SDHDP6.Rmd

One of the possible biological explanations for the interaction effect is the linkage disequilibrium. Europeans are known to have stronger LD between markers than Africans, and such difference can lead to an interaction effect between a tag SNP and expression level. Consider an eQTL s_1 that affects the expression y regardless of the population. Let s_2 be in LD with s_1 for Europeans, but not for Africans. Then, the variant s_2 will affect y on European chromosome through s_1 but not on African chromosome. In other words, s_2 will have different effects on y on each population, which is interpreted as an interaction between ancestry s_2 on y.

An example from the real data is illustrated below for gene SDHDP6. The black points are from s_1 rs654818. Among the SNPs that showed no significant interaction effect, rs654818 had the strongest genotype effect. For all local ancestries, it shows a decreasing effect of the minor allele. The red points are from s_2 rs679429 that showed the strongest interaction effect of local ancestry and genotype. When $L = 0$ (European gene), the genotypes from the two SNPs are almost the same, showing high LD, and therefore they both show decreasing effect of minor allele. When $L = 1$, they show less LD, and when $L = 2$ (African gene), the two genotypes show a completely different pattern. Therefore, we can suspect that the strong LD among Europeans lets the strongest eQTL to pass on its effects onto another variant, leading to an interaction effect.

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## [1] "1 with 20 snps"
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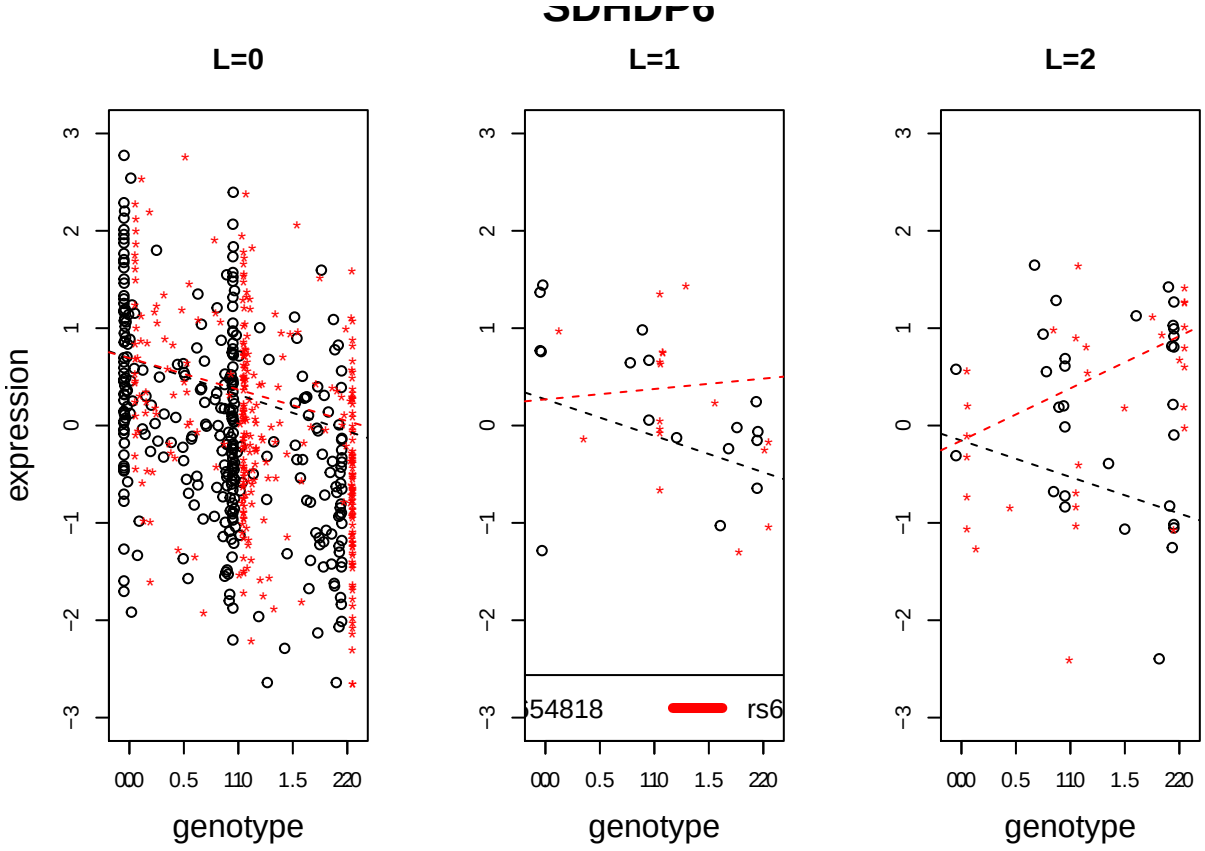

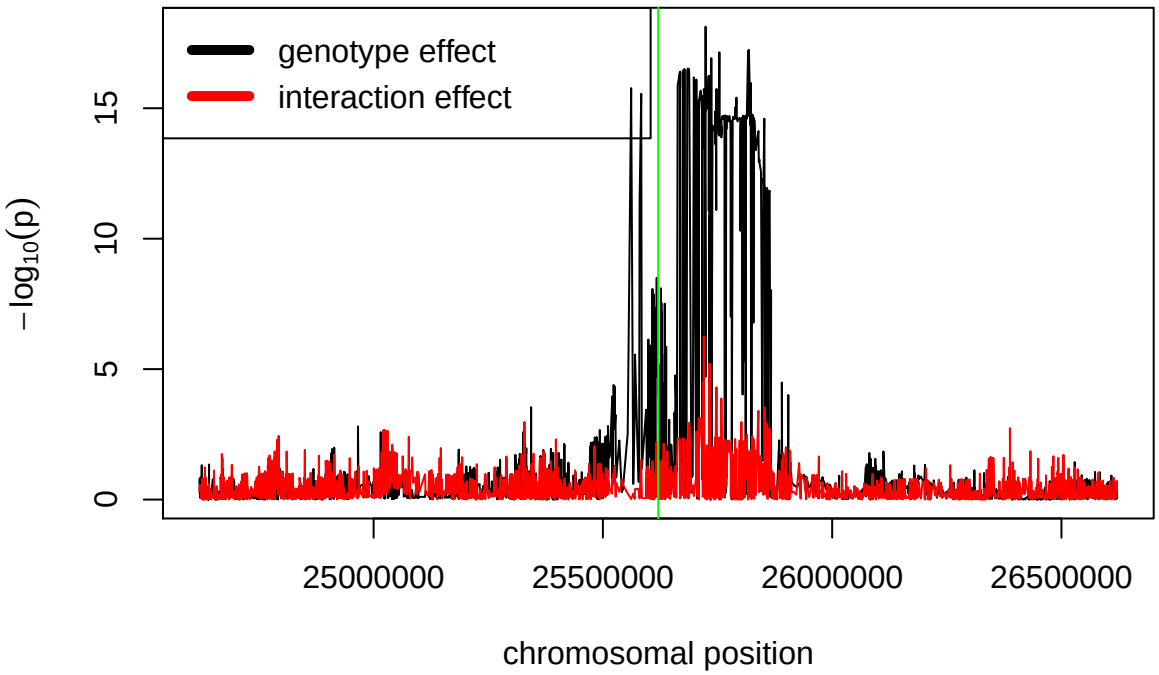
genotype effect
interaction effect
$-\log_{10}(p)$
0
5
10
15
25000000
25500000
26000000
26500000
chromosomal position