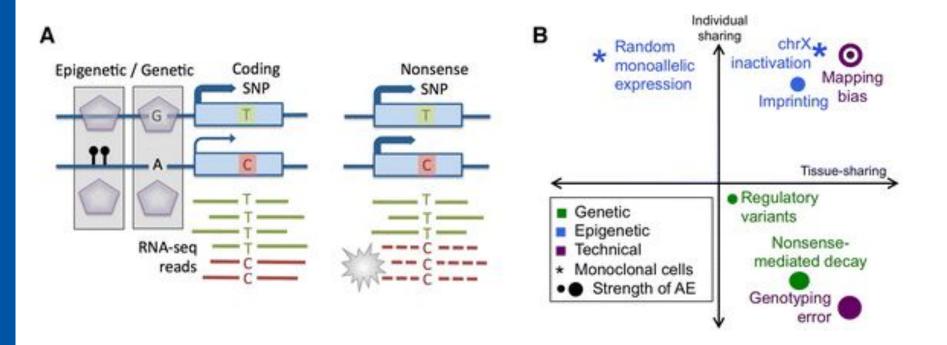
# Identifying Allele-Specific Expression (ASE) by bulk RNA sequencing

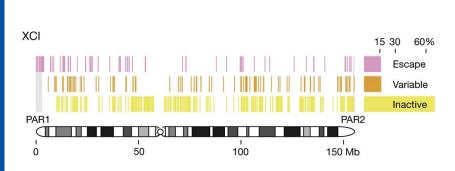
24 July 2023
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### Allele-Specific Expression (ASE)



Allelic expression and its sources. **a** Schematic illustration of AE. **b** Biological sources of AE, with the x-axis denoting the approximate sharing of AE across tissues of an individual, and the y-axis having the estimated sharing of AE signal in one tissue across different individuals [5, 8, 12, 13, 15]. *SNP* single-nucleotide polymorphism

### Landscape of X chromosome inactivation across human tissues



### Study samples



Random XCI

XCI in population/GTEx: Multi-tissue RNA-seq 29 tissue types 449 individuals 681 genes assessed



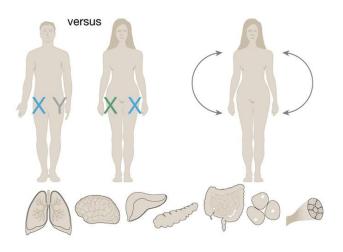
Fully skewed XCI

XCI within an individual/GTEx: WGS + RNA-seq 16 tissue types 1 female individual 186 genes assessed



Single active X chromosome

XCI in single cells: WES/WGS + scRNA-seq 940 single cells 2 cell types 4 female individuals 165 genes assessed



### Approach

Sex-biased expression in GTEx population samples

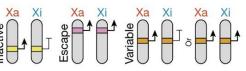
Allelic expression within a GTEx individual with highly skewed XCI Allelic expression in chrX utilizing phasing

### Genic XCI status

Tissue heterogeneity

Cellular heterogeneity

Impacts of incomplete XCI



X-chromosomal genes subject to XCI (inactive), escaping from XCI (escape), and variably escaping from XCI (variable)



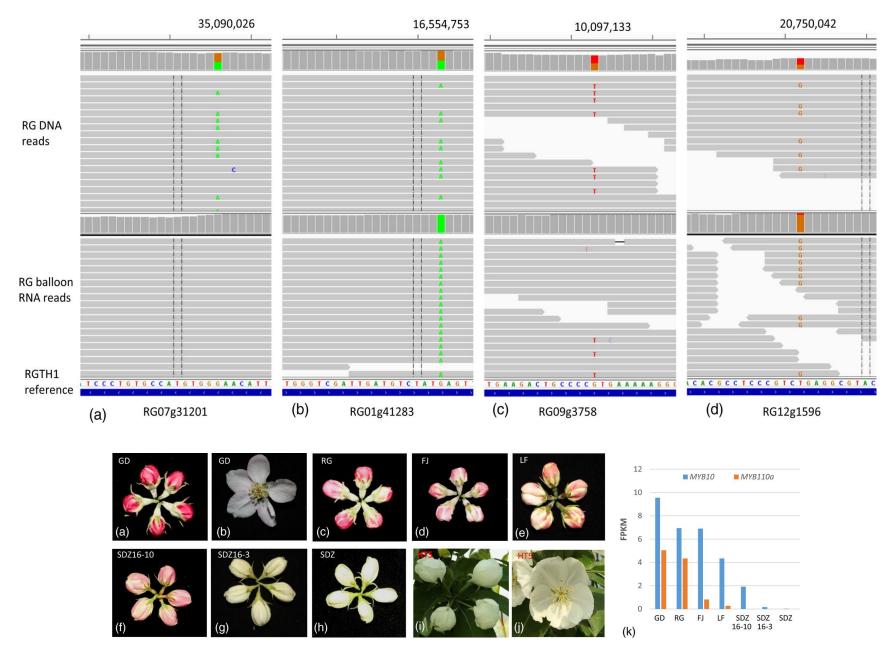
Active and inactive X chromosomes within a cell



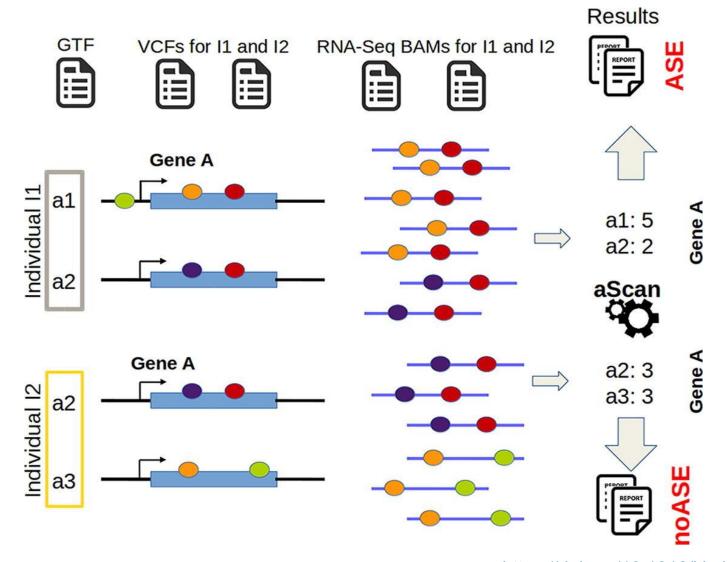
Cells with different X chromosomes

designated for inactivation

Transposon insertions regulate genome-wide allele-specific expression and underpin flower colour variations in apple (Malus spp.)



### aScan: A Novel Method for the Study of Allele Specific Expression in Single Individuals



## ASEP: Gene-based detection of allele-specific expression across individuals in a population by RNA sequencing

