



Icahn  
School of  
Medicine at  
Mount  
Sinai

*The Tisch  
Cancer Institute*



*Black Family  
Stem Cell Institute*

*Skin Biology  
and Diseases  
Resource-based  
Center*

*Department of Cell,  
Developmental and  
Regenerative Biology*



# Introduction to the Bioinformatics for Next Generation Sequencing (BiNGS) Shared Resource Facility

---

**BFSCI/CDRB/SBDRC**

**Sep 15, 2022**

**Dan Hasson**

# Acknowledgments

---



- Ramon Parsons
- Elena Ezhkova
- Sarah Millar
- Marek Mlodzik
- Sharon Mias
- Caba Amanda
- Evelina Berman
- Genevieve Joseph
- Donnaann Chiode
- Chan-Bene Lin
- Parbattie (Pearl) Arnold-Sukhram
- Emily Bernstein
- Geeta Leung
- Ed Hicks
- Nicole Jefferson
- Cepeda Nyomi
- Eugene Folder
- Lili Gai

# Mission

---



- To provide high-end data analysis service at reduced cost to support TCI-, BFSCI-, CDRB-, and SBDRC-affiliated faculty members in the planning, analysis, integration and interpretation of NGS datasets tailored to their research.
- We advise on NGS experimental design and protocols.
- Provide a broad range of bioinformatic analyses (standard and customized) for multiple applications spanning transcriptomics, epigenomics, and genomics.
- Provide extensive training platforms.

# Our Team



**Ernesto Guccione**  
Co-Director



**Dan Hasson**  
Co-Director



**Deniz Demircioglu**  
Lead Bioinformatician



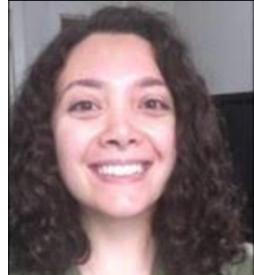
**Saul Carcamo**  
Bioinformatician



**Xuedi Wang**  
Bioinformatician



**Gargi Damle**  
Junior Bioinformatician



**Gulay Ulukaya**  
Junior Bioinformatician



**Charles Coleman**  
Junior Bioinformatician



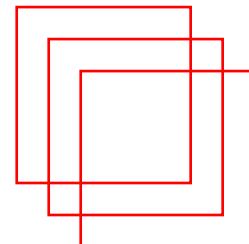
**Aman Agarwal**  
Junior Bioinformatician



**Lewis Tomalin**  
Bioinformatician



**Alexander Tsankov**  
Consultant



Actively recruiting  
3x bioinformaticians

# The Goal

---



Create an **informal long-term relationship** between your lab and the core where we;

- Advise on experimental design, perform data analysis and support data generation for publication.
- Our bioinformaticians becomes part of your lab participating in lab meetings and reading literature specific to your studies so their analysis is more informed.
- We meet regularly to discuss results and to set goals for data analysis.
- We are incorporated into your grants.
- We train your lab members and provide tools so they can dig deeper into the data.



Icahn  
School of  
Medicine at  
Mount  
Sinai

*The Tisch  
Cancer Institute*



*Black Family  
Stem Cell Institute*

*Skin Biology  
and Diseases  
Resource-based  
Center*

*Department of Cell,  
Developmental and  
Regenerative Biology*



# Overview of BiNGS services

---



# Transcriptomics



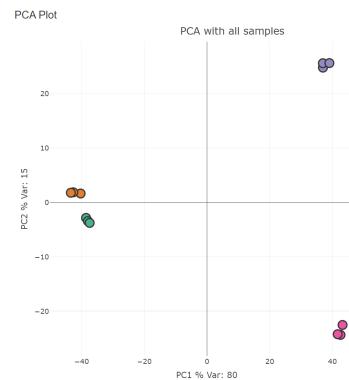
- Bulk RNA-seq
- Alternative Splicing Analysis
- Alternative Promoter Analysis
- Iso-seq
- Transcriptional Analysis of TCGA and other public datasets

# Bulk RNA-seq



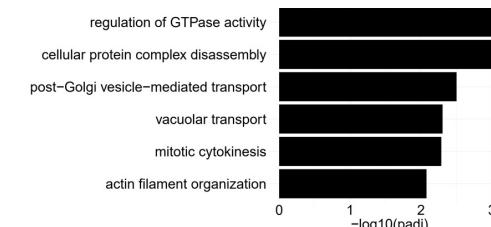
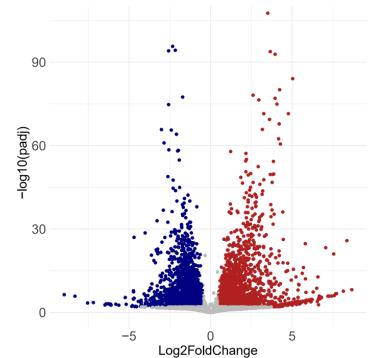
## Standard analysis:

- Evaluation of reads quality and alignment statistics
- Samples normalization using internal controls or computational methods
- A link to a UCSC genome browser session for all normalized datasets
- Normalized read counts (TPM)
- Assessment of sample similarity (PCA plot)
- Differential gene expression (tables and interactive heatmaps)
- Gene Set Enrichment Analysis (GSEA), Gene-Ontology terms and pathway enrichment analysis



## Customized analysis:

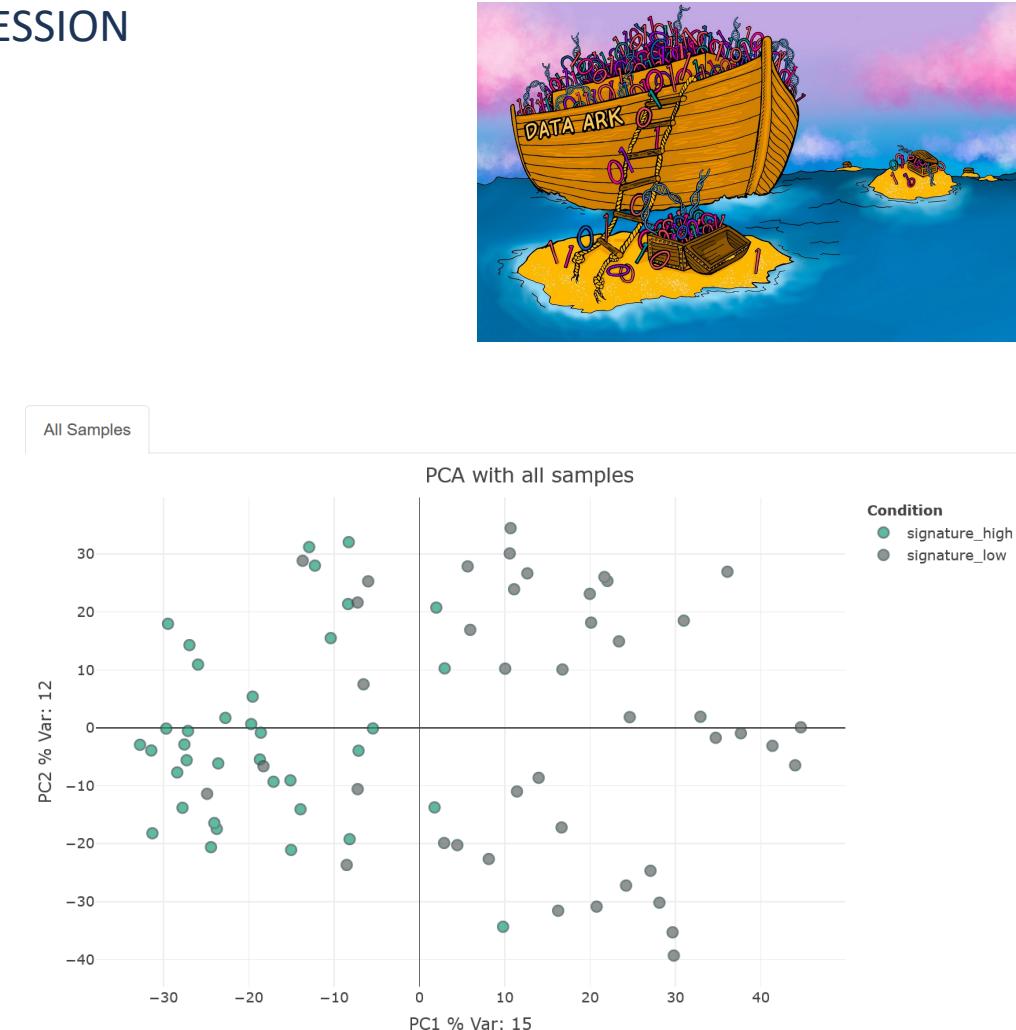
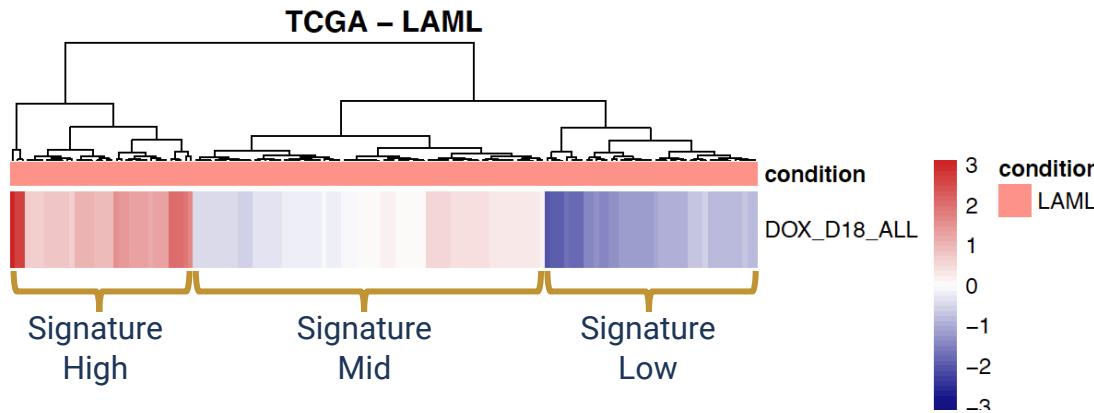
- Motif discovery
- Gene expression modules
- Data integration (e.g. ATAC-seq, ChIP-seq)
- Data integration with publicly available resources (e.g. ENCODE, TCGA)
- Publication quality figures



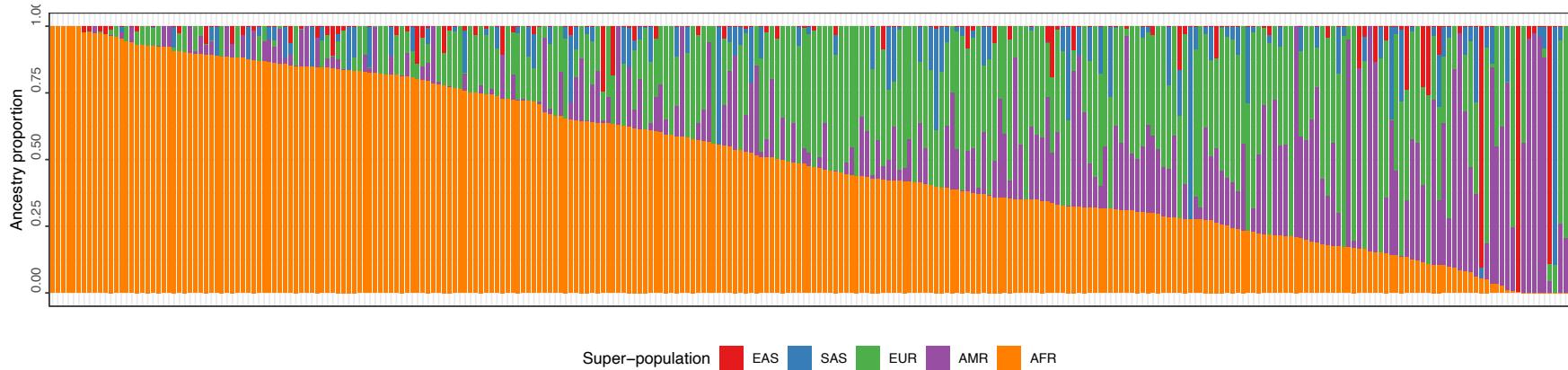
# TCGA Signature Analysis (or other publicly available datasets)



CLUSTERING OF TCGA DATA BASED ON DIFFERENTIAL GENE EXPRESSION SIGNATURE (ssGSEA) FROM A CELL LINE UPON PERTURBATION



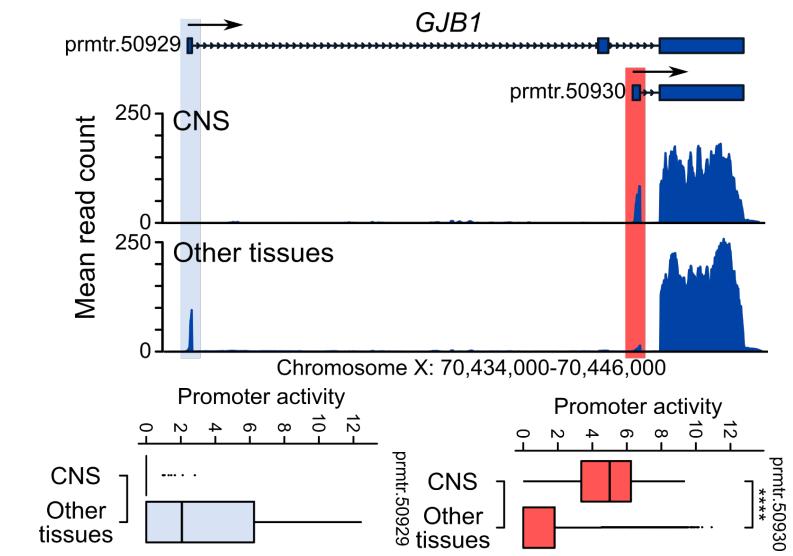
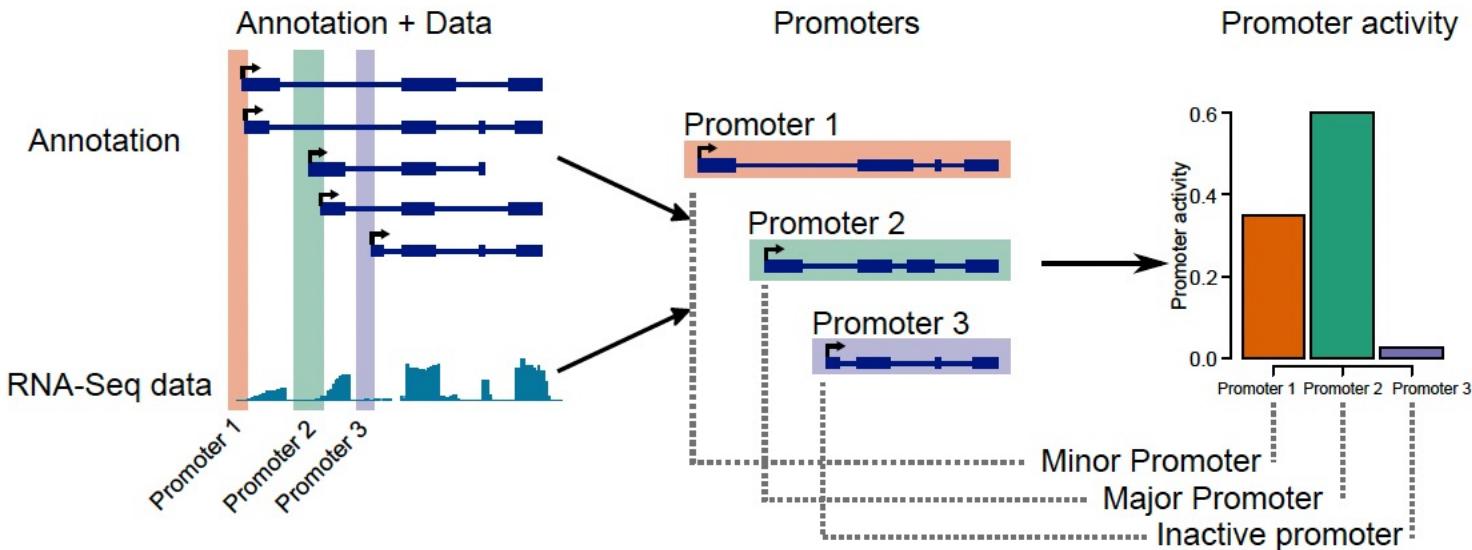
# Ancestry analysis (bulk RNA-seq)



# Alternative Promoters



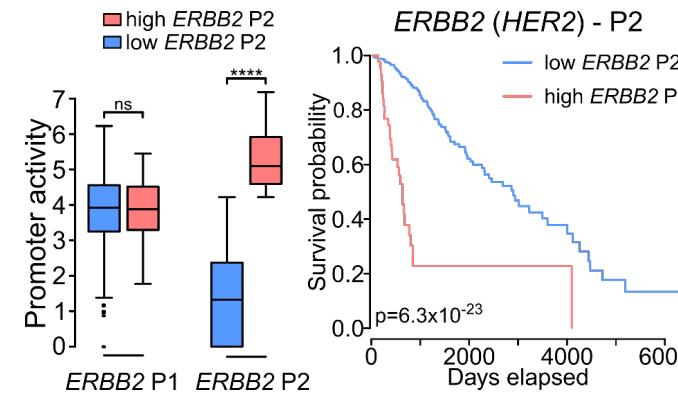
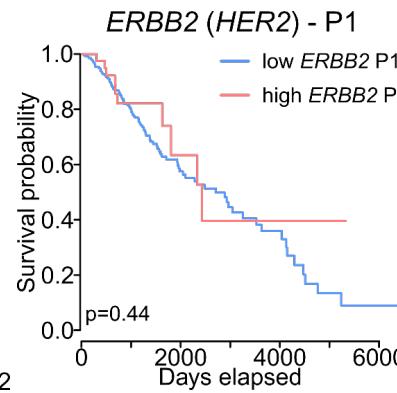
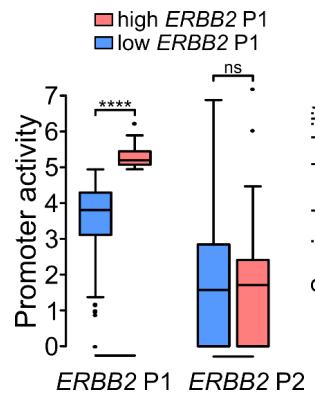
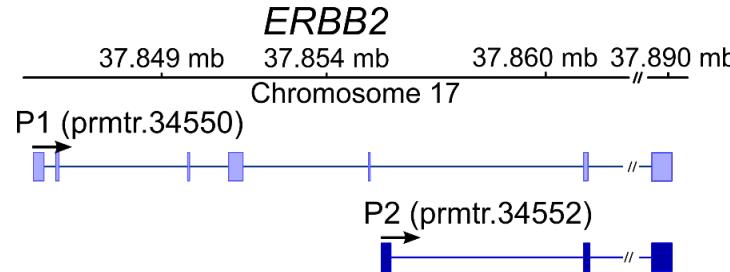
A Pan-cancer Transcriptome Analysis Reveals Pervasive Regulation through Alternative Promoters



# Alternative Promoters (TCGA)



A Pan-cancer Transcriptome Analysis Reveals Pervasive Regulation through Alternative Promoters



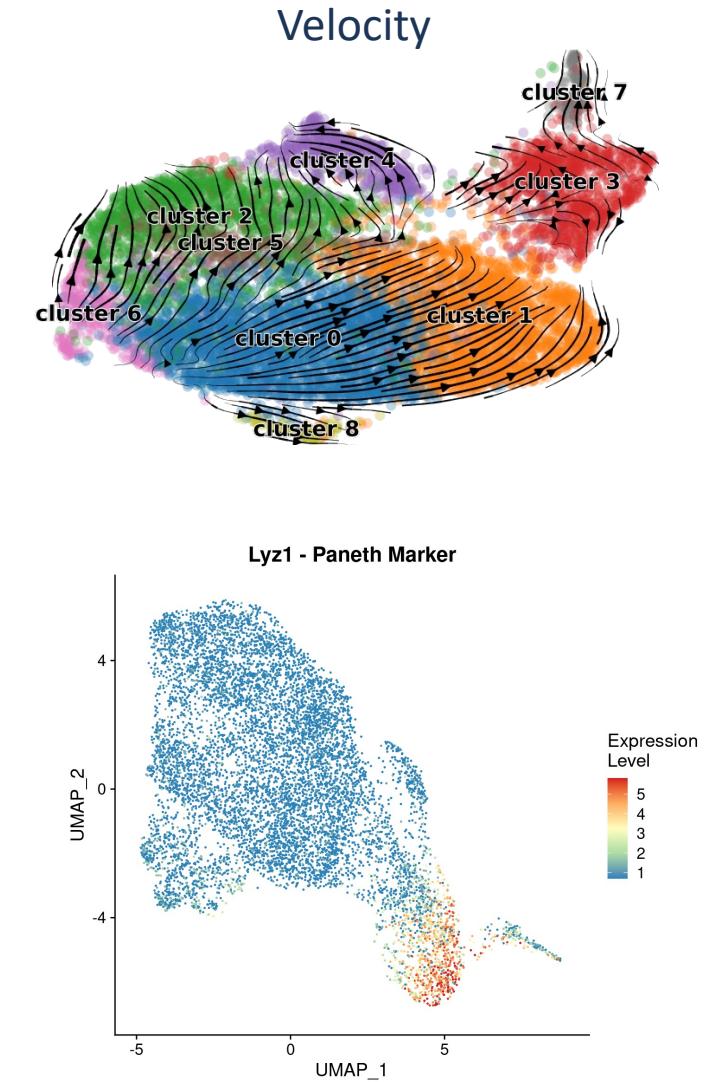
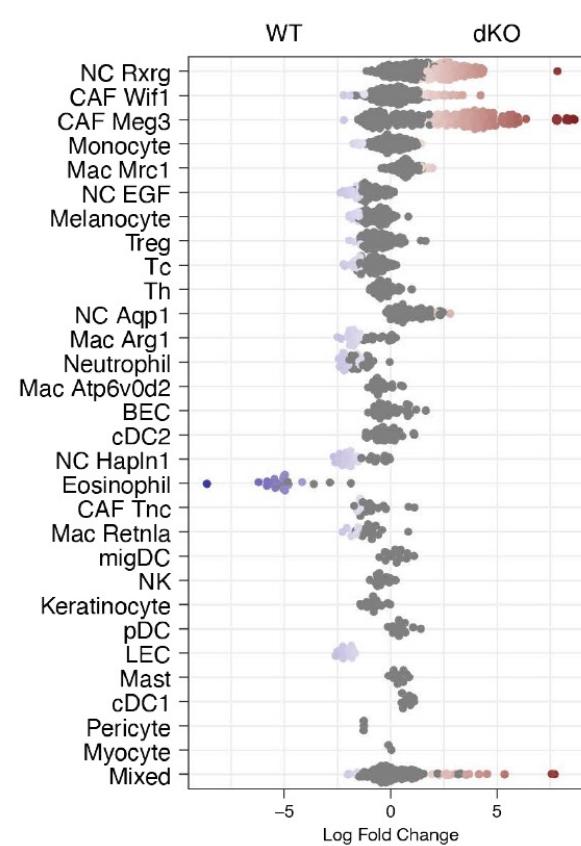
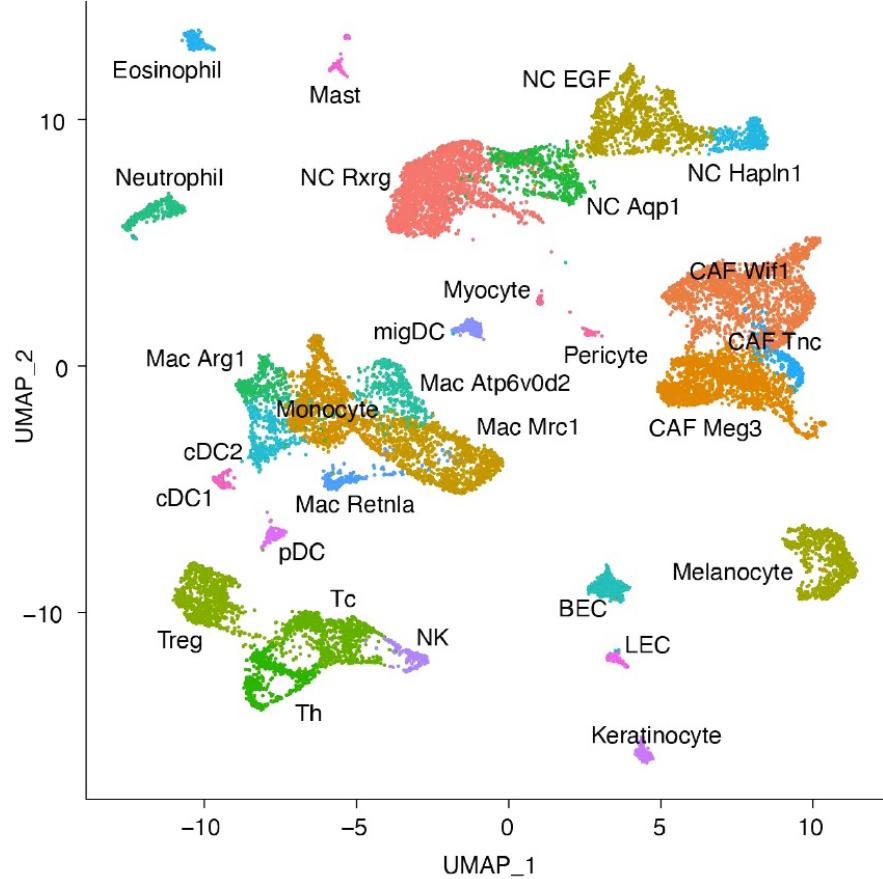


# Single Cell Platforms

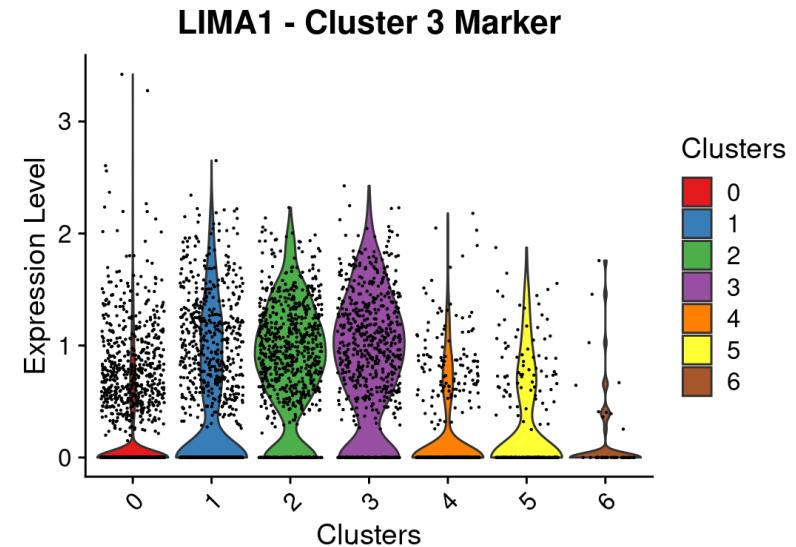
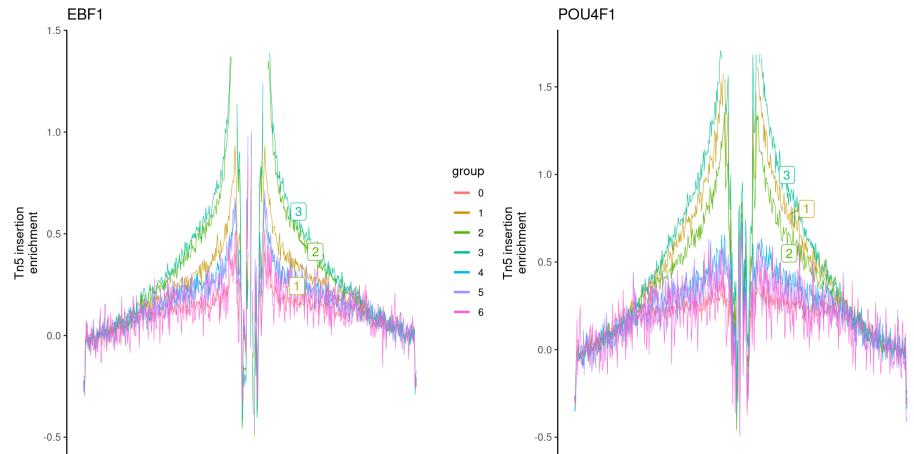
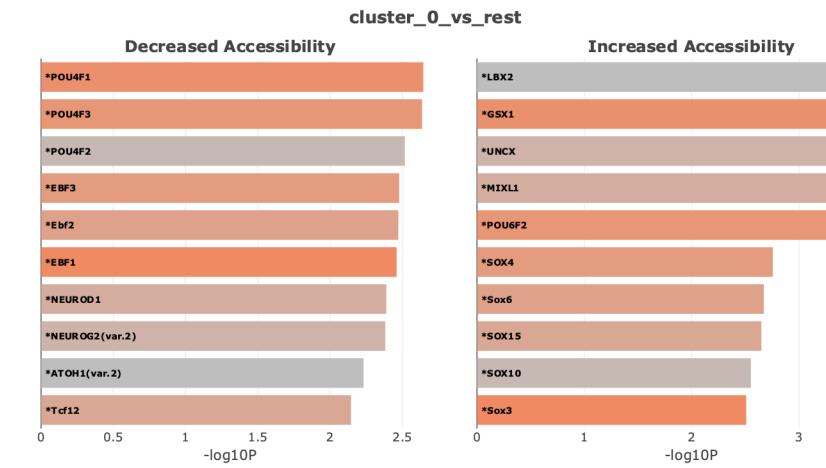
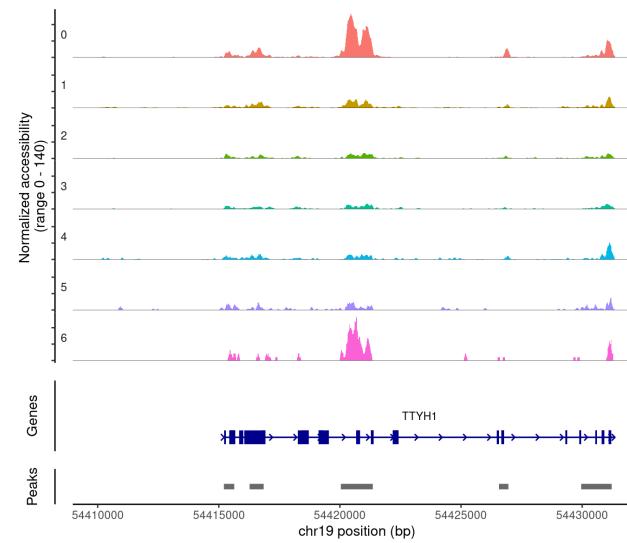
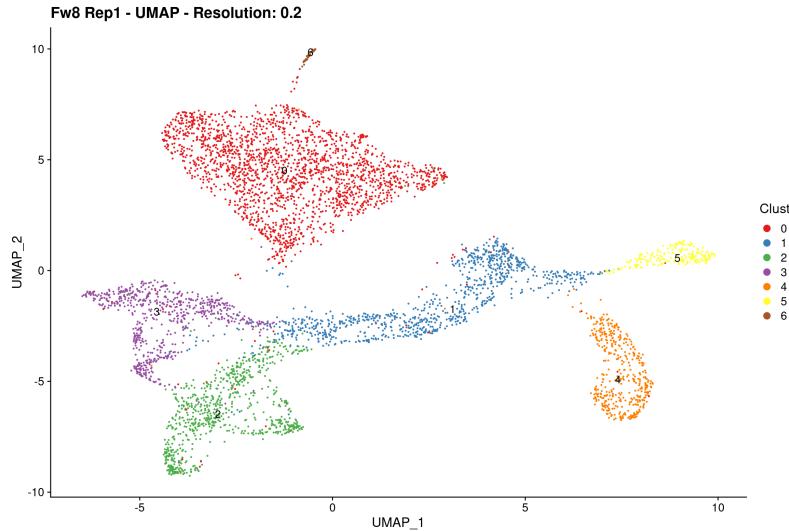


- scRNA-seq
- scATAC-seq
- Single Nucleus Multiome ATAC + RNA
- Spatial Transcriptomics

# Single Cell RNA-Seq



# Single Cell ATAC-Seq

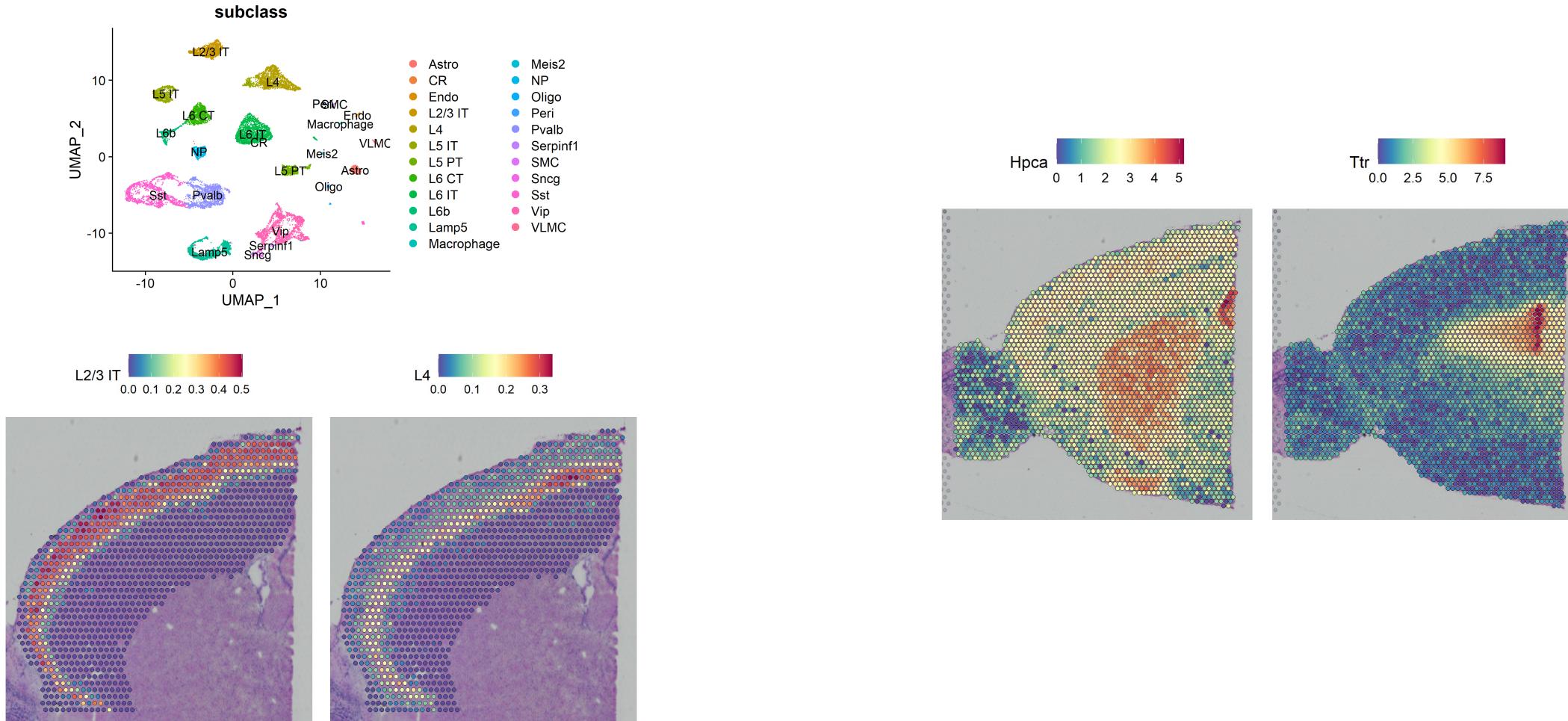


# Spatial Transcriptomics



Single-cell data from mouse cortical cells, generated with the SMART-seq2 protocol.

Cell types identified based on single-cell data, can be used to 'deconvolute' the underlying composition of cell-types in the spatial omic spots.

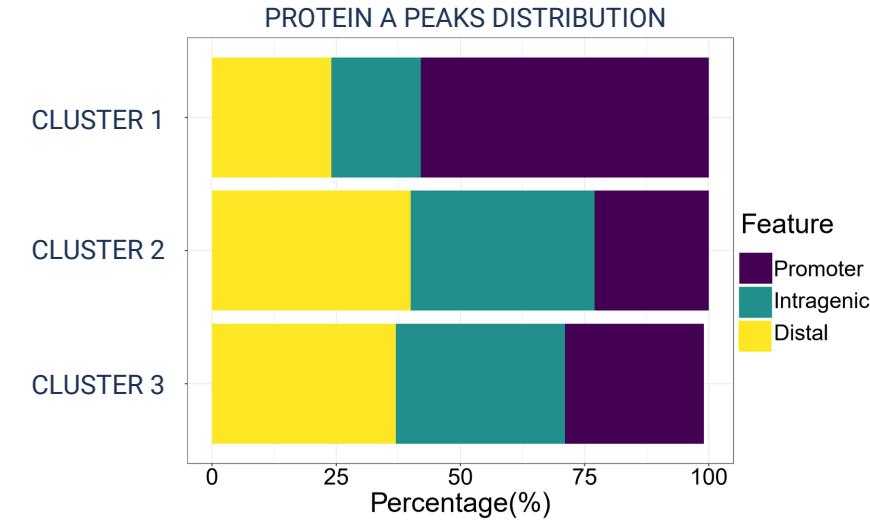
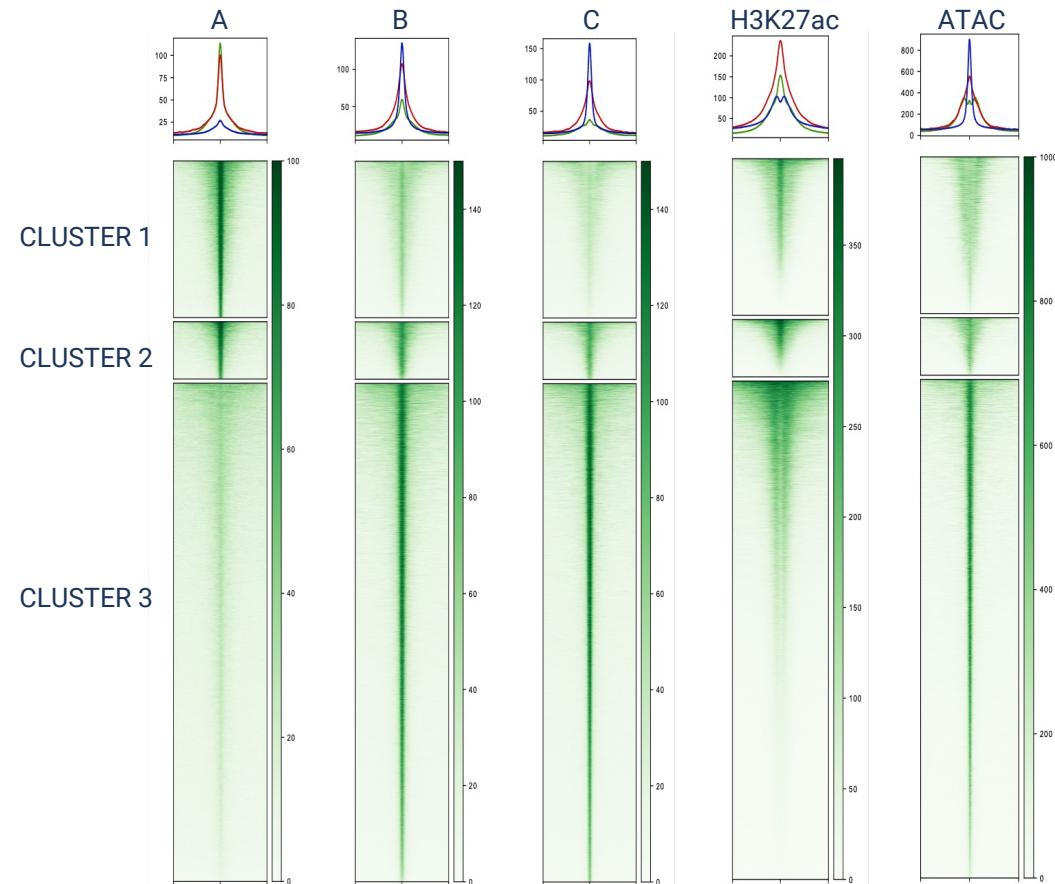




# Epigenetics



- ChIP-seq
- Bulk ATAC-Seq
- CUT&RUN and CUT&Tag
- HiC/HiChIP
- Methy Seq



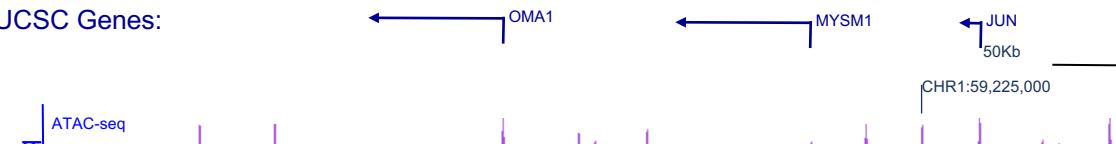
**CLUSTER 1 MOTIF ANALYSIS**

Motif	Name	P-val	Rank
ATGACTCATC	AP-1(FOSL2)	1e-18728	1(3)
TAACCCACAAA	RUNX2	1e-2008	2
CCCAATTGTT	SOX15(SOX10)	1e-1132	3(4)
CCACTAGGGGCC	CTCF(CTCF)	1e-1109	4(2)
SACTTCCCGT	ETV2(FLI1)	1e-904	5(2)

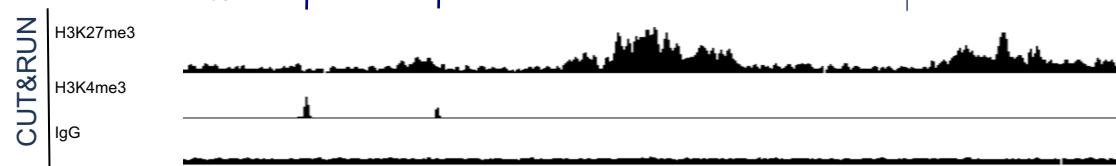
# ChIP-Seq/Cut&Run



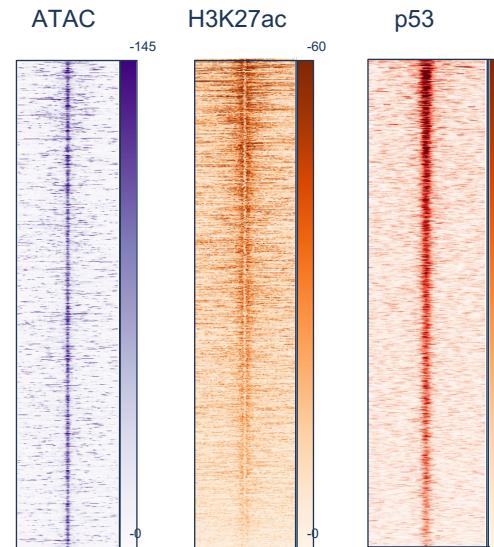
A UCSC Genes:



B

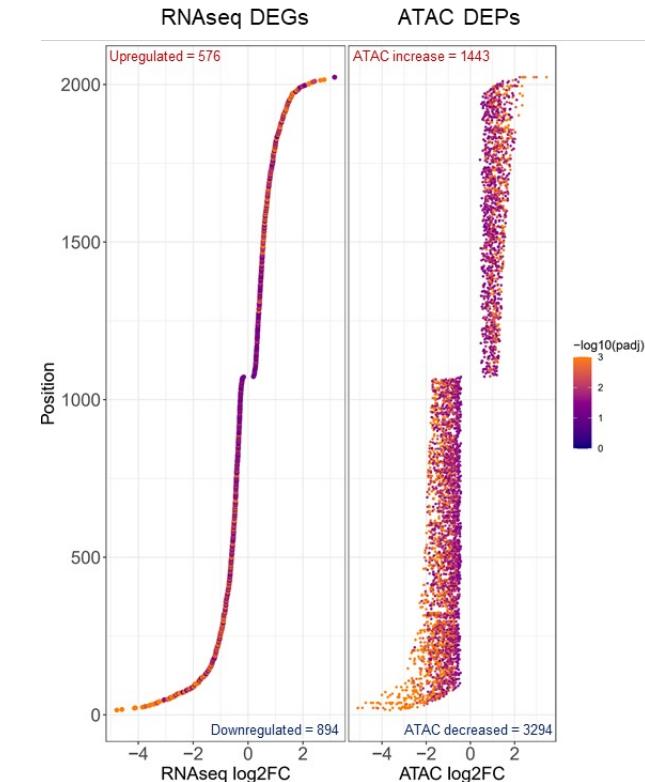


C

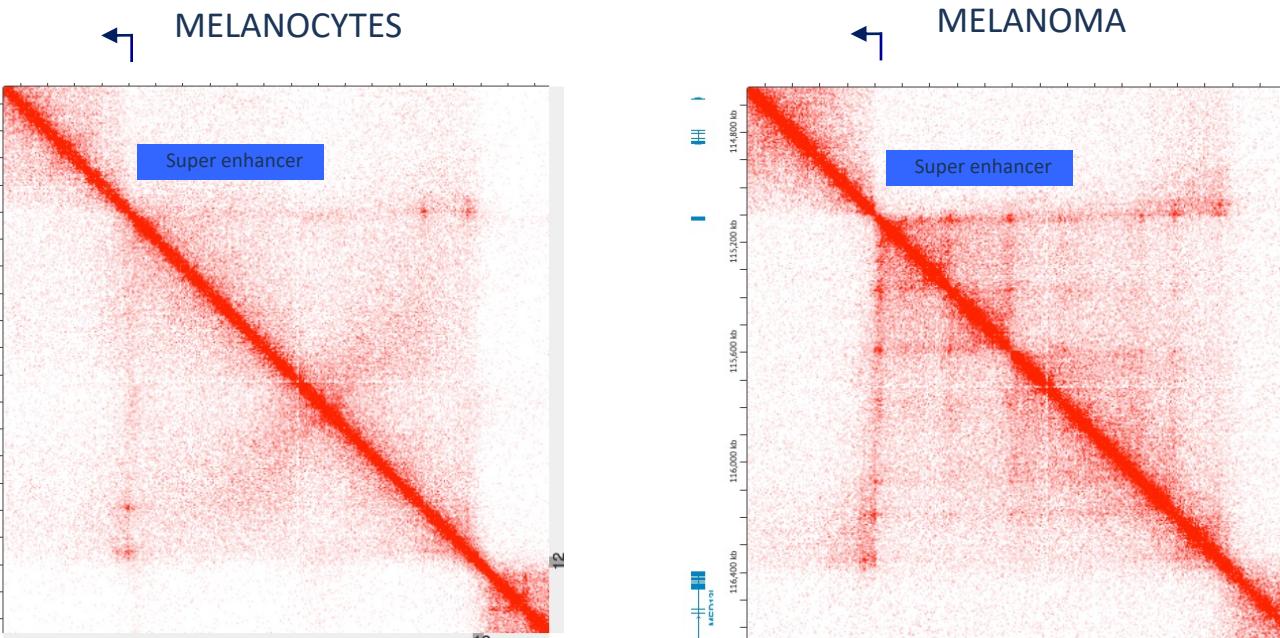
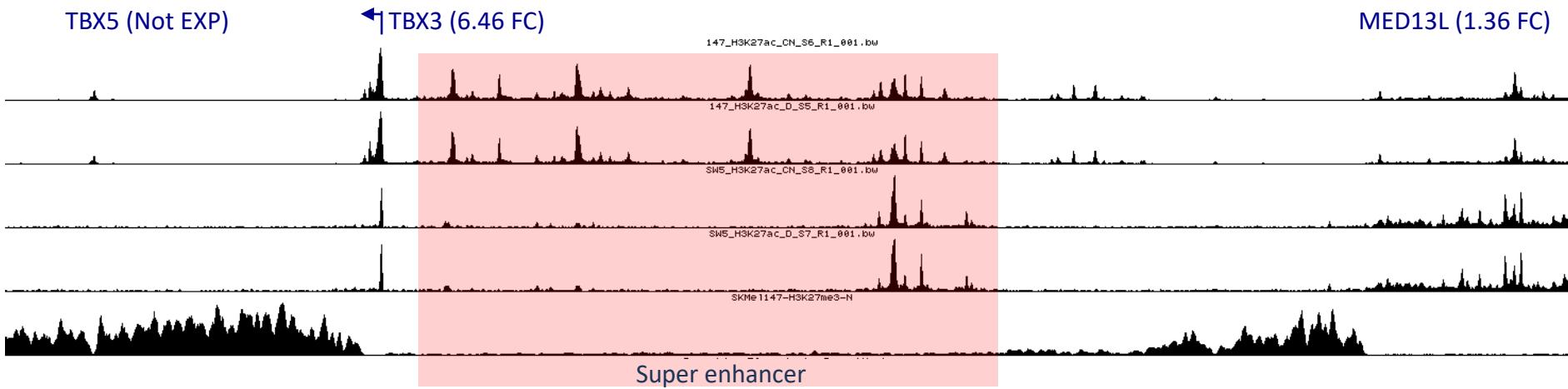


D

RNAseq-ATACseq Association



# HiC/HiCHIP



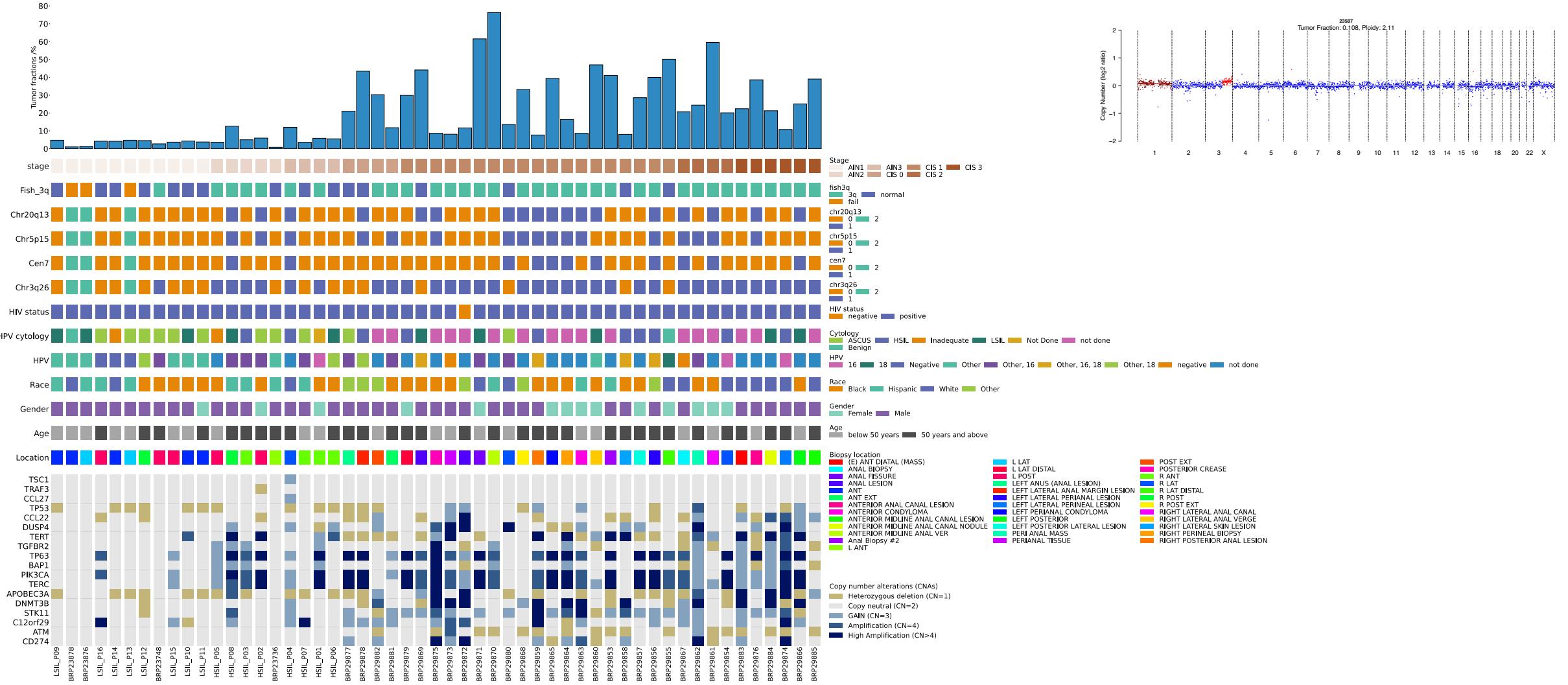
LONG RANGE CHROMATIN INTERACTION BETWEEN GENE PROMOTER AND A SUPER ENHANCER OBSERVED DIFFERENTIALLY IN MELANOMA CELLS



# Genomics

- Whole Exome Seq
- Whole Genome Seq

# Ultra Low Pass WGS for CNA discovery



With Keith Sigel

# Skin GLOW (Skin-Gene Level Omic Web Tool)



## Skin-GLOW (Skin-Gene Level Omics Web Resource)

Search for gene name / Ensembl ID

Gene Identifier:

Select the data type to visualize

- Normalized Counts (DESeq2)
- Transcripts per Million (TPM)
- Raw Counts

Select the sample groups to visualize

- Cell type
- Sequencing Batch

Select the color palette to use

- Dark2
- Set1
- Set2
- Set3

Select the plot\_type to use

- Box Plot
- Box Plot with Dots
- Violin Plot
- Violin Plot with Dots

Plot gene expression on log scale



Icahn  
School of  
Medicine at  
Mount  
Sinai

Skin Biology  
and Diseases  
Resource-based  
Center

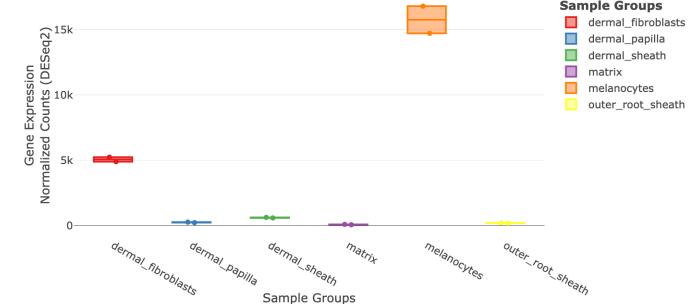
Grant number: P30 AR079200

### Dermal Sheath

Heitman et al, Science 2020 (GSE136996)

Pubmed / GEO

Mitf Gene Expression

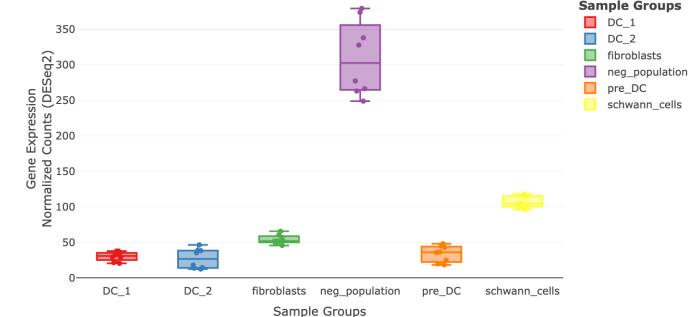


### Embryonic Skin (Hair-GEL)

Sennett et al, Dev Cell 2015 (GSE122026)

Pubmed / GEO / Hair-GEL

Mitf Gene Expression

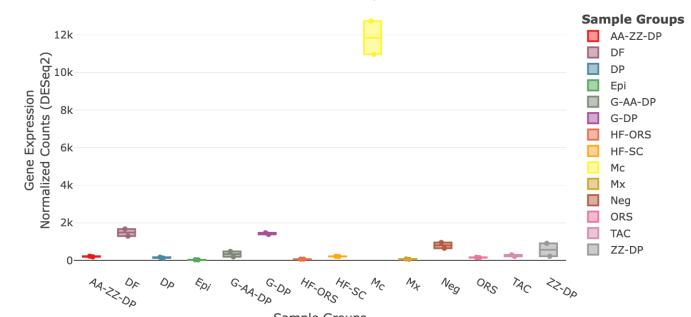


### Neonatal Mouse Skin P5

Rezza et al, Cell Rep 2016 (GSE77197)

Pubmed / GEO

Mitf Gene Expression

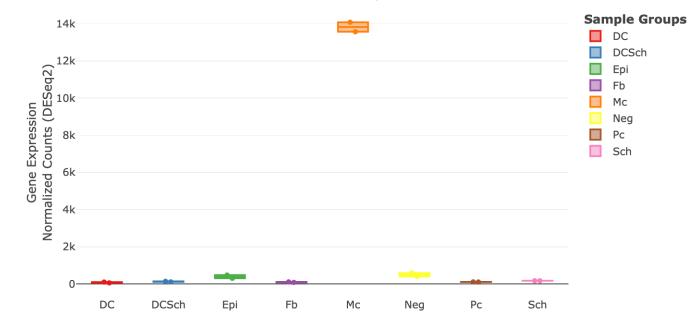


### Embryonic Mouse Skin E14.5

Rezza et al, Cell Rep 2016 (GSE70288)

Pubmed / GEO

Mitf Gene Expression



## **We offer two optional payment structures:**

1. Hourly (short term): \$95 an hour for TCI and BFSCI/CDRB/SBDRC members.
2. % effort (long term): Investigator pays % effort of one of our bioinformaticians (grants)

**Priorities:** Long term projects; First-come, first-served; and based on bioinformaticians expertise and availability.

# Submit a Project ([BiNGS.mssm.edu/submit-a-project/](http://BiNGS.mssm.edu/submit-a-project/))



## Project Details

**Project Name \***

**Name of PI \***

First

Last

**Email of PI \***

Email

Confirm Email

**Name of Leading Scientist (Contact Person) \***

First

Last

**Email of Leading Scientist (Contact Person) \***

Email

Confirm Email

**Species \***

**Analysis Type \***

- ChIP-Sequencing
- ATAC-Sequencing
- Cut&Run- and Cut&Tag-Sequencing
- scATAC-Sequencing
- RNA-Sequencing
- Alternative Splicing
- Alternative Promoter
- Transcriptomic Analysis of TCGA and Other Public Cancer Datasets
- Whole Genome Sequencing (WGS)
- Whole Exome Sequencing (WES)

Note: More than one analysis type can be selected.

**Project Description \***

0 of 1000 max words.

**Number of Samples and Sample Details \***

**Comments**

**Submit**

Open office hours:

Mondays 4-6pm

Please contact us to schedule!

Zoom ID: 409 848 3549



Icahn  
School of  
Medicine at  
Mount  
Sinai

*The Tisch  
Cancer Institute*



*Black Family  
Stem Cell Institute*

*Skin Biology  
and Diseases  
Resource-based  
Center*

*Department of Cell,  
Developmental and  
Regenerative Biology*



# LOOKING FORWARD TO WORKING TOGETHER. THANK YOU!

---

**[BiNGS.mssm.edu](http://BiNGS.mssm.edu)**