Supplementary Materials

Table S1: Cutoffs for BF corresponding to type I error rate of 0.01 obtained from simulated data (ρ = 0, 0.3, 0.7, and 0.99) and GAW19 data on ULK4 and ZNF280D genes (ρ = 0.55). A cutoff is obtained for each combination of Freq, # Hap, and ρ values. A haplotype is considered rare if its frequency is less than or equal to 0.02.

Method	Freq	# Hap	ρ = 0, 0.3	ρ = 0.55, 0.7, 0.99
Bivariate LBL	rare	<= 8	3.54	2.33
	rare	> 8	1.97	1.67
	common	<= 8	0.64	0.71
	common	> 8	0.83	0.68
Univariate LBL	rare	<= 8	9.17	5.18
	rare	> 8	6.16	4.68
	common	<= 8	2.92	3.43
	common	> 8	3.42	3.22

Freq: Haplotype frequency, # Hap: Number of haplotypes in the window/block

Table S2: Empirical type I error rates (in %) calculated using the simulated data. Cutoffs for BF used for declaring significance correspond to type I error rate of 1% and are listed in Table S1.

Setting	Haplotype	Freq	$\rho = 0$	$\rho = 0.3$	$\rho = 0.7$	$\rho = 0.99$
1	h01100	0.300	0.2	0.2	0.2	0.2
1	h10100	0.005	1.2	0.6	1.6	1.2
1	h11011	0.010	0.9	1.1	1.2	0.8
1	h11100	0.155	0.5	0.5	0.3	0.5
1	h11111	0.110	1.0	0.8	1.5	0.3
2	h01010	0.060	1.9	1.3	1.4	0.7
2	h01100	0.250	0.4	0.3	0.0	0.0
2	h10000	0.080	1.6	0.8	0.7	0.9
2	h10100	0.005	0.7	1.0	1.1	0.3
2	h11011	0.010	1.5	0.6	0.6	0.3
2	h11100	0.090	1.9	1.4	0.6	0.4
2	h11101	0.085	1.0	1.3	0.6	0.6
2	h11111	0.100	0.6	0.5	0.2	0.5
3	h00111	0.070	0.9	1.4	0.7	0.4
3	h01000	0.020	0.6	1.0	0.6	0.3
3	h01011	0.050	1.2	1.4	1.2	1.0
3	h01101	0.060	1.3	1.8	1.1	0.2
3	h01110	0.140	0.9	0.2	0.3	0.1
3	h10010	0.080	1.0	1.1	0.8	0.4
3	h10100	0.005	1.0	1.1	0.6	0.1
3	h11011	0.010	1.3	0.9	0.9	0.2
3	h11101	0.090	1.2	0.7	0.9	0.1
3	h11110	0.130	0.8	0.4	0.4	0.3
3	h11111	0.100	1.1	0.7	0.3	0.1

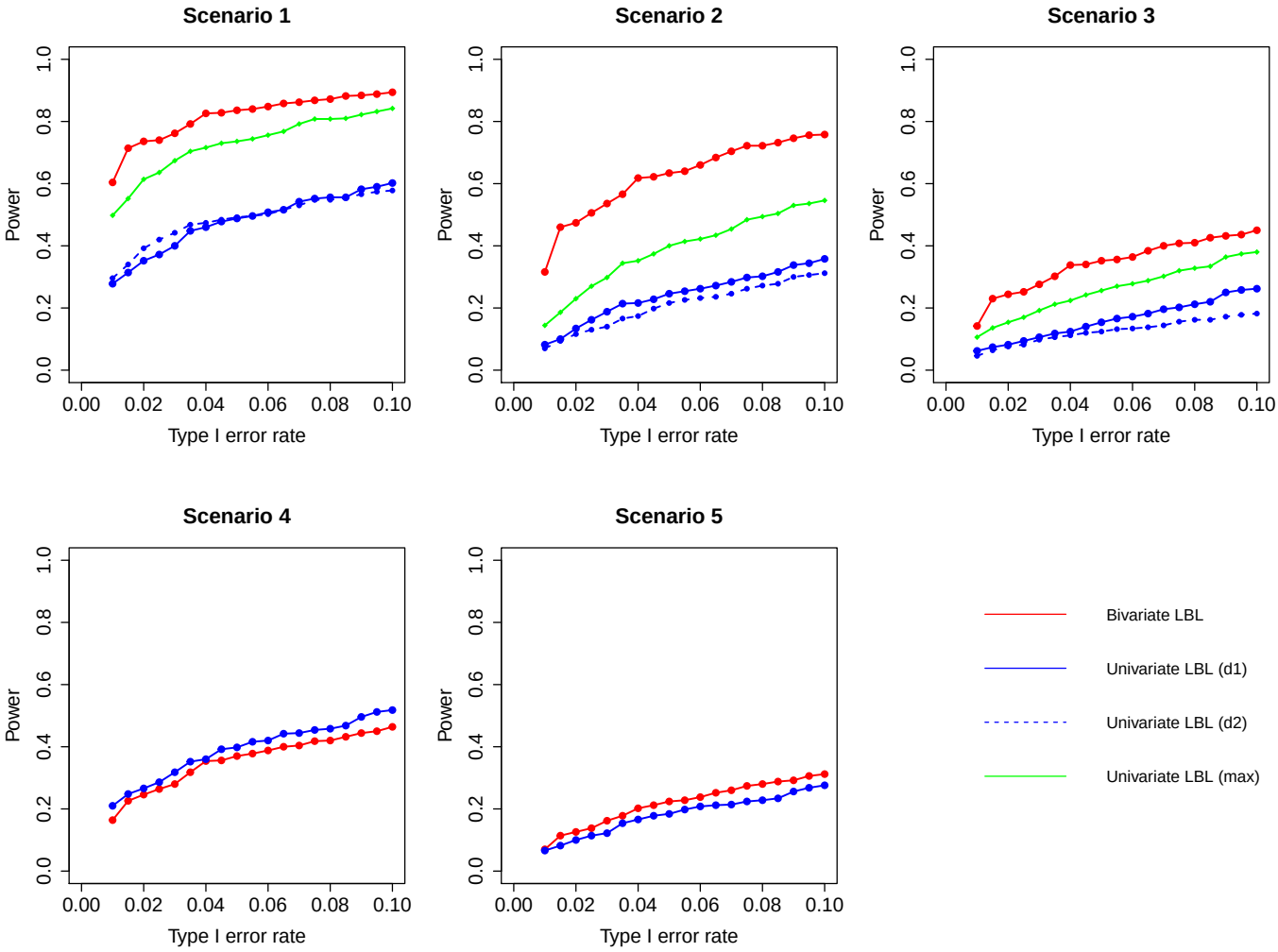

Figure S1: Simulation results for hypothesis 1 under setting 2 (9 haplotypes) and $\rho = 0$. The scenarios are listed in Table 1. d1=disease 1, d2=disease 2.

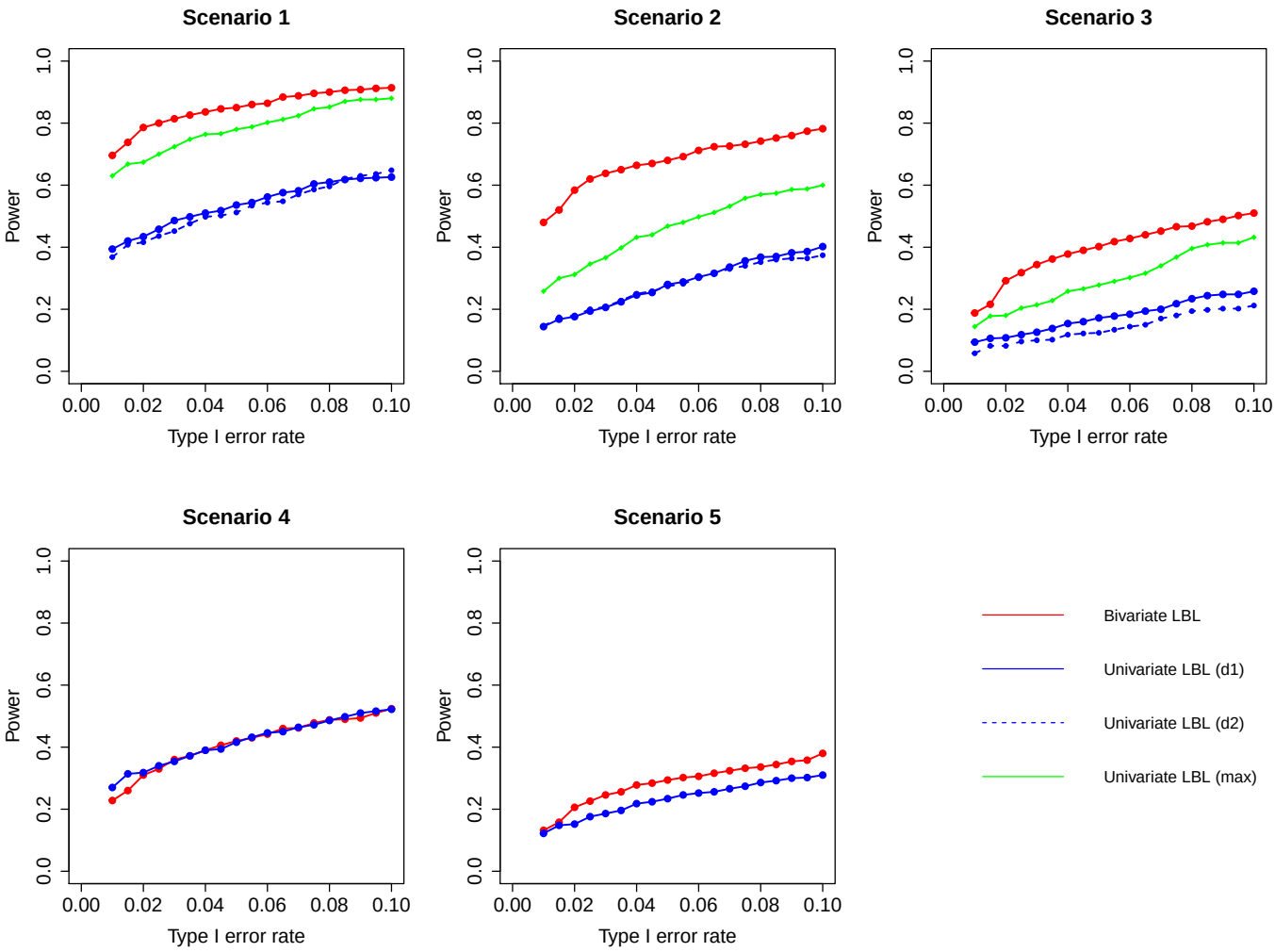

Figure S2: Simulation results for hypothesis 1 under setting 2 (9 haplotypes) and $\rho = 0.3$. The scenarios are listed in Table 1. d1=disease 1, d2=disease 2.

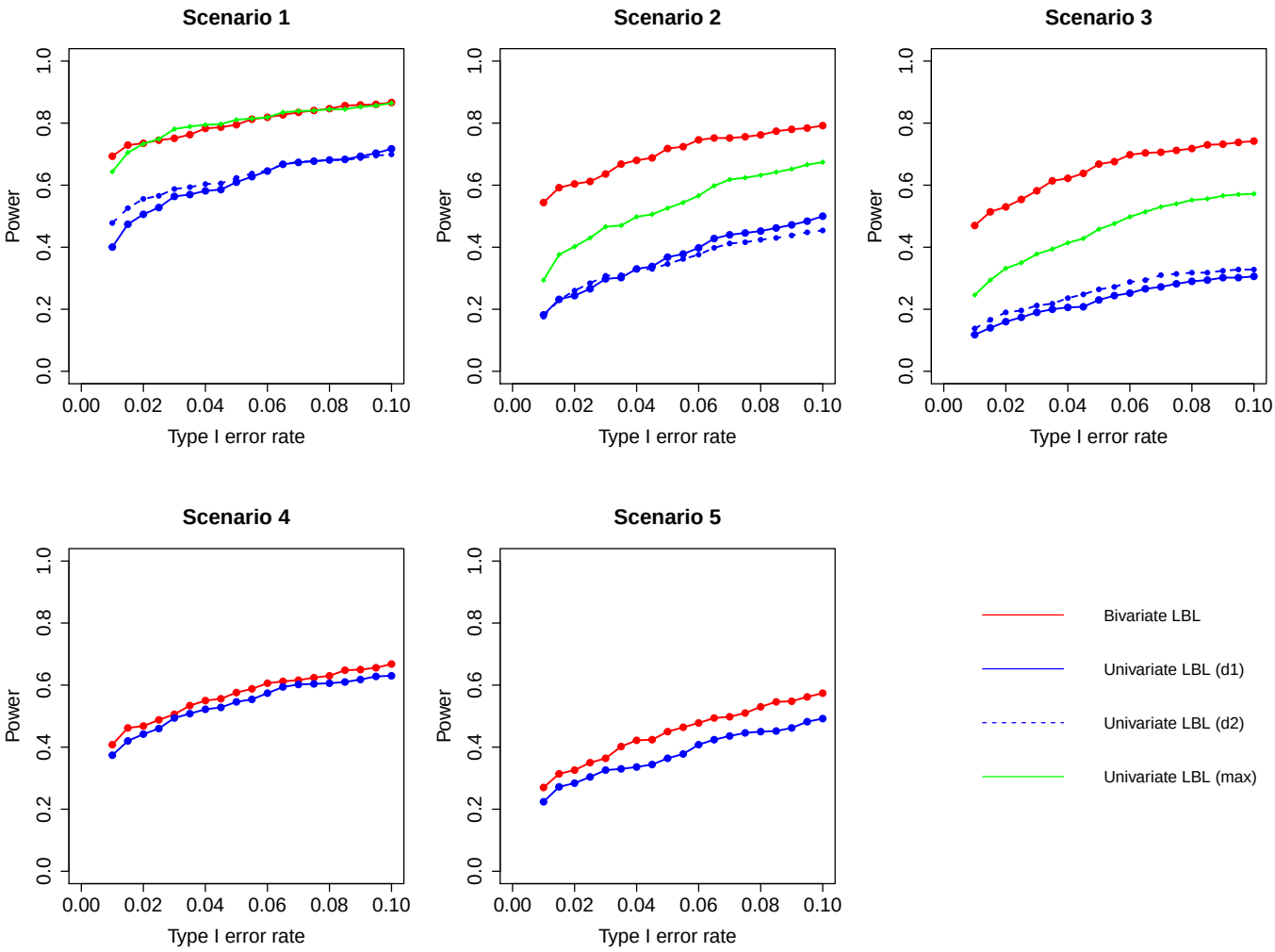

Figure S3: Simulation results for hypothesis 1 under setting 2 (9 haplotypes) and $\rho = 0.7$. The scenarios are listed in Table 1. d1=disease 1, d2=disease 2.

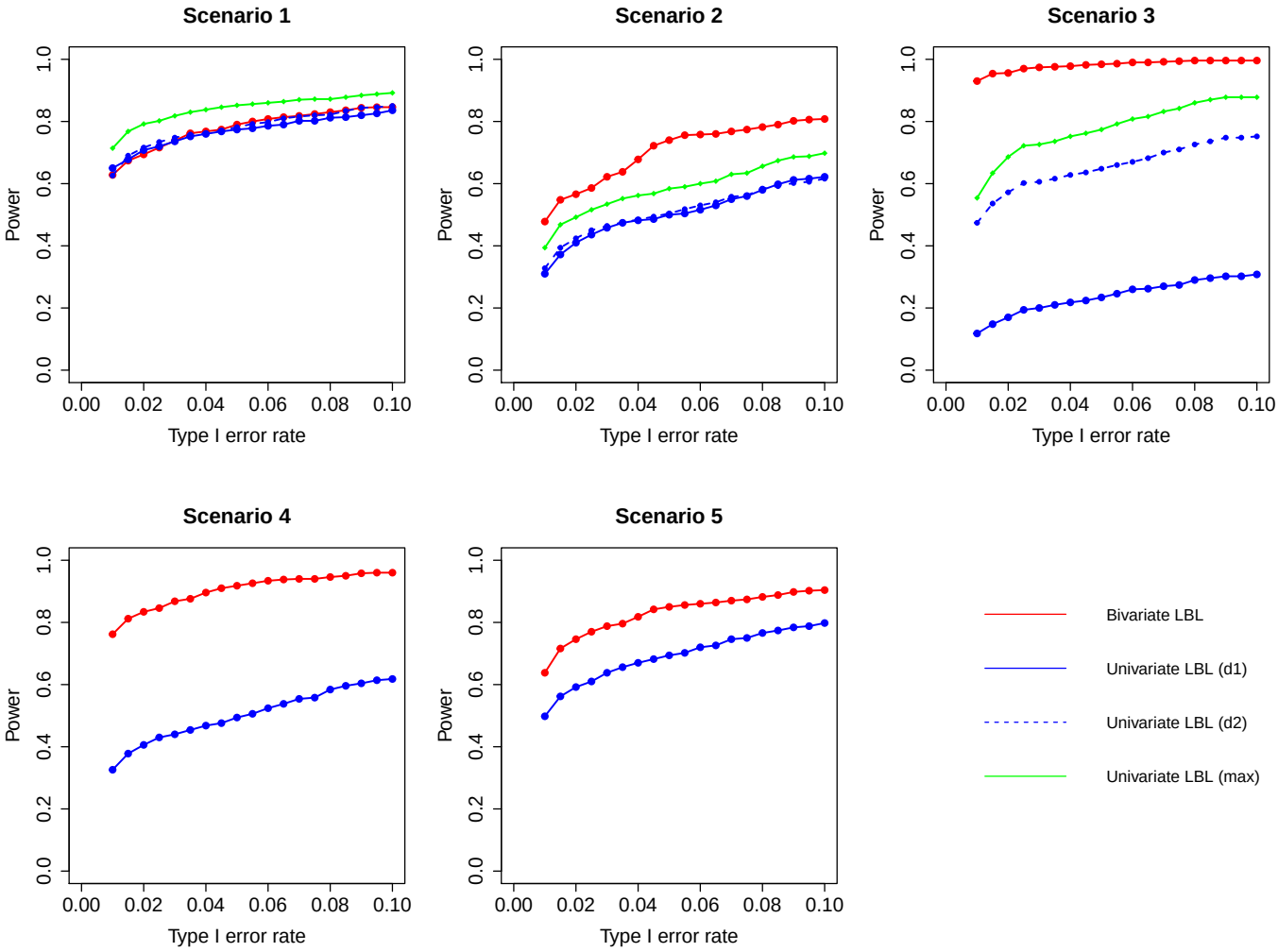

Figure S4: Simulation results for hypothesis 1 under setting 2 (9 haplotypes) and $\rho = 0.99$. The scenarios are listed in Table 1. d1=disease 1, d2=disease 2.

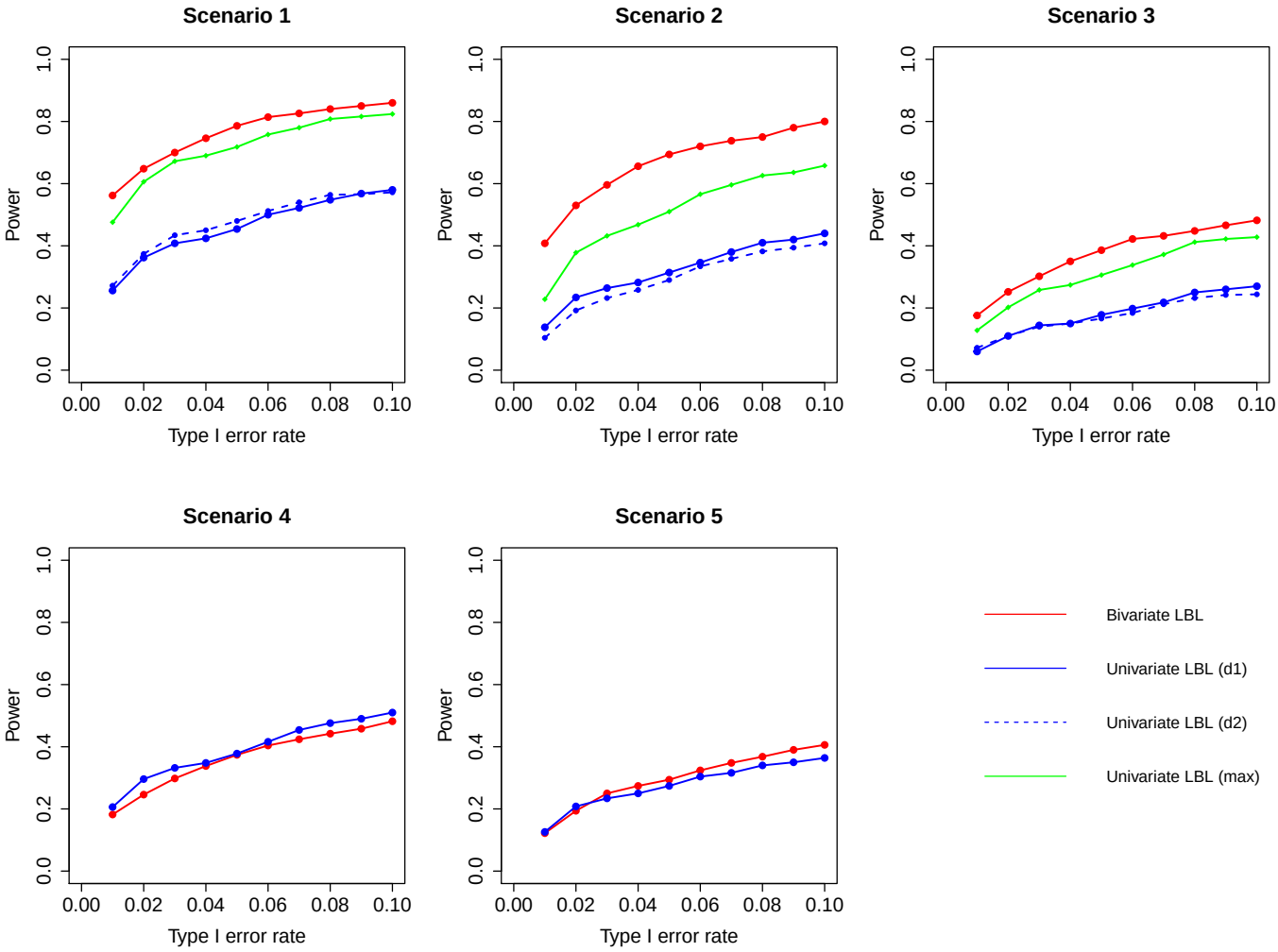

Figure S5: Simulation results for hypothesis 2 under setting 2 (9 haplotypes) and $\rho = 0$. The scenarios are listed in Table 1. d1=disease 1, d2=disease 2.

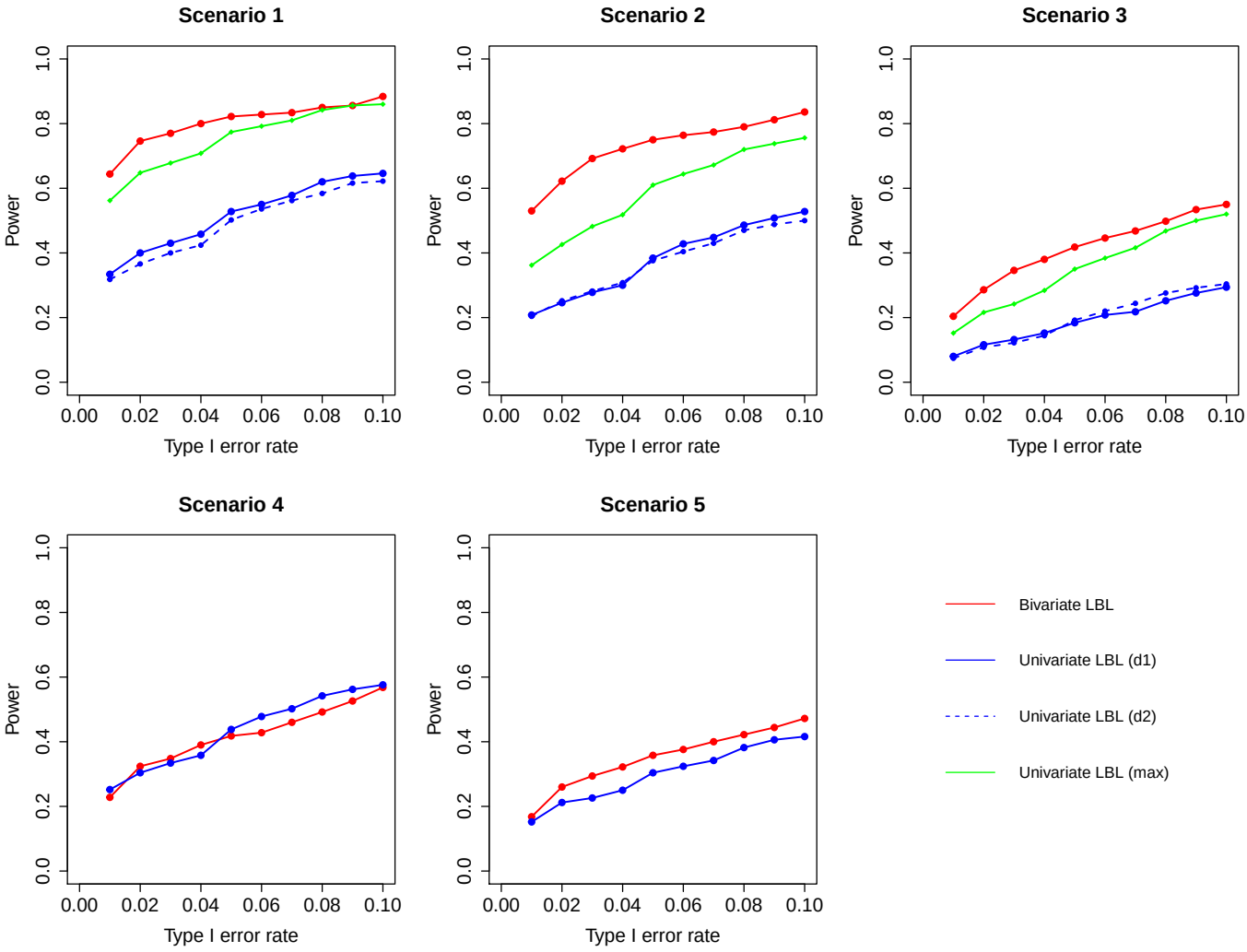

Figure S6: Simulation results for hypothesis 2 under setting 2 (9 haplotypes) and $\rho = 0.3$. The scenarios are listed in Table 1. d1=disease 1, d2=disease 2.

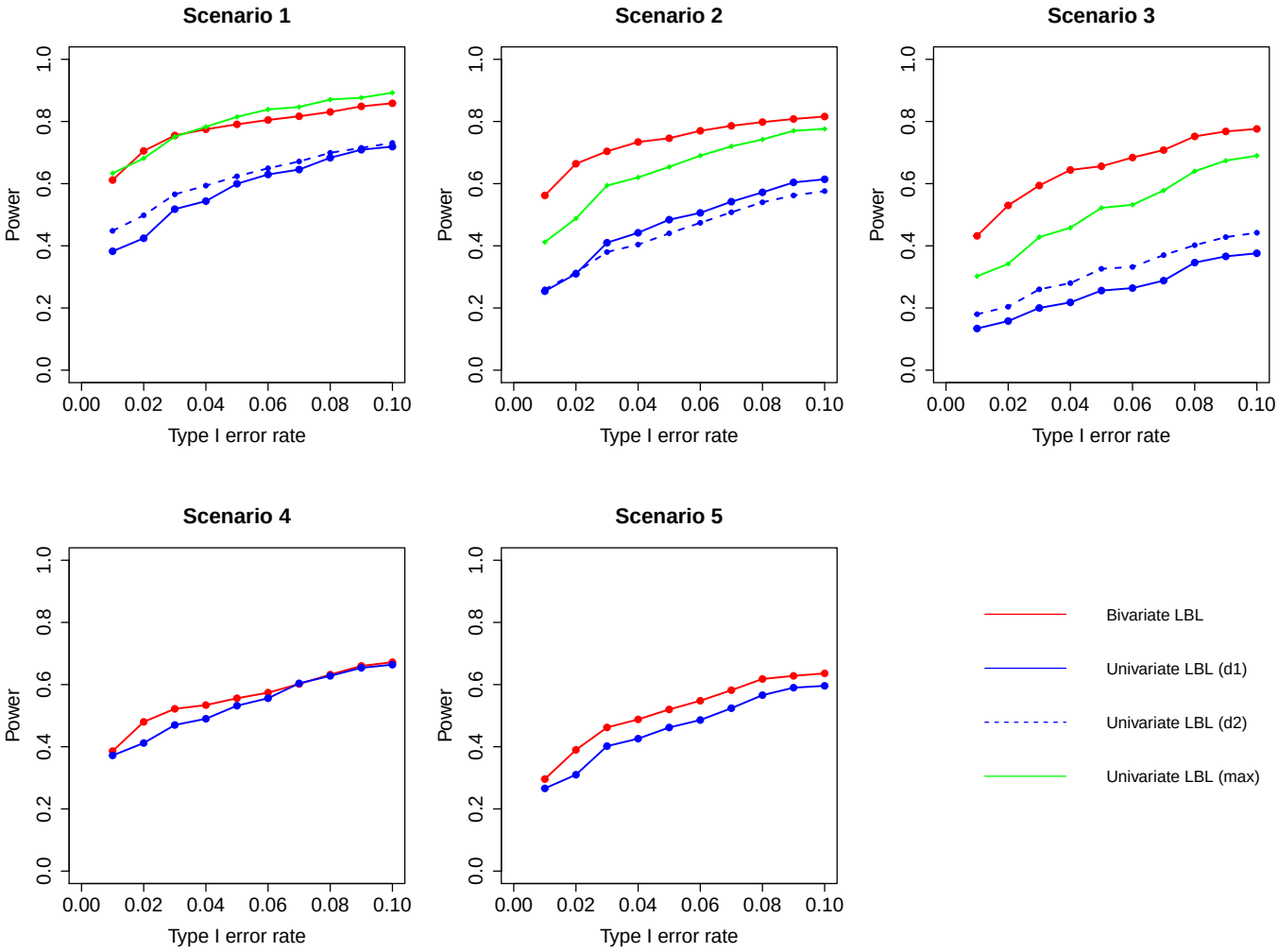

Figure S7: Simulation results for hypothesis 2 under setting 2 (9 haplotypes) and $\rho = 0.7$. The scenarios are listed in Table 1. d1=disease 1, d2=disease 2.

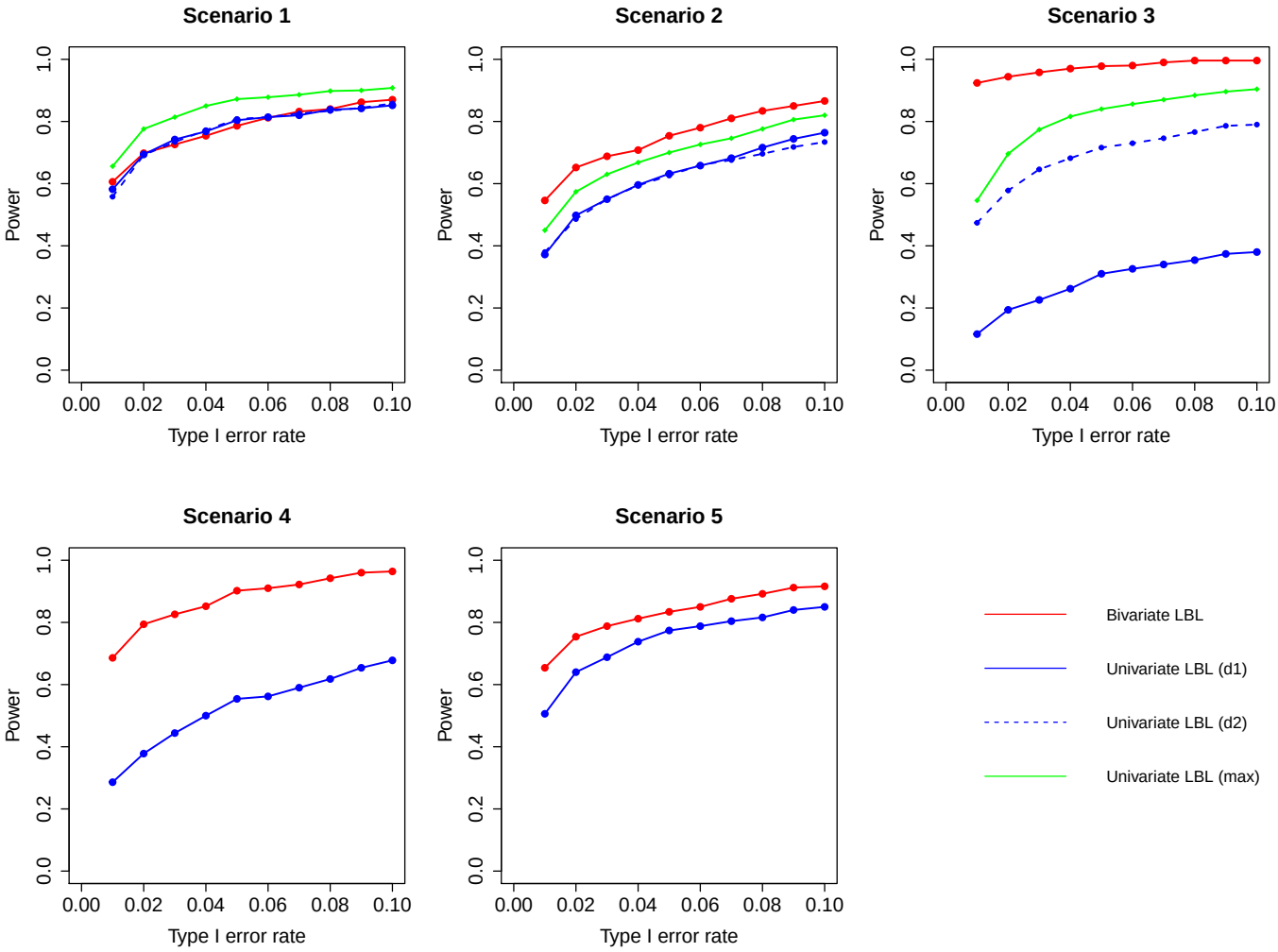

Figure S8: Simulation results for hypothesis 2 under setting 2 (9 haplotypes) and $\rho = 0.99$. The scenarios are listed in Table 1. d1=disease 1, d2=disease 2.

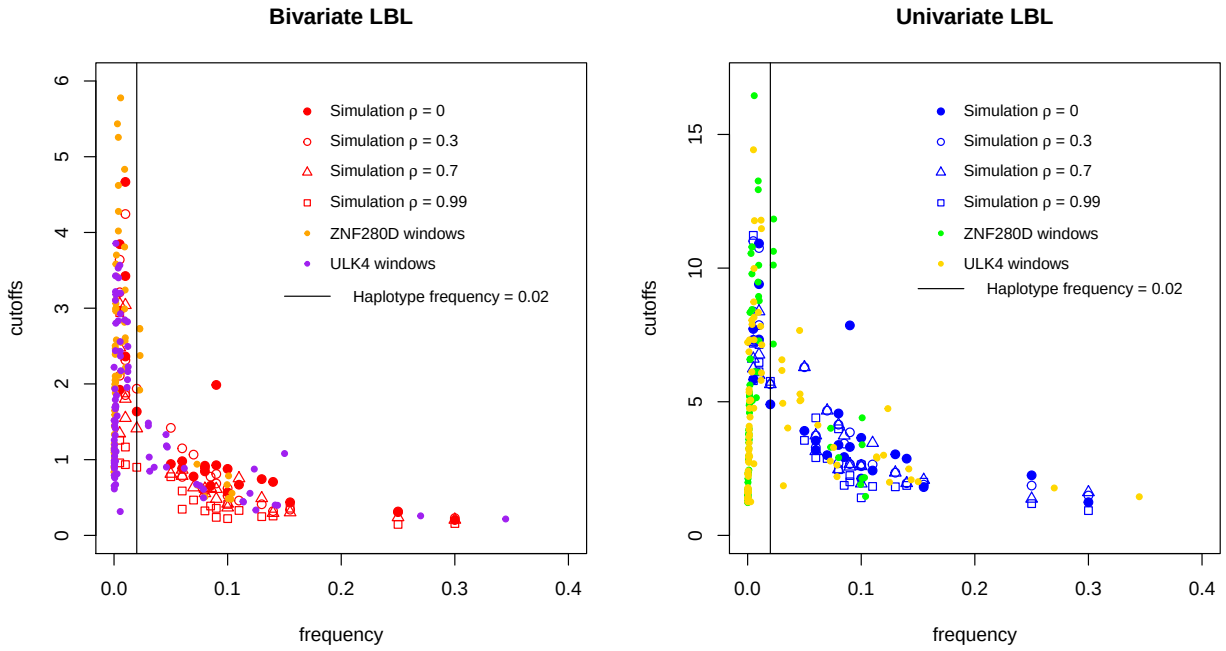

Figure S9: Cutoffs for BF vs. haplotype frequency. Cutoffs corresponding to type I error rate of 0.01 obtained from simulated data and GAW19 data on ULK4 and ZNF280D genes. A haplotype is considered rare if its frequency is less than or equal to 0.02.

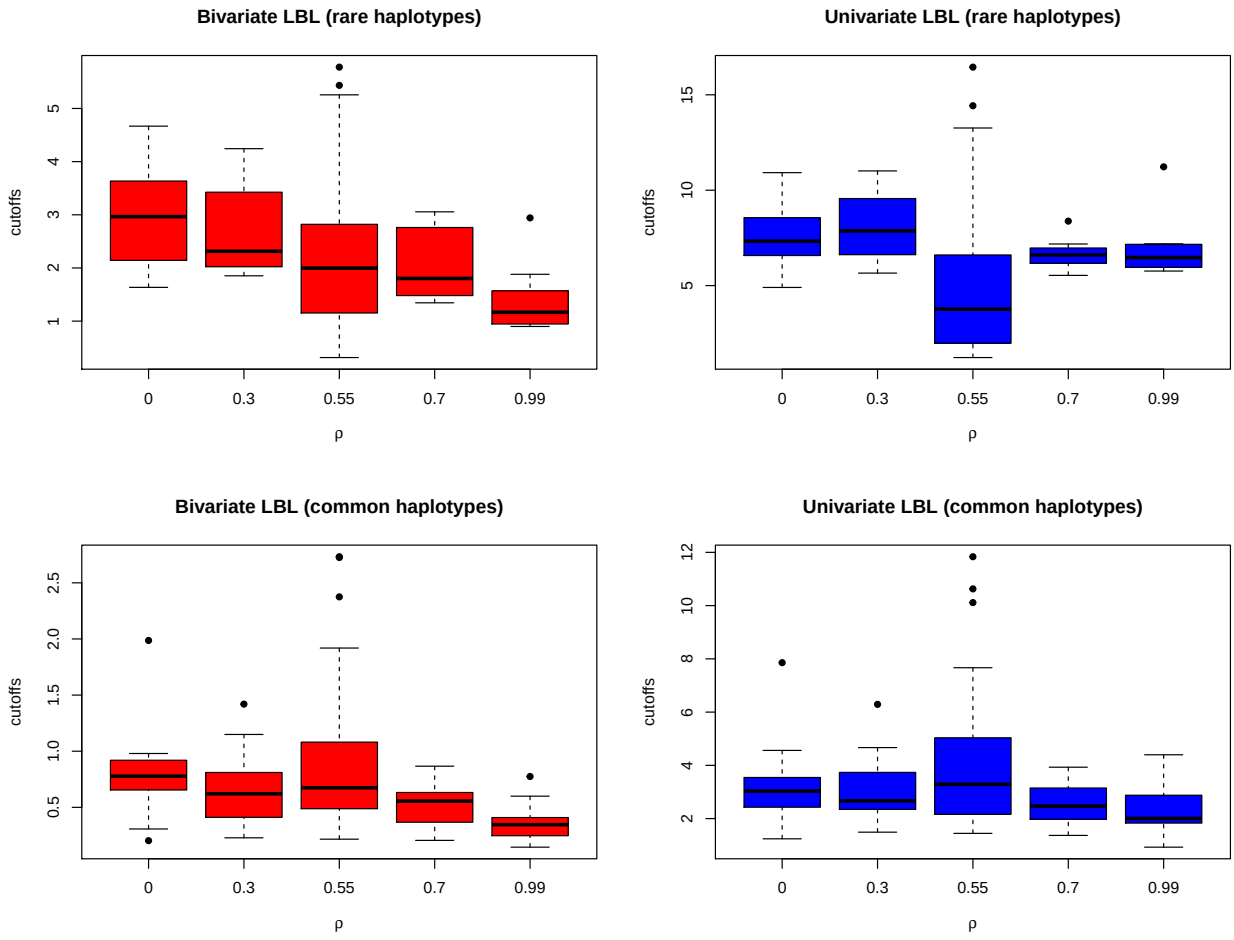

Figure S10: Cutoffs for BF vs. correlation coefficient, ρ. Cutoffs correspond to type I error rate of 0.01 obtained from simulated data and GAW19 data on ULK4 and ZNF280D genes. A haplotype is considered rare if its frequency is less than or equal to 0.02.

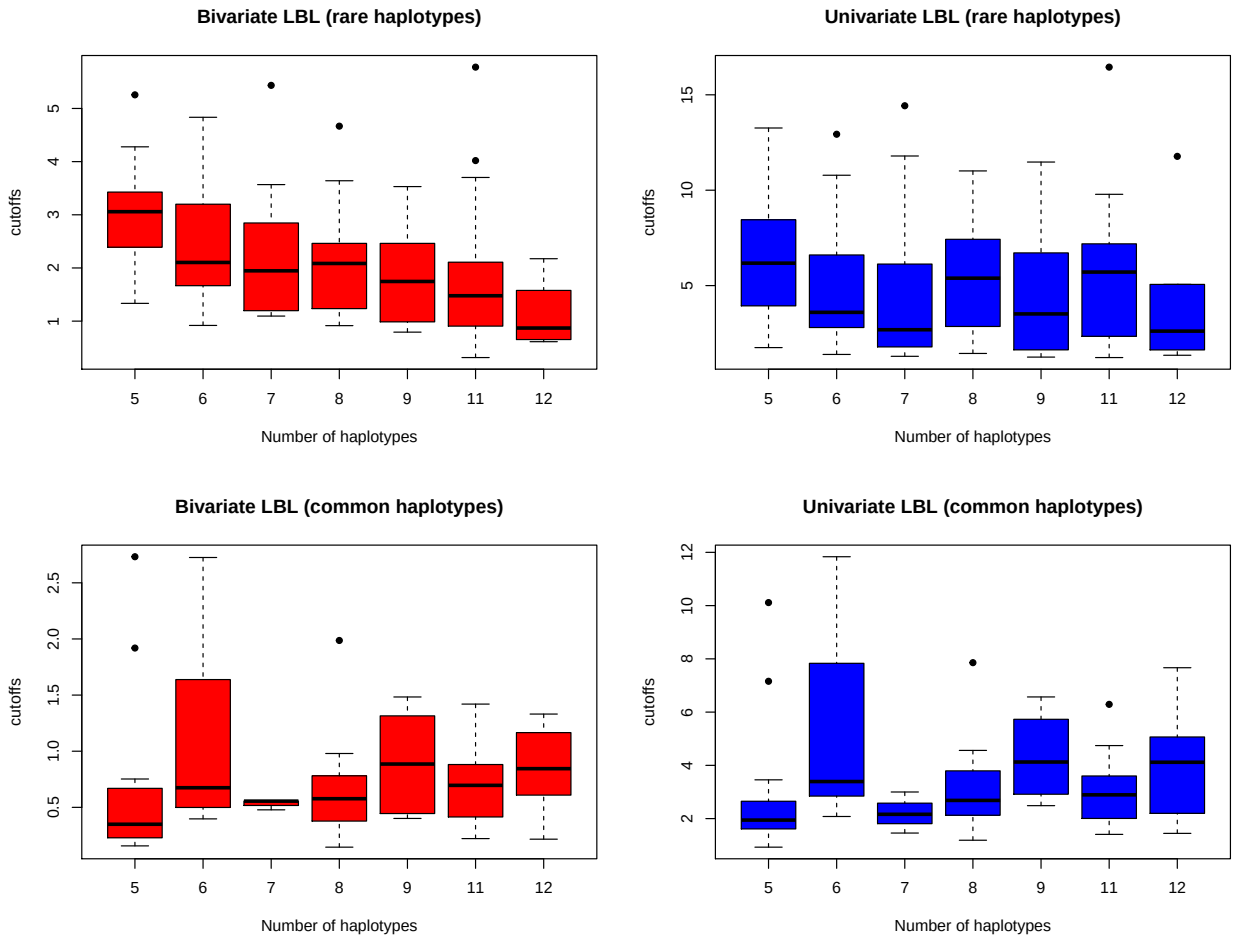

Figure S11: Cutoffs for BF vs. Number of haplotypes in the corresponding haplotype block. Cutoffs correspond to type I error rate of 0.01 obtained from simulated data and GAW19 data on ULK4 and ZNF280D genes. A haplotype is considered rare if its frequency is less than or equal to 0.02.