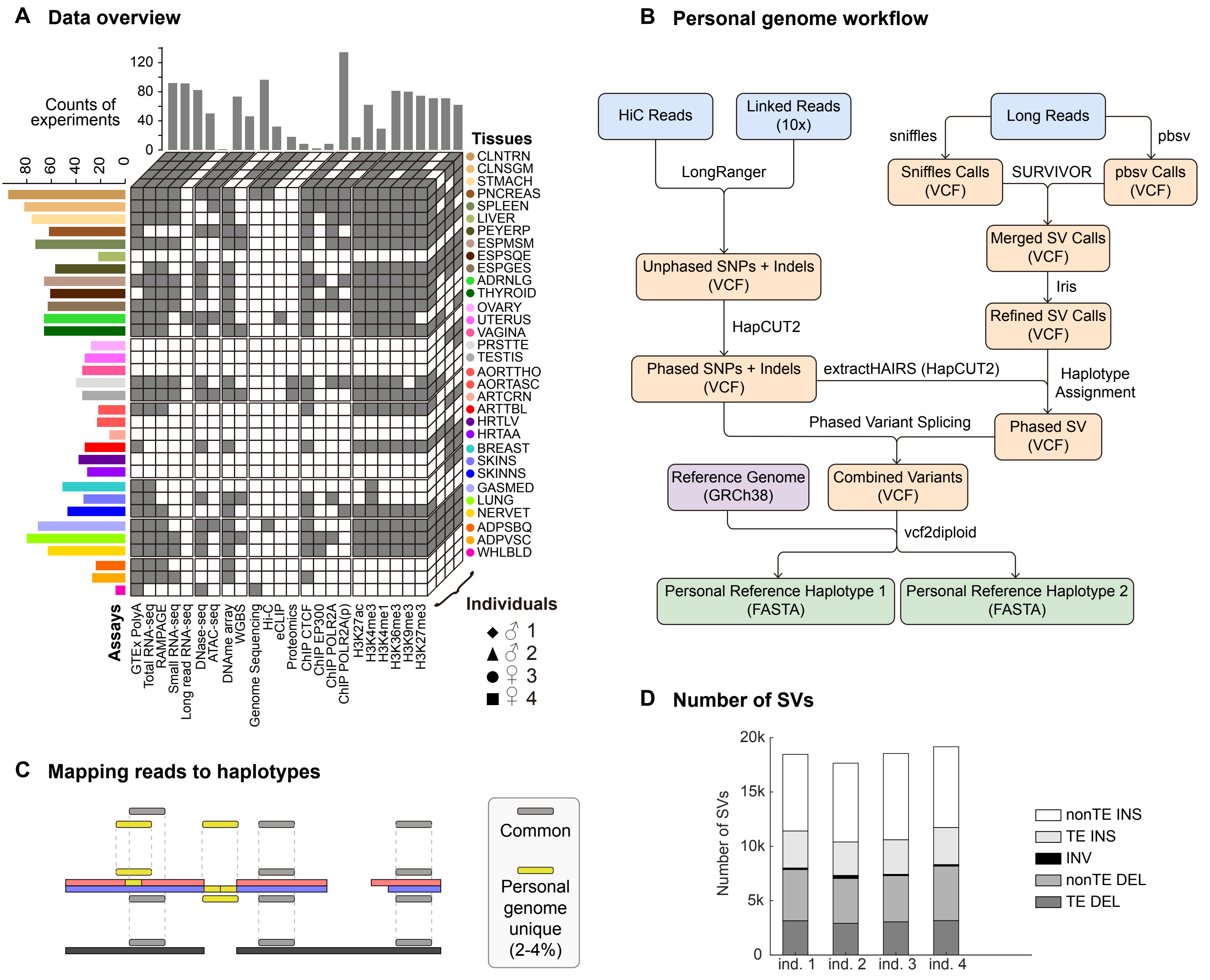
A Data overview
Counts of experiments
120
80
40
0
80
60
40
20
0
Tissues
CLNTRN
CLNSGM
STMACH
PNCREAS
SPLEEN
LIVER
PEYERP
ESPMSM
ESPSQE
ESPGES
ADRNLG
THYROID
OVARY
UTERUS
VAGINA
PRSTTE
TESTIS
AORTTHO
AORTASC
ARTCRN
ARTTBL
HRTLV
HRTAA
BREAST
SKINS
SKINNS
GASMED
LUNG
NERVET
ADPSBQ
ADPVSC
WHLBLD
Assays
GTEx PolyA
Total RNA-seq
RAMPAGE
Small RNA-seq
Long read RNA-seq
DNase-seq
ATAC-seq
DNAme array
WGBS
Genome Sequencing
Hi-C
eCLIP
Proteomics
ChIP CTCF
ChIP EP300
ChIP POLR2A
ChIP POLR2A(p)
H3K27ac
H3K4me3
H3K4me1
H3K36me3
H3K9me3
H3K27me3
Individuals
◆ ♂ 1
▲ ♂ 2
● ♀ 3
■ ♀ 4
B Personal genome workflow
HiC Reads
Linked Reads (10x)
Long Reads
sniffles
pbsv
LongRanger
Sniffles Calls (VCF)
SURVIVOR
pbsv Calls (VCF)
Merged SV Calls (VCF)
Iris
Refined SV Calls (VCF)
Unphased SNPs + Indels (VCF)
HapCUT2
Phased SNPs + Indels (VCF)
extractHAIRS (HapCUT2)
Haplotype Assignment
Phased SV (VCF)
Phased Variant Splicing
Reference Genome (GRCh38)
Combined Variants (VCF)
vcf2diploid
Personal Reference Haplotype 1 (FASTA)
Personal Reference Haplotype 2 (FASTA)
C Mapping reads to haplotypes
Common
Personal genome unique (2-4%)
D Number of SVs
Number of SVs
20k
15k
10k
5k
0
ind. 1
ind. 2
ind. 3
ind. 4
nonTE INS
TE INS
INV
nonTE DEL
TE DEL