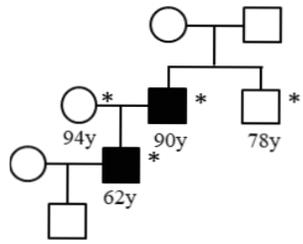
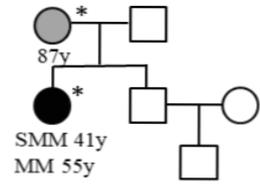


Supplementary Figures

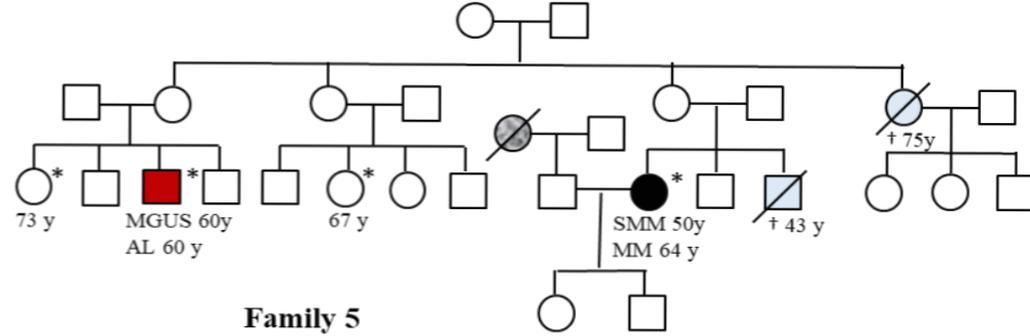
Family 1



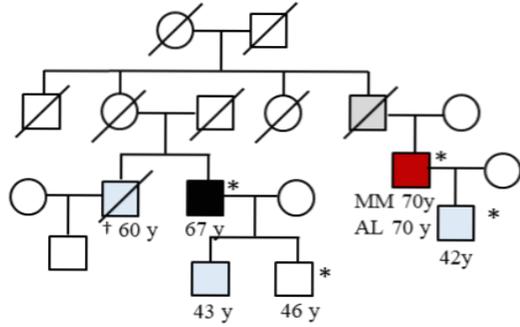
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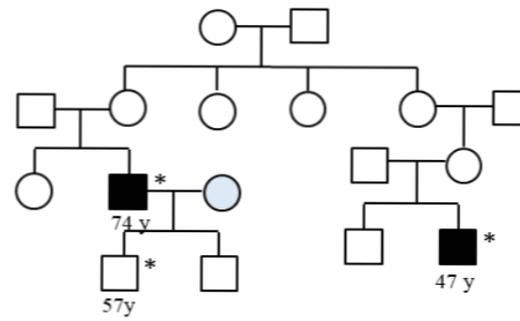
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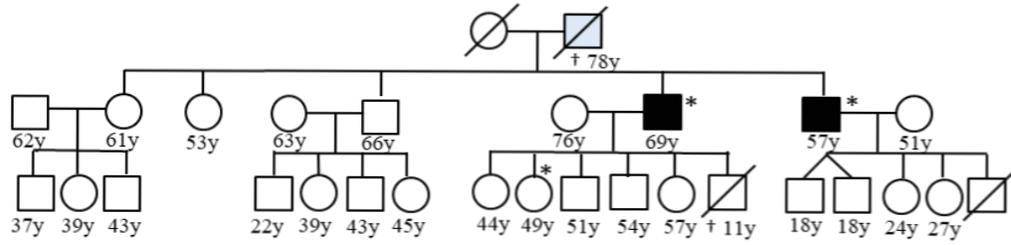
Family 4



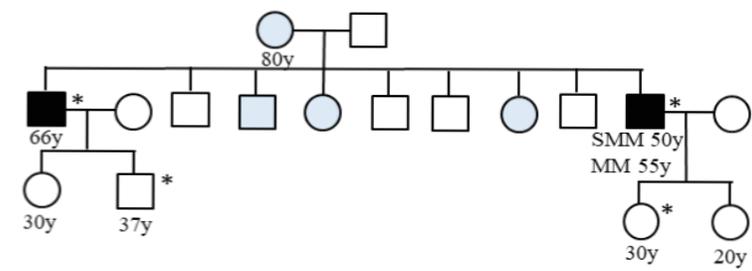
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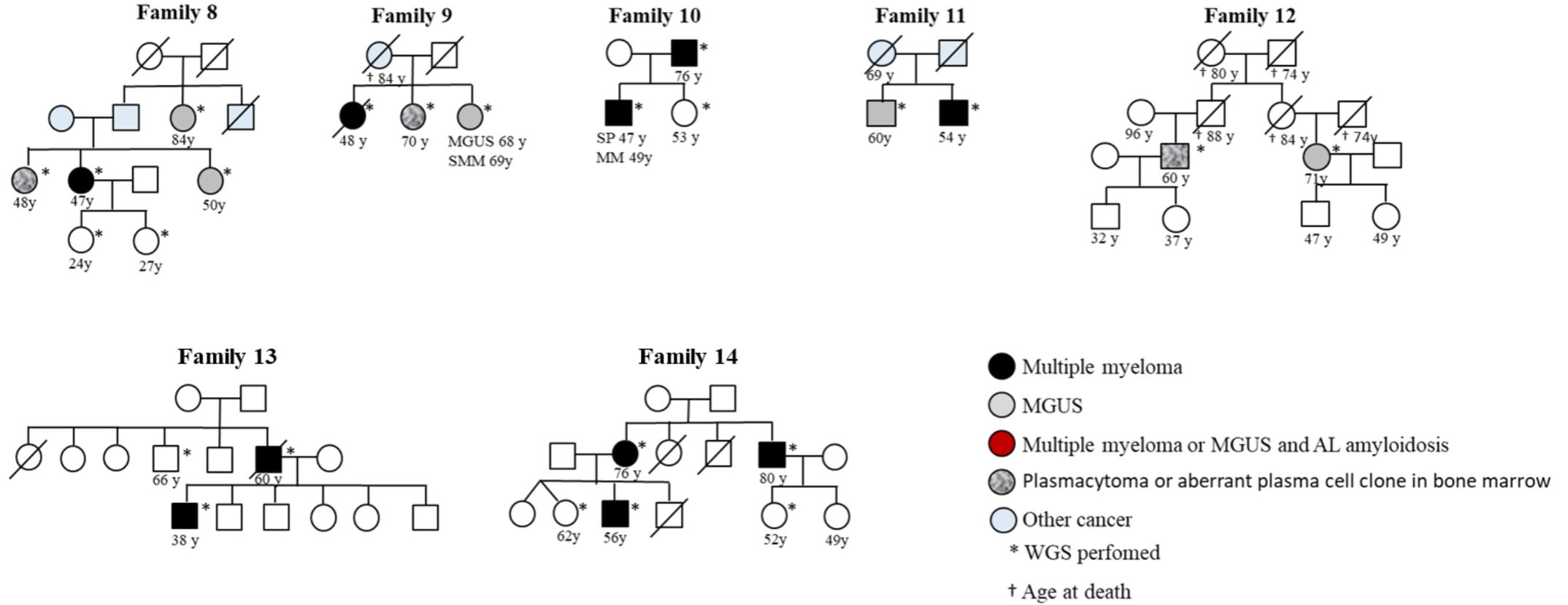


Family 6

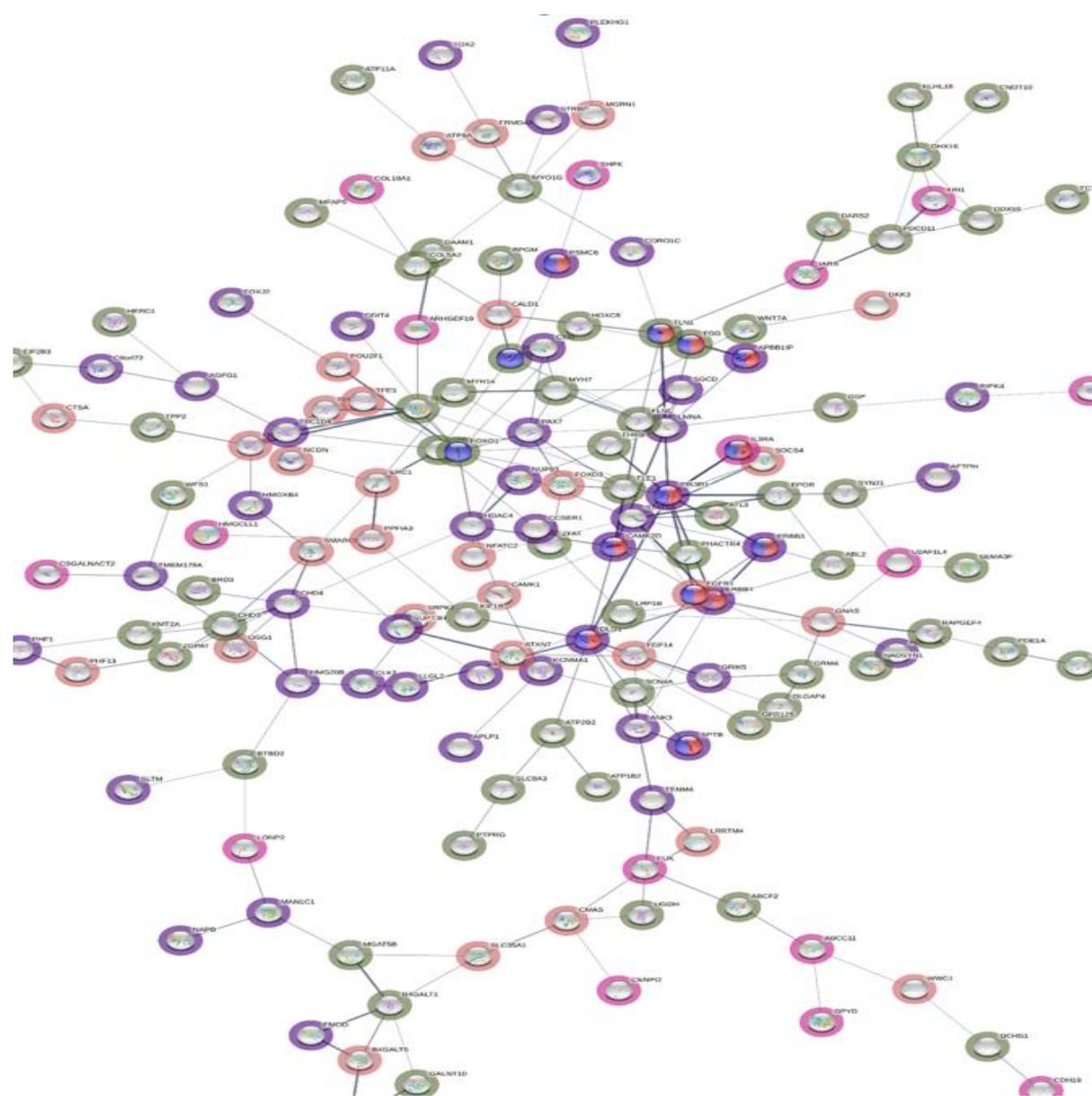


Family 7



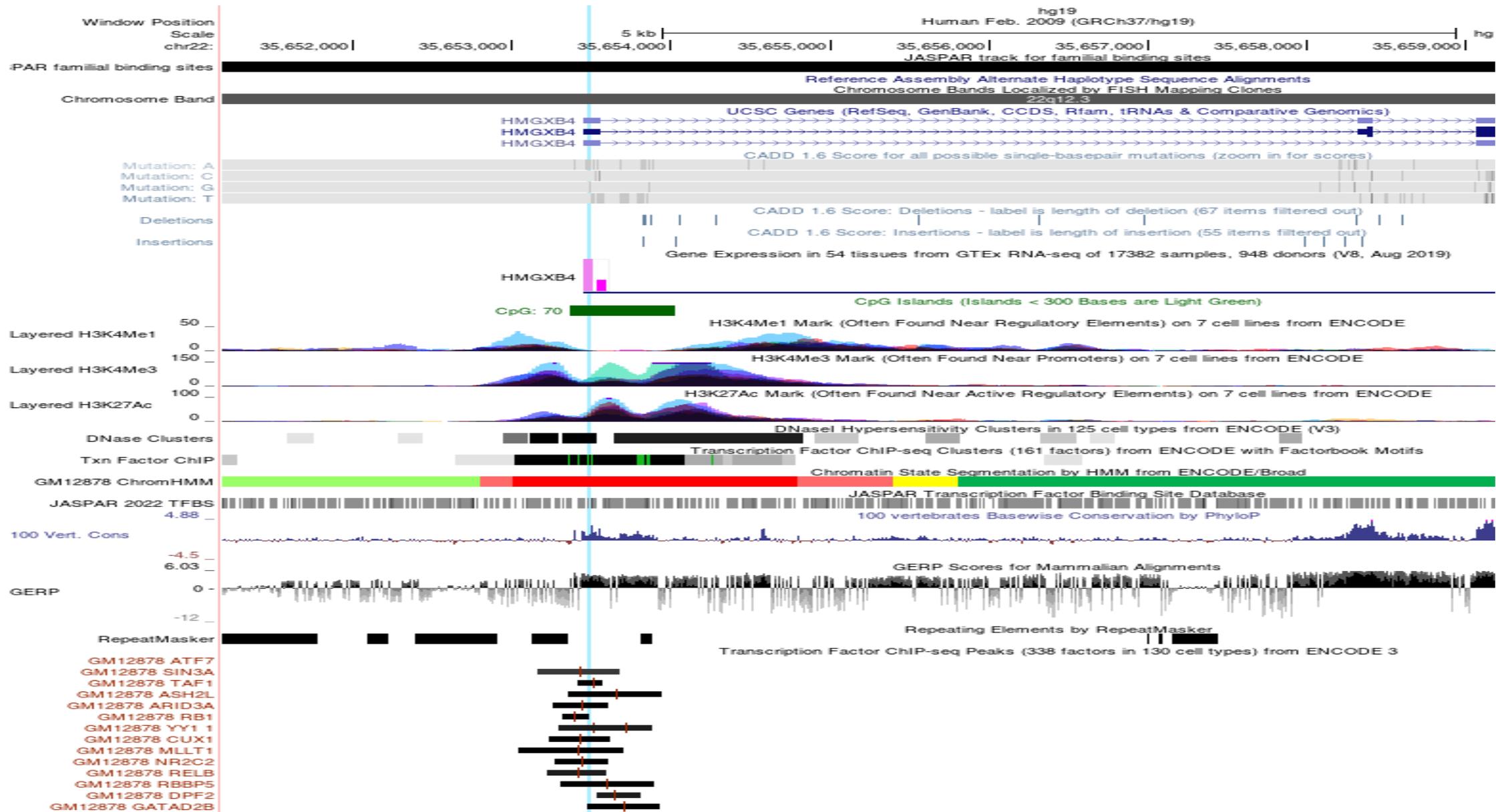


Supplementary Figure S1. Family pedigrees of the 14 families investigated in the study.

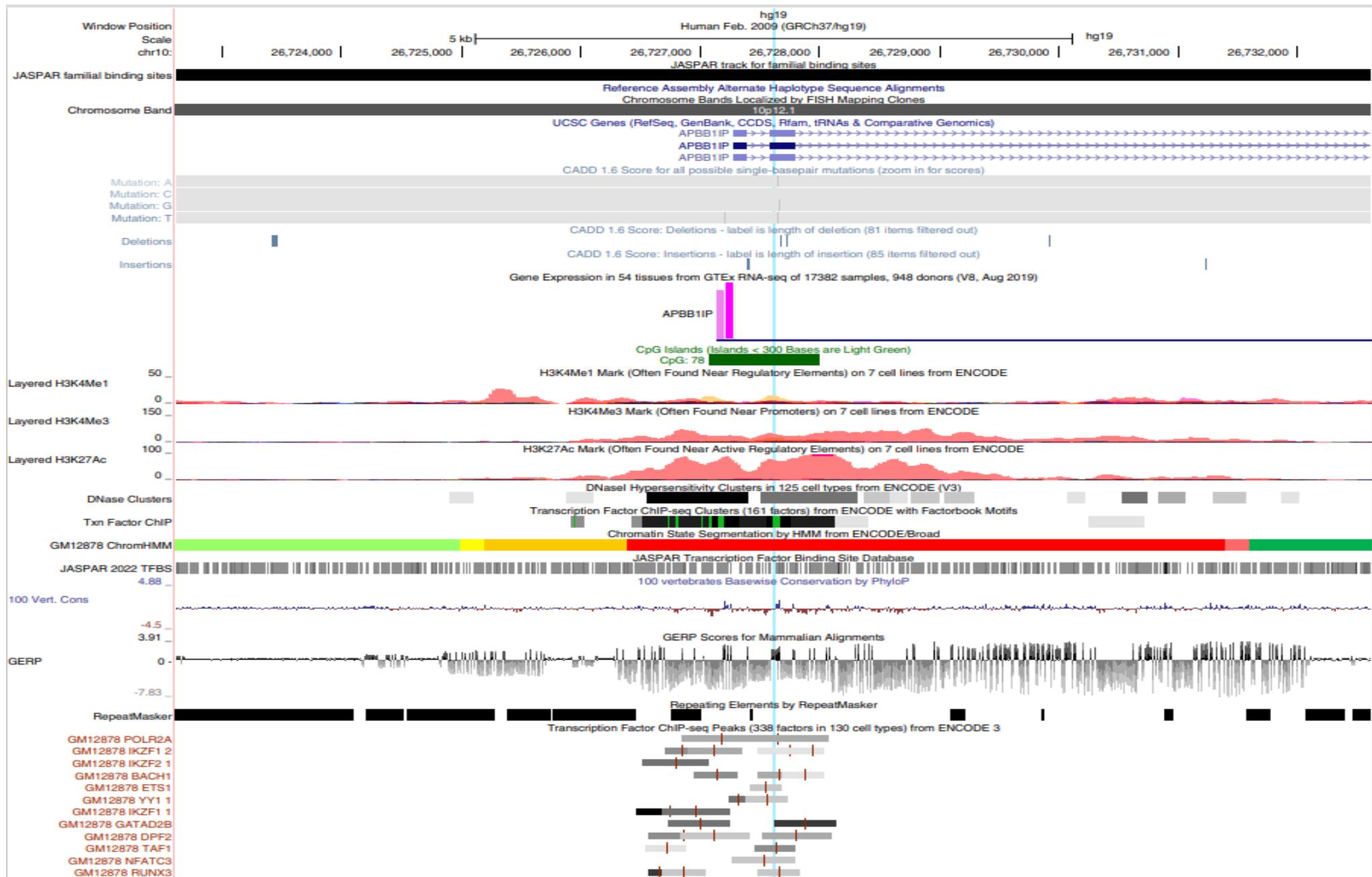


Supplementary Figure S2. Protein interaction network of a combination of genes with 5' and 3' UTR variants and genes with missense and loss-of-function variants generated by STRING. Proteins surrounded by green halo are missense and those surrounded by magenta are loss-of-function (LoF) variant related proteins. 5'UTR and 3'UTR variant proteins are surrounded by indigo and light red haloes respectively

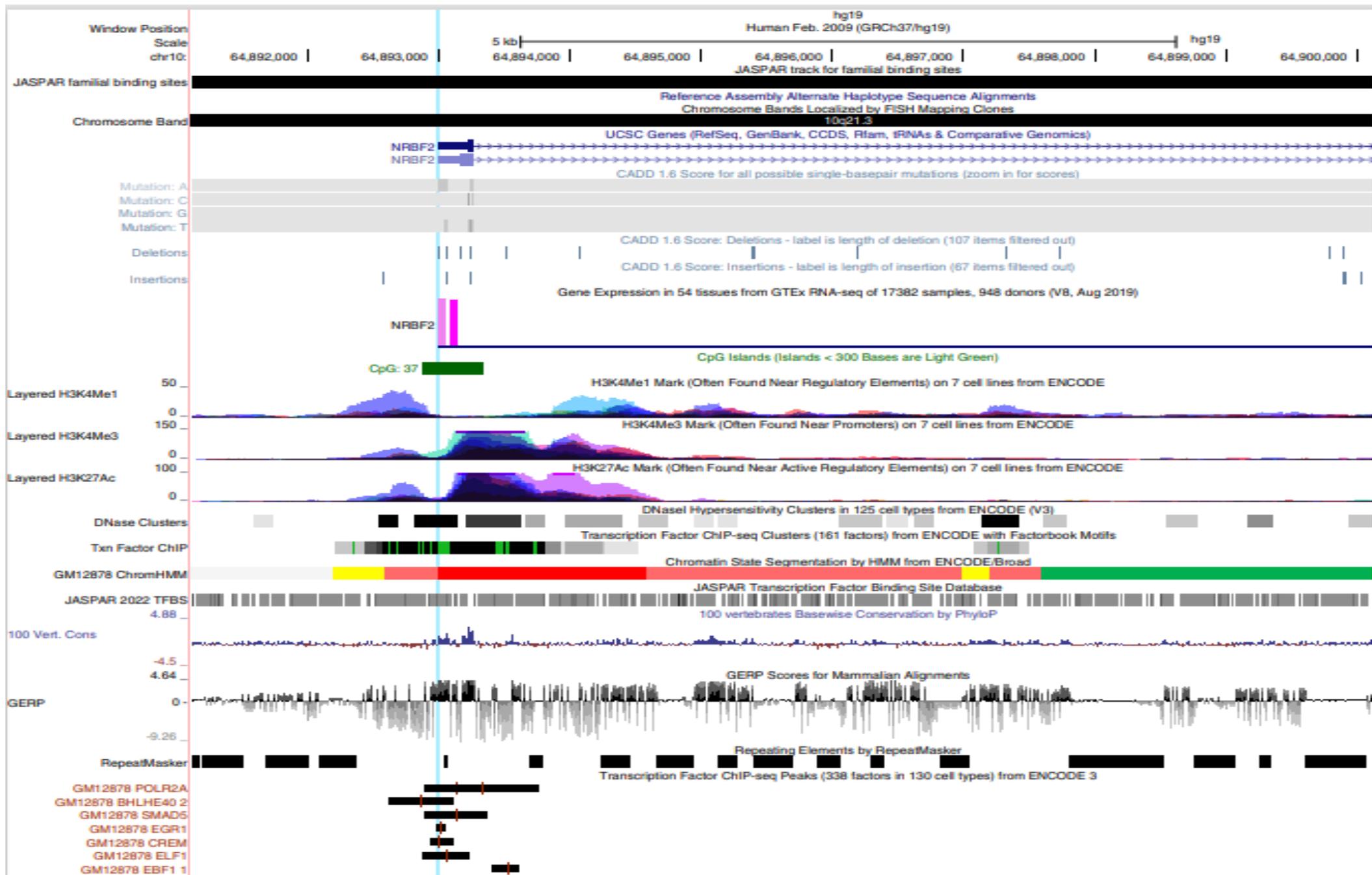
Supplementary Figures S3–S21: UCSC plots of top 5' UTR and MAPK pathway variants



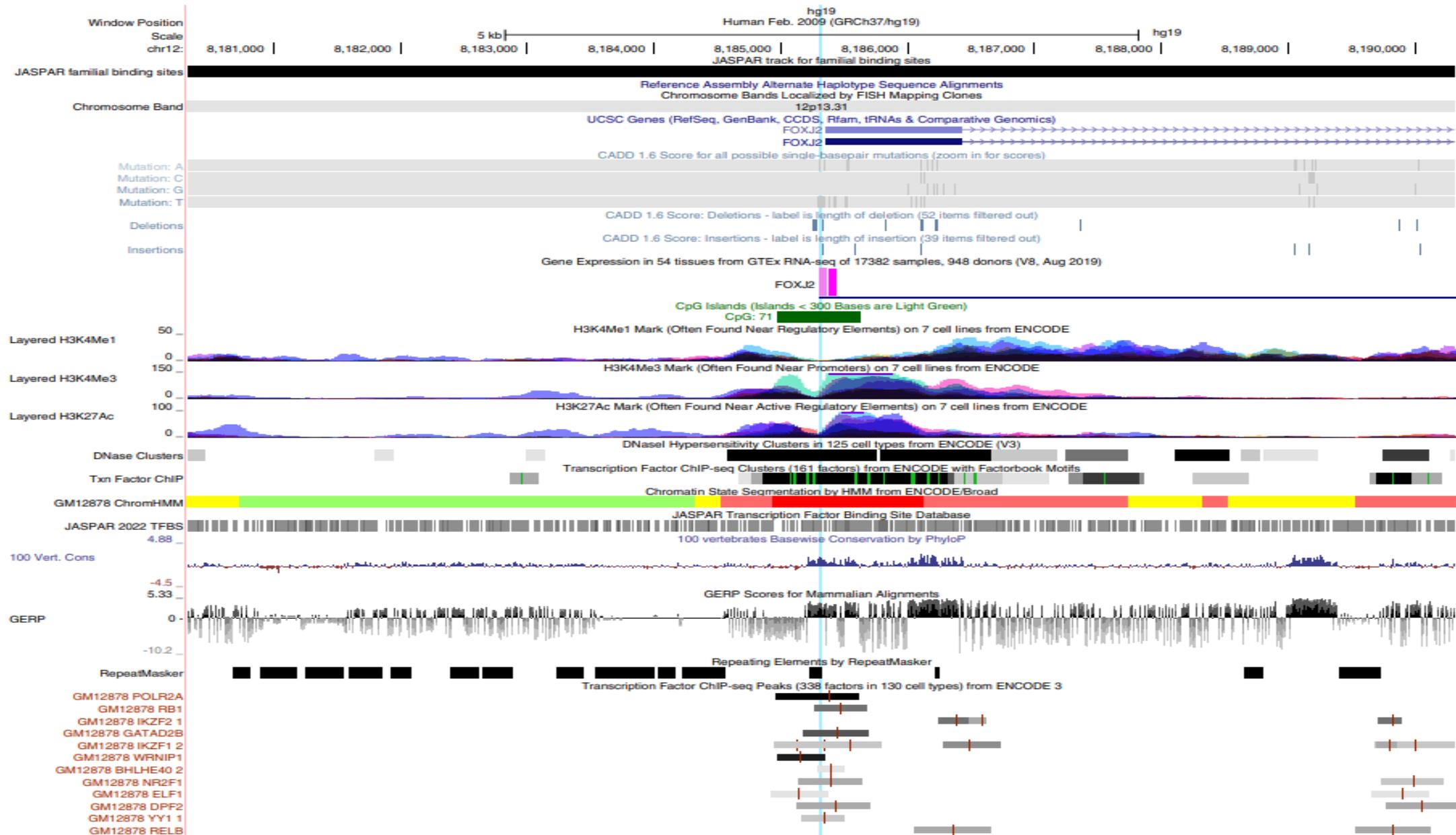
Supplementary Figure S3 HMGXB4:22_35653479_C_A



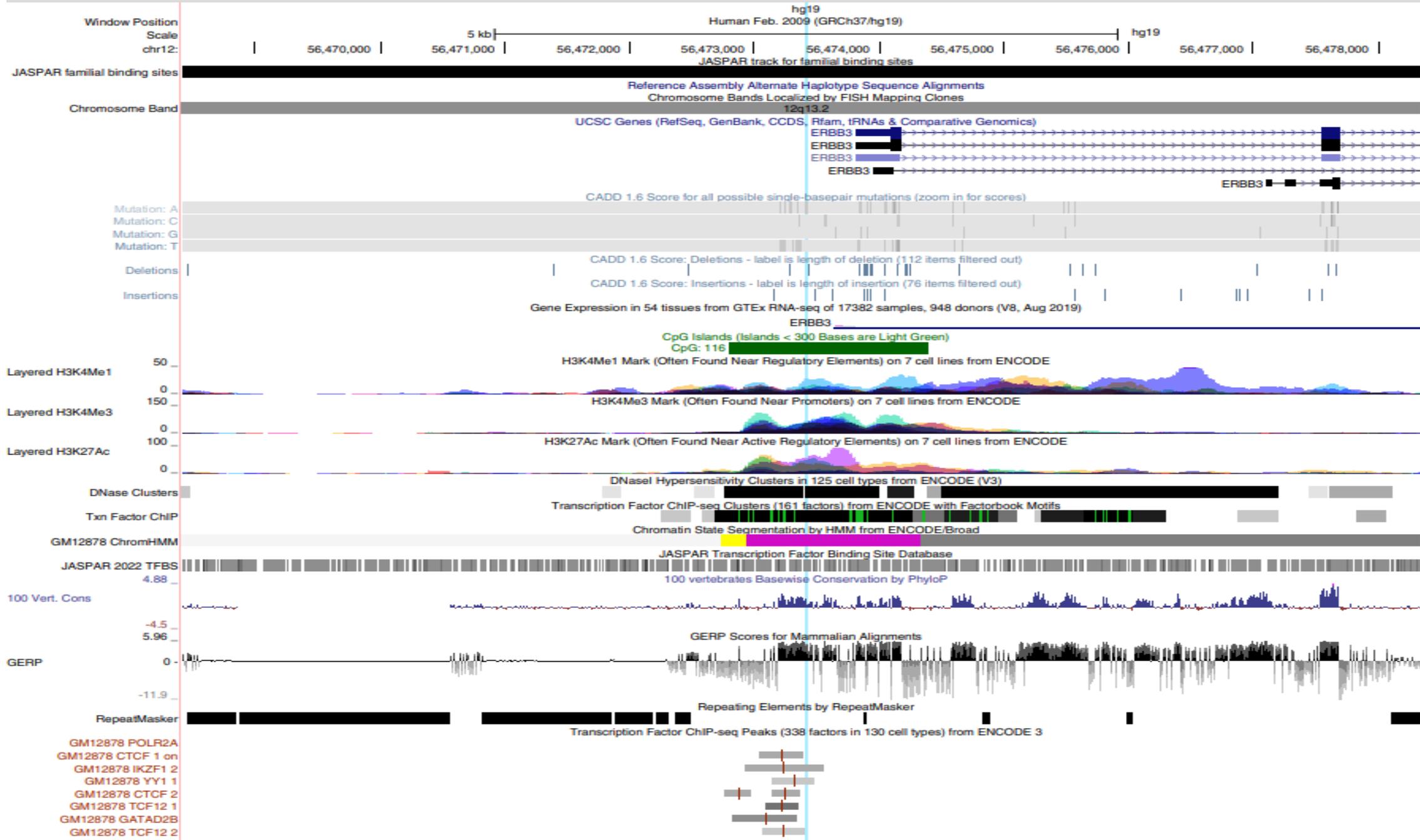
Supplementary Figure S4. *APBB1IP*:10_26727608_C_G



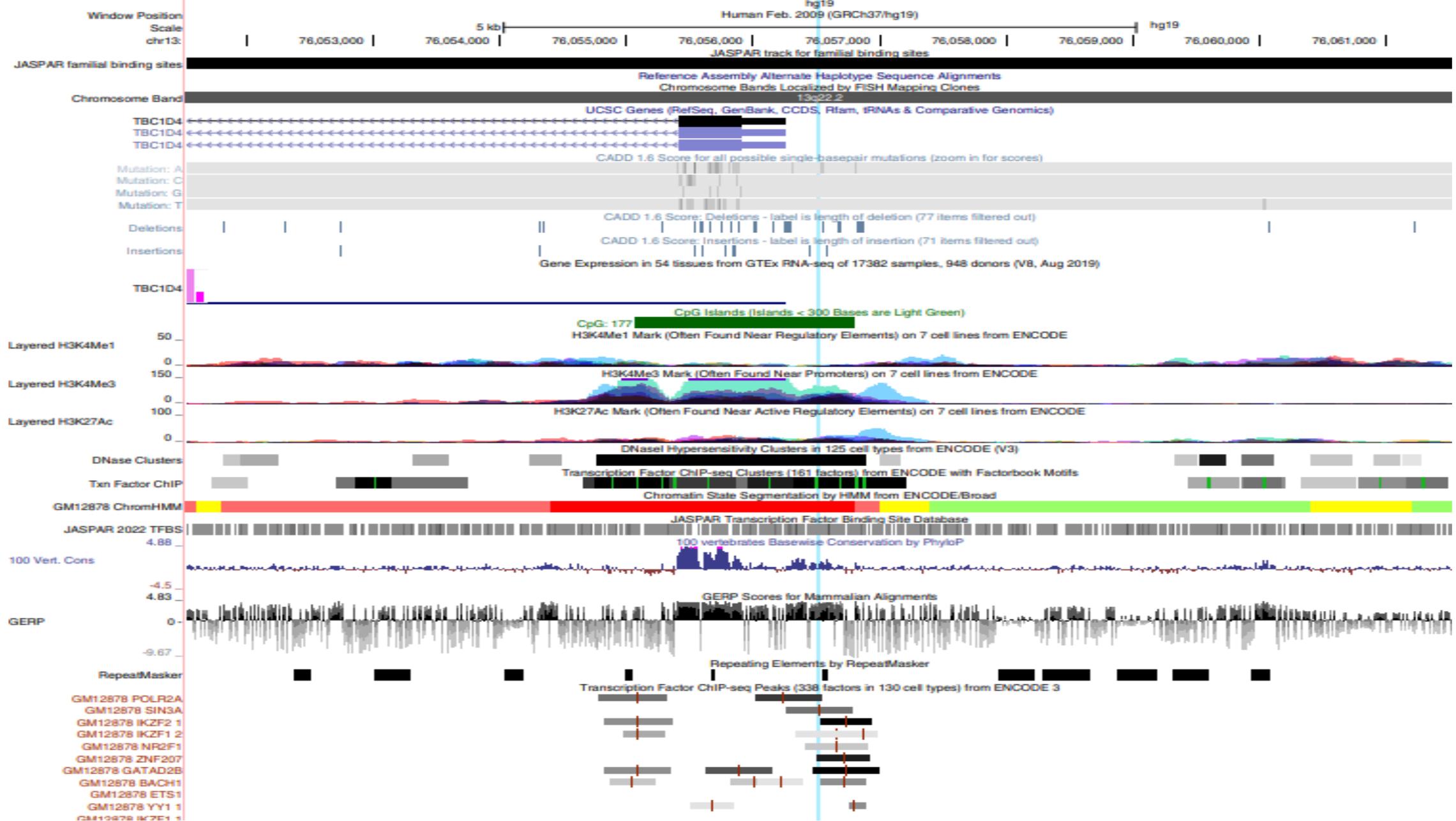
Supplementary Figure S5. NRBF2: 10_64893005_T_C



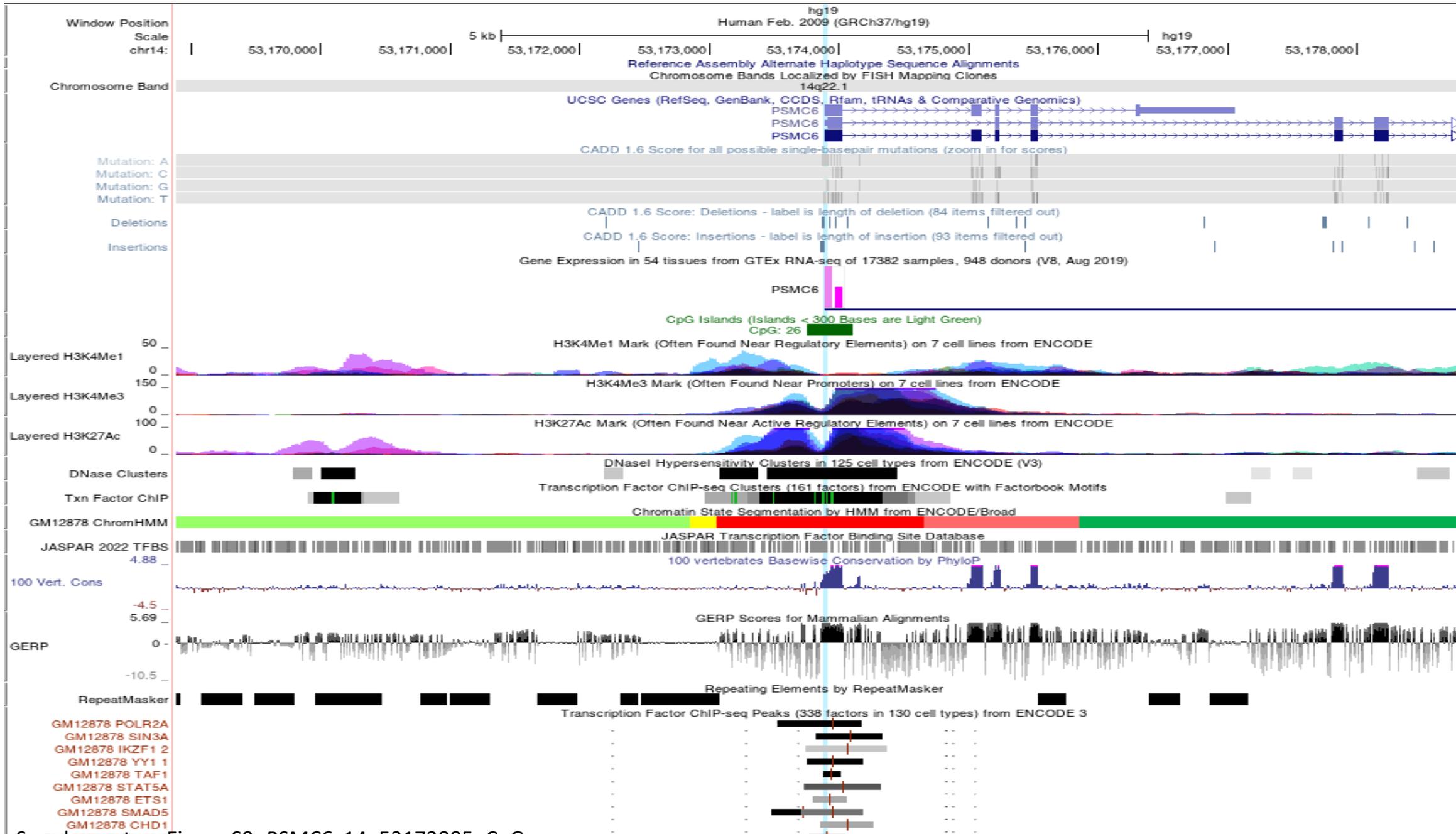
Supplementary Figure S6. *FOXJ2:12_8185317_GGAGCC_G*



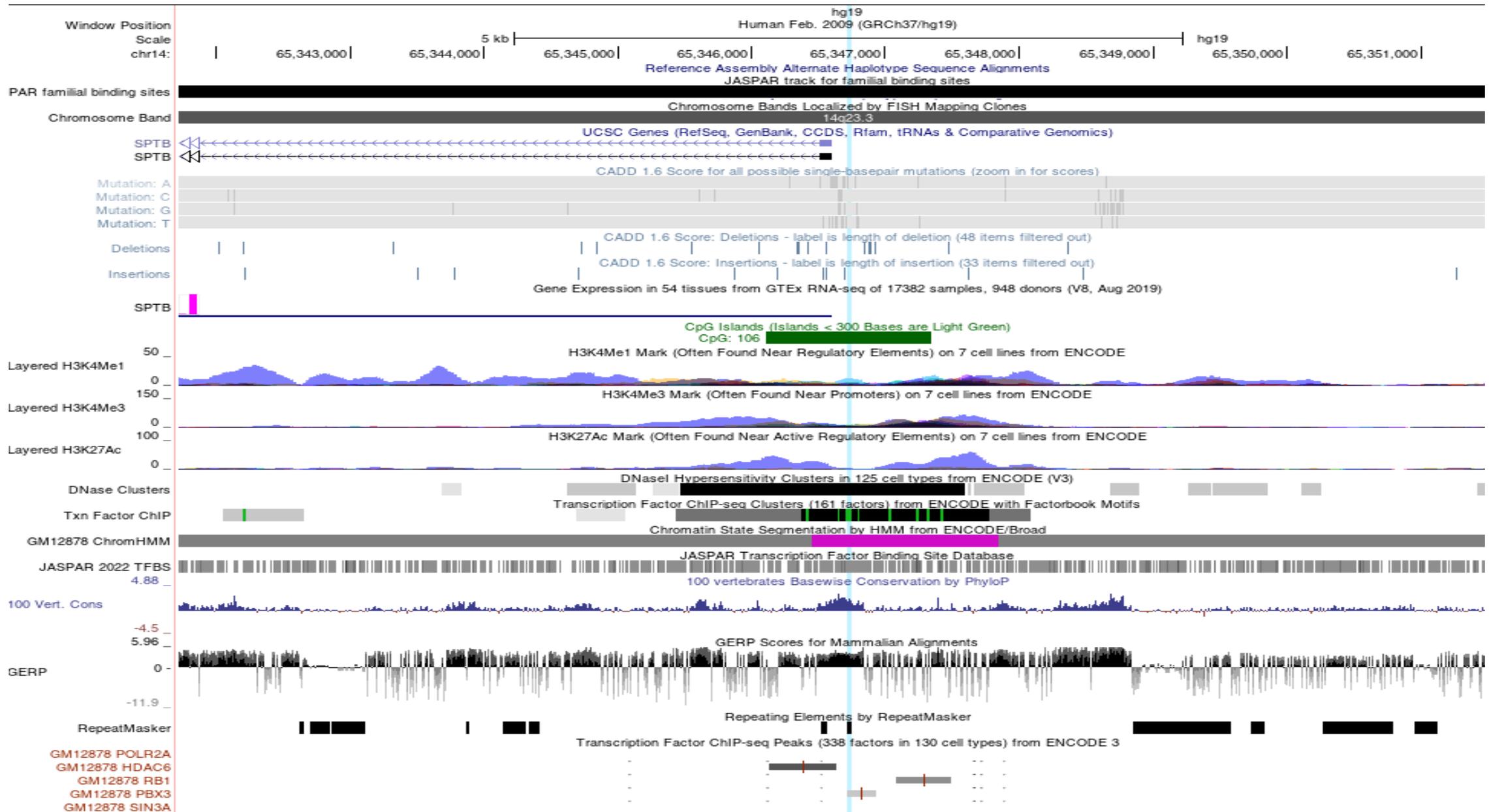
Supplementary Figure S7. ERBB3:12_56473408_C_T



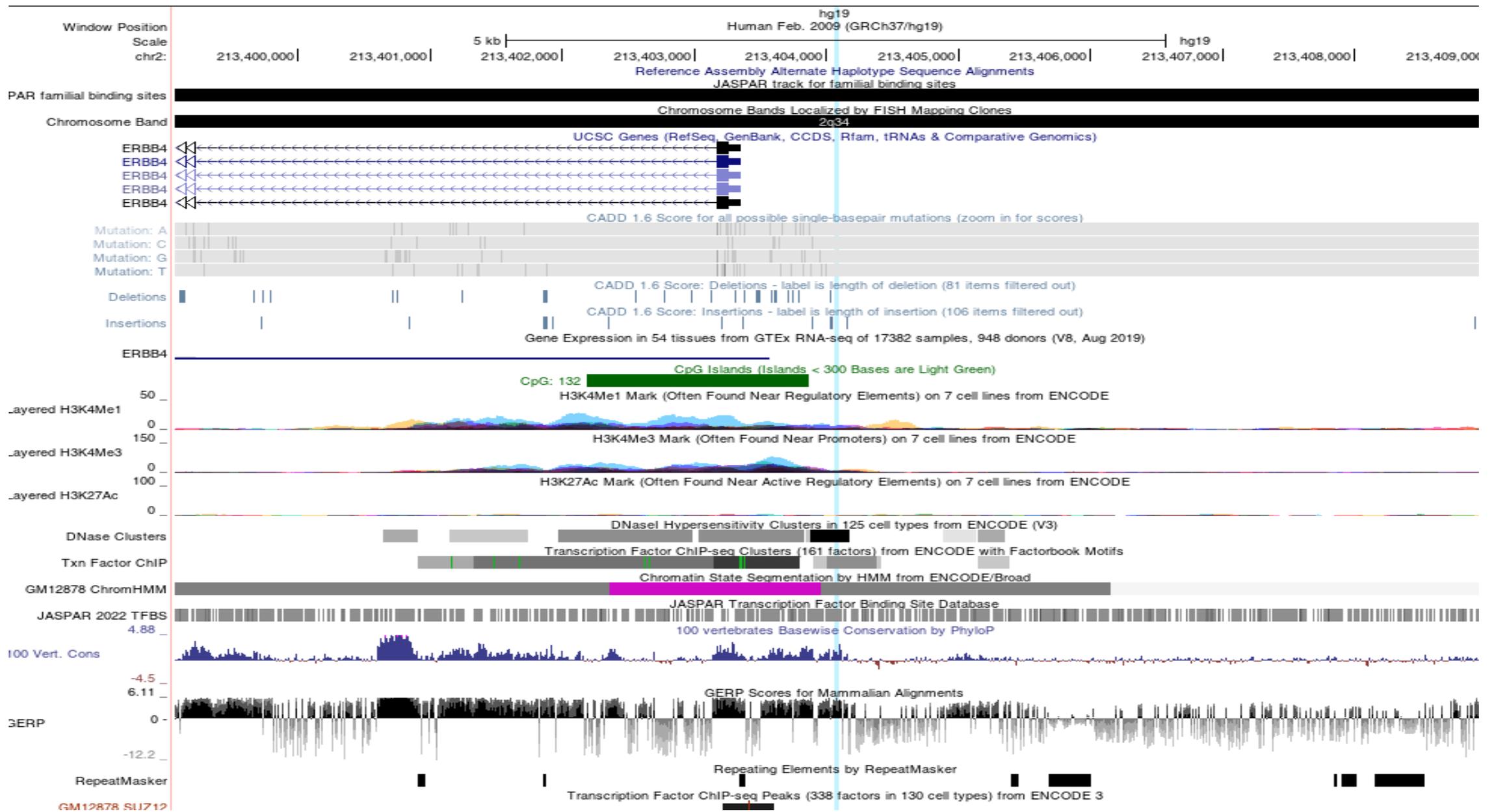
Supplementary Figure S8. *TBC1D4*: 13_76056522_G_A



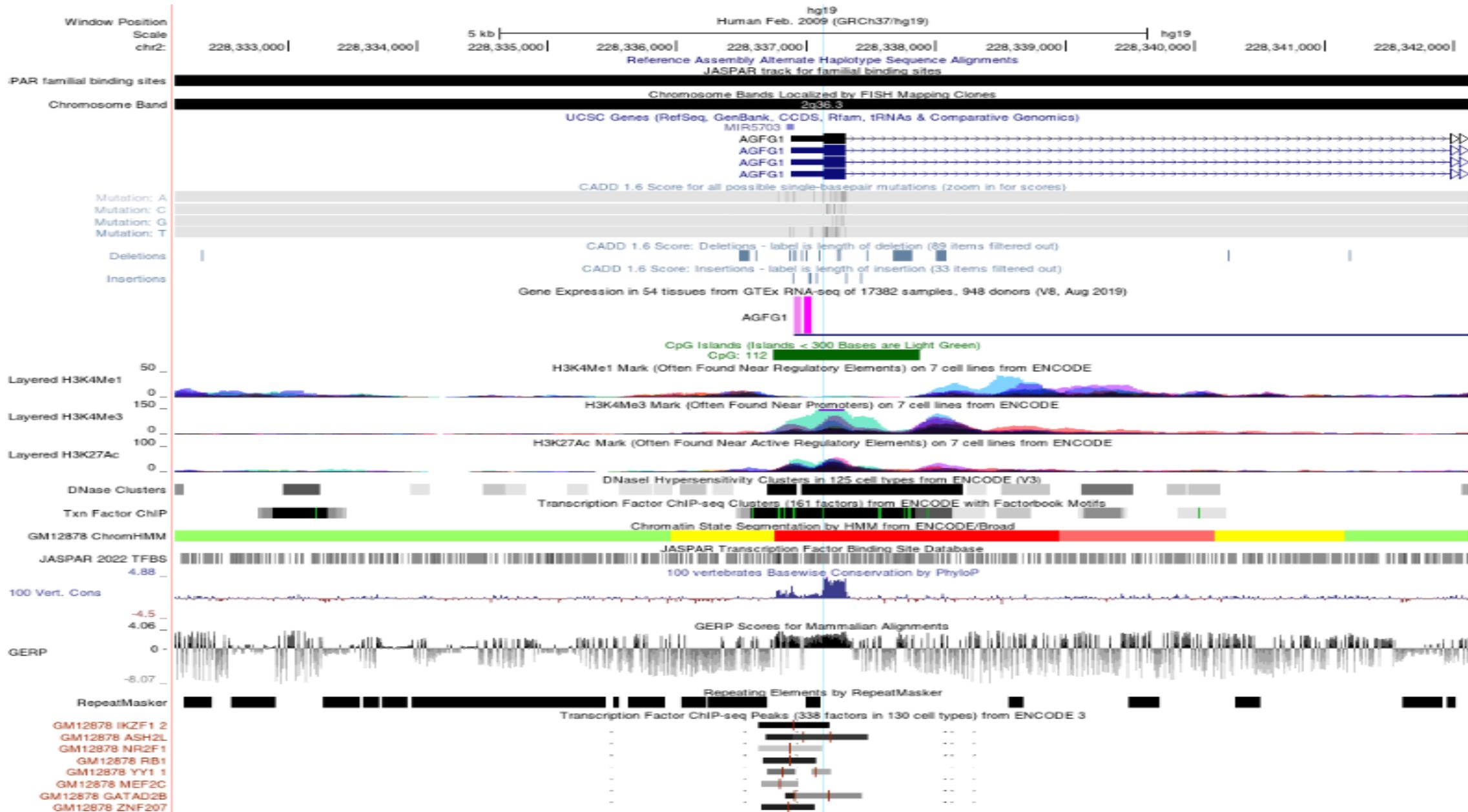
Supplementary Figure S9. *PSMC6*: 14_53173885_C_G



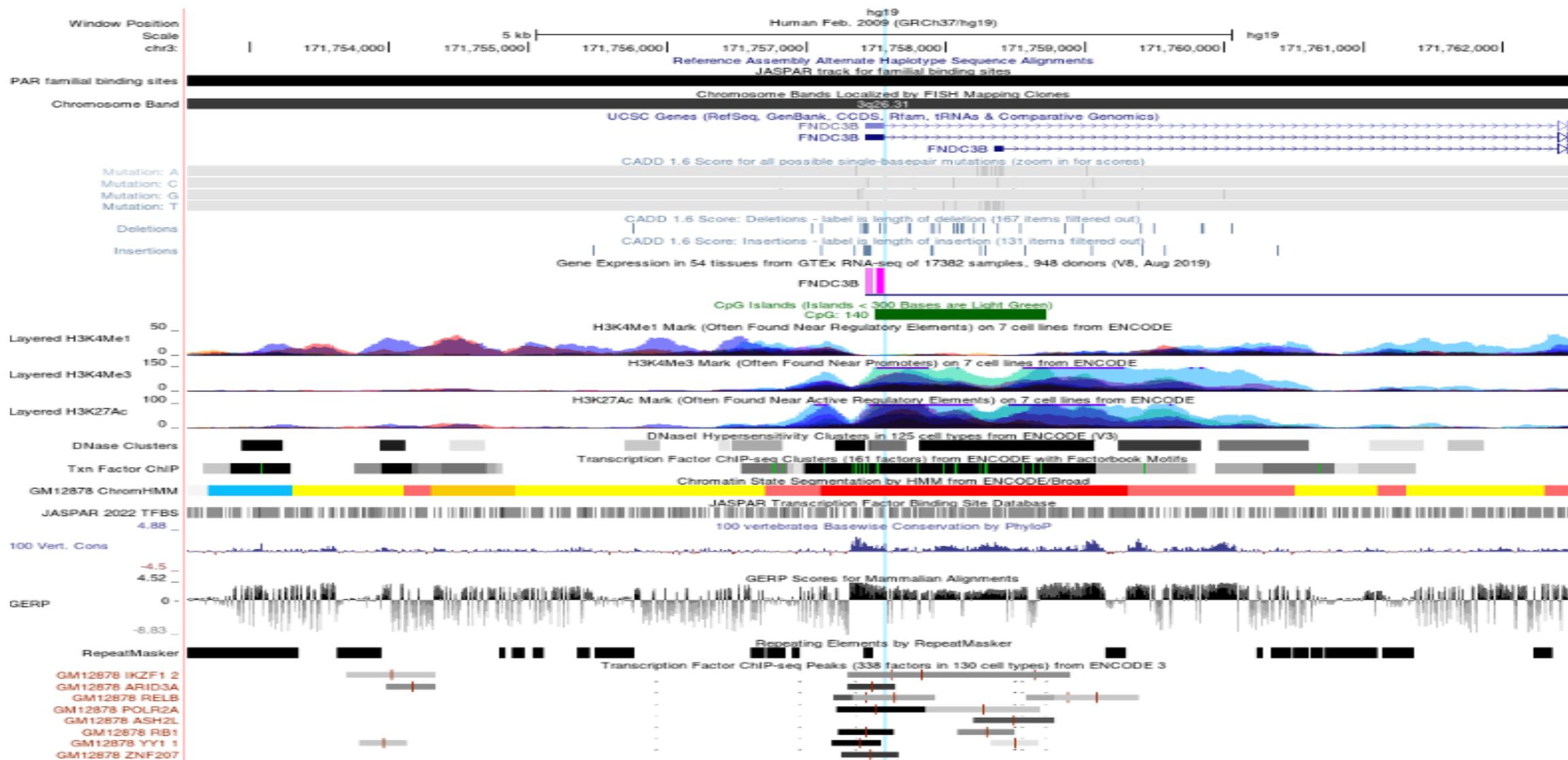
Supplementary Figure S10. *SPTB*: 14_65346721_C_A



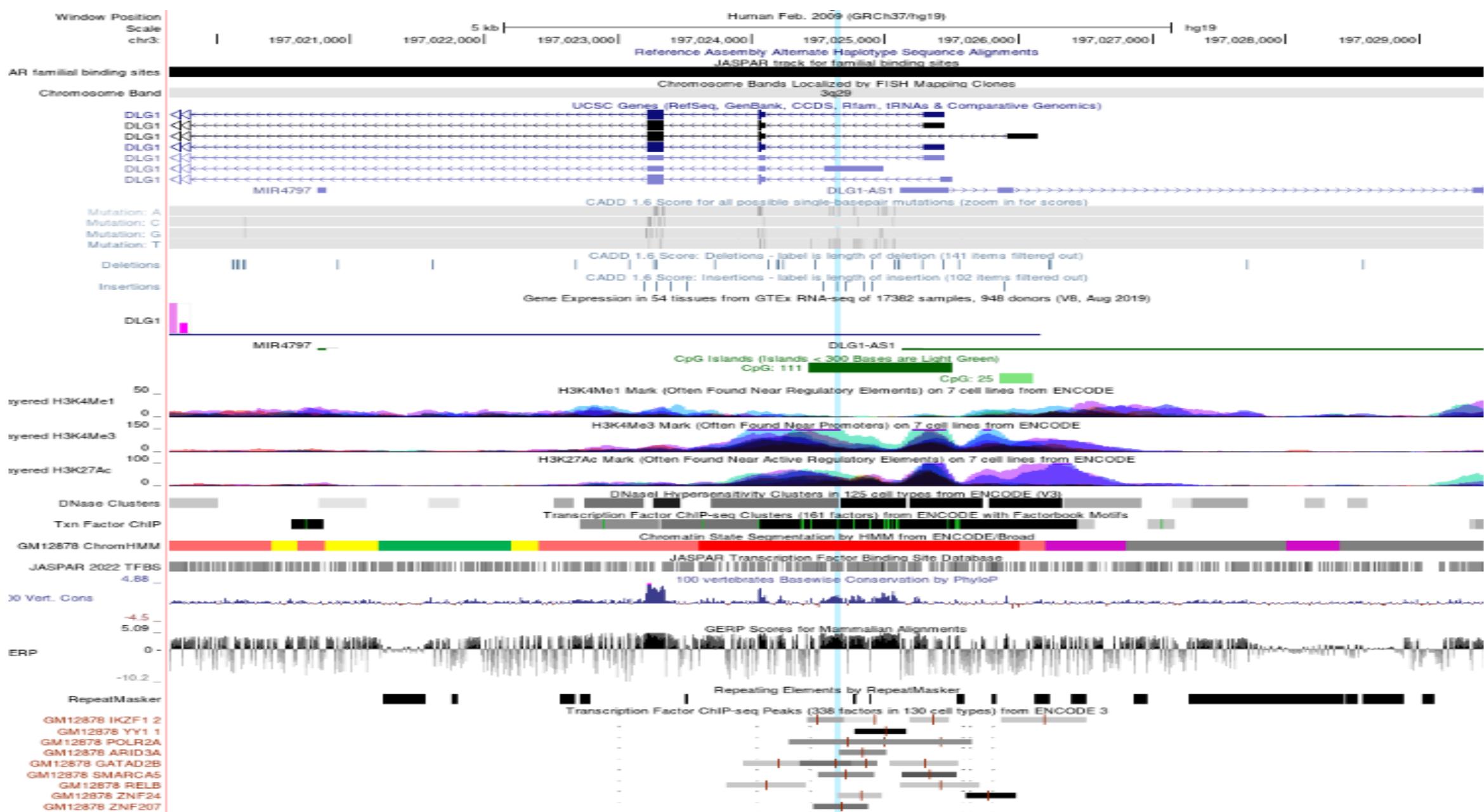
Supplementary Figure S12. *ERBB4*: 2_213404066_C_T



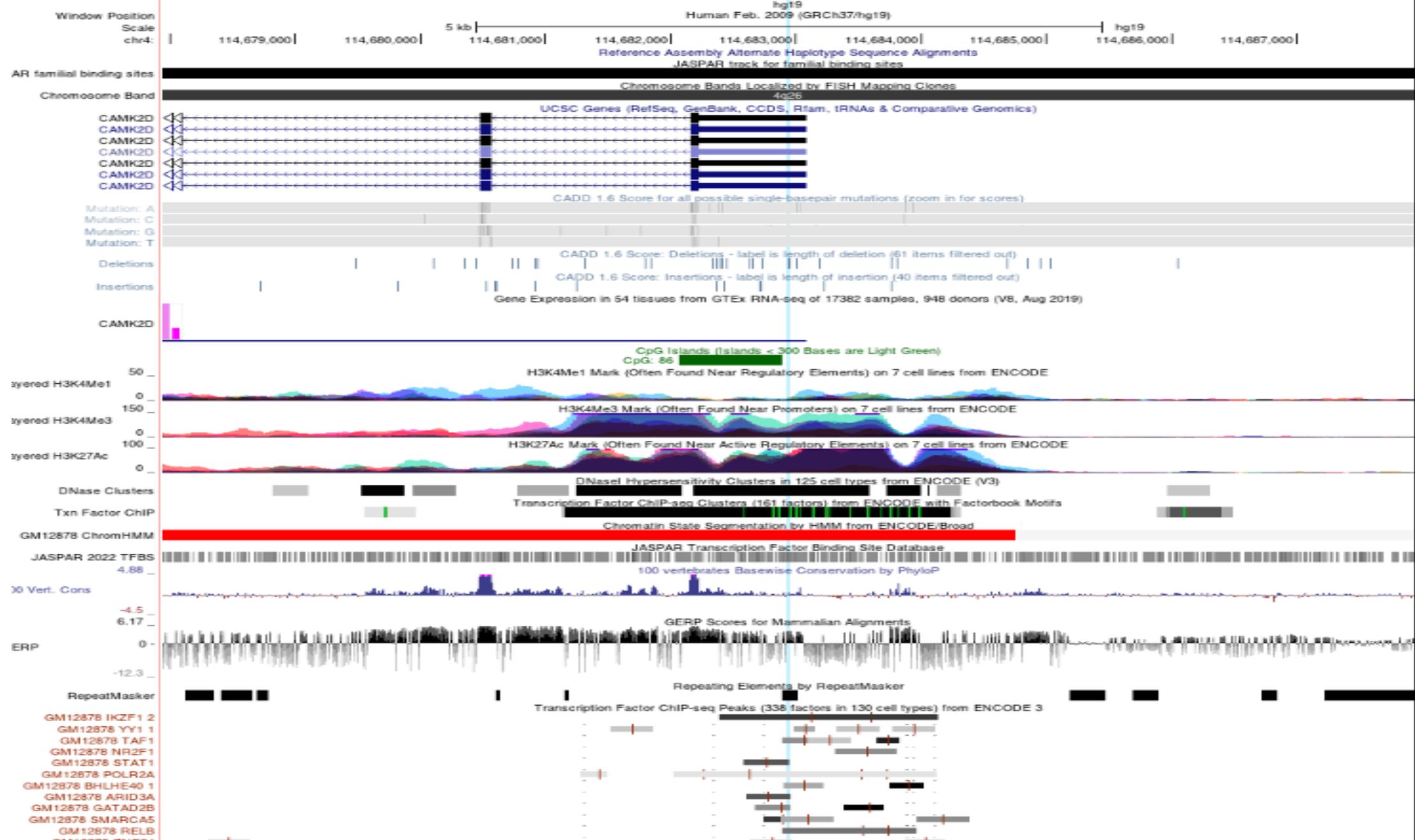
Supplementary Figure S13. *AGFG1*: 2_228337132_G_A



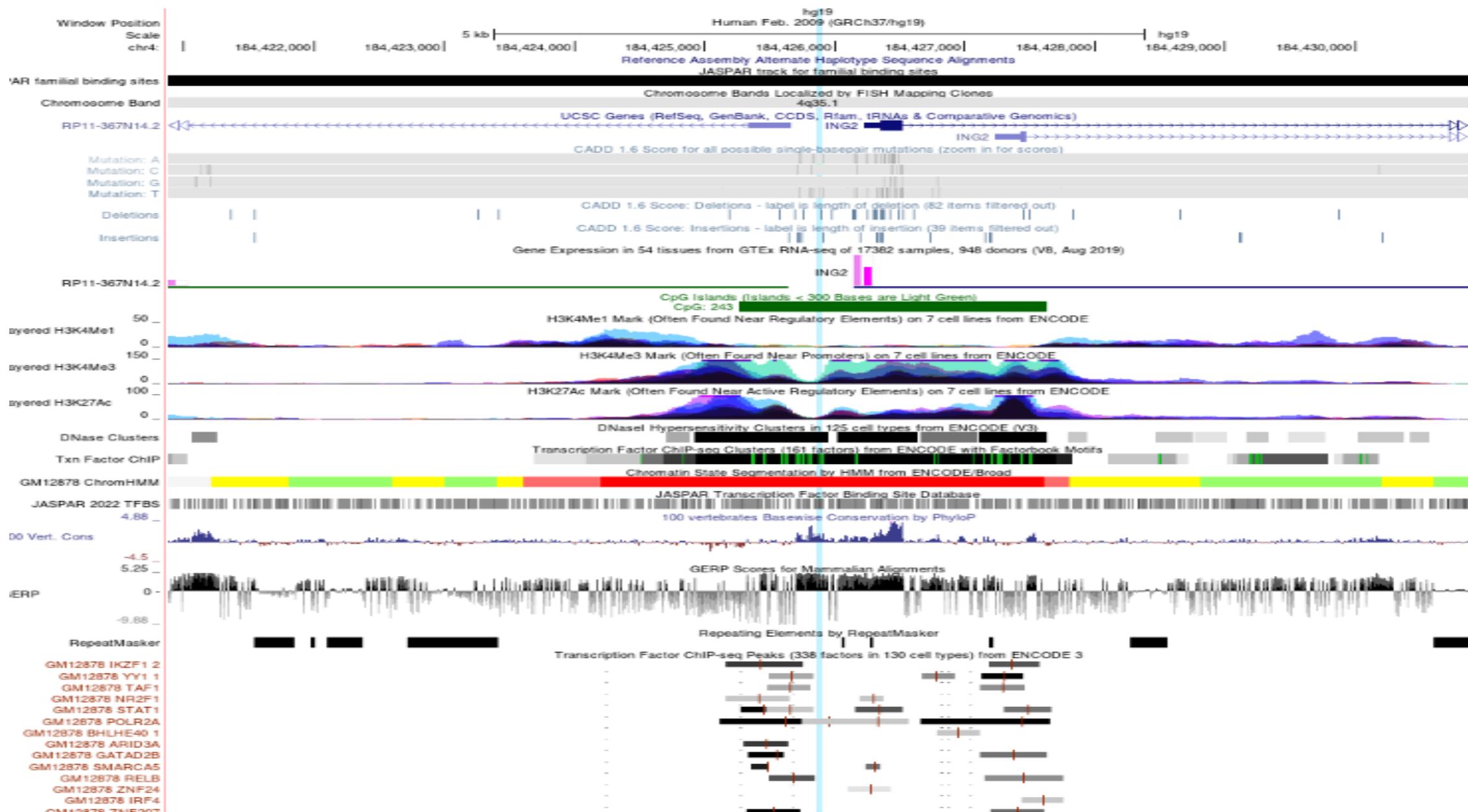
Supplementary Figure S14. *FNDC3B*: 3_171757553_C_A



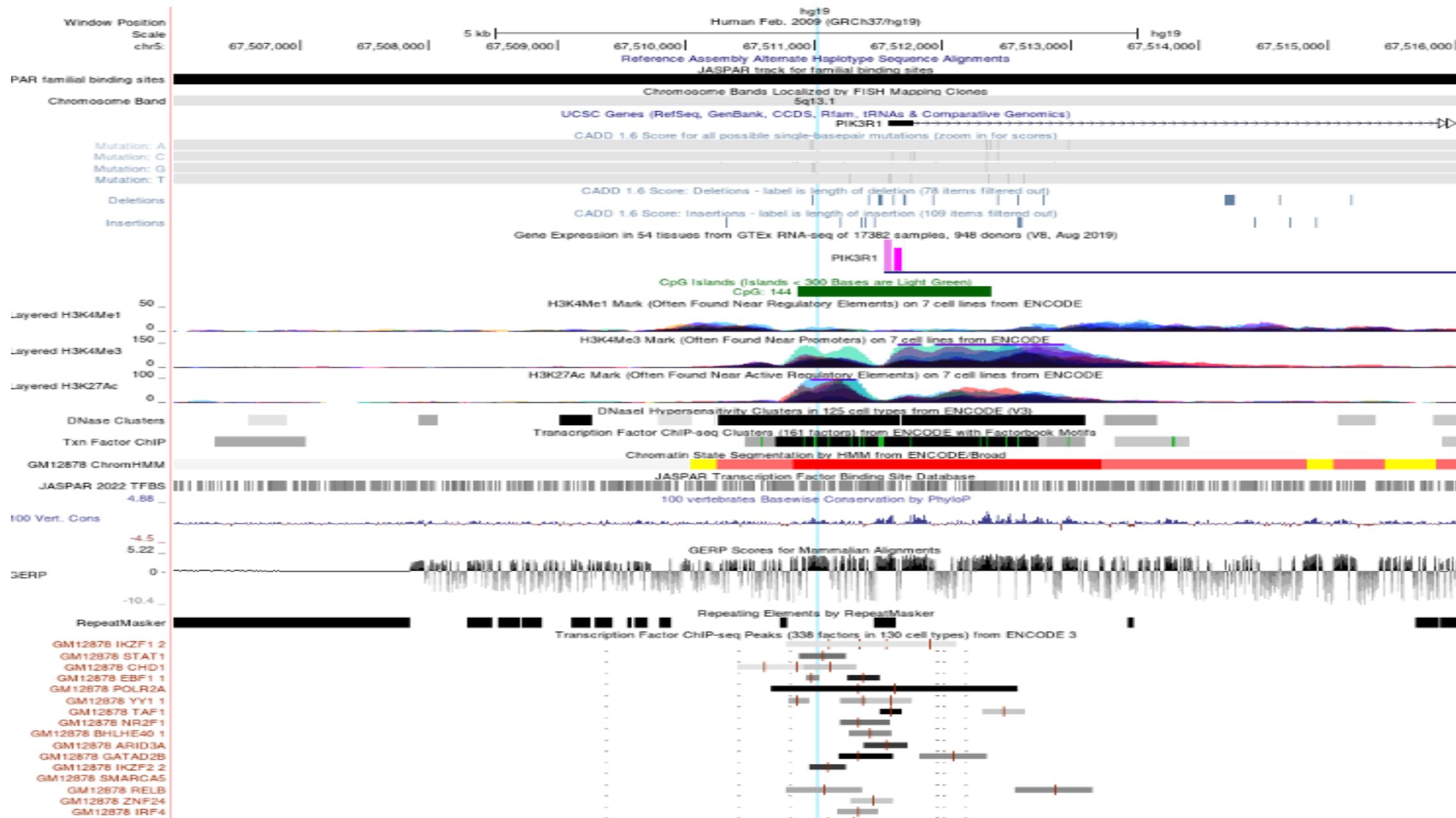
Supplementary Figure S15. *DLG1*: 3_197024641_C_T



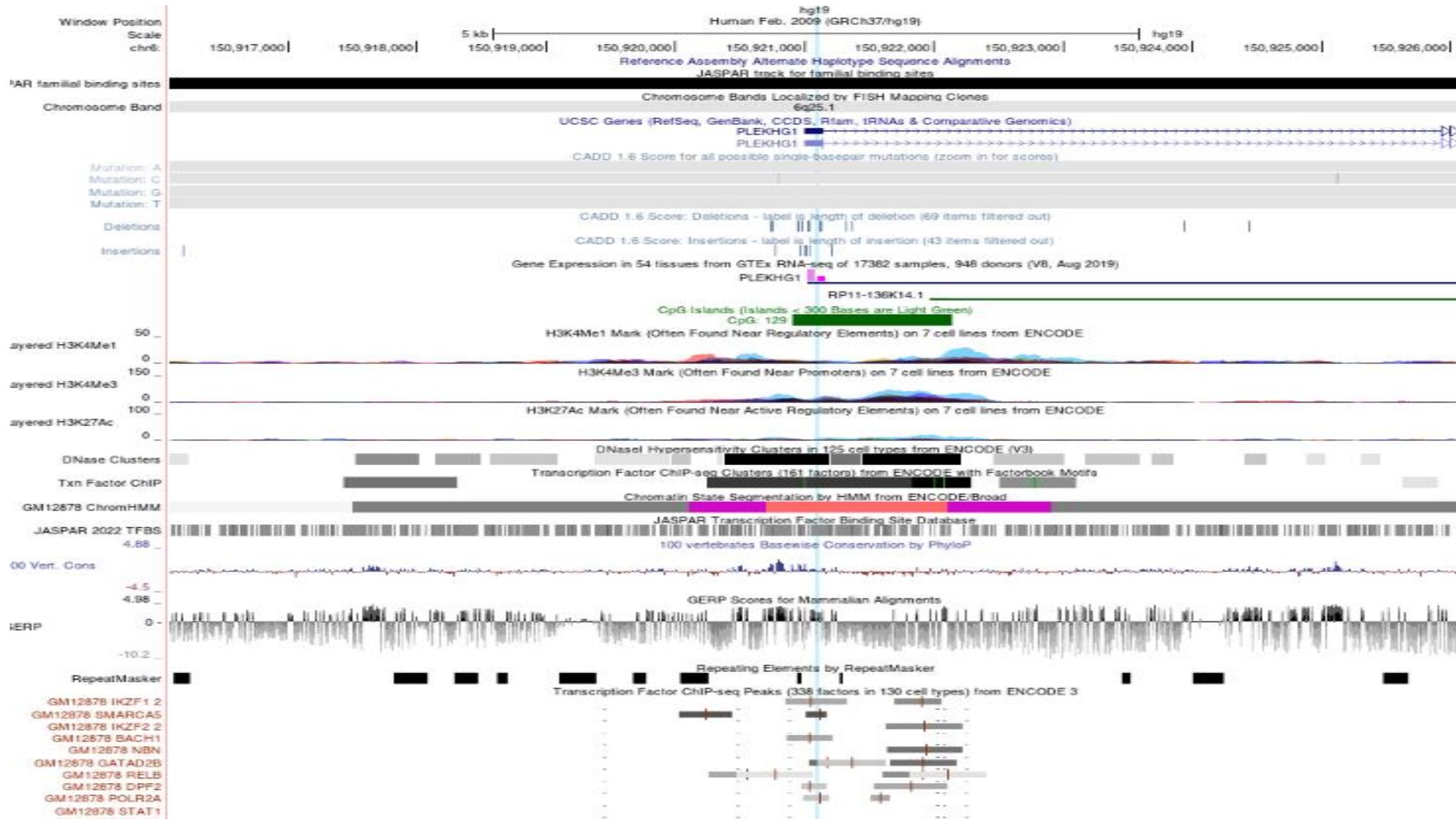
Supplementary Figure S16. *CAMK2D*: 4_114682943_TCCTCCTCCGCG_T



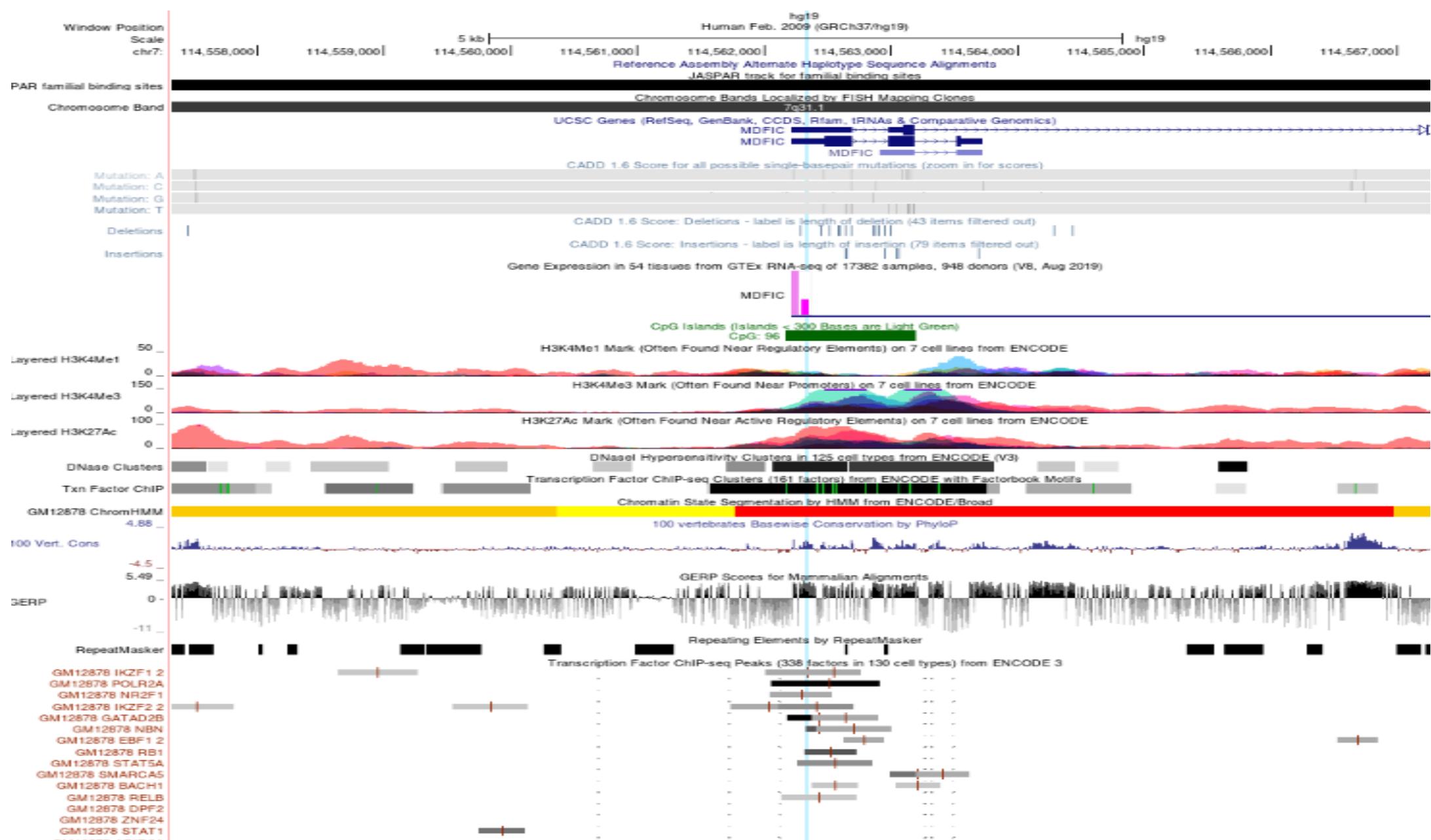
Supplementary Figure S17. *ING2*: 4_184425877_C_A



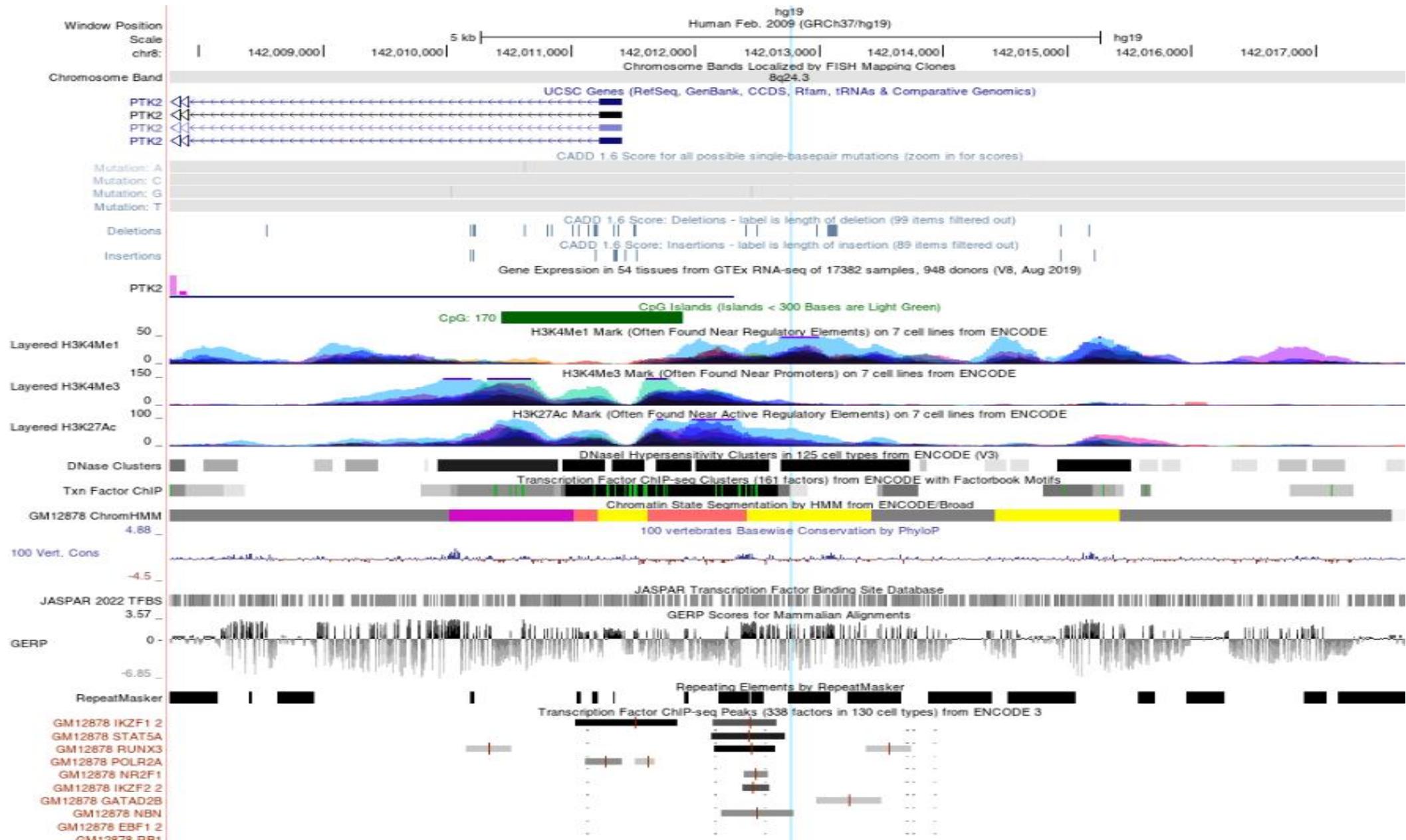
Supplementary Figure S18. *PIK3R1*: 5_67511017_G_C



Supplementary Figure S19. *PLEKHG1*: 6_150921086_G_A



Supplementary Figure S20. *MDFIC*: 7_114562322_C_G



Supplementary Figure S21. *PTK2*: 8_142012766_C_T