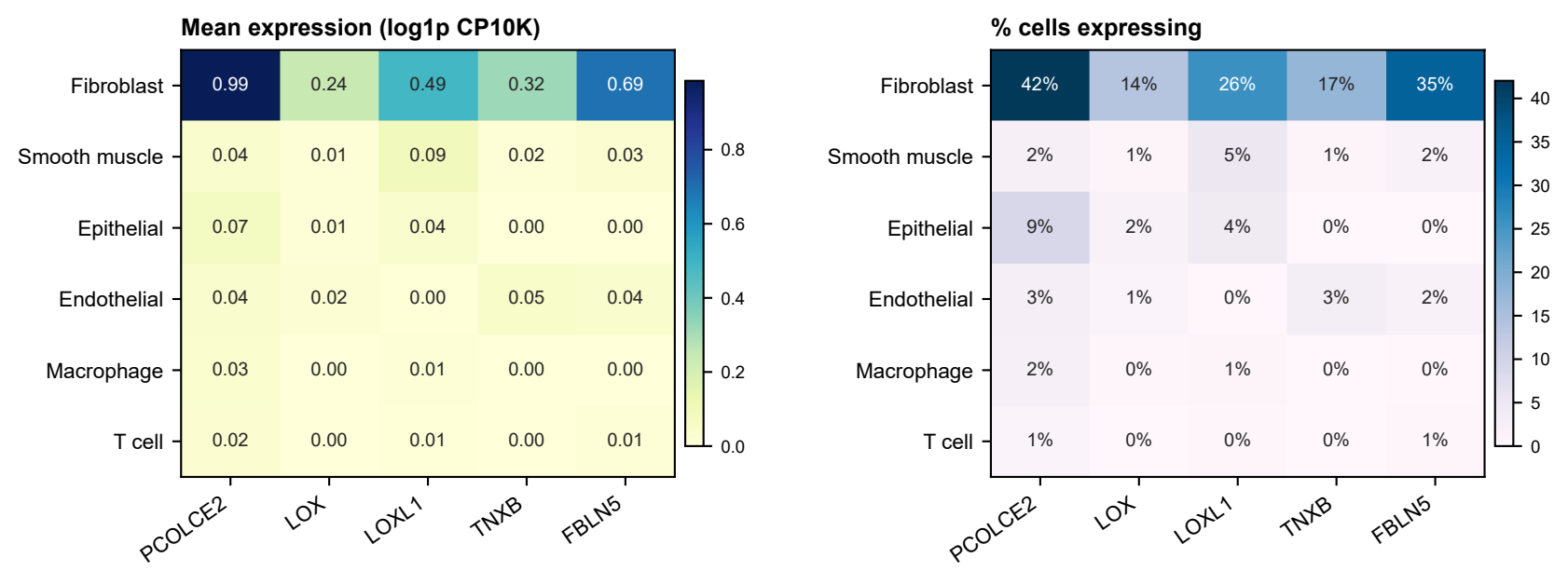
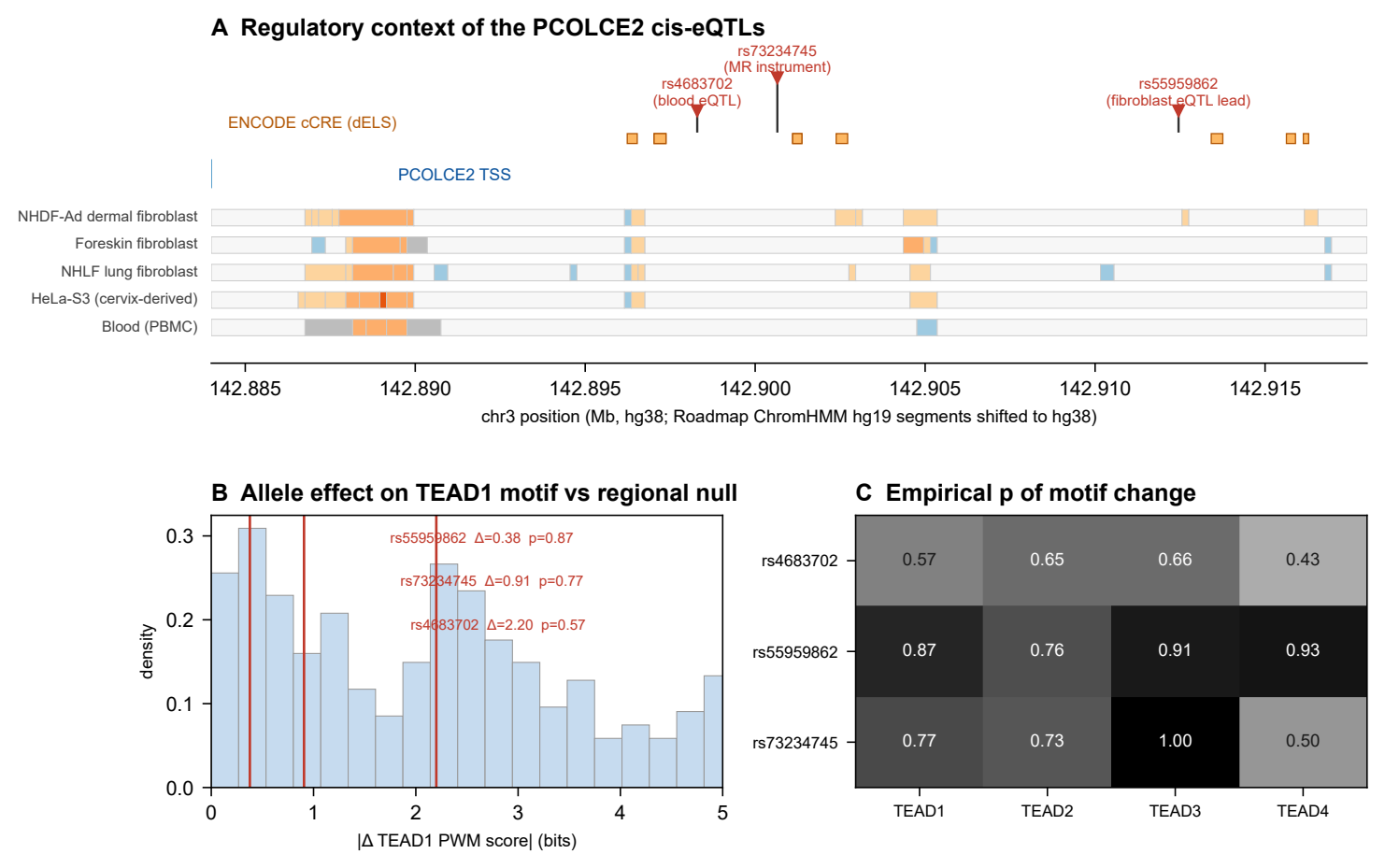

Mean expression (log1p CP10K)
Fibroblast
Smooth muscle
Epithelial
Endothelial
Macrophage
T cell
0.99
0.24
0.49
0.32
0.69
0.04
0.01
0.09
0.02
0.03
0.07
0.01
0.04
0.00
0.00
0.04
0.02
0.00
0.05
0.04
0.03
0.00
0.01
0.00
0.00
0.02
0.00
0.01
0.00
0.01
0.8
0.6
0.4
0.2
0.0
PCOLCE2
LOX
LOXL1
TNXB
FBLN5
% cells expressing
Fibroblast
Smooth muscle
Epithelial
Endothelial
Macrophage
T cell
42%
14%
26%
17%
35%
2%
1%
5%
1%
2%
9%
2%
4%
0%
0%
3%
1%
0%
3%
2%
2%
0%
1%
0%
0%
1%
0%
0%
0%
1%
40
35
30
25
20
15
10
5
0
PCOLCE2
LOX
LOXL1
TNXB
FBLN5


A Regulatory context of the PCOLCE2 cis-eQTLs
rs73234745
(MR instrument)
rs4683702
(blood eQTL)
rs55959862
(fibroblast eQTL lead)
ENCODE cCRE (dELS)
PCOLCE2 TSS
NHDF-Ad dermal fibroblast
Foreskin fibroblast
NHLF lung fibroblast
HeLa-S3 (cervix-derived)
Blood (PBMC)
142.885
142.890
142.895
142.900
142.905
142.910
142.915
chr3 position (Mb, hg38; Roadmap ChromHMM hg19 segments shifted to hg38)
B Allele effect on TEAD1 motif vs regional null
rs55959862 Δ=0.38 p=0.87
rs73234745 Δ=0.91 p=0.77
rs4683702 Δ=2.20 p=0.57
density
0.0
0.1
0.2
0.3
0
1
2
3
4
5
|Δ TEAD1 PWM score| (bits)
C Empirical p of motif change
rs4683702
rs55959862
rs73234745
0.57
0.65
0.66
0.43
0.87
0.76
0.91
0.93
0.77
0.73
1.00
0.50
TEAD1
TEAD2
TEAD3
TEAD4