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

Supplementary File 3

Version 2.0.0

The Wellcome Trust Case Control Consortium

December 18, 2009

Plots of CNV calls and association testing results produced by the CNVtools approach

The following plots show the CNV intensity distributions for each cohort at all of the CNVs in Table 3 and Supplementary Table 13. The CNV intensity distributions for each cohort are given in separate sub-plots. Where a cohort consists of samples from two sources (Blood derived DNA and Cell Line DNA) then two subplots are given, one each for samples from each source. The posterior probability for each CNV class is overlayed onto the intensity distributions. The minor allele frequency estimate for the cohort and the p-values for the additive test of the cohort versus the two control cohorts and the expanded reference panels (see Supplementary Table 9) are given in the title of each subplot. If either or both of the 2 p-values (restricted or expanded) is < 0.001 then the title is colored red. The sample size of the cohort and the case and control sample sizes in the expanded reference analysis are given below each subplot. The bottom middle sub-plot first shows the CNV name and position. The "gender chi-square" category shows the goodness-of-fit test statistic for association between gender and copy number calls, which is distributed under the null of no association between gender and genotype as a chi-squared random variable with (n-1) degrees of freedom where n is the number of copy number classes. The "Dup Concordance" category shows the concordance at this CNV between the duplicate samples included in this study. This "Stratified: FALSE" indicates that regions of origin were not included as covariates. The P-value testing association between region and copy number call, denoted as "P-geography", is only shown when a stratified test of association was used. The last row shows the normalization and the summary method (either an initial PCA or mean signal summary across CNV probes (see Supplementary Table 8) , always followed by a linear discriminant analysis step).

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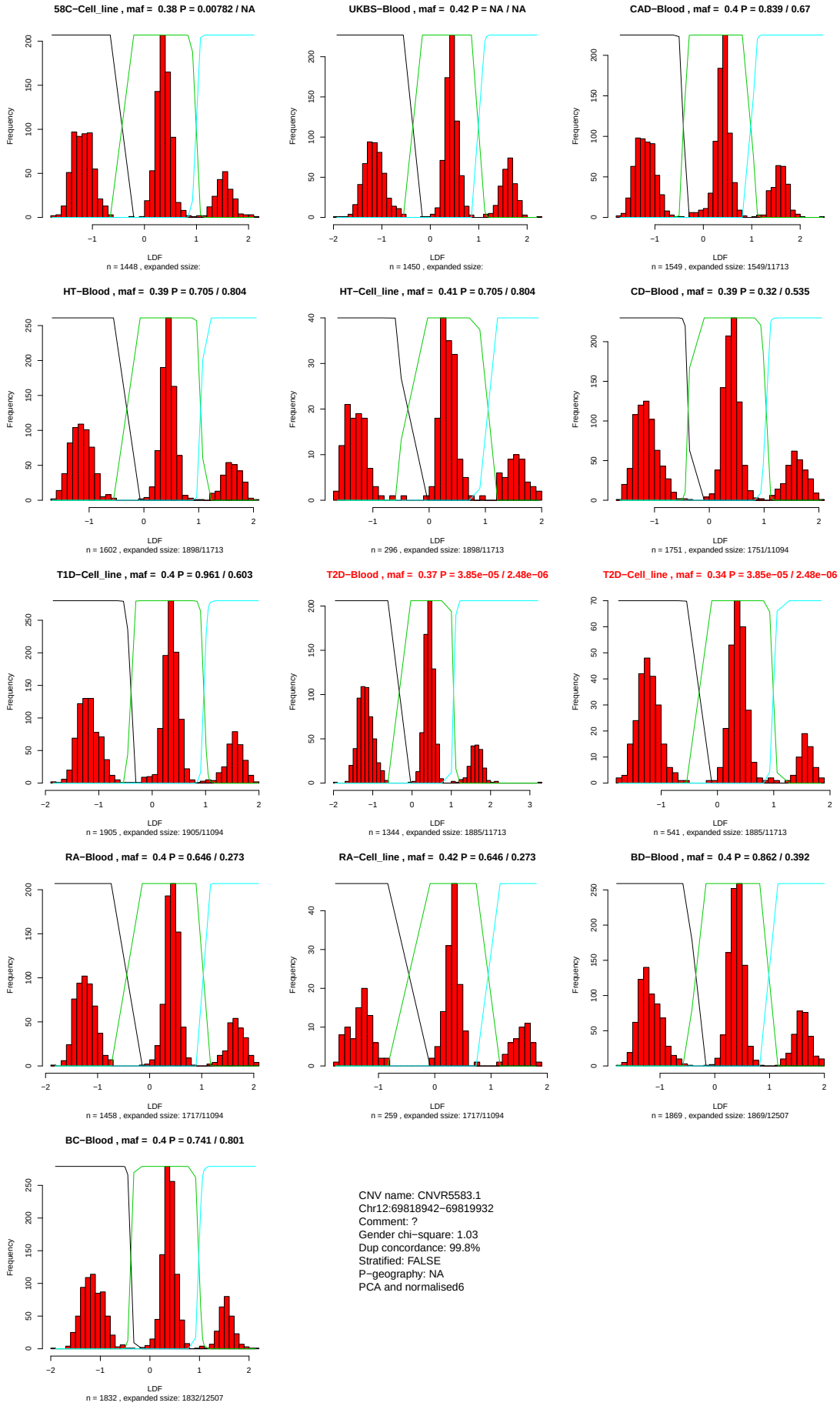
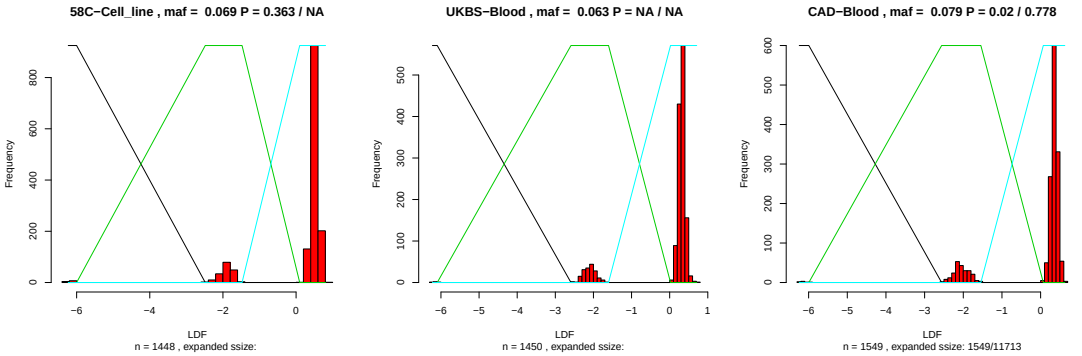
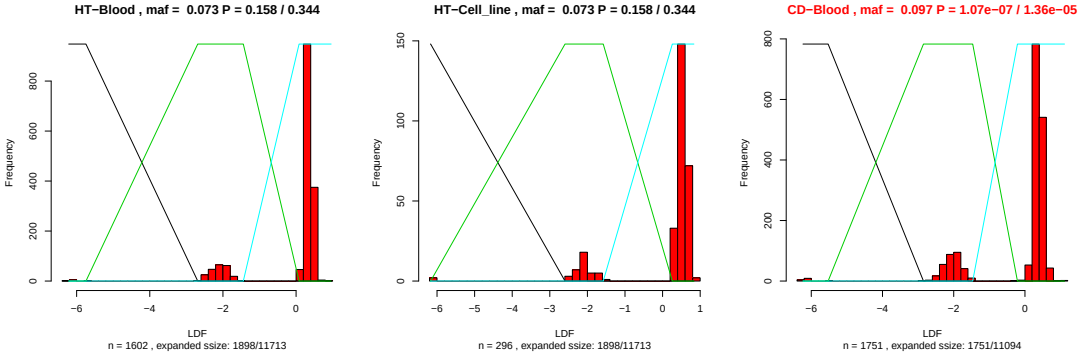
1 CNVR5583.1

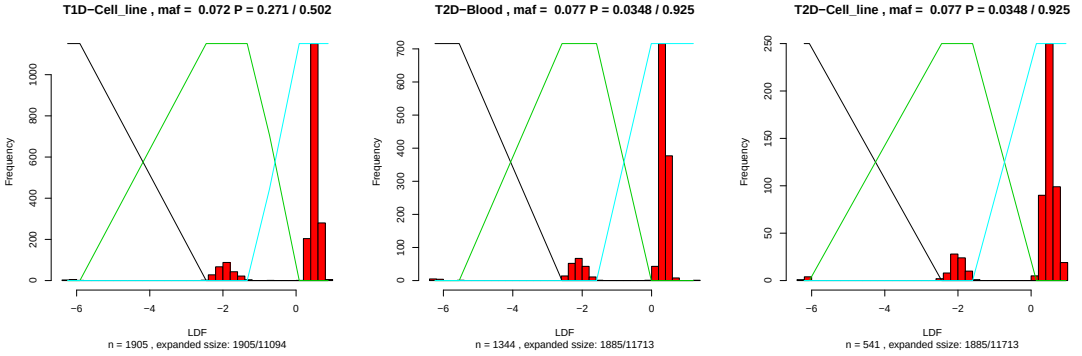


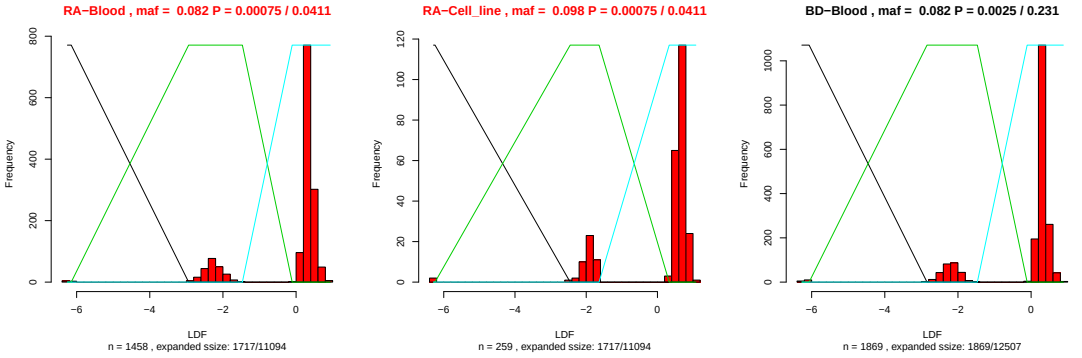
CNV name: CNVR5583.1
Chr12:69818942−69819932
Comment: ?
Gender chi−square: 1.03
Dup concordance: 99.8%
Stratified: FALSE
P−geography: NA
PCA and normalised6

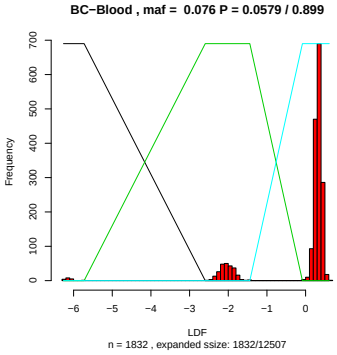
2 CNVR2646.1





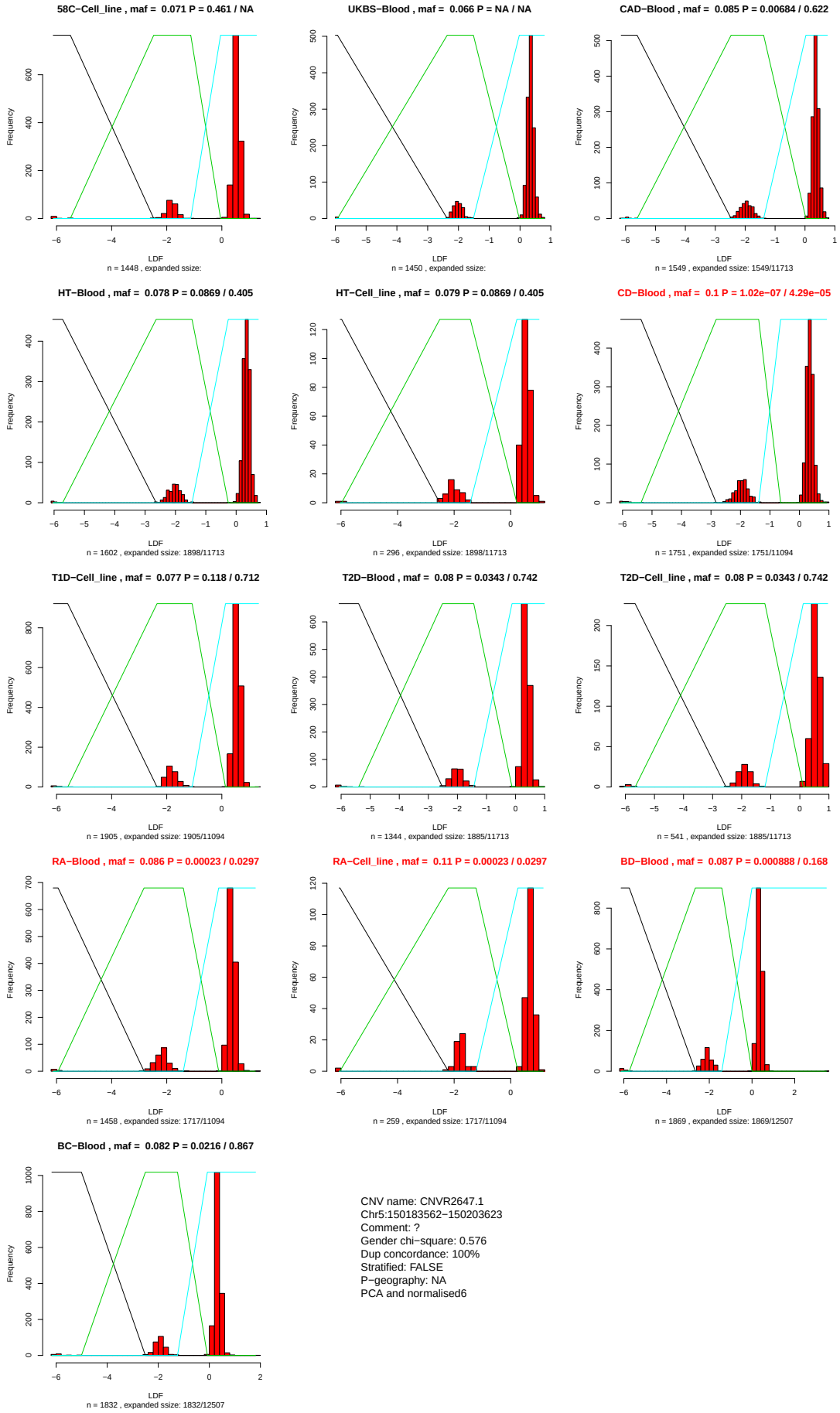






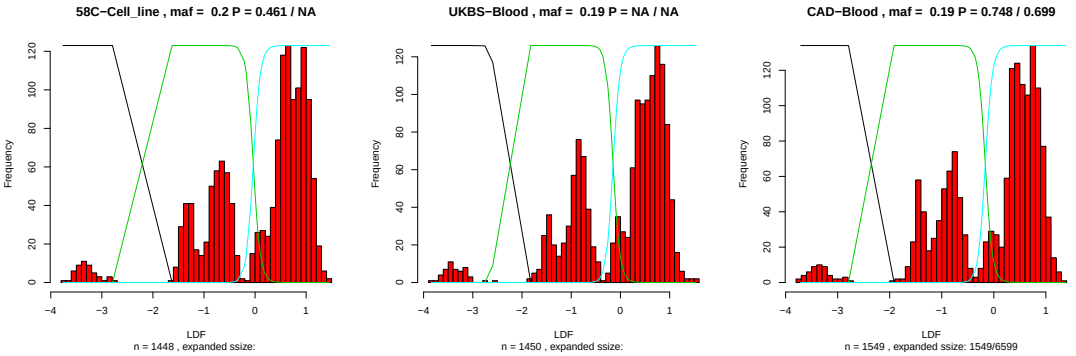
CNV name: CNVR2646.1
Chr5:150157836−150161778
Comment: ?
Gender chi−square: 0.515
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
PCA and normalised6

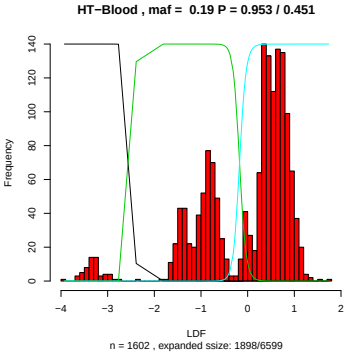
3 CNVR2647.1

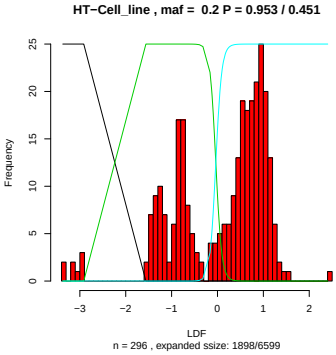


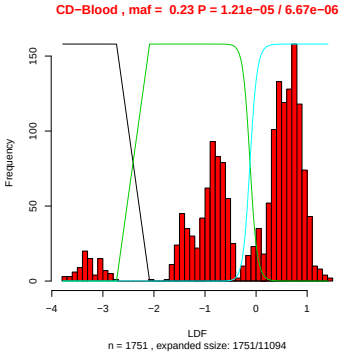
CNV name: CNVR2647.1
Chr5:150183562−150203623
Comment: ?
Gender chi−square: 0.576
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
PCA and normalised6

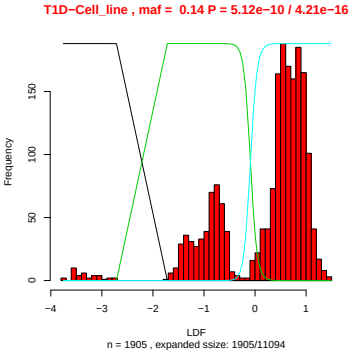
4 CNVR2841.20

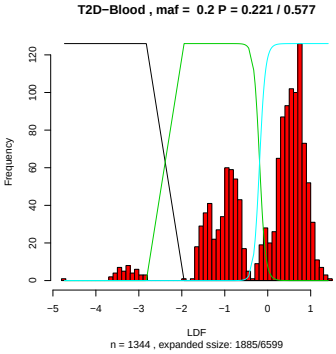


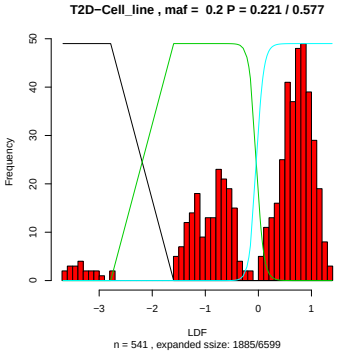


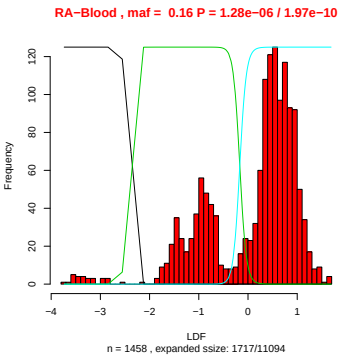


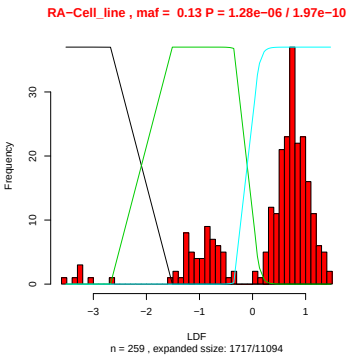


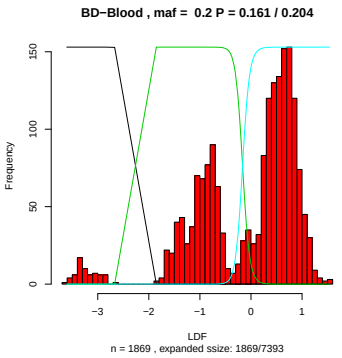


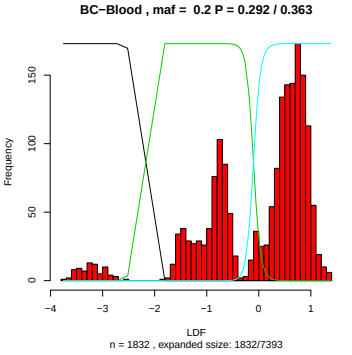






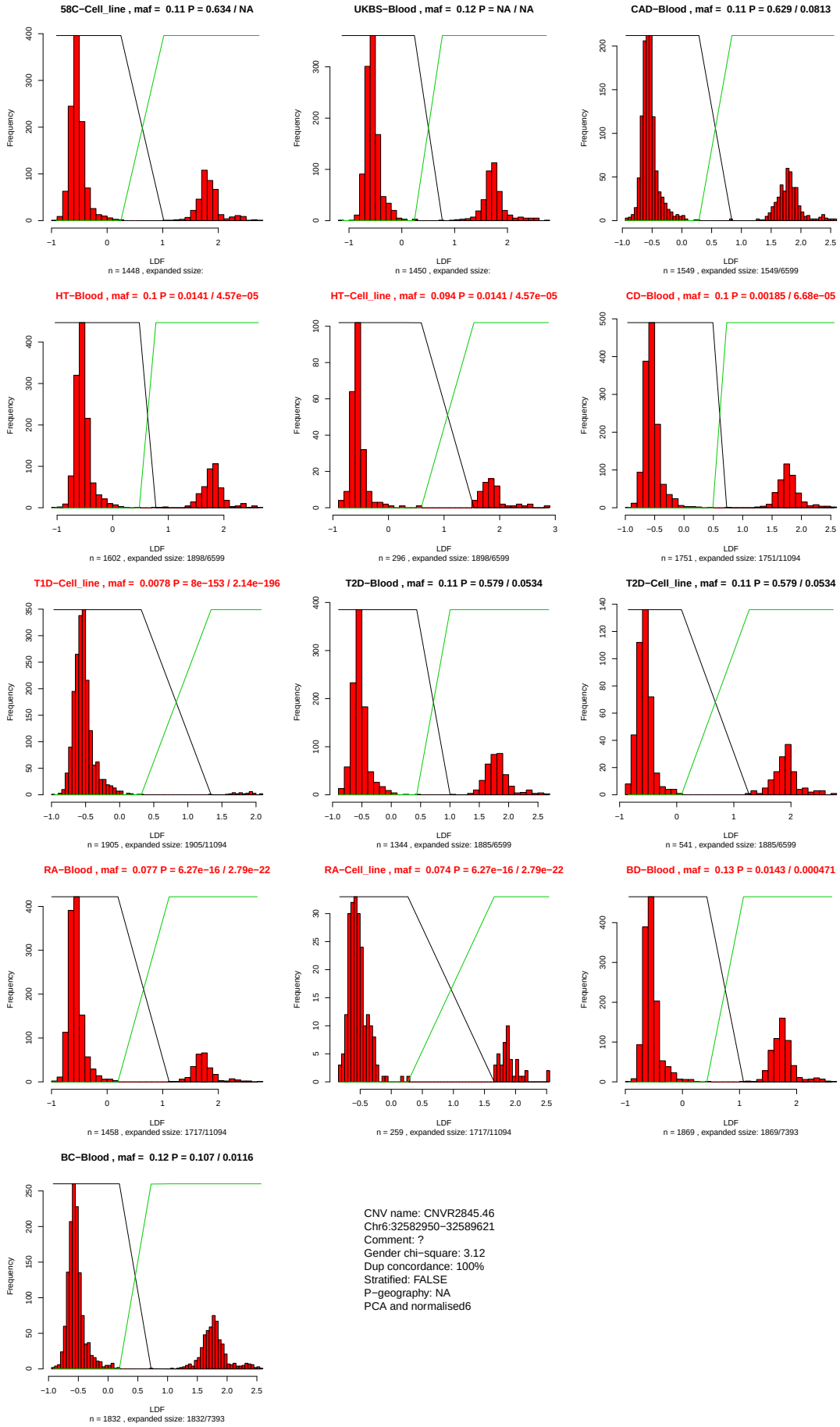






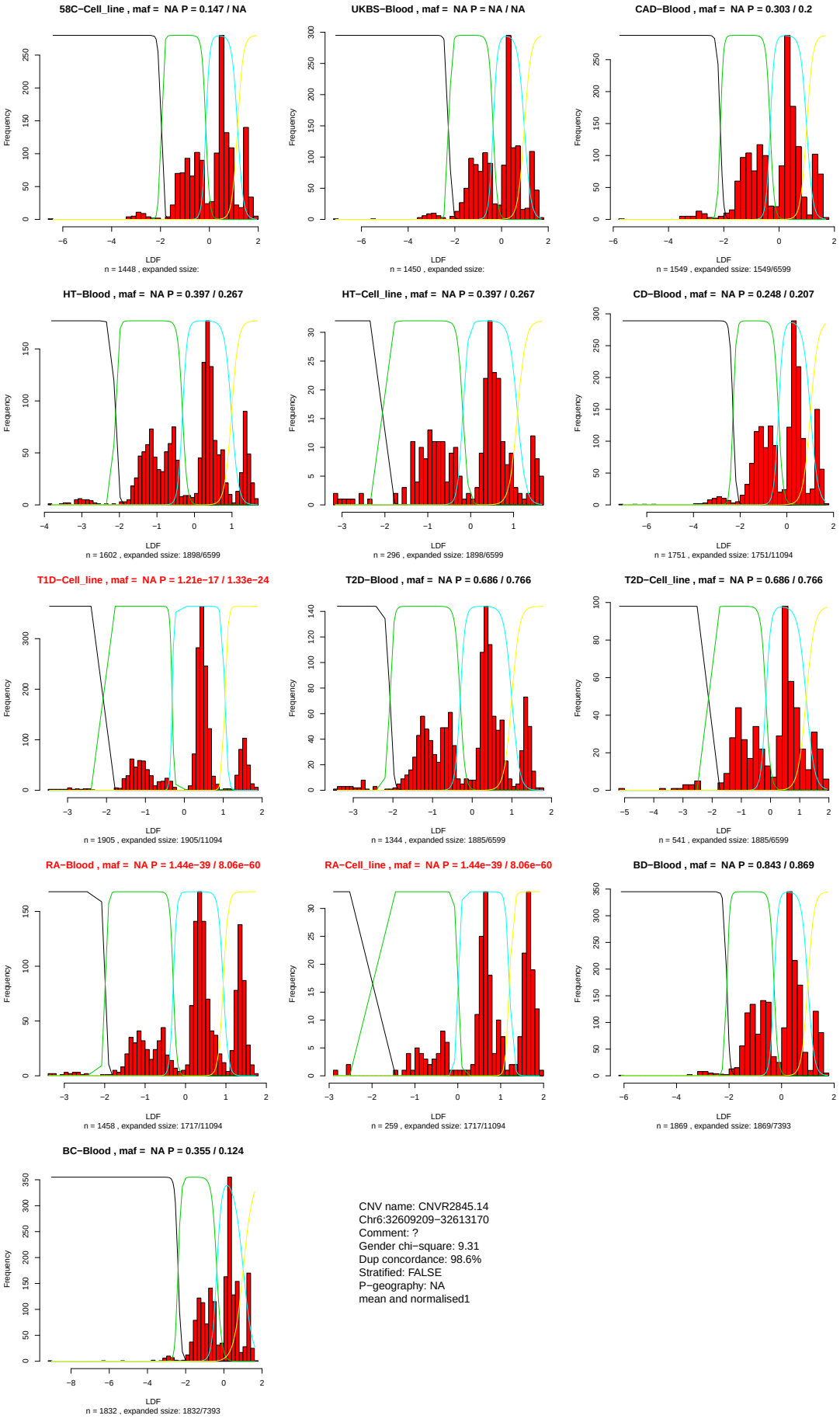
CNV name: CNVR2841.20
Chr6:31416574−31421640
Comment: ?
Gender chi−square: 1
Dup concordance: 98.6%
Stratified: FALSE
P−geography: NA
PCA and normalised6

5 CNVR2845.46



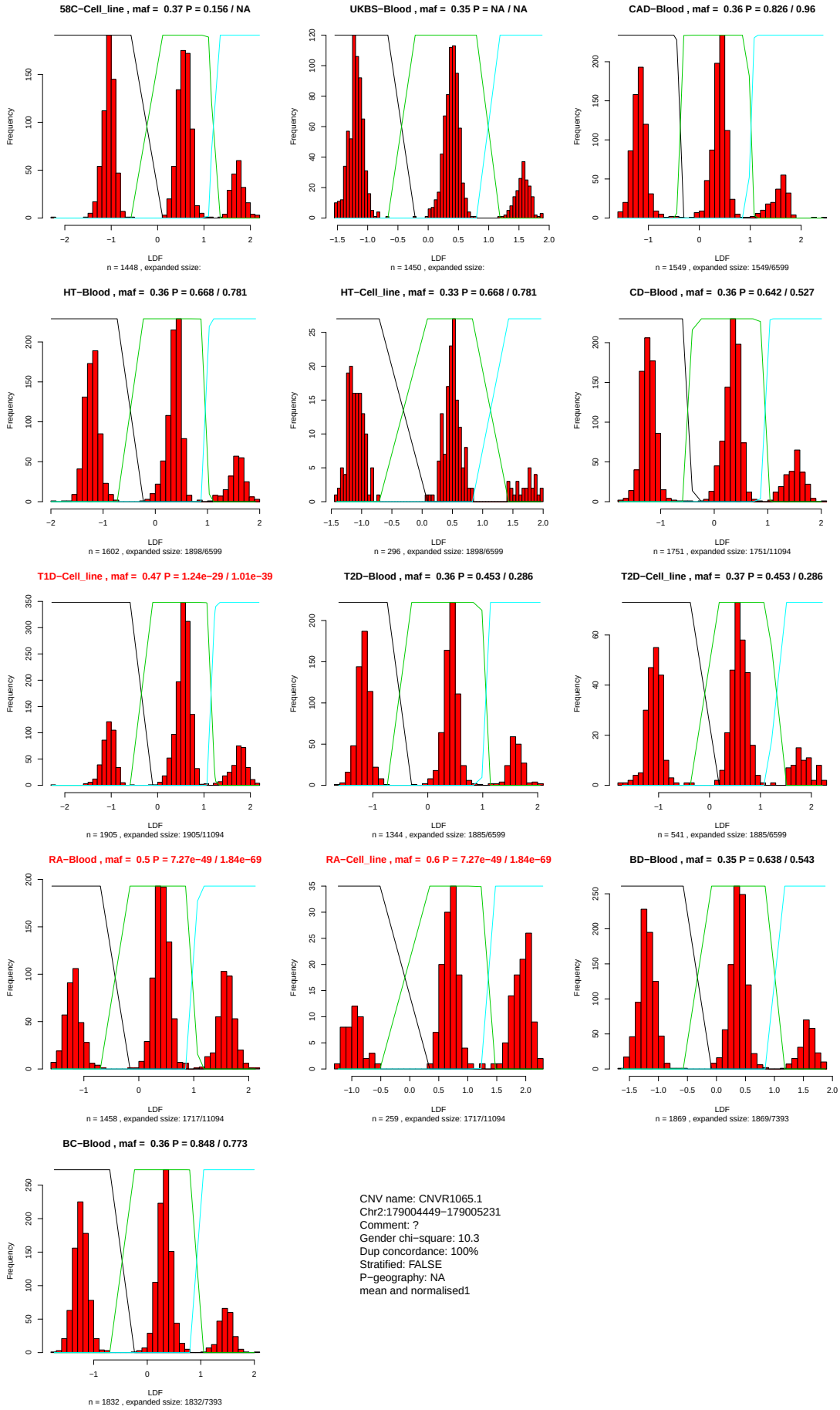
CNV name: CNVR2845.46
Chr6:32582950−32589621
Comment: ?
Gender chi−square: 3.12
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
PCA and normalised6

6 CNVR2845.14



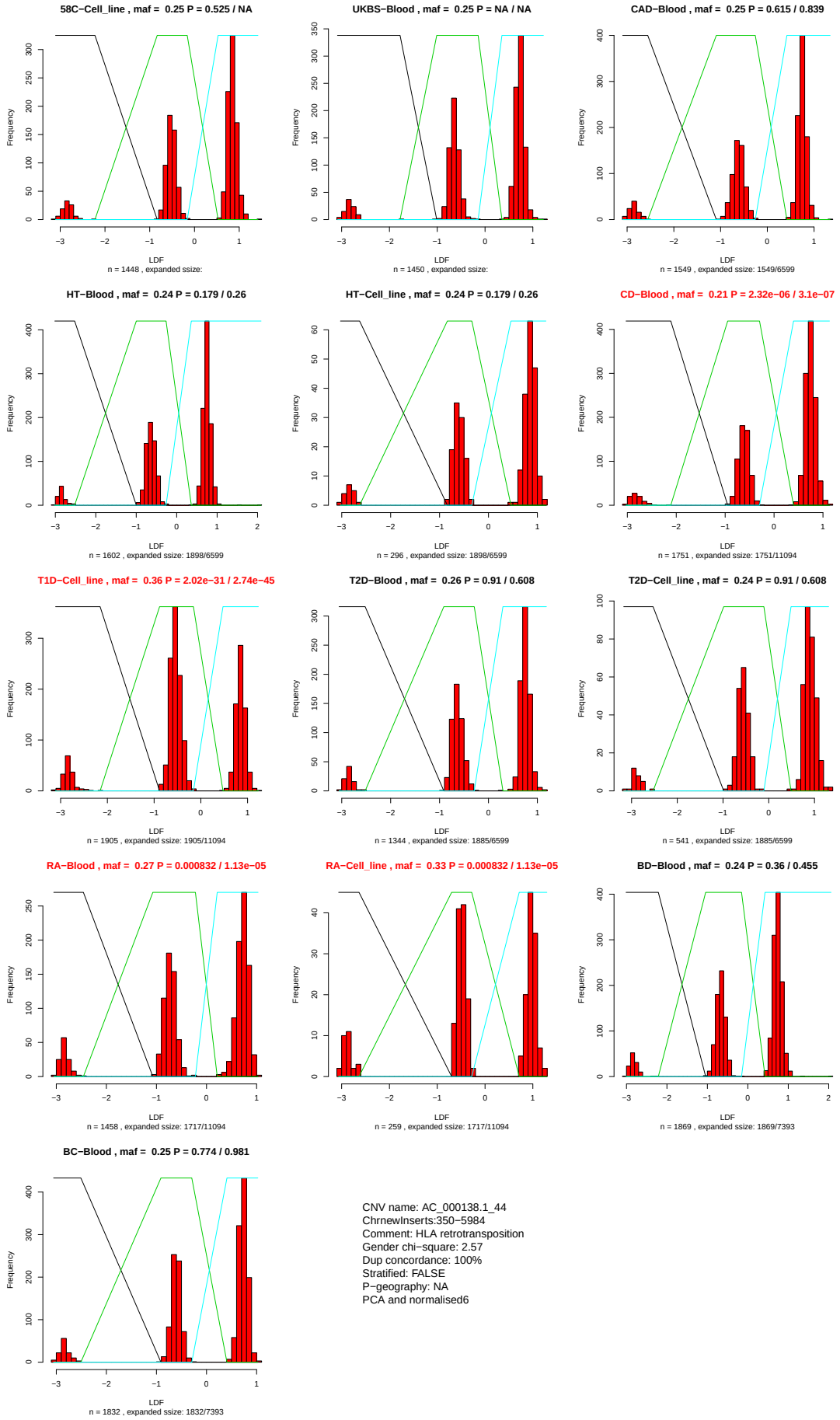
CNV name: CNVR2845.14
Chr6:32609209−32613170
Comment: ?
Gender chi−square: 9.31
Dup concordance: 98.6%
Stratified: FALSE
P−geography: NA
mean and normalised1

7 CNVR1065.1



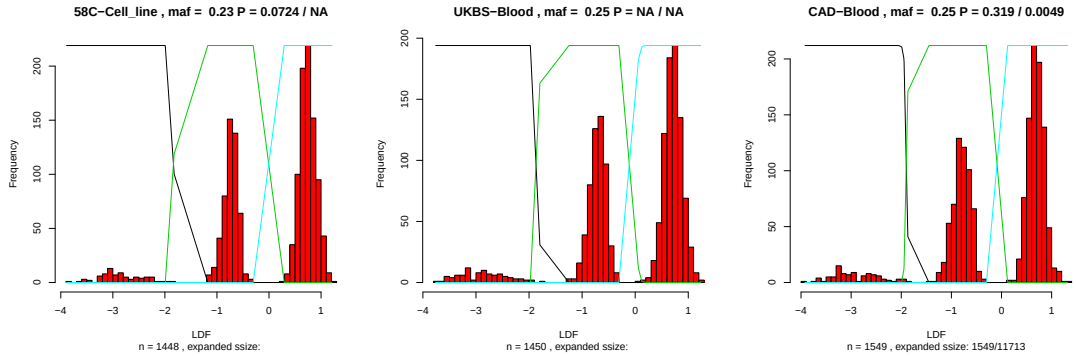
CNV name: CNVR1065.1
Chr2:179004449−179005231
Comment: ?
Gender chi−square: 10.3
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
mean and normalised1

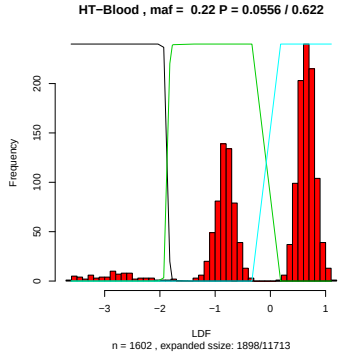
8 AC_000138.1_44

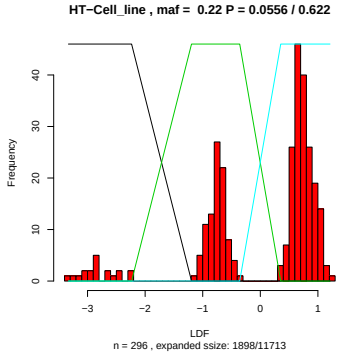


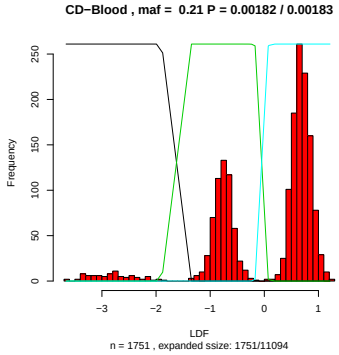
CNV name: AC_000138.1_44
ChrnewInserts:350−5984
Comment: HLA retrotransposition
Gender chi−square: 2.57
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
PCA and normalised6

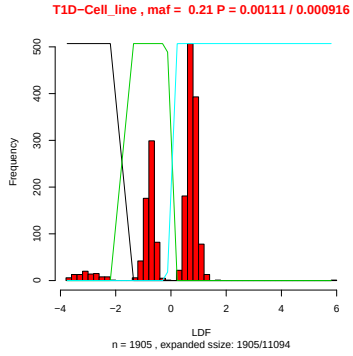
9 CNVR7113.6

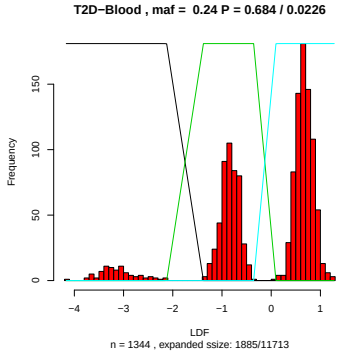


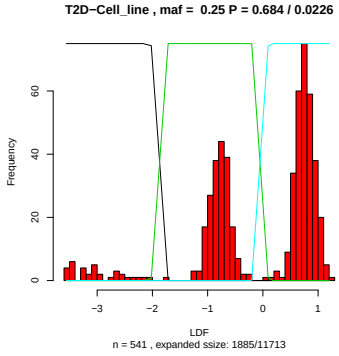


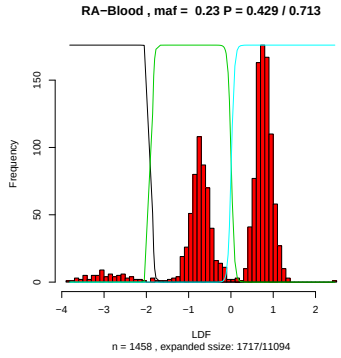


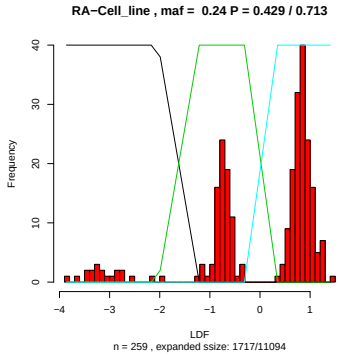


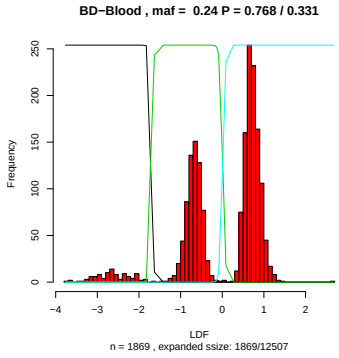


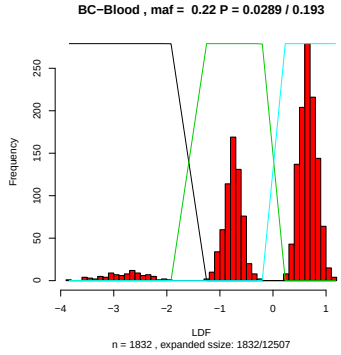






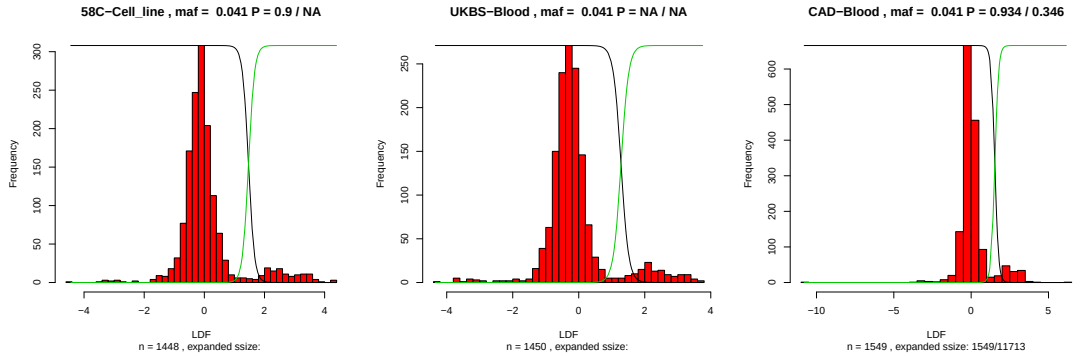


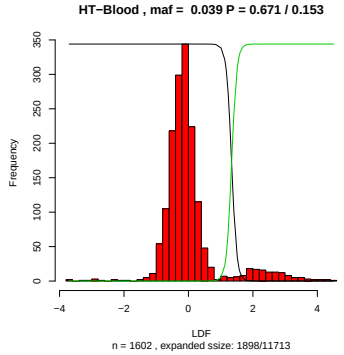


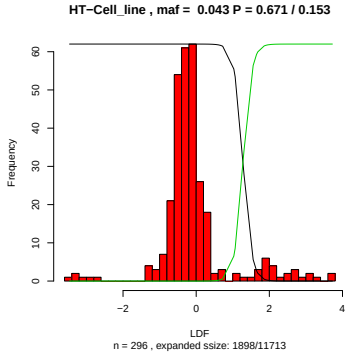


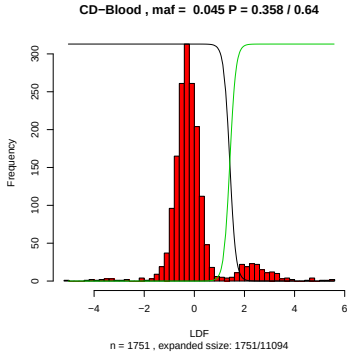
CNV name: CNVR7113.6
Chr17:40930407−40964305
Comment: ?
Gender chi−square: 0.317
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
PCA and normalised6

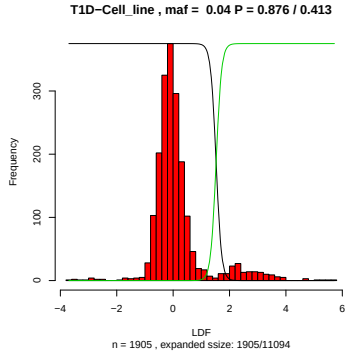
10 CNVR73.9

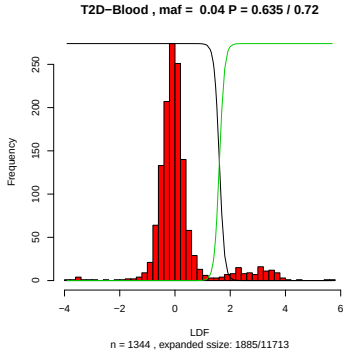


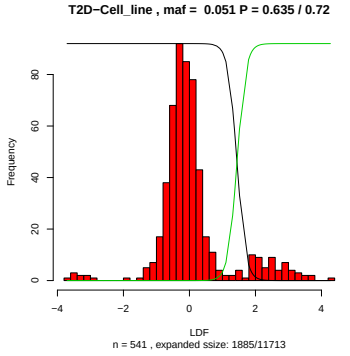


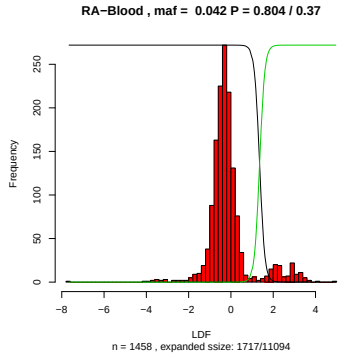


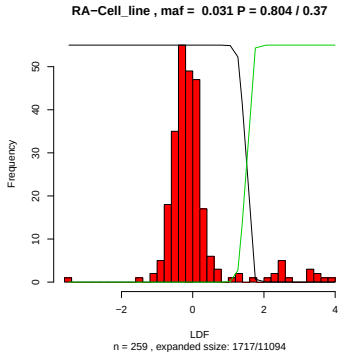


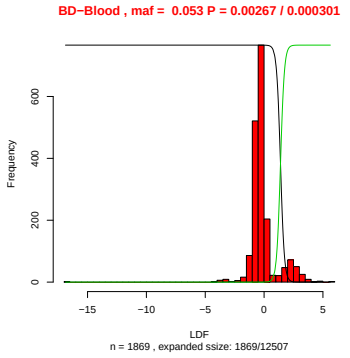


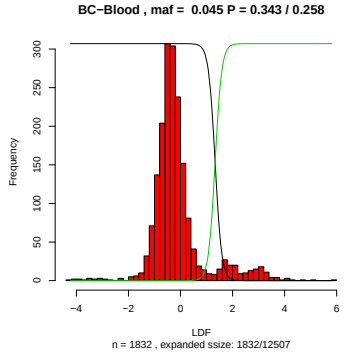






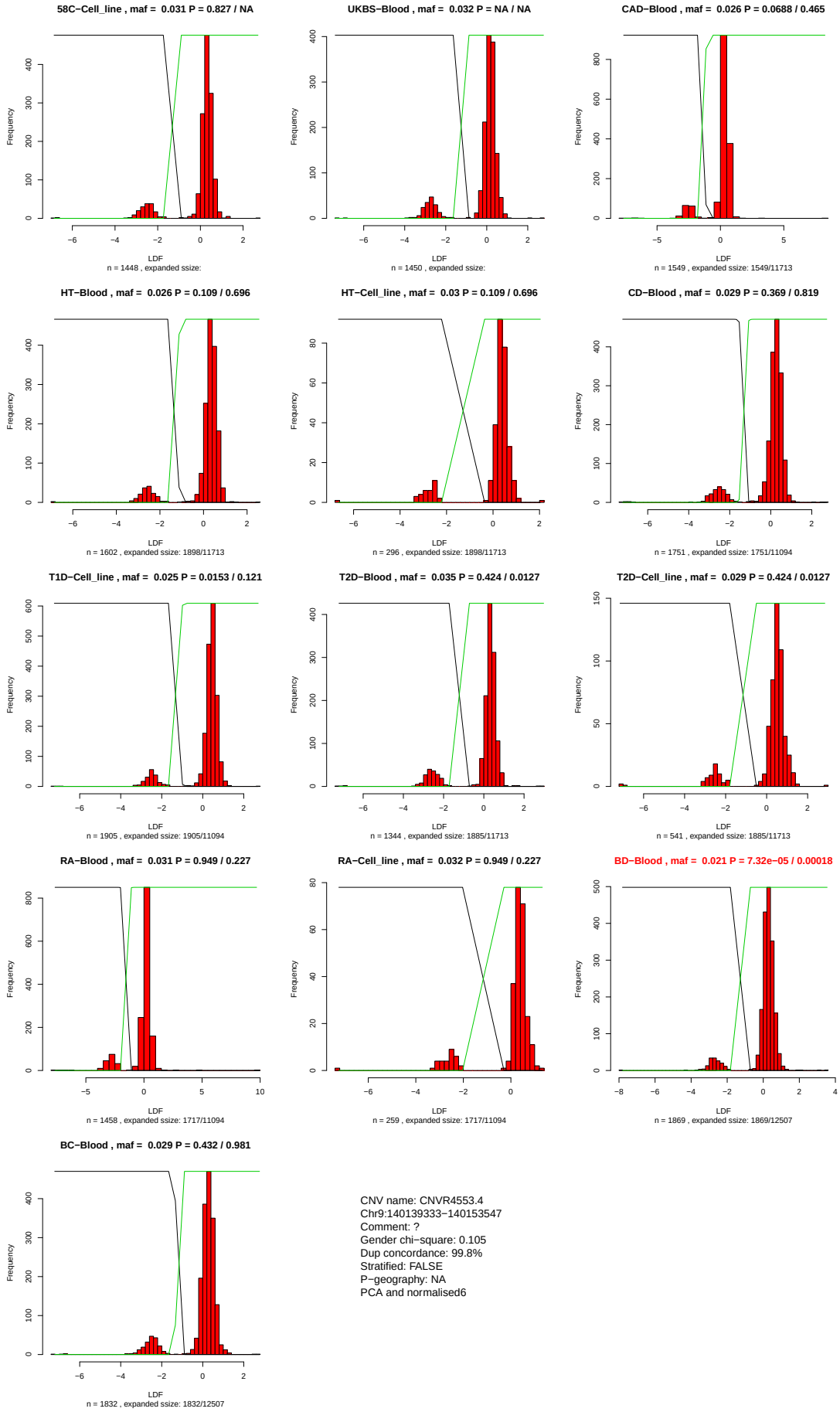






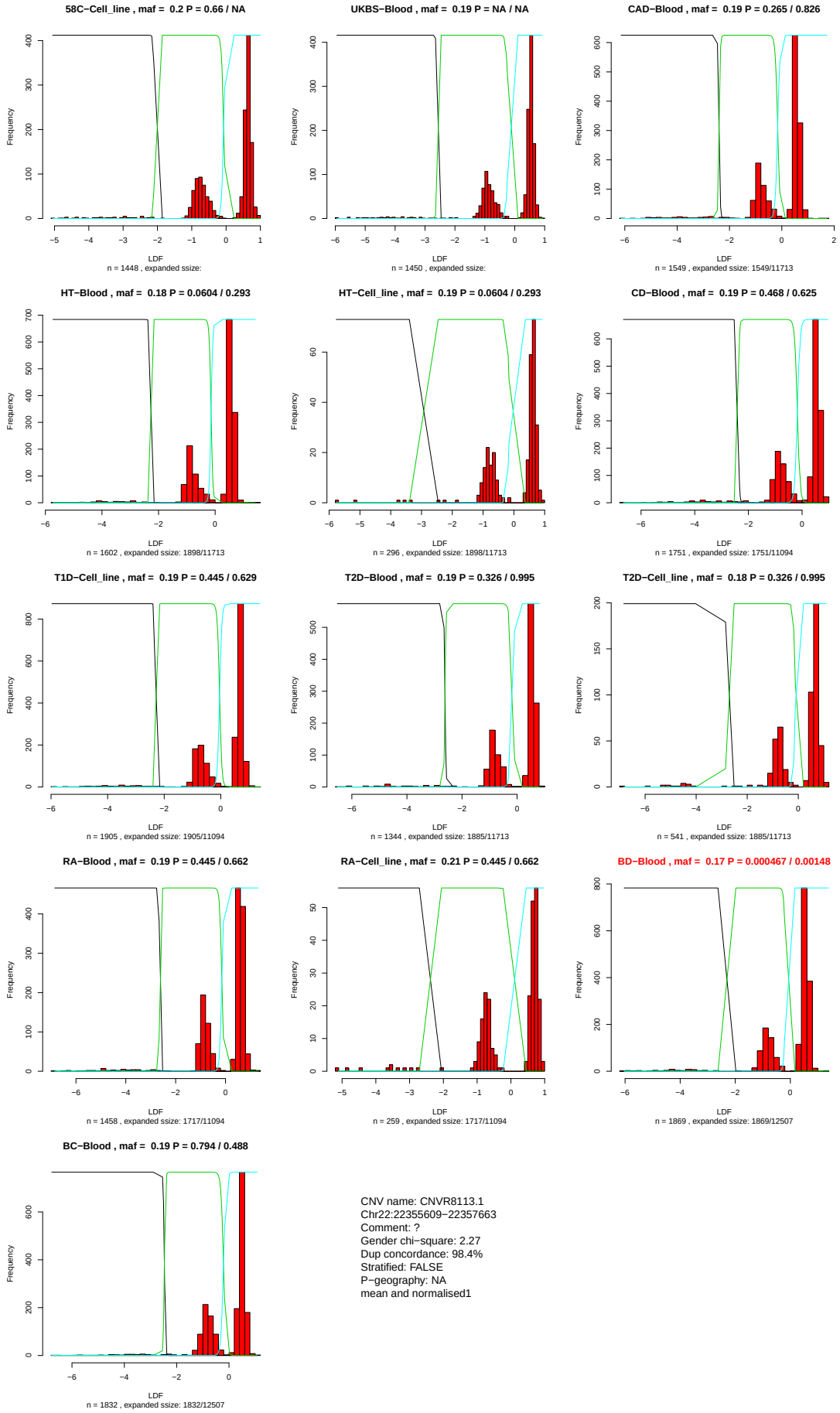
CNV name: CNVR73.9
Chr1:13094201−13113075
Comment: ?
Gender chi−square: 3.28
Dup concordance: 98.8%
Stratified: FALSE
P−geography: NA
PCA and normalised1

11 CNVR4553.4



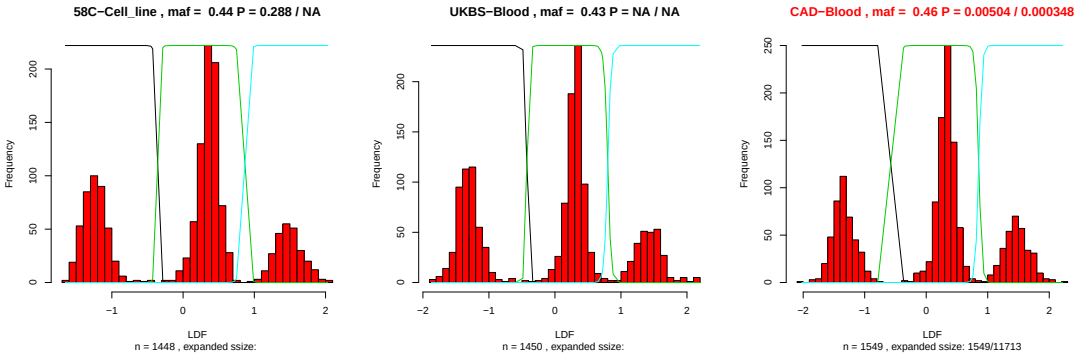
CNV name: CNVR4553.4
Chr9:140139333−140153547
Comment: ?
Gender chi−square: 0.105
Dup concordance: 99.8%
Stratified: FALSE
P−geography: NA
PCA and normalised6

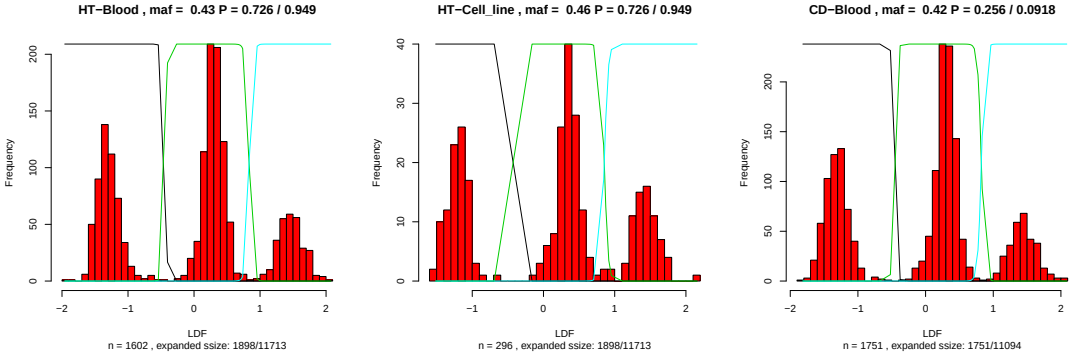
12 CNVR8113.1

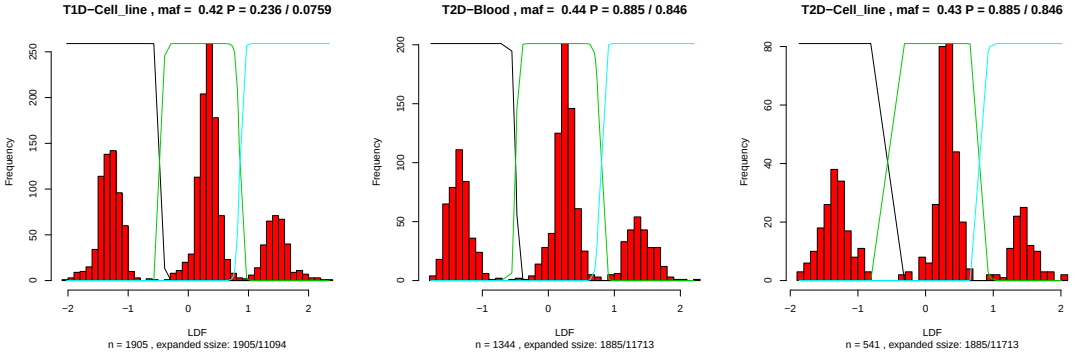


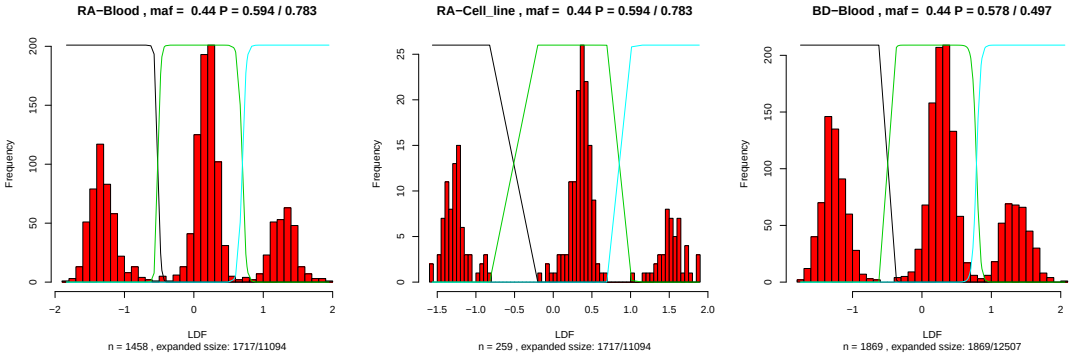
CNV name: CNVR8113.1
Chr22:22355609−22357663
Comment: ?
Gender chi−square: 2.27
Dup concordance: 98.4%
Stratified: FALSE
P−geography: NA
mean and normalised1

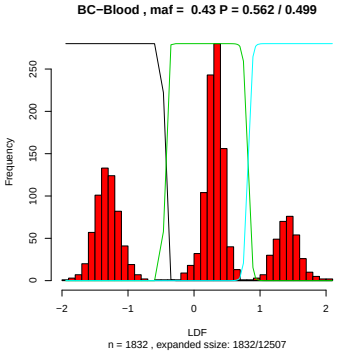
13 CNVR765.1





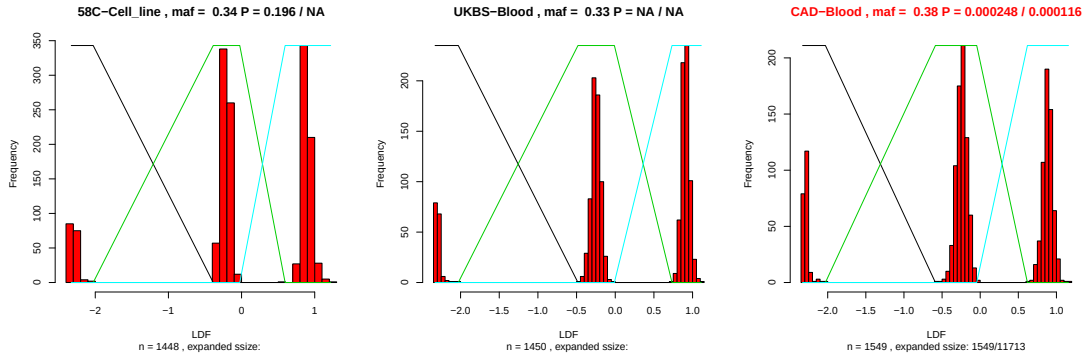


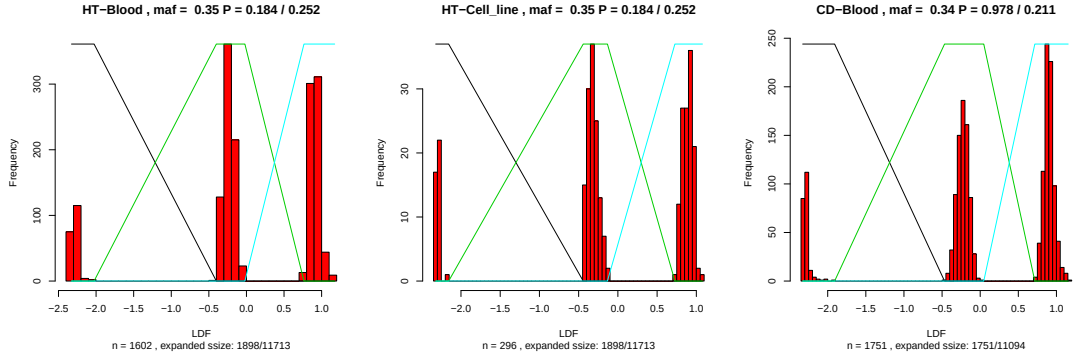


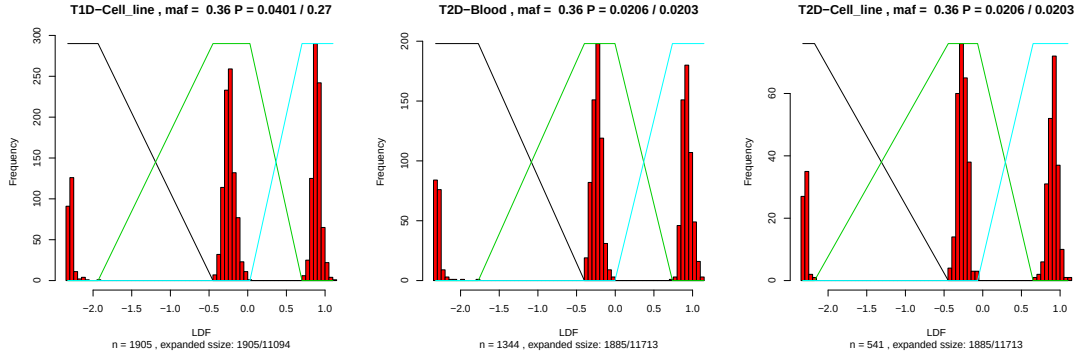


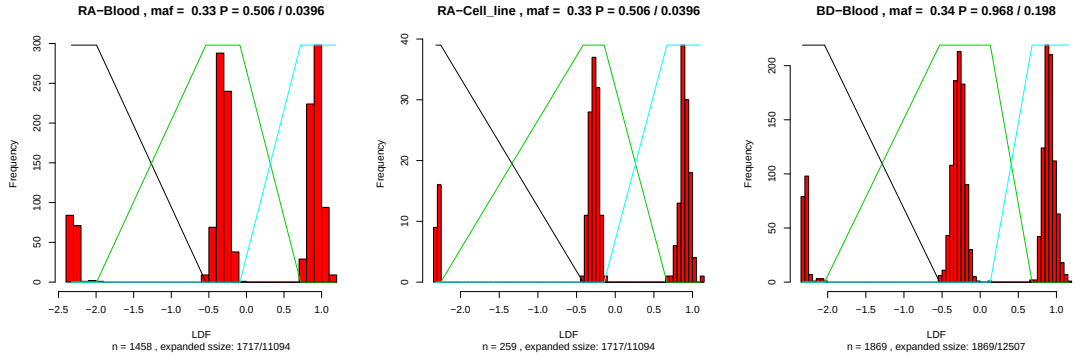
CNV name: CNVR765.1
Chr2:41817888−41818771
Comment: ?
Gender chi−square: 6.11
Dup concordance: 99.8%
Stratified: FALSE
P−geography: NA
PCA and normalised6

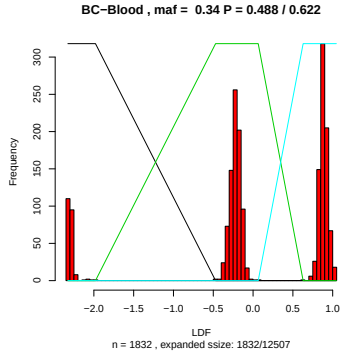
14 CNVR1152.1





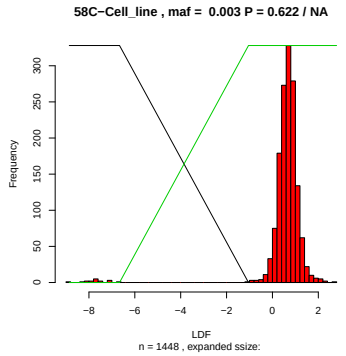


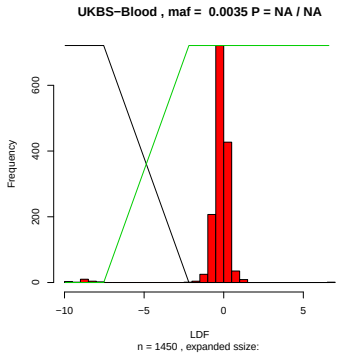


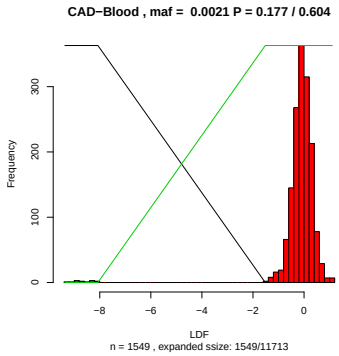


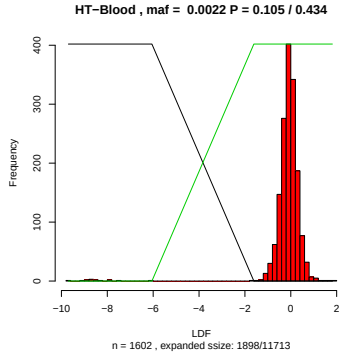
CNV name: CNVR1152.1
Chr2:226873606−226879267
Comment: ?
Gender chi−square: 7.75
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
PCA and normalised6

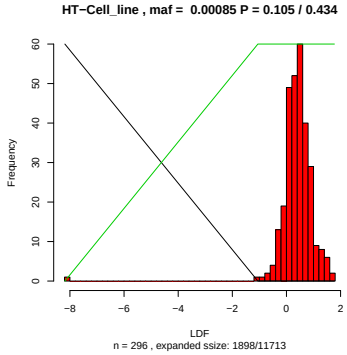
15 CNVR164.1

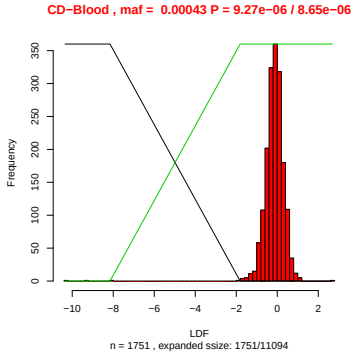


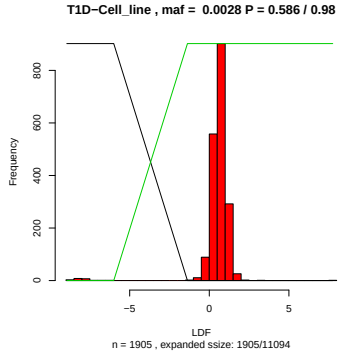


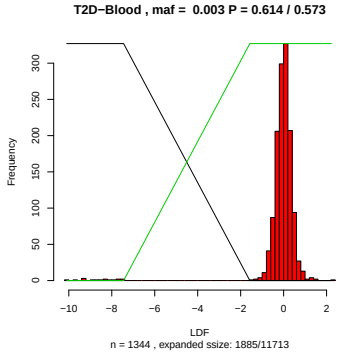


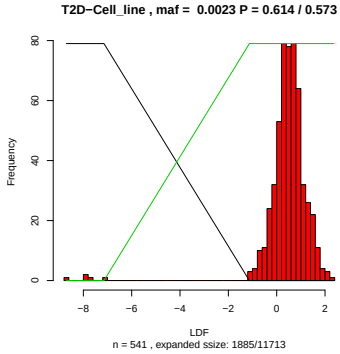


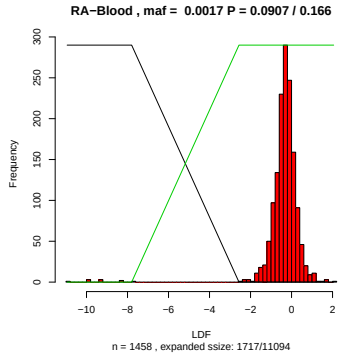


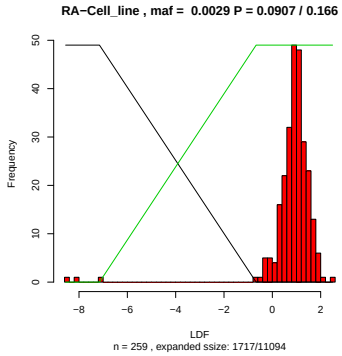


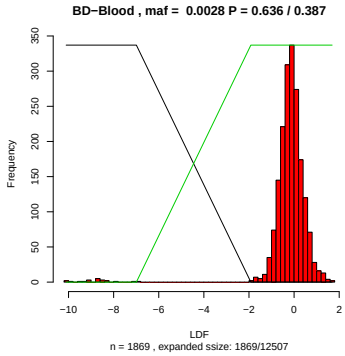


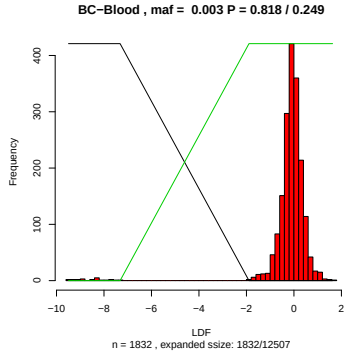






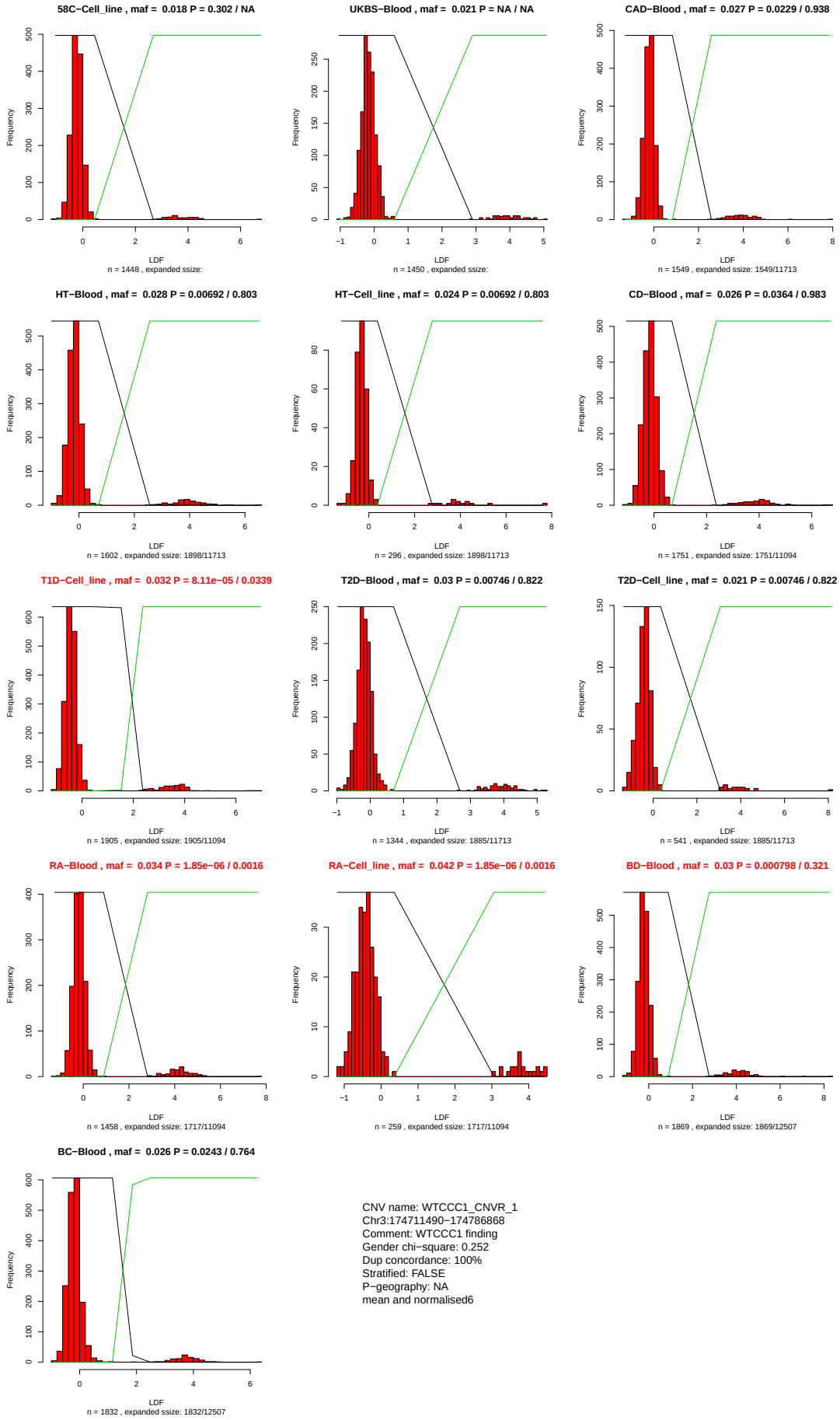






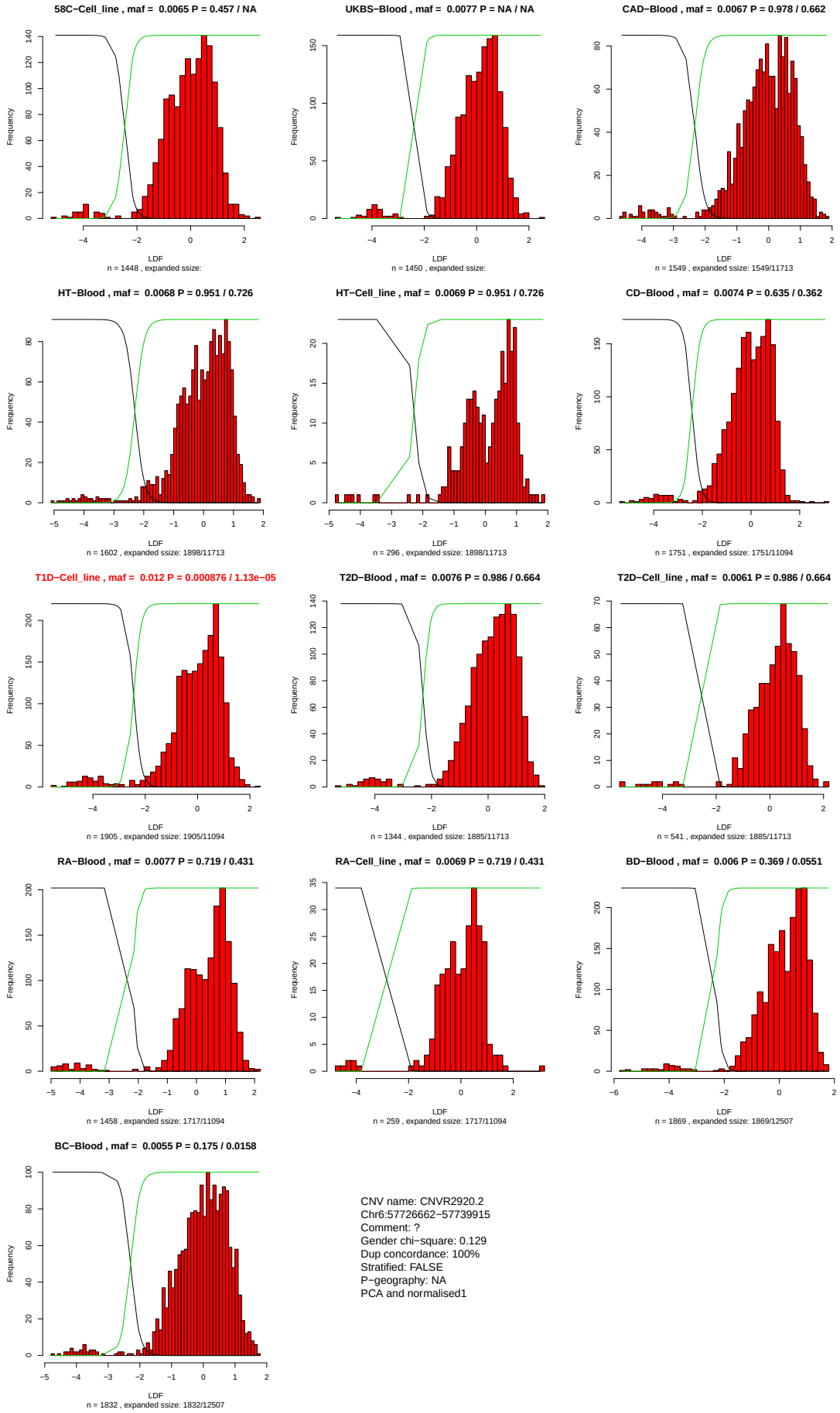
CNV name: CNVR164.1
Chr1:45407164−45418038
Comment: ?
Gender chi−square: 0.0119
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
PCA and normalised6

16 WTCCC1_CNVR_1



CNV name: WTCCC1_CNVR_1
Chr3:174711490−174786868
Comment: WTCCC1 finding
Gender chi−square: 0.252
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
mean and normalised6

17 CNVR2920.2



CNV name: CNVR2920.2
Chr6:57726662−57739915
Comment: ?
Gender chi−square: 0.129
Dup concordance: 100%
Stratified: FALSE
P−geography: NA
PCA and normalised1