

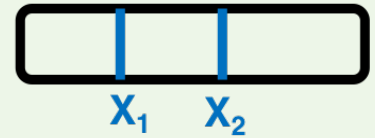
❖ Simulation of phenotypic values (**100** iterations)

$$\mathbf{y} = \mathbf{X}_1\beta_1 + \mathbf{X}_2\beta_2 + \mathbf{X}_3\beta_3 + \mathbf{Z}\mathbf{u} + \mathbf{e}, (\text{Eq. 1})$$

\mathbf{y} : phenotypic values $\mathbf{X}_1\beta_1, \mathbf{X}_2\beta_2, \mathbf{X}_3\beta_3$: fixed effects (marker effects)
 $\mathbf{Z}\mathbf{u}$: random effects (polygene effects) \mathbf{e} : residuals

• \mathbf{X}_1 and \mathbf{X}_2 belong to the same haplotype block.

one haplotype block



❖ We assumed 2 scenarios.

① **Coupling** : $\text{cor}(\mathbf{X}_1, \mathbf{X}_2) > 0 \rightarrow \beta_1 = \beta_2$, $\text{cor}(\mathbf{X}_1, \mathbf{X}_2) < 0 \rightarrow \beta_1 = -\beta_2$

② **Repulsion** : $\text{cor}(\mathbf{X}_1, \mathbf{X}_2) > 0 \rightarrow \beta_1 = -\beta_2$, $\text{cor}(\mathbf{X}_1, \mathbf{X}_2) < 0 \rightarrow \beta_1 = \beta_2$



❖ Perform GWAS with 4 methods.

① Our proposed method, **RAINBOW** (R package RAINBOWR)

② The **single-SNP** method (R package rrBLUP)

③ **Haplotype-based** method introduced by Yano *et al.*, 2016

④ **SNP-set** method, SKAT (R package SKAT)



❖ Evaluate the results with the following summary statistics

① $-\log_{10}(p)$ and $-\log_{10}(p_\alpha)$, correspond to the **detection power**

② **Recall, Precision, and F-measure**

③ **AUC** (area under the curve) for **regions around causals**