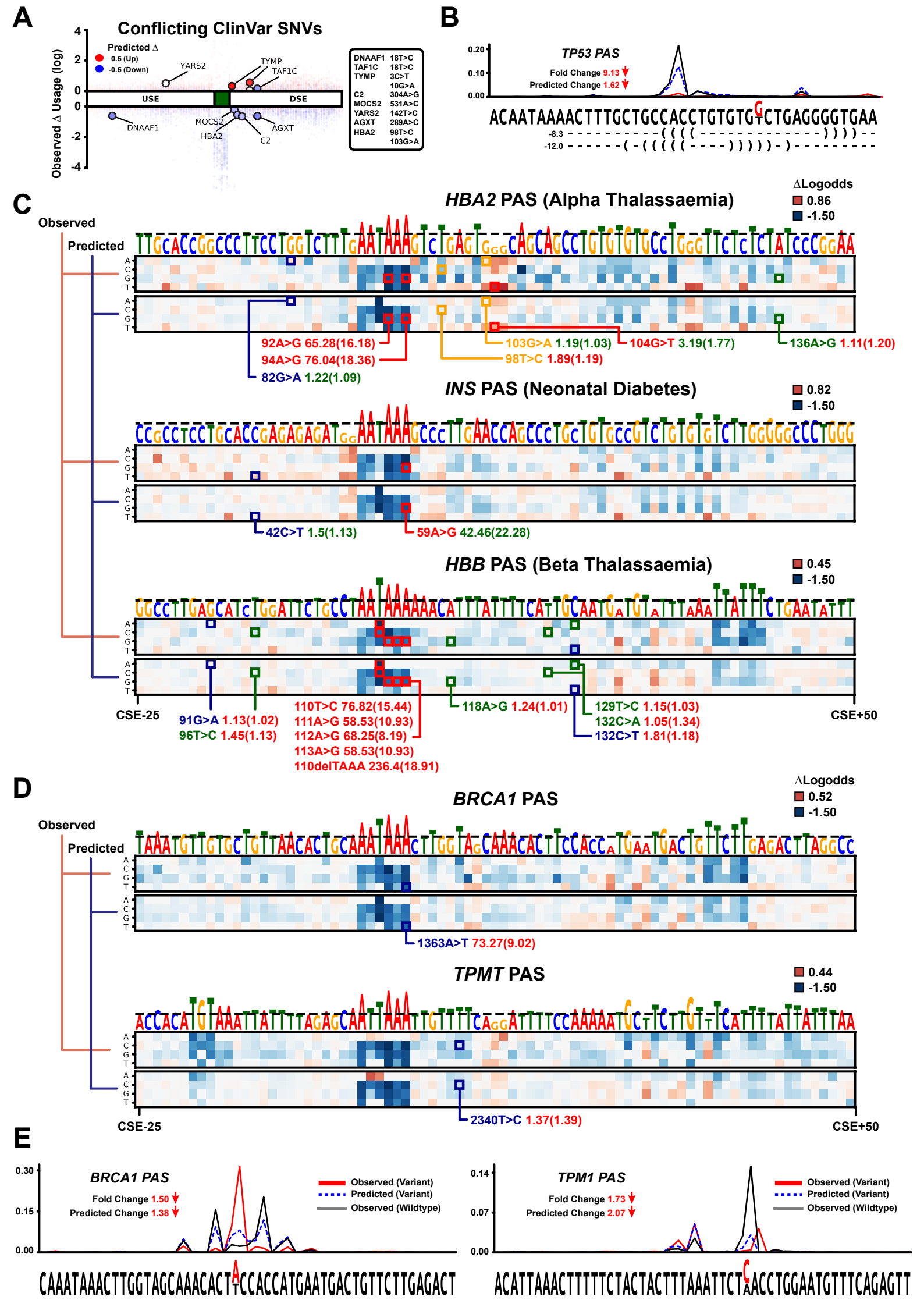
A
Conflicting ClinVar SNVs
Predicted Δ
0.5 (Up)
-0.5 (Down)
Observed Δ Usage (log)
USE
DSE
YARS2
TYMP
TAF1C
DNAAF1
MOCS2
HBA2
C2
AGXT
DNAAF1 18T>C
TAF1C 18T>C
TYMP 3C>T
10G>A
C2 304A>G
MOCS2 531A>C
YARS2 142T>C
AGXT 289A>C
HBA2 98T>C
103G>A
B
TP53 PAS
Fold Change 9.13
Predicted Change 1.62
ACAATAAAACTTTGCTGCCACCTGTGTGCTGAGGGGTGAA
C
HBA2 PAS (Alpha Thalassaemia)
ΔLogodds
0.86
-1.50
Observed
Predicted
92A>G 65.28(16.18)
94A>G 76.04(18.36)
82G>A 1.22(1.09)
103G>A 1.19(1.03)
98T>C 1.89(1.19)
104G>T 3.19(1.77)
136A>G 1.11(1.20)
INS PAS (Neonatal Diabetes)
0.82
-1.50
42C>T 1.5(1.13)
59A>G 42.46(22.28)
HBB PAS (Beta Thalassaemia)
0.45
-1.50
CSE-25
CSE+50
91G>A 1.13(1.02)
96T>C 1.45(1.13)
110T>C 76.82(15.44)
111A>G 58.53(10.93)
112A>G 68.25(8.19)
113A>G 58.53(10.93)
110delTAAA 236.4(18.91)
118A>G 1.24(1.01)
129T>C 1.15(1.03)
132C>A 1.05(1.34)
132C>T 1.81(1.18)
D
BRCA1 PAS
ΔLogodds
0.52
-1.50
Observed
Predicted
1363A>T 73.27(9.02)
TPMT PAS
0.44
-1.50
CSE-25
CSE+50
2340T>C 1.37(1.39)
E
BRCA1 PAS
Fold Change 1.50
Predicted Change 1.38
Observed (Variant)
Predicted (Variant)
Observed (Wildtype)
CAAATAAACTTGGTAGCAAACACTACCACCATGAATGACTGTTCTTGAGACT
TPM1 PAS
Fold Change 1.73
Predicted Change 2.07
ACATTAAACTTTTTCTACTACTTTAAATTCTACCTGGAATGTTTCAGAGTT