Recessive deleterious variation has a limited impact on signals of adaptive introgression in human populations

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Introduction

- A higher frequency of archaic variants and longer introgressed tracts are the typical signatures of adaptive introgression (AI)
- If two populations carry recessive deleterious variants private to them, a heterosis effect occurs upon admixture that reduces the deleterious effects (Fig. 1)
- Heterosis effect can increase the introgressed ancestry, while the extent it affects the summary statistics used in practice to detect adaptive introgression is unknown
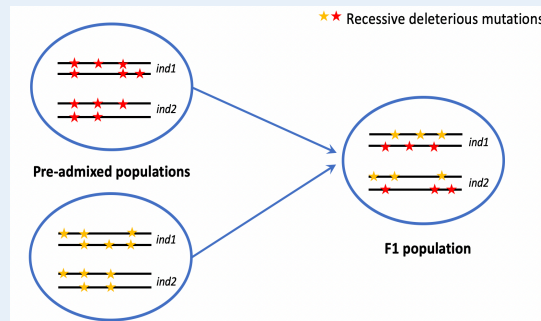

Fig. 1: Heterosis effect from an increase in heterozygosity due to admixture

Results

- The presence of recessive, deleterious variants can substantially increase the mean and variance of AI summary statistics upon admixture, similar to the effect of AI with beneficial mutation
- This leads to a higher false positive rates (FPR) for all AI summary statistics examined in this work
- In humans, most of the previously identified to be AI candidate are less prone to false detection, with the exception of two gene regions (*HYAL2* and *HLA*, Fig 2)
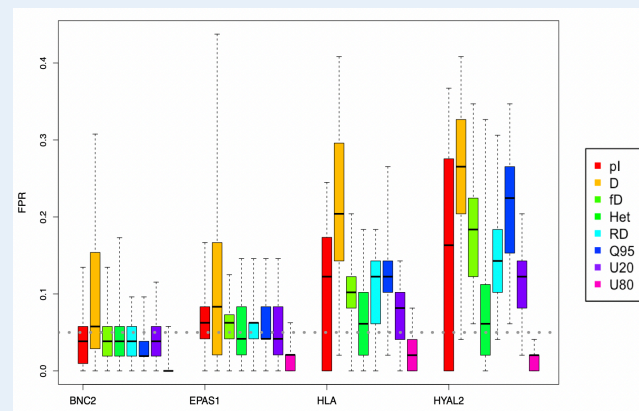

Fig. 2: FPRs for summary statistics from human AI candidate regions

- High false positive in the outlier regions are mainly contributed by simultaneous high exon density and low recombination rate (Fig. 3)
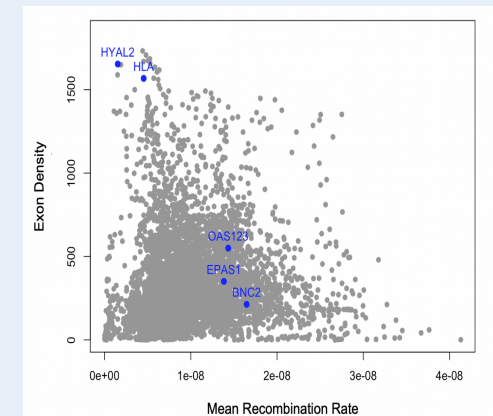

Fig. 3 Human exon density and recombination rate in 5MB windows

- Null model incorporating realistic distribution of fitness effect is recommended for future AI studies, especially for organisms with compact genome or complex demography

Manuscript and Data Availability

https://www.biorxiv.org/content/10.1101/2020.01.13.905174v1