



Школа «Науки о
данных»

Трек «Биоинформатика» 25-26
октября

UCSC Genome Browser

UCSC Genome Browser Home

http://genome.ucsc.edu/ 

UCSC Genome Browser Home +

UCSC Genome Bioinformatics

Genomes (circled in red) [Blat](#) - [Tables](#) - [Gene Sorter](#) - [PCR](#) - [VisiGene](#) - [Proteome](#) - [Session](#) - [FAQ](#) - [Help](#)

Genome Browser

ENCODE

Neandertal

Blat

Table Browser

Gene Sorter

In Silico PCR

Genome Graphs

Galaxy

VisiGene

Proteome Browser

Utilities

Downloads

Release Log

About the UCSC Genome Bioinformatics Site

Welcome to the UCSC Genome Browser website. This site contains the reference sequence and working draft assemblies for a large collection of genomes. It also provides portals to the [ENCODE](#) and [Neandertal](#) projects.

We encourage you to explore these sequences with our tools. The [Genome Browser](#) zooms and scrolls over chromosomes, showing the work of annotators worldwide. The [Gene Sorter](#) shows expression, homology and other information on groups of genes that can be related in many ways. [Blat](#) quickly maps your sequence to the genome. The [Table Browser](#) provides convenient access to the underlying database. [VisiGene](#) lets you browse through a large collection of *in situ* mouse and frog images to examine expression patterns. [Genome Graphs](#) allows you to upload and display genome-wide data sets.

The UCSC Genome Browser is developed and maintained by the Genome Bioinformatics Group, a cross-departmental team within the Center for Biomolecular Science and Engineering ([CBSE](#)) at the University of California Santa Cruz ([UCSC](#)). If you have feedback or questions concerning the tools or data on this website, feel free to contact us on our [public mailing list](#).

News  [News Archives ▶](#)

To receive announcements of new genome assembly releases, new software features, updates and training seminars by email, subscribe to the [genome-announce](#) mailing list.

10 February 2012 - dbSNP 135 Available for hg19

We are pleased to announce the release of four tracks derived from dbSNP build 135, available on the human assembly (GRCh37/hg19). dbSNP build 135 is available at NCBI. The new tracks contain additional annotation data not included in previous dbSNP tracks, with corresponding coloring and filtering options in the Genome Browser.

As for dbSNP build 132, there are four tracks in this release. One is a track containing all mappings of reference SNPs to the human assembly, labeled "All SNPs (135)". The other three tracks are subsets of this track and show interesting and easily defined subsets of dbSNP:

Done

UCSC Genome Browser

Human (Homo sapiens) Genome Browser Gateway

http://genome.ucsc.edu/cgi-bin/hgGateway

Human (Homo sapiens) Genome B... +

Home Genomes Blat Tables Gene Sorter PCR Session FAQ Help

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.

clade	genome	assembly	position or search term	gene
Mammal	Human	Feb. 2009 (GRCh37/hg19)	chr21:33,031,597-33,041,570	submit
Mammal	Human	Feb. 2009 (GRCh37/hg19)		
Vertebrate	Chimp	Mar. 2006 (NCBI36/hg18)		
Deuterostome	Gorilla	May 2004 (NCBI35/hg17)		
Insect	Orangutan	July 2003 (NCBI34/hg16)		
Nematode	Gibbon			
Other	Rhesus			
	Marmoset			
	Mouse			
	Rat			
	Guinea pig			
	Rabbit			
	Pig			
	Sheep			
	Cow			
	Horse			
	Cat			
	Dog			
	Panda			
	Microbat			
	Elephant			

Interface settings to their defaults.

[configure tracks and display](#) [clear position](#)

About the Human Feb. 2009 (GRCh37) Assembly

The February 2009 human reference genome was produced by the [Genome Reference Consortium](#). For more information about the assembly, see [GRCh37](#) in the NCBI Assembly database.

Sample position queries

A genome position can be specified by a chromosome number, a band name, a contig ID, an EST or STS marker, a chromosomal coordinate, a genomic clone ID, an mRNA ID, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the [User's Guide](#) for more information.

Request:	Genome Browser Response:
chr7	Displays all of chromosome 7
chrUn_g1000212	Displays all of the unplaced contig g1000212
20p13	Displays region for band p13 on chr 20
chr3:1-1000000	Displays first million bases of chr 3, counting from p-arm telomere
chr3:1000000+2000	Displays a region of chr3 that spans 2000 bases, starting with position 1000000

ATP [submit](#)

ATP1A1

ATP1A1OS

ATP1A2

ATP1A3

ATP1A4

ATP1B1

ATP1B2

ATP1B3

ATP1B4

ATP2A1

ATP2A2

ATP2A3

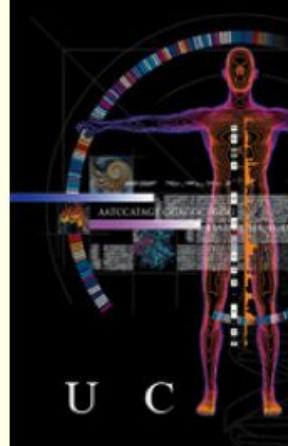
ATP2B1

ATP2B2

ATP2B3

ATP2B4

ATP2C1



U C

Human Gene ATP1A1 (uc010owv.1) Description and Page Index

Description: Homo sapiens ATPase, Na⁺/K⁺ transporting, alpha 1 polypeptide (ATP1A1), transcript variant 4, mRNA.

RefSeq Summary (NM_001160234): The protein encoded by this gene belongs to the family of P-type cation transport ATPases, and to the subfamily of Na⁺/K⁺-ATPases. Na⁺/K⁺-ATPase is an integral membrane protein responsible for establishing and maintaining the electrochemical gradients of Na and K ions across the plasma membrane. These gradients are essential for osmoregulation, for sodium-coupled transport of a variety of organic and inorganic molecules, and for electrical excitability of nerve and muscle. This enzyme is composed of two subunits, a large catalytic subunit (alpha) and a smaller glycoprotein subunit (beta). The catalytic subunit of Na⁺/K⁺-ATPase is encoded by multiple genes. This gene encodes an alpha 1 subunit. Multiple transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, May 2009].

Strand: + Genomic Size: 31594 Exon Count: 22 Coding Exon Count: 21

UCSC Genome Browser

Version v262

Viewing Tracks & Clade = mam



Human chr1:116,915,795-116,94...



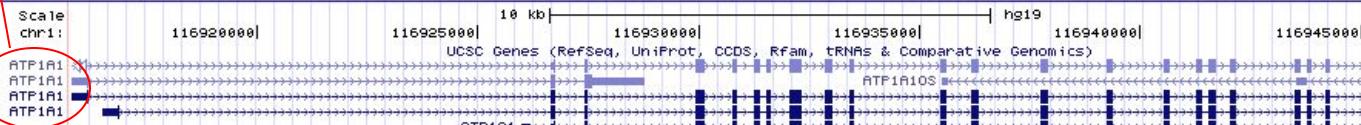
Home Genomes Blat Tables Gene Sorter PCR DNA Convert PDF/PS Session Ensembl NCBI Help

UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

move <<< << < > >> ZOOM in 1.5x 3x 10x base ZOOM out 1.5x 3x 10x

position/search chr1:116,915,795-116,947,396 gene jump clear size 31,602 bp. configure

chr1 (p13.1) 33 ip31.1 1q12 32.1 1q41 q43[44]



RefSeq Genes Human mRNAs Spliced ESTs 100 Layered H3K27Ac 0

H3K27Ac Mark (Often Found Near Active Regulatory Elements) on 7 cell lines from ENCODE

DNase Clusters Txn Factor ChIP 4 -

Digital DNaseI Hypersensitivity Clusters from ENCODE

Transcription Factor ChIP-seq from ENCODE

Placental Mammal Basewise Conservation by PhyloP

Mammal Cons 8 -4 -

Multiz Alignments of 46 Vertebrates

Rhesus Mouse Dog Elephant Opossum Chicken X_tropicalis Zebrafish

Common SNPs(135) Simple Nucleotide Polymorphisms (dbSNP 135) Found in >= 1% of Samples

Common SNPs(132) Simple Nucleotide Polymorphisms (dbSNP 132) Found in >= 1% of Samples

RepeatMasker Repeating Elements by RepeatMasker

move start

Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options.

move end

Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.

< 2.0 >

< 2.0 >

Done

UCSC Genome Browser

Regulation

ENCODE Regulation... <input type="button" value="show"/>	18 CD34 DnaseI <input type="button" value="hide"/>	CpG Islands <input type="button" value="hide"/>	ENC DNA Methyl... <input type="button" value="hide"/>	ENC DNase/FAIRE... <input type="button" value="hide"/>	ENC Histone... <input type="button" value="hide"/>
ENC RNA Binding... <input type="button" value="hide"/>	ENC TF Binding... <input type="button" value="hide"/>	18 ORegAnno <input type="button" value="hide"/>	Stanf Nucleosome <input type="button" value="hide"/>	SUNY SwitchGear <input type="button" value="hide"/>	17 SwitchGear TSS <input type="button" value="hide"/>
TFBS Conserved <input type="button" value="hide"/>	TS miRNA sites <input type="button" value="hide"/>	UMMS Brain Hist <input type="button" value="hide"/>	Vista Enhancers <input type="button" value="hide"/>	18 NKI Nuc Lamina... <input type="button" value="hide"/>	18 UCSF Brain Methyl <input type="button" value="hide"/>

Comparative Genomics

Conservation <input type="button" value="full"/>	18 Cons Indels MmCf <input type="button" value="hide"/>	GERP <input type="button" value="hide"/>	18 Evo Cpg <input type="button" value="hide"/>	Primate Chain/Net <input type="button" value="hide"/>	Placental Chain/Net <input type="button" value="hide"/>
hg19Patch2 Chain/Net <input type="button" value="hide"/>	Vertebrate Chain/Net <input type="button" value="hide"/>				

Neandertal Assembly and Analysis

<input type="button" value="+"/>					
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Variation and Repeats

Common SNPs(135) <input type="button" value="dense"/>	Flagged SNPs(135) <input type="button" value="hide"/>	Mult. SNPs(135) <input type="button" value="hide"/>	All SNPs(135) <input type="button" value="hide"/>	Common SNPs(132) <input type="button" value="dense"/>	Flagged SNPs(132) <input type="button" value="hide"/>
Mult. SNPs(132) <input type="button" value="hide"/>	All SNPs(132) <input type="button" value="hide"/>	SNPs (131) <input type="button" value="hide"/>	Arrays <input type="button" value="hide"/>	GIS DNA PET <input type="button" value="hide"/>	HAIB Genotype <input type="button" value="hide"/>
18 SNP Arrays <input type="button" value="hide"/>	HGDP Allele Freq <input type="button" value="hide"/>	18 HapMap SNPs <input type="button" value="hide"/>	DGV Struct Var <input type="button" value="hide"/>	Segmental Dups <input type="button" value="hide"/>	RepeatMasker <input type="button" value="dense"/>
Interrupted Rpts <input type="button" value="hide"/>	Simple Repeats <input type="button" value="hide"/>	Microsatellite <input type="button" value="hide"/>	Self Chain <input type="button" value="hide"/>	18 Genome Variants <input type="button" value="hide"/>	NumtS Sequence <input type="button" value="hide"/>

UCSC Genome Browser

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http://genome.ucsc.edu/ 

UCSC Genome Browser Home +

UCSC Genome Bioinformatics

Genomes - Blat - Tables - Gene Sorter - PCR - VisiGene - Proteome - Session - FAQ - Help

VERTEBRATES - Complete annotation sets

- [Human](#) (circled in red)
- [Cat](#)
- [Chicken](#)
- [Chimpanzee](#)
- [Cow](#)
- [Dog](#)
- [Elephant](#)
- [Fugu](#)
- [Gibbon](#)
- [Gorilla](#)
- [Guinea pig](#)
- [Horse](#)
- [Lamprey](#)
- [Lizard](#)
- [Marmoset](#)
- [Medaka](#)
- [Microbat](#)
- [Mouse](#)
- [Opossum](#)
- [Orangutan](#)
- [Panda](#)
- [Pig](#)
- [Platypus](#)
- [Rabbit](#)
- [Rat](#)
- [Rhesus](#)
- [Sheep](#)
- [Stickleback](#)
- [Tetraodon](#)
- [Turkey](#)
- [Wallaby](#)
- [X. tropicalis](#)
- [Zebra finch](#)
- [Zebrafish](#)

VERTEBRATES - Sequence downloads only

- [Armadillo](#)
- [Bushbaby](#)
- [European hedgehog](#)
- [Shrew](#)
- [Tenrec](#)
- [Tree shrew](#)
- [J. Craig Venter](#)

DEUTEROSTOMES

- [C. intestinalis](#)
- [Lancelet](#)
- [S. purpuratus](#)

INSECTS

- [A. gambiae](#)
- [A. mellifera](#)
- [D. ananassae](#)
- [D. erecta](#)
- [D. grimshawi](#)
- [D. melanogaster](#)
- [D. mojavensis](#)
- [D. persimilis](#)
- [D. pseudoobscura](#)
- [D. sechellia](#)
- [D. simulans](#)
- [D. virilis](#)
- [D. willistoni](#)
- [D. yakuba](#)
- [T. castaneum](#)

YEAST AND OTHERS

- [S. cerevisiae](#)
- [Sea hare](#)
- [Denisova](#)

OTHER DOWNLOADS

- [Shared Data \(Protein DBs, hgFixed, visiGene\)](#)
- [LiftOver Files](#)
- [ENCODE Project Files \(Genome-wide Phase\)](#)
- [ENCODE Project Files \(Pilot Phase\)](#)

Working draft assemblies for a large
olls over chromosomes, showing the
on groups of genes that can be
s convenient access to the
images to examine expression

cross-departmental team within the
z ([UCSC](#)). If you have feedback or
list.

News Archives ►

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The human assembly
on data not included in previous

of reference SNPs to the human
sting and easily defined subsets of

Done

Complete human genome

Human Genome

Feb. 2009 (hg19, GRCh37)

- Full data set
- Data set by chromosome
- Annotation database
- GC percent data
- Protein database for hg19
- SNP135-masked FASTA files
- SNP132-masked FASTA files
- SNP131-masked FASTA files
- LiftOver files
- Pairwise Alignments
 - Human self alignments
 - Human/Chimp (panTro3)
 - Human/Chimp (panTro2)
 - Human/Gorilla (gorGor3)
 - Human/Gorilla (gorGor1)
 - Human/Orangutan (ponAbe2)
 - Human/Gibbon (nomLeu1)
 - Human/Rhesus (rheMac2)
 - Human/Baboon (papHam1)
 - Human/Marmoset (calJac3)
 - Human/Marmoset (calJac1)
 - Human/Bushbaby (otoGar1)
 - Human/Mouse lemur (micMurl1)
 - Human/Tree shrew (tupBel1)
 - Human/Shrew (sorAral1)
 - Human/Mouse (mm9)
 - Human/Rat (rn4)
 - Human/Squirrel (speTri1)
 - Human/Guinea pig (cavPor3)
 - Human/Rabbit (oryCun2)
- Multiple Alignments
 - Multiple alignments of 45 vertebrate genomes with Human
 - Conservation scores for alignments of 45 vertebrate genomes with Human
 - Basewise conservation scores (phyloP) of 45 vertebrate genomes with Human
 - FASTA alignments of 45 vertebrate genomes with Human for CDS regions

Name	Last modified	Size	Description
Parent Directory		-	
chromAtp.tar.gz	20-Mar-2009 09:02	538K	
chromFa.tar.gz	20-Mar-2009 09:21	905M	
chromFaMasked.tar.gz	20-Mar-2009 09:30	477M	
chromOut.tar.gz	20-Mar-2009 09:03	163M	
chromTrf.tar.gz	20-Mar-2009 09:30	7.6M	
est.fa.gz	11-Feb-2012 23:01	1.4G	
est.fa.gz.md5	11-Feb-2012 23:01	44	
hg19.2bit	08-Mar-2009 15:29	778M	
md5sum.txt	29-Jul-2009 10:04	457	
mrna.fa.gz	11-Feb-2012 22:33	166M	
mrna.fa.gz.md5	11-Feb-2012 22:33	45	
refMrna.fa.gz	11-Feb-2012 23:01	40M	
refMrna.fa.gz.md5	11-Feb-2012 23:01	48	
upstream1000.fa.gz	12-Feb-2012 20:32	7.5M	
upstream1000.fa.gz.md5	12-Feb-2012 20:32	53	
upstream2000.fa.gz	12-Feb-2012 20:34	14M	
upstream2000.fa.gz.md5	12-Feb-2012 20:34	53	
upstream5000.fa.gz	12-Feb-2012 20:37	35M	
upstream5000.fa.gz.md5	12-Feb-2012 20:37	53	
xenoMrna.fa.gz	11-Feb-2012 22:41	1.9G	
xenoMrna.fa.gz.md5	11-Feb-2012 22:41	49	

chromFa.tar.gz - The assembly sequence in one file per chromosome.
Repeats from RepeatMasker and Tandem Repeats Finder (with period of 12 or less) are shown in lower case; non-repeating sequence is shown in upper case.

chromFaMasked.tar.gz - The assembly sequence in one file per chromosome. Repeats are masked by capital Ns; non-repeating sequence is shown in upper case.

chromOut.tar.gz - RepeatMasker .out files (one file per chromosome). RepeatMasker was run with the -s (sensitive) setting. Using: Jan 29 2009 (open-3-2-7) version of RepeatMasker and RELEASE 20090120 of library RepeatMaskerLib.embl

Transposable Elements

- 45% of the human genome is occupied by transposons and transposon-like repetitive elements.
- Barbara McClintock (1902-1992) in 50s.
- Nobel prize in 1983

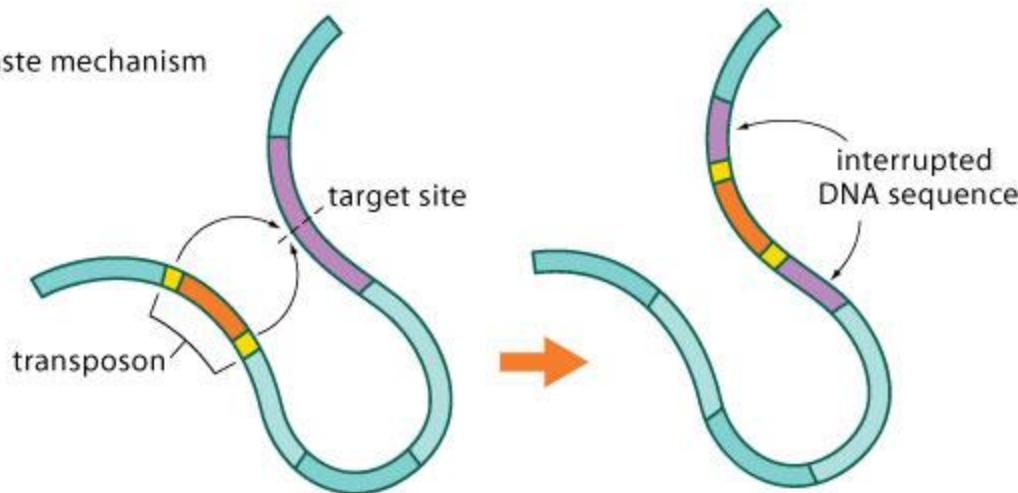


Gregory G. Dimijian, M.D./Photo Researchers

Two methods of transposition:

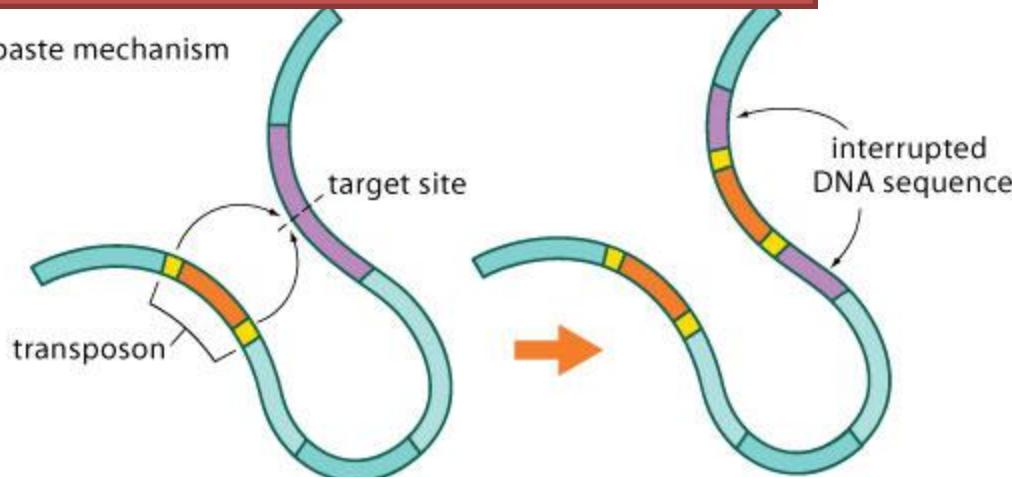
1. Cut-and-paste mechanism

Class II



Class I – retrotransposons (via RNA intermediate)

2. Copy-and-paste mechanism

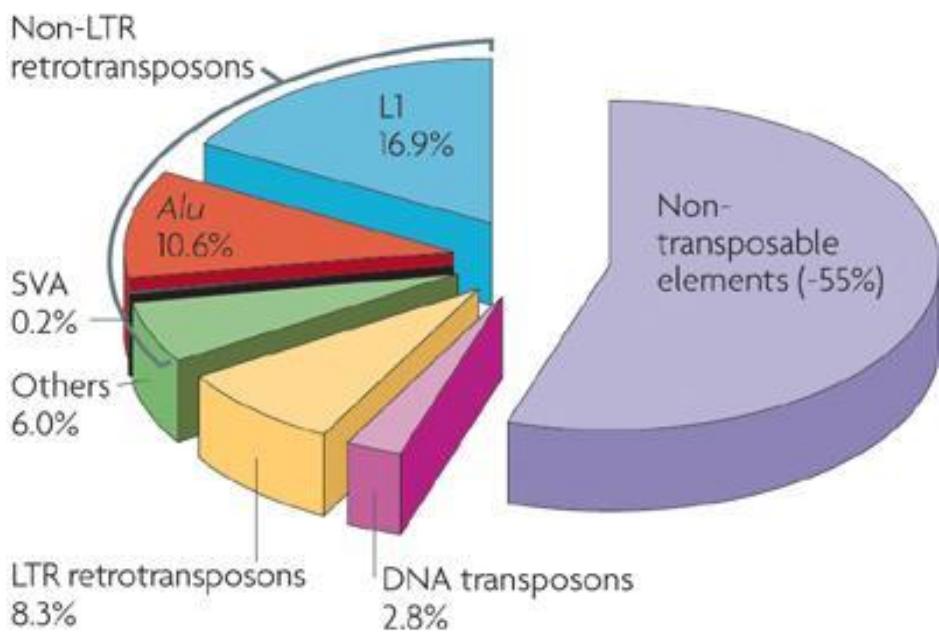


Схожесть с ретровирусами
Retrovirus reverse transcripton

<http://www.youtube.com/watch?v=eS1GODinO8w>

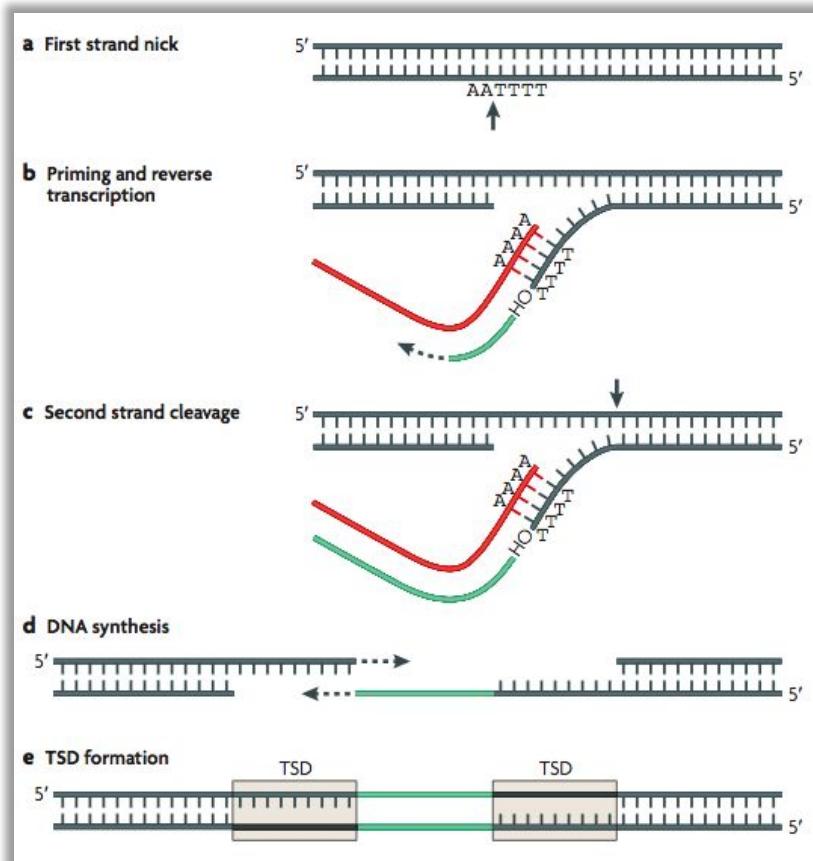
TABLE 17.1**Categorization of Transposable Elements by Transposition Mechanism**

Category	Examples	Host Organism
I. Cut-and-paste transposons	IS elements (e.g., IS50) Composite transposons (e.g., <i>Tn5</i>) <i>Ac/Ds</i> elements <i>P</i> elements <i>hobo</i> elements <i>piggyBac</i> <i>Sleeping Beauty</i>	Bacteria Bacteria Maize <i>Drosophila</i> <i>Drosophila</i> moth salmon
II. Replicative transposons	<i>Tn3</i> elements	Bacteria
III. Retrotransposons		
A. Retroviruslike elements (also called long terminal repeat, or LTR, retrotransposons)	<i>Ty1</i> <i>copia</i> <i>gypsy</i>	Yeast <i>Drosophila</i> <i>Drosophila</i>
B. Retroposons	<i>F</i> , <i>G</i> , and <i>I</i> elements Telomeric retroposons LINEs (e.g., <i>L1</i>) SINEs (e.g., <i>Alu</i>)	<i>Drosophila</i> <i>Drosophila</i> Humans Humans

a**b****Alu****SVA**

The impact of retrotransposons on human genome evolution

Richard Cordaux & Mark A. Batzer





Which transposable elements are active in the human genome?

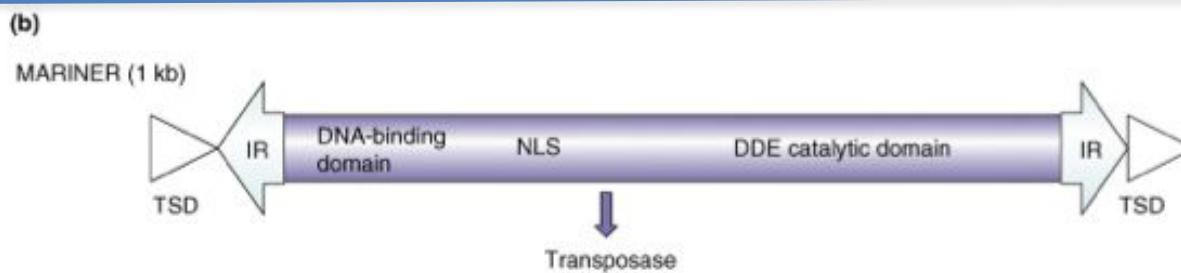
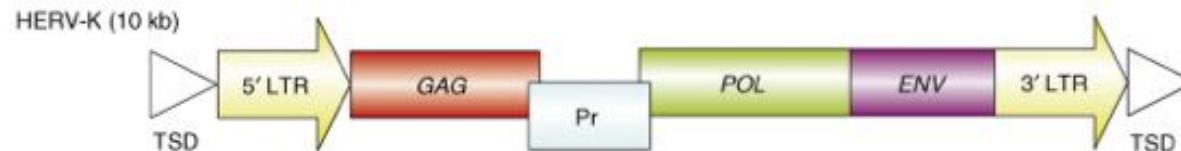
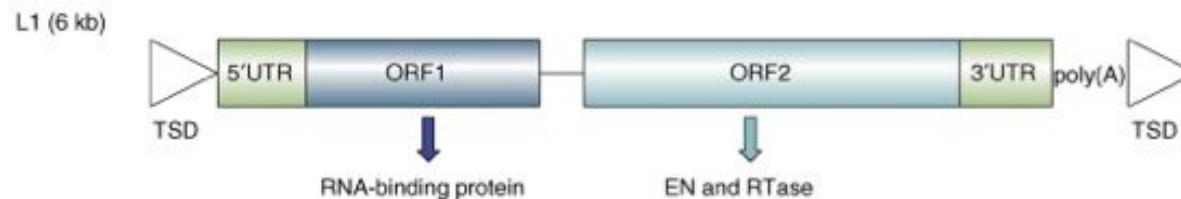
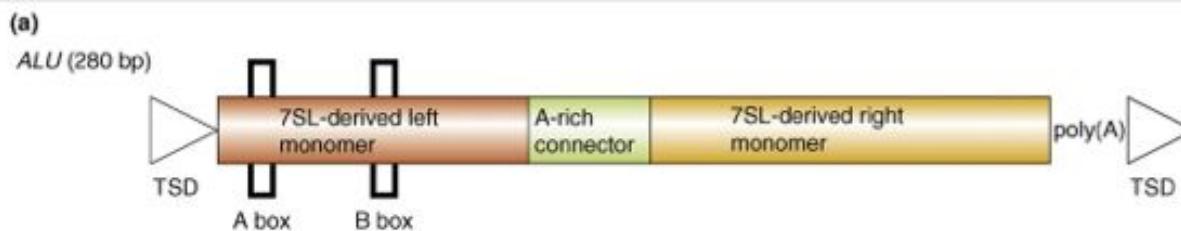
Ryan E. Mills^{1,2}, E. Andrew Bennett^{1,3}, Rebecca C. Iskow^{1,3} and Scott E. Devine^{1,2,3}

¹ Department of Biochemistry, Emory University School of Medicine, Atlanta, GA 30322, USA

² Center for Bioinformatics, Emory University School of Medicine, Atlanta, GA 30322, USA

³ Genetics and Molecular Biology Graduate Program, Emory University School of Medicine, Atlanta, GA 30322, USA

Although a large proportion (44%) of the human genome is occupied by transposons and transposon-like repetitive elements, only a small proportion (<0.05%) of these elements remain active today. Recent evidence indicates that ~35–40 subfamilies of *Alu*, L1 and SVA elements (and possibly HERV-K elements) remain actively mobile in the human genome. These active transposons are of great interest because they continue to produce genetic diversity in human populations and also cause human diseases by integrating into genes. In this review, we examine these active human transposons and explore mechanistic factors that influence their mobilization.



Non-Active

First Layer of Genome Annotation

UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

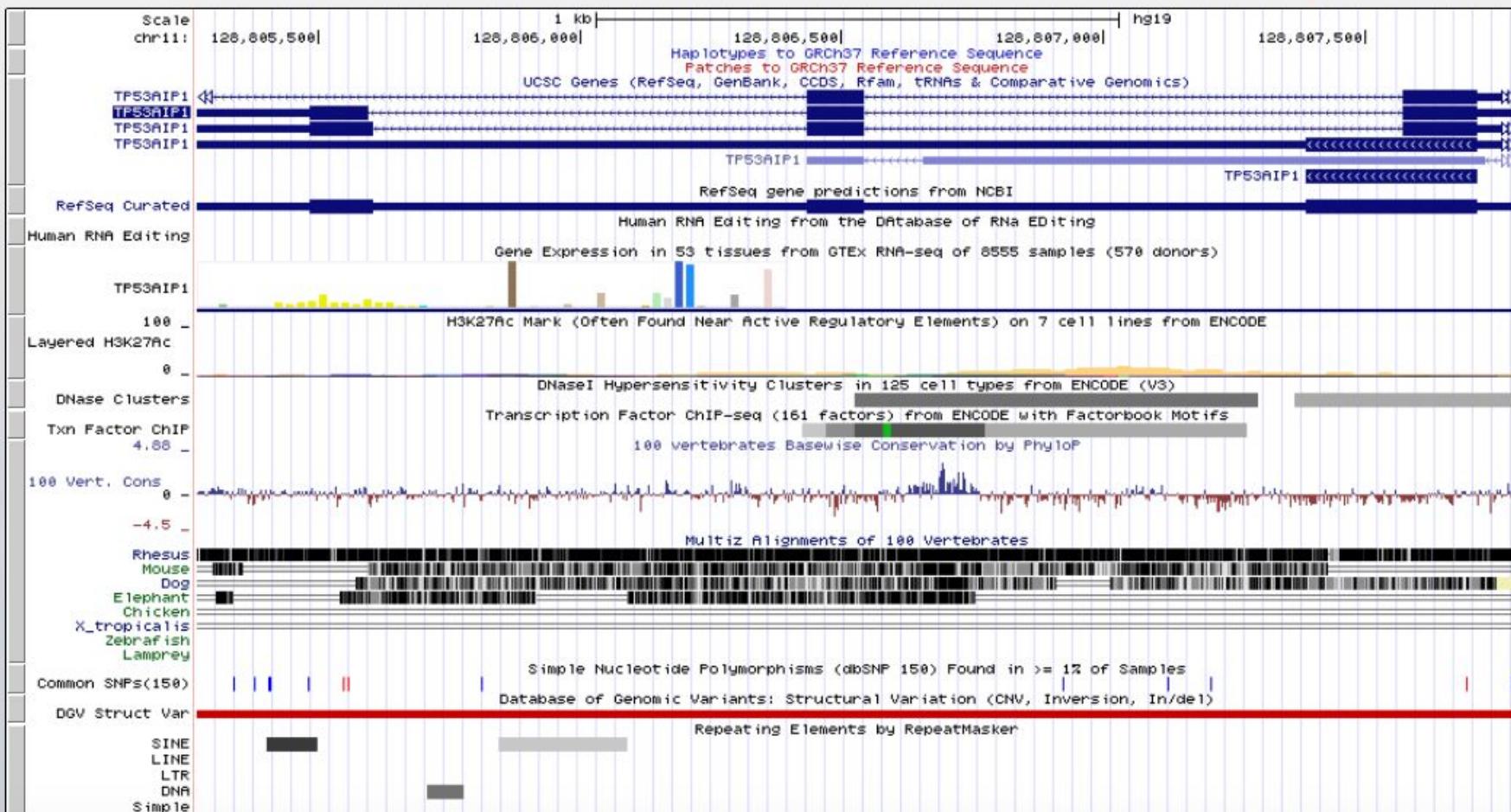
move <<< << < > >> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

chr11:128,805,270-128,807,789 2,520 bp.

enter position, gene symbol, HGVS or search terms

go

See us FREE @ ASHG Wed 7



Epigenetics

Levels of DNA Packaging in Eukaryotes

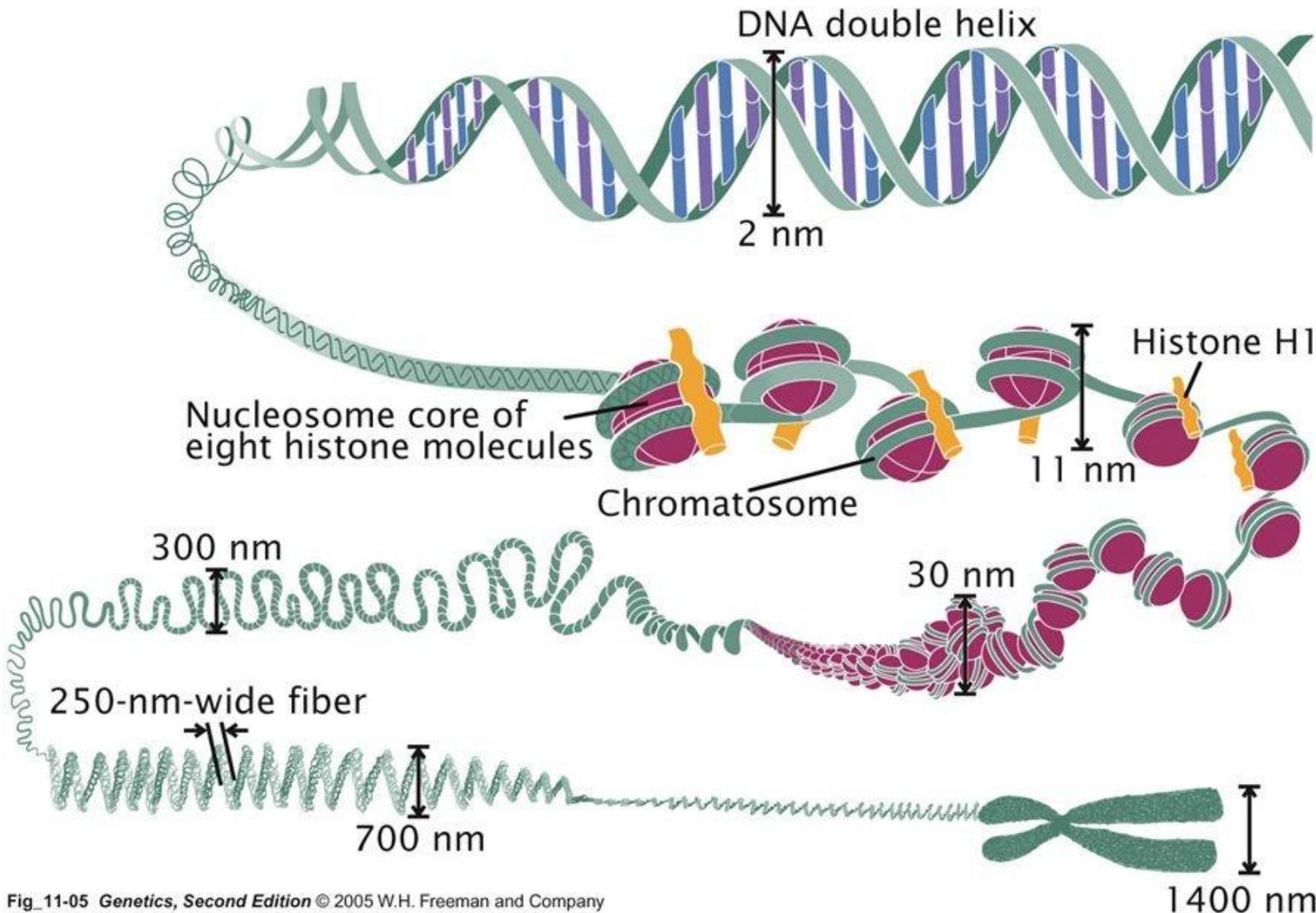
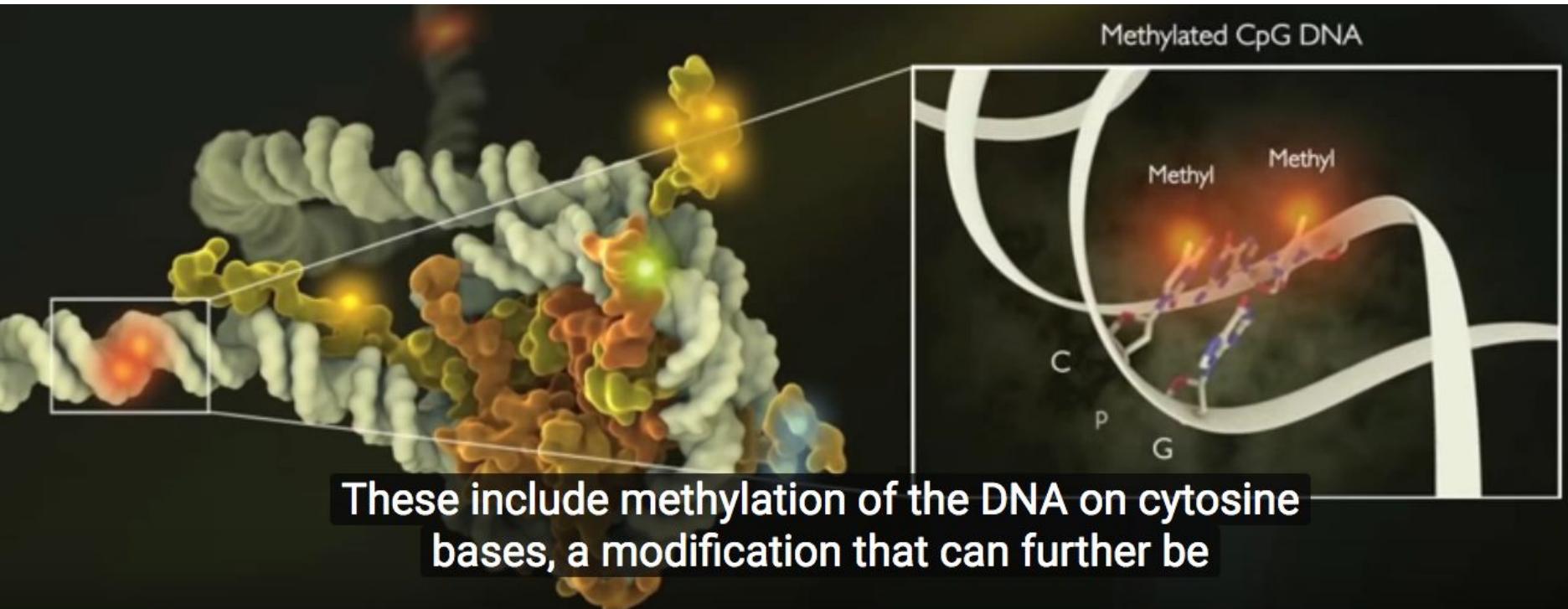
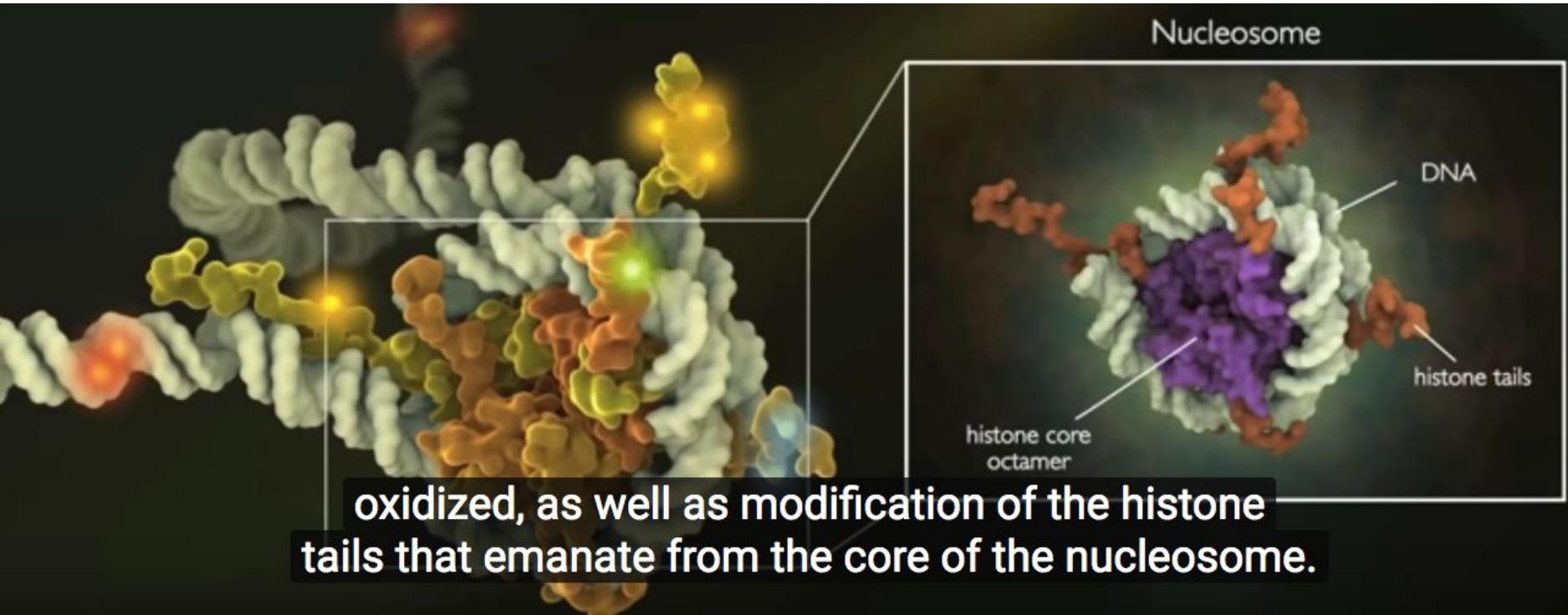


Fig. 11-05 *Genetics, Second Edition* © 2005 W.H. Freeman and Company

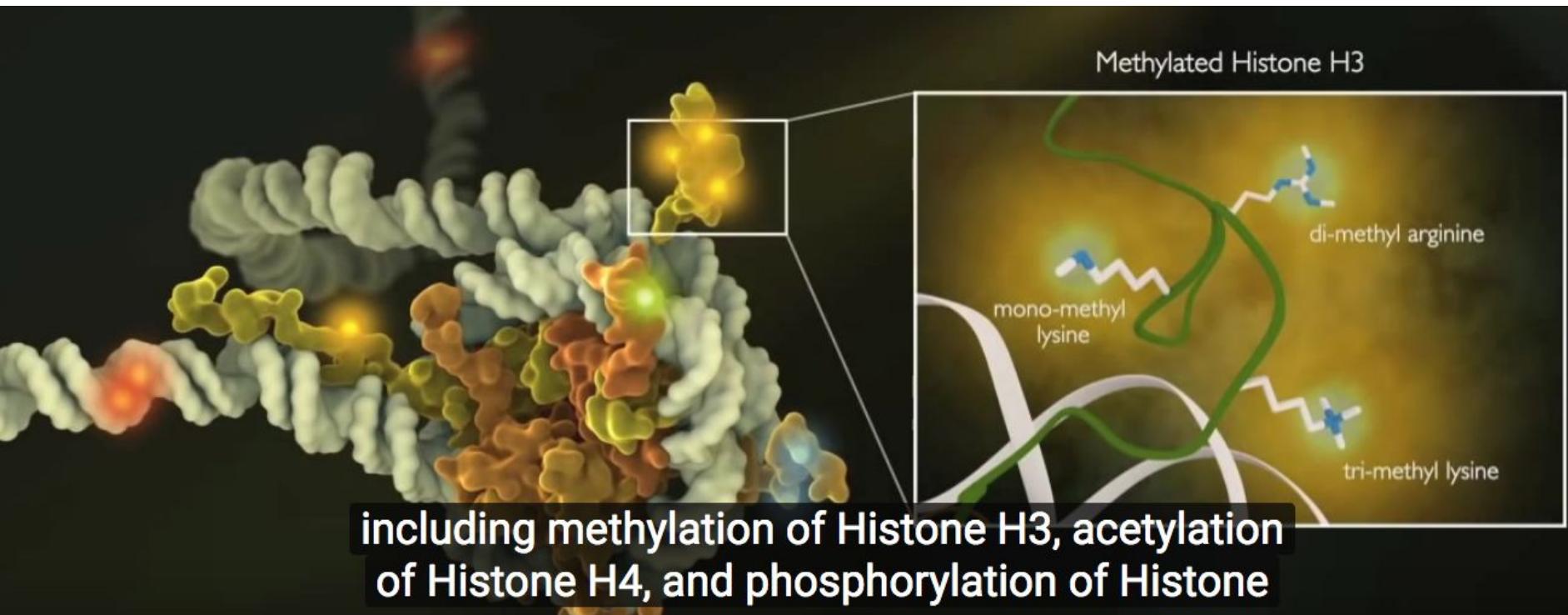
Second Layer of Genome Annotation



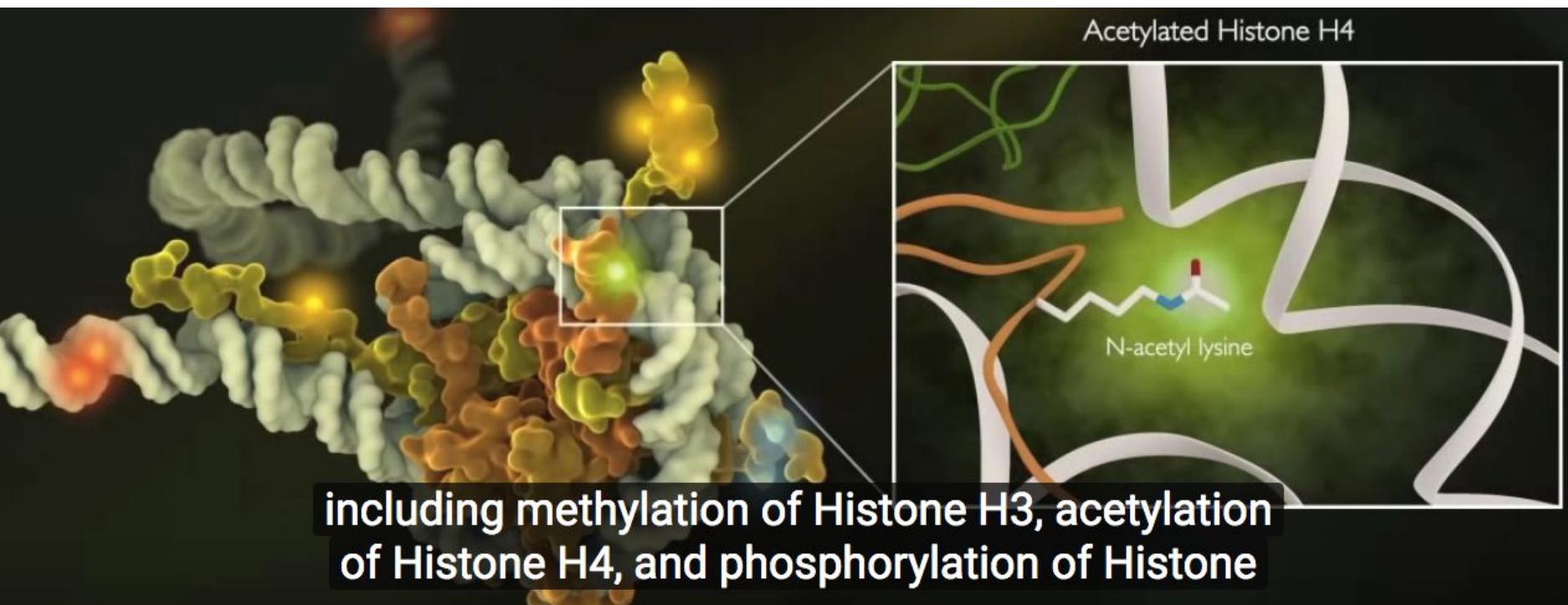
Second Layer of Genome Annotation



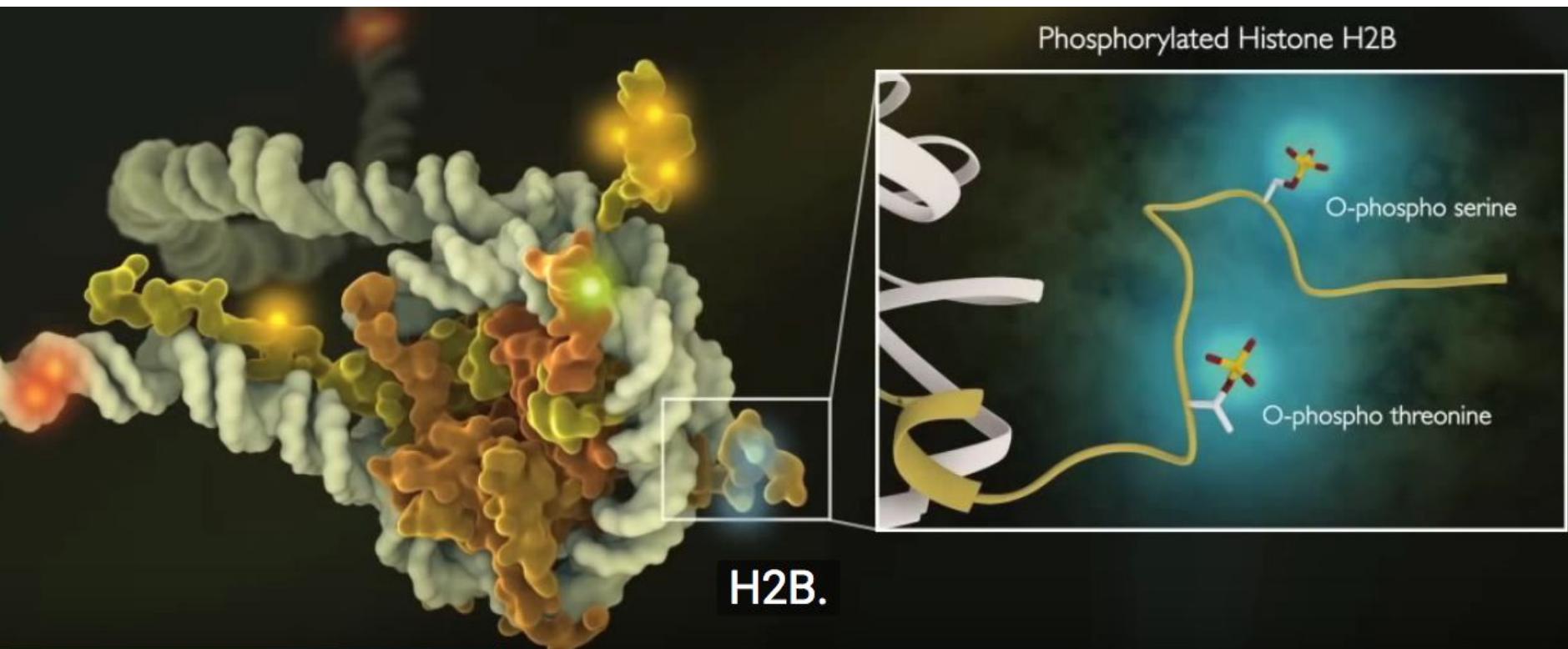
Second Layer of Genome Annotation



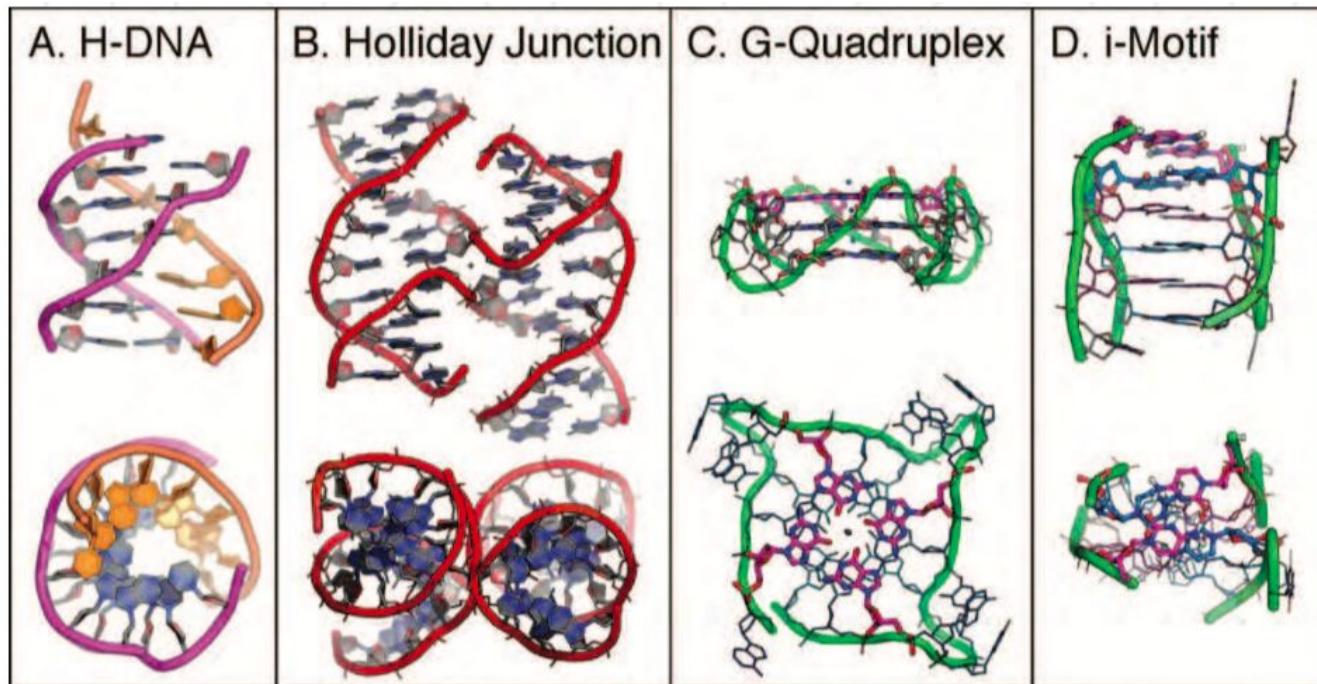
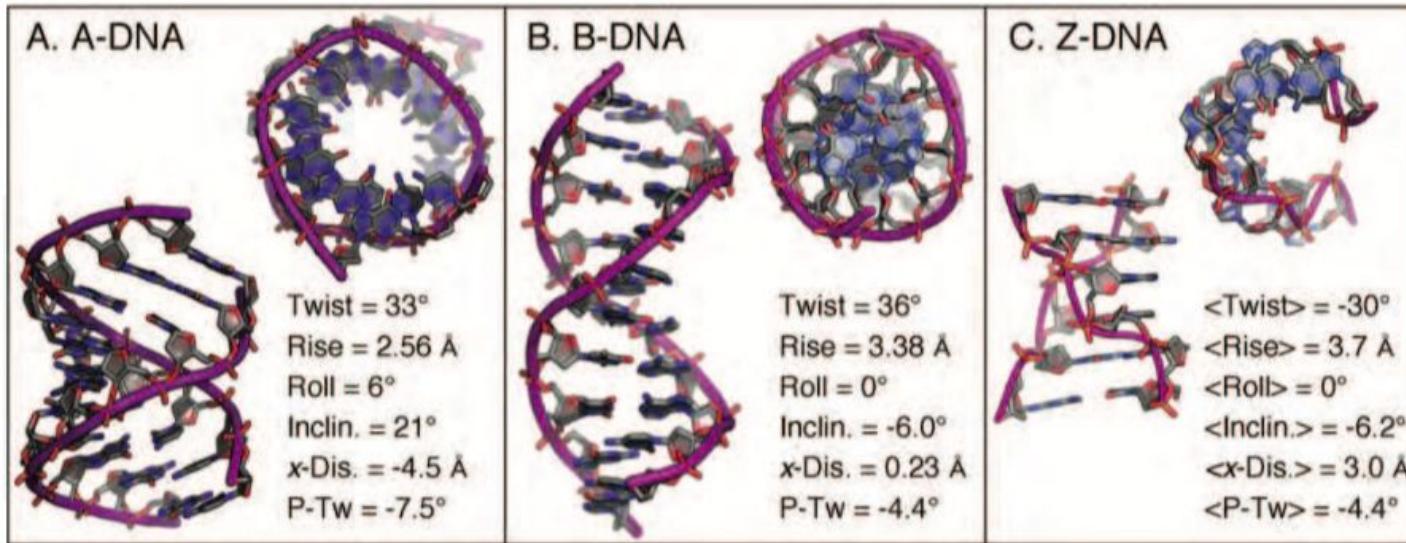
Second Layer of Genome Annotation



Second Layer of Genome Annotation



Third Layer of Genome Annotation

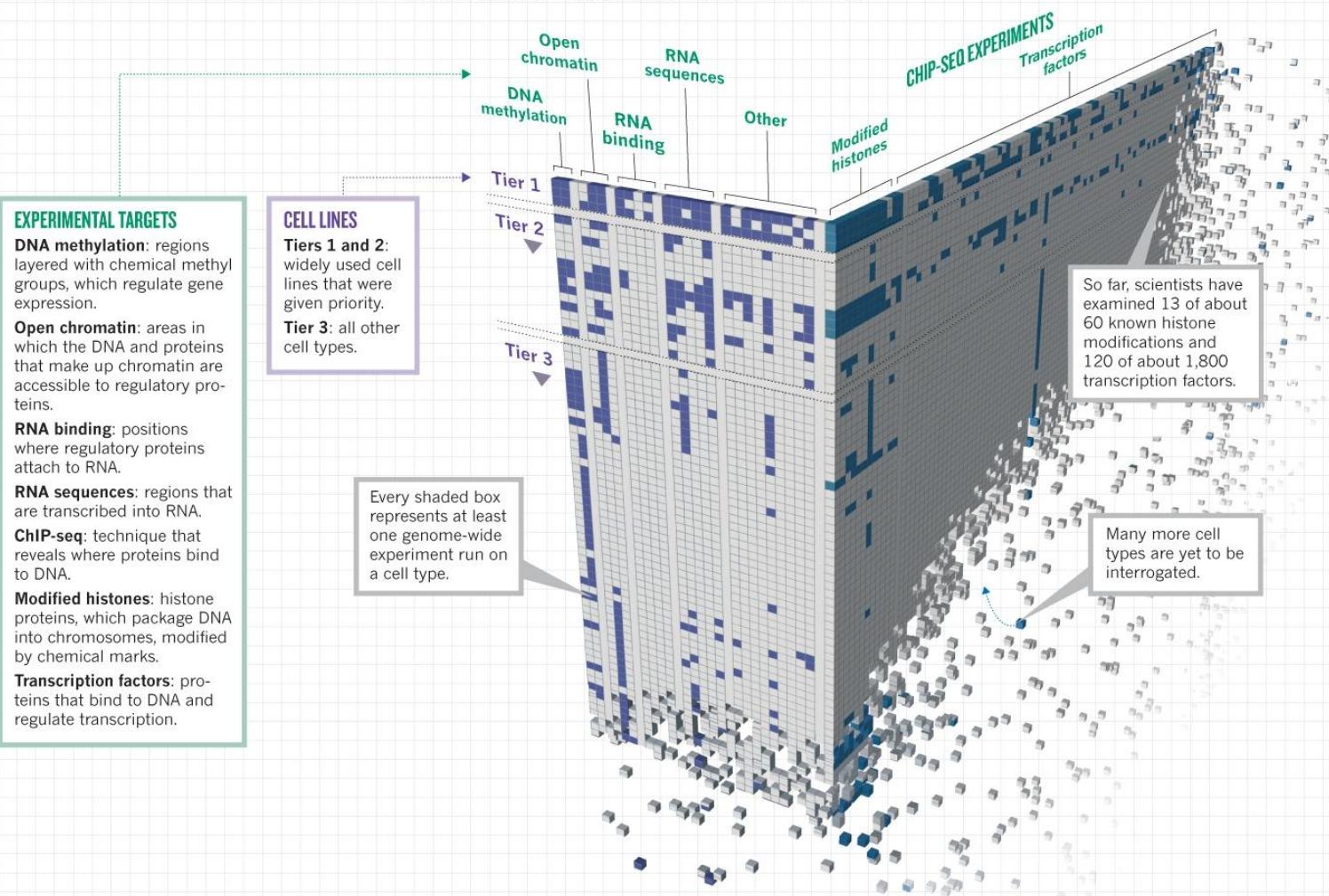


Data Accumulation

ENCODE: Encyclopedia of DNA Elements

MAKING A GENOME MANUAL

Scientists in the Encyclopedia of DNA Elements Consortium have applied 24 experiment types (across) to more than 150 cell lines (down) to assign functions to as many DNA regions as possible — but the project is still far from complete.

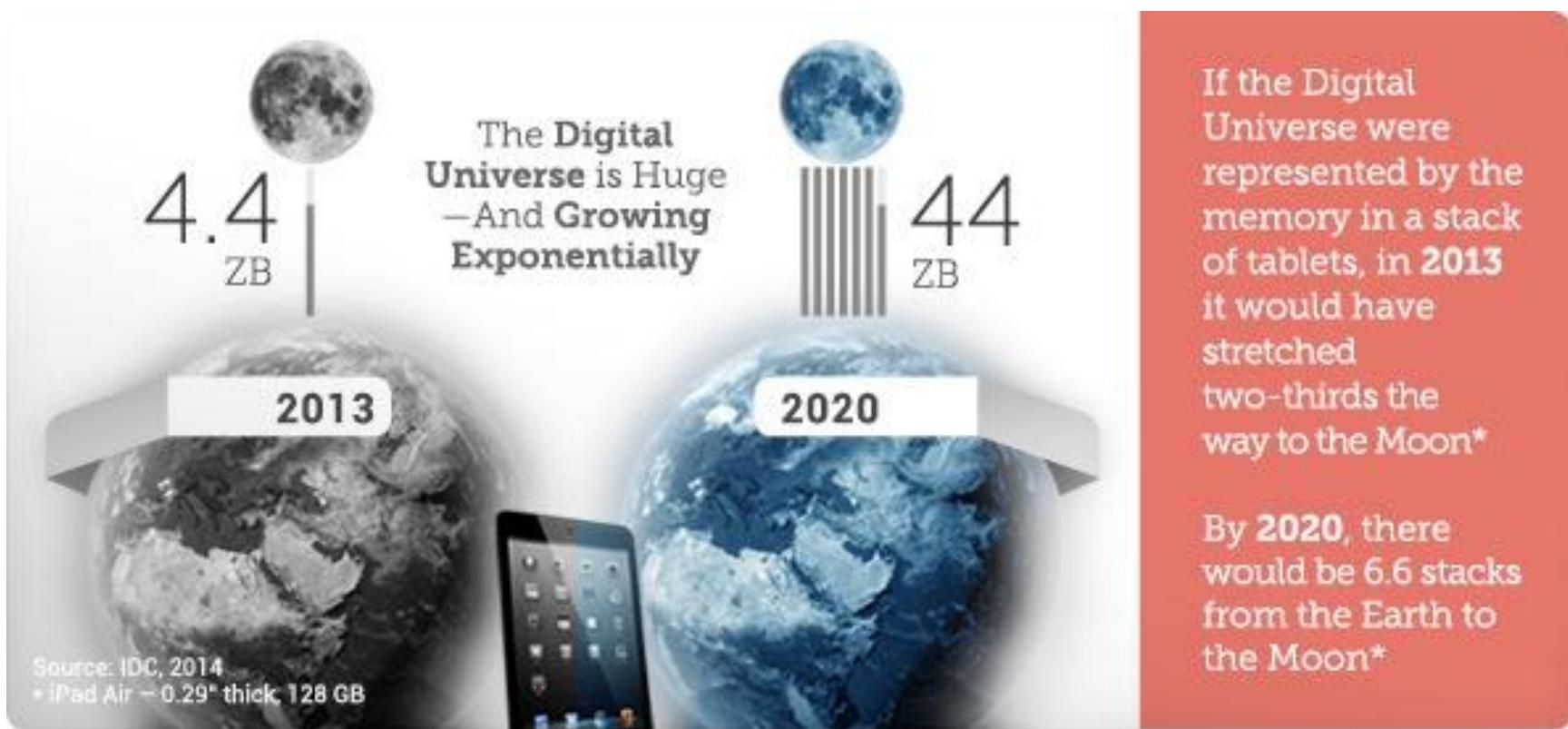


Digital Universe

- Like the Physical Universe the Digital Universe is also expanding but much faster doubling every two years – and by 2020 will be 44 zettabytes ($10^{\wedge} 21$)
- Every second a new 205 000 bytes come to being
- At the end of this lecture the digital universe will grow by 2 214 000 000 bytes or 2.2 GB.

Digital Universe

Data Universe Will Expand To 44 Trillion GBs By 2020

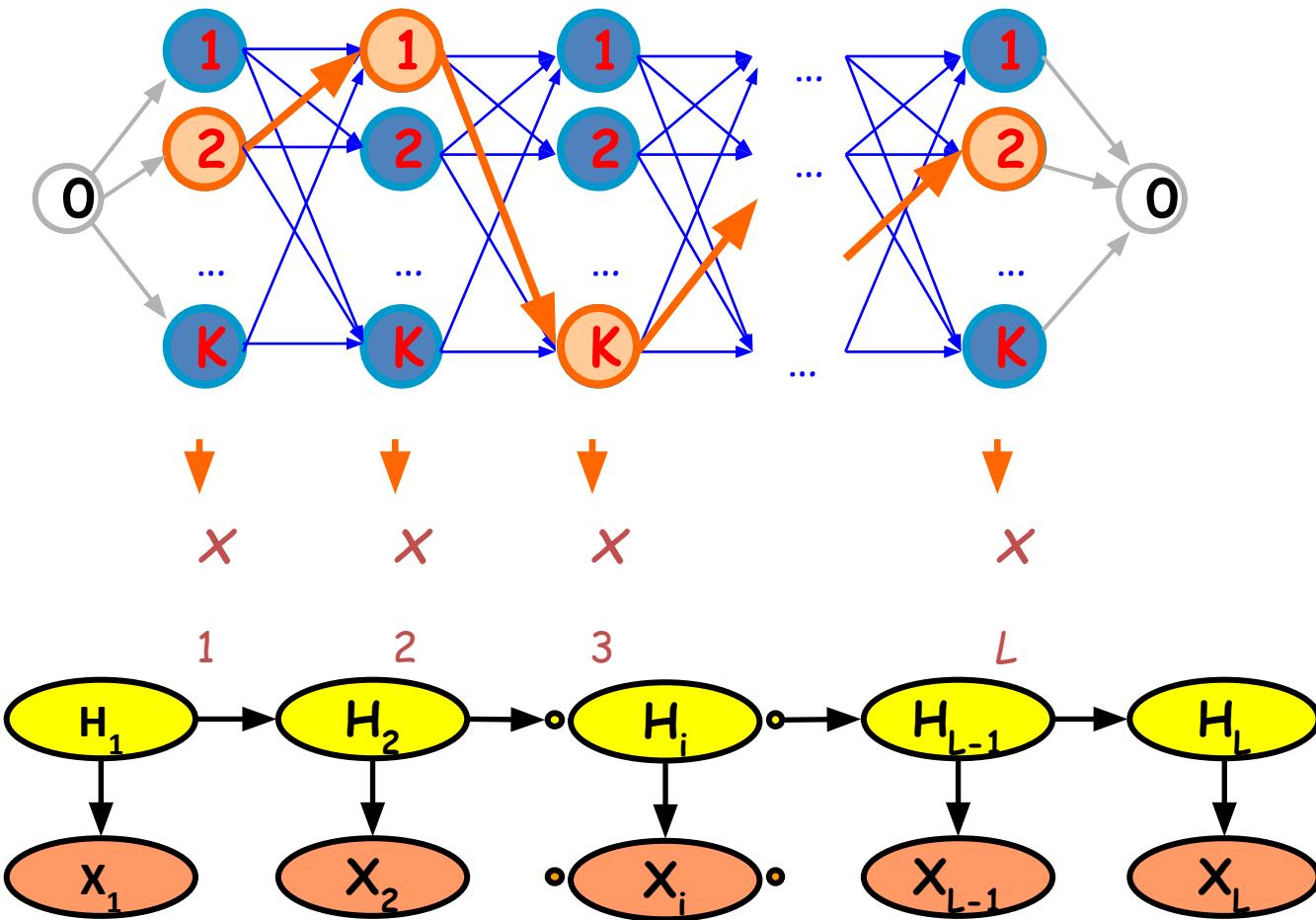


Что делать?

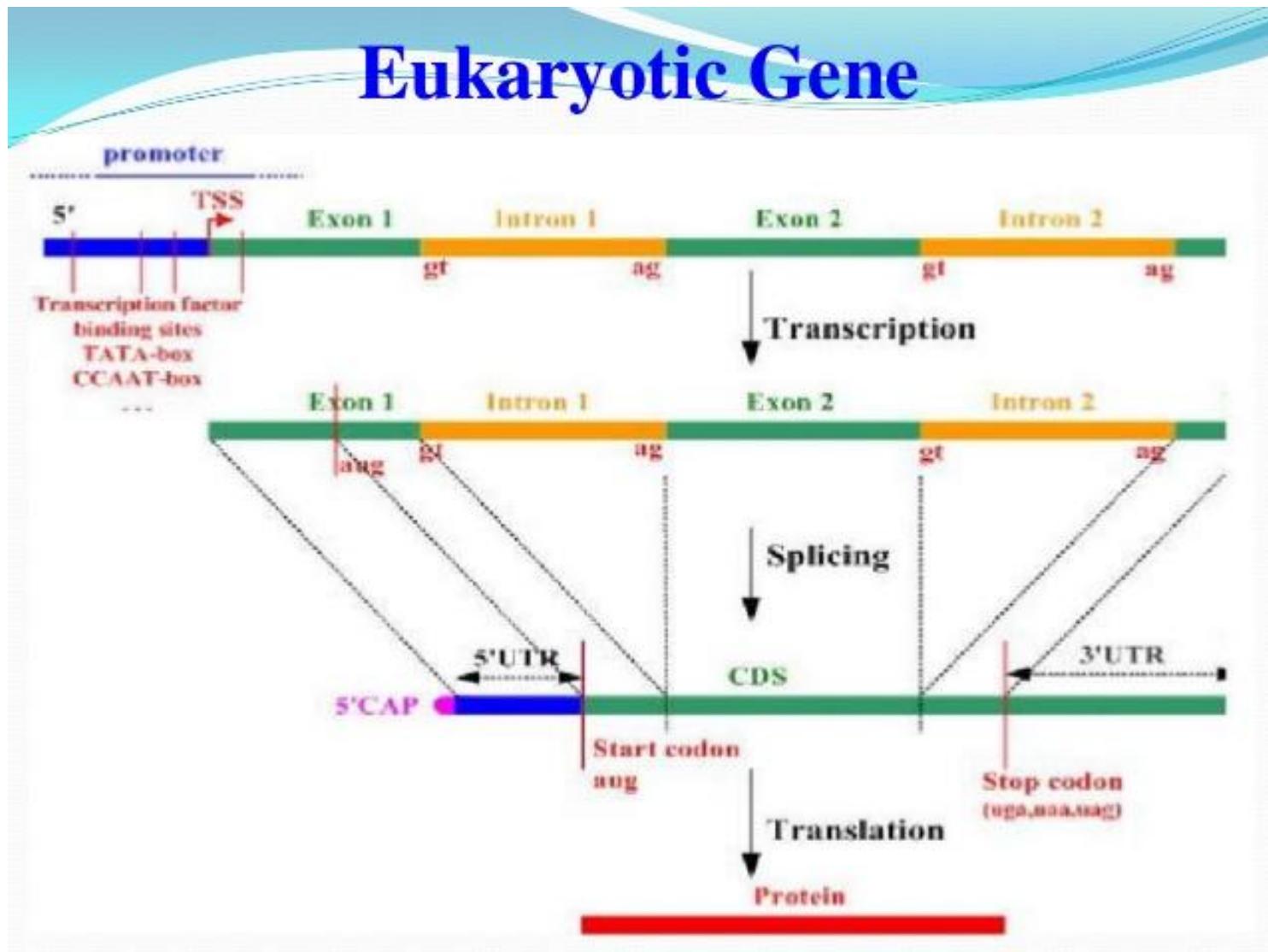


Что получилось? (Success Stories)

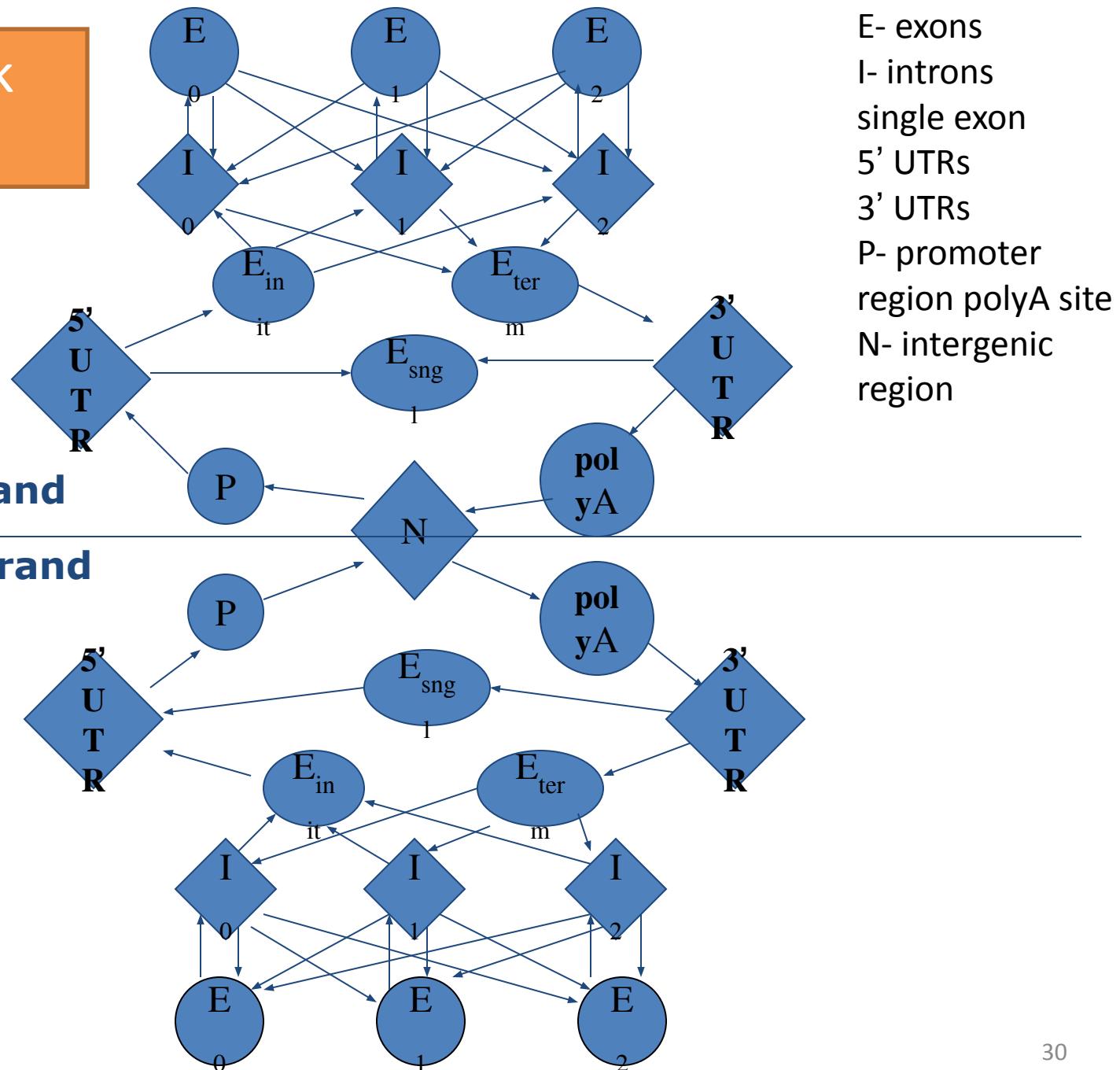
СКРЫТЫЕ ЦЕПИ МАРКОВА



Gene Prediction



GeneMark HMM



Promoter prediction

McPromoter

- Hidden Markov model with six interpolated Markov chain submodels
 - upstream 1 and 2,
 - TATA box, spacer,
 - Initiator
 - downstream.
 - Gaussian densities of DNA physicochemical properties.
- Neural network classifier

Deciphering the splicing code

Nature 2010

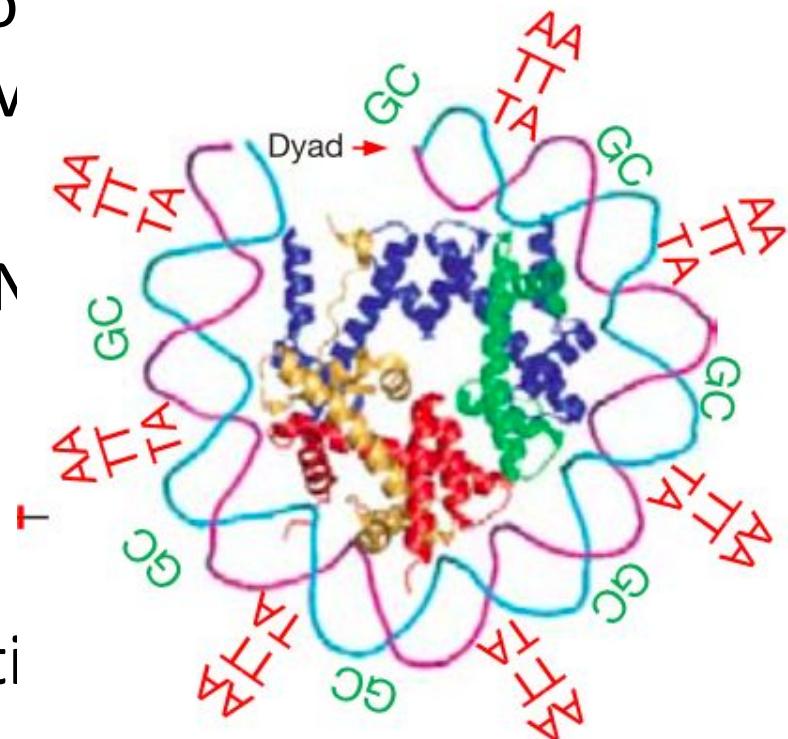
Yoseph Barash^{1,2*}, John A. Calarco^{2*}, Weijun Gao¹, Qun Pan², Xinchen Wang^{1,2}, Ofer Shai¹, Benjamin J. Blencowe²
& Brendan J. Frey^{1,2,3}

- predict tissue-dependent changes in alternative splicing for thousands of exons.
- 1,014 features: known motifs, new motifs, short motifs and features describing transcript structure
- trained on RNA-seq data
- single-layer logistic Bayesian network or neural network, or a weighted combination of single-layer decision trees.

A genomic code for nucleosome positioning

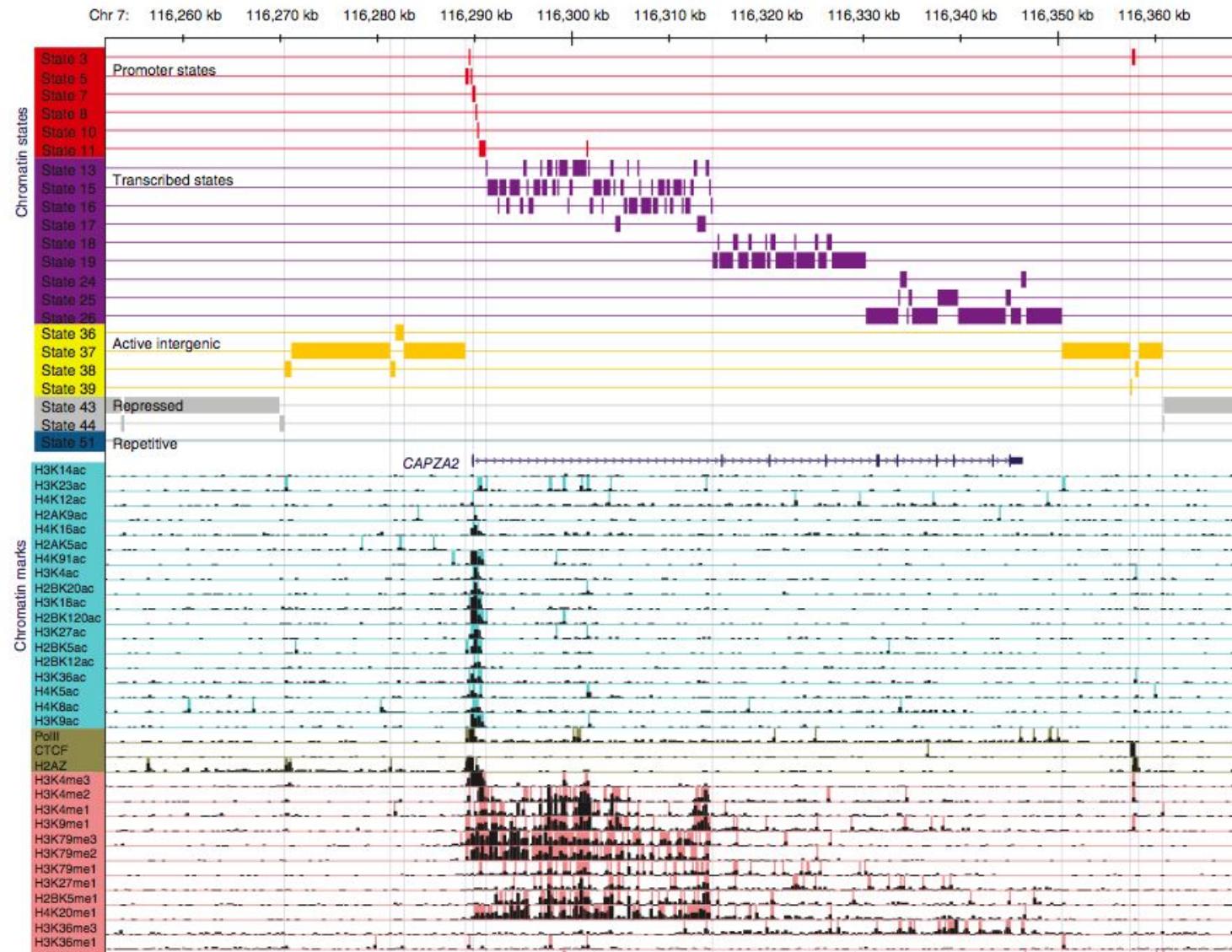
Eran Segal¹, Yvonne Fondufe-Mittendorf², Lingyi Chen², AnnChristine Thåström², Yair Field¹, Irene K. Moore², Ji-Ping Z. Wang³ & Jonathan Widom²

- Genome intrinsic organization can explain ~50% of the in vivo nucleosome positions
- Probabilistic nucleosome–DNA interaction model - built on dinucleotide distribution
- Thermodynamic model for predicting nucleosome positions genome-wide.

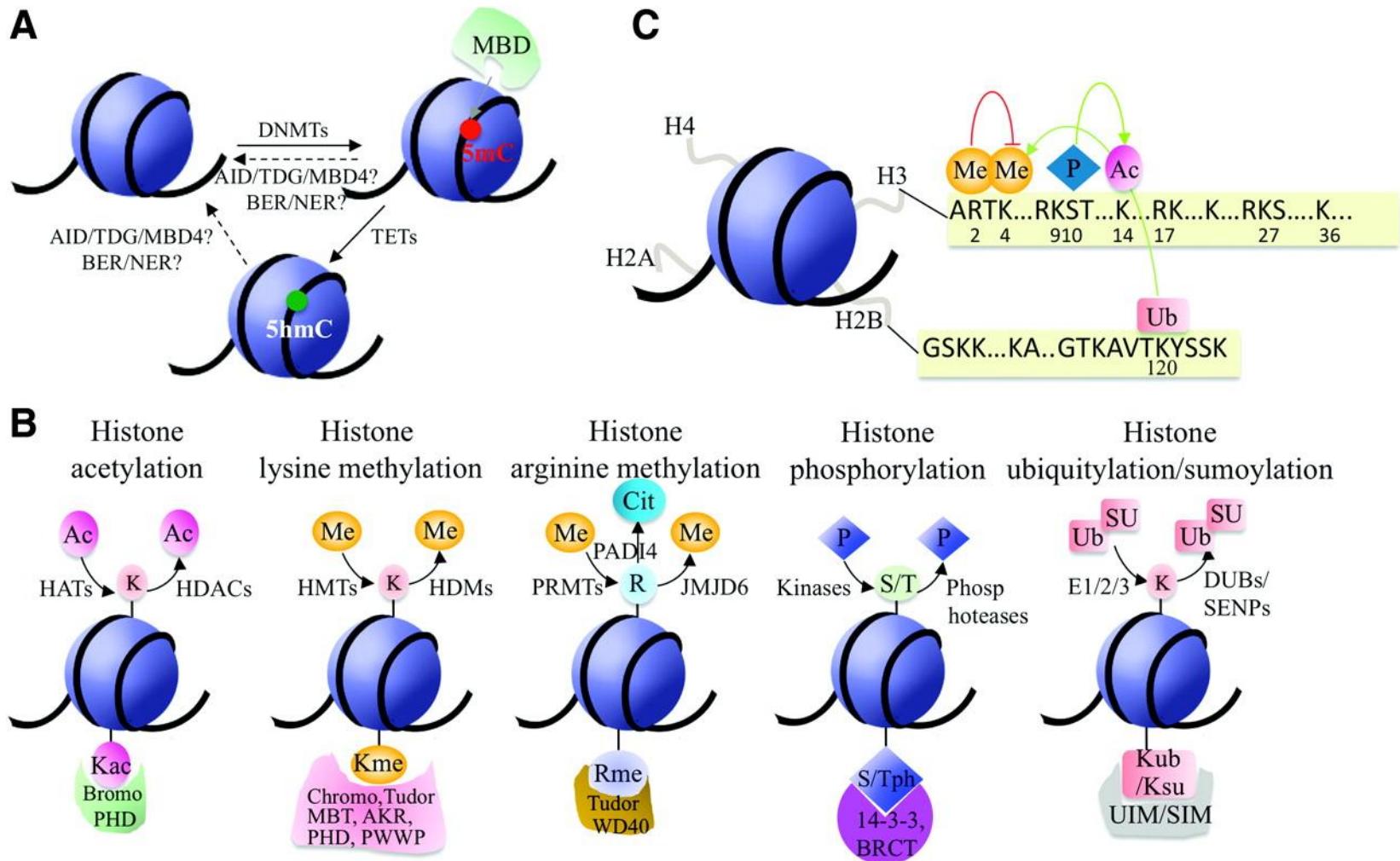


Discovery and characterization of chromatin states for systematic annotation of the human genome

Jason Ernst^{1,2} & Manolis Kellis^{1,2}



Schematic overview of epigenetic regulatory mechanisms.

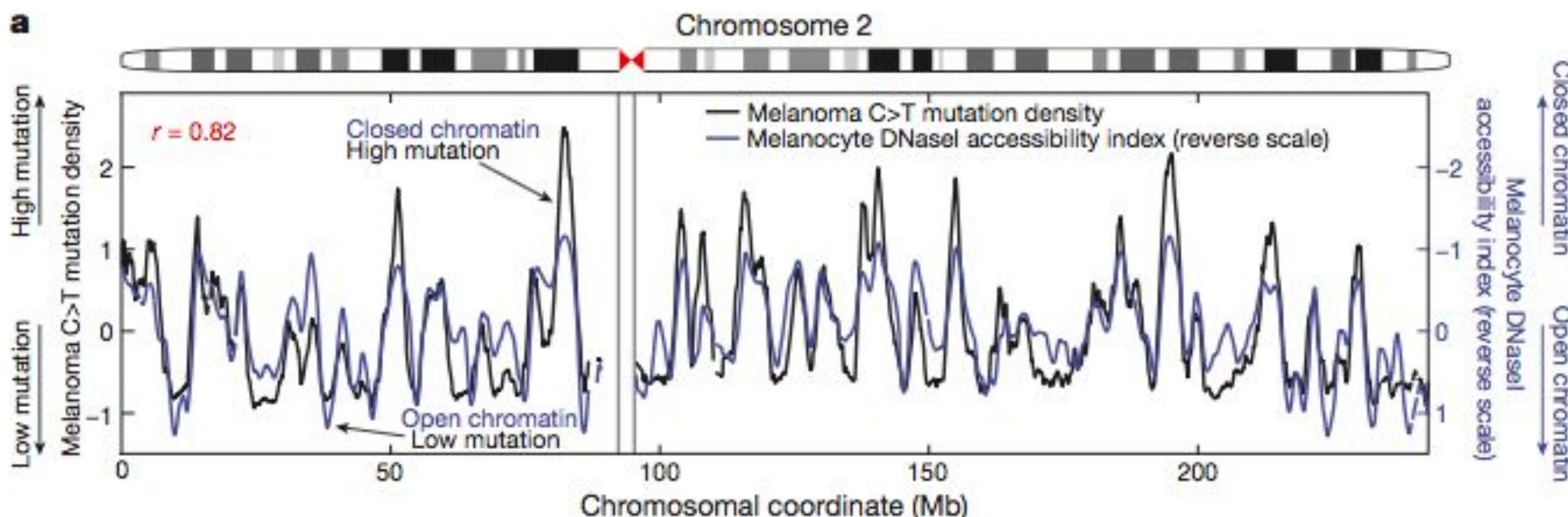


Yonggang Zhou et al. Circ Res. 2011;109:1067-1081

Cell-of-origin chromatin organization shapes the mutational landscape of cancer

Paz Polak^{1,2*}, Rosa Karlić^{3*}, Amnon Koren^{2,4}, Robert Thurman⁵, Richard Sandstrom⁵, Michael S. Lawrence², Alex Reynolds⁵, Eric Rynes⁵, Kristian Vlahoviček^{3,6}, John A. Stamatoyannopoulos^{5,7} & Shamil R. Sunyaev^{1,2}

Random Forest model predicts cancer mutation densities from epigenomic mark ups



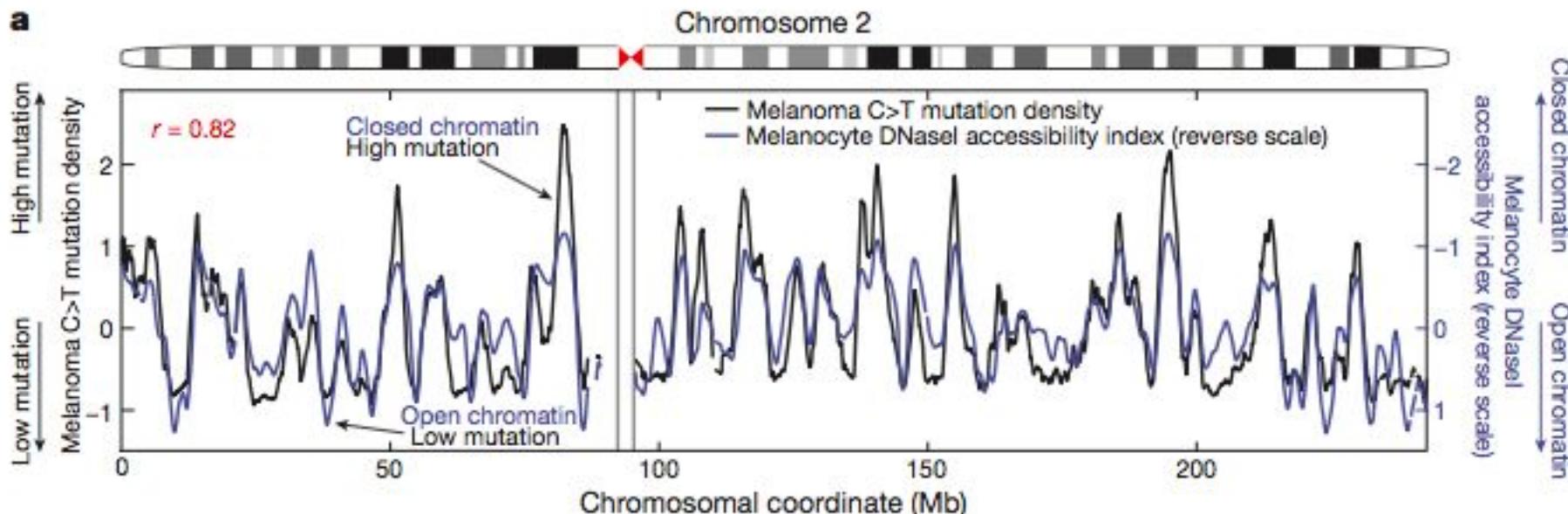
Cell-of-origin chromatin organization shapes the mutational landscape of cancer

Paz Polak^{1,2*}, Rosa Karlić^{3*}, Amnon Koren^{2,4}, Robert Thurman⁵, Richard Sandstrom⁵, Michael S. Lawrence², Alex Reynolds⁵, Eric Rynes⁵, Kristian Vlahoviček^{3,6}, John A. Stamatoyannopoulos^{5,7} & Shamil R. Sunyaev^{1,2}

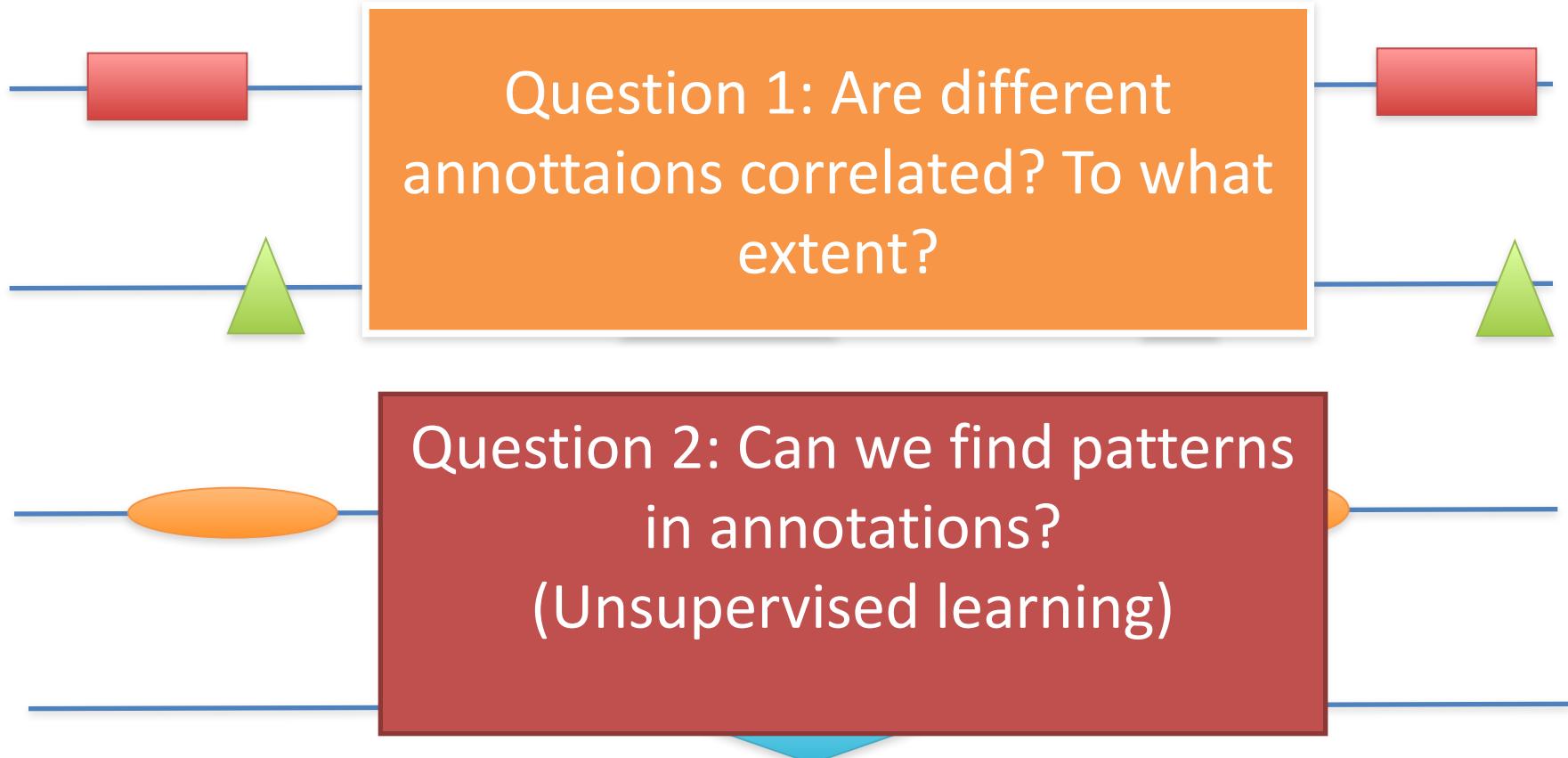
¹Division of Genetics, Department of Medicine, Brigham & Women's Hospital and Harvard Medical School, Boston, Massachusetts 02115, USA. ²The Broad Institute of MIT and Harvard, Cambridge, Massachusetts 02142, USA. ³Bioinformatics Group, Department of Molecular Biology, Division of Biology, Faculty of Science, University of Zagreb, Horvatovac 102a, 10000 Zagreb, Croatia. ⁴Department of Genetics, Harvard Medical School, Boston, Massachusetts 02115, USA. ⁵Department of Genome Sciences, University of Washington, Seattle, Washington 98195, USA. ⁶Department of Informatics, University of Oslo, P.O. Box 1080, Blindern, NO-0316 Oslo, Norway. ⁷Department of Medicine, Division of Oncology, University of Washington, Seattle, Washington 98195, USA.

*These authors contributed equally to this work.

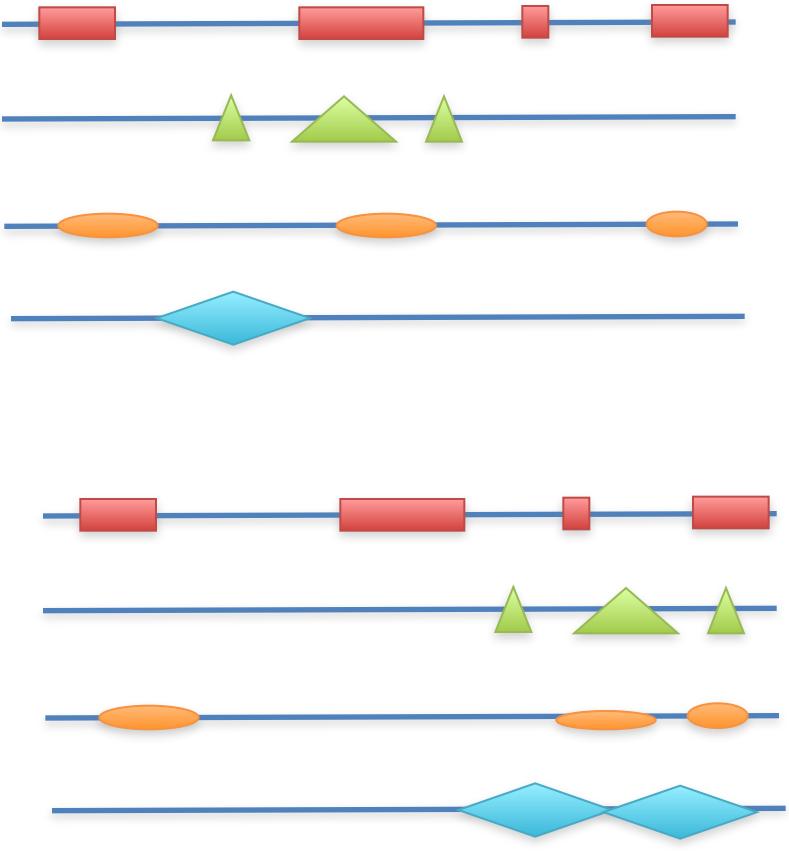
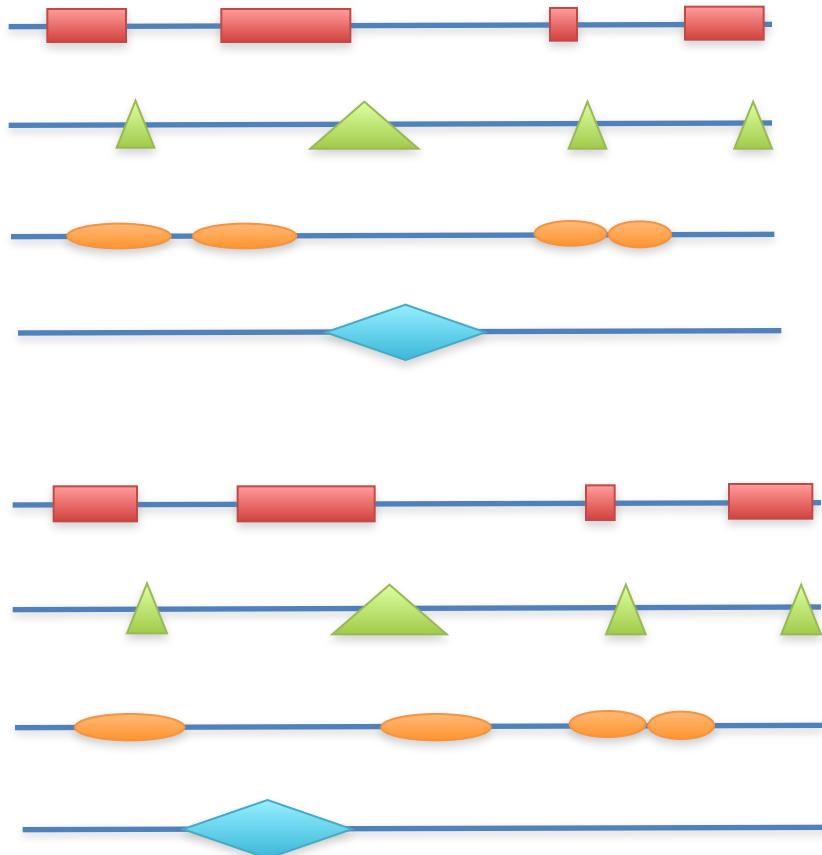
360 | NATURE | VOL 518 | 19 FEBRUARY 2015



We have many experimental genome-wide annotations



Annotations under different conditions



Как много данных?

- Roadmap Epigenomics
~ 3 000 полногеномных данных
- ENCODE Encyclopedia of Genomic Elements
~ 9000 полногеномных данных
- International Cancer Genome Consortium
~ 20 000 patients (~50 типов рака)
- The Cancer Genome Atlas
~ patients 11 000 (~33 типа рака)

Открытые вопросы

- Какие участки кода работают одновременно?
- Как переключать режимы работы клетки?
- Как перепрограммируется код для разных типов тканей?
- Сколько механизмов регуляции существует в клетках (надежда на универсальность)?



Спасибо за внимание

