

applied genomics

BIOL5382 - Applied Genomics Laboratory Course

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databases

- *how to access genomic information*
- *UCSC genome browser*
 - *case story for TBXT gene evolution*
- *EnsEMBL*
- *GTEX (genome tissue expression)*

<https://genome.ucsc.edu>

UCSC Genome Browser Home x + https://genome.ucsc.edu New Chrome available :

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UNIVERSITY OF CALIFORNIA SANTA CRUZ Genomics Institute UCSC Genome Browser

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Search genes, data, help docs and more... Search

Tools

- [Genome Browser](#) - Interactively visualize genomic data
- [BLAT](#) - Rapidly align sequences to the genome
- [In-Silico PCR](#) - Rapidly align PCR primer pairs to the genome
- [Table Browser](#) - Download and filter data from the Genome Browser
- [LiftOver](#) - Convert genome coordinates between assemblies
- [REST API](#) - Returns data requested in JSON format
- [Variant Annotation Integrator](#) - Annotate genomic variants
- [More tools...](#)

hg38 hg19 mm39

News

- Jun. 7, 2024 - [New GENCODE Versions tracks for hg19/hg38/mm39 \(V46/VM35\)](#)
- May. 22, 2024 - [New GENCODE gene tracks: V46 \(hg38\) - VM35 \(mm39\)](#)
- Apr. 25, 2024 - [New AbSplice Prediction Scores track for hg19](#)
- Mar. 26, 2024 - [New gnomAD v4 Constraint Metrics \(hg38\) and gnomAD Non-canc...](#)
- Mar. 07, 2024 - [New Prediction Scores super track and BayesDel track for hg19](#)
- Mar. 05, 2024 - [New JASPAR tracks: Human \(hg19/hg38\) - Mouse \(mm10/mm39\)](#)

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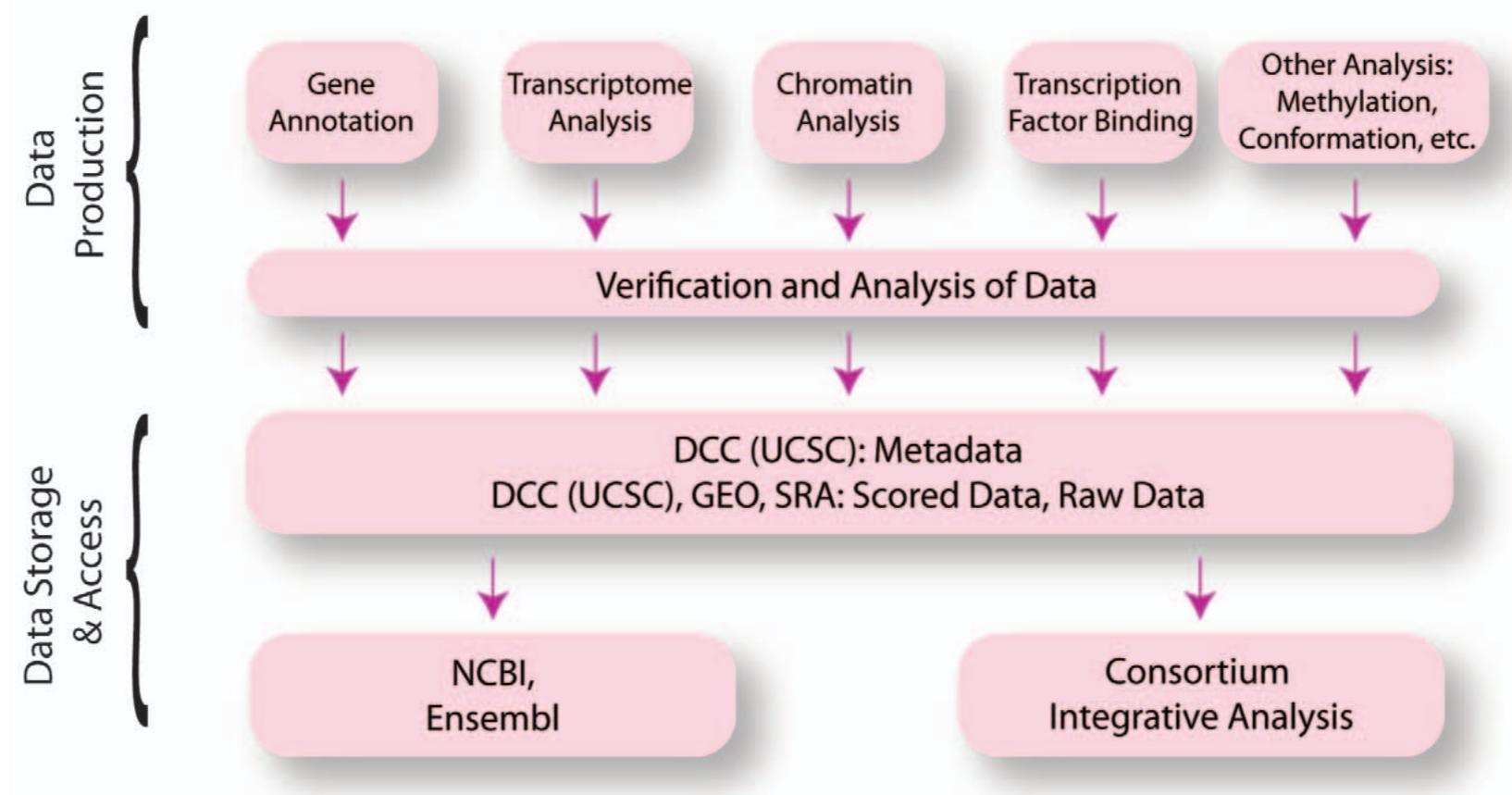
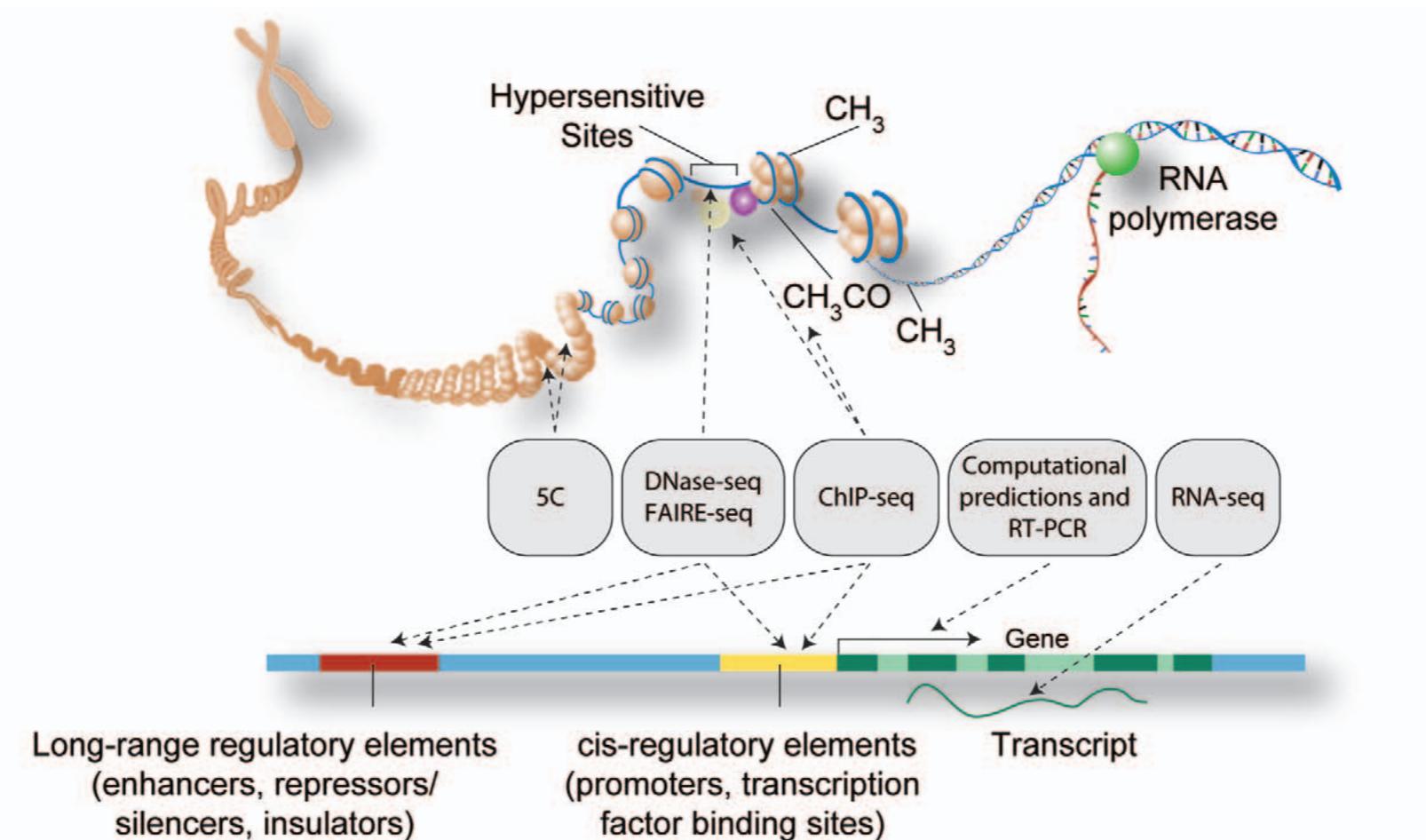
Meetings and Workshops: Come see us in person!

- NYU Langone Center for Human Genetics and Genomics - New York, NY. June 24, 2024.
- [McKusick Summer Course in Human and Mammalian Genetics and Genomics](#) - Bar Harbor, ME. July 24, 2024.
- [Brazilian Society for Medical Genetics](#) - Cuiaba, Brazil. August 28, 2024.
- Faculty of Medicine, University of Chile, West Campus - Santiago, Chile. Sep 3-4, 2024

Feel free to [contact us](#) if you are interested in attending a workshop, or meeting someone from the team to collaborate, get help, or ask any questions at the meetings.

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<https://news.ucsc.edu/2015/06/genome-anniversary.html>



genome browsing

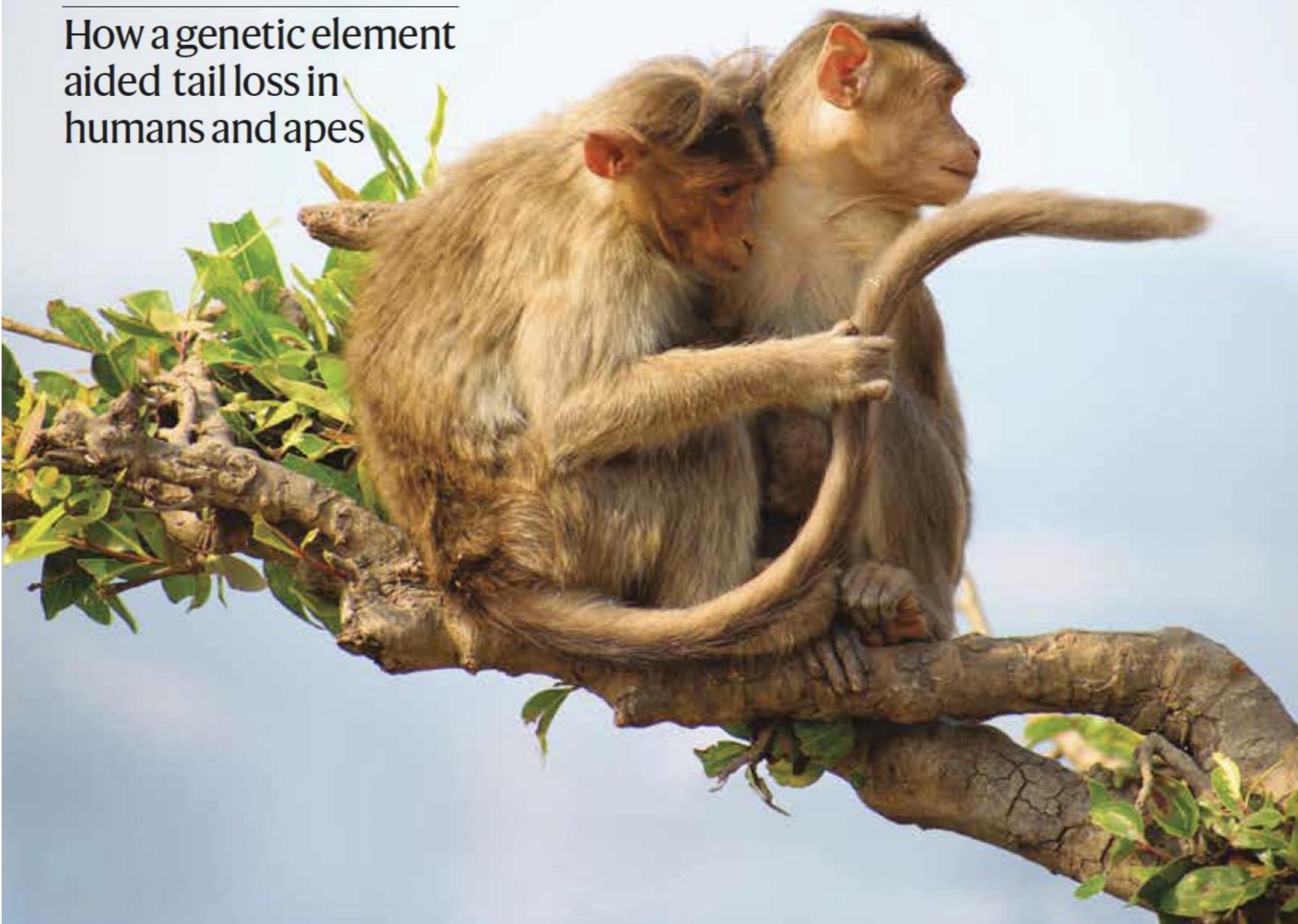
- *specific genes*
- *specific genomic regions*
- *first pass information on annotations (genes, repeats)*
- *expression across tissues*
- *conservation*
- *regulation*

The international journal of science / 29 February 2024

nature

TALE OF TAILS

How a genetic element
aided tail loss in
humans and apes



On the genetic basis of tail-loss evolution in humans and apes

<https://doi.org/10.1038/s41586-024-07095-8>

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Open access



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The loss of the tail is among the most notable anatomical changes to have occurred along the evolutionary lineage leading to humans and to the ‘anthropomorphous apes’^{1–3}, with a proposed role in contributing to human bipedalism^{4–6}. Yet, the genetic mechanism that facilitated tail-loss evolution in hominoids remains unknown. Here we present evidence that an individual insertion of an Alu element in the genome of the hominoid ancestor may have contributed to tail-loss evolution. We demonstrate that this Alu element—inserted into an intron of the *TBXT* gene^{7–9}—pairs with a neighbouring ancestral Alu element encoded in the reverse genomic orientation and leads to a hominoid-specific alternative splicing event. To study the effect of this splicing event, we generated multiple mouse models that express both full-length and exon-skipped isoforms of *Tbxt*, mimicking the expression pattern of its hominoid orthologue *TBXT*. Mice expressing both *Tbxt* isoforms exhibit a complete absence of the tail or a shortened tail depending on the relative abundance of *Tbxt* isoforms expressed at the embryonic tail bud. These results support the notion that the exon-skipped transcript is sufficient to induce a tail-loss phenotype. Moreover, mice expressing the exon-skipped *Tbxt* isoform develop neural tube defects, a condition that affects approximately 1 in 1,000 neonates in humans¹⁰. Thus, tail-loss evolution may have been associated with an adaptive cost of the potential for neural tube defects, which continue to affect human health today.

a personal story behind the science



<https://podcasts.apple.com/us/podcast/bo-xia-and-a-tale-of-tails/id1563415749?i=1000647438287>

Start with the Brachyury gene

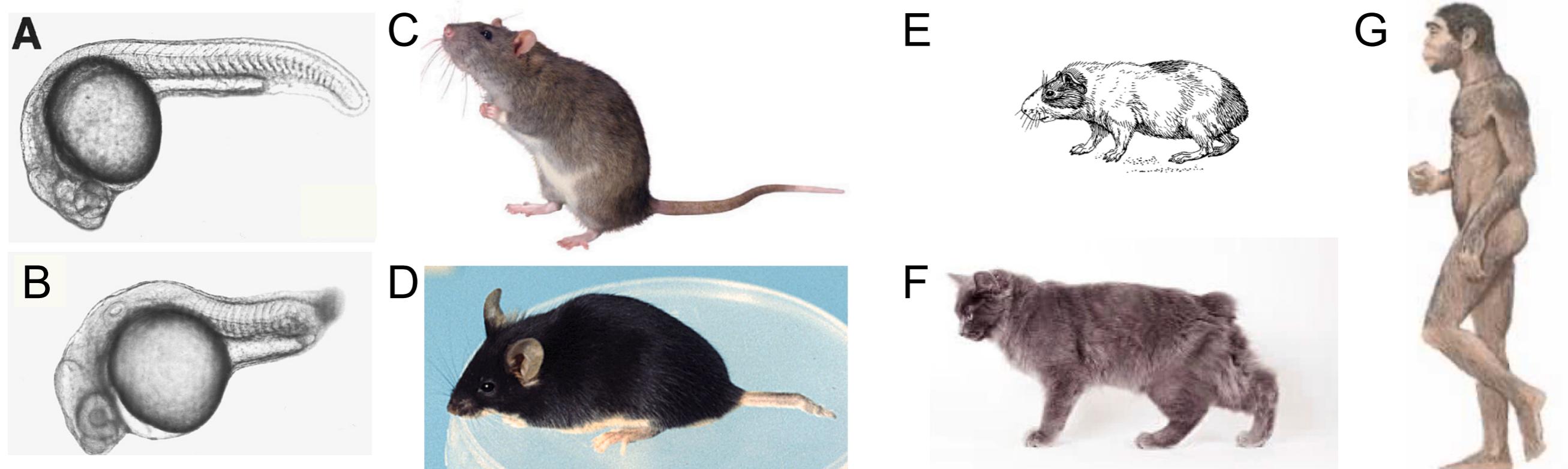


Fig. 3. The short-tailed and tailless animals. A - zebrafish, wild type; B - homozygotic *no tail* (Brachyury, T) mutant ([Halpern et al., 1997](#)); C - mice, wild type; D - mice, heterozygotic T mutant; E - guinea pig, wild type; cat, Manx, heterozygotic T mutant; G - hominid, wild type.

<https://ensembl.org>

Homo_sapiens - Ensembl gen x + https://useast.ensembl.org/Homo_sapiens/Info/Index New Chrome available

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Human (GRCh38.p14) ▾

Search Human (Homo sapiens)

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e.g. BRCA2 or 17:63992802-64038237 or rs699 or osteoarthritis

Genome assembly: GRCh38.p14 (GCA_000001405.29)

- More information and statistics
- Download DNA sequence (FASTA)
- Convert your data to GRCh38 coordinates
- Display your data in Ensembl

Other assemblies

GRCh37 Full Feb 2014 archive with BLAST, VEP and BioMart Go

Gene annotation

What can I find? Protein-coding and non-coding genes, splice variants, cDNA and protein sequences, non-coding RNAs.

- More about this genebuild
- Download FASTA files for genes, cDNAs, ncRNA, proteins
- Download GTF or GFF3 files for genes, cDNAs, ncRNA, proteins
- Update your old Ensembl IDs

Pax6 INS FOXP2 BRCA2 DMD ssh Example gene

View karyotype

Example region

Comparative genomics

What can I find? Homologues, gene trees, and whole genome alignments across multiple species.

- More about comparative analysis
- Download alignments (EMF)

Example gene tree

Regulation

What can I find? Regulatory features like enhancers and promoters, and regulatory activity including ATAC-seq and ChIP-seq tracks.

More about the Ensembl regulatory annotation

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ATCGAGCT ATCCAGCT ATCGAGAT Example variant

Example phenotype

Variation

What can I find? Short sequence variants and longer structural variants; disease and other phenotypes

- More about variation in Ensembl
- Download all variants (GVF)
- Variant Effect Predictor

Ve!P

I Agree

<https://epigenomegateway.wustl.edu>

WashU Epigenome Browser x +

epigenomegateway.wustl.edu/browser/ Finish update :

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WashU Epigenome Browser Documentation Switch to the 'old' browser

CHOOSE A GENOME LOAD A SESSION

Please select a genome

 Human
hg19
hg38
t2t-chm13-v1.1
t2t-chm13-v2.0

 Chimp
panTro6
panTro5
panTro4

 Gorilla
gorGor4
gorGor3

 Gibbon
nomLeu3

 Baboon
papAnu2

 Rhesus
rheMac10
rheMac8
rheMac3
rheMac2

 Crab-Eating Macaque

 Marmoset

 Cow