Screenshots of Variants chosen

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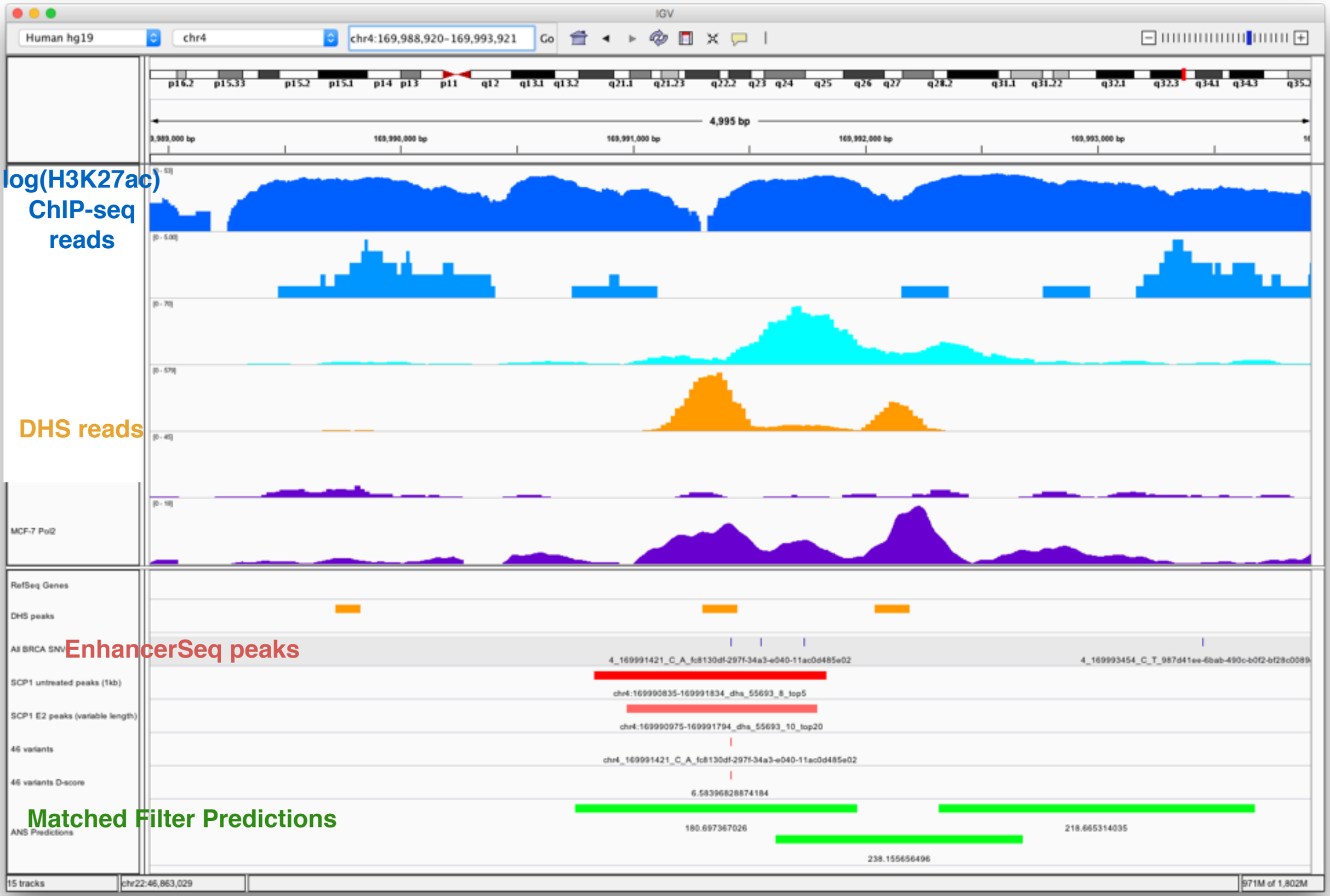
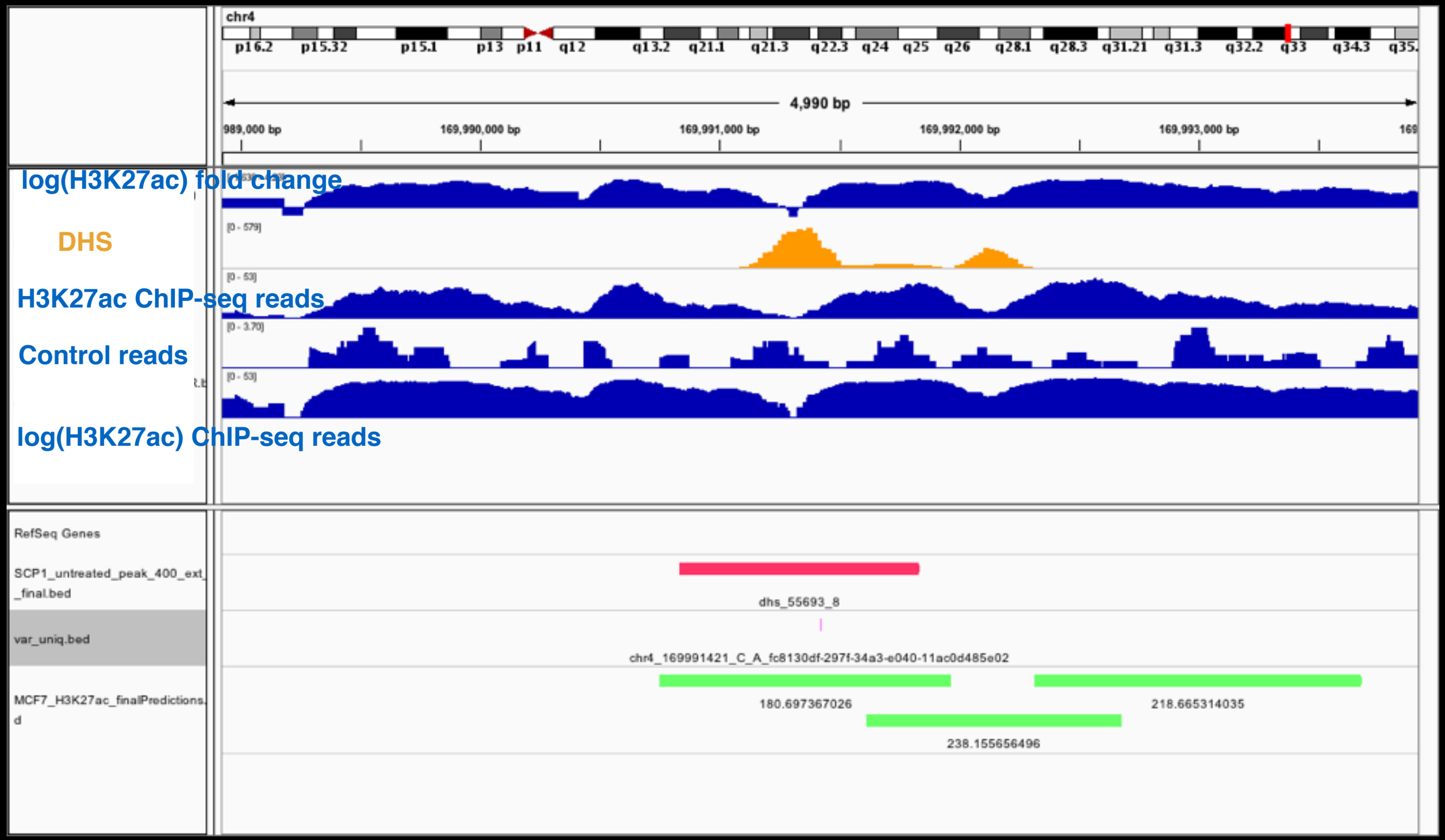
P3-VAR

Region1


IGV
Human hg19
chr4
chr4:169,988,920-169,993,921
Go
4,995 bp
169,990,000 bp
169,991,000 bp
169,992,000 bp
169,993,000 bp
log(H3K27ac) ChIP-seq reads
DHS reads
MCF-7 Pol2
RefSeq Genes
DHS peaks
All BRCA SNV
EnhancerSeq peaks
4_169991421_C_A_fc8130df-297f-34a3-e040-11ac0d485e02
4_169993454_C_T_987d41ee-6bab-490c-b0f2-bf28c0089
SCP1 untreated peaks (1kb)
chr4:169990835-169991834_dhs_55693_8_top5
SCP1 E2 peaks (variable length)
chr4:169990975-169991794_dhs_55693_10_top20
46 variants
chr4_169991421_C_A_fc8130df-297f-34a3-e040-11ac0d485e02
46 variants D-score
6.58396828874184
Matched Filter Predictions
ANS Predictions
180.697367026
218.665314035
238.155656496
15 tracks
chr22:46,863,029
971M of 1,802M

Same region1

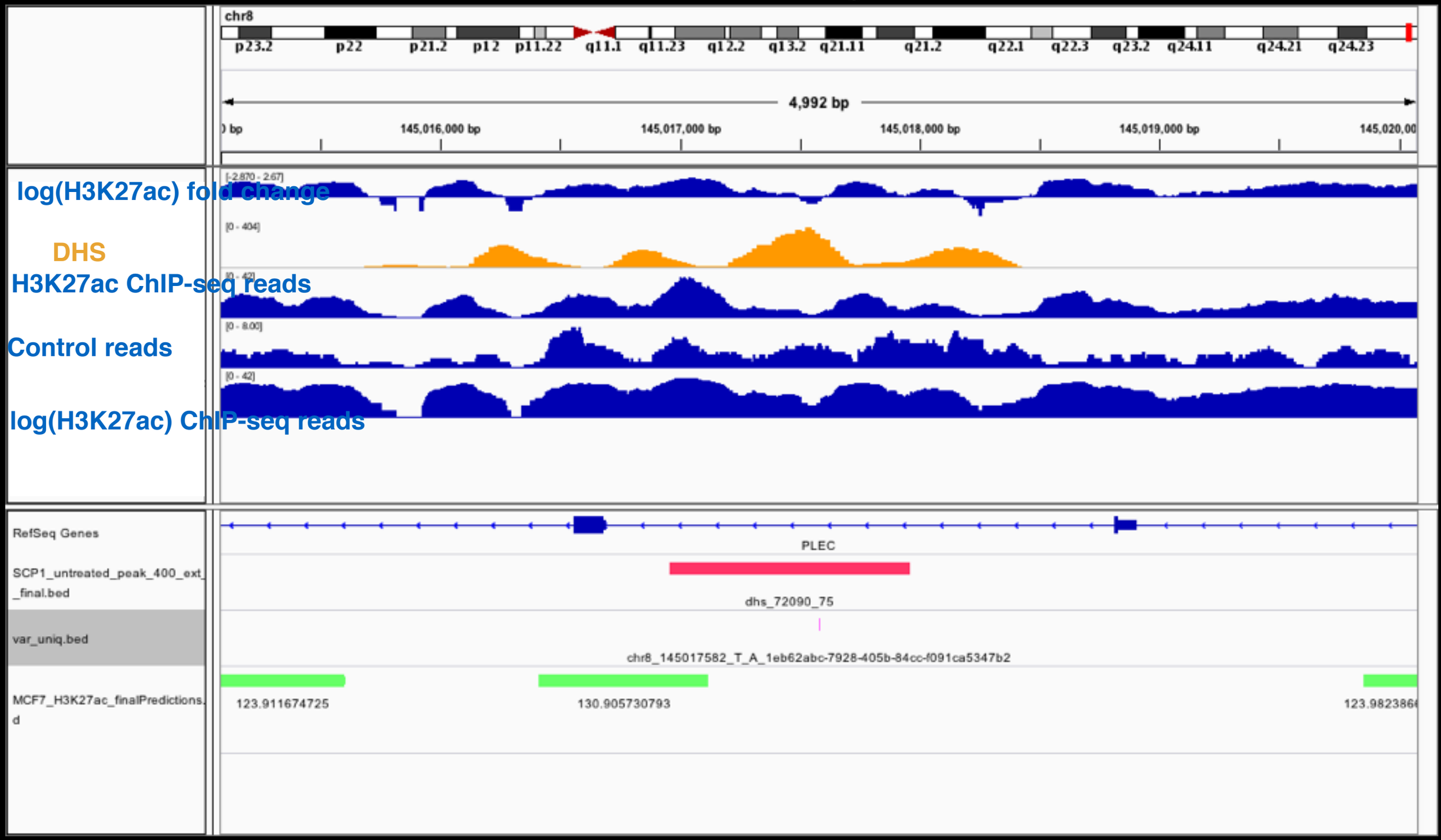


Good prediction and enhancer to test/vary.

Region2

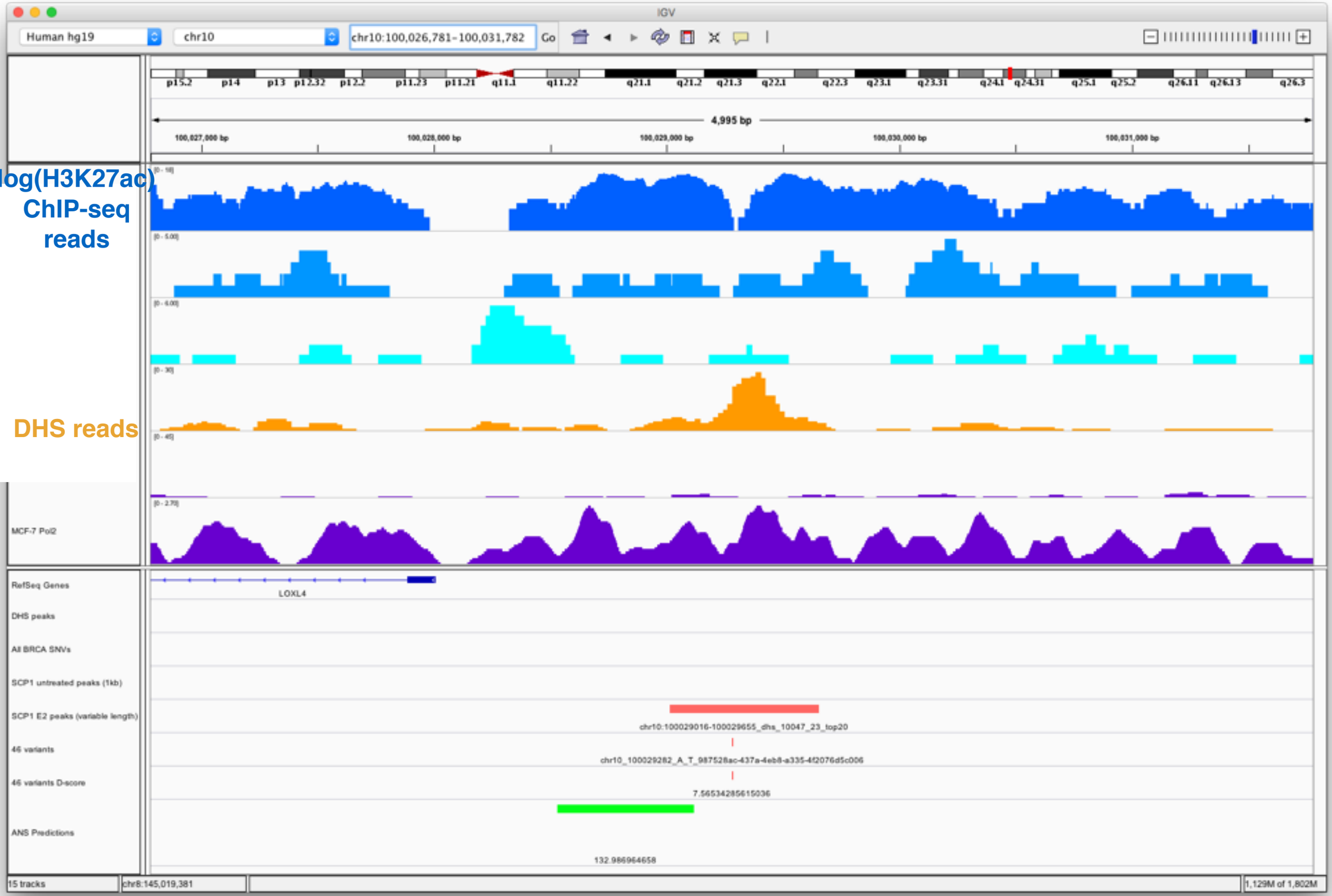

IGV
Human hg19
chr8
chr8:145,015,081-145,020,082
Go
4,995 bp
145,016,000 bp
145,017,000 bp
145,018,000 bp
145,019,000 bp
145,020,000
log(H3K27ac) ChIP-seq reads
DHS reads
MCF-7 Pol2
RefSeq Genes
PLEC
MIR661
DHS peaks
All BRCA SNVs
8_145017582_T_A_1eb62abc-7928-405b-84cc-f091ca5347b2
SCP1 untreated peaks (1kb)
chr8:145016962-145017961_dhs_72090_75_top20
SCP1 E2 peaks (variable length)
46 variants
chr8_145017582_T_A_1eb62abc-7928-405b-84cc-f091ca5347b2
46 variants D-score
4.605219929002
ANS Predictions
123.911674725
130.905730793
123.98238661
15 tracks
chr4:169,992,357
1,056M of 1,802M

Same region2

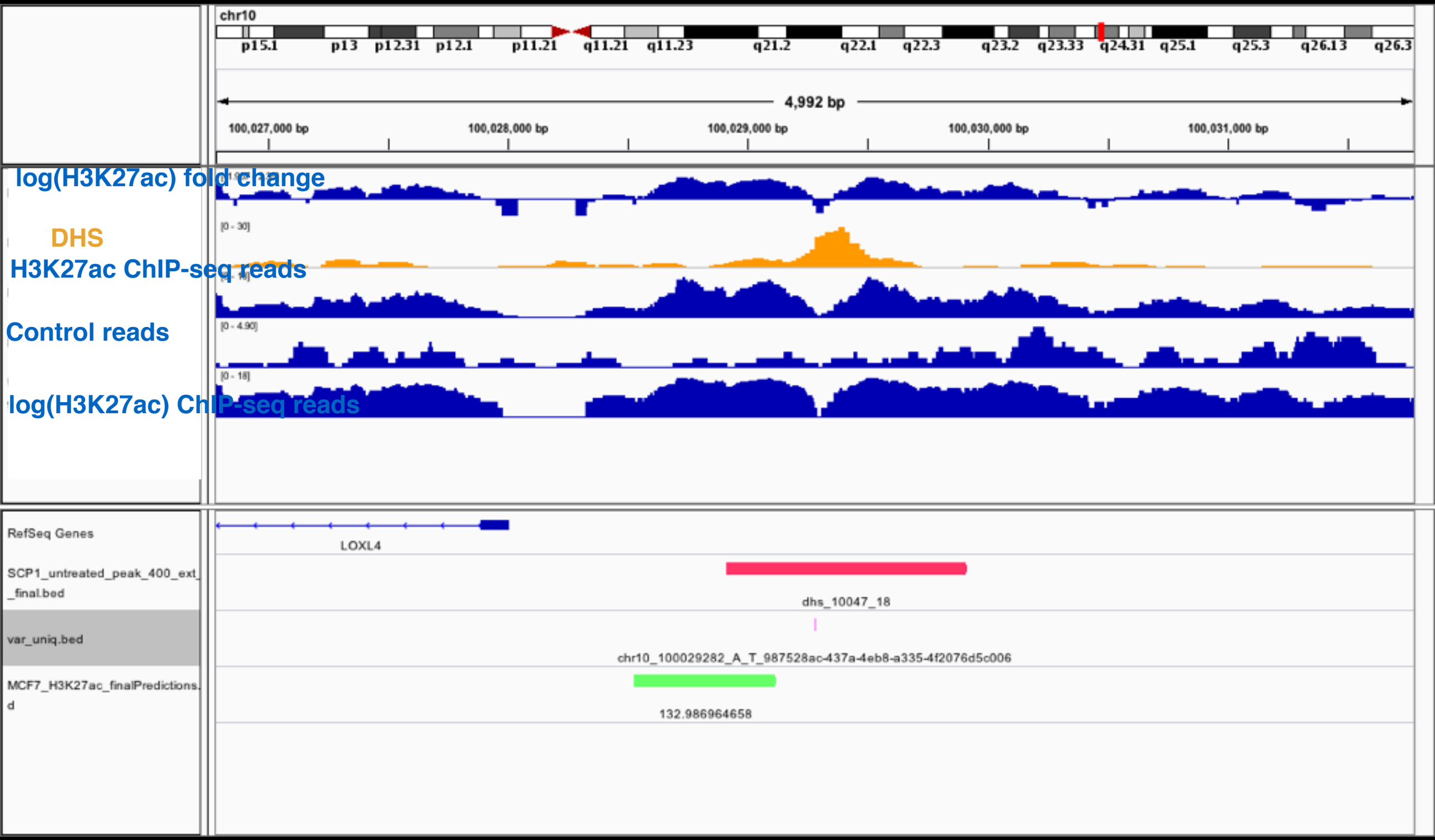


Prediction looks better on log(fold change) plot than EnhancerSeq peak - cannot change our prediction

Region3

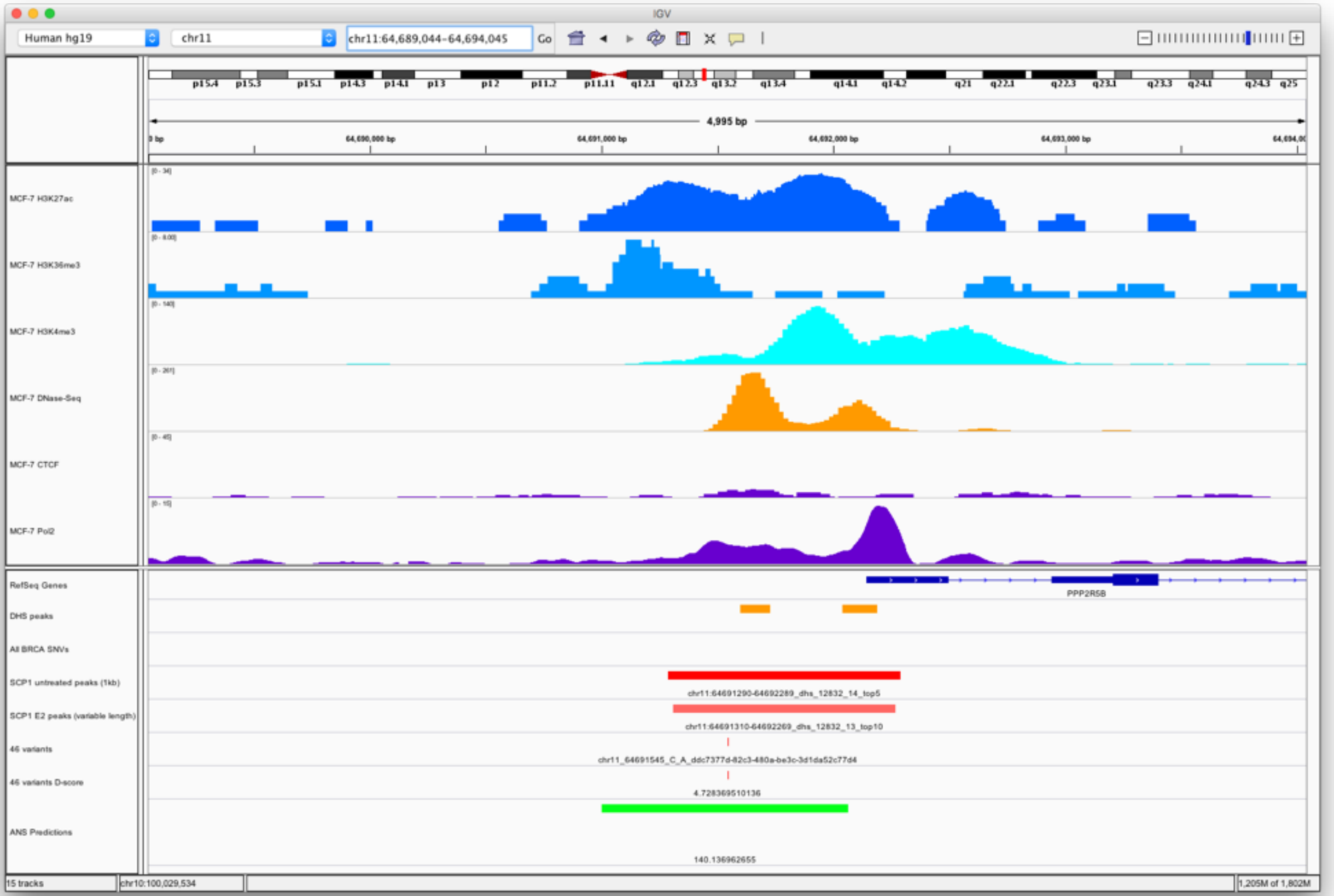

IGV
Human hg19
chr10
chr10:100,026,781-100,031,782
Go
4,995 bp
100,027,000 bp
100,028,000 bp
100,029,000 bp
100,030,000 bp
100,031,000 bp
log(H3K27ac) ChIP-seq reads
DHS reads
MCF-7 Pol2
RefSeq Genes
LOXL4
DHS peaks
All BRCA SNVs
SCP1 untreated peaks (1kb)
SCP1 E2 peaks (variable length)
chr10:100029016-100029655_dhs_10047_23_top20
46 variants
chr10_100029282_A_T_98752fac-437a-4eb8-a335-4f2076d5c006
46 variants D-score
7.56534285615036
ANS Predictions
132.986964658
15 tracks
chr8:145,019,381
1,129M of 1,802M

Same region



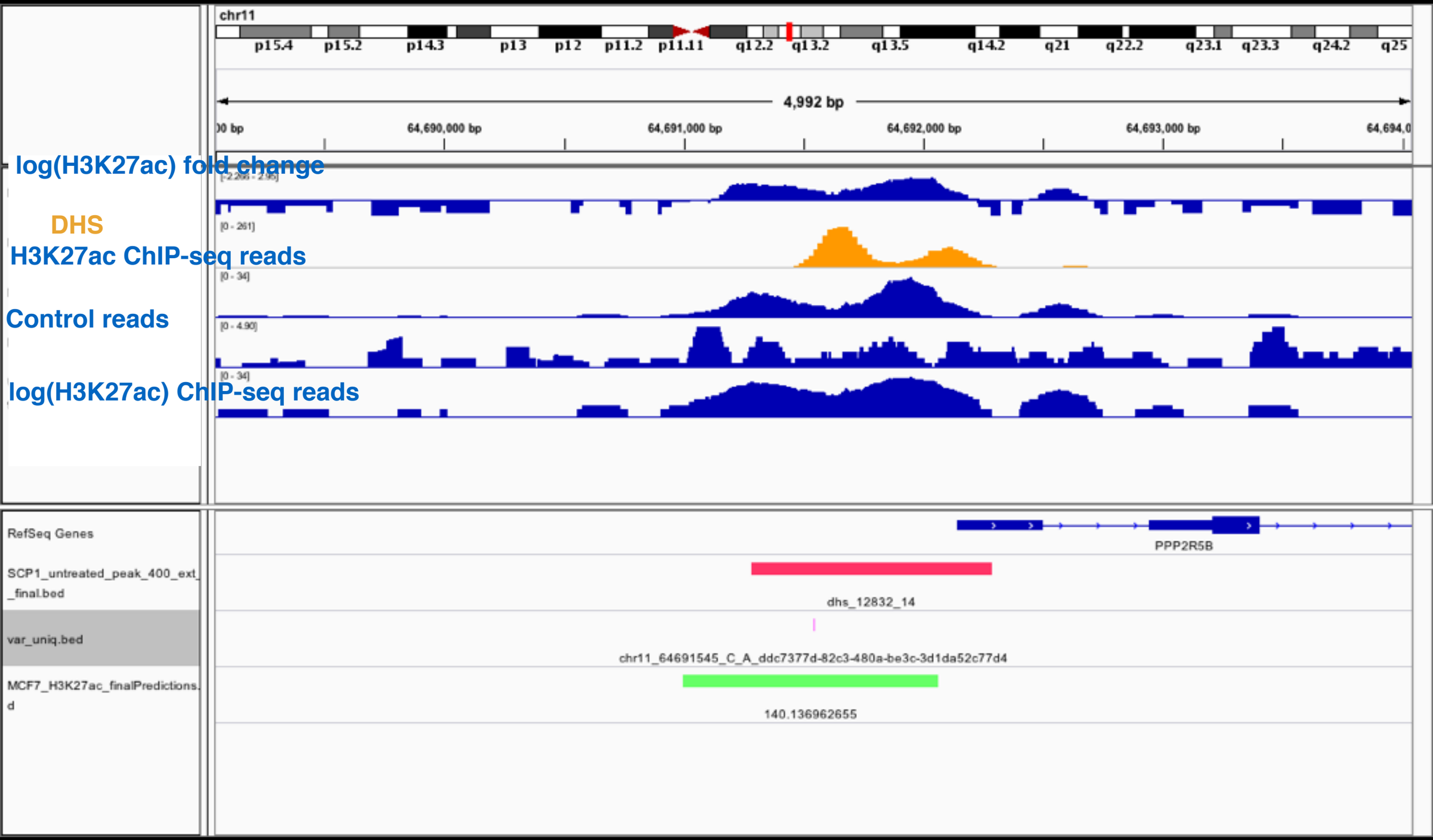
Most probably the EnhancerSeq peak fell below the MF score cutoff

Region4



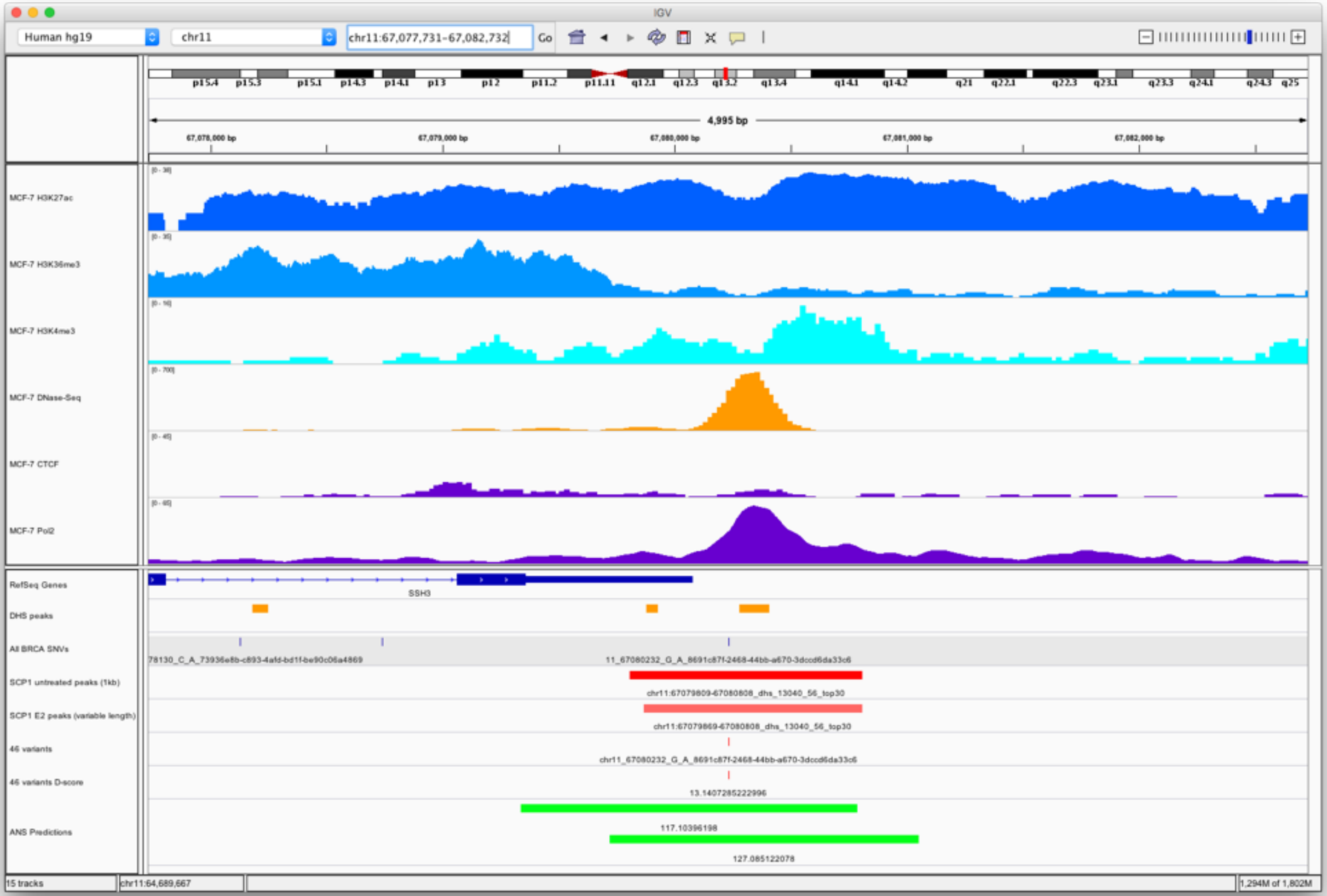
Possibly a promoter region

Same Region4

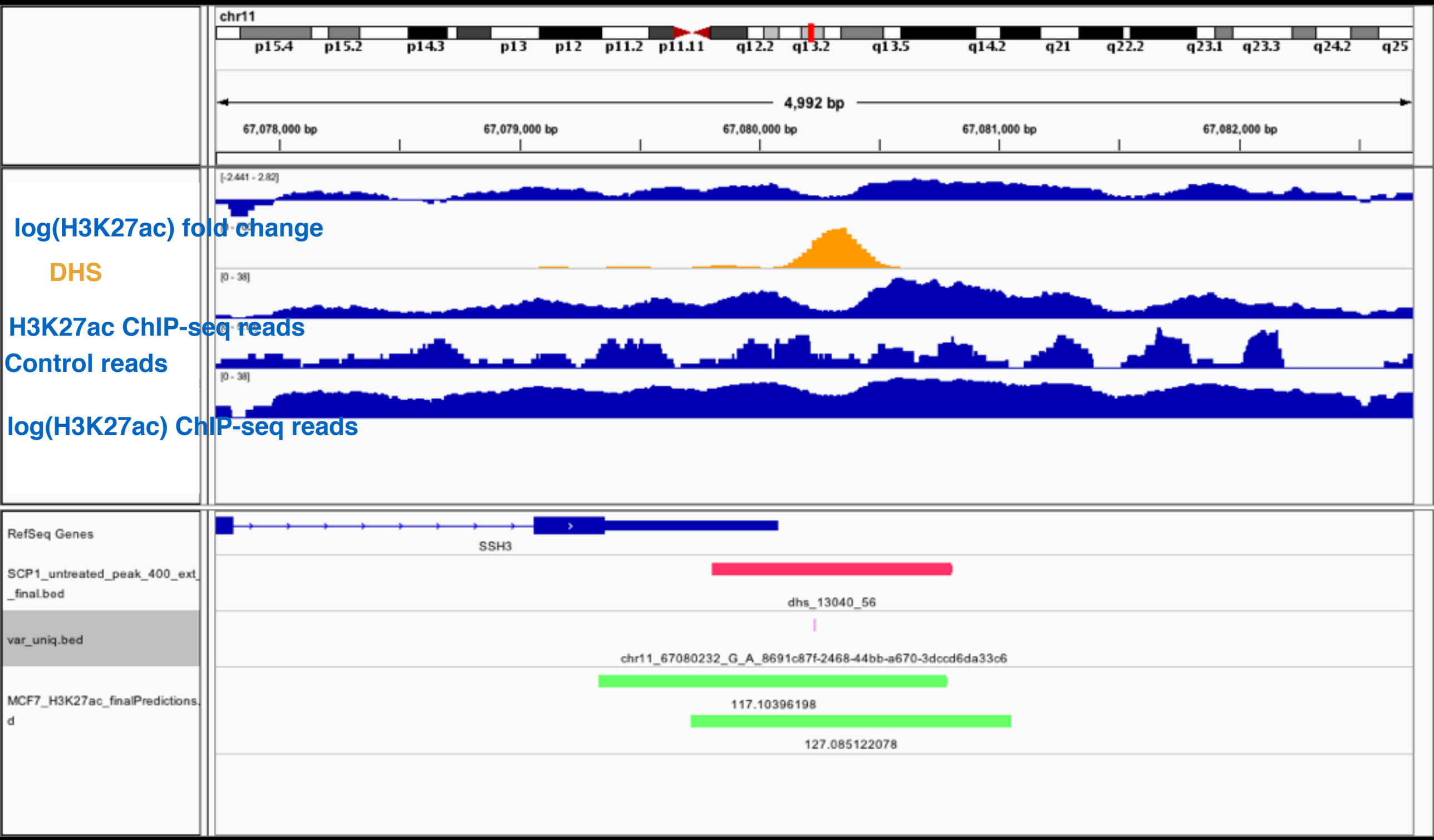


Good prediction by all measures

Region5

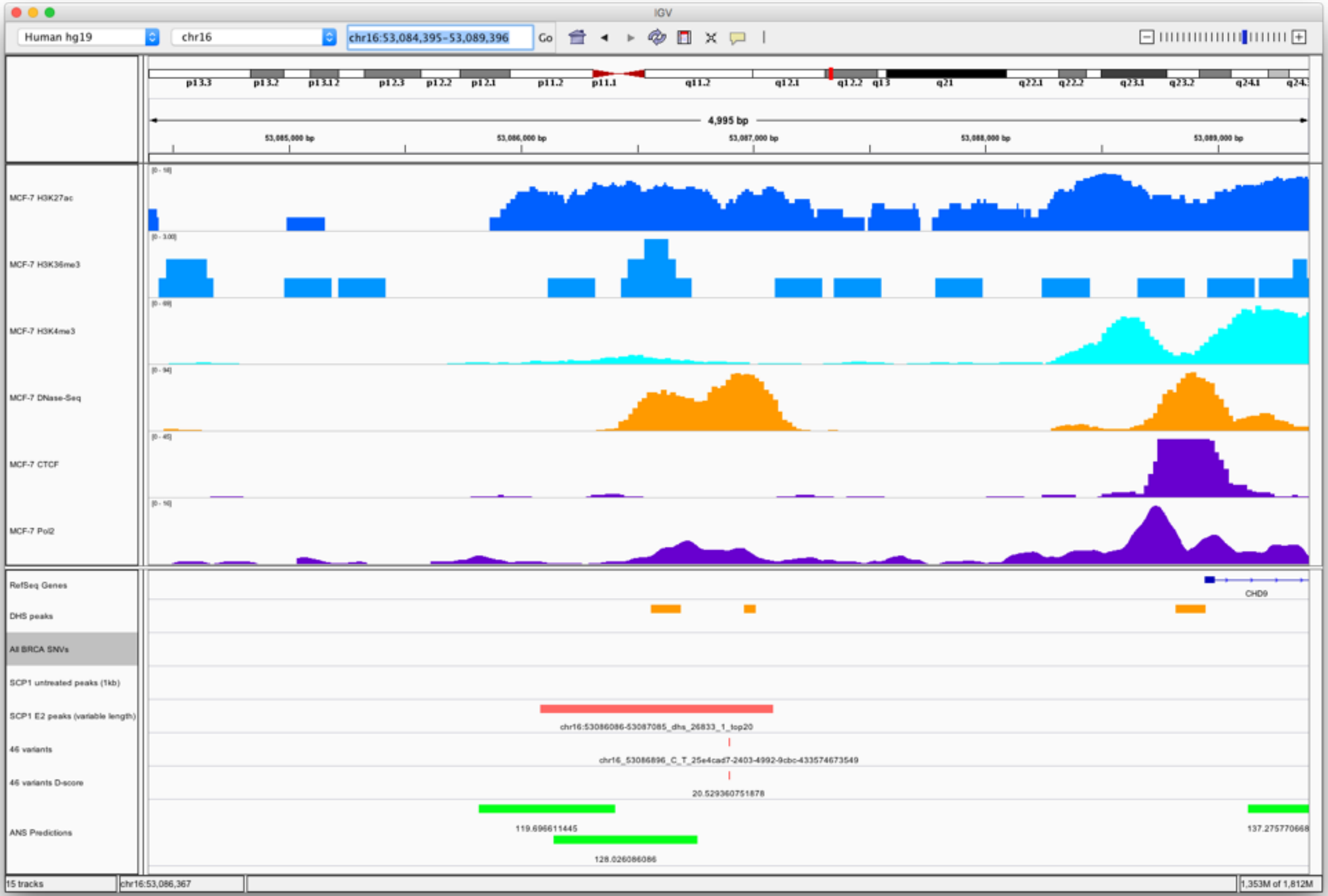

IGV
Human hg19
chr11
chr11:67,077,731-67,082,732
Go
p15.4 p15.3 p15.1 p14.3 p14.1 p13 p12 p11.2 p11.11 q12.1 q12.3 q13.2 q13.4 q14.1 q14.2 q21 q22.1 q22.3 q23.1 q23.3 q24.1 q24.3 q25
4,995 bp
67,078,000 bp
67,079,000 bp
67,080,000 bp
67,081,000 bp
67,082,000 bp
MCF-7 H3K27ac
MCF-7 H3K36me3
MCF-7 H3K4me3
MCF-7 DNase-Seq
MCF-7 CTCF
MCF-7 Pol2
RefSeq Genes
SSH3
DHS peaks
All BRCA SNVs
78130_C_A_73936e8b-c893-4afd-bd1f-be90c06a4869
11_67080232_G_A_8691c87f-2468-44bb-a670-3dccd6da33c6
SCP1 untreated peaks (1kb)
chr11:67079809-67080808_dhs_13040_56_top30
SCP1 E2 peaks (variable length)
chr11:67079869-67080808_dhs_13040_56_top30
46 variants
chr11_67080232_G_A_8691c87f-2468-44bb-a670-3dccd6da33c6
46 variants D-score
13.1407285222996
ANS Predictions
117.10396198
127.085122078
15 tracks
chr11:64,689,667
1,294M of 1,802M

Same Region5



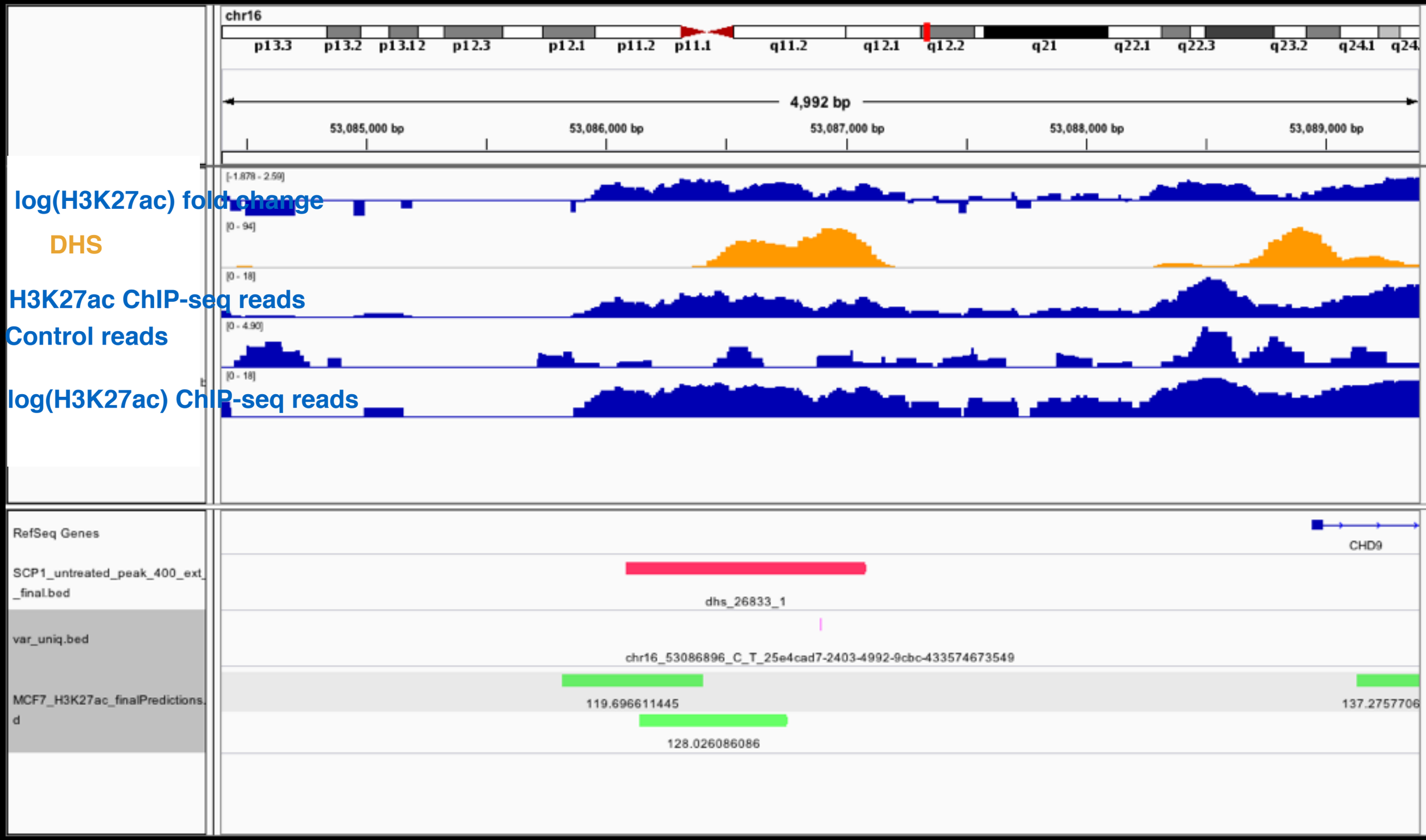
Good prediction by all measures

Region6



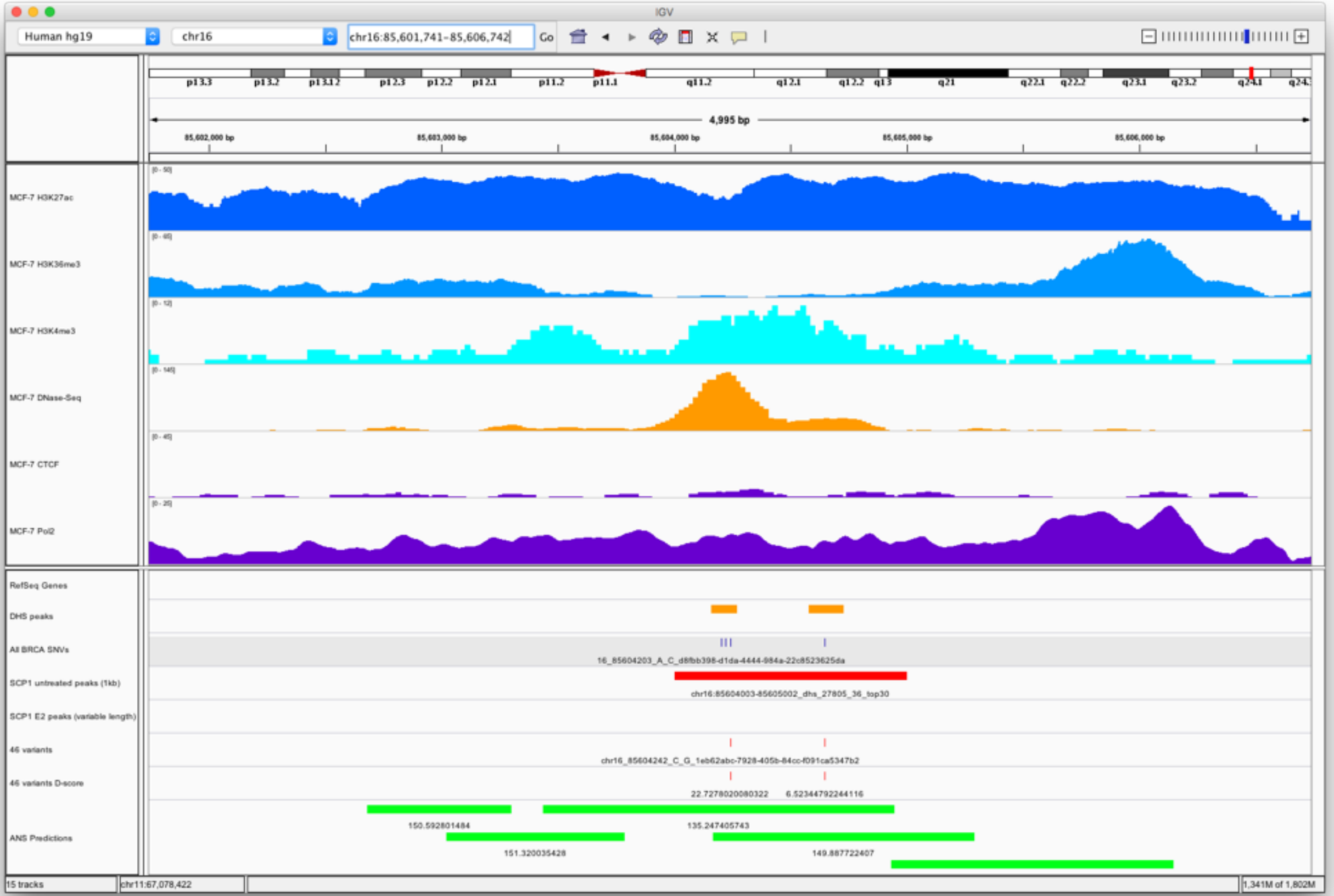
High D-score

Same Region6



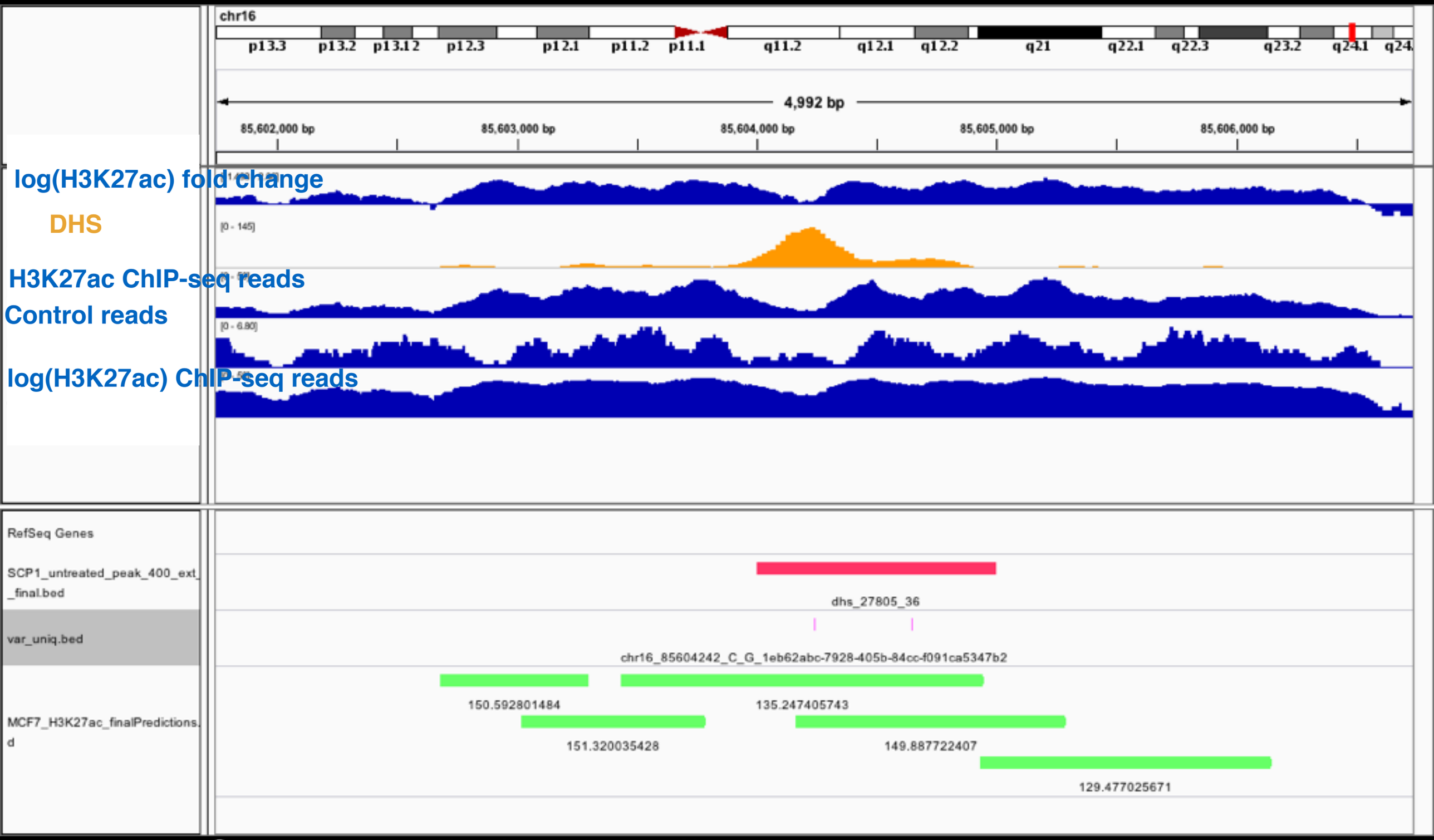
Most probably the EnhancerSeq peak fell below the MF score cutoff/flat in that region.

Region7



High D-score

Same Region7

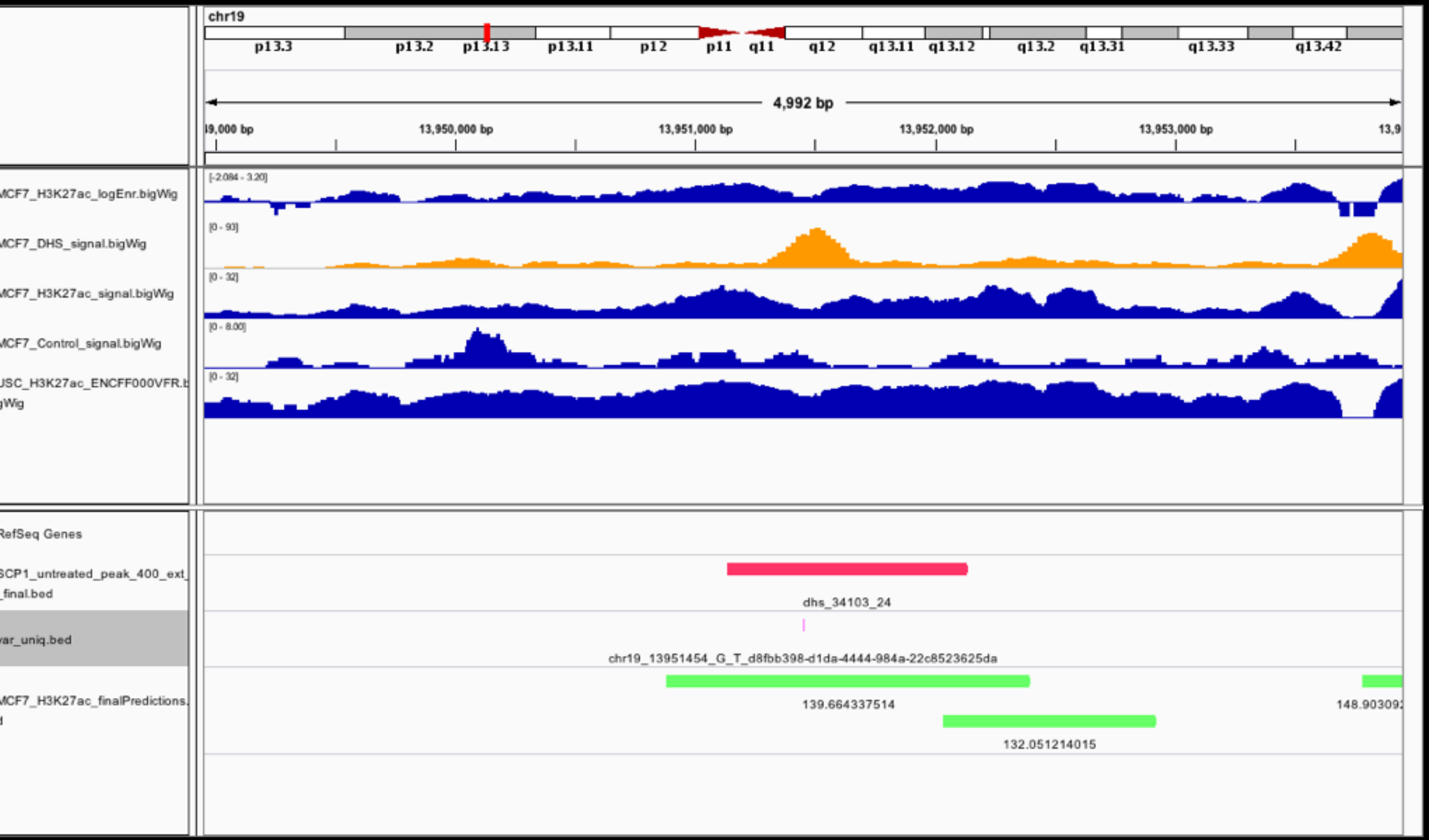


Good prediction by all measures

Region8

High D-score

Same Region8

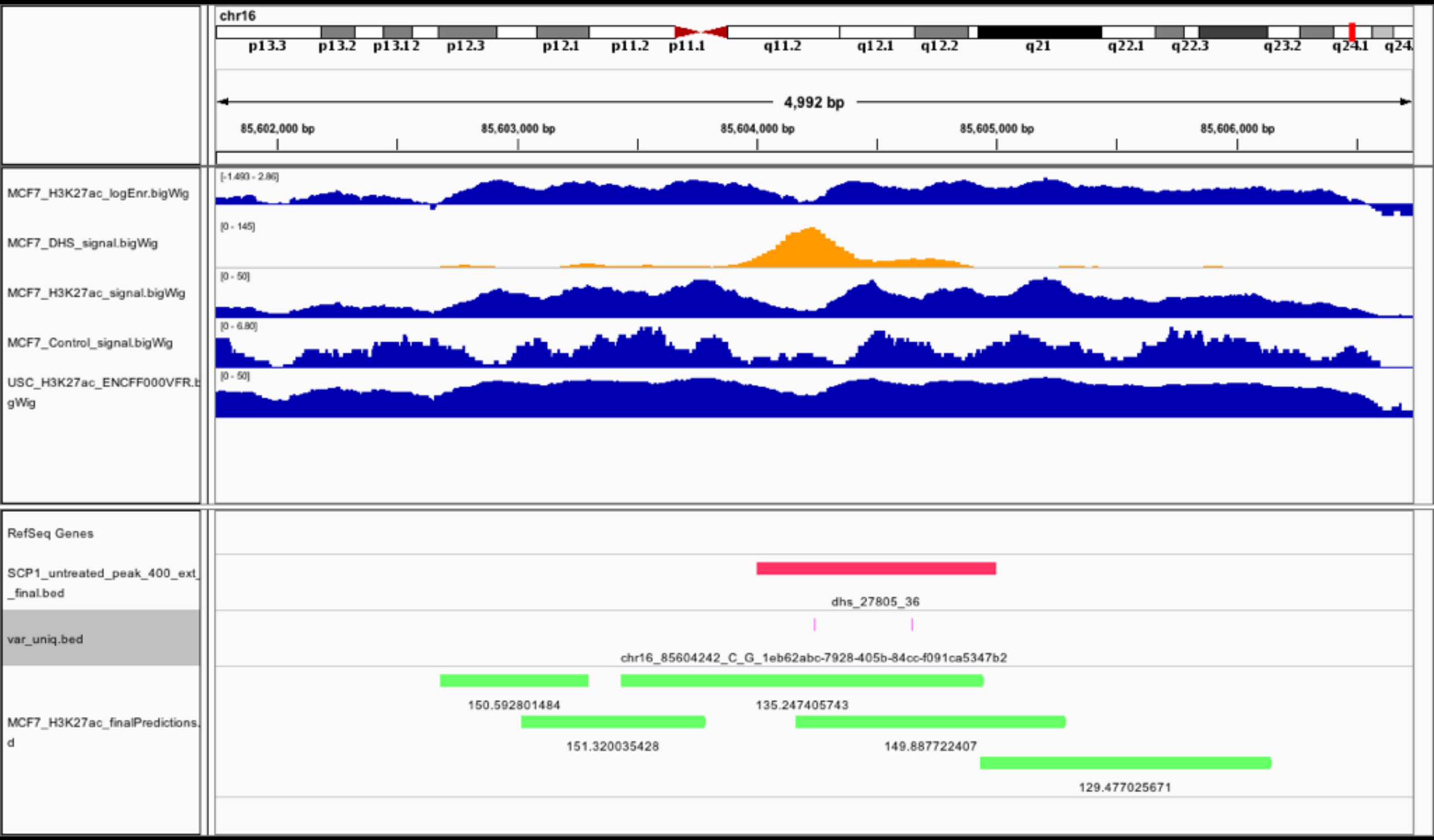


Good prediction by all measures

Region8

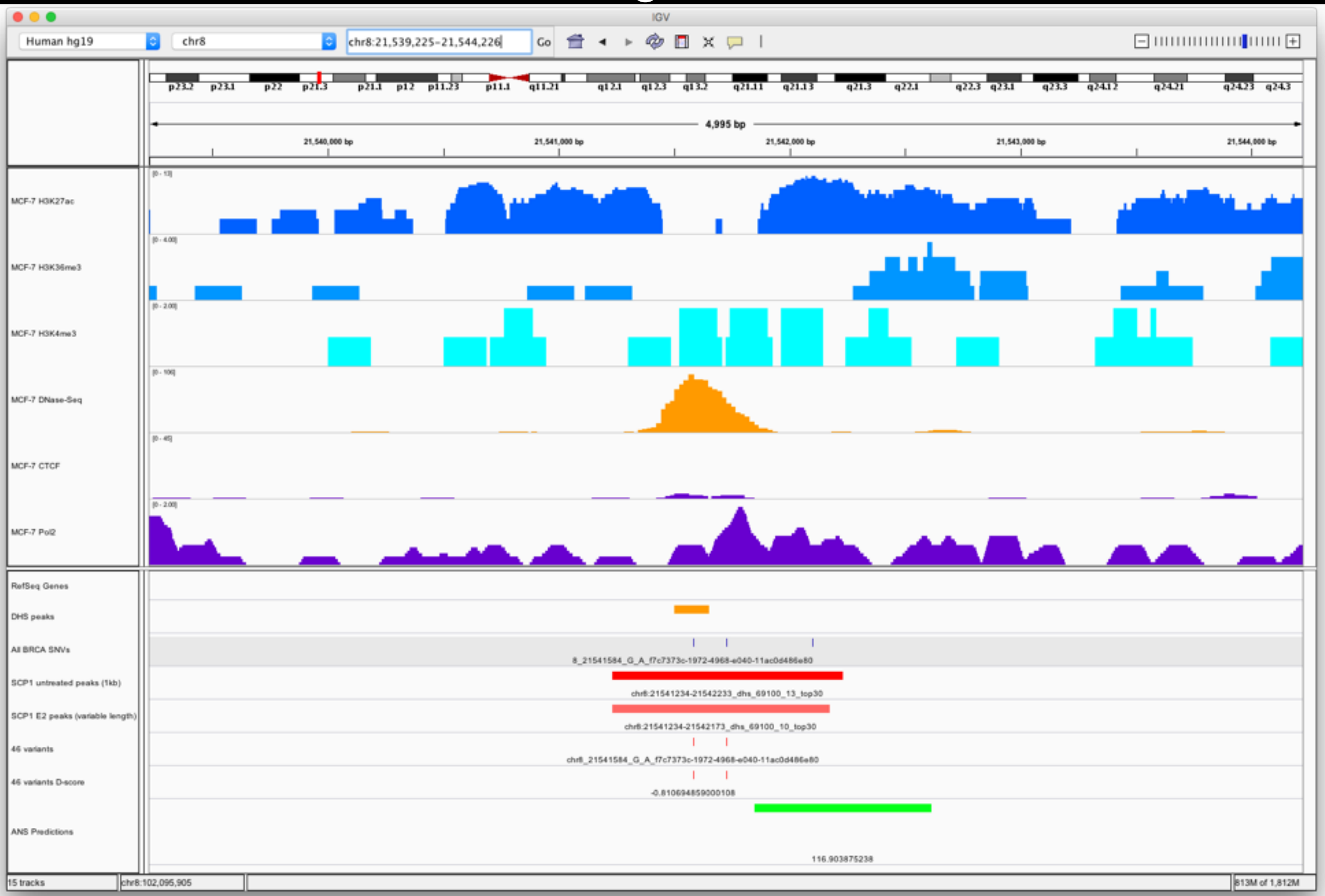

IGV
Human hg19
chr19
chr19:56,108,576-56,113,577
Go
p13.3
p13.2
p13.13
p13.11
p12
p11
q11
q12
q13.11
q13.12
q13.2
q13.31
q13.32
q13.33
q13.41
q13.42
q13.43
4,995 bp
56,109,000 bp
56,110,000 bp
56,111,000 bp
56,112,000 bp
56,113,000 bp
MCF-7 H3K27ac
MCF-7 H3K36me3
MCF-7 H3K4me3
MCF-7 DNase-Seq
MCF-7 CTCF
MCF-7 Pol2
RefSeq Genes
FIZ1
ZNF524
DHS peaks
All BRCA SNVs
10_C_T_5b7d5767-2037-4175-aec9-886776343181
19_56110377_C_T_cc4f9175-a8c8-424a-99d7-8bd11cae0e52
SCP1 untreated peaks (1kb)
chr19:56110709-56111708_dhs_36376_51_top5
SCP1 E2 peaks (variable length)
chr19:56110909-56111728_dhs_36376_53_top5
46 variants
chr19_56111077_T_G_f7d70fac-c76f-399e-e040-11ac0c482f9c
46 variants D-score
1.3889409235811
ANS Predictions
145.681563694
146.900055845
15 tracks
chr19:56,110,881
1,439M of 1,812M

Same Region8



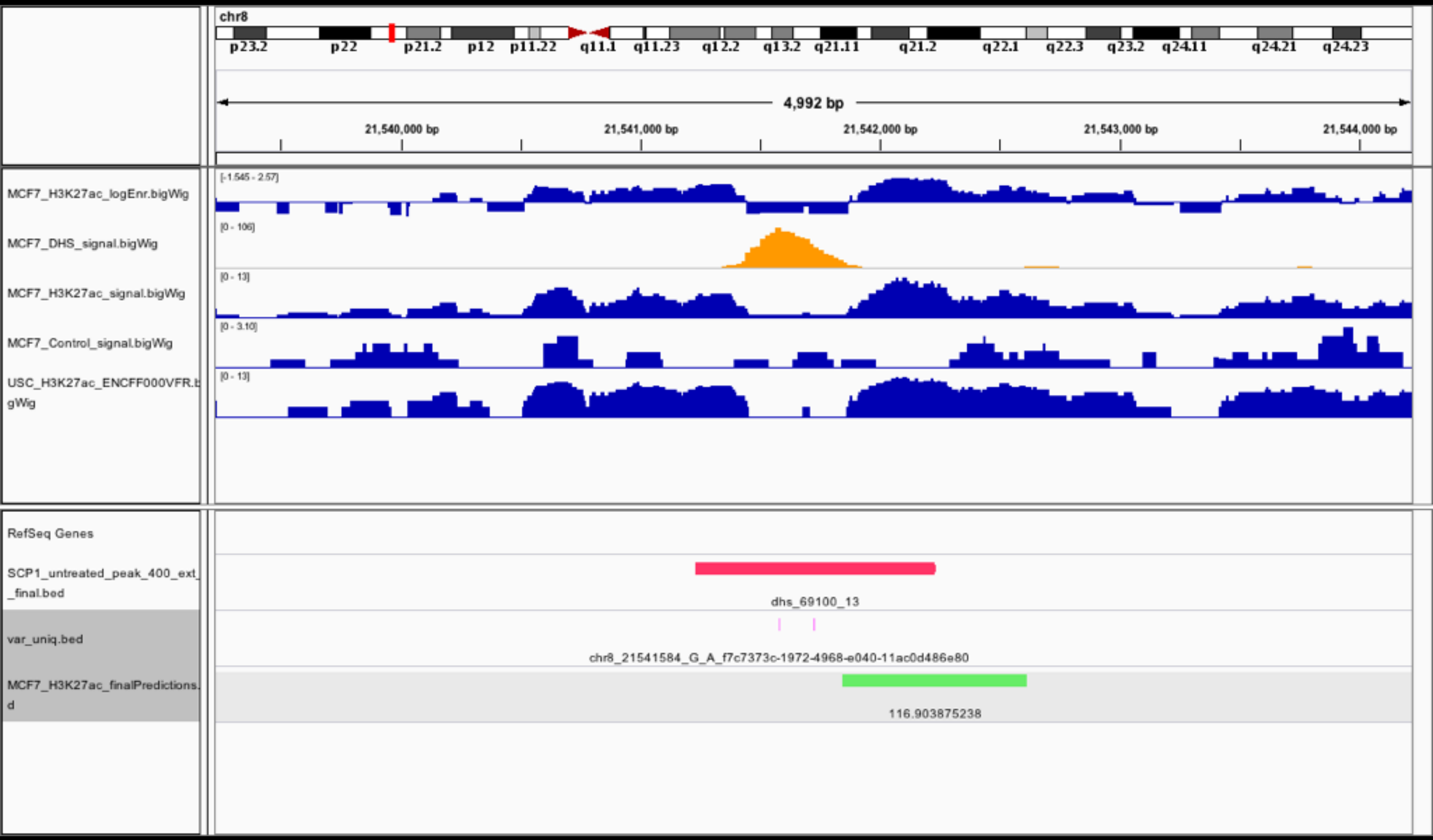
Now focusing just on what looked like bad predictions

Region9



Negative D-score (benefit)

Same Region9

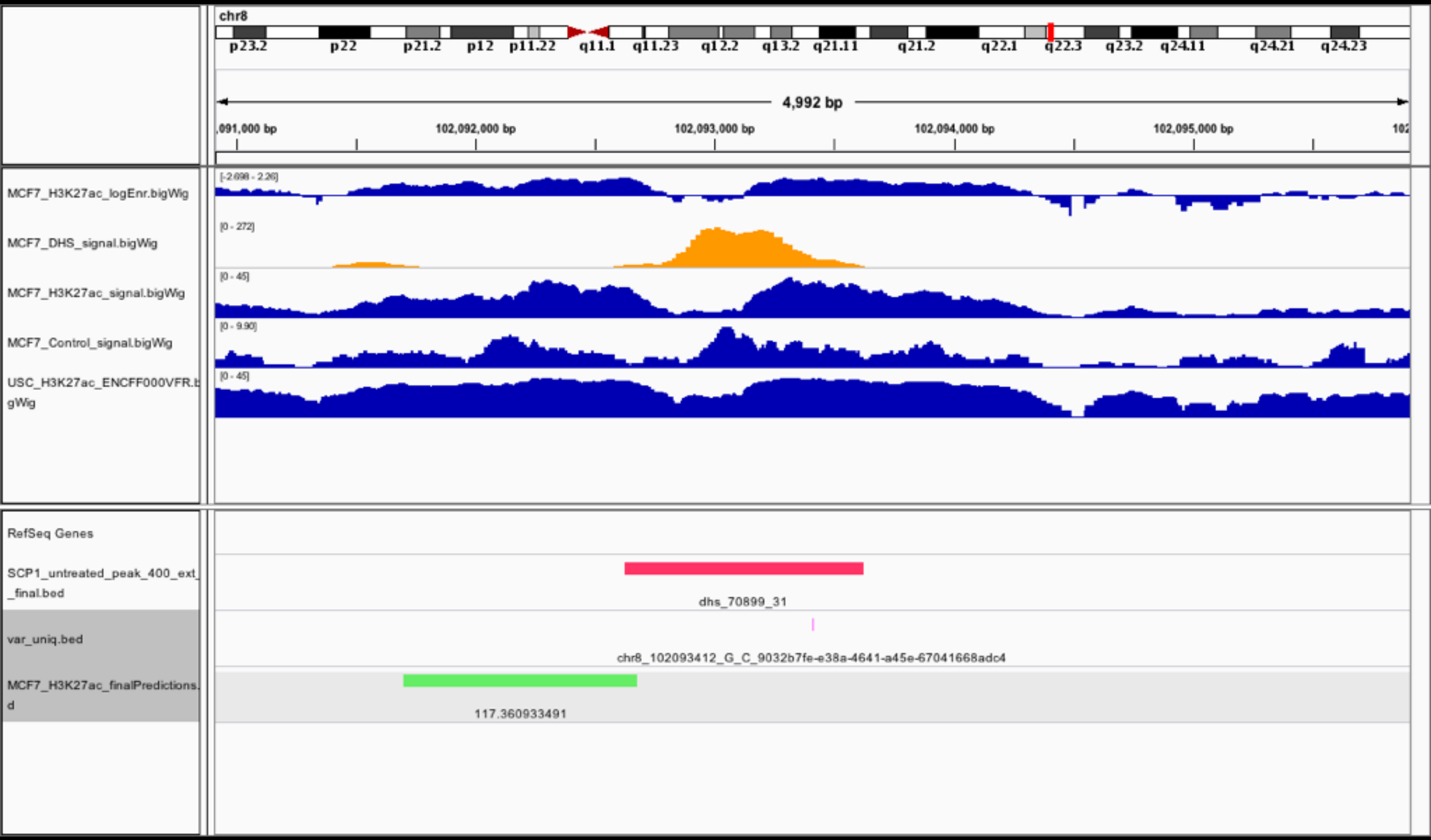


Not such a bad presentation in my opinion (DHS and H3K27ac just point to different regions).

Region 10

Good enhancer, variant off-centered

Same Region 10



The DHS peak is below enhancer prediction threshold