Properties of Runs of Homozygosity as a measure of Identity by Descent

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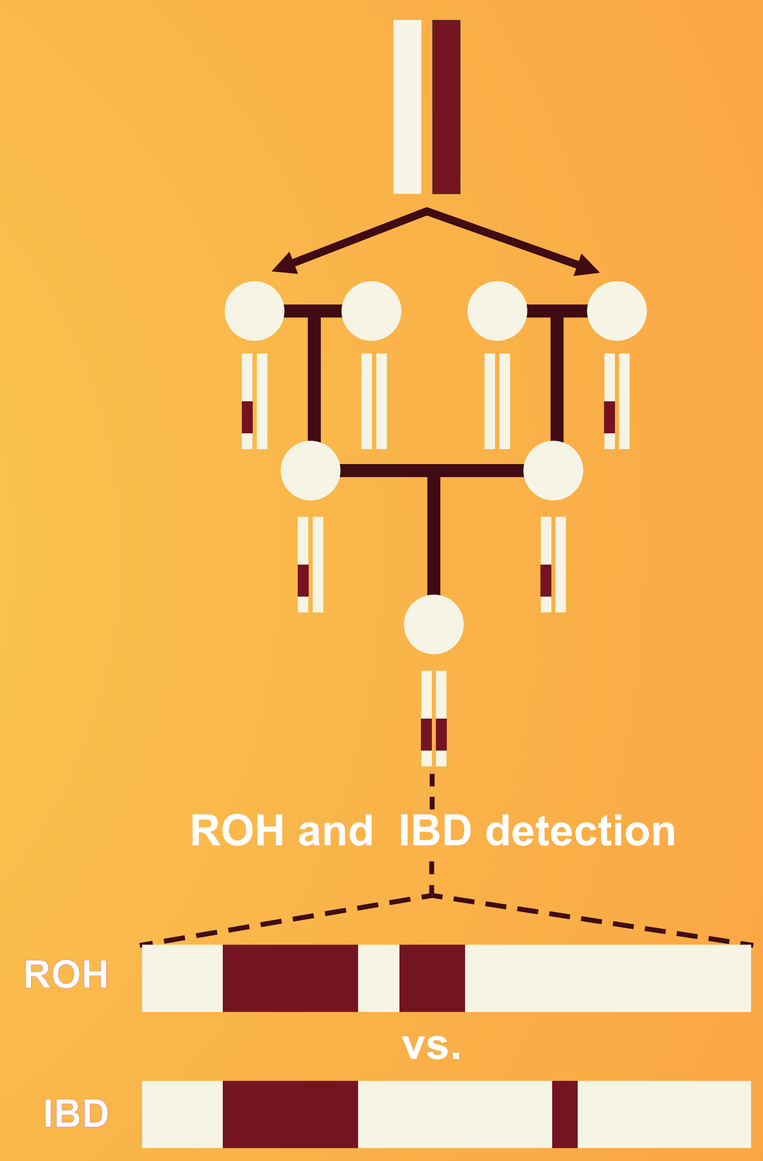
Introduction

Runs of Homozygosity (ROH) are commonly used to estimate Identity-by-descent (IBD). However, the method's accuracy at the segment level in livestock populations has only been evaluated in a few studies.

Thus, the aim of this study was to determine to what extent ROH are truly IBD and estimate the proportion of IBD segments that go undetected in a simulated livestock population



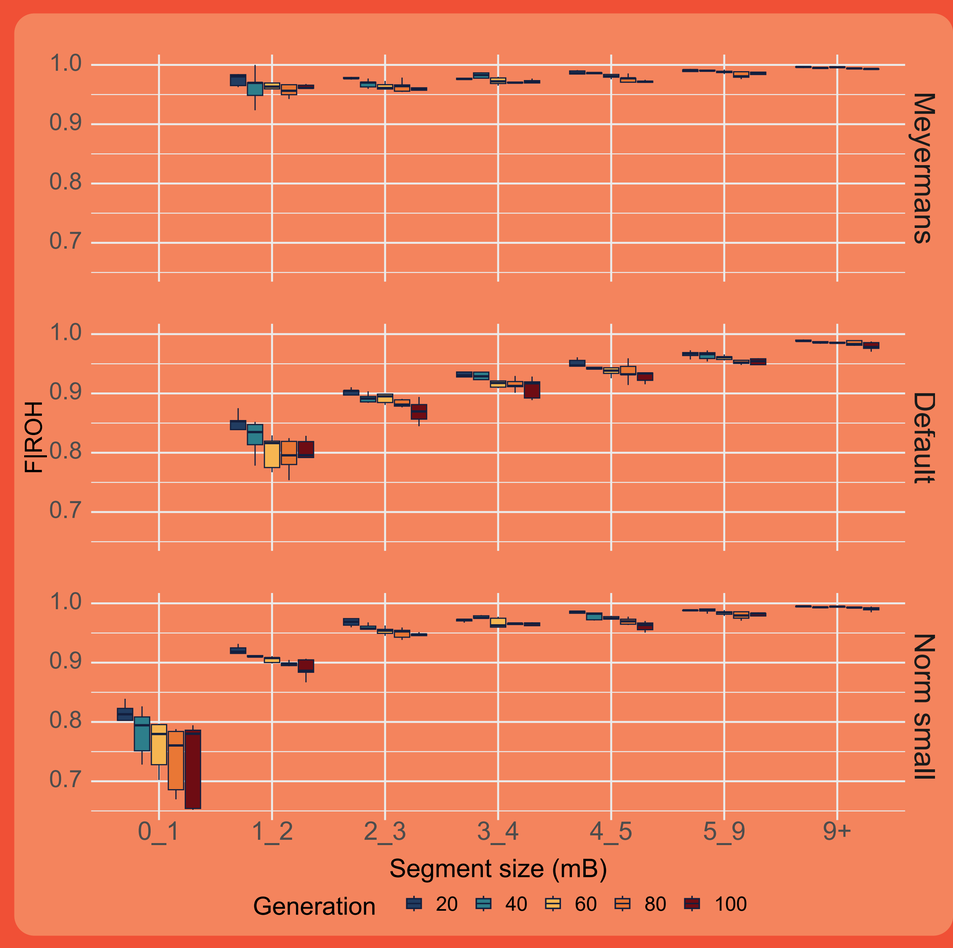
Method

Using ADAM we simulated a population with one 100cM chromosome for 100 generations. One scenario had NE = 100, and one had NE = 200. The genome had 10k SNPs and 10k founder markers used to track IBD .

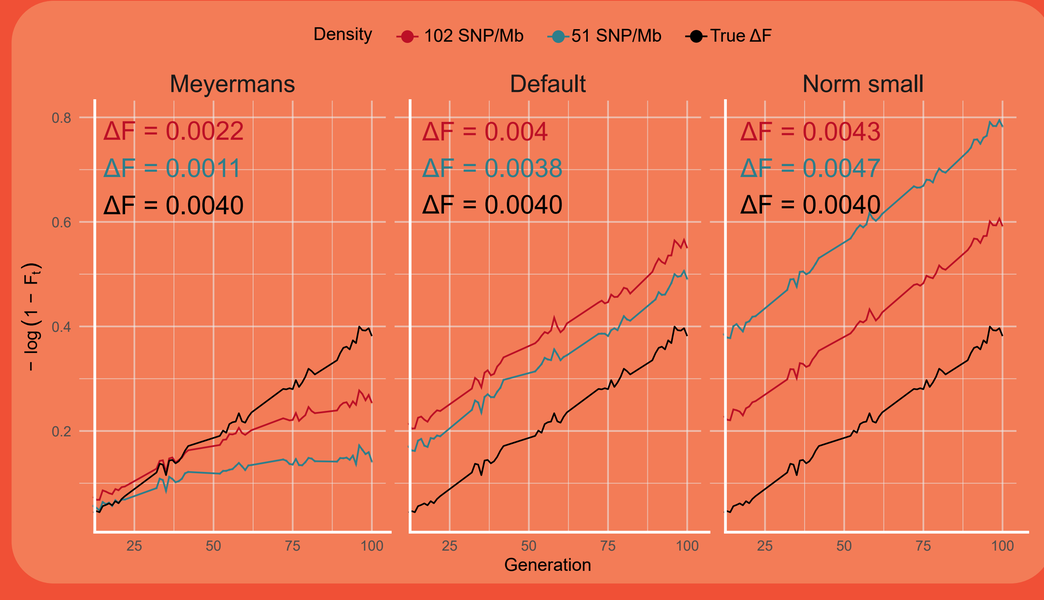
To detect ROH we used PLINK with different parameter settings such as the default, recommendations from Meyermans et al. (2020), and settings with a low minimum length threshold (Norm small).

The analysis consisted of the true positive rate, IBD sensitivity, inbreeding within ROH, correlation between true and estimated inbreeding, and comparing true and estimated rate of inbreeding.

Inbreeding within ROH



Rate of inbreeding



Conclusions

- For a 150k and 300k SNP chip ROH is a reliable indicator for IBD when ROHs are longer than 1 Mb.
- The accurate detection of long segments yields high correlations between estimated and true inbreeding. The high correlations implies that individual differences in inbreeding are accurately predicted by F_{ROH}.
- Furthermore, the regression coefficients being close to one signify that a 1% variation in F_{ROH} correspond to a 1% variation in F_{IBD}.
- Detecting shorter ROHs is essential for correctly estimating the rate of inbreeding in a population.

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