

Uniformly collected and processed functional genomics assays provide a way to interpret non-coding variants in terms of tissue of action

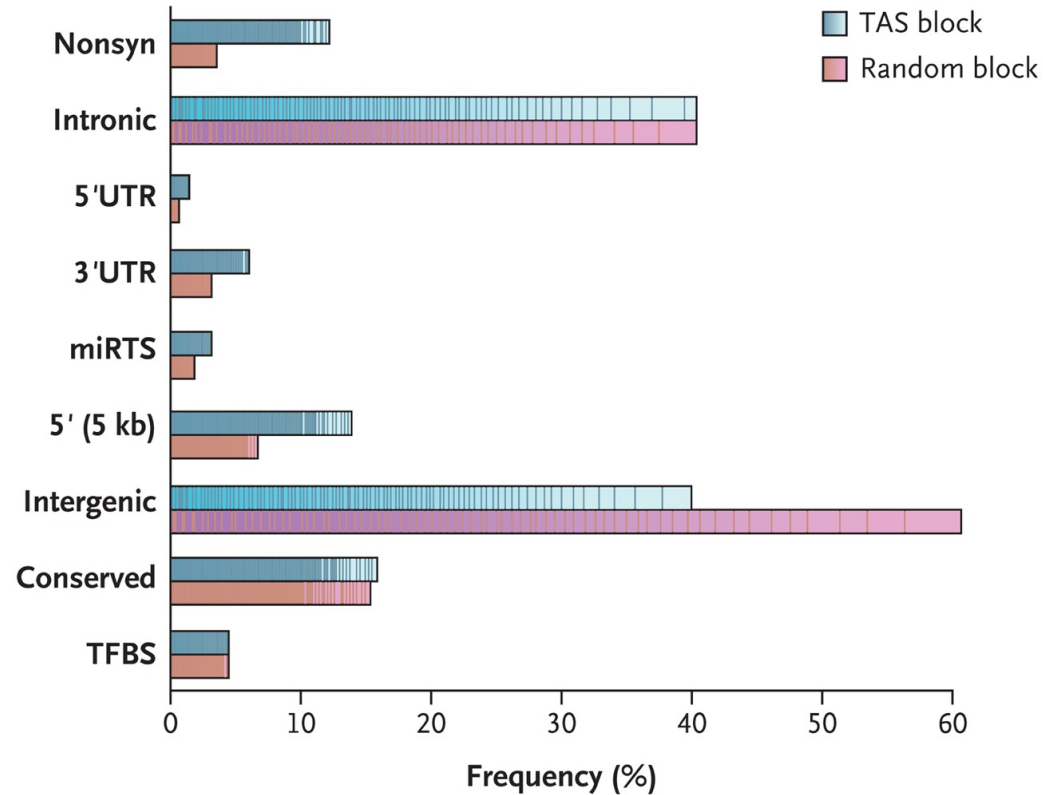
Fabio Navarro – Yale University
ASHG 2018

Agenda



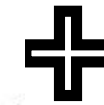
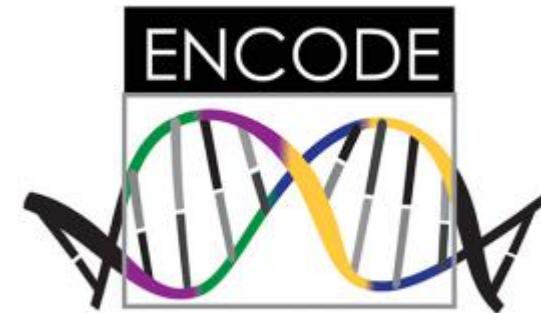
- What is EN-TEEx?
- The EN-TEEx resource
- Our first findings

EN-TE_x is the follow up of GT_{EX} and ENCODE



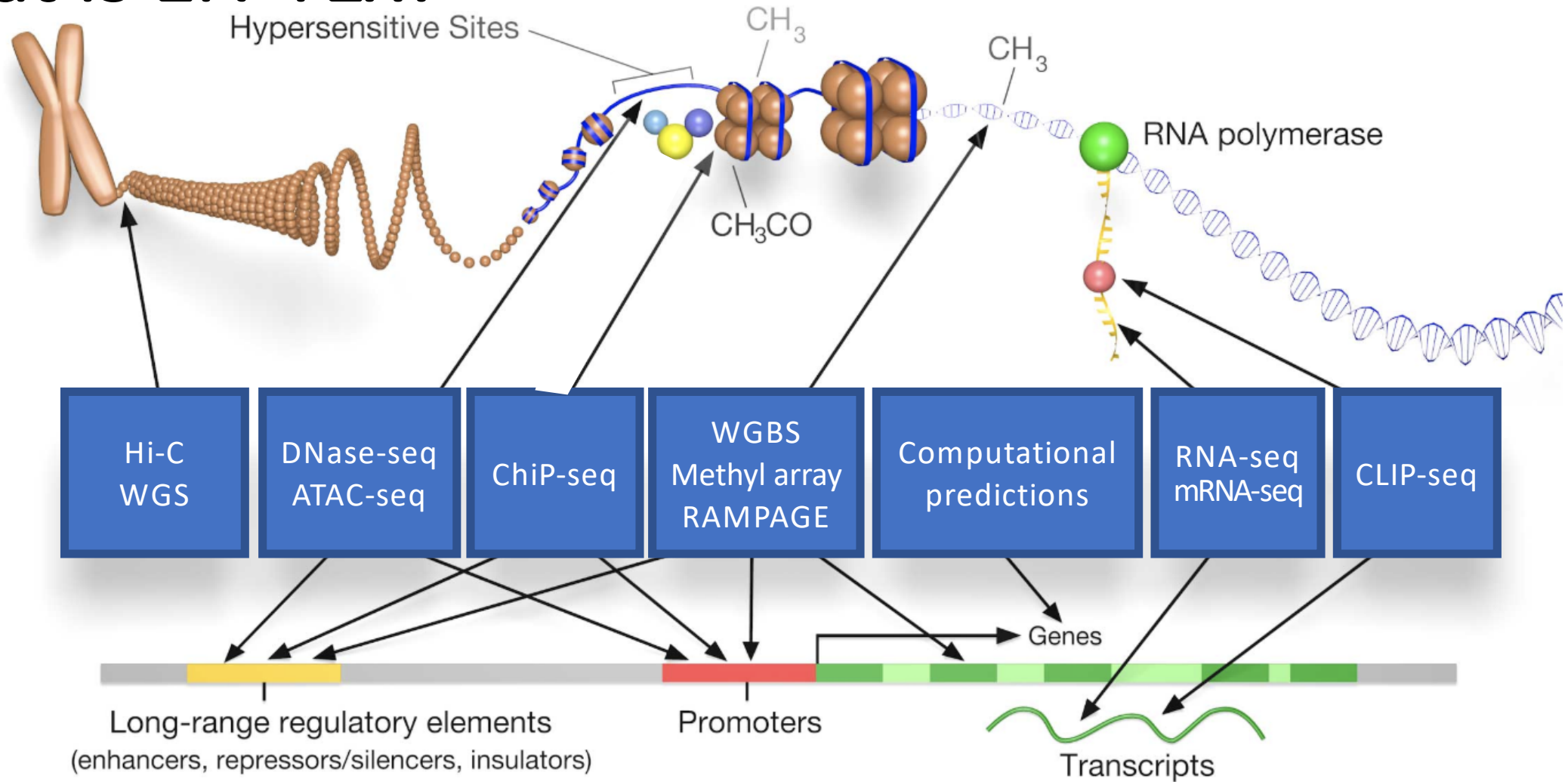
Encyclopedia of DNA Elements

Genotype-Tissue Expression

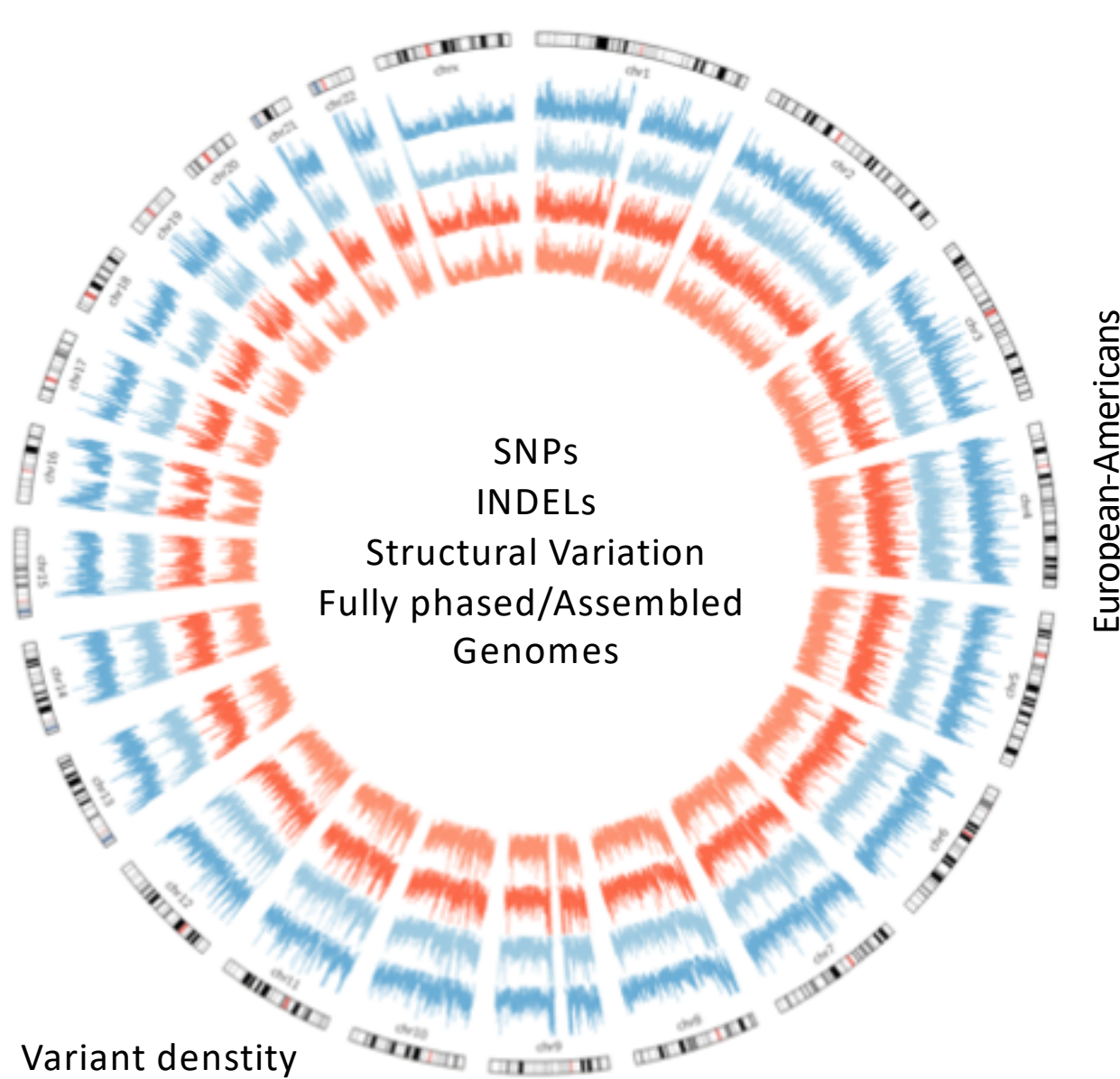


Genomewide association studies and assessment of the risk of disease. (2010). Genomewide association studies and assessment of the risk of disease., 363(2), 166–176. <http://doi.org/10.1056/NEJMra0905980>

What is EN-TE_x?



What is EN-TE_x?



ENC001
37 years old



ENC002
54 years old



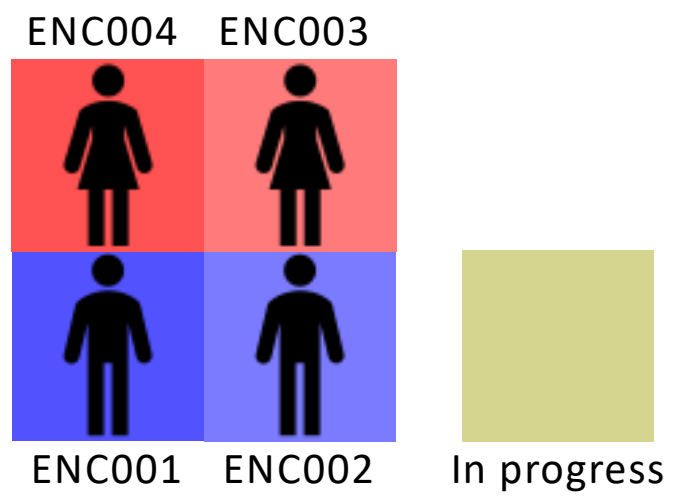
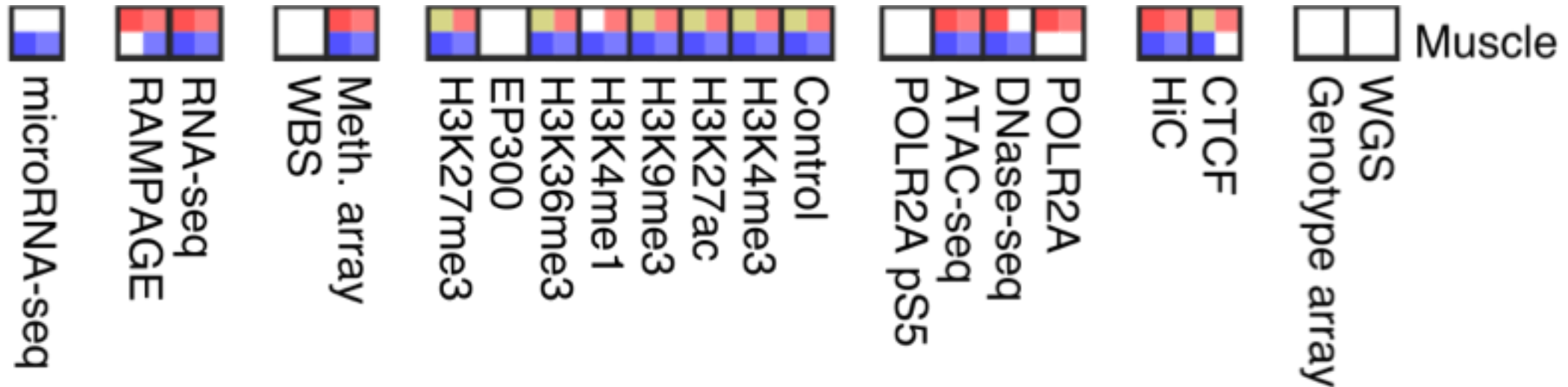
ENC003
53 years old



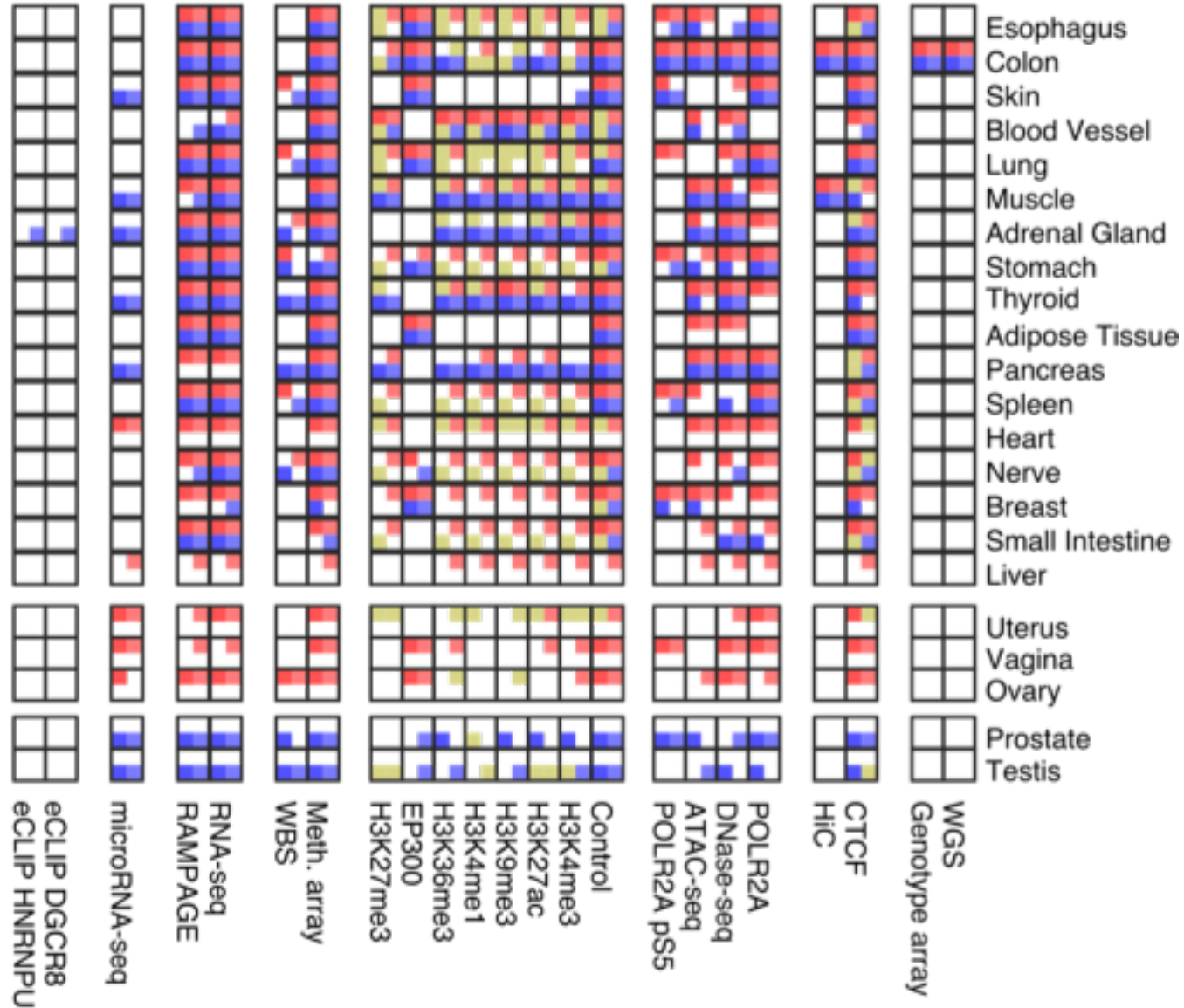
ENC004
51 years old

- Muscles**
 - Gastrocnemius medialis
 - Stomach
 - Transverse colon
 - Peyer's patch
 - Sigmoid colon
- Alimentary**
 - Right lobe of liver
 - Body of pancreas
 - Esophagus squamous epithelium
 - Gastroesophageal sphincter
 - Esophagus muscularis mucosa
- Genital**
 - Testis
 - Prostate gland
 - Uterus
 - Vagina
 - Ovary
- Endocrine**
 - Thyroid gland
 - Adrenal gland
 - Spleen
- Cardio**
 - Right atrium auricular region
 - Heart left ventricle
 - Thoracic aorta
 - Ascending aorta
- Nervous**
 - Omental fat pad
 - Subcutaneous adipose tissue
 - Tibial nerve
- Integumentary**
 - Suprapubic skin
 - Lower leg skin
 - Breast epithelium
- Respiratory**
 - Upper lobe of left lung

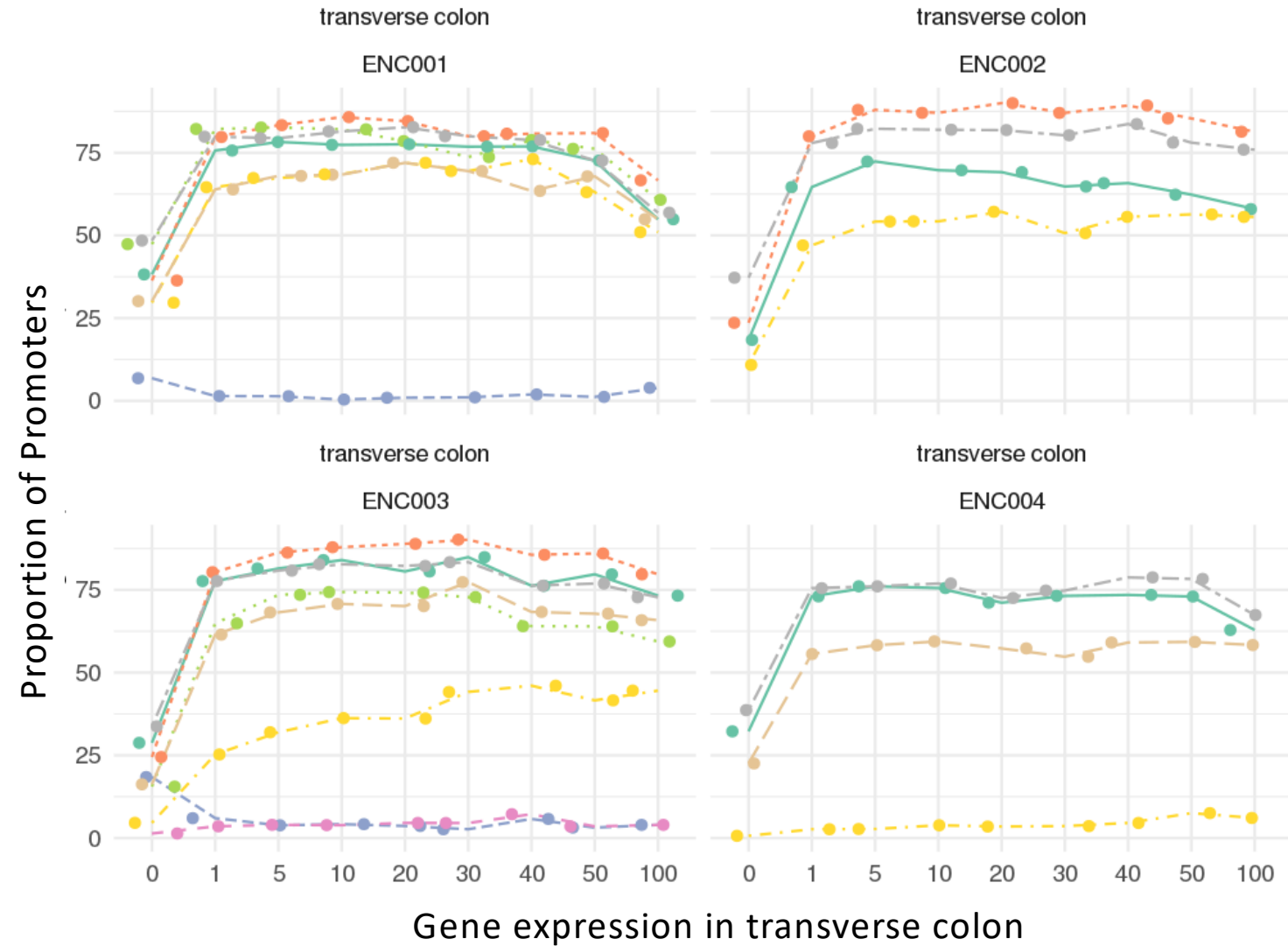
The EN-TE_x dataset



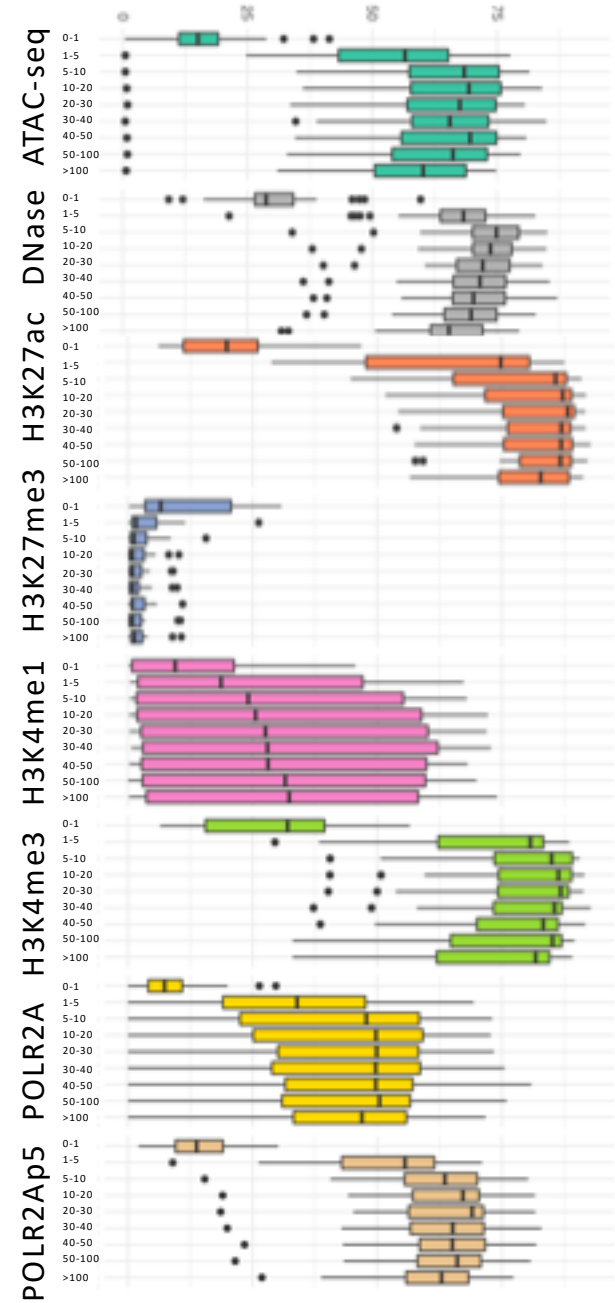
The EN-TEEx dataset



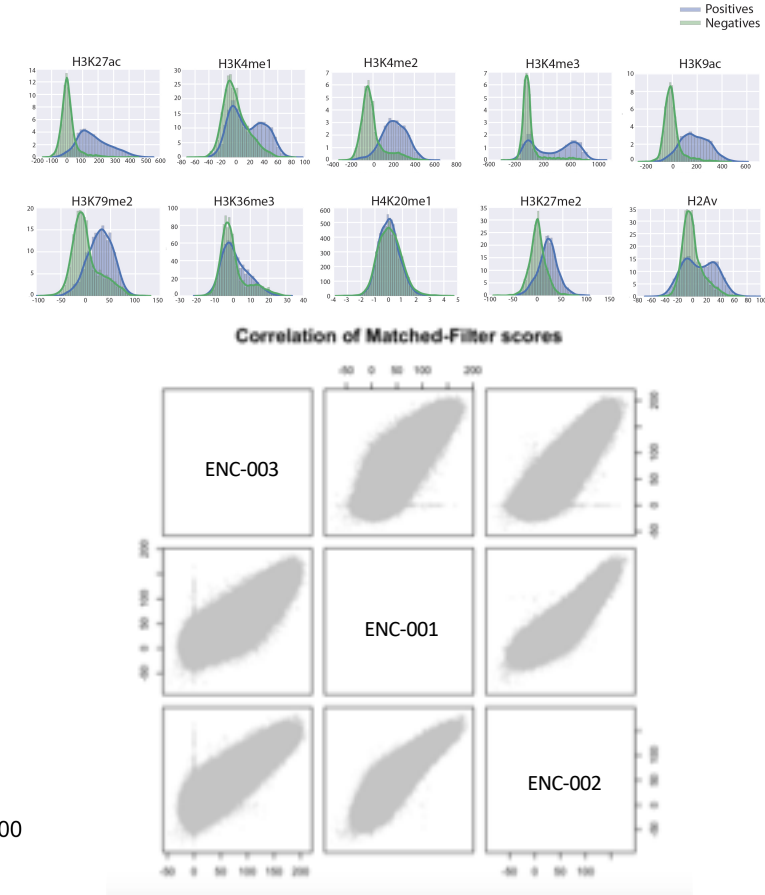
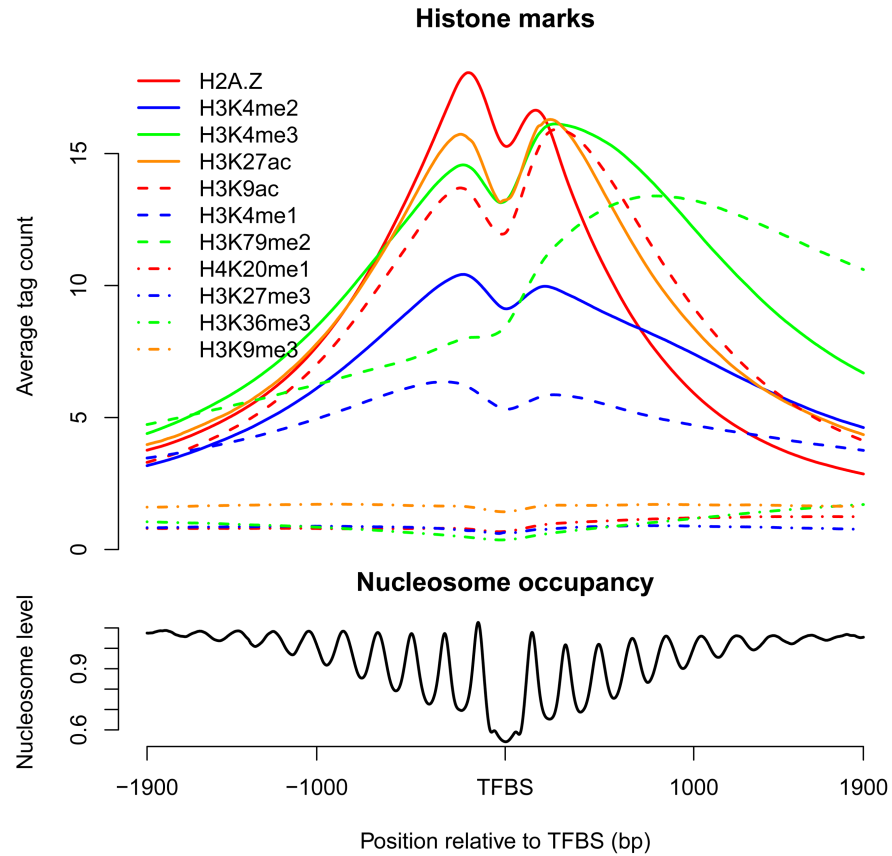
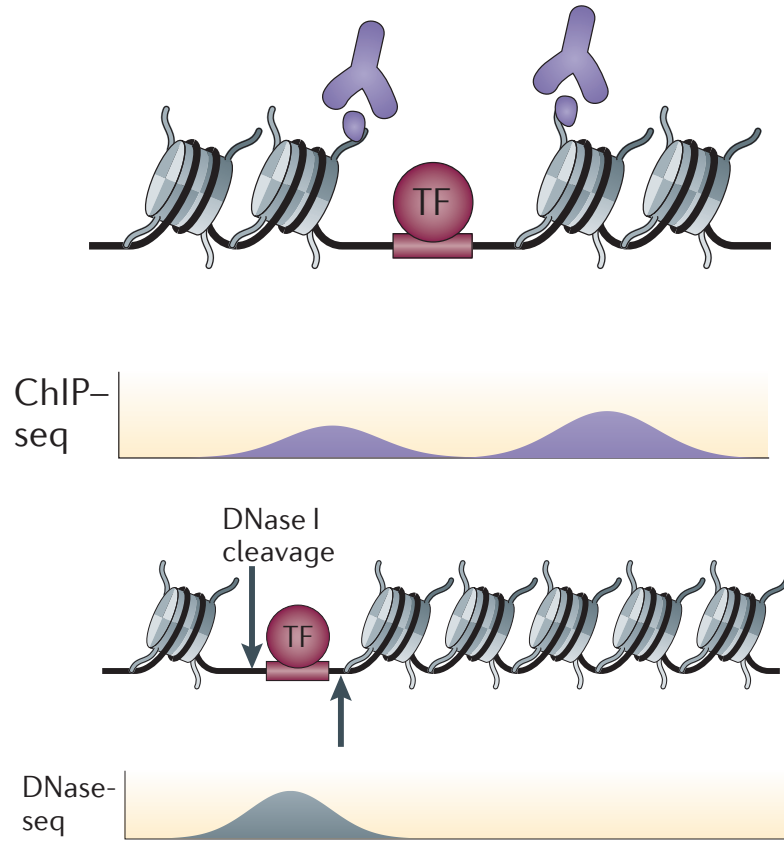
Variation across tissues



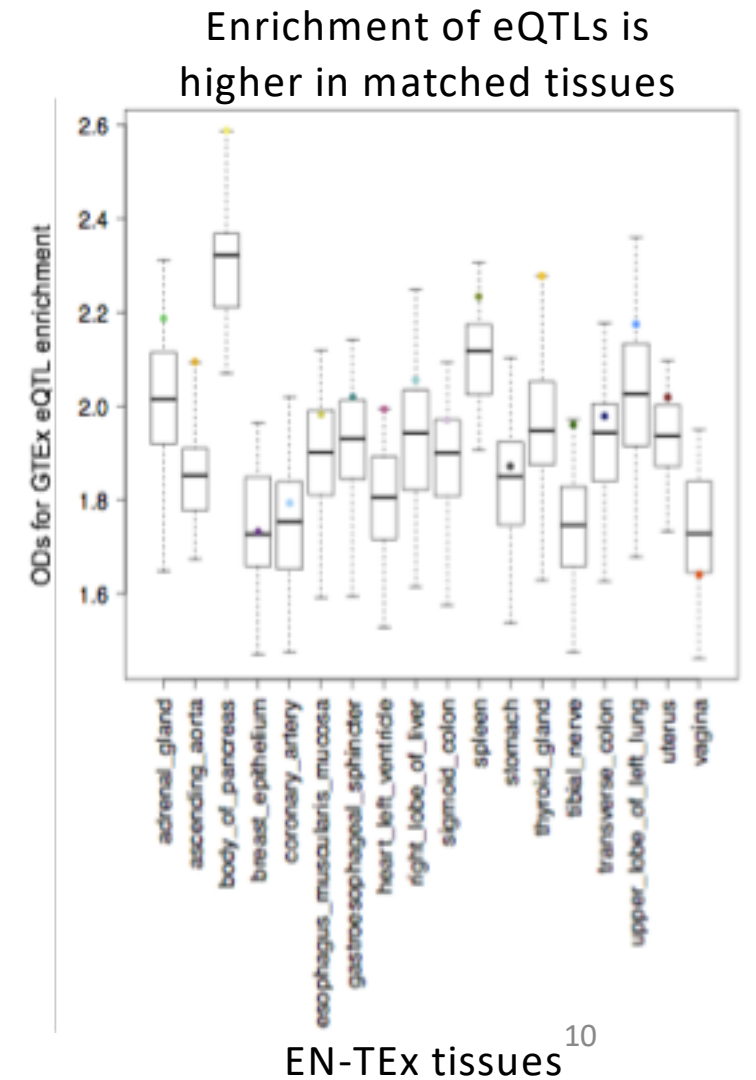
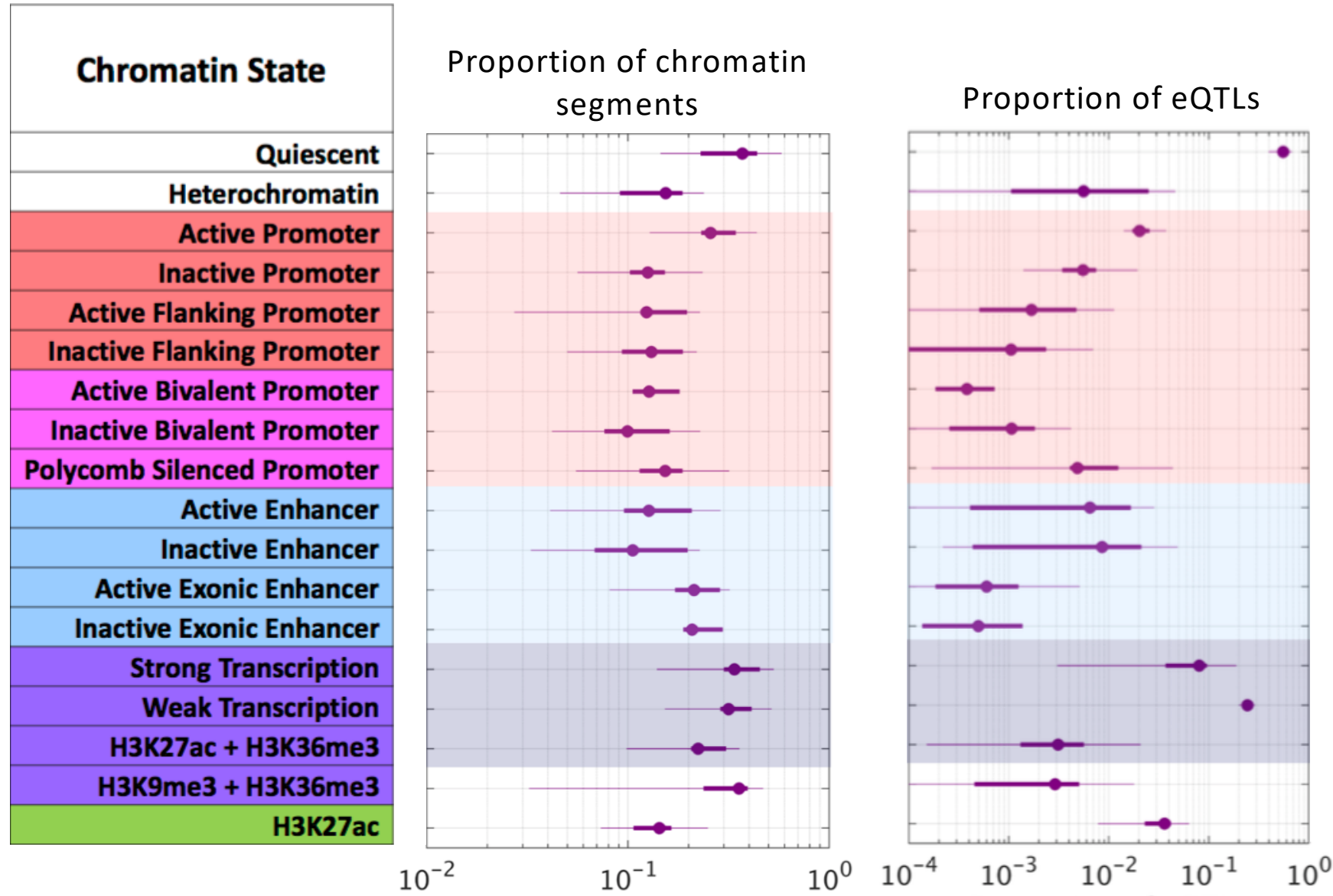
Proportion of Promoters
Across all EN-TEx samples



Integrative methods for calling regulatory regions



eQTLs are enriched in tissue regulatory regions



Acknowledgements

The background features a large, light blue DNA double helix structure that spans across the top and middle of the page. Below it, there is a stylized illustration of a chromosome, showing a blue thread with yellow bands representing chromatin loops. The overall aesthetic is clean and scientific.

Analysis and Data Generation Labs:

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ENCODE Consortium

NHGRI

GENCODE

GTEX consortium

Roadmap Epigenomics project