

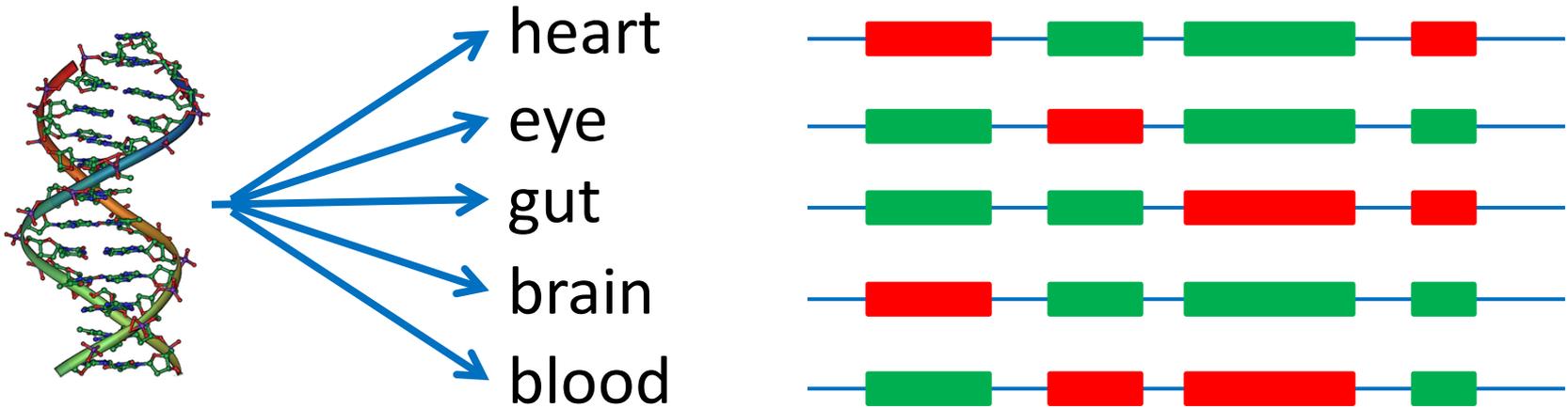
The NIH Roadmap Epigenomics Program: A Community Epigenomics Resource

Lisa Helbling Chadwick, Ph.D.
(chadwickL@niehs.nih.gov)

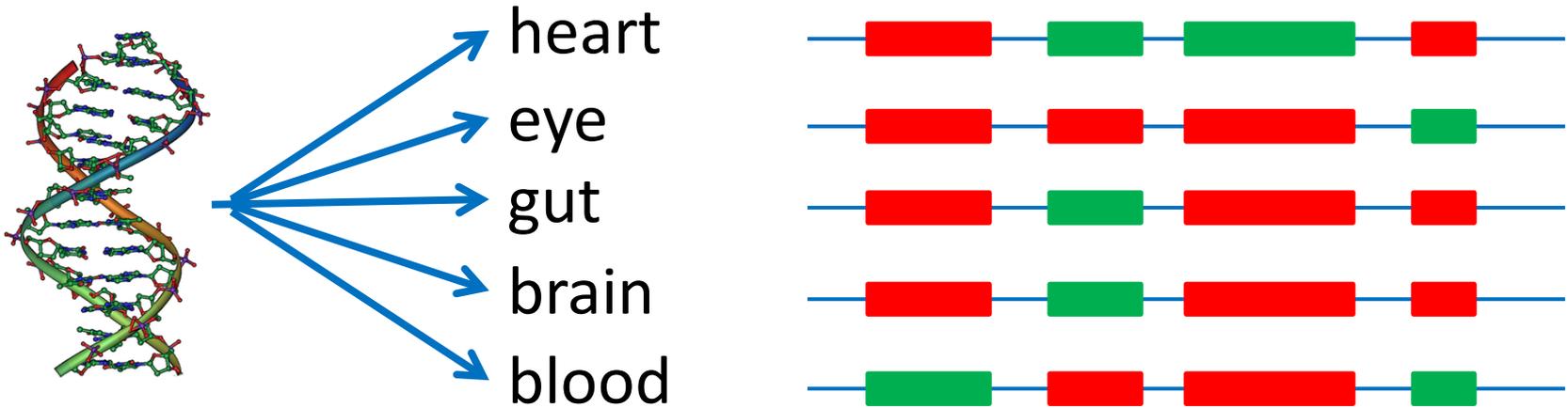
Program Director, NIH Roadmap Epigenomics Program
National Institute of Environmental Health Sciences



One genome, many cell types, many epigenetic programs



Epigenetic changes may be associated with disease



What is the NIH Roadmap Epigenomics Program?

Research to transform our understanding of how epigenetics contributes to disease
(<http://commonfund.nih.gov/epigenomics>)

- Epigenomics of Human Health and Disease
- Technology Development in Epigenetics
- Discovery of Novel Epigenetic Marks
- Computational analyses of Reference Epigenomic data
- Functional Epigenomics (epigenomic manipulation)
- Technology Development - *in vivo* epigenetic imaging

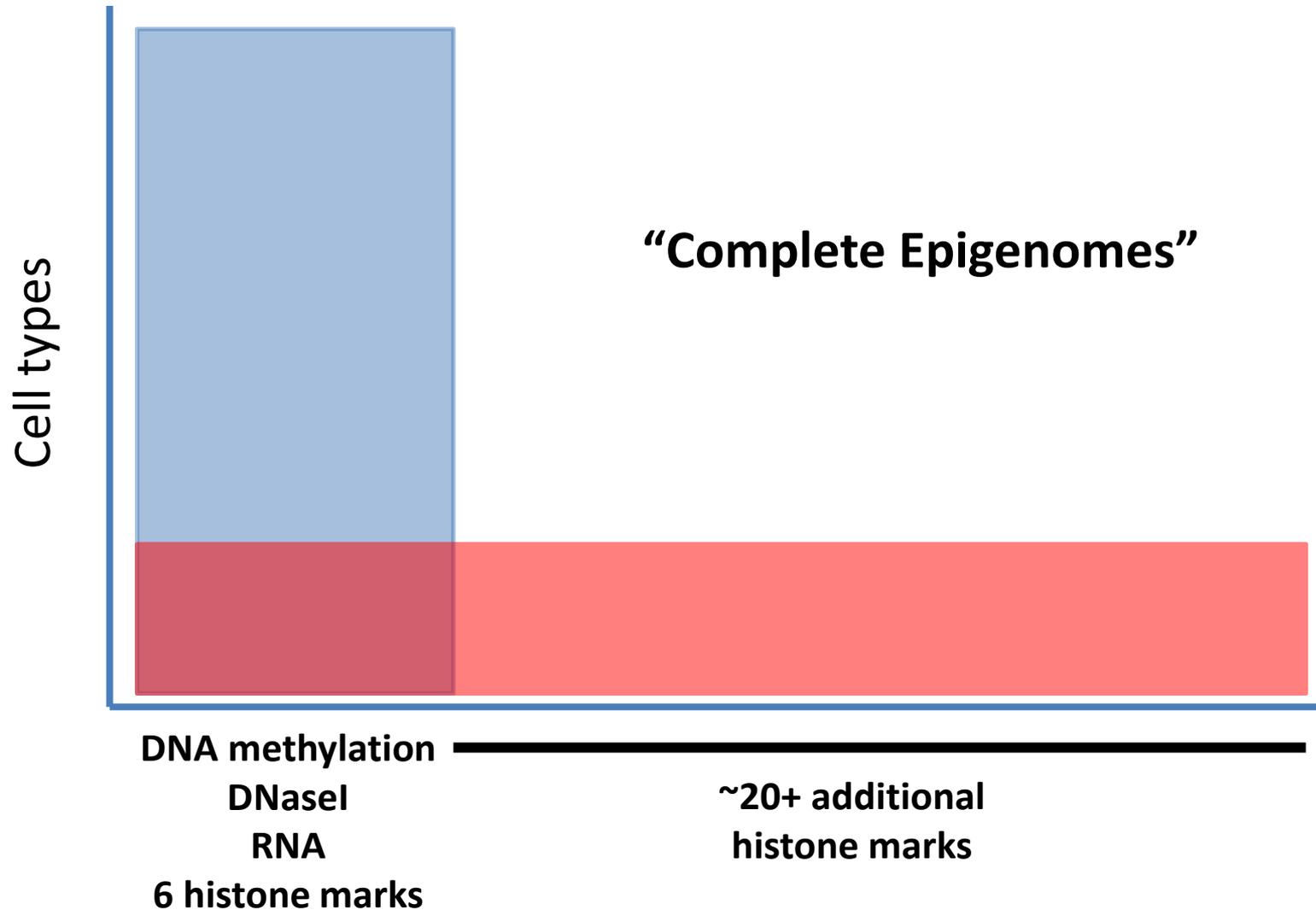


The Reference Epigenome Mapping Consortium:

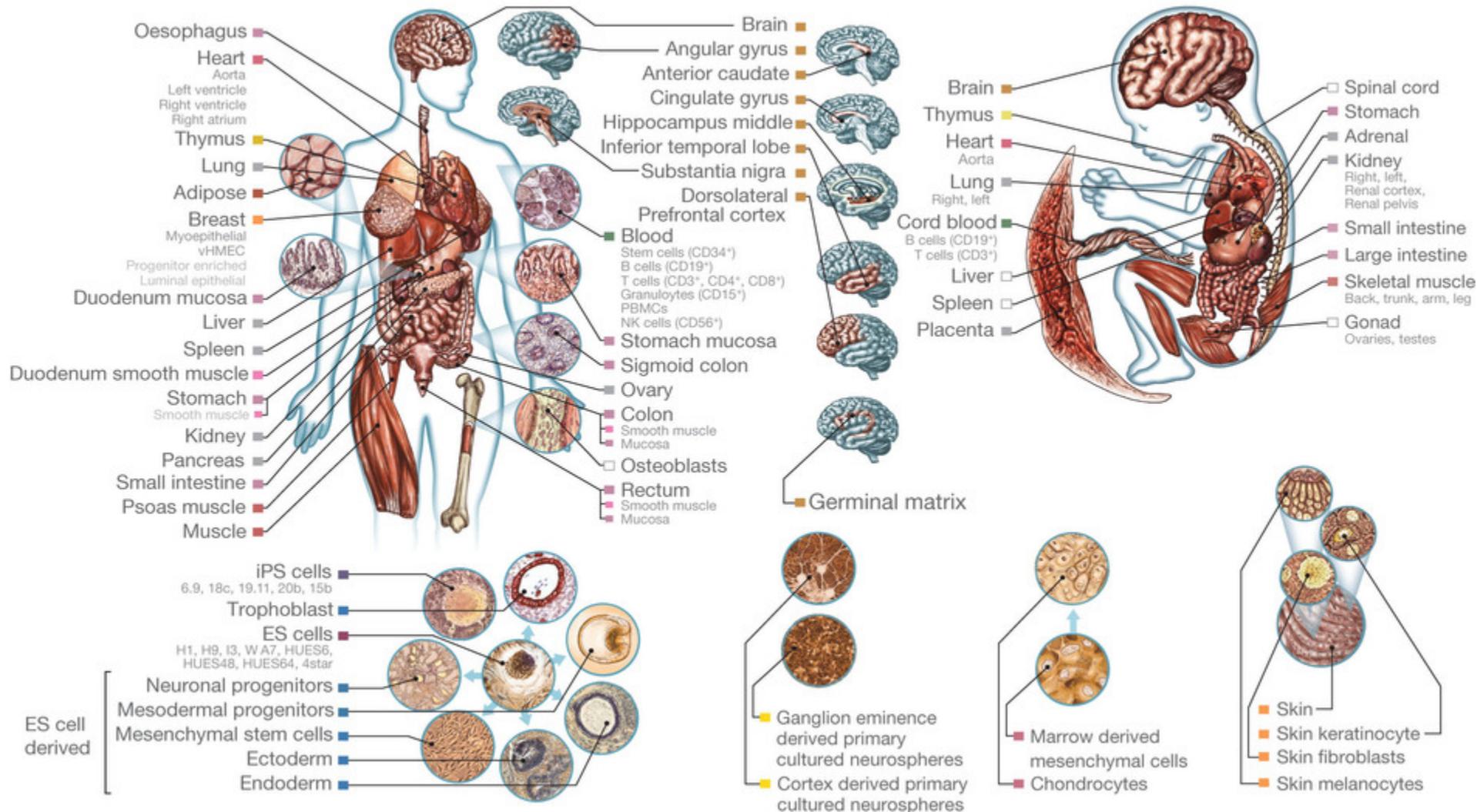
- Reference Epigenome Mapping Centers
 - Brad Bernstein & Alex Meissner* (Broad)
 - Joe Costello* (UCSF)
 - Bing Ren* (UCSD)
 - John Stamatoyannopoulos* (Washington)
- Epigenomics Data Analysis and Coordination Center
 - Aleks Milosavljevic* (Baylor)

A community resource of epigenetic data in primary human cells/tissues

Broad and deep mapping of epigenetic profiles in human cells and tissues

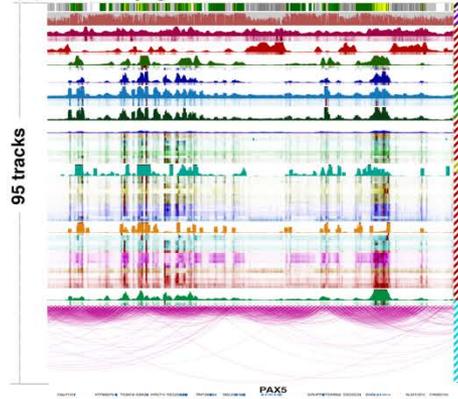


Broad and deep mapping of epigenetic profiles in over 100 human primary cells and tissues

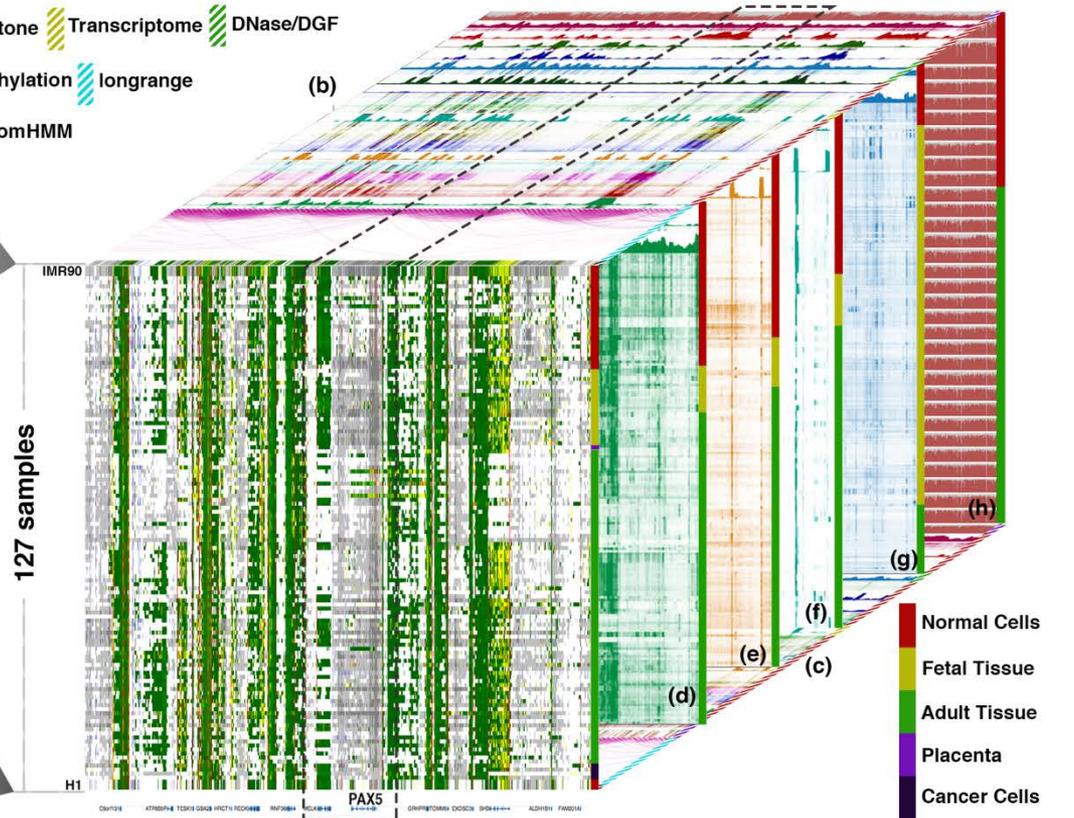


(a) complete epigenomes overview

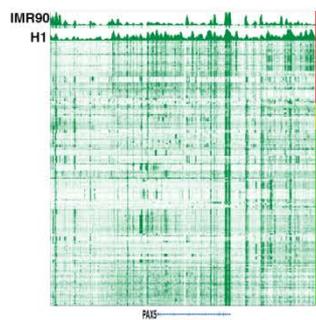
(b) IMR90 epigenome



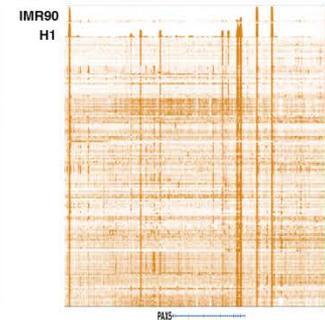
(c) H1 epigenome



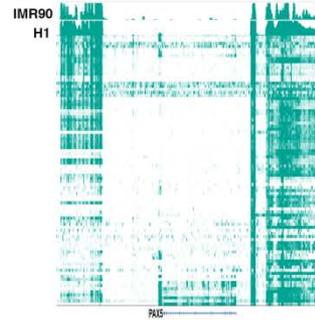
(d) H3K4me1



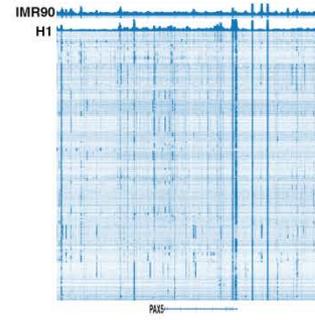
(e) H3K4me3



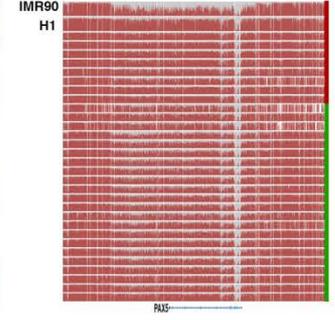
(f) RNA-seq



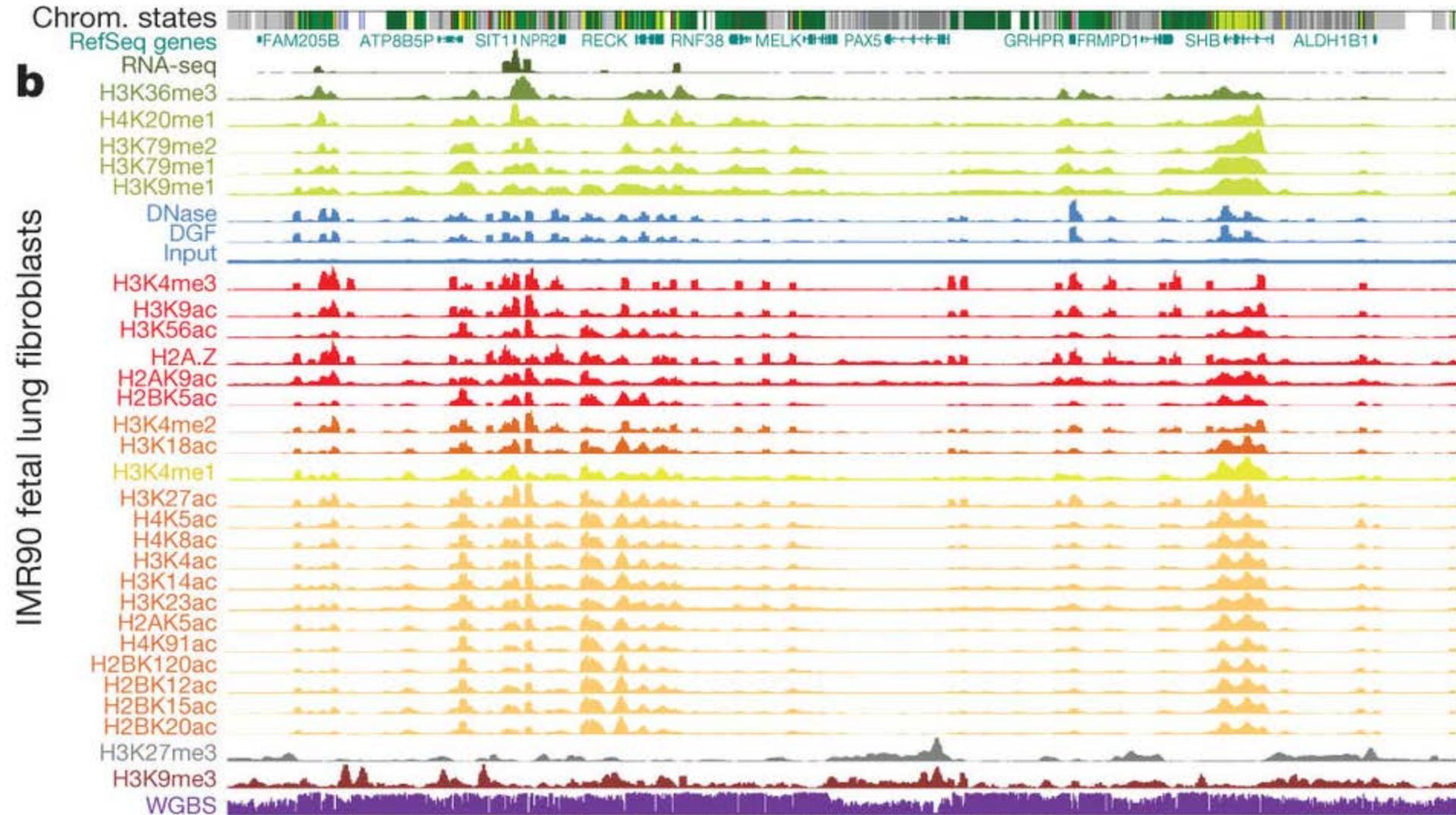
(g) DNase-seq



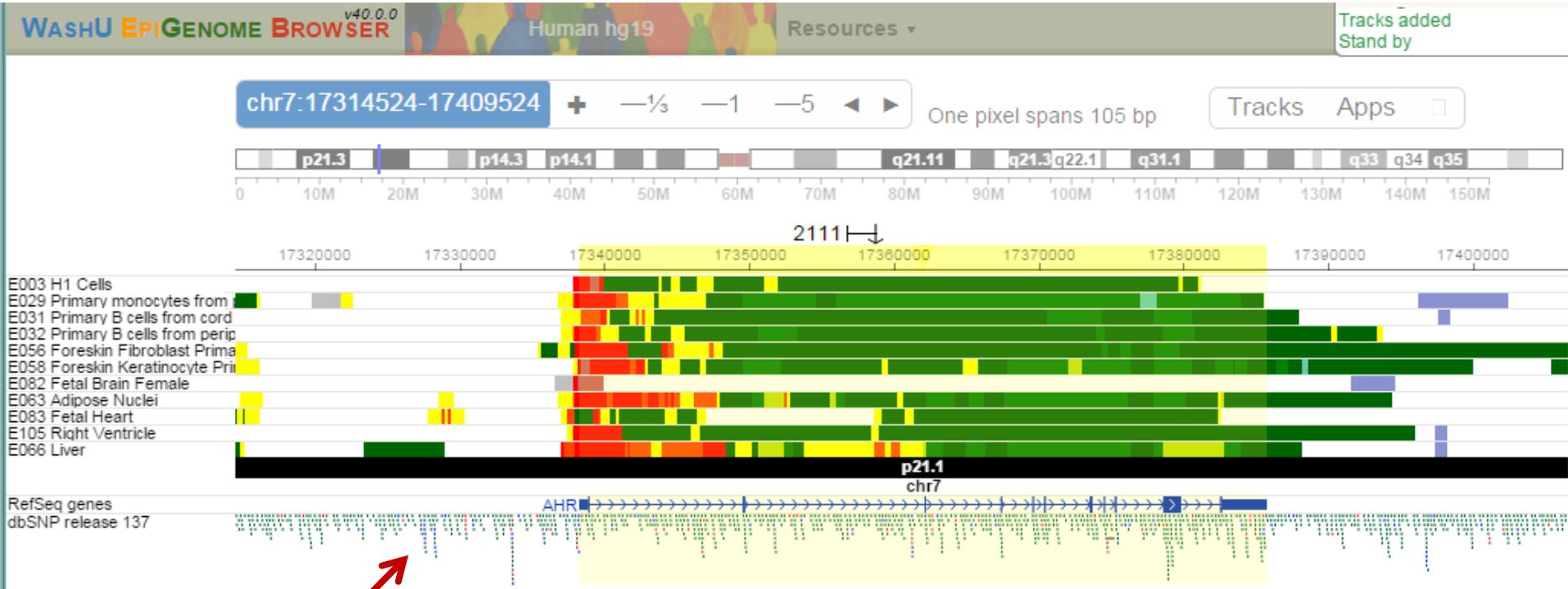
(h) WGBS



Chromatin state tracks summarize information from epigenomic maps



Chromatin state tracks summarize information from epigenomic maps



SNPs



Active TSS



Active transcription



Poised TSS



quiescent



Enhancer

nature

THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE

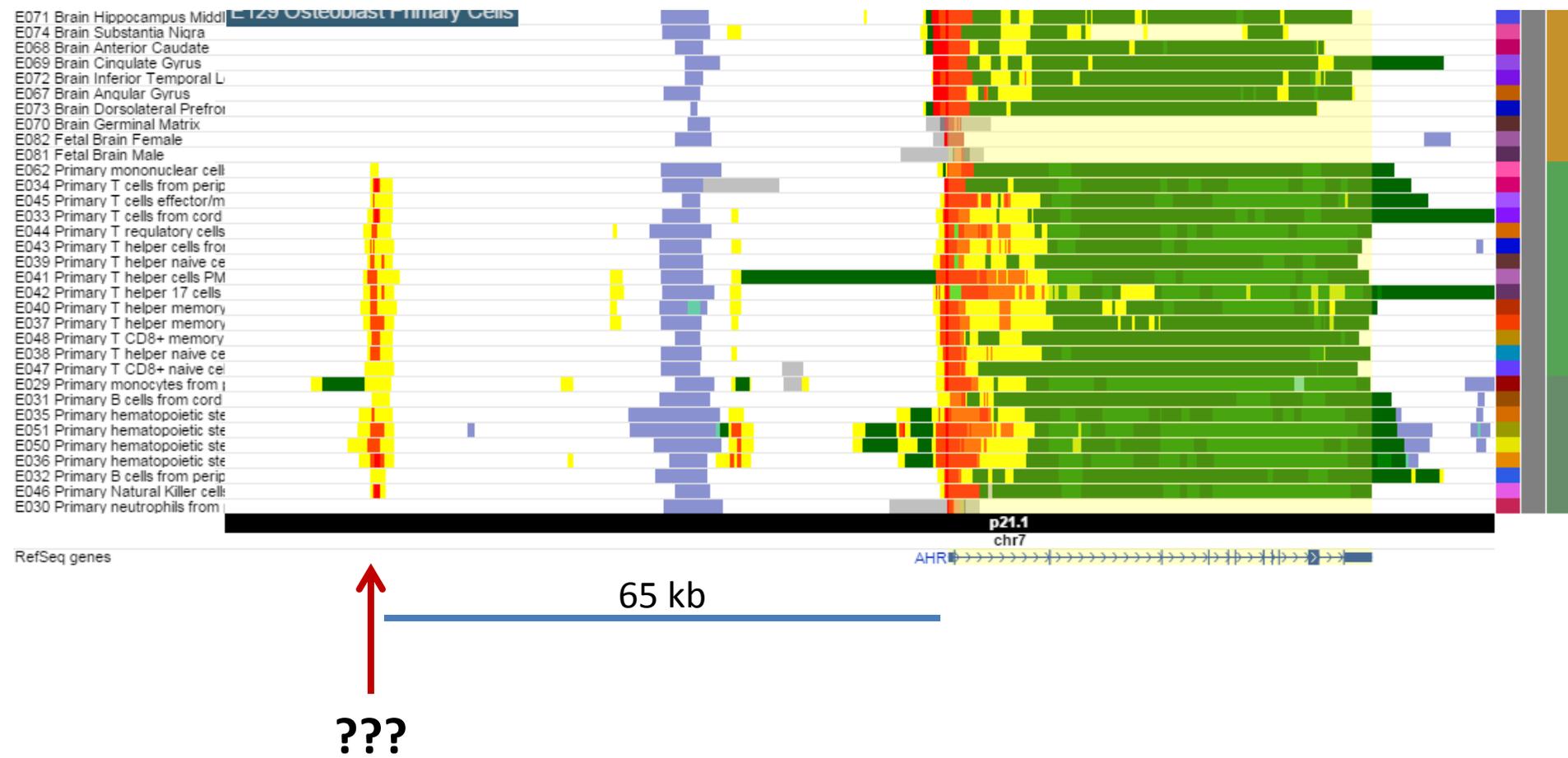


A lot of data, a lot of papers published

Find out:

- What data are there?
- What did we learn from these data about how the epigenome varies across normal tissues, and across differentiation?
- How can we use the data to identify regulatory modules?
- What are some things you do with these data?

I got a highly significant SNP in my GWAS... in the middle of nowhere!



Browse many articles by topical “threads” at [nature.com/epigenomeroadmap](https://www.nature.com/epigenomeroadmap)

The image displays a user interface for the Nature Epigenome Roadmap. It is divided into two main sections: "Thread articles" on the left and "Research papers" on the right. A central vertical column contains a series of colored squares (green, blue, purple, pink) and white squares, likely representing filters or a navigation menu.

Thread articles

- THREAD 1**
1. Annotation of the non-coding genome
[Highlight referenced papers >](#)
- THREAD 2**
2. Relationship between different epigenomic marks: DNA accessibility and methylation, histone marks, and RNA
[Highlight referenced papers >](#)
- THREAD 3**
3. Epigenomic changes during differentiation and development
[Highlight referenced papers >](#)
- THREAD 4**
4. Regulatory models: networks, motifs, modules, sequence drivers and predictive models
[Highlight referenced papers >](#)
- THREAD 5**
5. Interpreting variation: GWAS, cancer, genotype, evolution and allelic
[Highlight referenced papers >](#)

Research papers

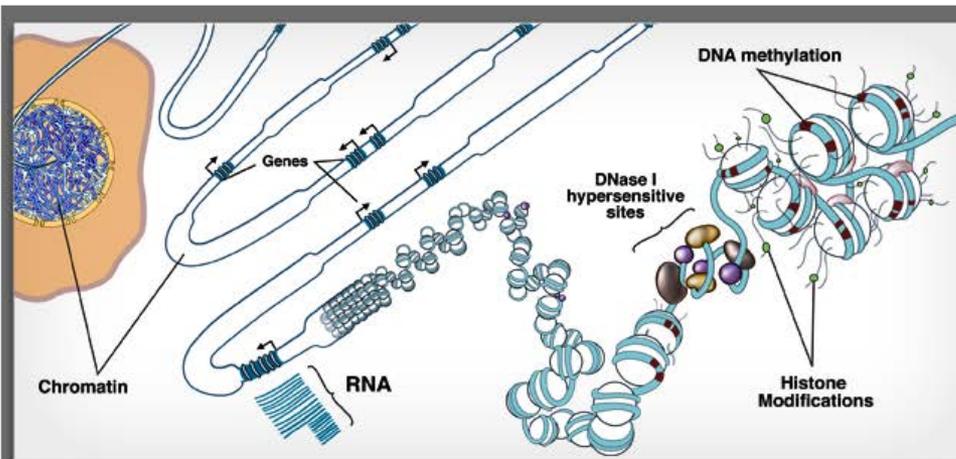
- Nature**
Conserved epigenomic signals in mice and humans reveal immune basis of Alzheimer's disease
Elizabetha Gjoneska, Andreas R. Pfenning, Hansruedi Mathys, Gerald Quon, Anshul Kundaje *et al.*
[Highlight associated threads](#)
- Nature Communications**
The meta-epigenomic structure of purified human stem cell populations is defined at cis-regulatory sequences
N. Ari Wijetunga, Fabien Delahaye, Yong M. Zhao, Aaron Golden, Jessica C. Mar *et al.*
[Highlight associated threads](#)
- Nature**
Genetic and epigenetic fine mapping of causal autoimmune disease variants
Kyle Kai-How Farh, Alexander Marson, Jiang Zhu, Markus Klei, William J. Housley *et al.*
[Highlight associated threads](#)
- Nature Communications**
Epigenomic footprints across 111 reference epigenomes reveal tissue-specific epigenetic regulation of lincRNAs
Viren Amin, R. Alan Harris, Vitor Onuchic, Andrew R. Jackson, Tim Charneck *et al.*
[Highlight associated threads](#)

I would like to look at this data. Where do I go?



Search: GO

HOME PARTICIPANTS BROWSE DATA PROTOCOLS COMPLETE EPIGENOMES TOOLS PUBLICATIONS



NIH Roadmap Epigenomics Mapping Consortium

The NIH Roadmap Epigenomics Mapping Consortium was launched with the goal of producing a public resource of human epigenomic data to catalyze basic biology and disease-oriented research. The Consortium leverages experimental pipelines built around next-generation sequencing technologies to map DNA methylation, histone modifications, chromatin accessibility and small RNA transcripts in stem cells and primary ex vivo tissues selected to represent the normal counterparts of tissues and organ systems frequently involved in human disease. The Consortium expects to deliver a collection of normal epigenomes that will provide a framework or reference for comparison and integration within a broad array of future studies. The

INTEGRATIVE ANALYSIS of 111 REFERENCE HUMAN EPIGENOMES

- Brain
 - Angular gyrus
 - Anterior cingulate
 - Cingulate gyrus
 - Hippocampus middle
 - Inferior temporal lobe
 - Substantia Nigra
 - Dorsolateral Prefrontal Cortex
- Heart
 - Aorta
- Lung
 - High cell
- Cord blood
 - CD34+ CD38-
 - CD34+ CD38+
- Liver
- Spleen
- Placenta
- Blood
 - CD34+ CD38-
 - CD34+ CD38+
 - CD34+ CD38+ CD45+
 - CD34+ CD38+ CD45+
 - CD34+ CD38+ CD45+
 - CD34+ CD38+ CD45+
- Stomach mucosa
- Sigmoid colon
- Ovary
- Colon

VIEW DATA

VIEW/DOWNLOAD QUICK LINKS

Genome Browsers

- <http://genomebrowser.wustl.edu/>
- <http://epigenomegateway.wustl.edu/>

Data Repositories

- [NCBI Epigenomics Gateway](#)
- [Epigenome Atlas](#)

Lots of choices!

Just give me the data. That's it.



Gene Expression Omnibus

[GEO Publications](#) | [FAQ](#) | [MIAME](#) | [Email GEO](#)

[Login](#)

[NCBI](#) » [GEO](#) » [NIH Roadmap Epigenomics](#)

NIH Roadmap Epigenomics Project Data Listings

This page summarizes Roadmap Epigenomics project data that are available through the GEO repository. Additional data will be added as it becomes available.

For data usage terms and conditions, please refer to
[Epigenomics Data Access Policies](#)

More information and resources are available:

- [Roadmap Epigenomics Homepage](#)
- [Epigenome Browser](#)
- [Epigenome Atlas](#)
- [NCBI Epigenomics Gateway](#)

[FTP download](#)

[How to use this page](#)

Samples

Studies

Matrix

Search

3135 results

[View tracks](#)

[Export](#)

Page of 63 [>](#) [>>](#) Page size

GEO Accession ^	Sample	Experiment	Track <input type="checkbox"/>	Center	Download	Embargo end date
GSM409307	H1 cell line	H3K4me1	<input type="checkbox"/>	UCSD	sra bam,bed,wig	2010-05-03
GSM409308	H1 cell line	H3K4me3	<input type="checkbox"/>	UCSD	sra bam,bed,wig	2010-05-03
GSM409312	H1 cell line	H3K36me3	<input type="checkbox"/>	UCSD	sra bam,bed,wig	2010-05-03
GSM410807	H1 cell line	H3K9ac	<input type="checkbox"/>	UCSF-UBC	sra bam,bed,wig	2010-05-03
GSM410808	H1 cell line	H3K4me3	<input type="checkbox"/>	UCSF-UBC	sra bam,bed,wig	2010-05-03
GSM428286	H1 cell line	MRE-Seq	<input type="checkbox"/>	UCSF-UBC	sra bam,bed,wig	2010-10-07
GSM428289	H1 cell line	ChIP-Seq input	<input type="checkbox"/>	UCSF-UBC	sra bam,bed,wig	2010-05-03
GSM428291	H1 cell line	H3K9me3	<input type="checkbox"/>	UCSF-UBC	sra bam,bed,wig	2010-05-03
GSM428295	H1 cell line	H3K27me3	<input type="checkbox"/>	UCSF-UBC	sra bam,bed,wig	2010-05-03
GSM428296	H1 cell line	H3K36me3	<input type="checkbox"/>	UCSF-UBC	sra bam,bed,wig	2010-05-03

I just want to look at the data! (and I have the patience to deal with the UCSC browser)

Home Genomes Genome Browser Tools Mirrors Downloads My Data Help About Us

Human (*Homo sapiens*) Genome Browser Gateway

The UCSC Genome Browser was created by the [Genome Bioinformatics Group of UC Santa Cruz](#).
Software Copyright (c) The Regents of the University of California. All rights reserved.

group genome assembly position search term

Mammal Human Feb. 2009 (GRCh37/hg19) chr7:17,290,776-17,433,275 enter position, gene symbol or search terms submit

[Click here to reset](#) the browser user interface settings to their defaults.

track search add custom tracks track hubs configure tracks and display

Public Hubs My Hubs

Enter search terms to find in public track hub description pages:

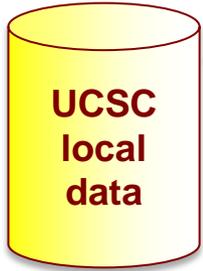
Search Public Hubs

Displayed list **restricted by search terms:** roadmap Show All Hubs

Clicking Connect redirects to the gateway page of the selected hub's default assembly.

Display	Hub Name	Description	Assemblies
<input type="button" value="Connect"/>	Roadmap Epigenomics Data Complete Collection at Wash U VizHub	Roadmap Epigenomics Human Epigenome Atlas Data Complete Collection, VizHub at Washington University in St. Louis	hg19
<input type="button" value="Connect"/>	ENCODE Analysis Hub	ENCODE Integrative Analysis Data Hub	hg19
<input type="button" value="Connect"/>	CEMT (CEEHRC)	Epigenomic Data tracks from BCGSC, Vancouver	hg19
<input type="button" value="Connect"/>	Roadmap Epigenomics Integrative Analysis Hub	Roadmap Epigenomics Integrative Analysis Hub at Washington University in St. Louis	hg19

Remote hub



**UCSC
local
data**

DNase Clusters



**Roadmap
Remote
Hub data**

30
3K27me3 Solexa-8039

30
BI H1 H3K4me3 Solexa

0

ENCODE

BS

MeDIP

MRE

K27me3

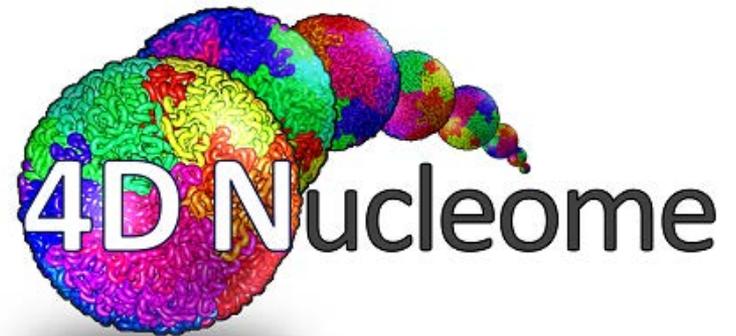
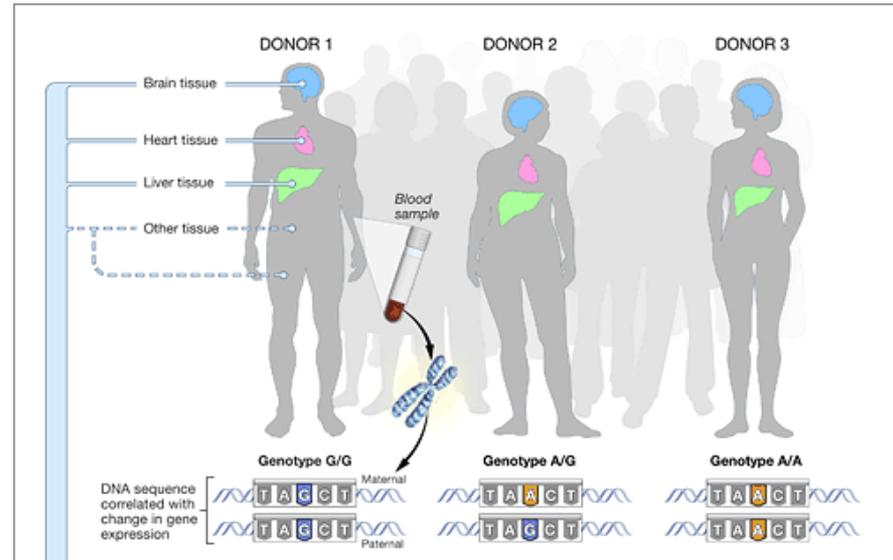
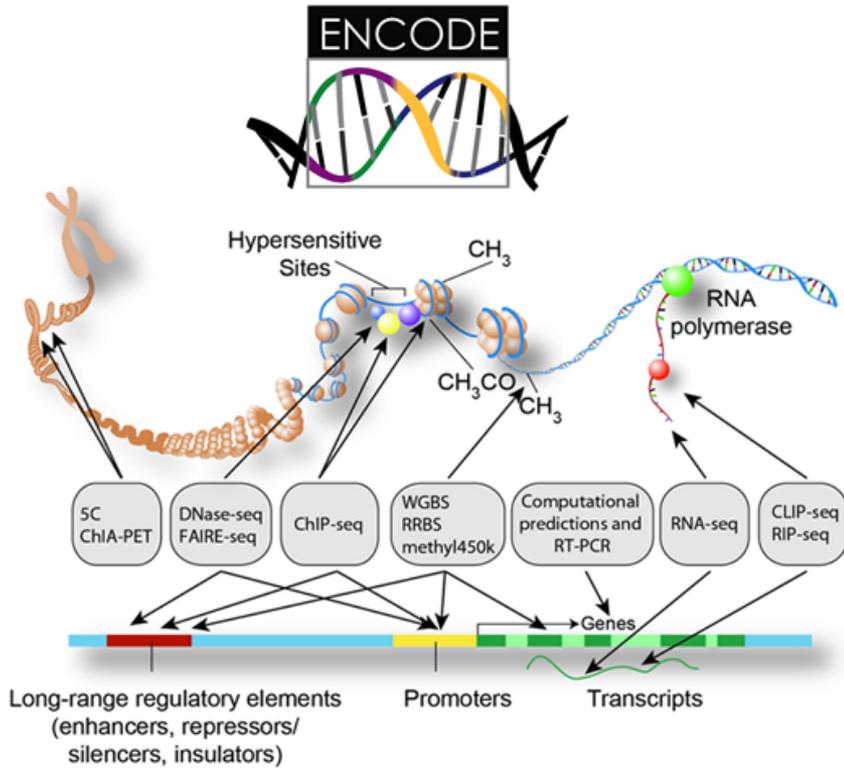
K4me3

Input

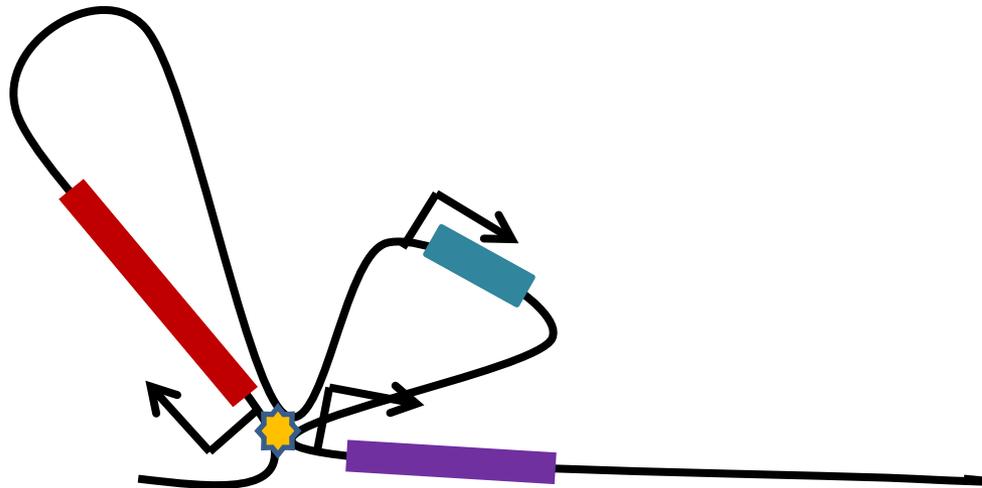
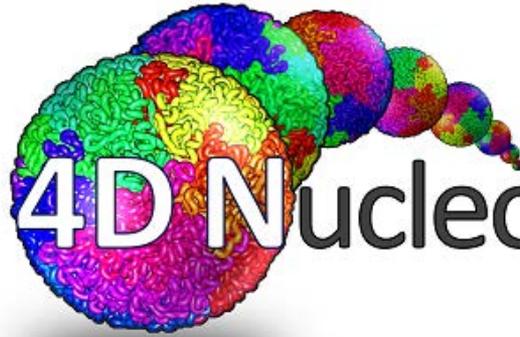
Another option: Next-gen epigenome browser @ WashU

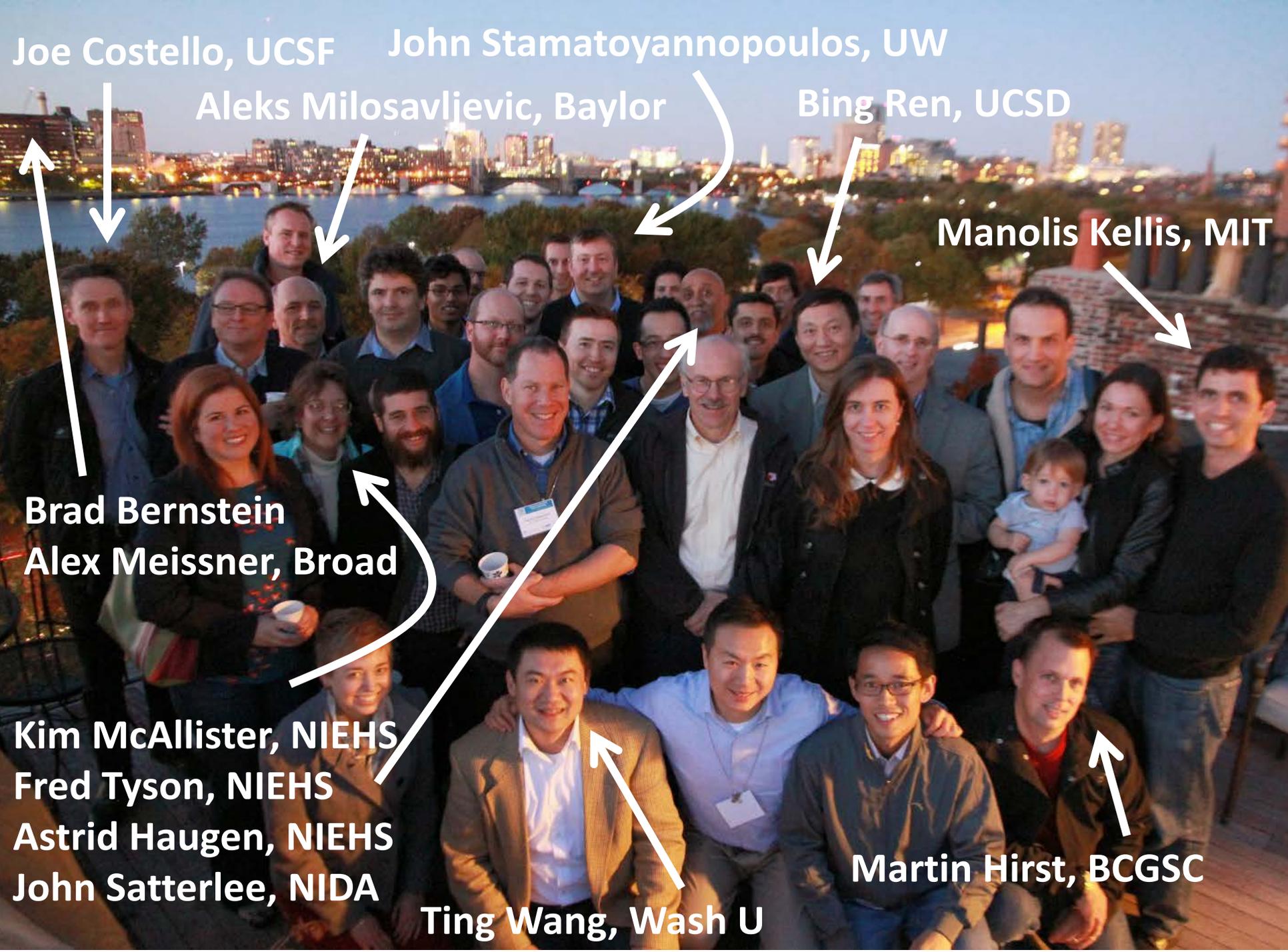


Lots of other large, public data sets



What is 4D Nucleome





Joe Costello, UCSF

John Stamatoyannopoulos, UW

Aleks Milosavljevic, Baylor

Bing Ren, UCSD

Manolis Kellis, MIT

Brad Bernstein

Alex Meissner, Broad

Kim McAllister, NIEHS

Fred Tyson, NIEHS

Astrid Haugen, NIEHS

John Satterlee, NIDA

Ting Wang, Wash U

Martin Hirst, BCGSC