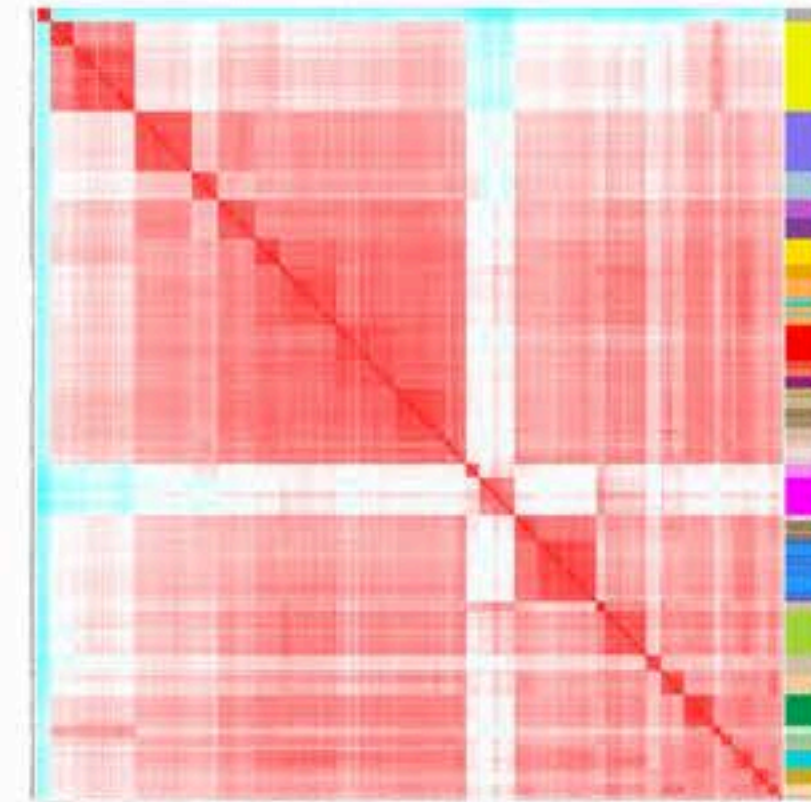
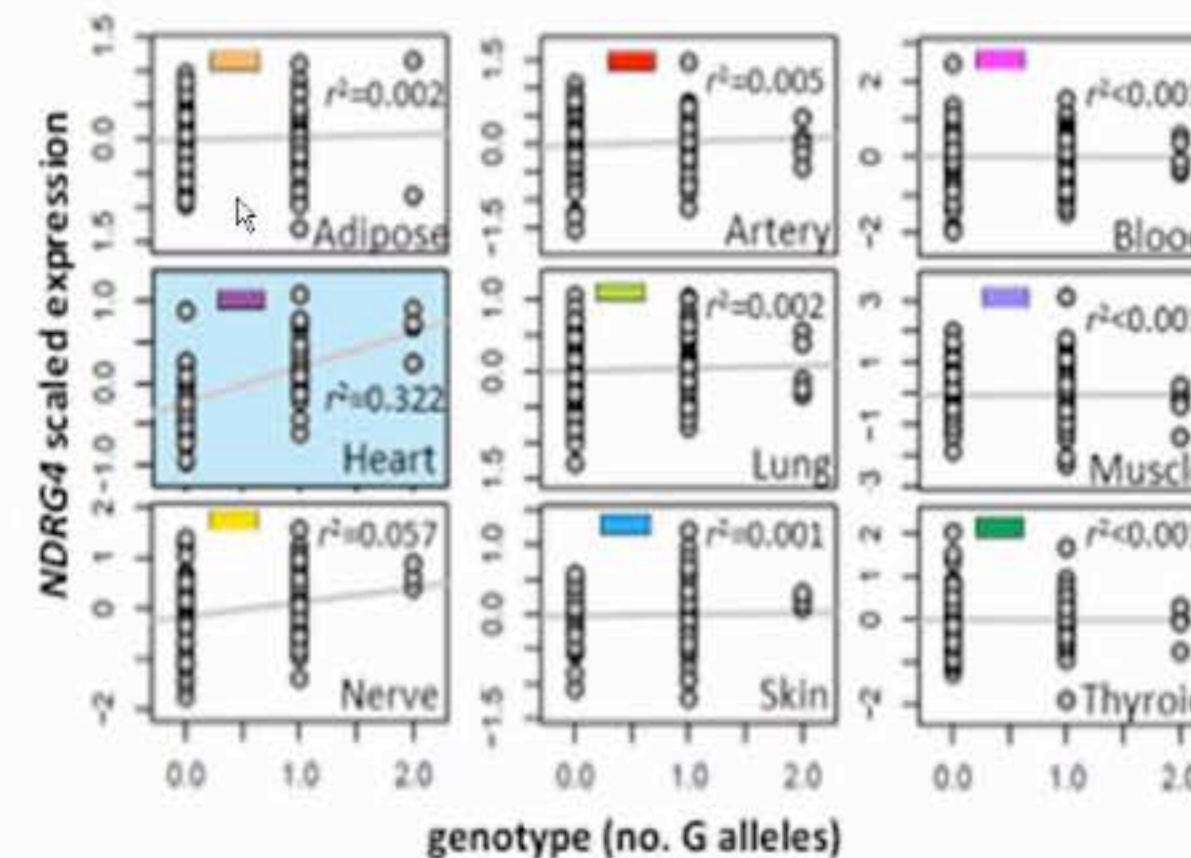


# GTEx Project Goals

Characterize the regulatory architecture of human genome by understanding the role of genetic variation on gene expression variation across a wide range of non-diseased human tissues.



- Create an atlas of human tissue gene expression
- Comprehensive resource database of *cis*- and *trans*-eQTLs to enable studies of role of genetic variation on gene regulation across tissues; interpret GWAS studies





# Scope - Primary Data Types

## ❑ 960 post-mortem donors

- up to 53 tissues/donor (45 main sites)

## ❑ DNA sequence each donor

- Whole genome (WGS) and whole exome (WES)

## ❑ RNA-sequencing on >25,000 tissues (~25 average/donor)

## ❑ Associated clinical and histopathological information

## ❑ Enhanced GTEx (eGTEx)

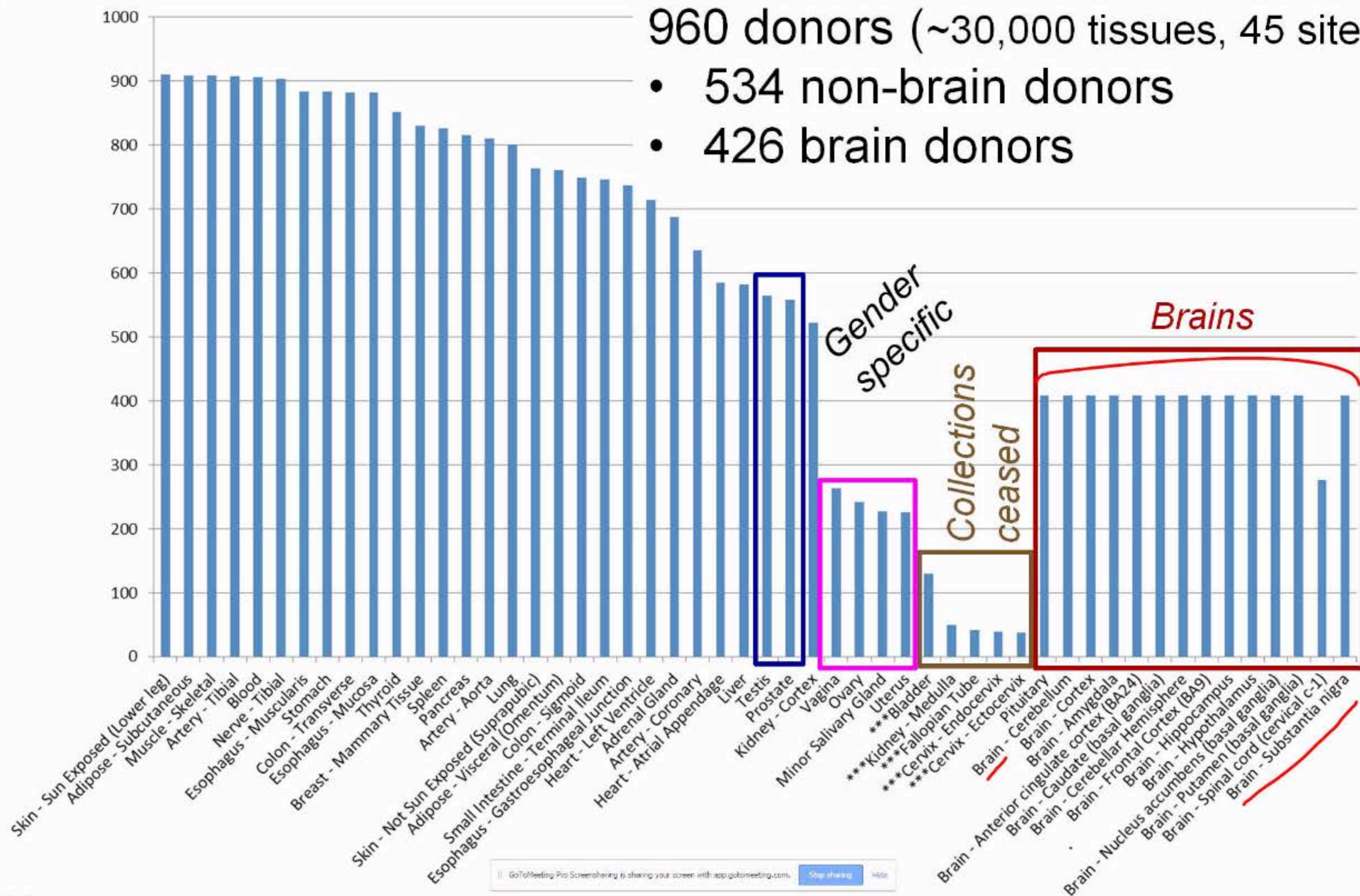
- Protein quantifications (x 2)
- Methylation (x2)
- Histone modifications
- Dnase-seq
- mmPCR-seq (deep ASE)
- Somatic DNA seq (deep exome seq)
- Analysis of telomere structure



# Tissues collected

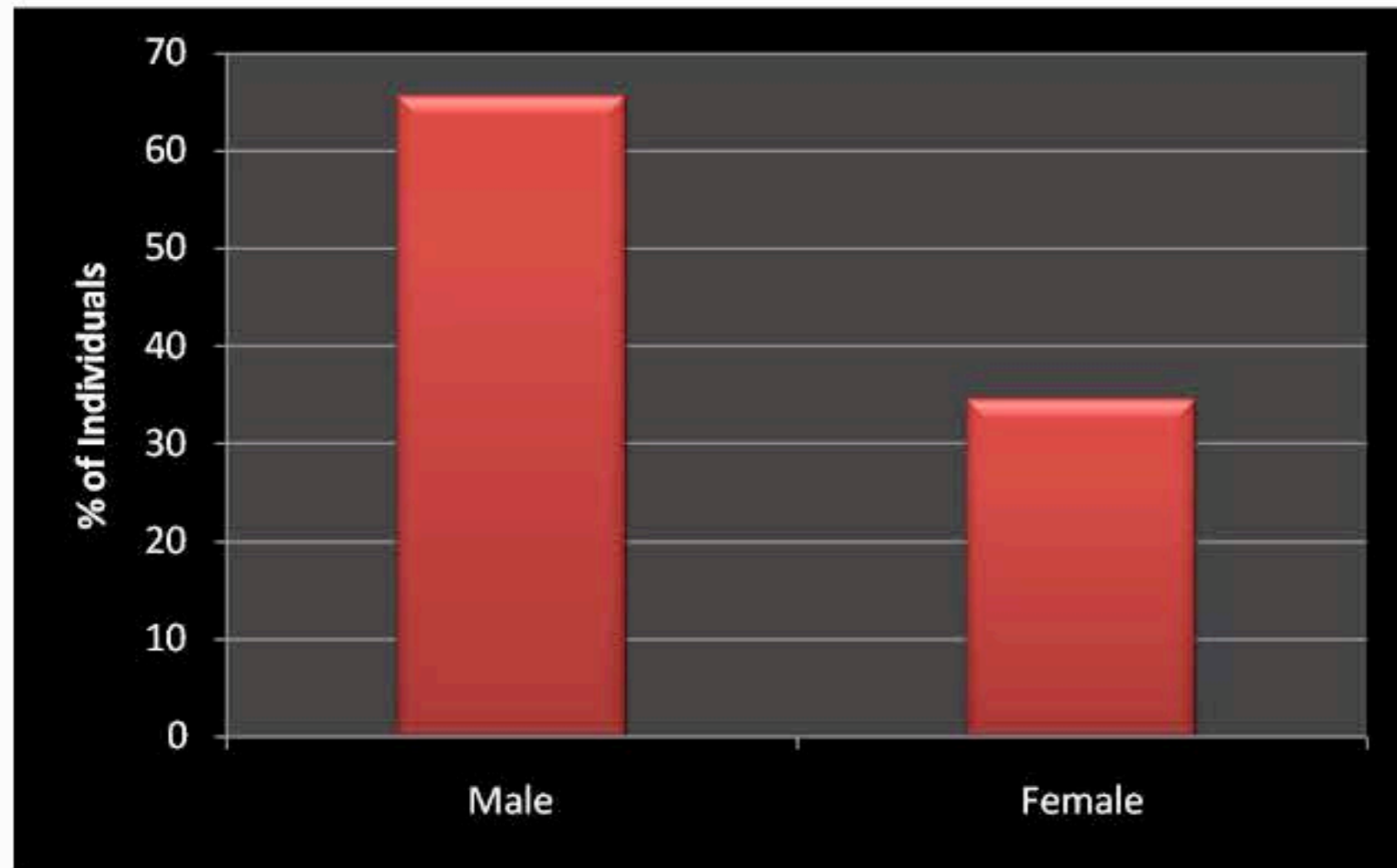
960 donors (~30,000 tissues, 45 sites)

- 534 non-brain donors
- 426 brain donors

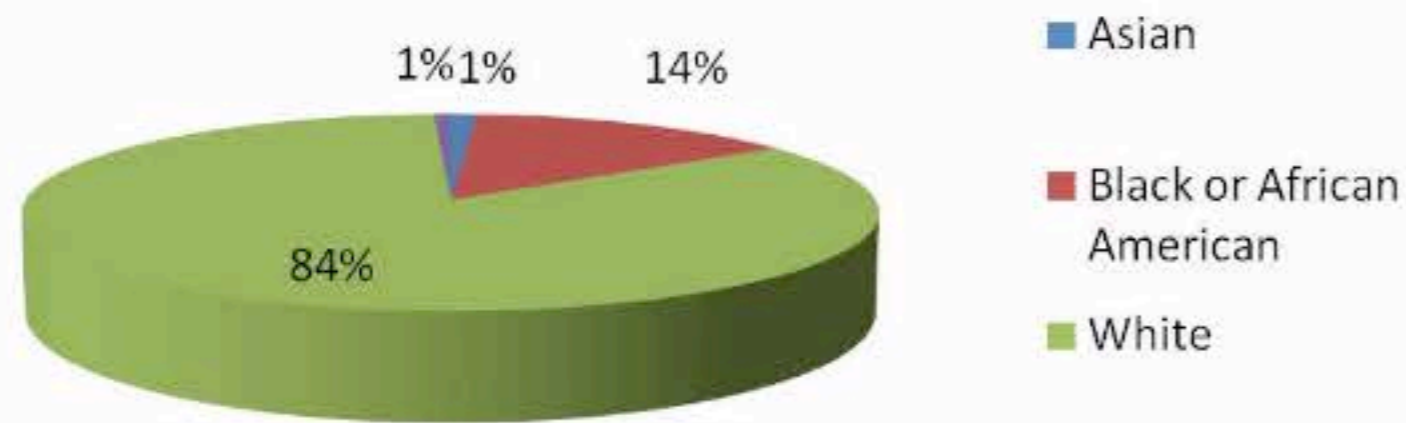




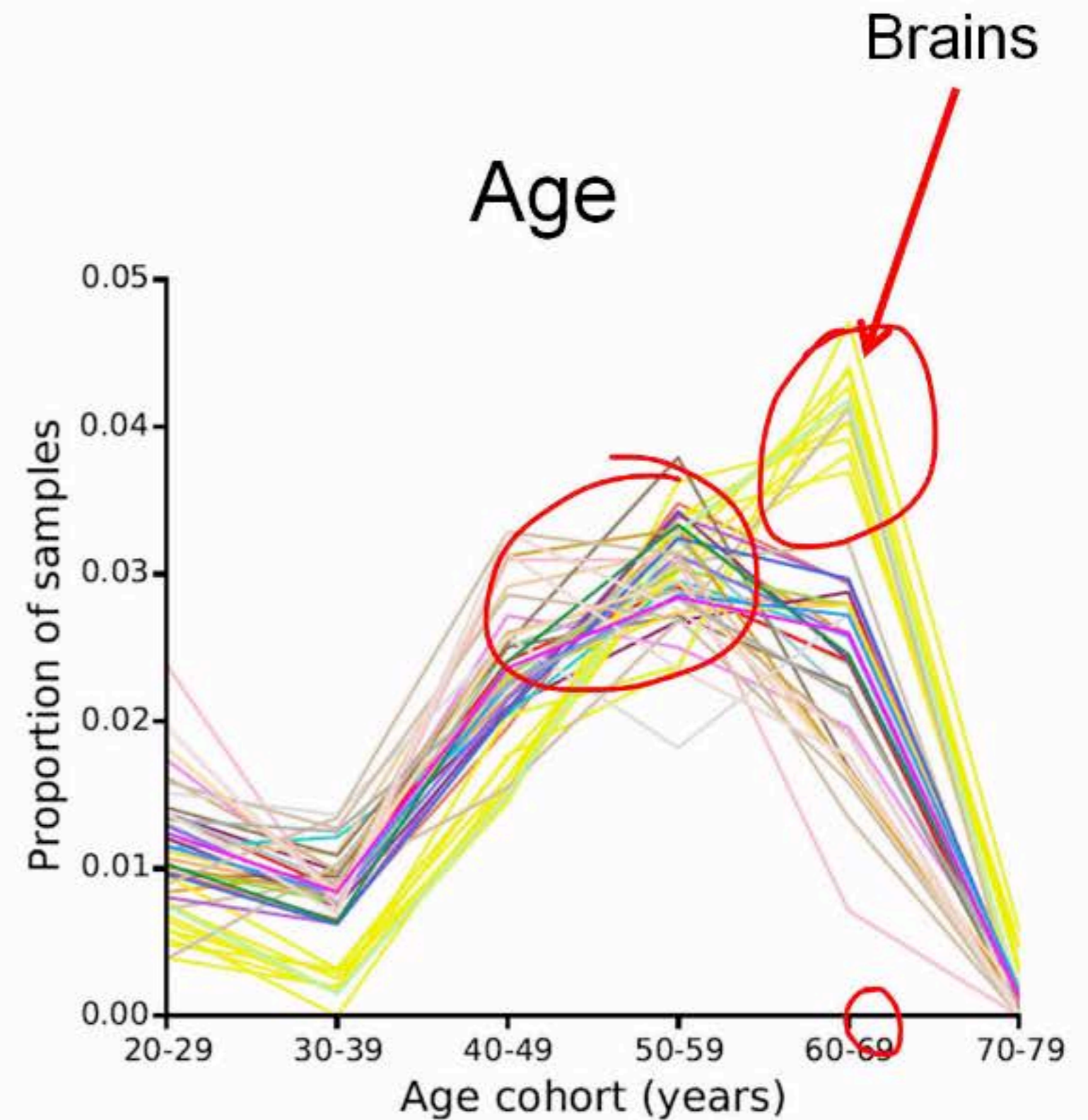
# Donor Demographics



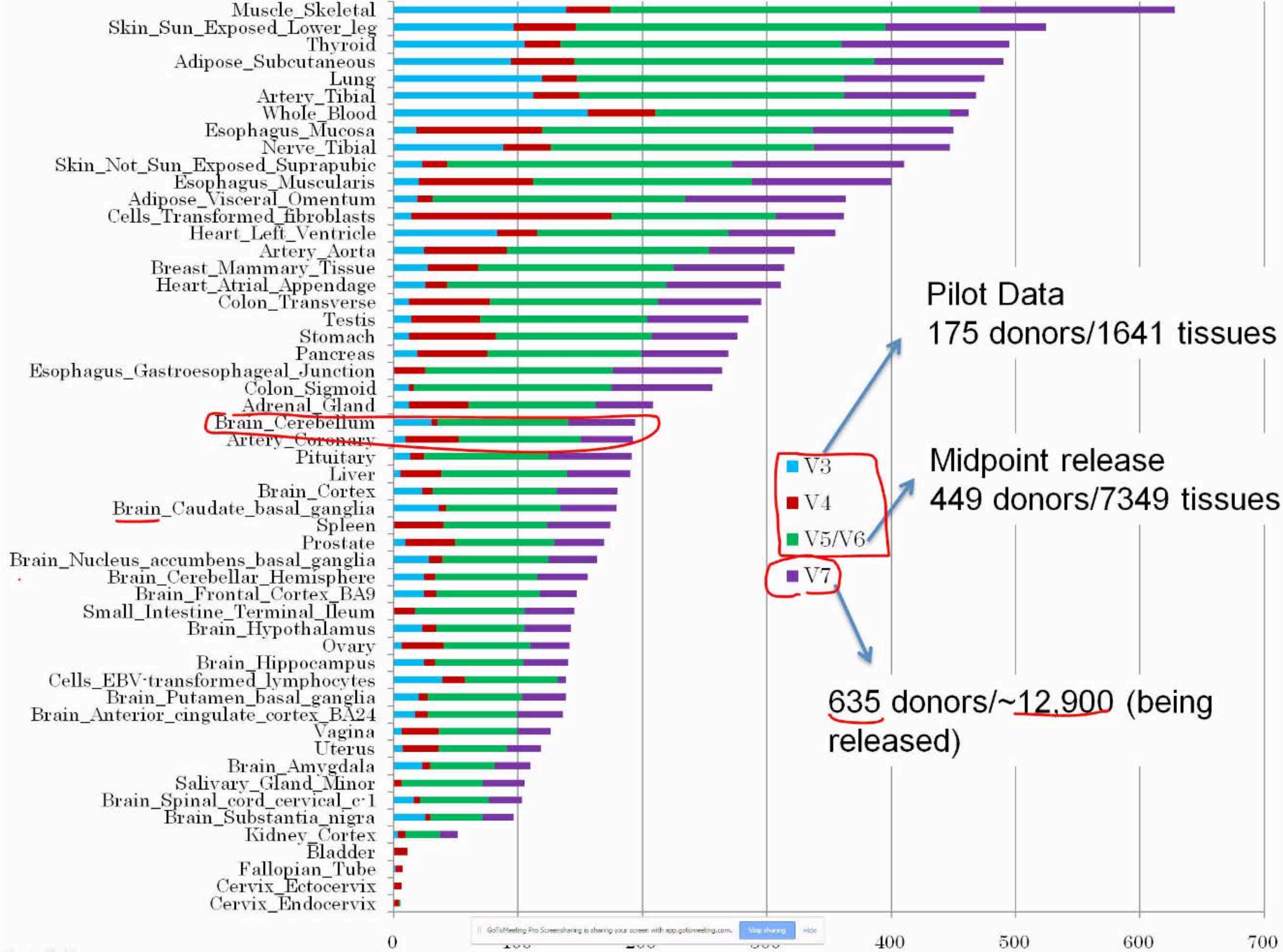
Gender



Race

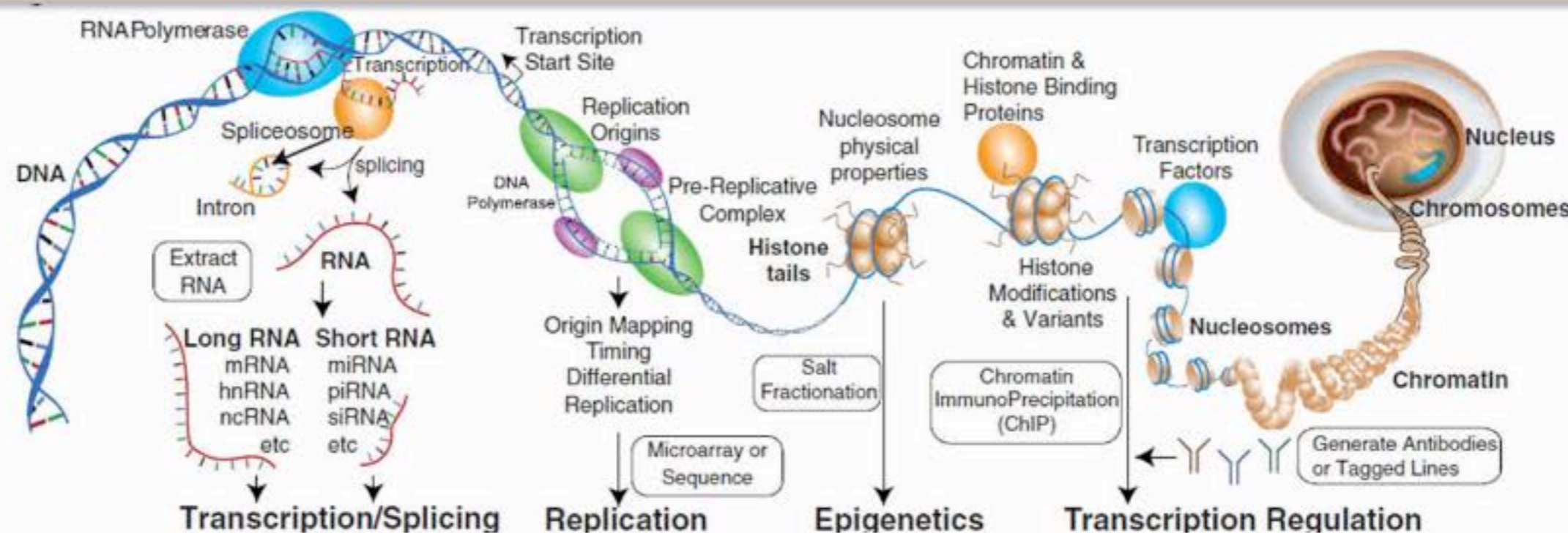








# eGTEx – enhancing GTEx



## Enhancing GTEx with molecular analyses of stored biospecimens (U01) RFA-RM-12-009

PI Name	Institution Name	Title
AKEY, JOSHUA MICHAEL	UNIVERSITY OF WASHINGTON	<a href="#">Comprehensively assessing human somatic variability and its influence on gene exp</a>
FEINBERG, ANDREW P	JOHNS HOPKINS UNIVERSITY	<a href="#">Strategic mapping of tissue and population methylation for mental health research</a>
KELLIS, MANOLIS	MASSACHUSETTS INSTITUTE OF TECHNOLOGY	<a href="#">Epigenomic variation atlas across human tissues and individuals in GTEx</a>
LI, JIN BILLY	STANFORD UNIVERSITY	<a href="#">HIGH RESOLUTION ALLELE SPECIFIC EXPRESSION ASSAYS</a>
PIERCE, BRANDON LEE	THE UNIVERSITY OF CHICAGO	<a href="#">Telomere length and chromosomal instability across various tissue types</a>
SNYDER, MICHAEL P TANG, HUA	STANFORD UNIVERSITY	<a href="#">Genotype-Tissue-Protein: proteomic variation and quantitative trait loci (pQTL)</a>
STAMATOYANNOPOULOS, JOHN A	UNIVERSITY OF WASHINGTON	<a href="#">High-resolution mapping of DNaseI hypersensitive regulatory DNA in GTEx samples</a>
STRANGER, BARBARA E	UNIVERSITY OF CHICAGO	<a href="#">The genetic basis of cross-tissue protein expression variability in humans</a>