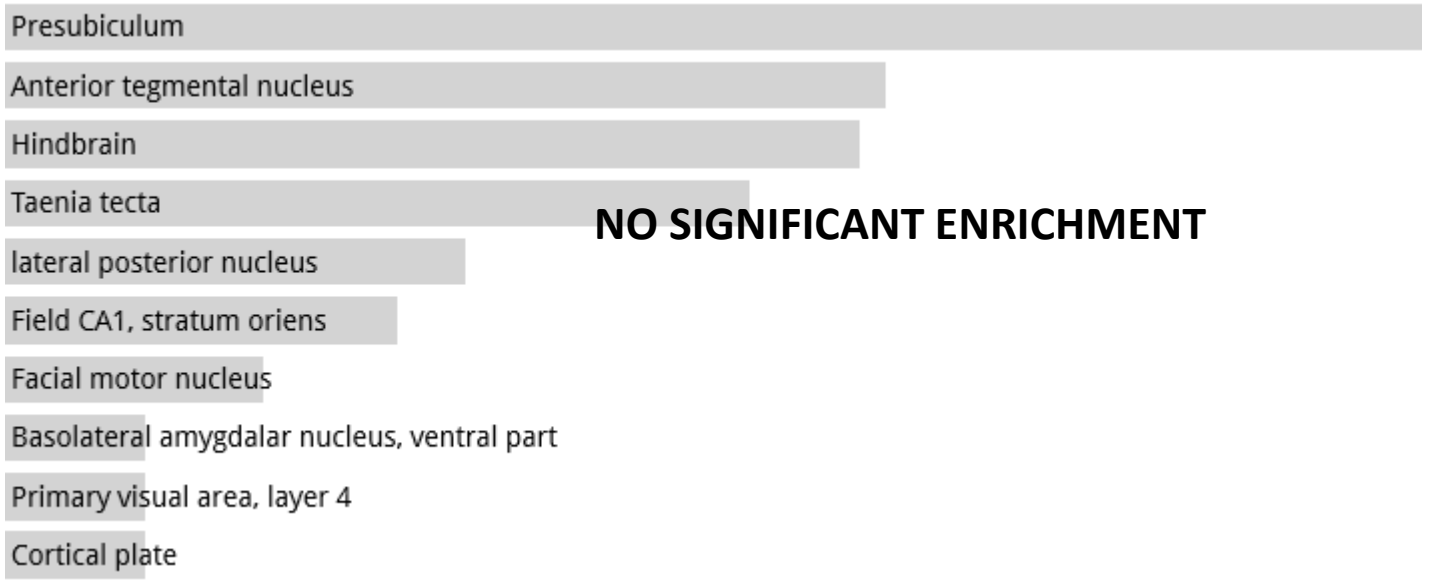
Disease Perturbations from GEO down random 4000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 11

Allen Brain Atlas up random 4000 genes



NO SIGNIFICANT ENRICHMENT

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

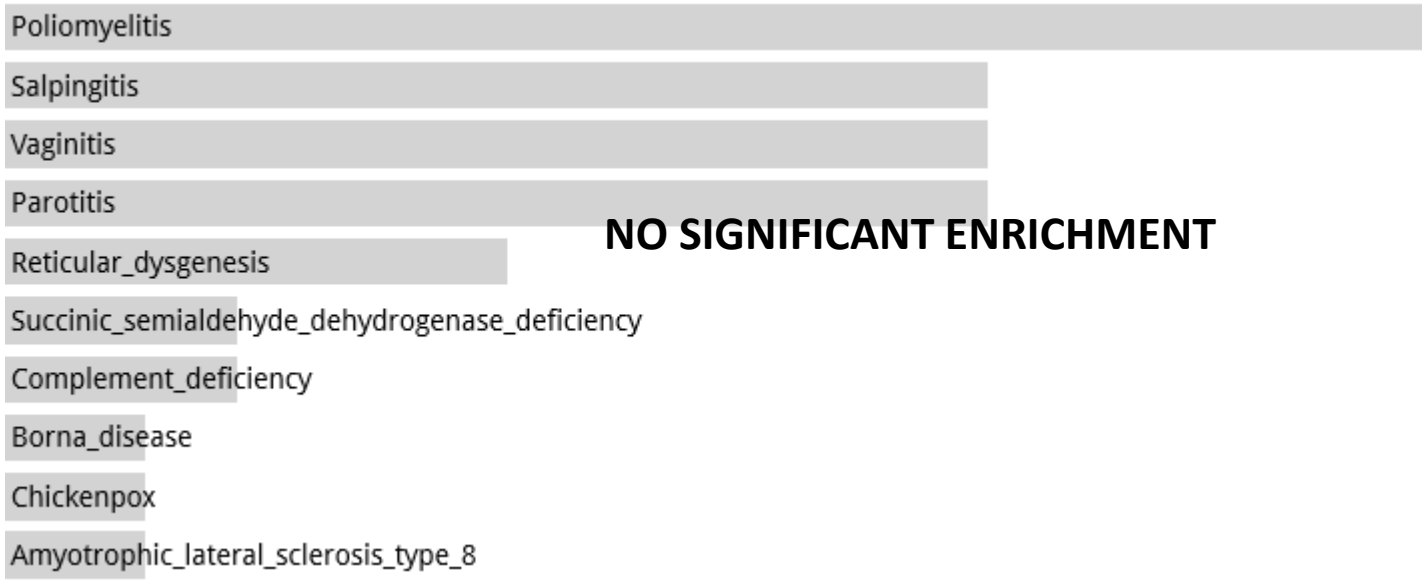
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

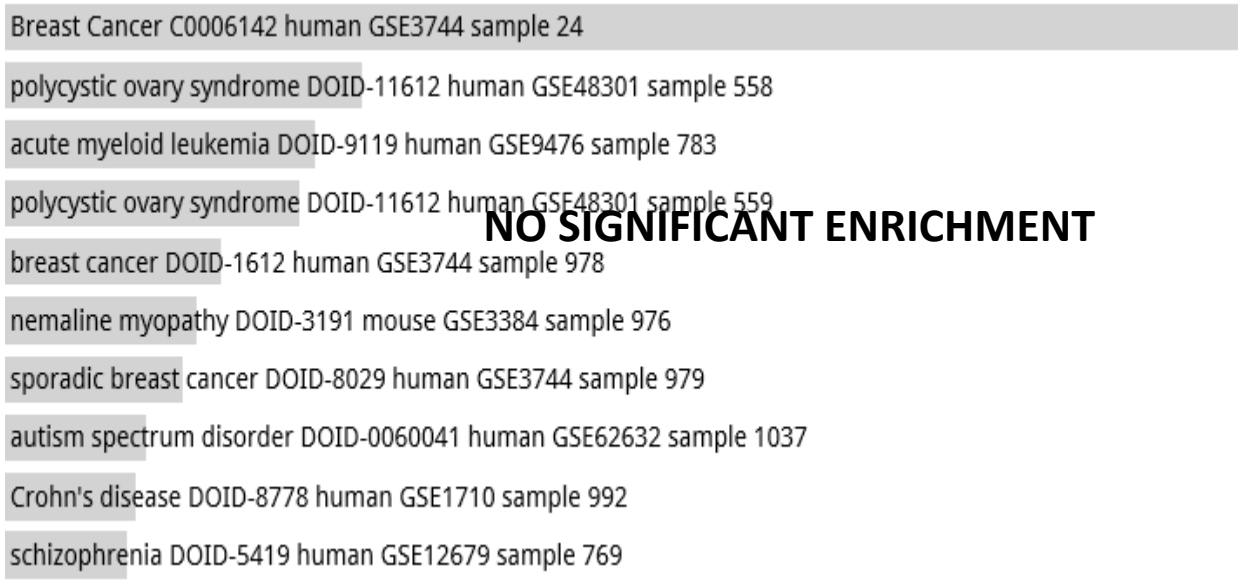
Random Set 12

Jensen DISEASES random 4000 genes



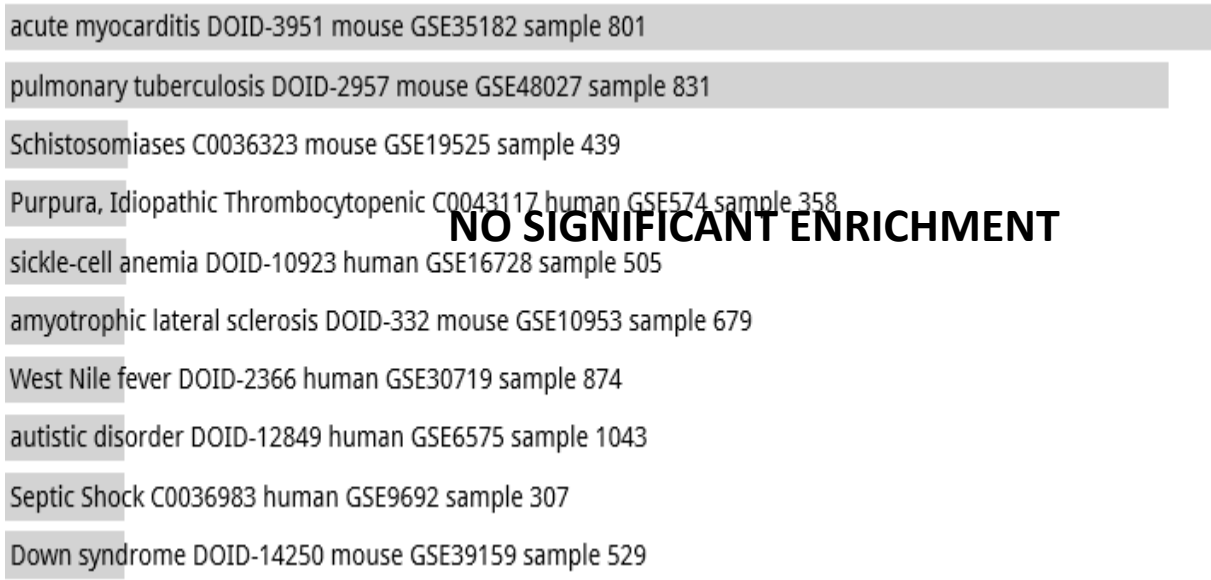
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 4000 genes



NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 4000 genes



NO SIGNIFICANT ENRICHMENT

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

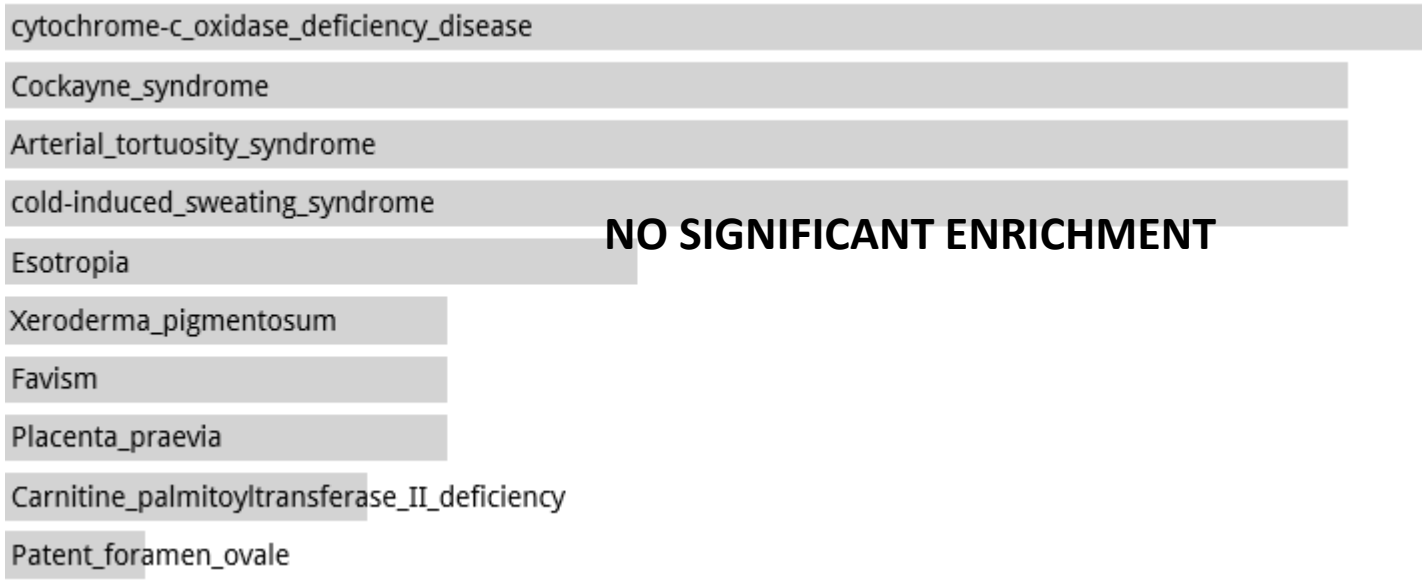
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

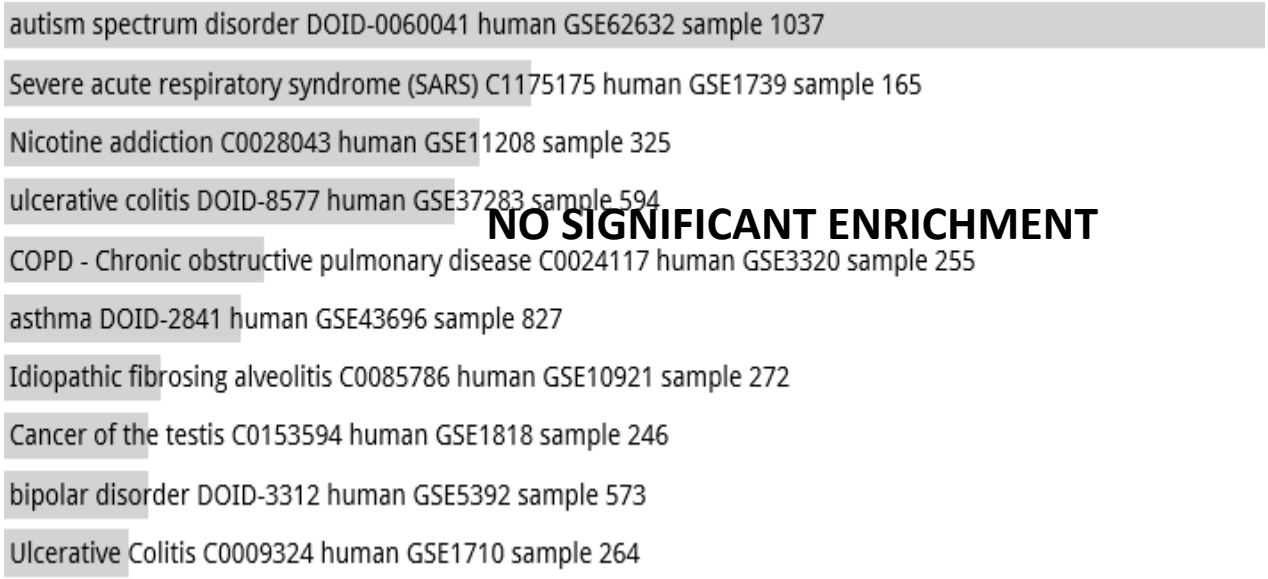
Random Set 13

Jensen DISEASES random 4000 genes



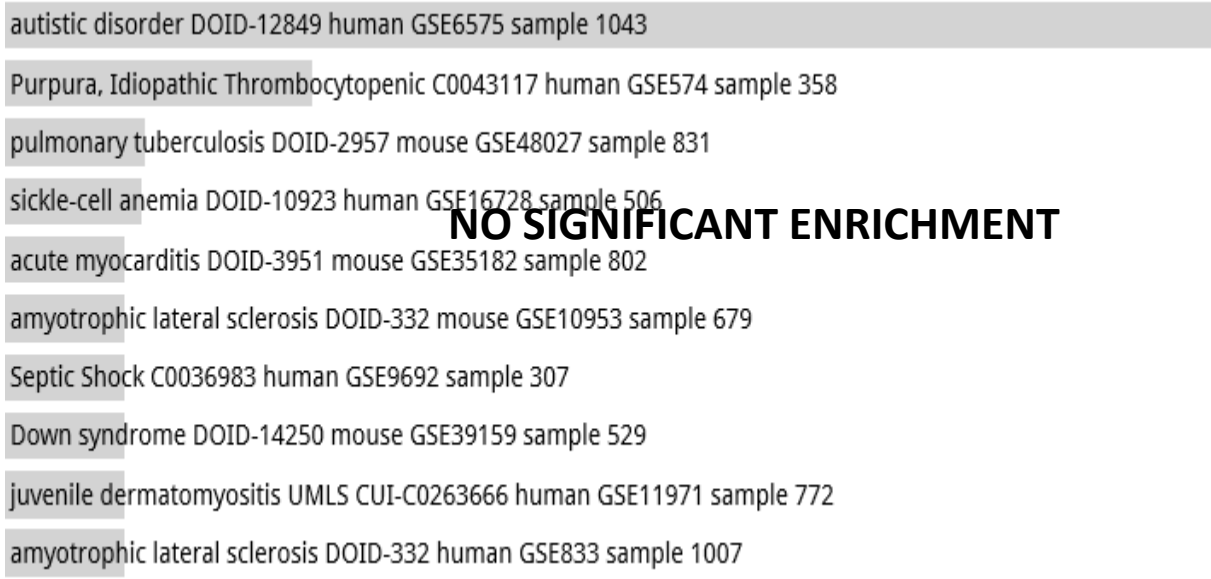
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 4000 genes



NO SIGNIFICANT ENRICHMENT

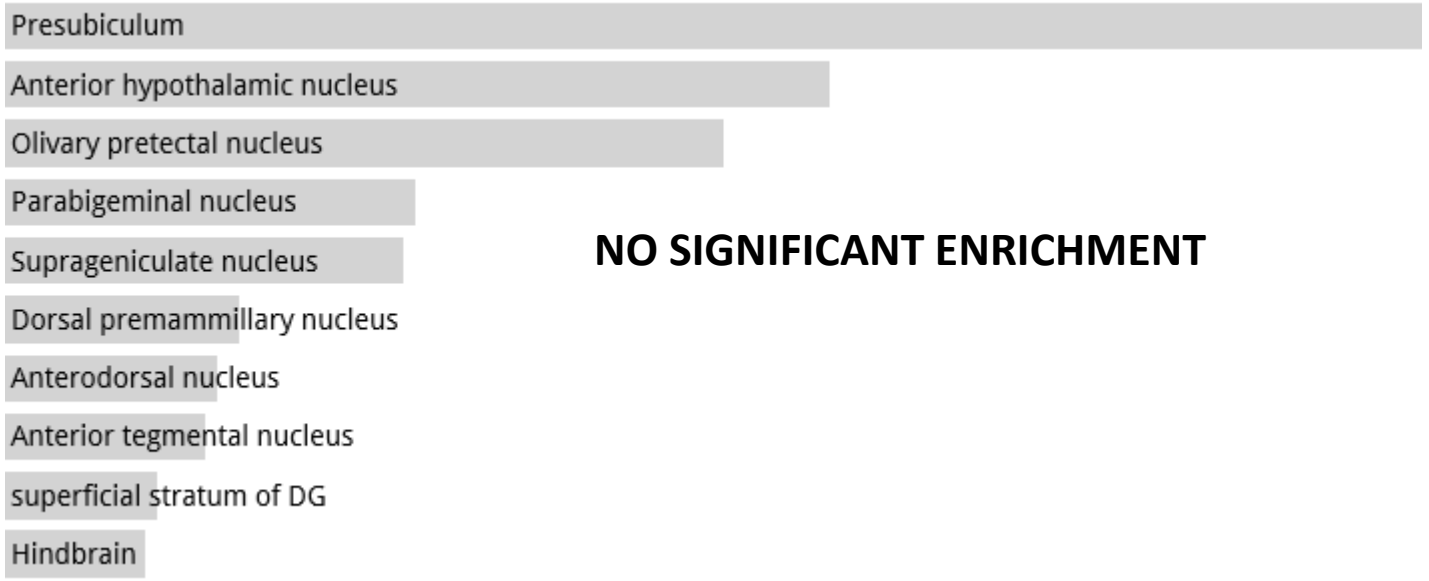
Disease Perturbations from GEO down random 4000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 13

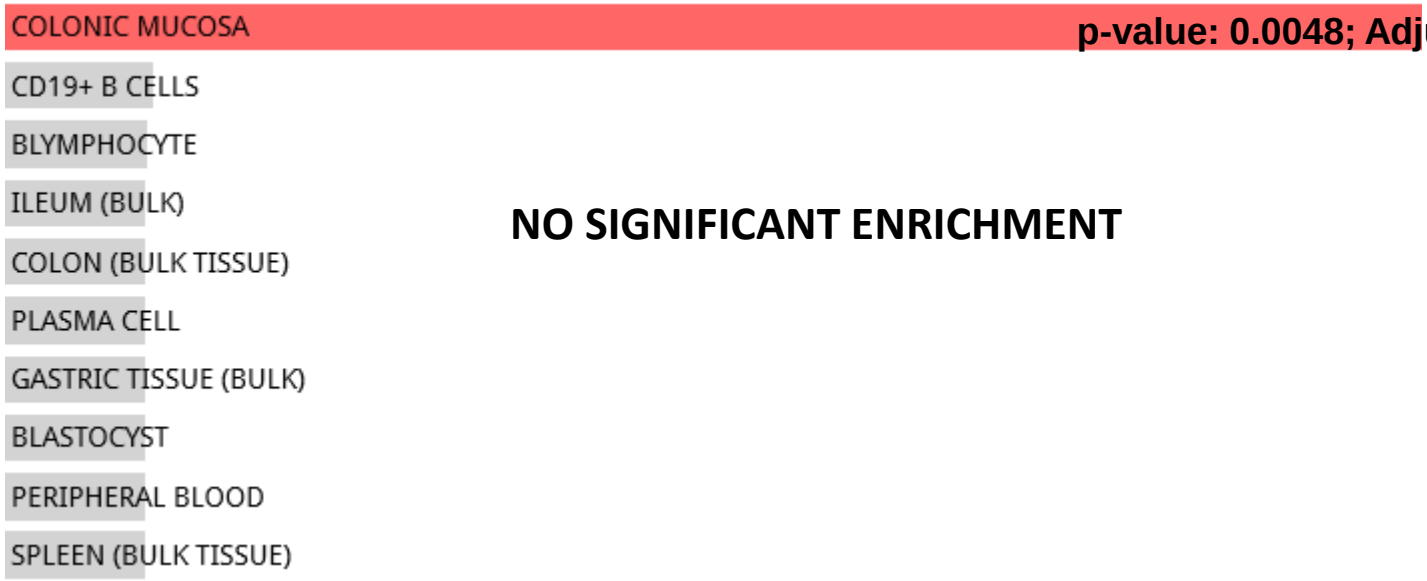
Allen Brain Atlas up random 4000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 14

ARCHS4 Tissues random 4000 genes



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

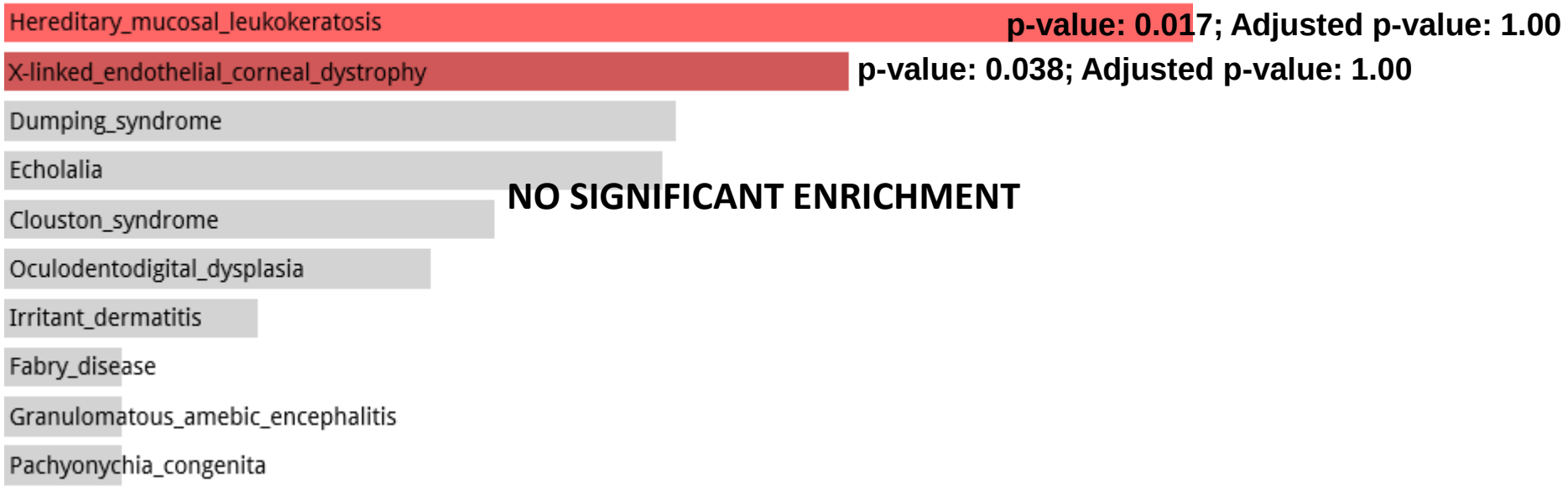
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

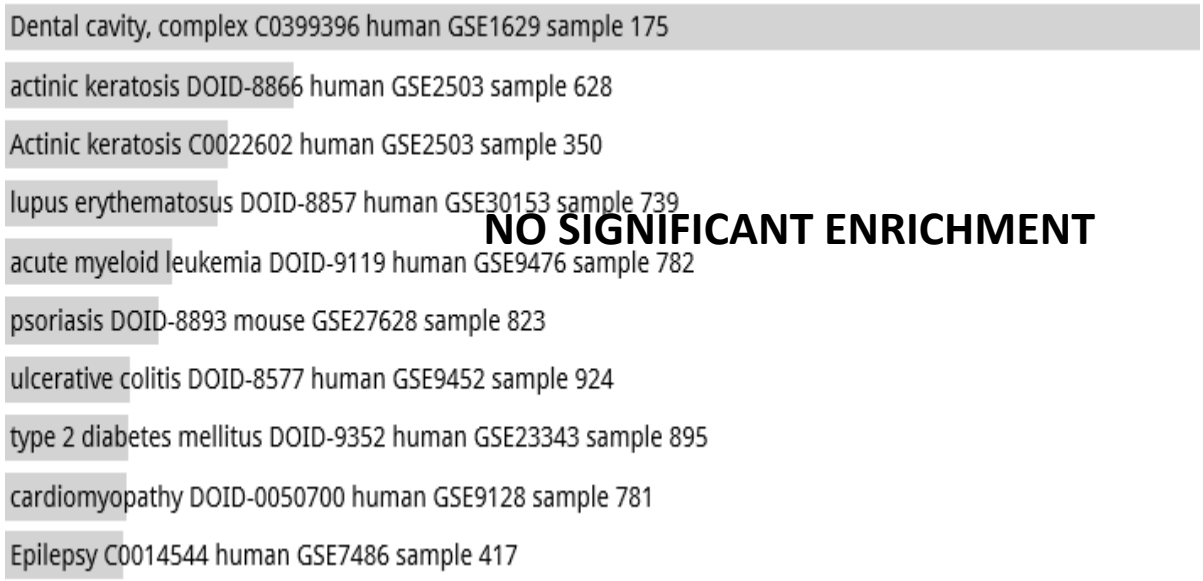
Random Set 14

Jensen DISEASES random 4000 genes



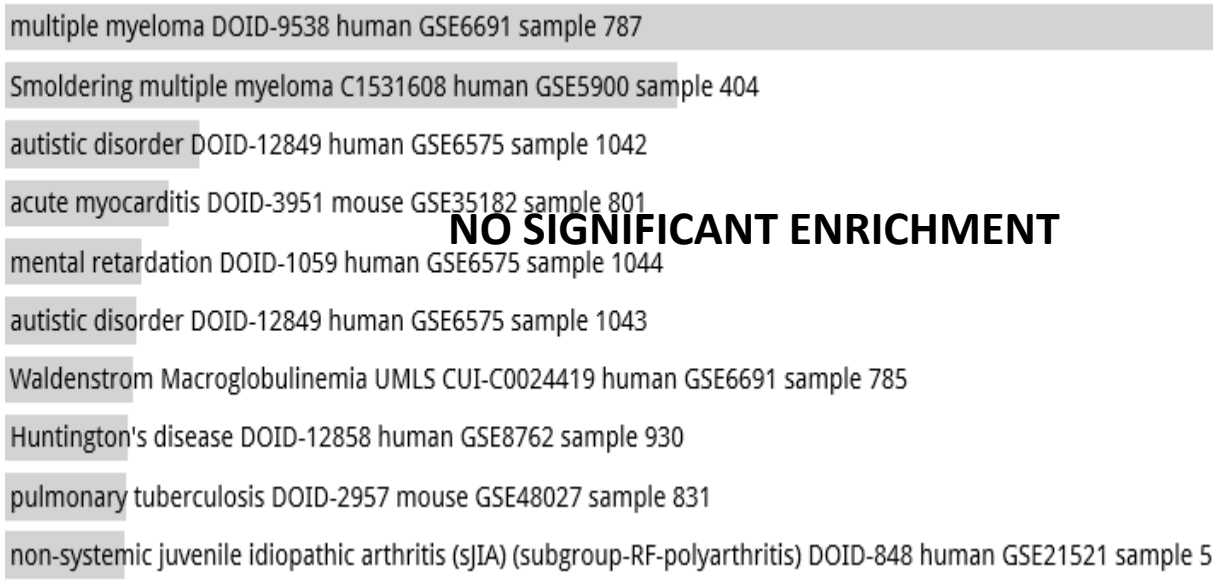
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 4000 genes



NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 4000 genes



NO SIGNIFICANT ENRICHMENT

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

Lateral reticular nucleus, parvicellular part

mantle zone of r9Ve

r4 part of magnocellular medial vestibular nucleus

periventricular stratum of r8Tr

superficial stratum of r7Lim

NO SIGNIFICANT ENRICHMENT

Random Set 15

ARCHS4 Tissues random 4000 genes

COLON (BULK TISSUE)
GASTRIC TISSUE (BULK)
PREFRONTAL CORTEX
ILEUM (BULK)
LUNG (BULK TISSUE)
NEURONAL EPITHELIUM
OMENTUM
LIVER (BULK TISSUE)
AMNIOTIC FLUID
RENAL CORTEX

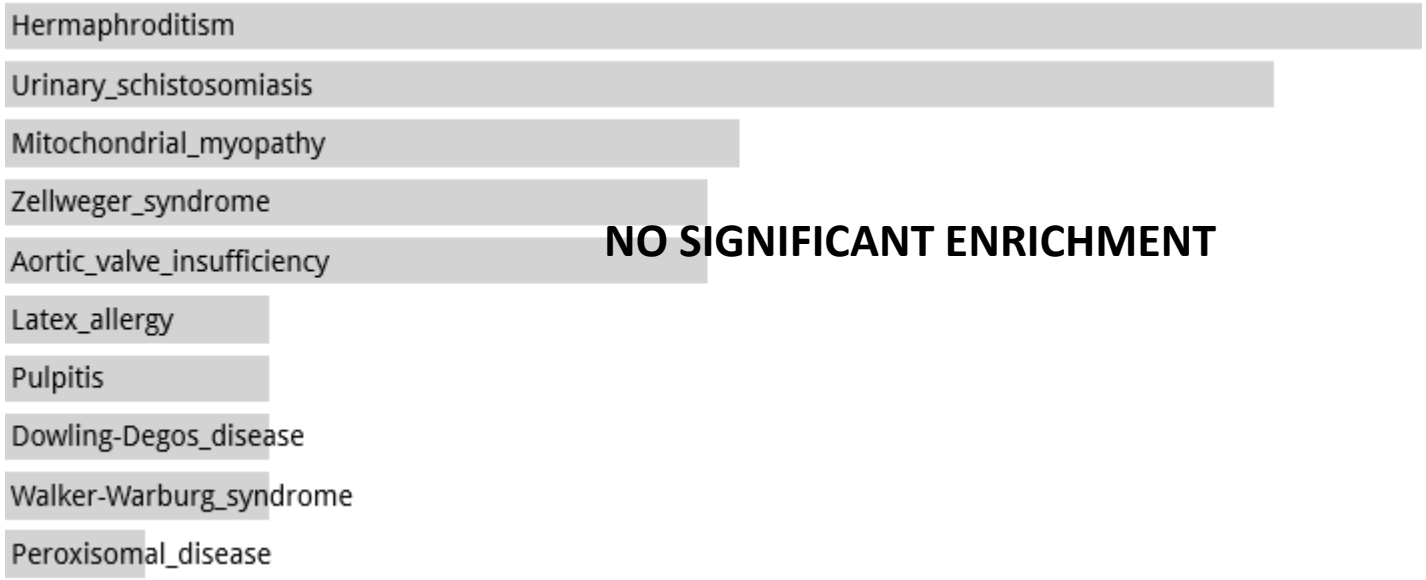
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

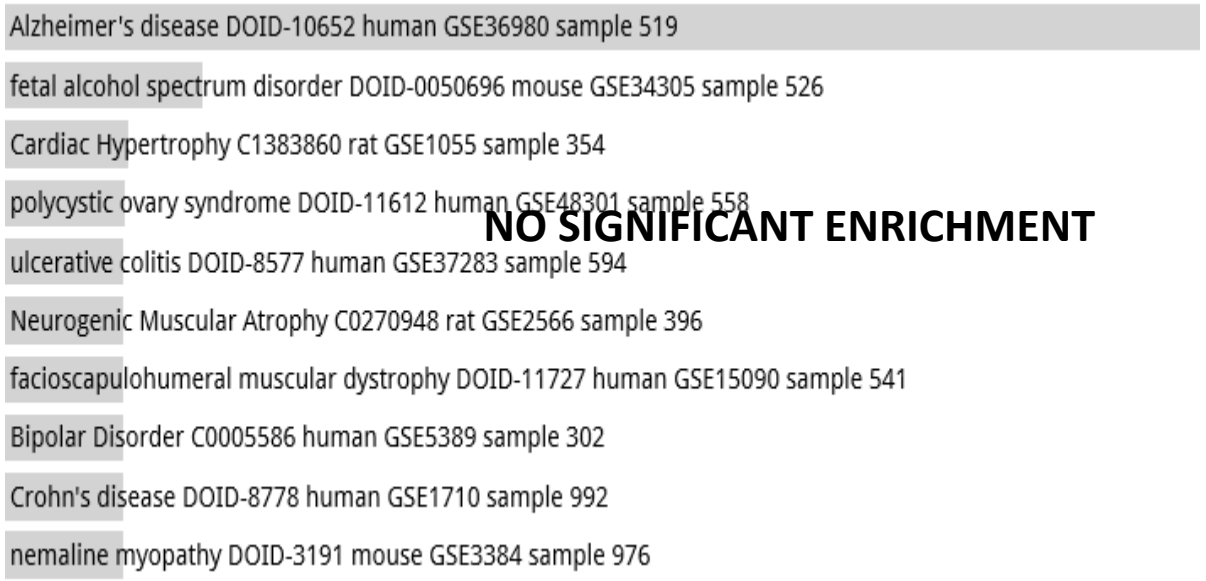
Random Set 15

Jensen DISEASES random 4000 genes



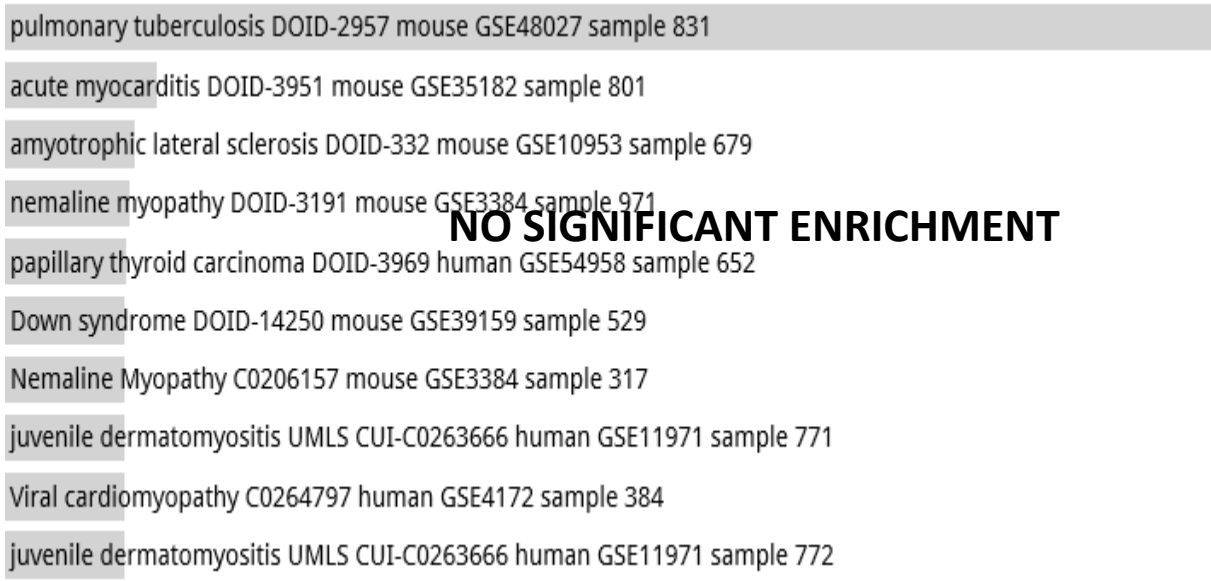
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 4000 genes



NO SIGNIFICANT ENRICHMENT

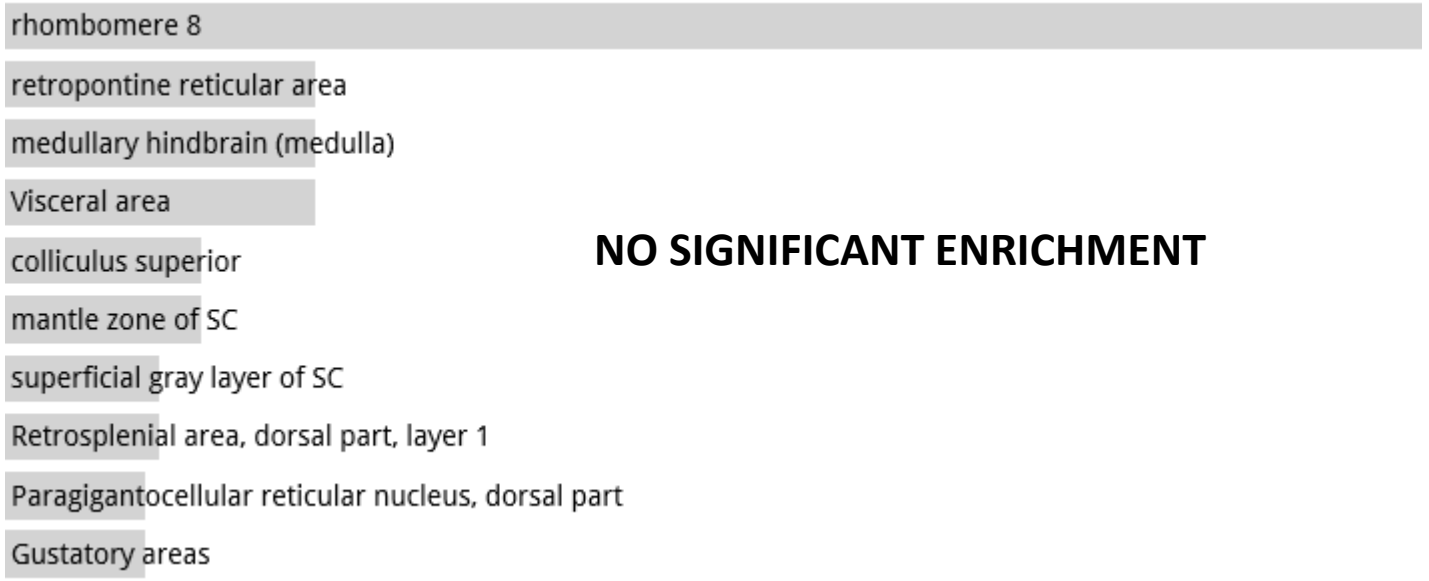
Disease Perturbations from GEO down random 4000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 15

Allen Brain Atlas up random 4000 genes



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

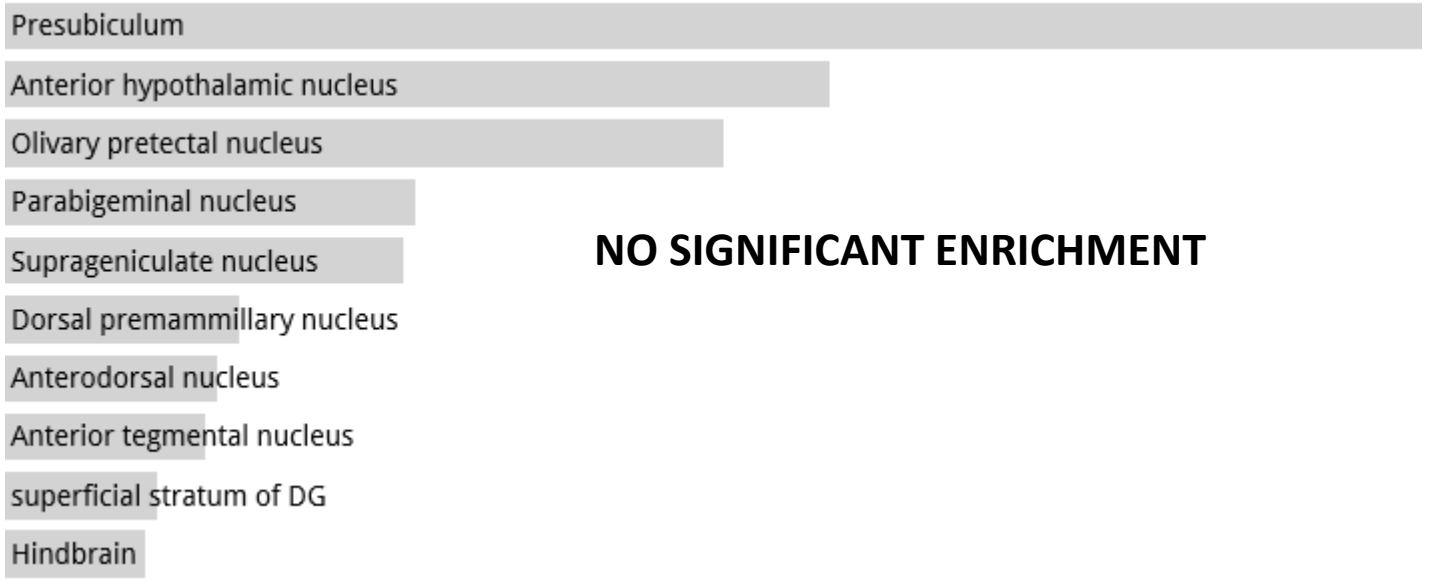
Disease Perturbations from GEO down random 4000 genes

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
Schistosomiases 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

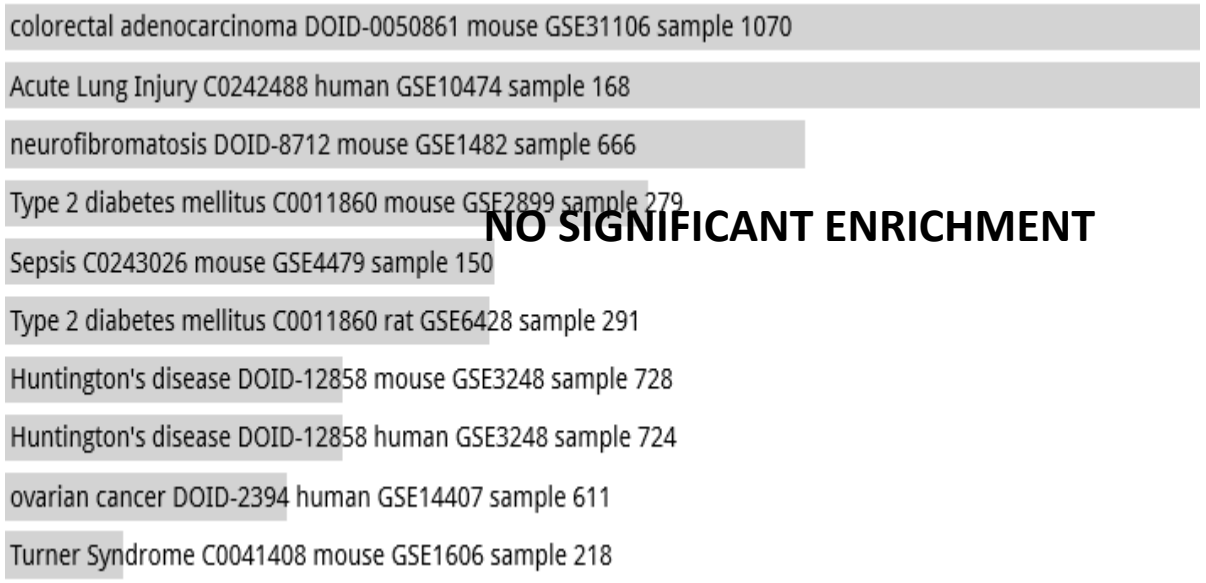
NO DATA AVAILABLE

Random Set 17

Jensen DISEASES random 4000 genes

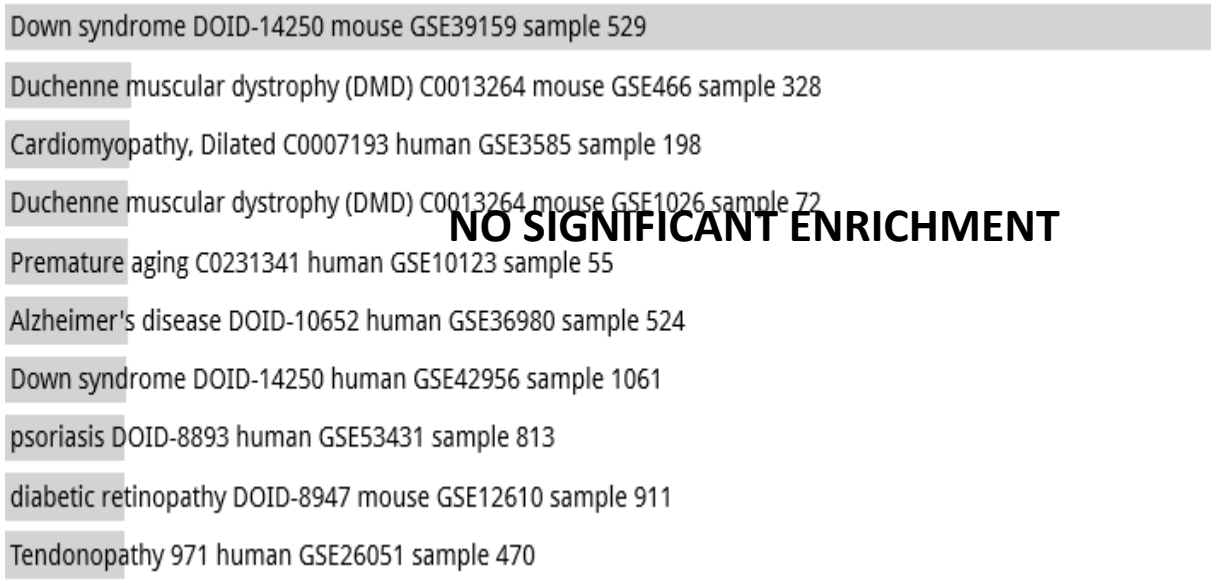
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 4000 genes



NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO down random 4000 genes



NO SIGNIFICANT ENRICHMENT

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

NO DATA AVAILABLE

Disease Perturbations from GEO down random 4000 genes

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

NO DATA AVAILABLE

Disease Perturbations from GEO down random 4000 genes

NO DATA AVAILABLE

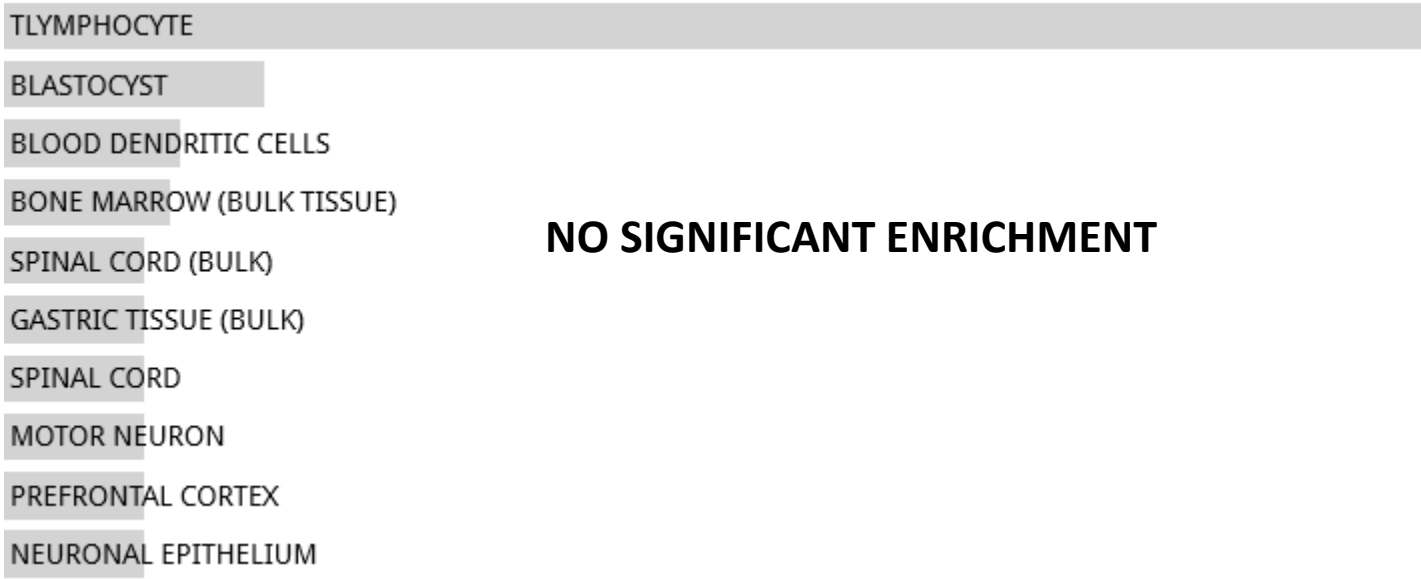
Random Set 19

Allen Brain Atlas up random 4000 genes

NO DATA AVAILABLE

Random Set 20

ARCHS4 Tissues random 4000 genes



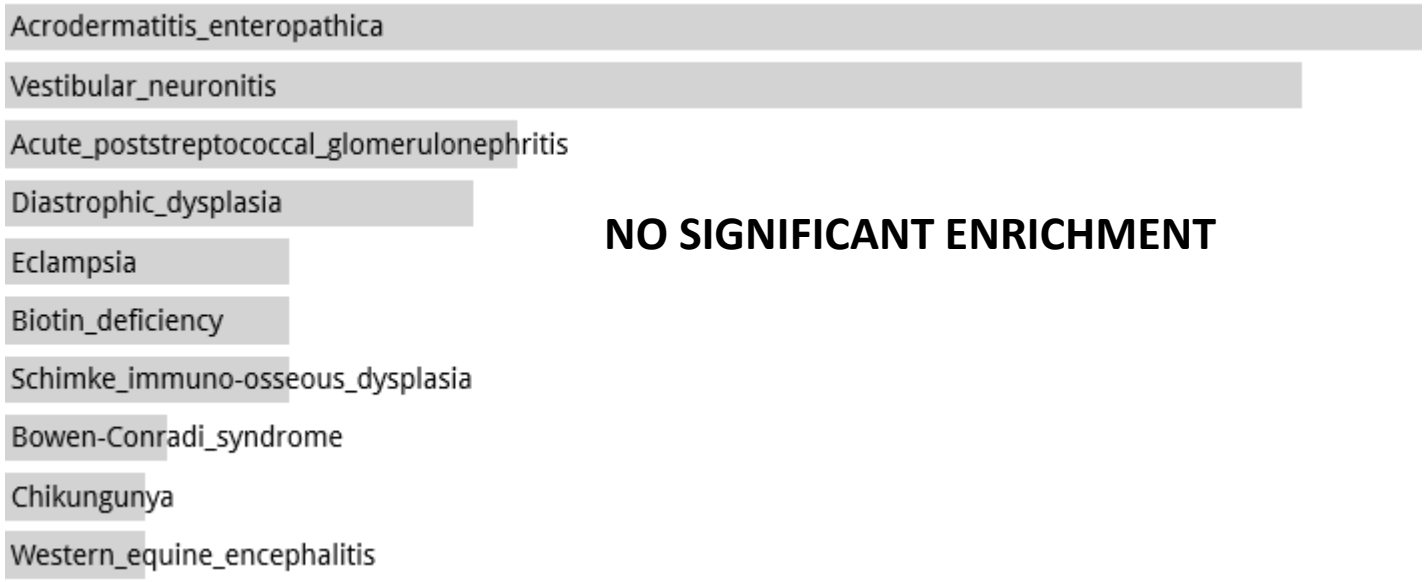
NO SIGNIFICANT ENRICHMENT

Jensen TISSUES random 4000 genes

NO DATA AVAILABLE

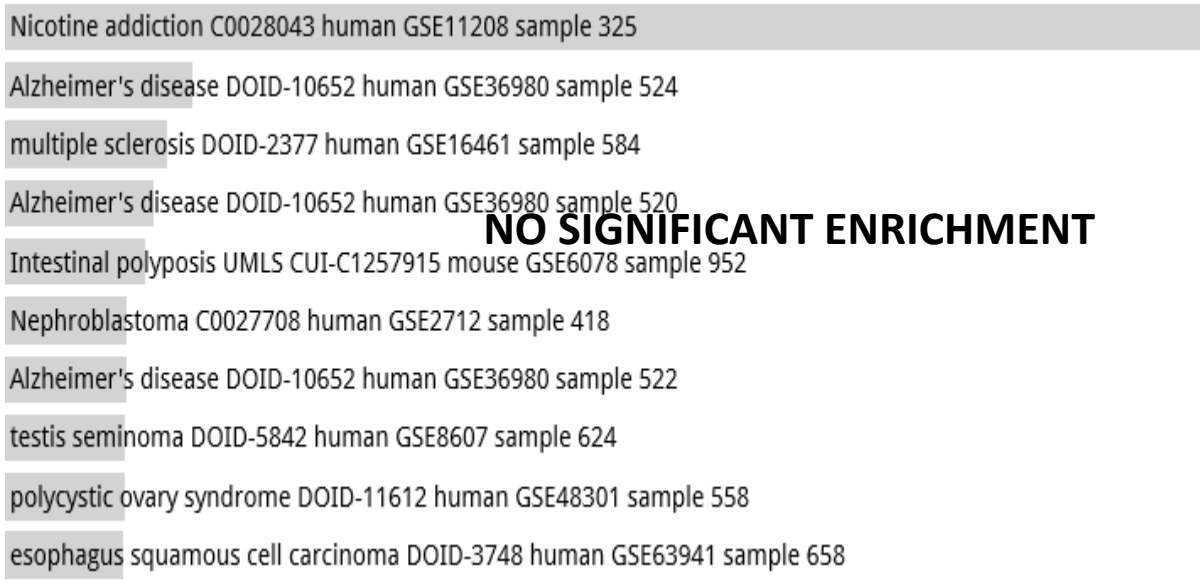
Random Set 20

Jensen DISEASES random 4000 genes



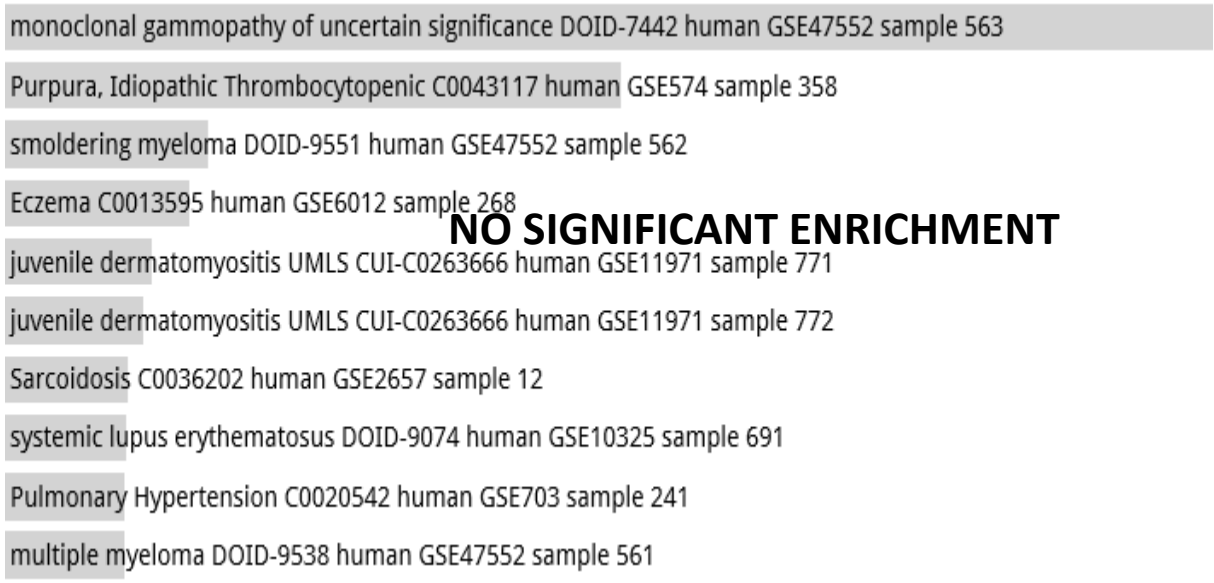
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 4000 genes



NO SIGNIFICANT ENRICHMENT

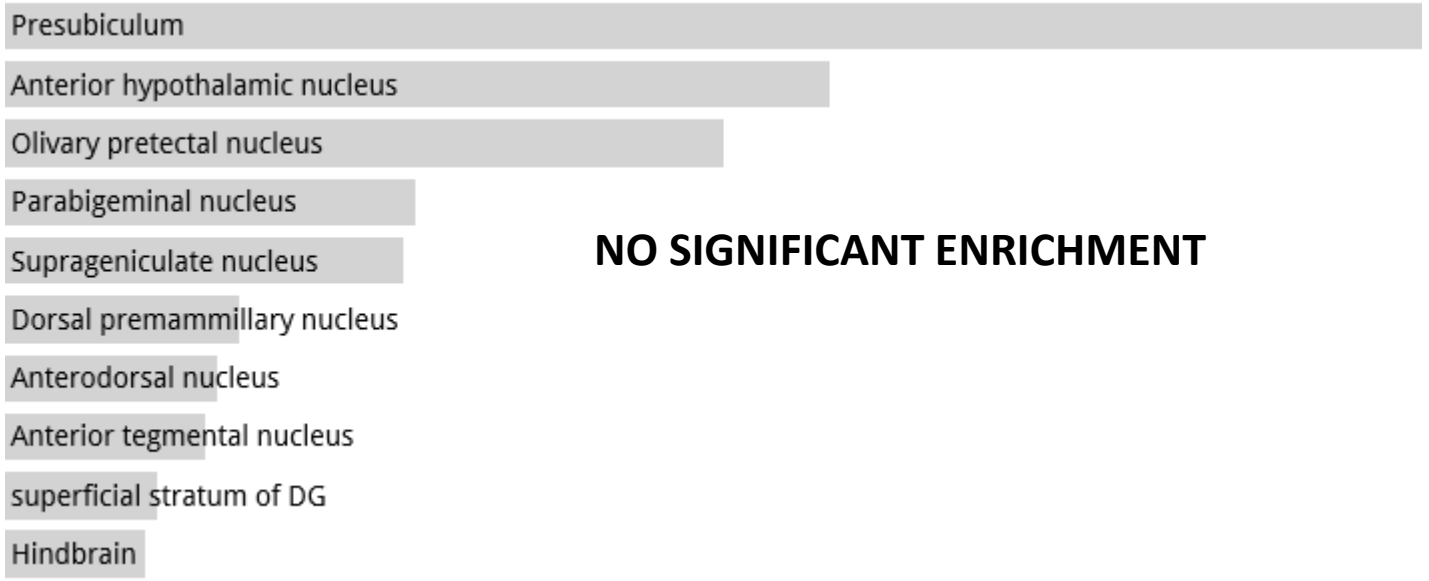
Disease Perturbations from GEO down random 4000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 20

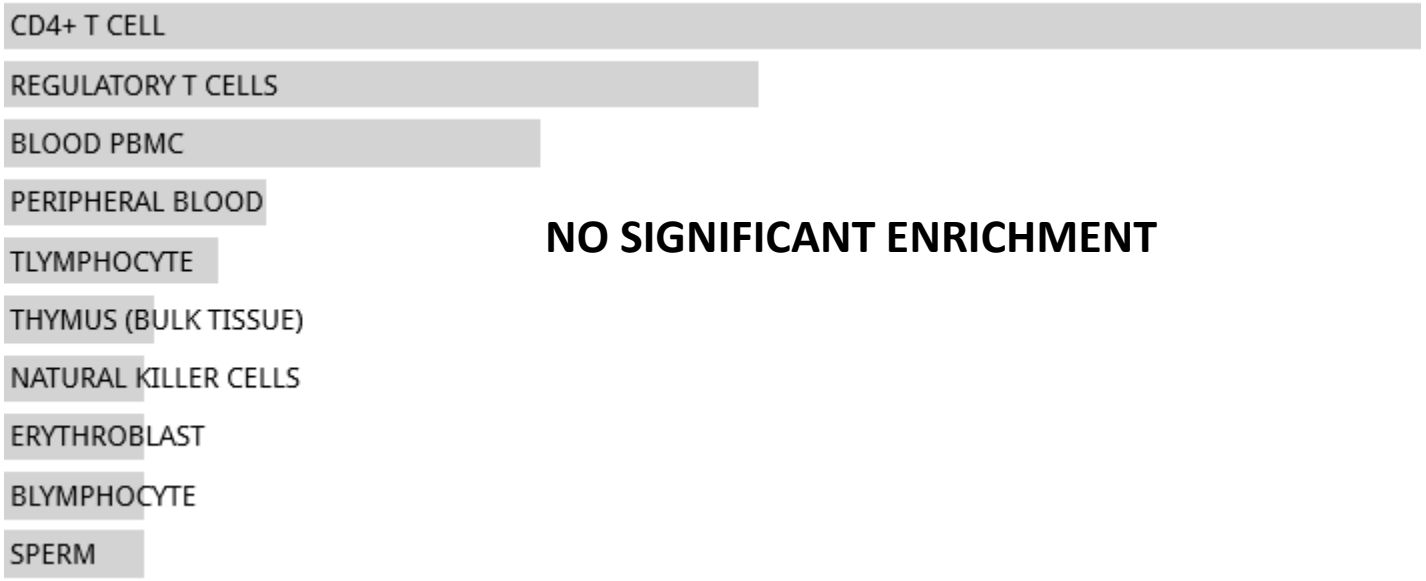
Allen Brain Atlas up random 4000 genes



NO SIGNIFICANT ENRICHMENT

Random Set 21

ARCHS4 Tissues random 2847 genes



NO SIGNIFICANT ENRICHMENT

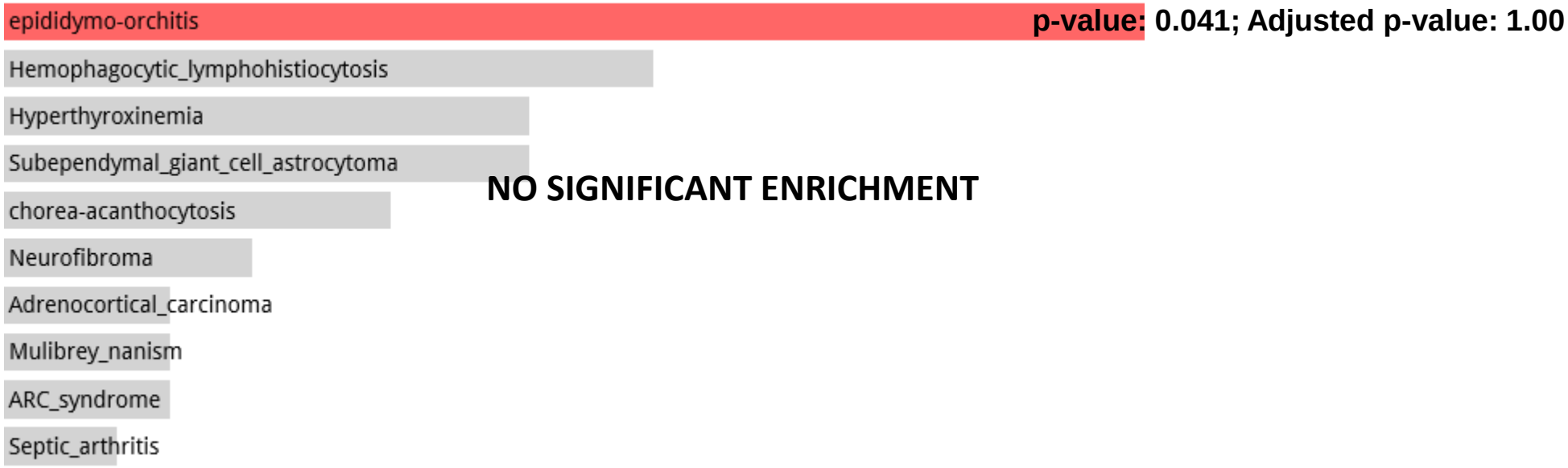
Jensen TISSUES random 2847 genes

p-value: 0.042; Adjusted p-value: 1.00

NO SIGNIFICANT ENRICHMENT

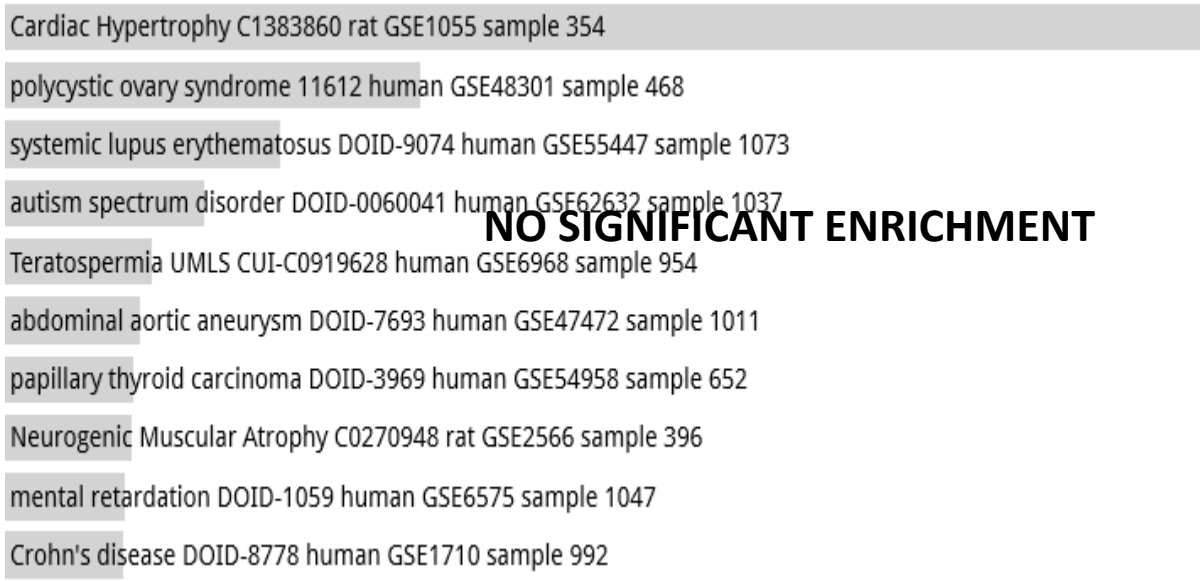
Random Set 21

Jensen DISEASES random 2847 genes



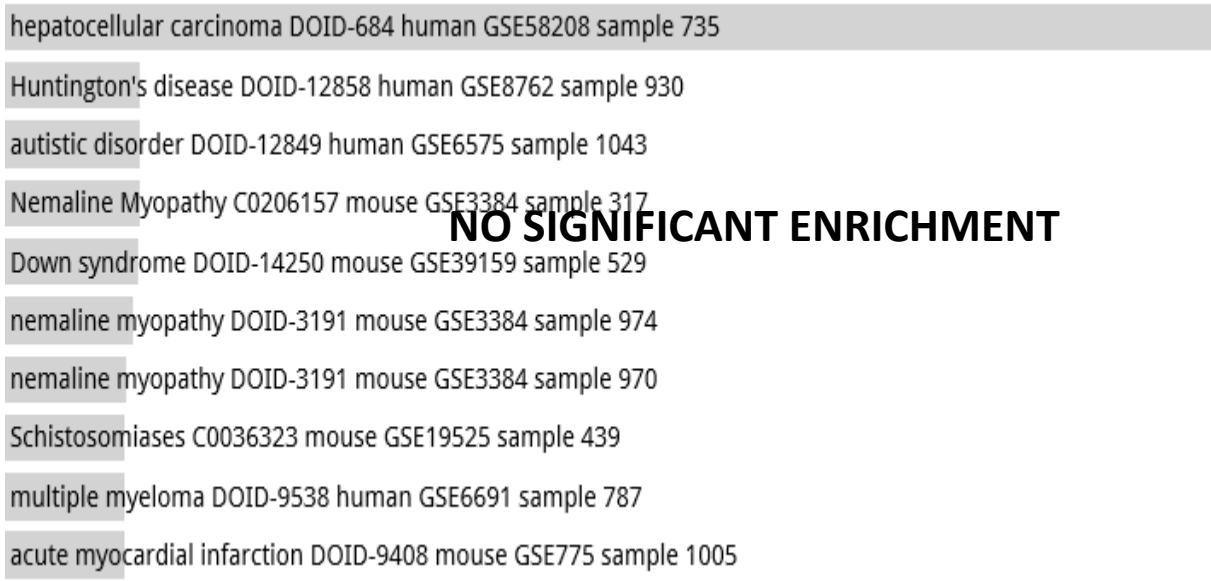
NO SIGNIFICANT ENRICHMENT

Disease Perturbations from GEO up random 2847 genes



NO SIGNIFICANT ENRICHMENT

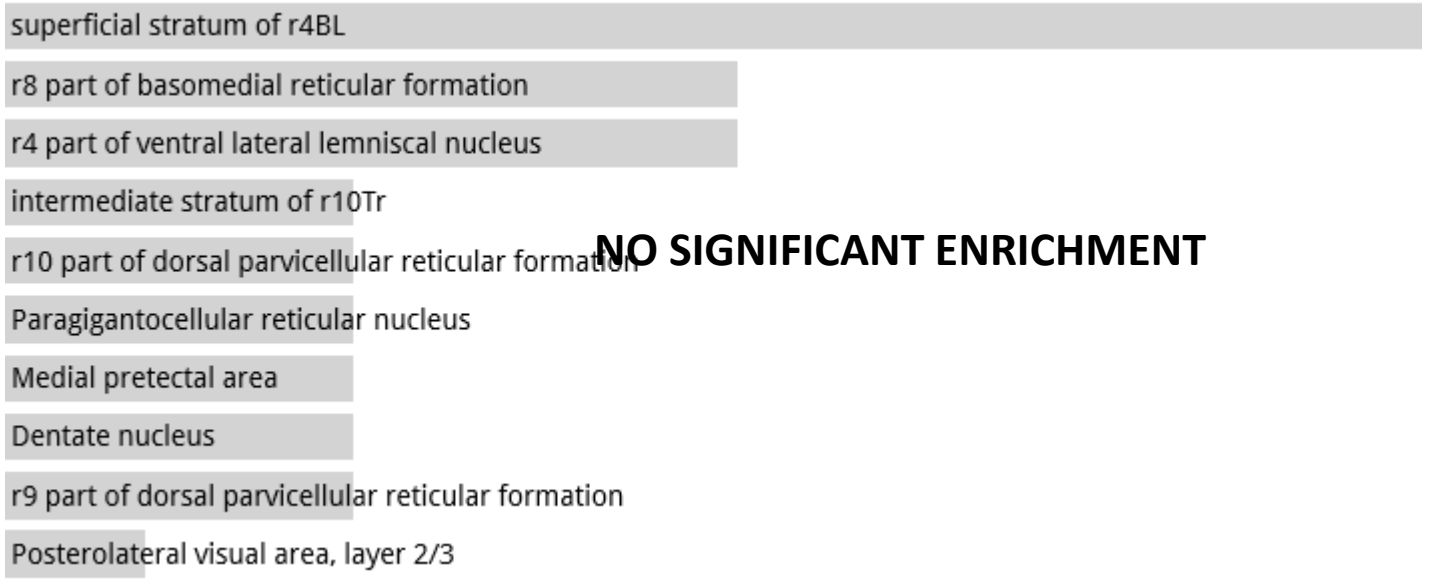
Disease Perturbations from GEO down random 2847 genes



NO SIGNIFICANT ENRICHMENT

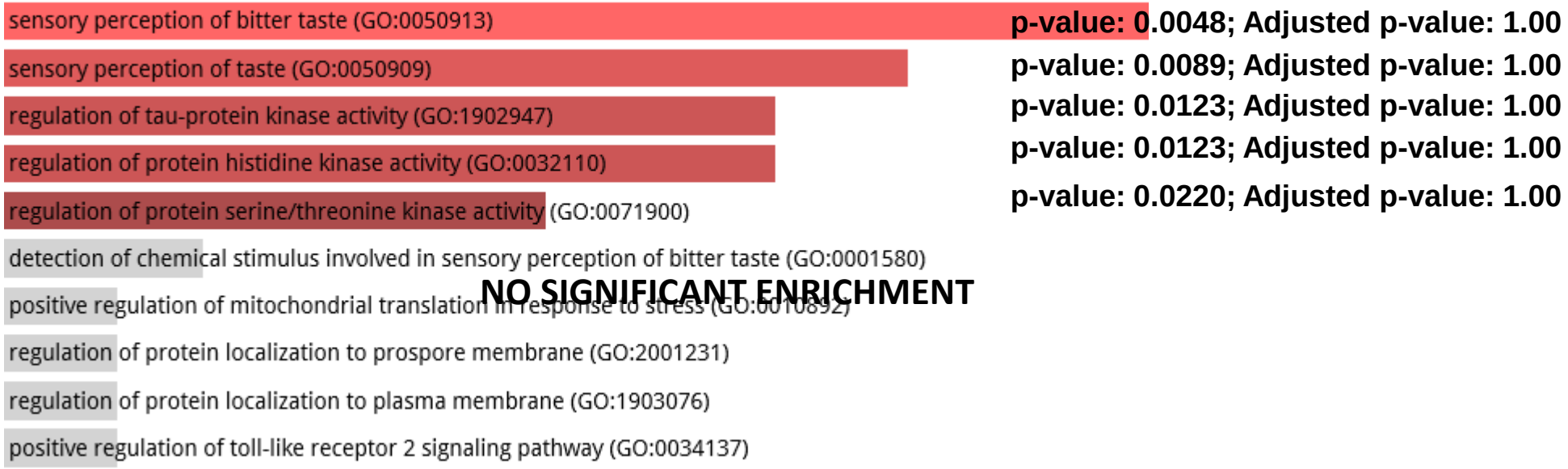
Random Set 21

Allen Brain Atlas up random 2847 genes



NO SIGNIFICANT ENRICHMENT

GO Biological Process 2017b 2847 genes

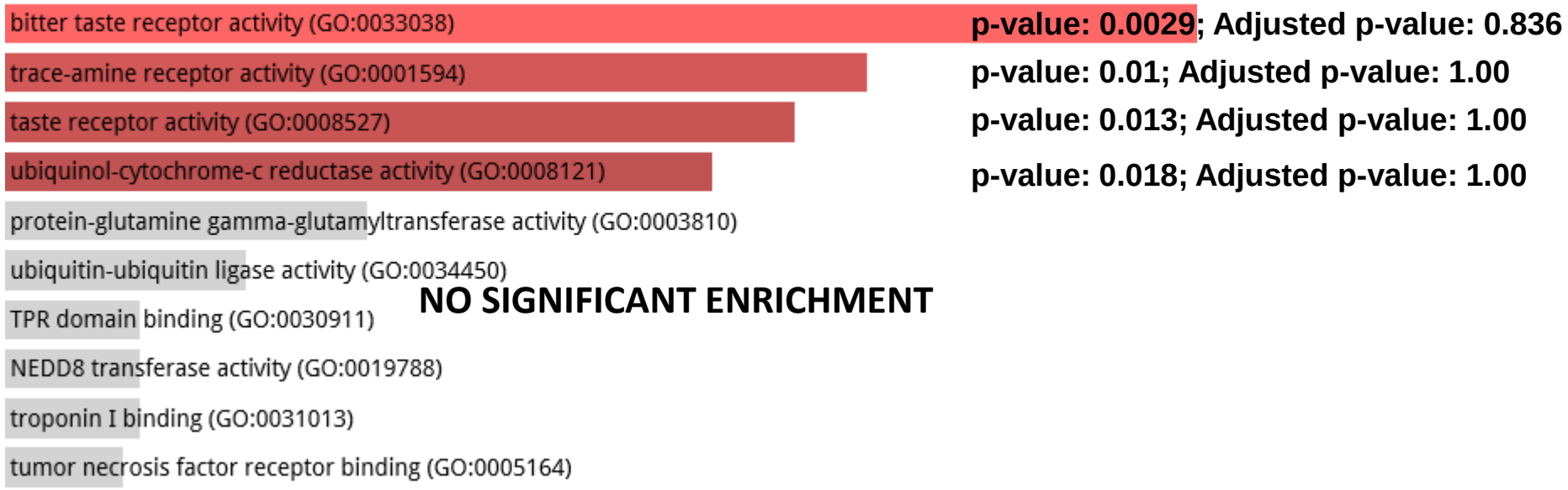


p-value: 0.0048; Adjusted p-value: 1.00
p-value: 0.0089; Adjusted p-value: 1.00
p-value: 0.0123; Adjusted p-value: 1.00
p-value: 0.0123; Adjusted p-value: 1.00
p-value: 0.0220; Adjusted p-value: 1.00

NO SIGNIFICANT ENRICHMENT

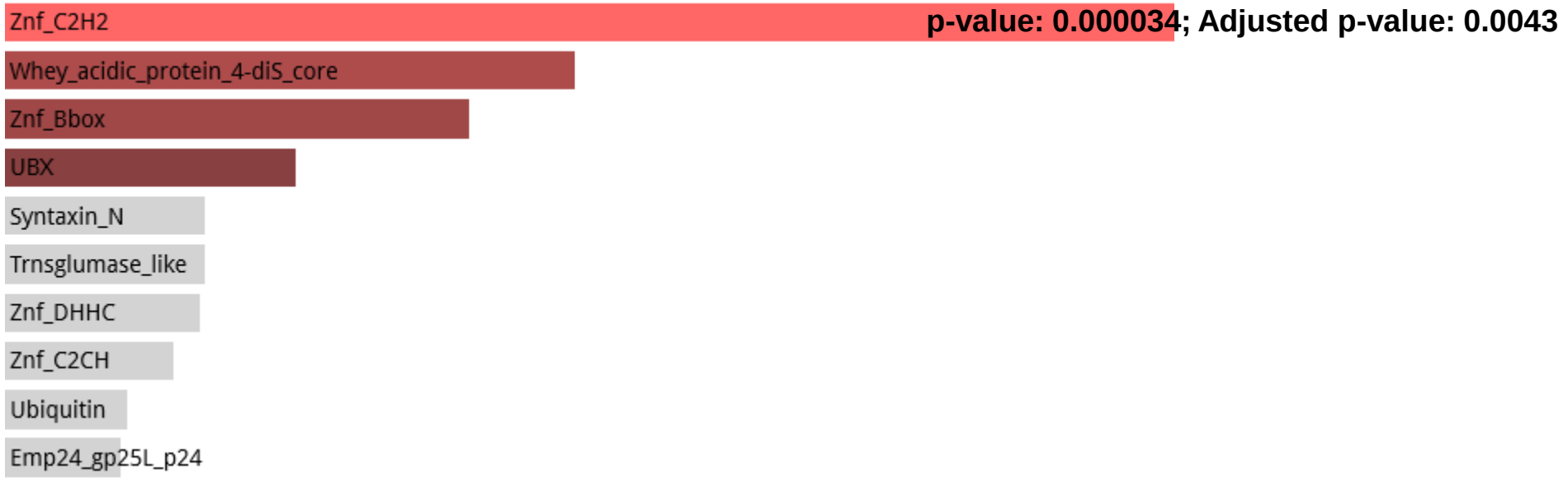
Random Set 21

GO Molecular Function 2017 2847 genes



NO SIGNIFICANT ENRICHMENT

Pfam InterPro Domains 2847 genes



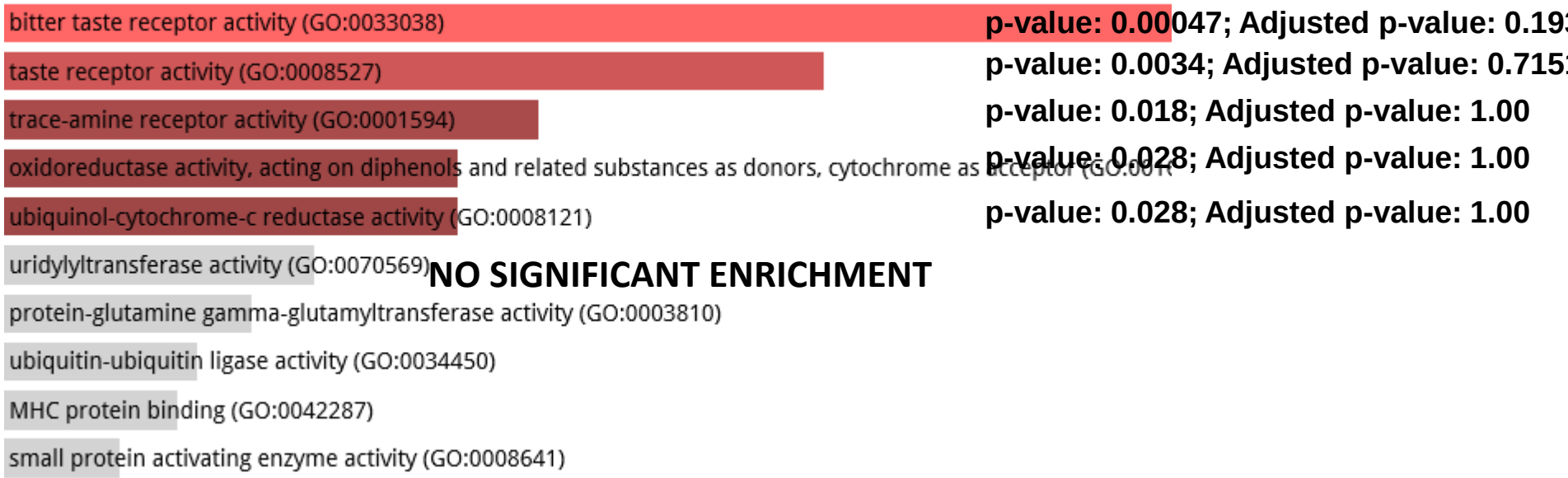
Random Set 21

GWAS Catalog 2019 2847 genes

p-value: 0.00049; Adjusted p-value: 0.31

NO SIGNIFICANT ENRICHMENT

GO Biological Process 2018 2847 genes



p-value: 0.00047; Adjusted p-value: 0.1939

p-value: 0.0034; Adjusted p-value: 0.7151

p-value: 0.018; Adjusted p-value: 1.00

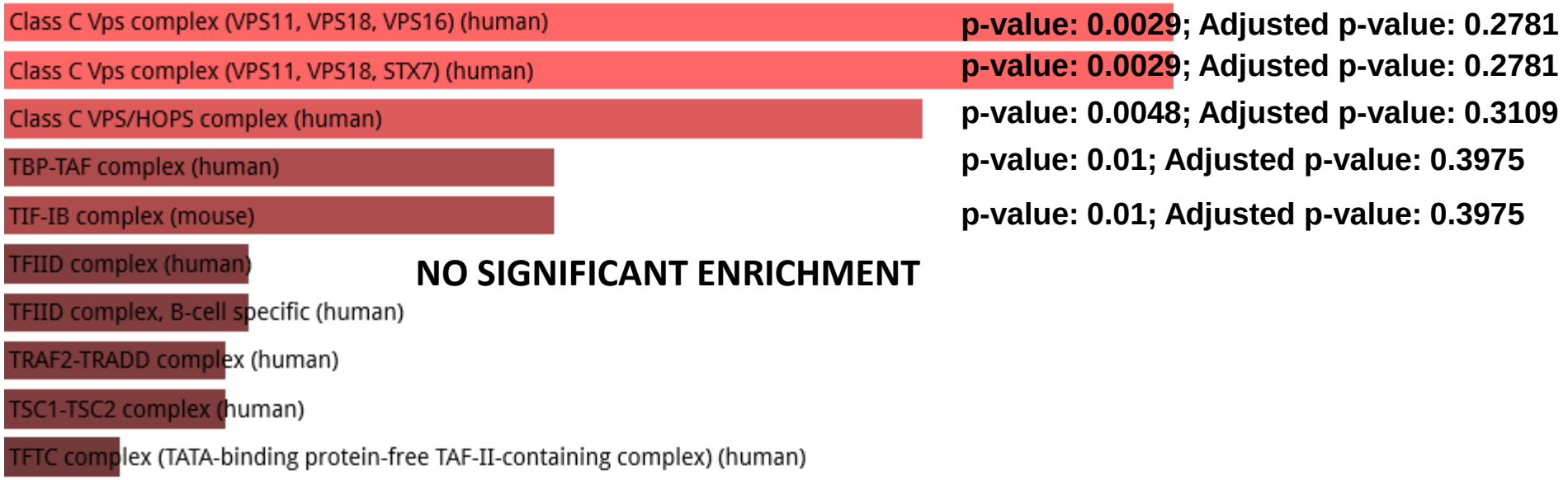
p-value: 0.028; Adjusted p-value: 1.00

p-value: 0.028; Adjusted p-value: 1.00

NO SIGNIFICANT ENRICHMENT

Random Set 21

CORUM 2847 genes



p-value: 0.0029; Adjusted p-value: 0.2781

p-value: 0.0029; Adjusted p-value: 0.2781

p-value: 0.0048; Adjusted p-value: 0.3109

p-value: 0.01; Adjusted p-value: 0.3975

p-value: 0.01; Adjusted p-value: 0.3975

NO SIGNIFICANT ENRICHMENT

CORUM database of protein complexes identified by mass spectrometry