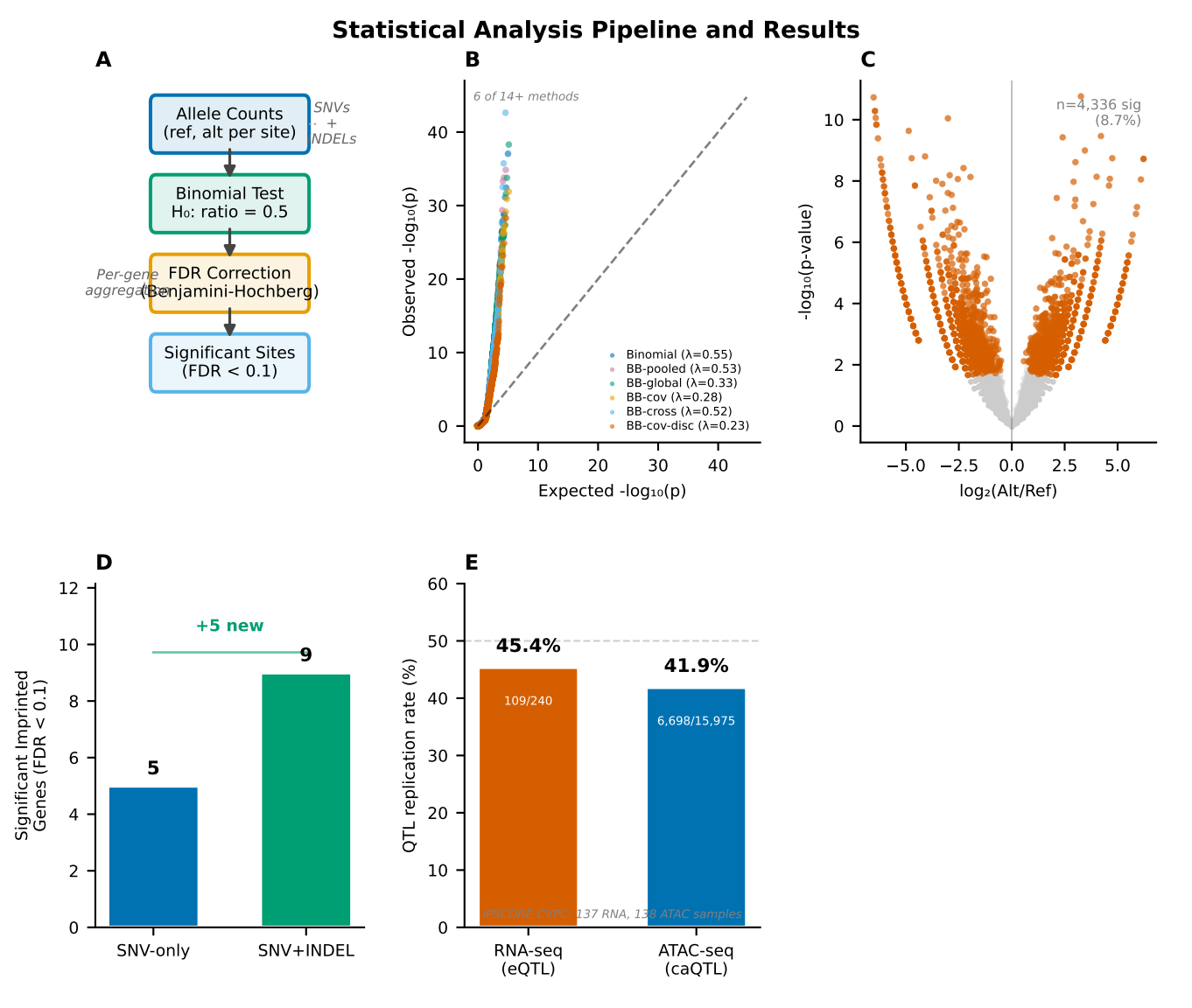
Statistical Analysis Pipeline and Results
A
Allele Counts
(ref, alt per site)
SNVs
+
INDELs
Binomial Test
H₀: ratio = 0.5
Per-gene
aggregation
FDR Correction
(Benjamini-Hochberg)
Significant Sites
(FDR < 0.1)
B
6 of 14+ methods
Observed -log₁₀(p)
Expected -log₁₀(p)
Binomial (λ=0.55)
BB-pooled (λ=0.53)
BB-global (λ=0.33)
BB-cov (λ=0.28)
BB-cross (λ=0.52)
BB-cov-disc (λ=0.23)
C
n=4,336 sig
(8.7%)
-log₁₀(p-value)
log₂(Alt/Ref)
D
Significant Imprinted
Genes (FDR < 0.1)
+5 new
5
9
SNV-only
SNV+INDEL
E
QTL replication rate (%)
45.4%
109/240
41.9%
6,698/15,975
iPSCORE CVPC: 137 RNA, 138 ATAC samples
RNA-seq
(eQTL)
ATAC-seq
(caQTL)