Additional figures for:

Human genomic regions with exceptionally high or low levels of population differentiation identified from 911 whole-genome sequences

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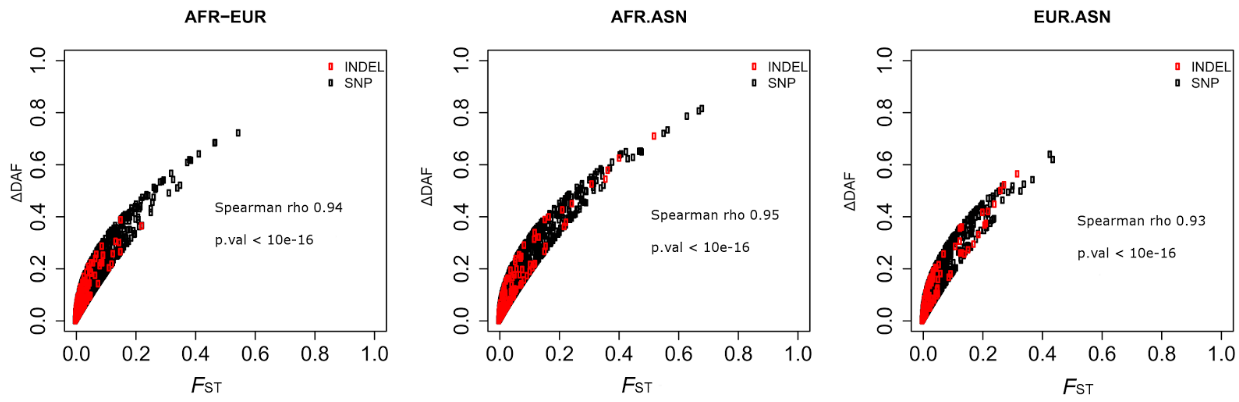
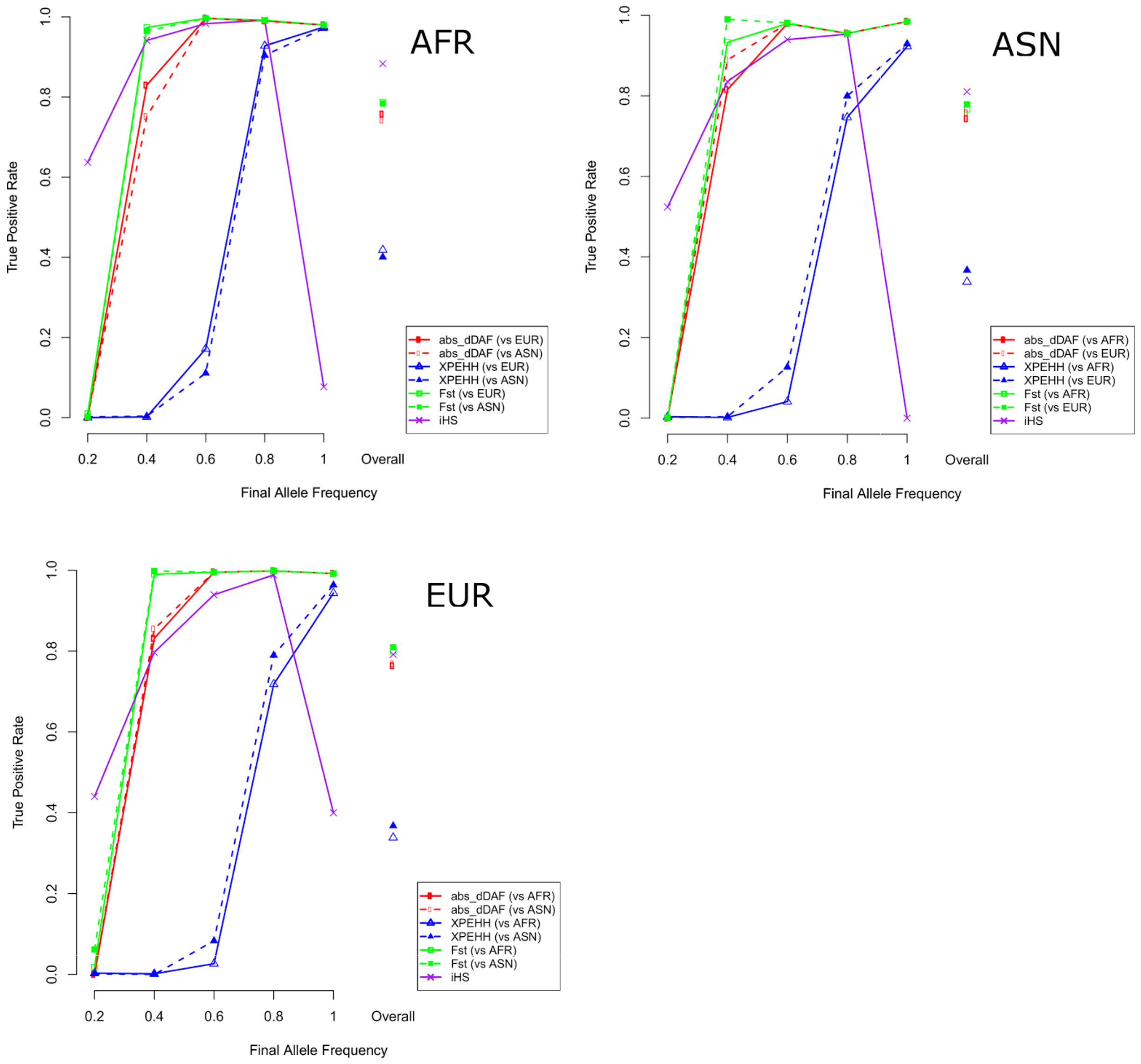
This file includes Supplementary Figures 1 to 16

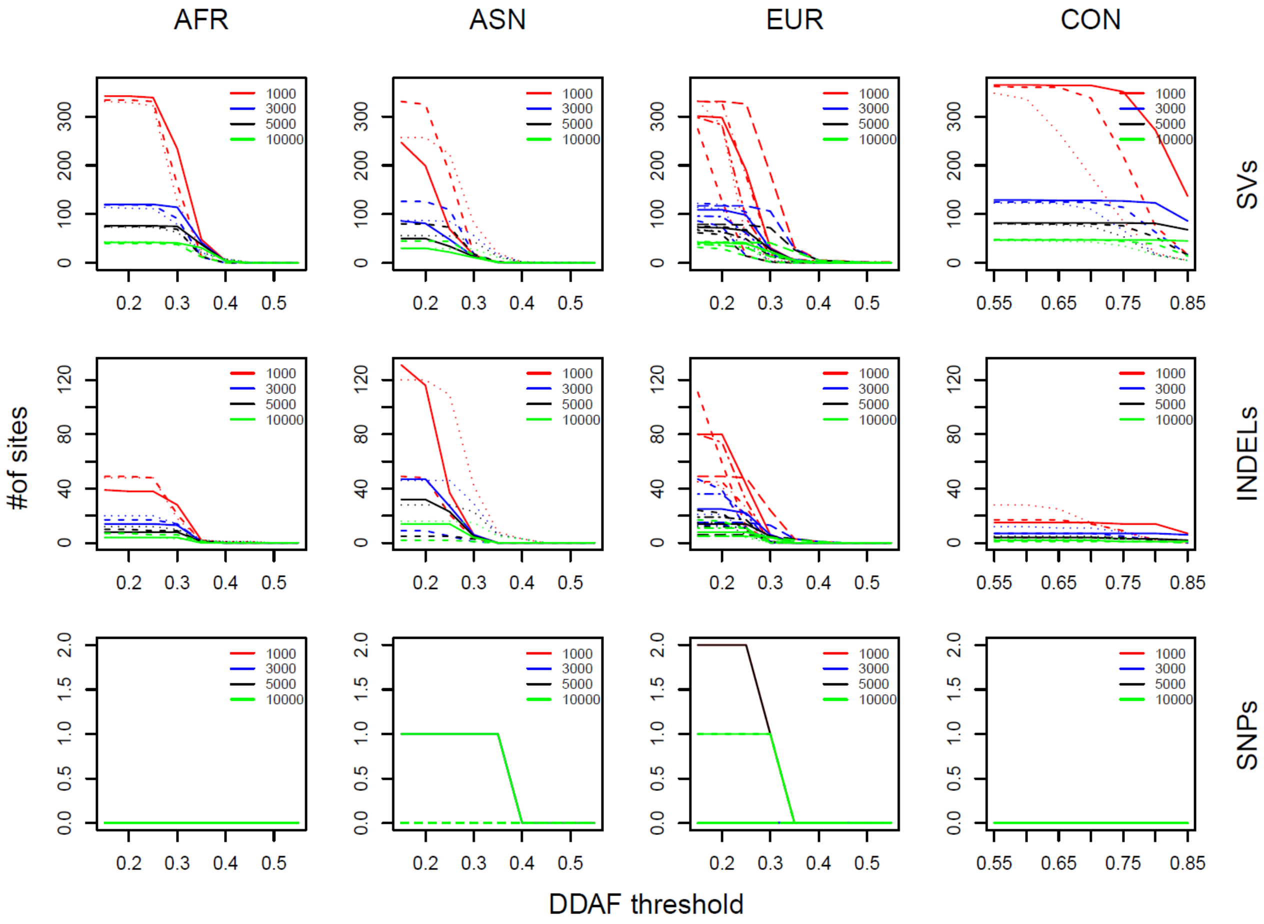
Supplementary Figure 1. Correlation between F_{ST} and ΔDAF a set of 5,000 random sites from chromosome 20



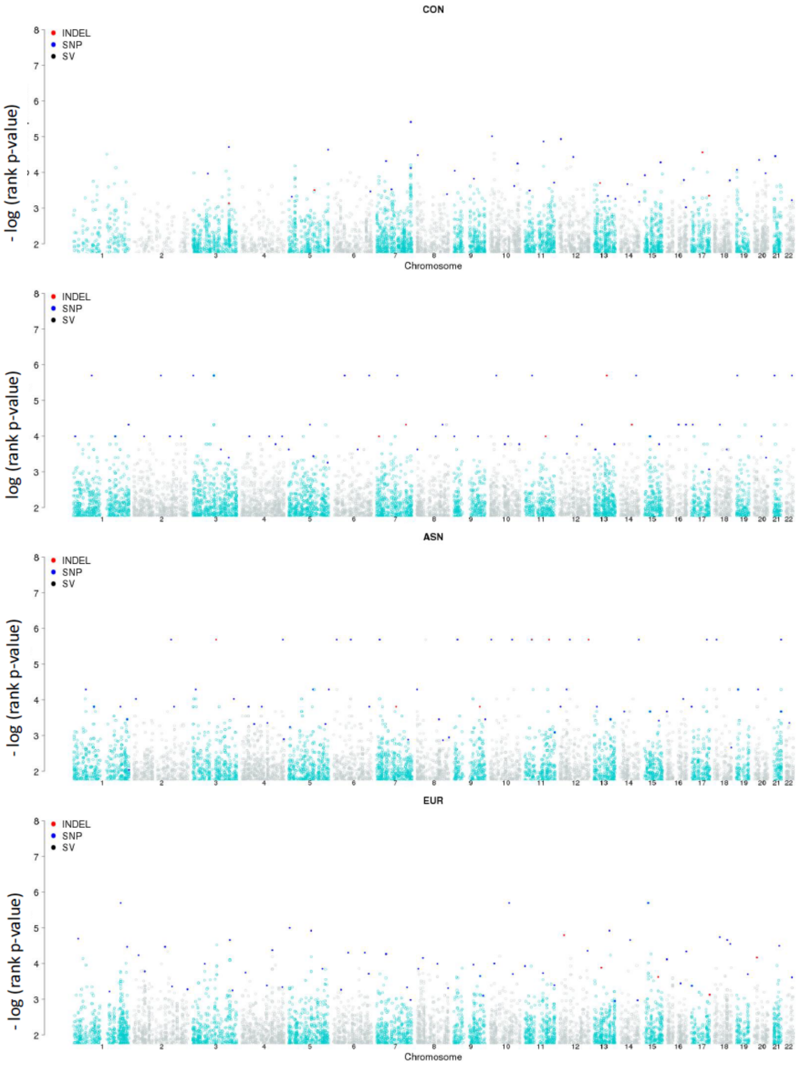
Supplementary Figure 2 . ΔDAF power to detect selection in simulated data. (See manuscript for details)



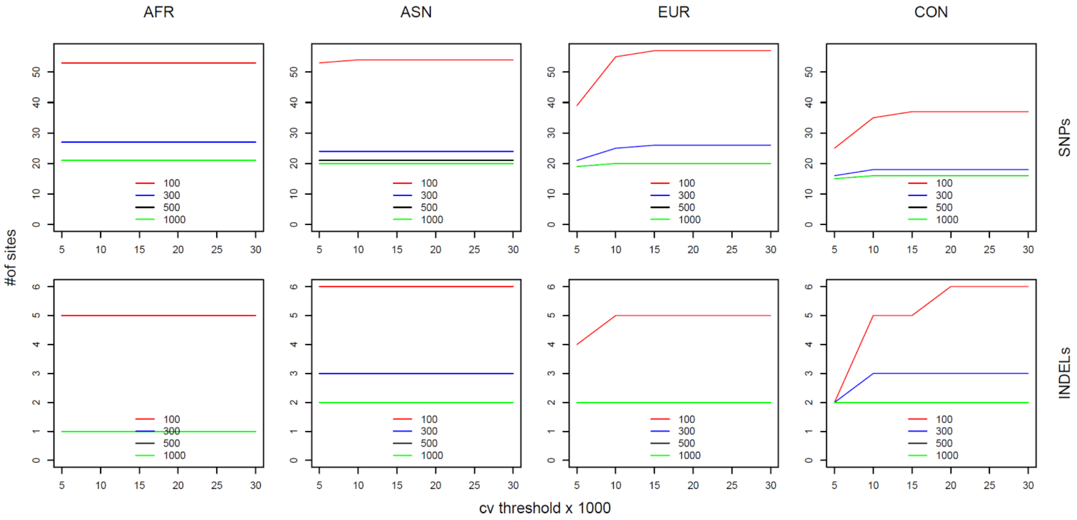
Supplementary Figure 3. Number of HighD sites according to different ΔDAF threshold and windows size.



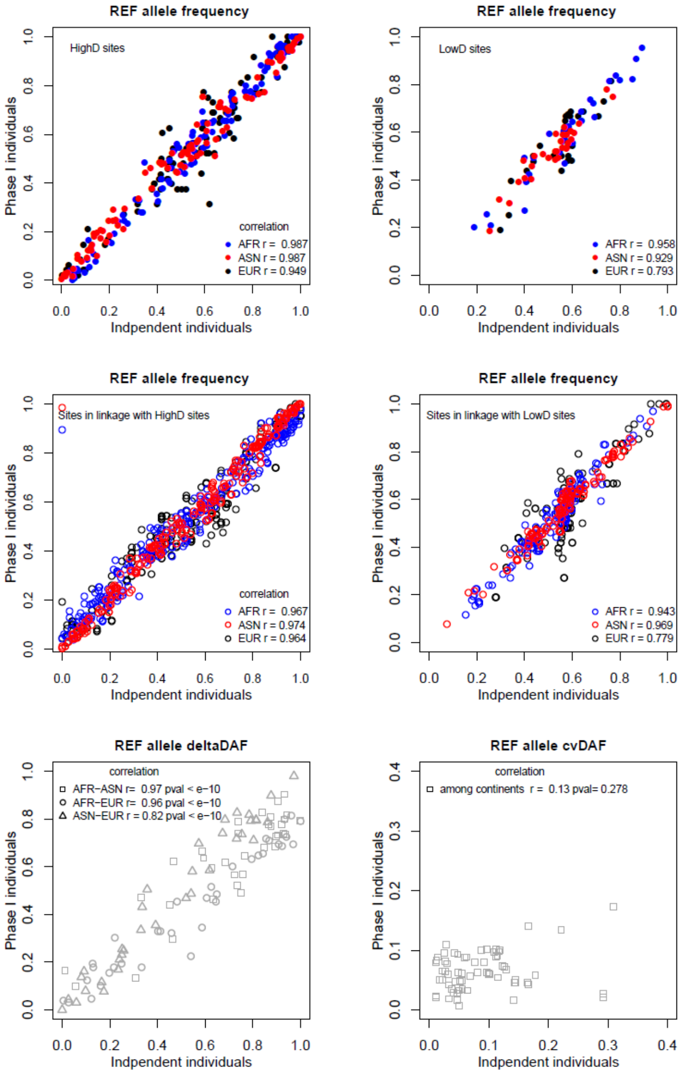
Supplementary Figure 4. Manhattan plots of rank p-values for cvDAFs at continental and populations levels. Chromosomes are represented by alternating turquoise and grey colors; red, blue and black dots represent INDEL, SNP and SV sites, respectively, that have been identified as low differentiated (LowD sites).



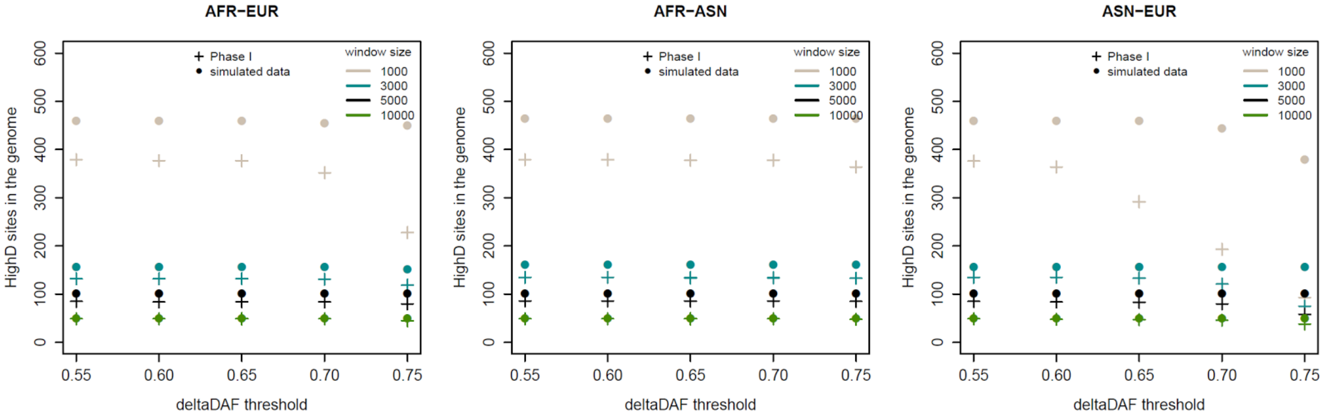
Supplementary Figure 5. Number of LowD sites according to different ΔDAF thresholds and windows sizes.



Supplementary Figure 6. Validation results in independent HapMap samples.

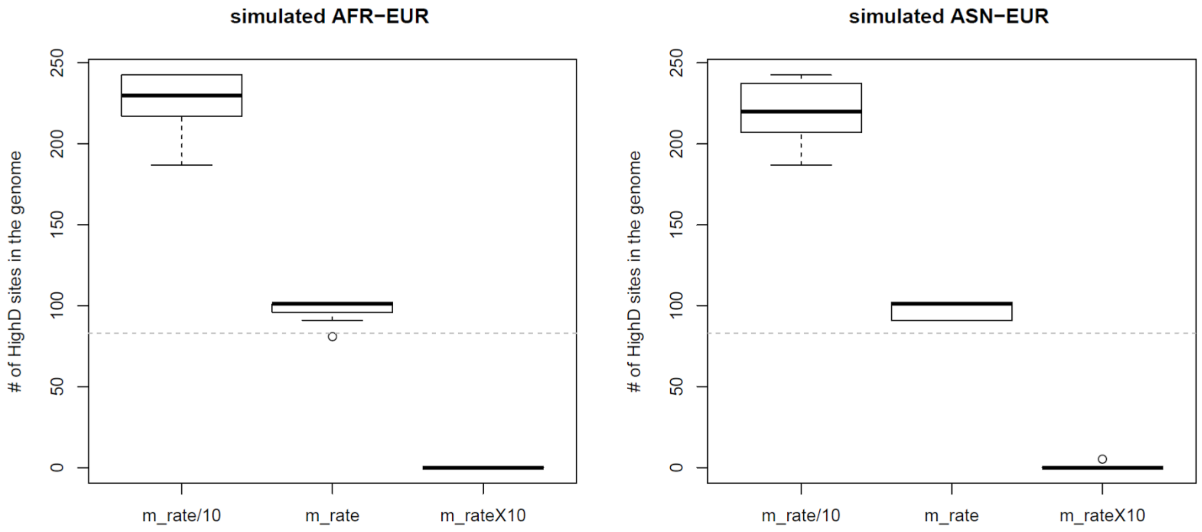


Supplementary Figure 7. Expected and observed number of HighD sites under different conditions of window sizes and ΔDAF thresholds.

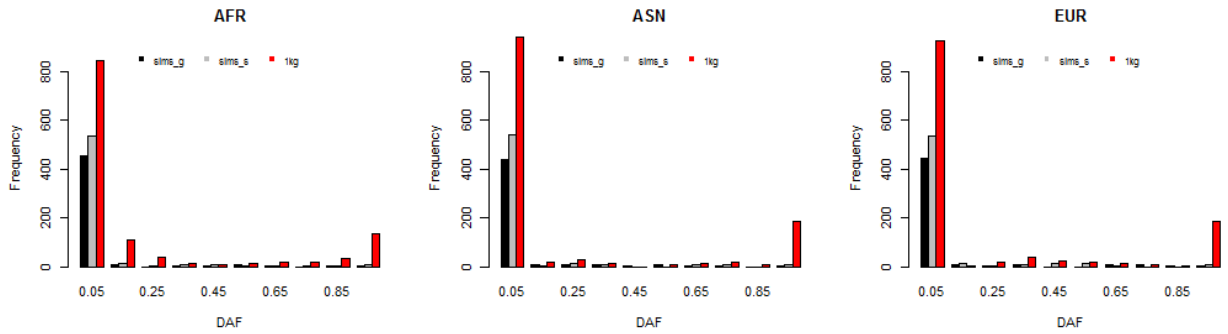


Supplementary Figure 8. Sensitivity to migration rate (m_rate) of the expected number of HighD sites from simulations under neutrality and comparison of two different demographic models. (a) Simulations of the AFR-EUR and ASN-EUR comparisons under the model proposed by Gravel and colleagues (Gravel S, et al. PNAS 2011, 108(29):11983-11988) as a complement to what presented in the main text. Dashed line represents number of observed HighD sites. (b) Comparison of allele frequency spectra of 1000 Genome data (red bars) with two published models (Gravel S, *et al.* PNAS 2011, 108(29):11983-11988 indicated as sims_g and Schaffner SF, *et al*. Genome Res 2005, 15(11):1576-1583 indicated as sims) and (c) number of expected HighD sites for simulated data under the model quoted above and another model (references X in the main text)

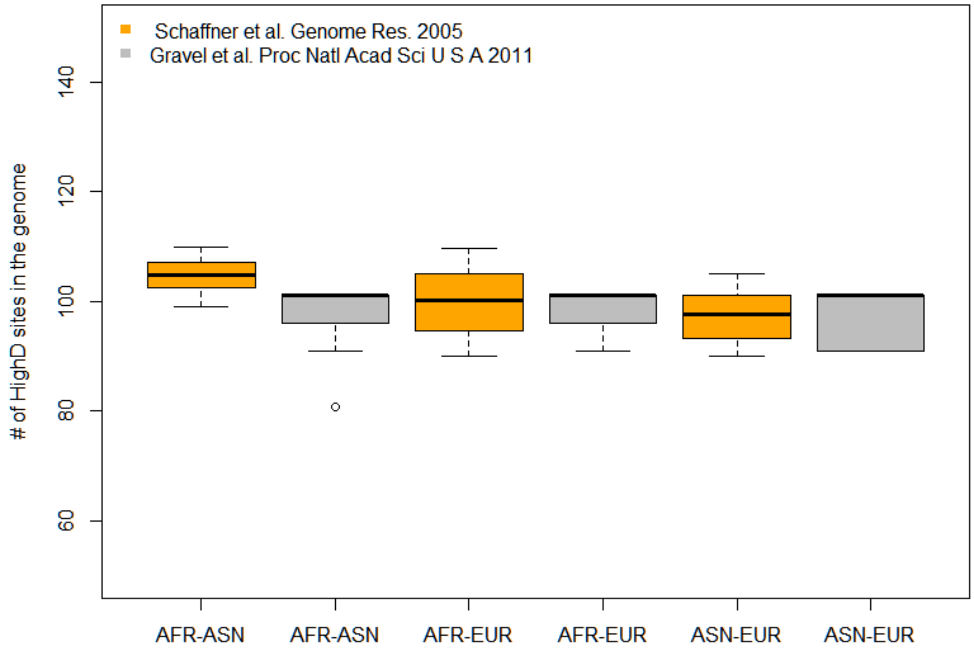
(a)



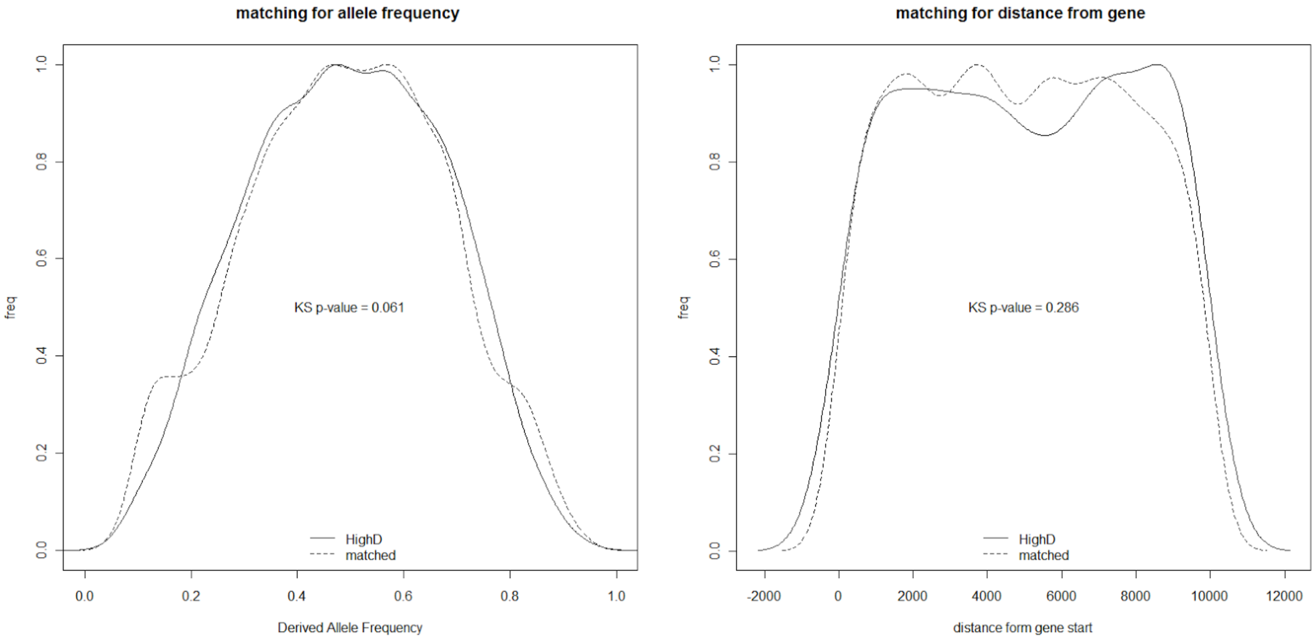
(b)


AFR
ASN
EUR
sims_g
sims_s
1kg
Frequency
0
200
400
600
800
0.05
0.25
0.45
0.65
0.85
DAF

(c)


Schaffner et al. Genome Res. 2005
Gravel et al. Proc Natl Acad Sci U S A 2011
# of HighD sites in the genome
60
80
100
120
140
AFR-ASN
AFR-ASN
AFR-EUR
AFR-EUR
ASN-EUR
ASN-EUR

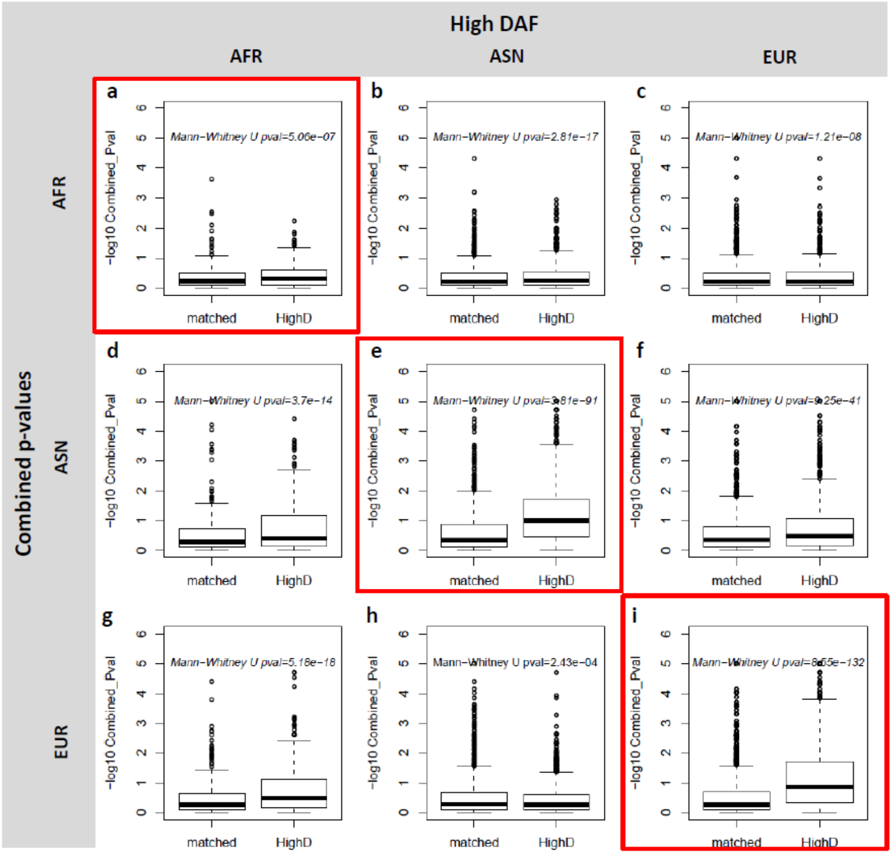
Supplementary Figure 9. Features of genomic sites matched for allele frequency and distance from gene.



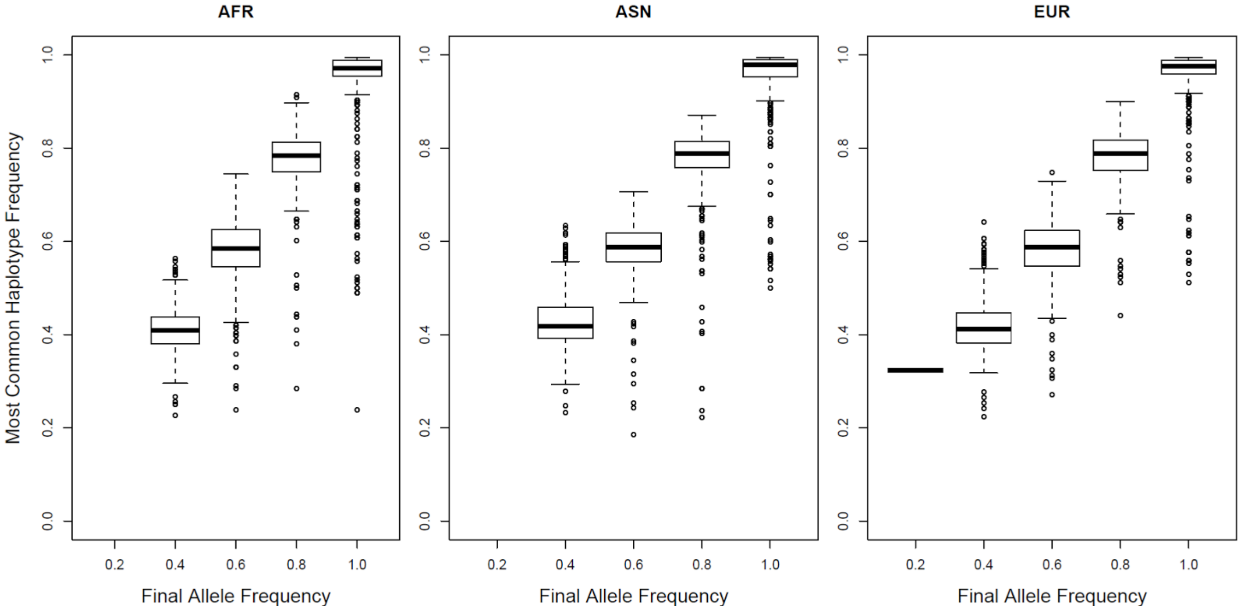
matching for allele frequency

matching for distance from gene

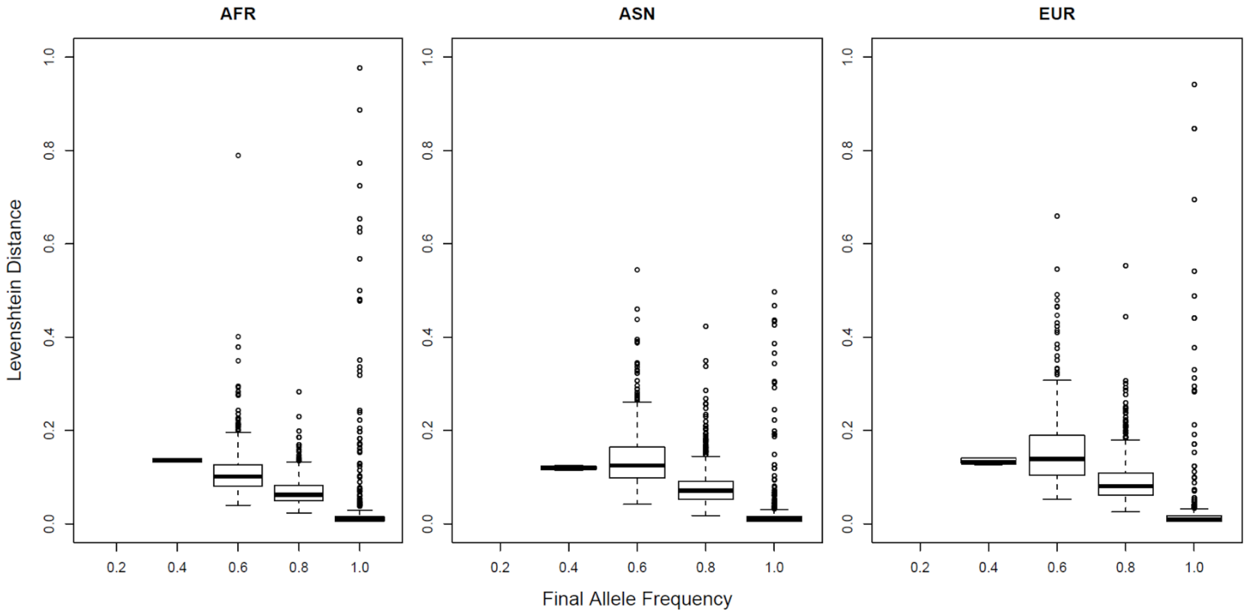
Supplementary Figure 10. Combined p-value for Tajima's D, Fay&Wu's H and Nielsen's CLR values in HighD sites and matched controls. Each column refer to a subset of HighD sites that have highest DAF in the population indicated in the head of the column; population indicated in rows are the population in which the combined p-values has been calculated for the set of sites in the column.



Supplementary Figure 11. Simulated data showing the frequency of the most common haplotype in 2kb surrounding a site under positive selection for a range of selective pressures leading to different final allele frequency at the selected site (on the x-axis). For hard sweeps (final allele frequency=1) there is mostly a single haplotype

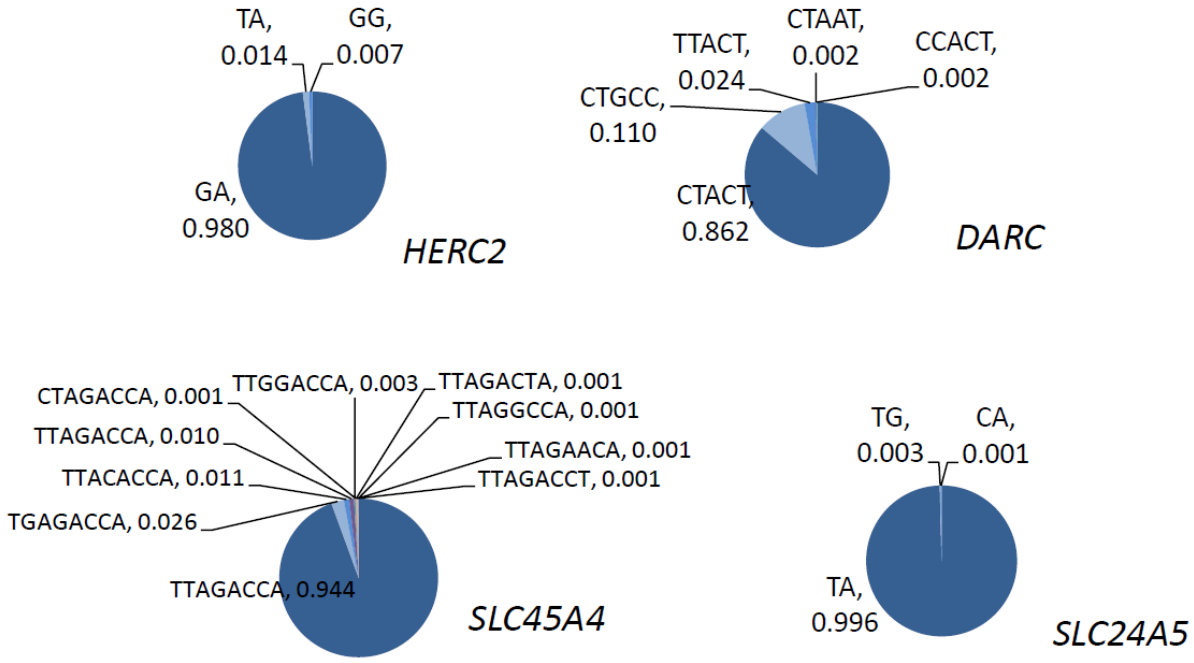


Supplementary Figure 12. Boxplots of Levenshtein distance from the major haplotype of all other haplotypes in 2kb surrounding sites under positive selection in simulated data. In the table below average values relative to boxplots. FaF=final allele frequency of the site under selection

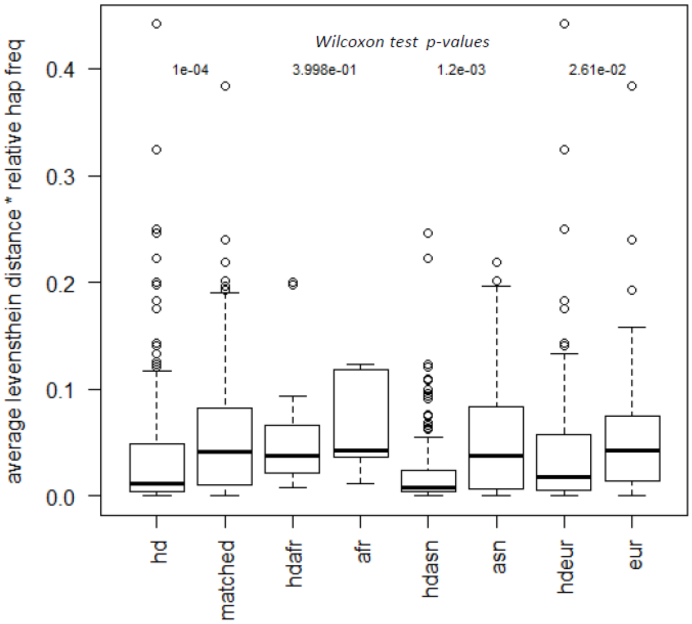


FaF	EUR	ASN	AFR
0.4	0.1340252	0.1198131	0.1356534
0.6	0.1547858	0.1388615	0.1105228
0.8	0.09283899	0.08026323	0.06947882
1.0	0.03144997	0.02643067	0.04296775

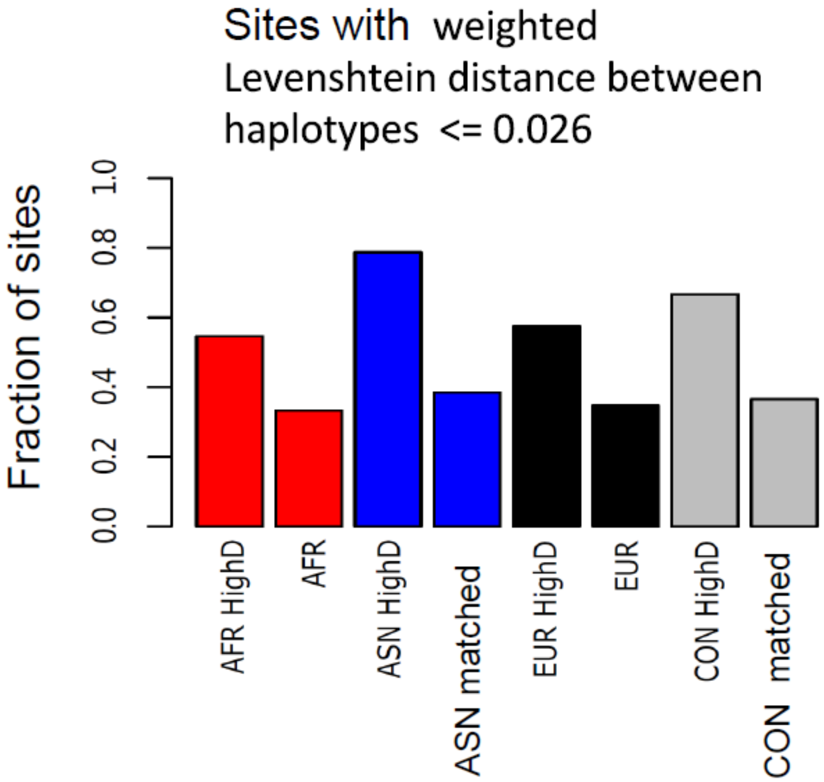
Supplementary Figure 13. Frequencies of haplotypes in 2 kb surrounding HighD sites demonstrated to be functional in examples of positive selection.



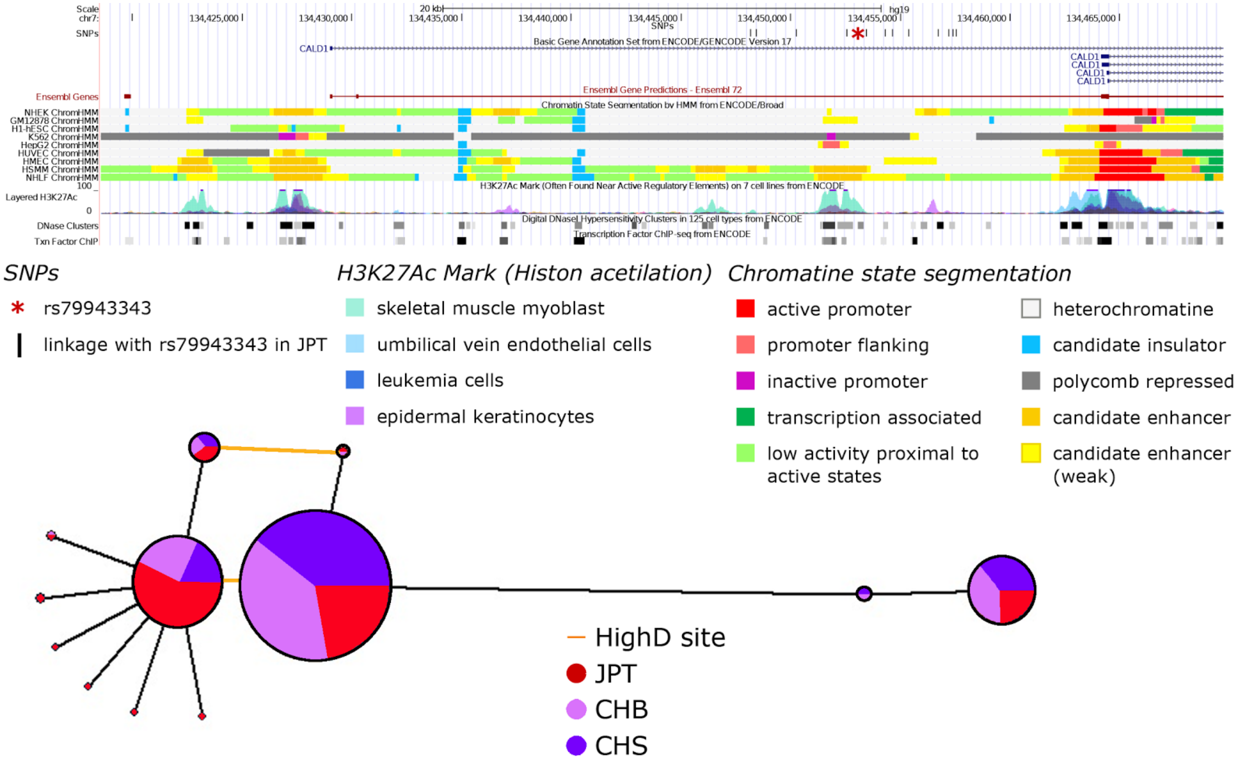
Supplementary Figure 14. Boxplots showing the distribution of weighted Levensthein distance in HighD sites and in matched controls for continental comparisons.



Supplementary Figure 15. Fraction of HighD and matched genomic sites with haplotypic features similar to sites accepted as examples of classic selective sweeps.



Supplementary Figure 16. Functional annotations in the genomic region surrounding the HighD site in *CALD1* and median joining network of haplotypes surrounding the site. Haplotypes are derived from sites in linkage disequilibrium (D’=1) with the HighD site in JPT populations.



Scale
chr7:
SNPs
20 kb
hg19
134,425,000
134,430,000
134,435,000
134,440,000
134,445,000
134,450,000
134,455,000
134,460,000
134,465,000
Basic Gene Annotation Set from ENCODE/GENCODE Version 17
CALD1
Ensembl Genes
Ensembl Gene Predictions - Ensembl 72
Chromatin State Segmentation by HMM from ENCODE/Broad
NHEK ChromHMM
GM12878 ChromHMM
H1-hESC ChromHMM
K562 ChromHMM
HepG2 ChromHMM
HUVEC ChromHMM
HMEC ChromHMM
HSMM ChromHMM
NHLF ChromHMM
100
Layered H3K27Ac
0
H3K27Ac Mark (Often Found Near Active Regulatory Elements) on 7 cell lines from ENCODE
DNase Clusters
Digital DNaseI Hypersensitivity Clusters in 125 cell types from ENCODE
Txn Factor ChIP
Transcription Factor ChIP-seq from ENCODE
SNPs
* rs79943343
| linkage with rs79943343 in JPT
H3K27Ac Mark (Histon acetilation)
skeletal muscle myoblast
umbilical vein endothelial cells
leukemia cells
epidermal keratinocytes
Chromatine state segmentation
active promoter
promoter flanking
inactive promoter
transcription associated
low activity proximal to active states
heterochromatine
candidate insulator
polycomb repressed
candidate enhancer
candidate enhancer (weak)
HighD site
JPT
CHB
CHS