Genome-wide association study of copy number variation in 16,000 cases of eight common diseases and 3,000 shared controls

Supplementary File 2

Version 2.0.0

The Wellcome Trust Case Control Consortium

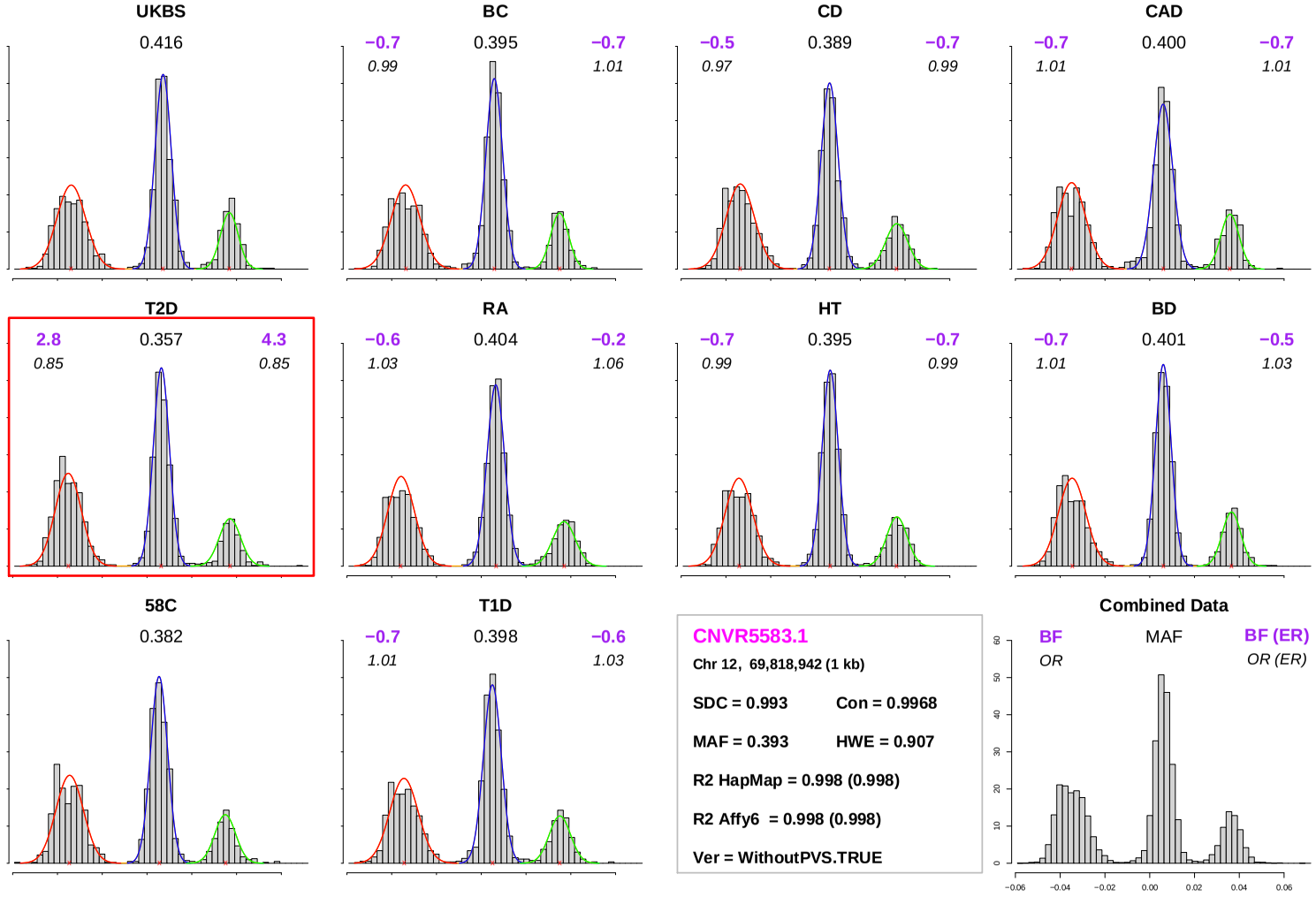
December 18, 2009

Plots of CNV calls and association testing results produced by the CNVCALL/CNVTEST approach

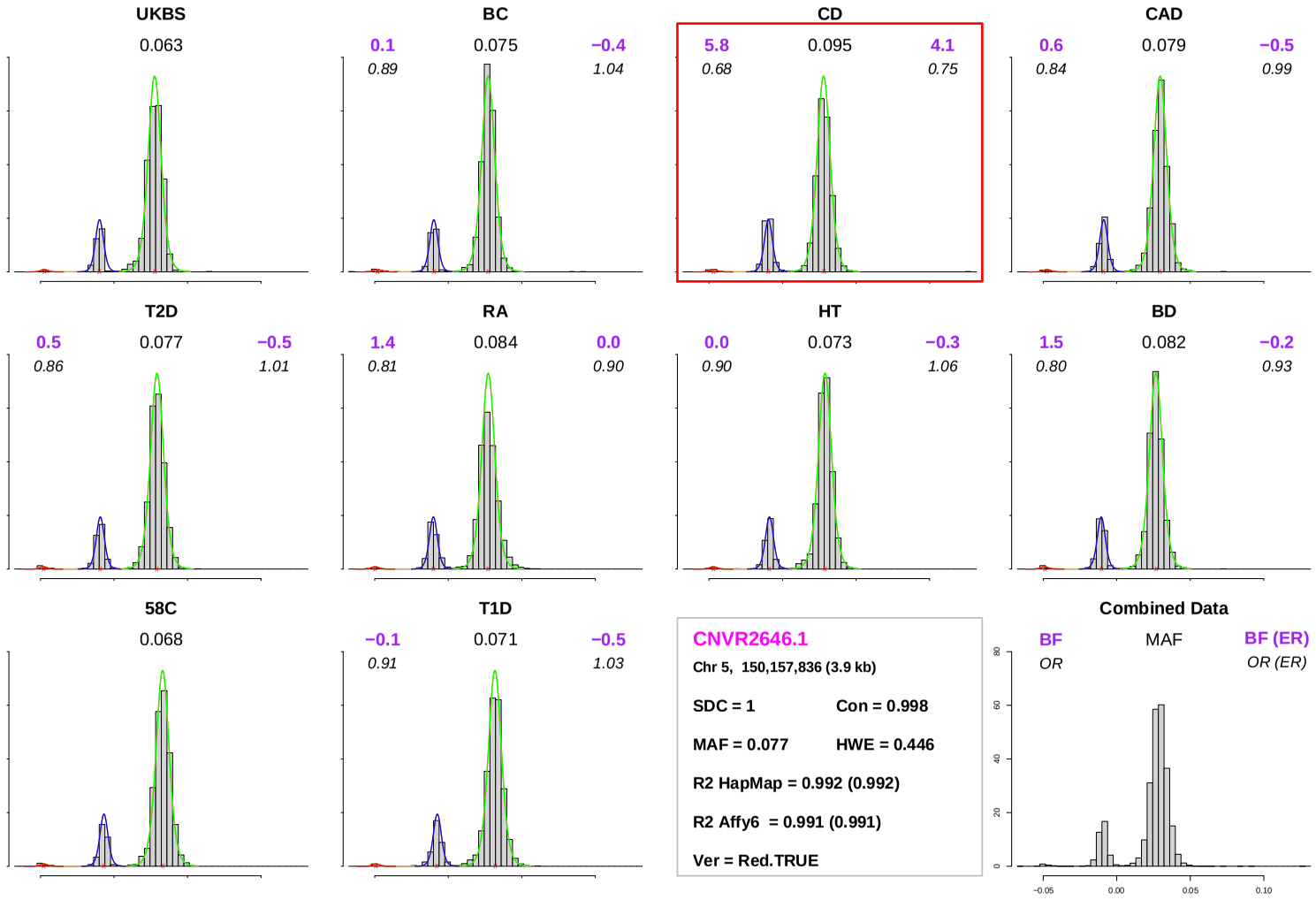
The following plots show the CNV intensity distributions for each cohort at all of the CNVs in Table 3 and Supplementary Table 13. For each CNV the CNV intensity distributions for each cohort are given in a separate sub-plot. The model fit (continuous coloured line) for each cohort is overlaid onto the intensity distributions. The means and variances of the CNV classes differ between cohorts but class proportions are set to the estimate of the overall class proportions. This aids visualization of effects since signals of association will show up as a difference between the model fit and the intensity histogram. The numbers at the top of each plot are as follows. Top left : $\log_{10}$ Bayes Factor (BF) for cohort versus the two control cohorts with associated estimate of the additive odds ratio for increasing copy number given below. Top right : $\log_{10}$ Bayes Factor for cohort versus the set of expanded reference panels (BF (ER)) (see Supplementary Table 9) with the associated estimate of the additive odds ratio for increasing copy number given below. Top middle : minor allele frequency estimate for that cohort. Case cohorts with either a BF or BF(ER) greater than 2.1 are highlighted by a red box around the subplot for that cohort. The CNV intensity distribution across all cohorts is given in the bottom right plot together with a legend for the 5 numbers included in each of the per-cohort subplots. The third subplot from the right on the bottom row gives information about the CNV shown in the rest of the subplots. The CNV names, chromosome, position and length of the CNV are given. The Strict Duplicate Concordance (SDC) is reported which is the ratio of all duplicate calls that agree divided by the total number of duplicates. The mean maximum posterior probability (or confidence (Con)) of the CNV genotype calls is given. The minor allele frequency (MAF) across all cohorts and the p-value for the test of Hardy-Weinberg Equilibrium (HWE) in all cohorts are shown. The maximum R^2 of the CNV to a HapMap SNP in a 1Mb region flanking the CNV is reported together with the maximum R^2 of the CNV to any HapMap SNP in brackets. The maximum R^2 of the CNV to a SNP on the Affymetrix 6.0 chip in a 1Mb region flanking the CNV is reported together with the maximum R^2 of the CNV to any Affymetrix 6.0 chip SNP in brackets. The version (Ver) of normalization used for the CNV is shown at the bottom of the subplot (see Supplementary Table 8 for more details).

Contents

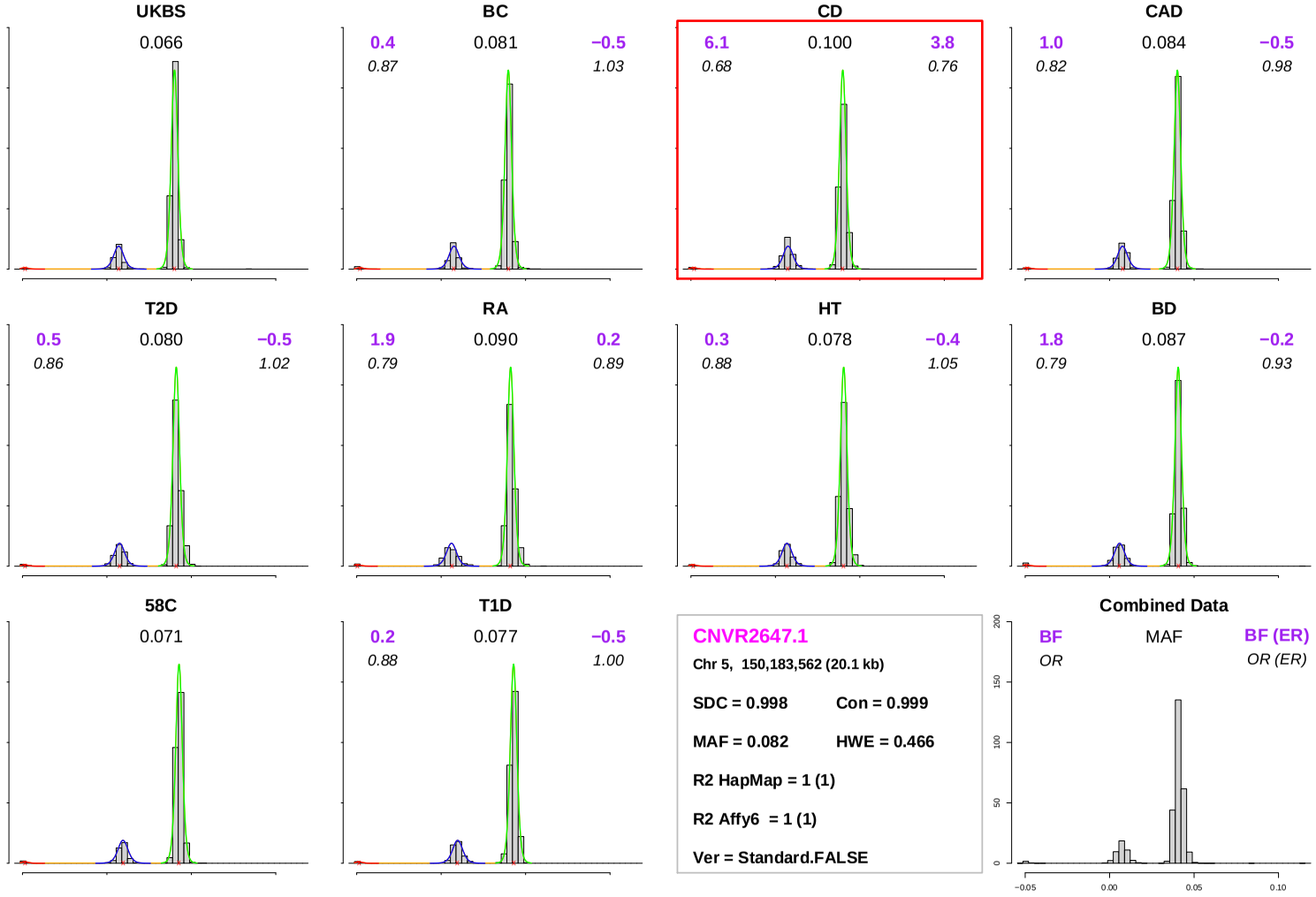
1 CNVR5583.1


UKBS
0.416
BC
−0.7 0.395 −0.7
0.99 1.01
CD
−0.5 0.389 −0.7
0.97 0.99
CAD
−0.7 0.400 −0.7
1.01 1.01
T2D
2.8 0.357 4.3
0.85 0.85
RA
−0.6 0.404 −0.2
1.03 1.06
HT
−0.7 0.395 −0.7
0.99 0.99
BD
−0.7 0.401 −0.5
1.01 1.03
58C
0.382
T1D
−0.7 0.398 −0.6
1.01 1.03
CNVR5583.1
Chr 12, 69,818,942 (1 kb)
SDC = 0.993 Con = 0.9968
MAF = 0.393 HWE = 0.907
R2 HapMap = 0.998 (0.998)
R2 Affy6 = 0.998 (0.998)
Ver = WithoutPVS.TRUE
Combined Data
BF MAF BF (ER)
OR OR (ER)

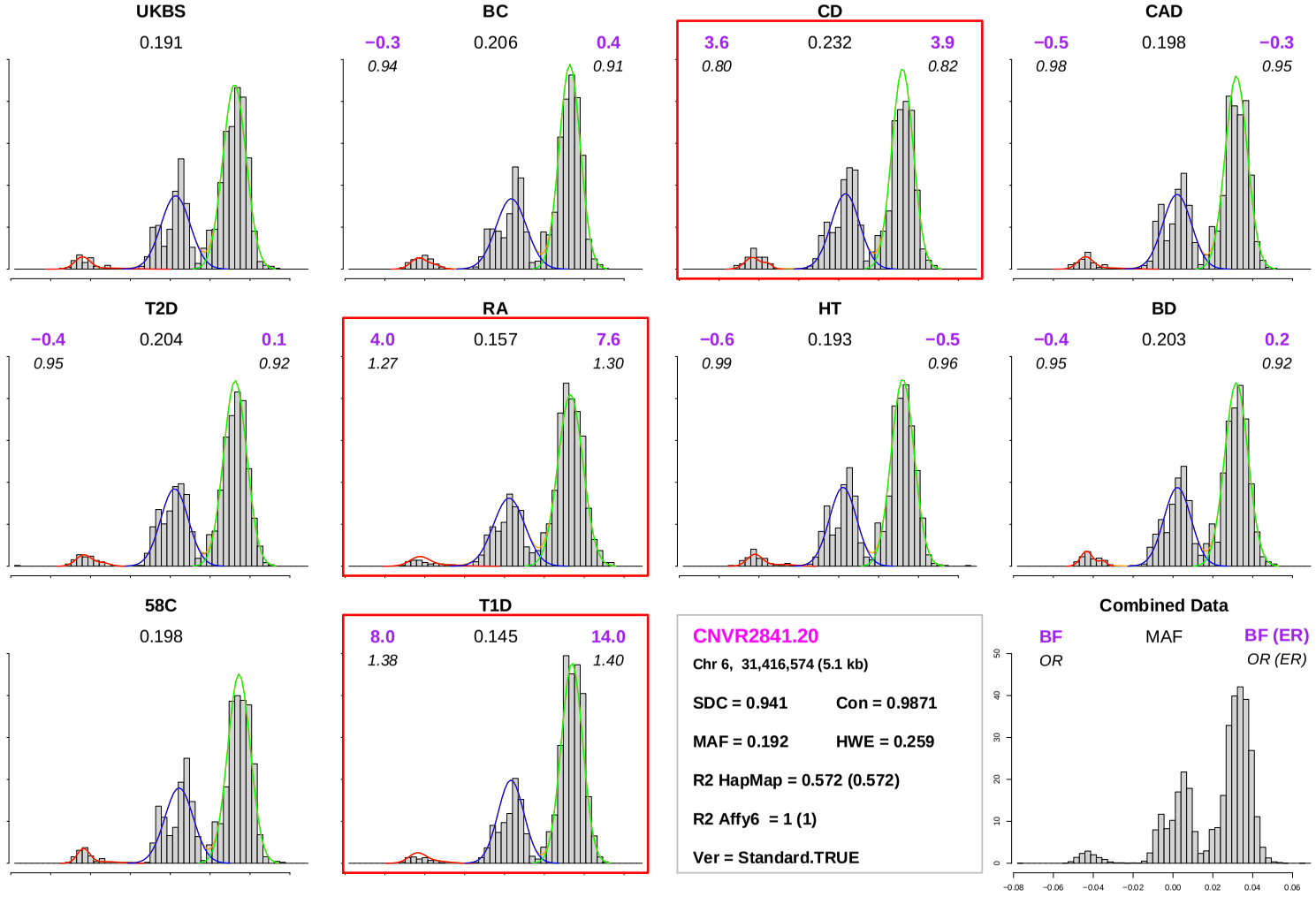
2 CNVR2646.1


UKBS
0.063
BC
0.1
0.075
−0.4
0.89
1.04
CD
5.8
0.095
4.1
0.68
0.75
CAD
0.6
0.079
−0.5
0.84
0.99
T2D
0.5
0.077
−0.5
0.86
1.01
RA
1.4
0.084
0.0
0.81
0.90
HT
0.0
0.073
−0.3
0.90
1.06
BD
1.5
0.082
−0.2
0.80
0.93
58C
0.068
T1D
−0.1
0.071
−0.5
0.91
1.03
CNVR2646.1
Chr 5, 150,157,836 (3.9 kb)
SDC = 1
Con = 0.998
MAF = 0.077
HWE = 0.446
R2 HapMap = 0.992 (0.992)
R2 Affy6 = 0.991 (0.991)
Ver = Red.TRUE
Combined Data
BF
MAF
BF (ER)
OR
OR (ER)
0
20
40
60
80
−0.05
0.00
0.05
0.10

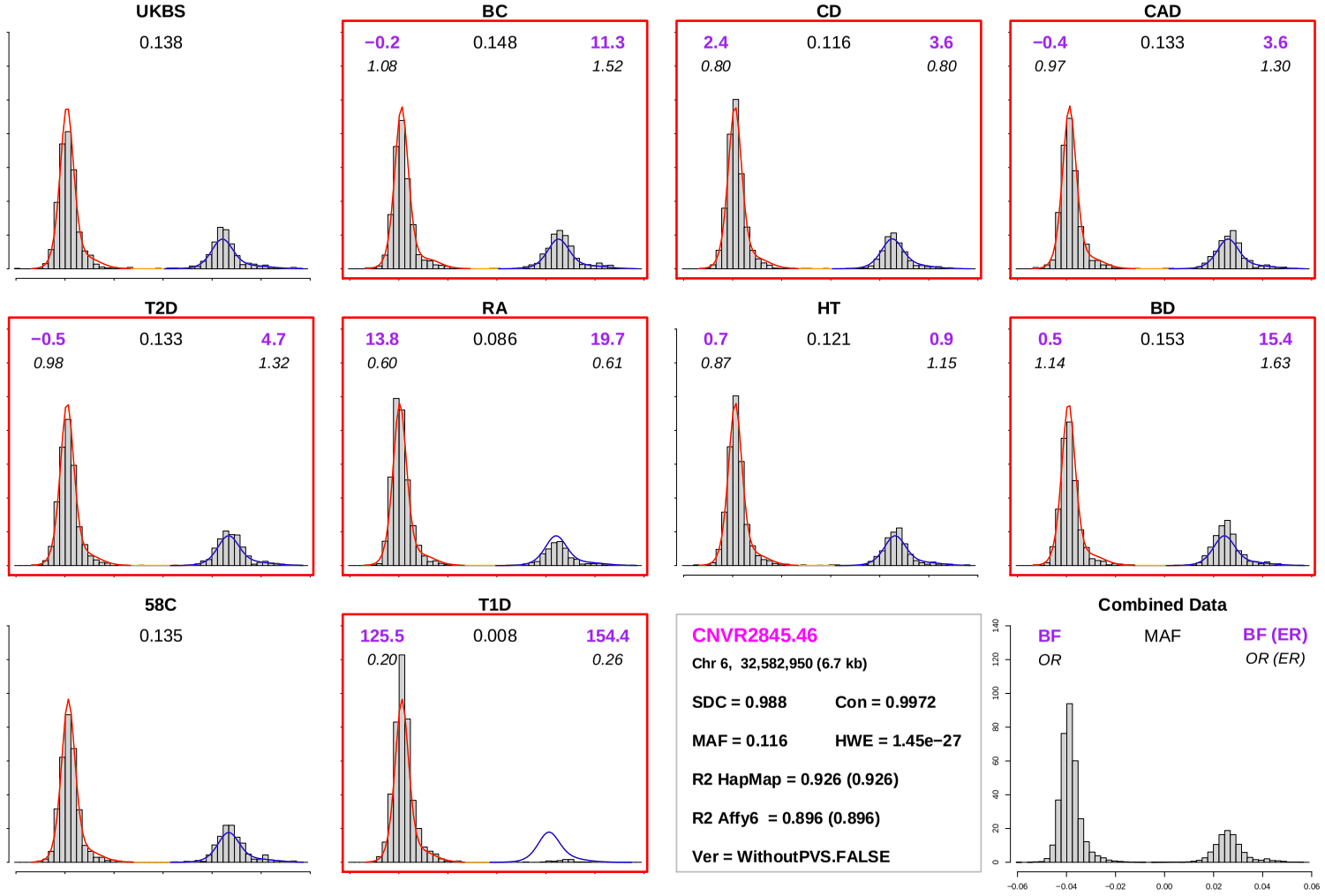
3 CNVR2647.1


UKBS
0.066
BC
0.4
0.081
−0.5
0.87
1.03
CD
6.1
0.100
3.8
0.68
0.76
CAD
1.0
0.084
−0.5
0.82
0.98
T2D
0.5
0.080
−0.5
0.86
1.02
RA
1.9
0.090
0.2
0.79
0.89
HT
0.3
0.078
−0.4
0.88
1.05
BD
1.8
0.087
−0.2
0.79
0.93
58C
0.071
T1D
0.2
0.077
−0.5
0.88
1.00
CNVR2647.1
Chr 5, 150,183,562 (20.1 kb)
SDC = 0.998
Con = 0.999
MAF = 0.082
HWE = 0.466
R2 HapMap = 1 (1)
R2 Affy6 = 1 (1)
Ver = Standard.FALSE
Combined Data
BF
MAF
BF (ER)
OR
OR (ER)
200
150
100
50
0
−0.05
0.00
0.05
0.10

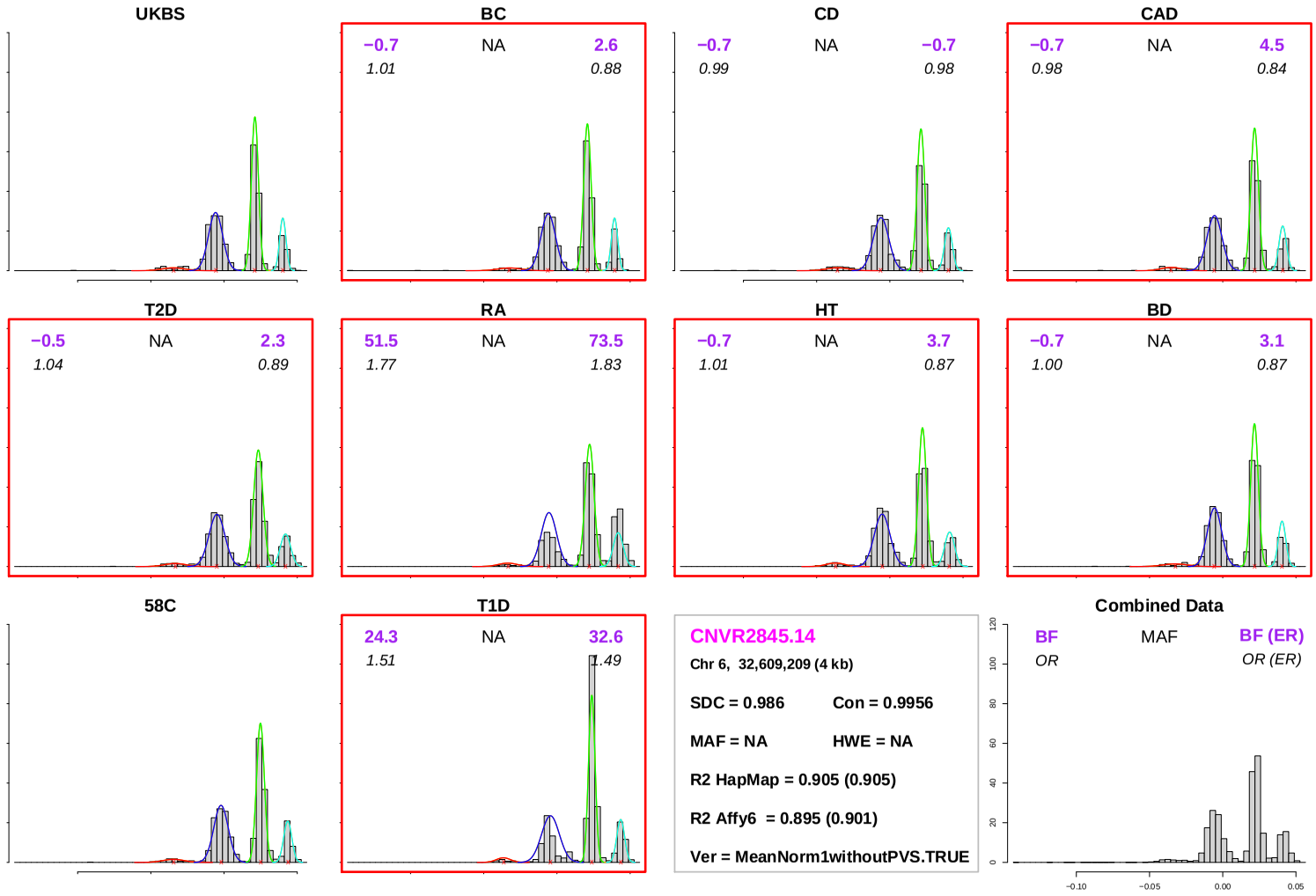
4 CNVR2841.20



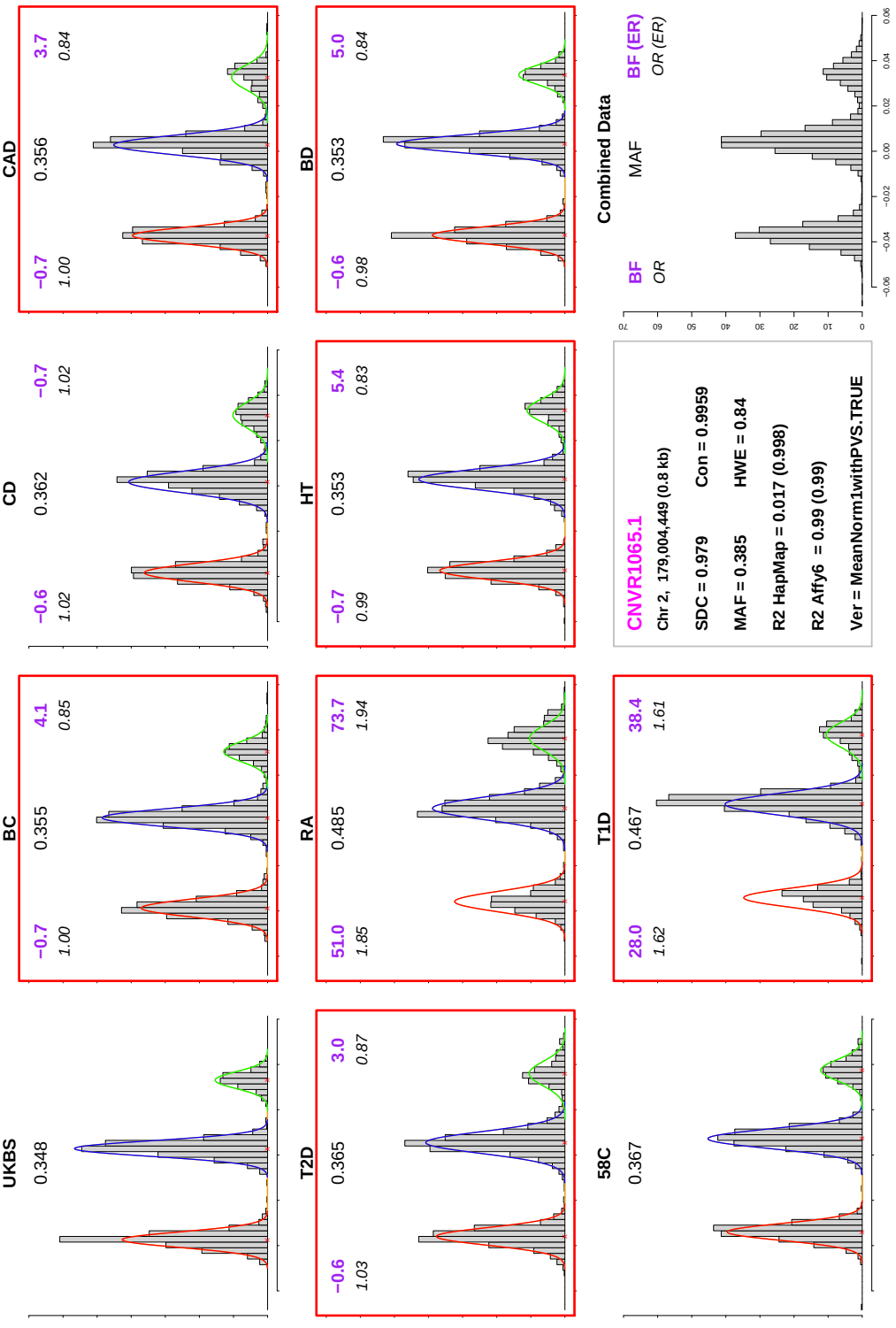
5 CNVR2845.46



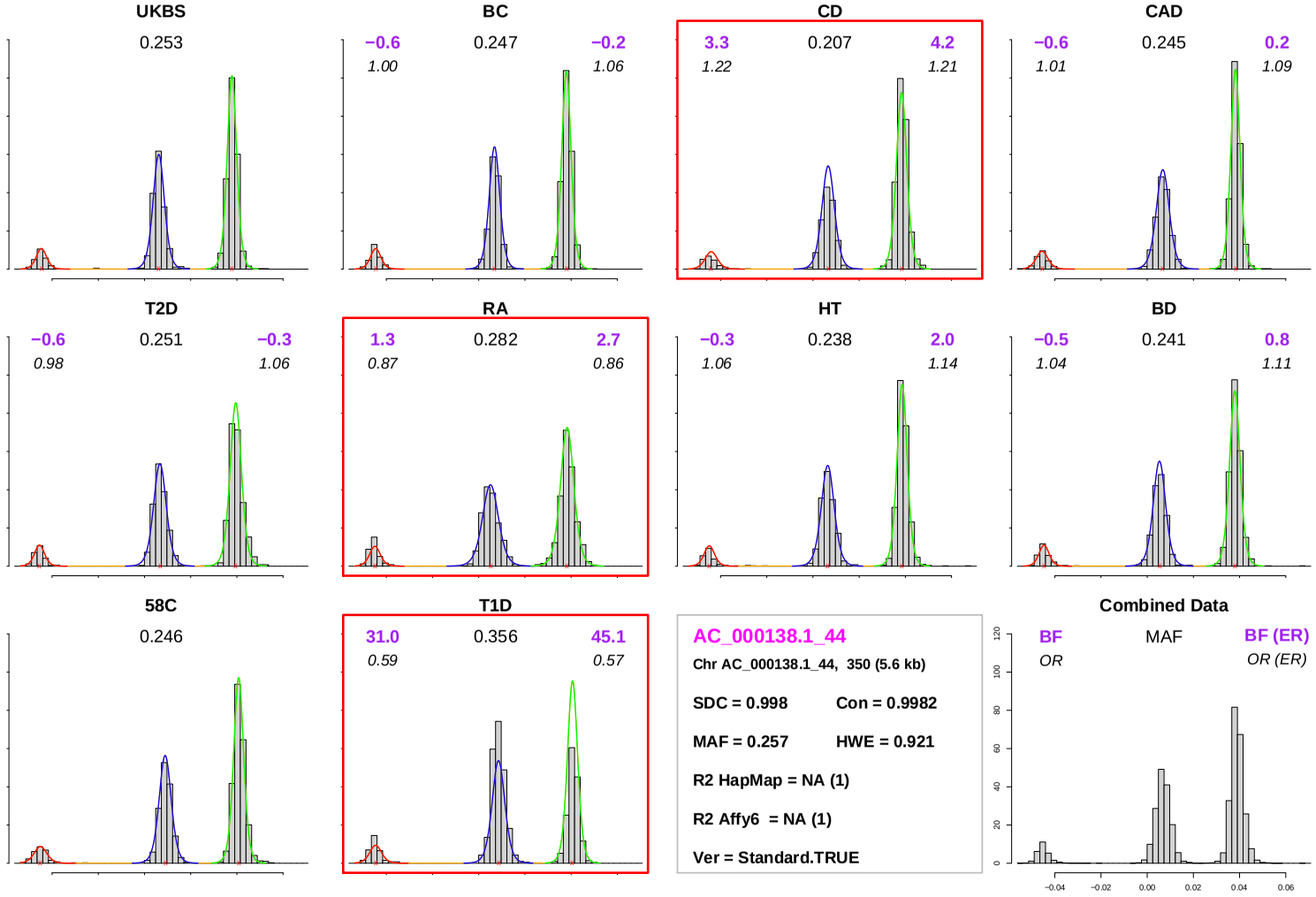
6 CNVR2845.14



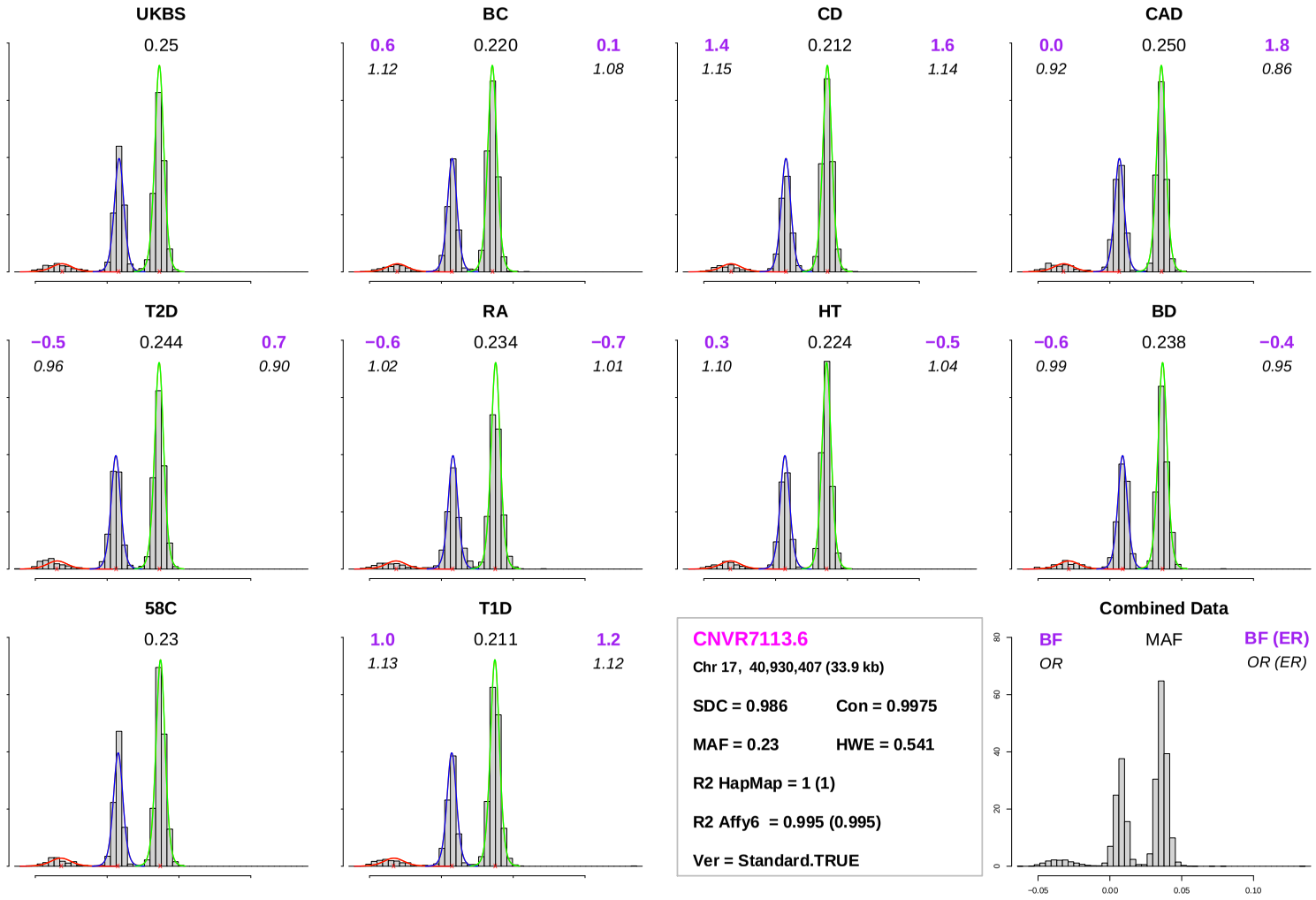
7 CNVR1065.1


UKBS
0.348
BC
0.355
−0.7
1.00
4.1
0.85
CD
0.362
−0.6
1.02
−0.7
1.02
CAD
0.356
−0.7
1.00
3.7
0.84
T2D
0.365
−0.6
1.03
3.0
0.87
RA
0.485
51.0
1.85
73.7
1.94
HT
0.353
−0.7
0.99
5.4
0.83
BD
0.353
−0.6
0.98
5.0
0.84
58C
0.367
T1D
0.467
28.0
1.62
38.4
1.61
CNVR1065.1
Chr 2, 179,004,449 (0.8 kb)
SDC = 0.979 Con = 0.9959
MAF = 0.385 HWE = 0.84
R2 HapMap = 0.017 (0.998)
R2 Affy6 = 0.99 (0.99)
Ver = MeanNorm1withPVS.TRUE
Combined Data
BF
OR
MAF
BF (ER)
OR (ER)

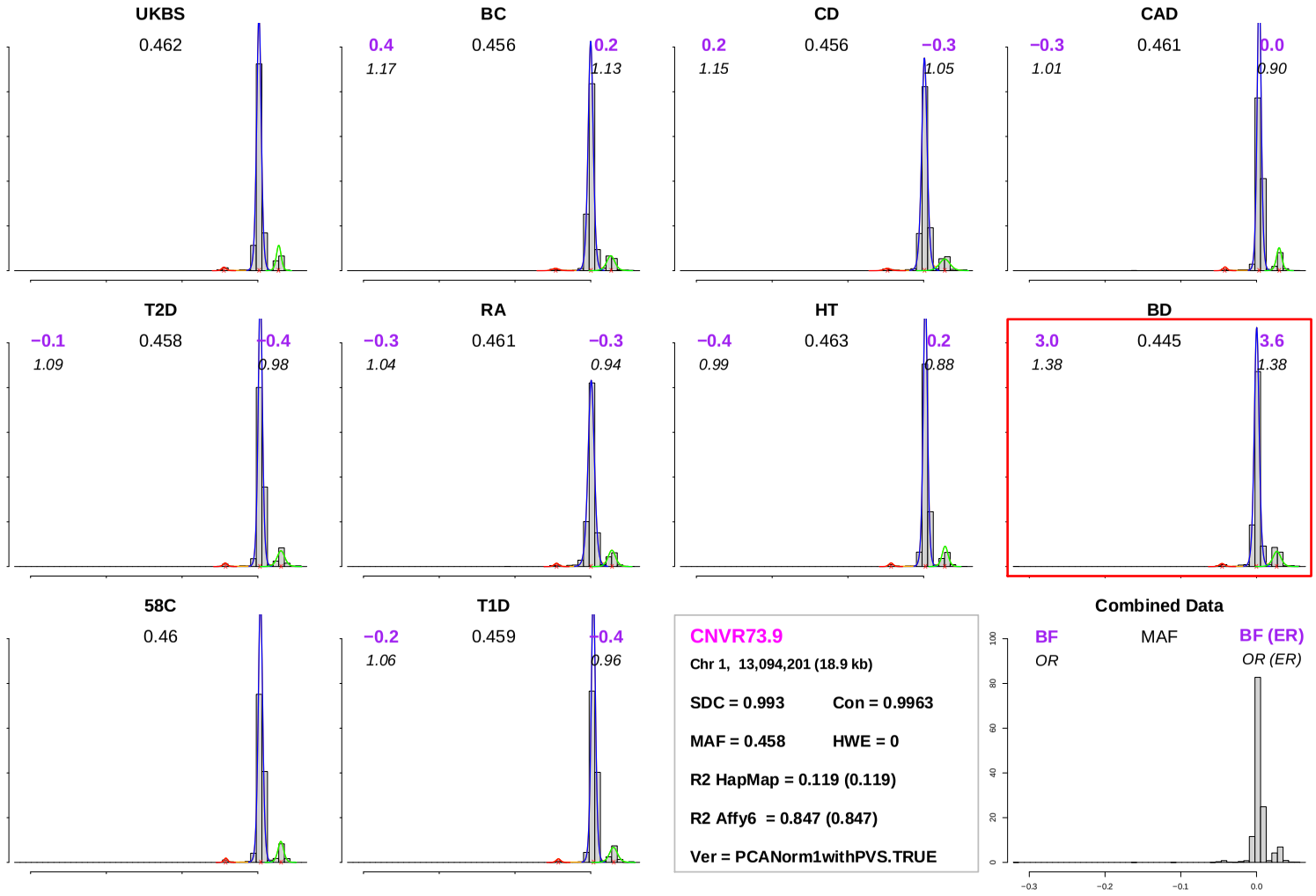
8 AC_000138.1_44



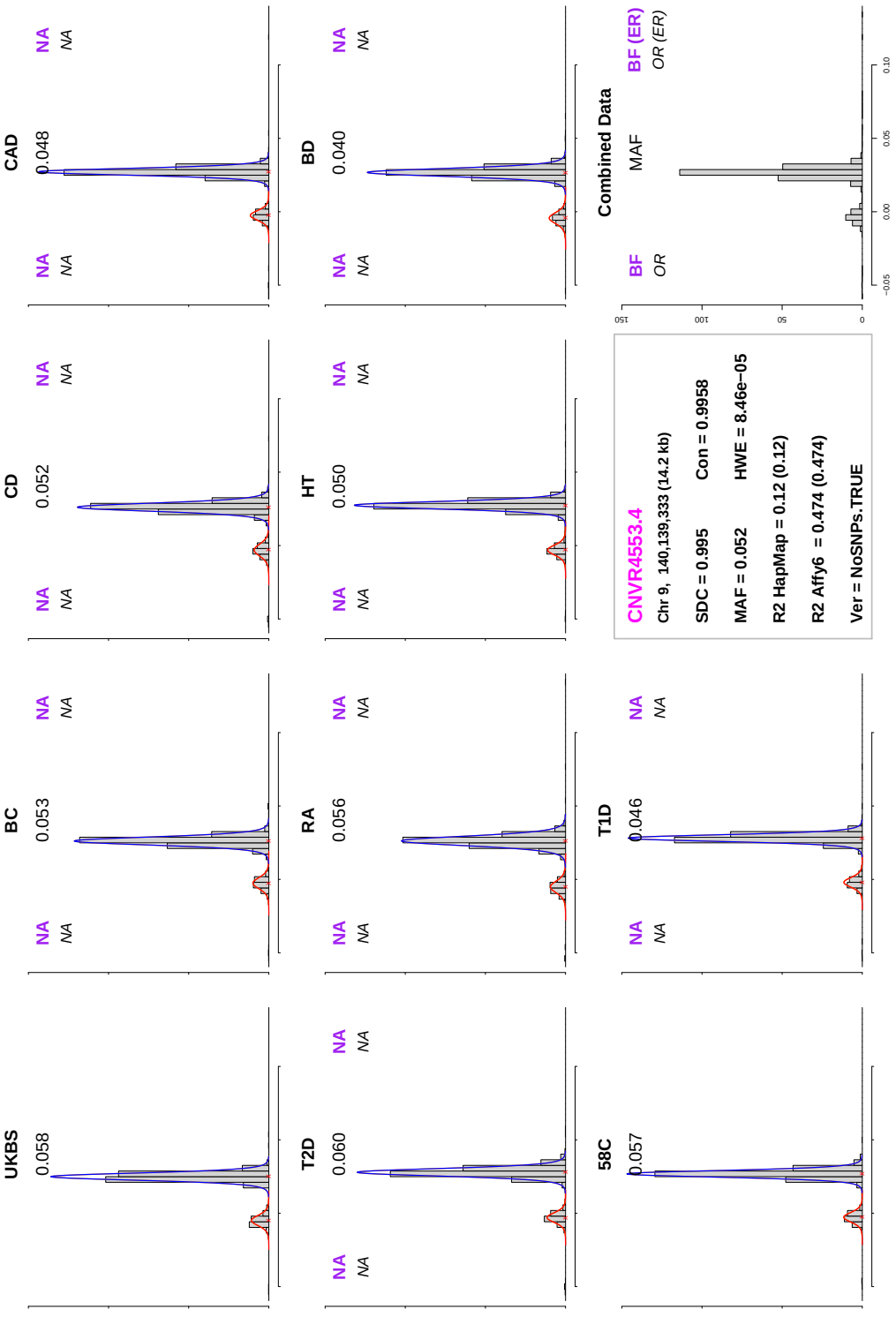
9 CNVR7113.6


UKBS
0.25
BC
0.6 0.220 0.1
1.12 1.08
CD
1.4 0.212 1.6
1.15 1.14
CAD
0.0 0.250 1.8
0.92 0.86
T2D
−0.5 0.244 0.7
0.96 0.90
RA
−0.6 0.234 −0.7
1.02 1.01
HT
0.3 0.224 −0.5
1.10 1.04
BD
−0.6 0.238 −0.4
0.99 0.95
58C
0.23
T1D
1.0 0.211 1.2
1.13 1.12
CNVR7113.6
Chr 17, 40,930,407 (33.9 kb)
SDC = 0.986 Con = 0.9975
MAF = 0.23 HWE = 0.541
R2 HapMap = 1 (1)
R2 Affy6 = 0.995 (0.995)
Ver = Standard.TRUE
Combined Data
BF MAF BF (ER)
OR OR (ER)
0 20 40 60 80
−0.05 0.00 0.05 0.10

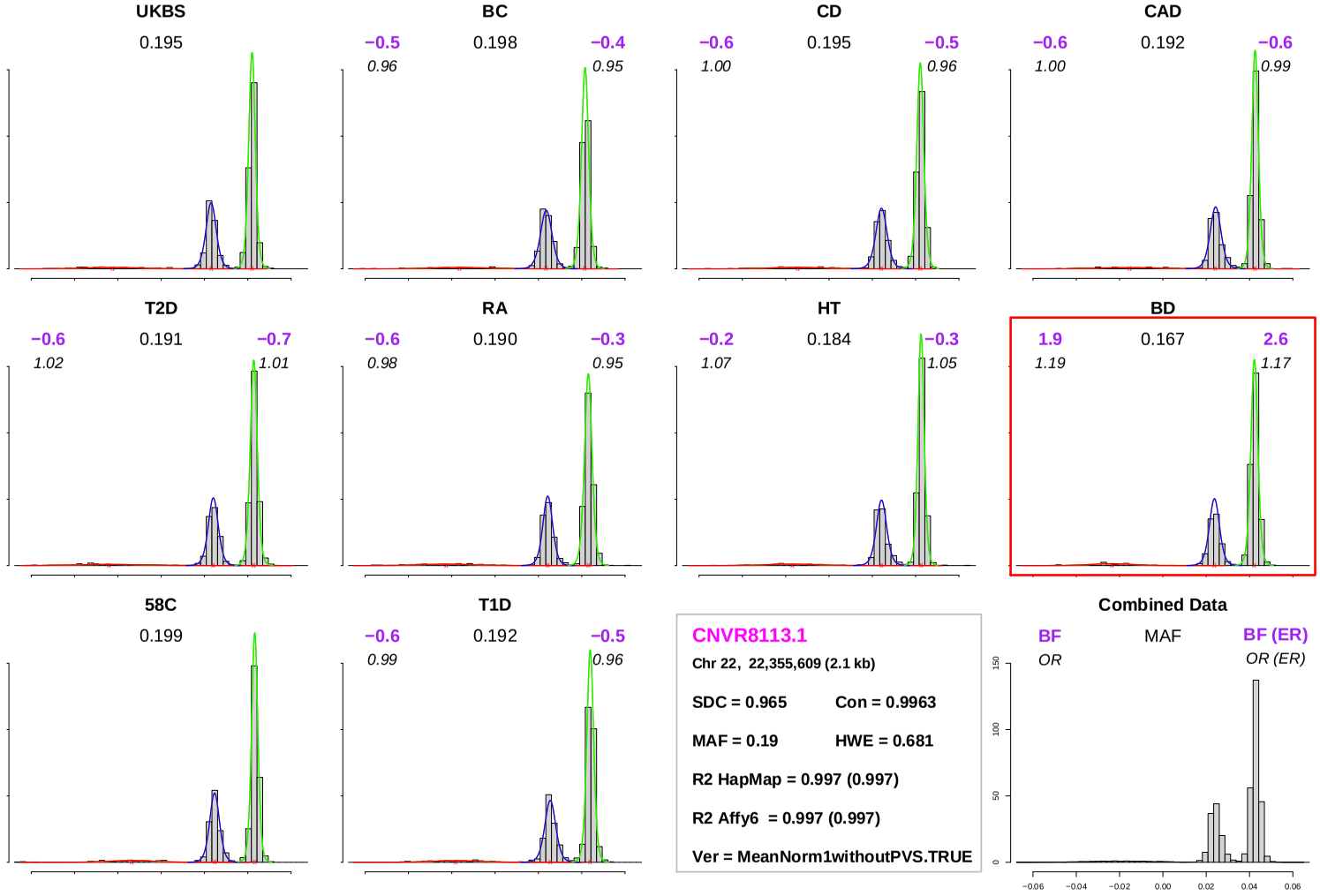
10 CNVR73.9


UKBS
0.462
BC
0.4
0.456
0.2
1.17
1.13
CD
0.2
0.456
−0.3
1.15
1.05
CAD
−0.3
0.461
0.0
1.01
0.90
T2D
−0.1
0.458
−0.4
1.09
0.98
RA
−0.3
0.461
−0.3
1.04
0.94
HT
−0.4
0.463
0.2
0.99
0.88
BD
3.0
0.445
3.6
1.38
1.38
58C
0.46
T1D
−0.2
0.459
−0.4
1.06
0.96
CNVR73.9
Chr 1, 13,094,201 (18.9 kb)
SDC = 0.993 Con = 0.9963
MAF = 0.458 HWE = 0
R2 HapMap = 0.119 (0.119)
R2 Affy6 = 0.847 (0.847)
Ver = PCANorm1withPVS.TRUE
Combined Data
BF
OR
MAF
BF (ER)
OR (ER)
100
80
60
40
20
0
−0.3
−0.2
−0.1
0.0

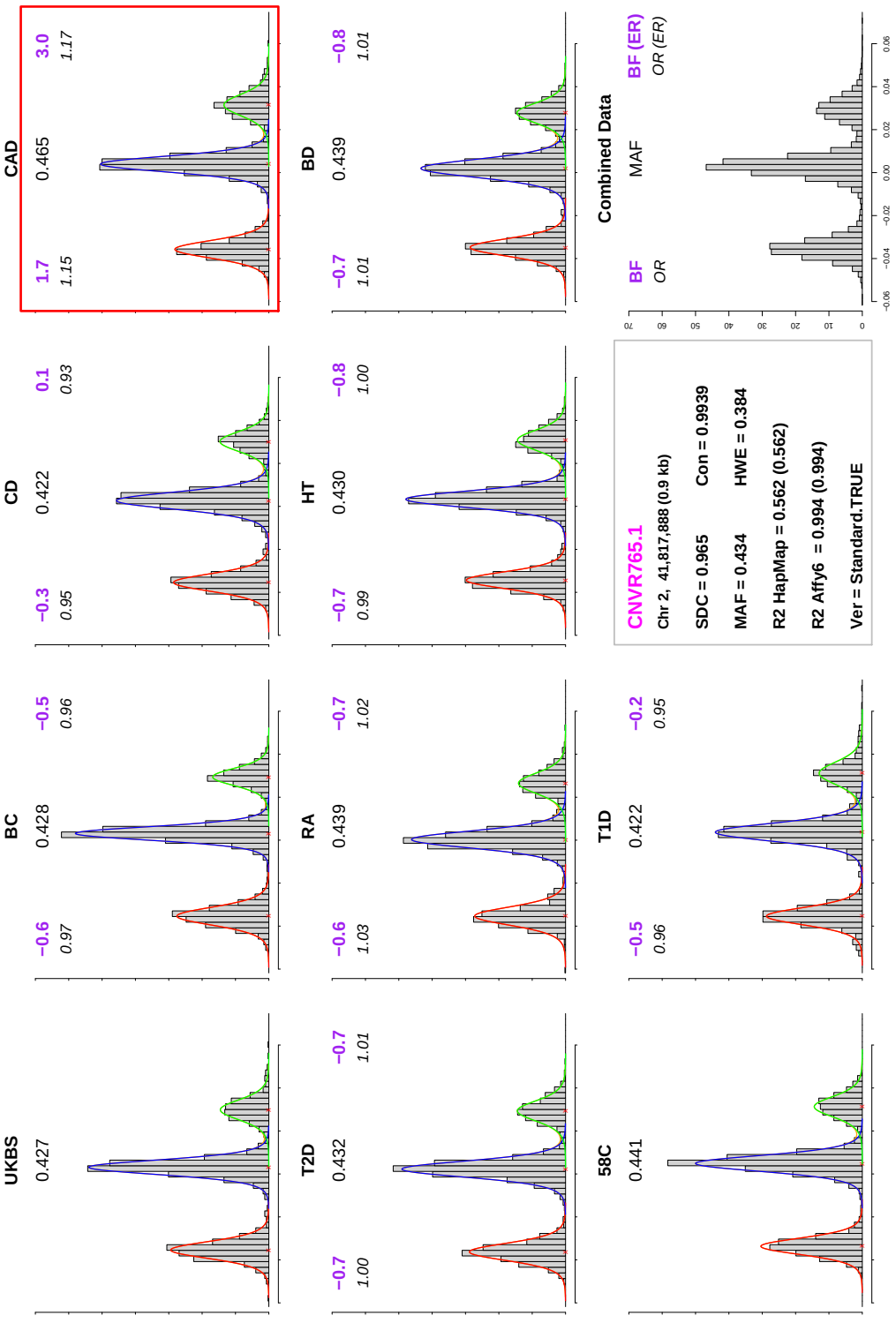
11 CNVR4553.4



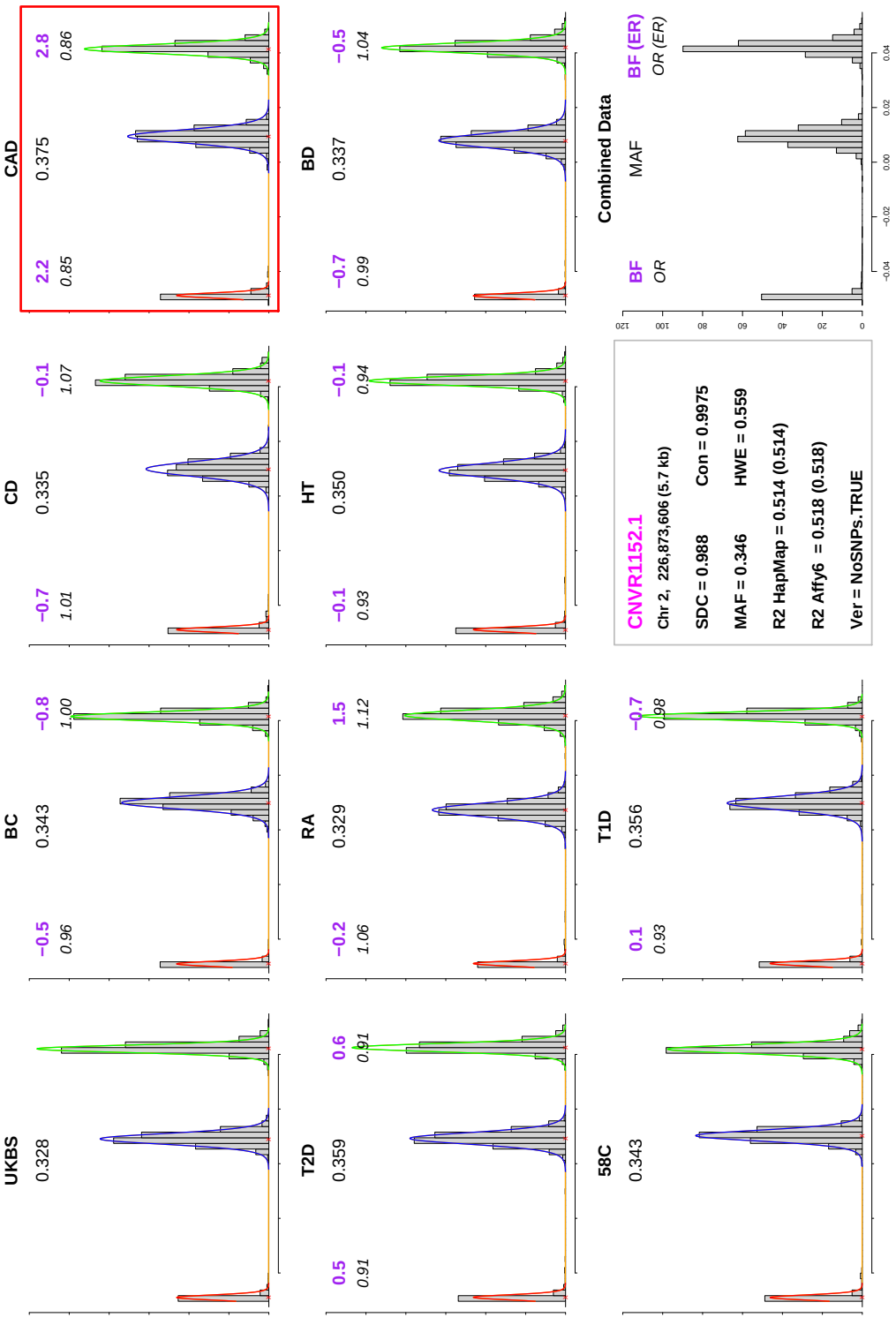
12 CNVR8113.1



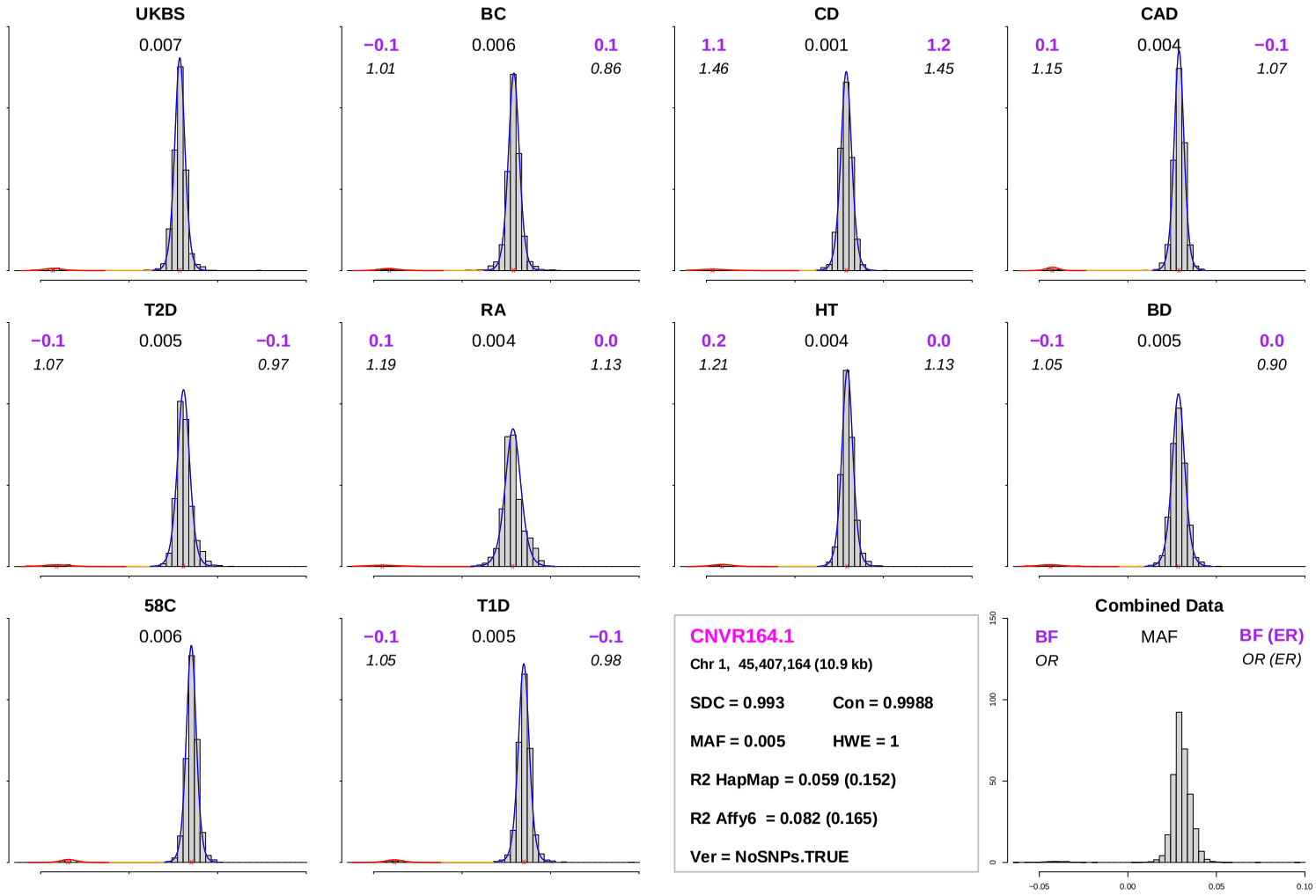
13 CNVR765.1


UKBS
0.427
BC
0.428
−0.6
0.97
−0.5
0.96
CD
0.422
−0.3
0.95
0.1
0.93
CAD
0.465
1.7
1.15
3.0
1.17
T2D
0.432
−0.7
1.00
−0.7
1.01
RA
0.439
−0.6
1.03
−0.7
1.02
HT
0.430
−0.7
0.99
−0.8
1.00
BD
0.439
−0.7
1.01
−0.8
1.01
58C
0.441
T1D
0.422
−0.5
0.96
−0.2
0.95
CNVR765.1
Chr 2, 41,817,888 (0.9 kb)
SDC = 0.965 Con = 0.9939
MAF = 0.434 HWE = 0.384
R2 HapMap = 0.562 (0.562)
R2 Affy6 = 0.994 (0.994)
Ver = Standard.TRUE
Combined Data
BF
OR
MAF
BF (ER)
OR (ER)

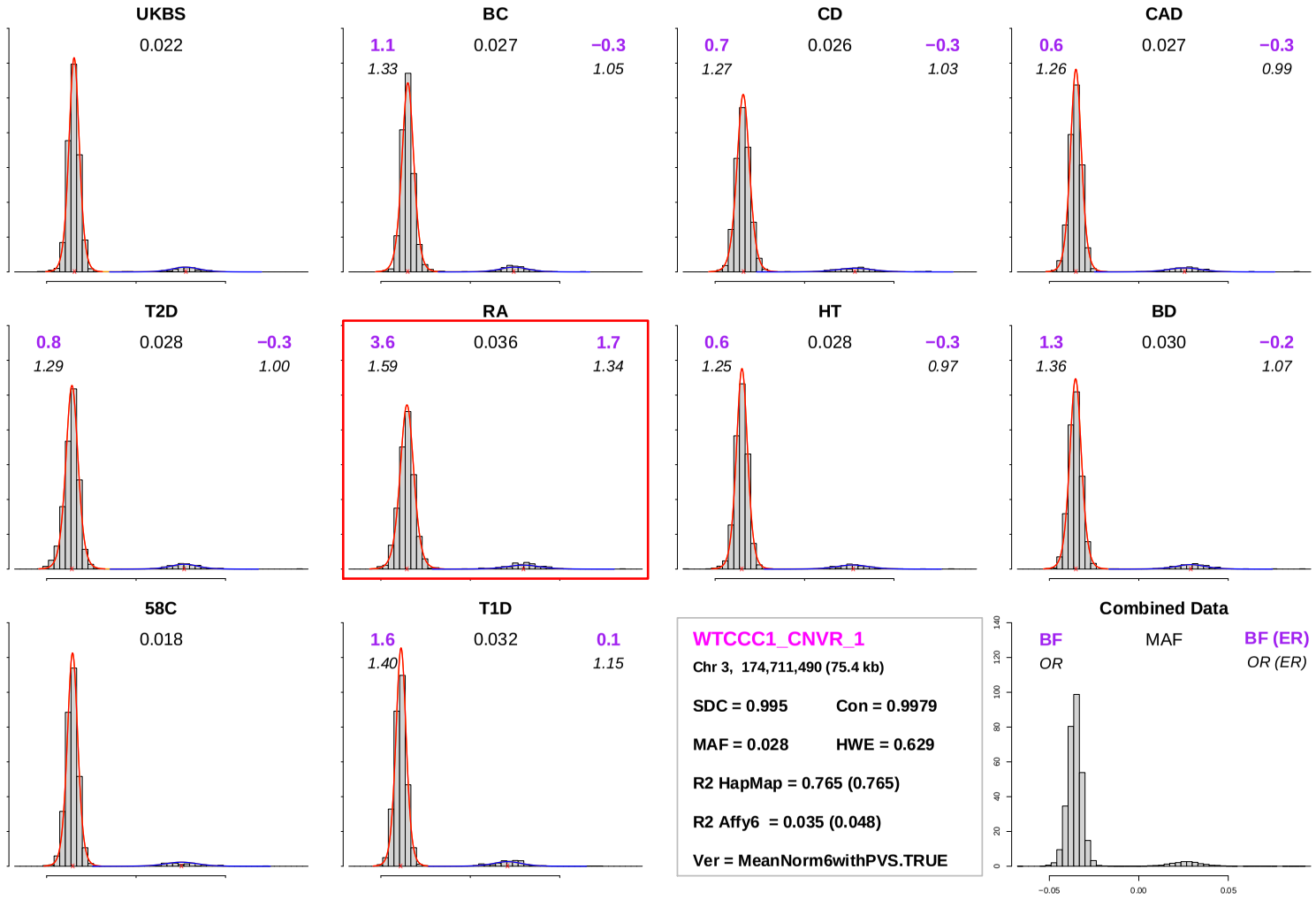
14 CNVR1152.1



15 CNVR164.1


UKBS
0.007
BC
−0.1 0.006 0.1
1.01 0.86
CD
1.1 0.001 1.2
1.46 1.45
CAD
0.1 0.004 −0.1
1.15 1.07
T2D
−0.1 0.005 −0.1
1.07 0.97
RA
0.1 0.004 0.0
1.19 1.13
HT
0.2 0.004 0.0
1.21 1.13
BD
−0.1 0.005 0.0
1.05 0.90
58C
0.006
T1D
−0.1 0.005 −0.1
1.05 0.98
CNVR164.1
Chr 1, 45,407,164 (10.9 kb)
SDC = 0.993 Con = 0.9988
MAF = 0.005 HWE = 1
R2 HapMap = 0.059 (0.152)
R2 Affy6 = 0.082 (0.165)
Ver = NoSNPs.TRUE
Combined Data
BF MAF BF (ER)
OR OR (ER)
150
100
50
0
−0.05 0.00 0.05 0.10

16 WTCCC1_CNVR_1



17 CNVR2920.2