

Influence of genomic architecture on the performance of association mapping models
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Joseph Powell

Background

- Association mapping
- Genomic architecture
 - Causal variants and markers
 - LD structure
 - Allele frequencies
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- Diverse range of models
 - Parameterization
 - Use of marker information

Background

Does genomic architecture affect the performance of association models?

How well do models perform under different sets of genomic architecture?

Objectives

Main Objective

Assess performance of association models across a spectrum of genomic architecture

↓

Identify situations that lead to the optimal performance of a model

Use observed information for model choice

Models

Regression-based $\rightarrow Y = 1'_n \mu + Xg + e$

Association models

- Single Marker
- Multiple markers
 - Main-order
 - Haplotypes (spectrum of parameterization*)

*Suppose we fit 3 markers (a,b,c)
 Main effects = a + b + c
 Haplotype = a + b + c
 + a:b + a:c + b:c
 + a:b:c + phase

Models

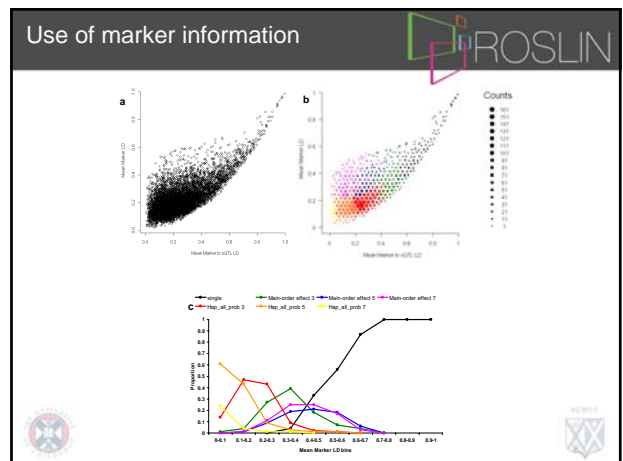
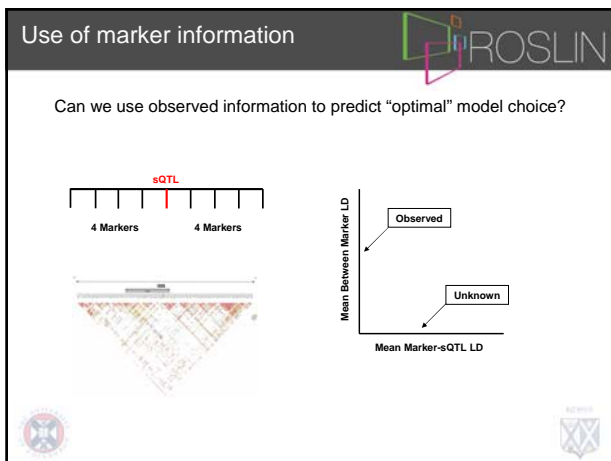
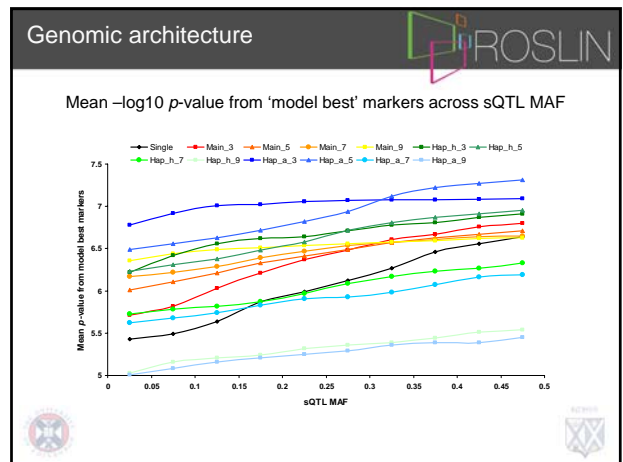
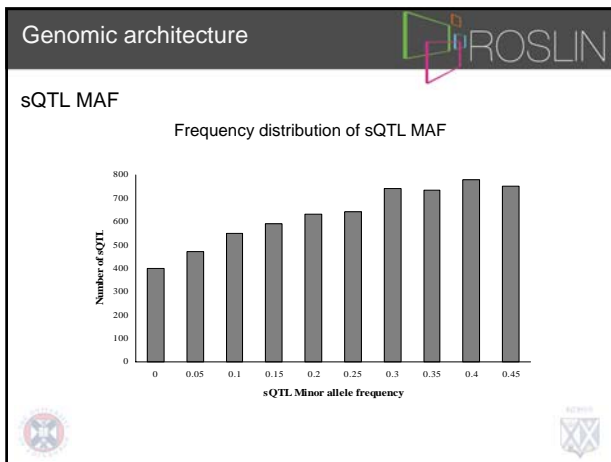
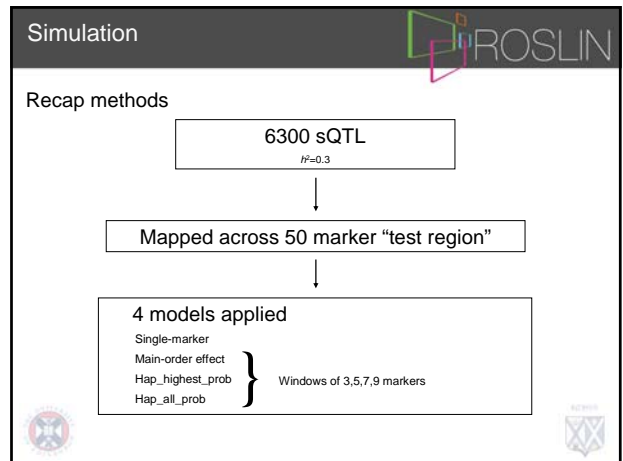
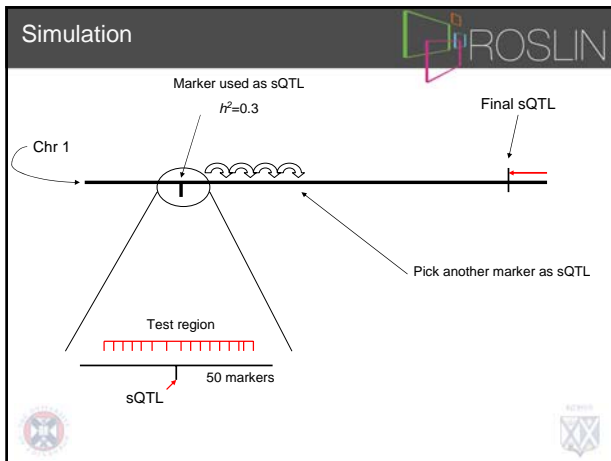
Difference between models is parameterization of marker information

$$X = n^*(1 \dots H)$$


1	1 1 2	0 0.5 0.5 0 0	0 0.12 0.12 0.26 0 0 0.5
0	0 2 2	0 0.5 0 0.5 0	0.16 0 0.84 0 0 0 0
0	0 2 1	0.5 0 0 0.5 0.5	0.12 0.12 0.12 0.12 0.52 0 0
2	2 1 1	0 0 0 0 1	0 0 0.2 0 0 0.6 0.2
2	2 0 0	0.5 0 0.5 0 0	0.14 0.14 0.17 0.17 0.1 0.1 0.18
1	1 0 1	0 0 1 0 0	0 0 1 0 0 0 0

Single Main-order Hap_highest_prob Hap_all_prob



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Main conclusions 

- Model performance affected by genomic architecture
- Optimal performance, relative to other models depends on localised conditions
- Observed information can be used to drive model choice



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