Association Testing with X Chromosome Data: Adding to Our Pipeline

Caitlin McHugh, with Tim Thornton
Adrienne Stilp
Matt Conomos
Cathy Laurie

Department of Biostatistics
University of Washington

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Outline

Update on adding X chromosome effects to the pipeline

- Should we adjust for random effects on the X + X chromosome PCs 1-2 when testing an autosomal SNP?
- Only if X effects are confounders.

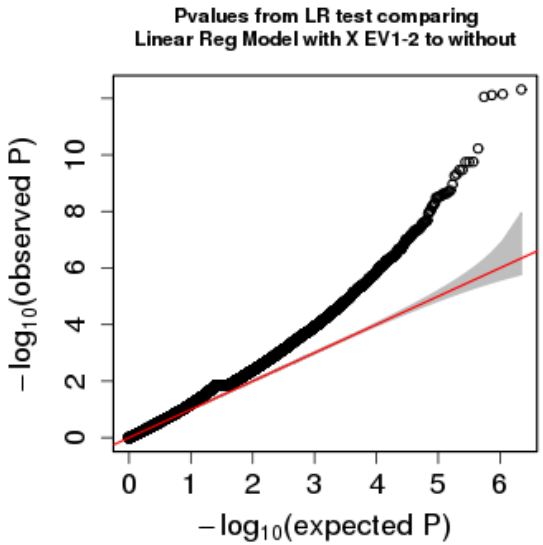
Are X chromosome effects confounders?

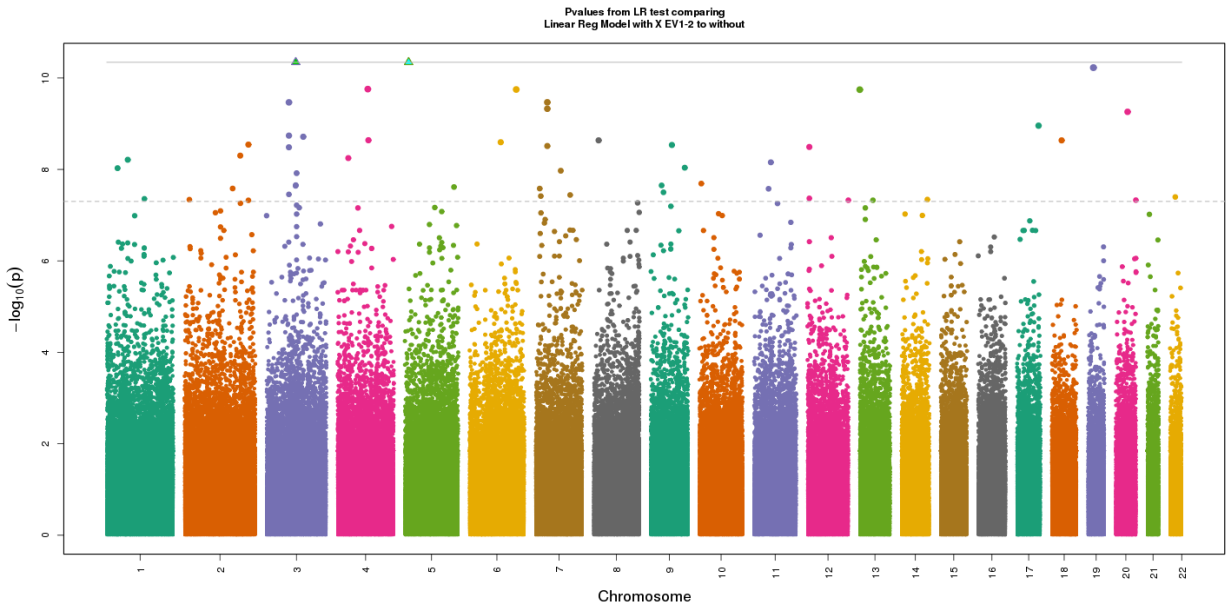
- In a set of unrelated samples, regress

$$\text{auto genotype} \sim \text{auto PC } 1-5$$
$$\text{auto genotype} \sim \text{auto PC } 1-5 + \text{X chr PC } 1-2$$

- Perform the likelihood ratio test (LRT) comparing these models.
- Examine QQ and Manhattan plots.

Results when testing auto genotypes





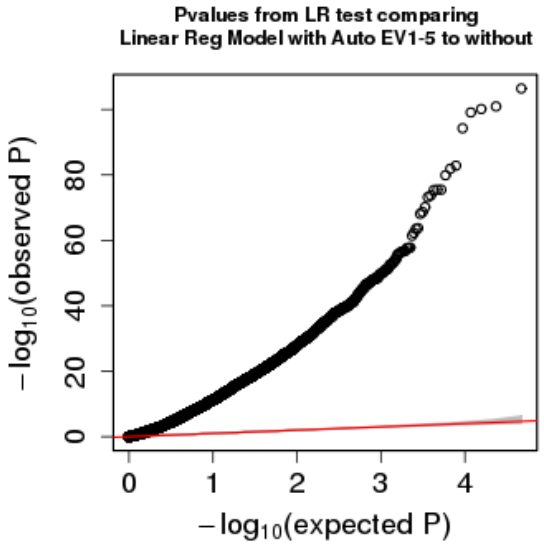
Are X chromosome effects confounders?

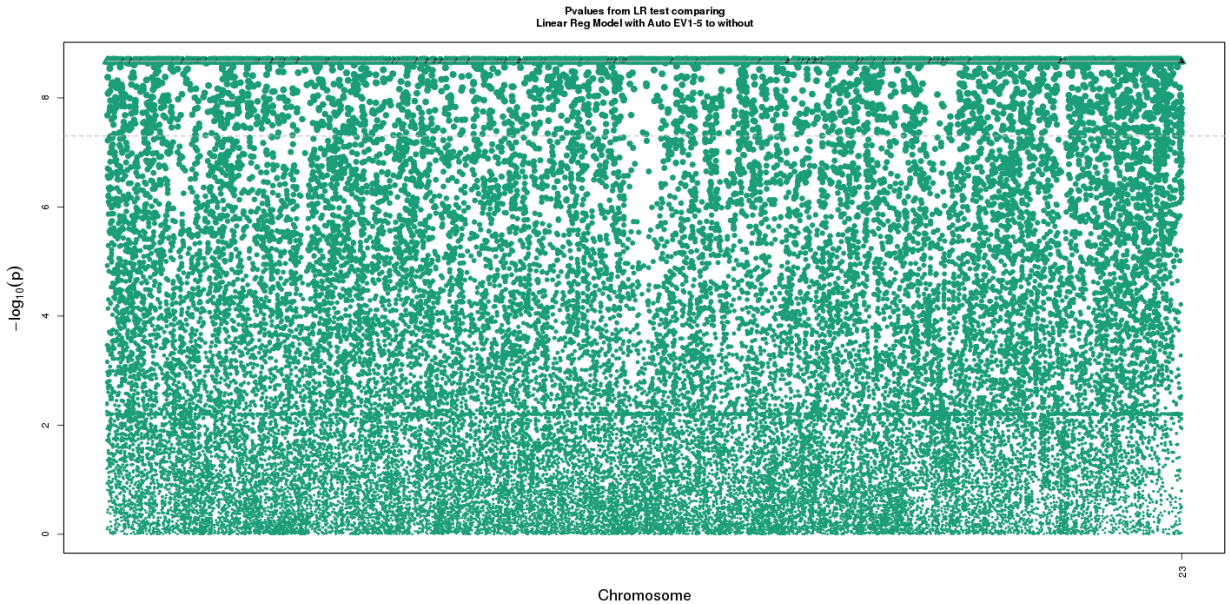
- In a set of unrelated samples, regress

$$\text{X genotype} \sim \text{X chr PC } 1-2$$
$$\text{X genotype} \sim \text{X chr PC } 1-2 + \text{auto PC } 1-5$$

- Perform LRT comparing these models.
- Examine QQ and Manhattan plots.

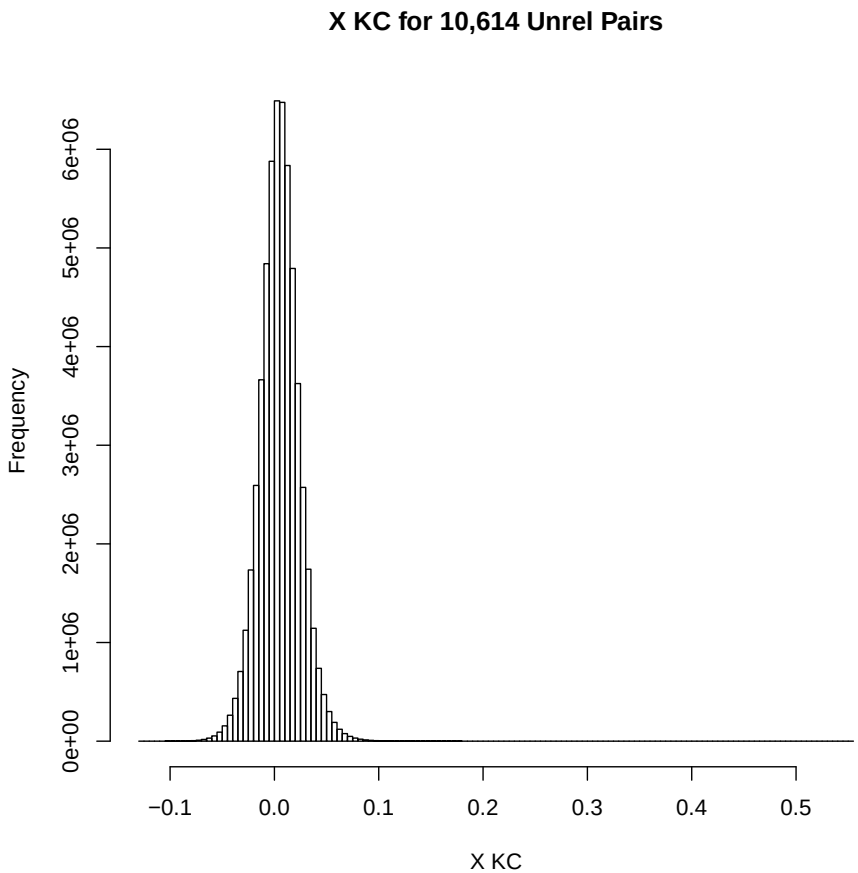
Results when testing X chromosome genotypes

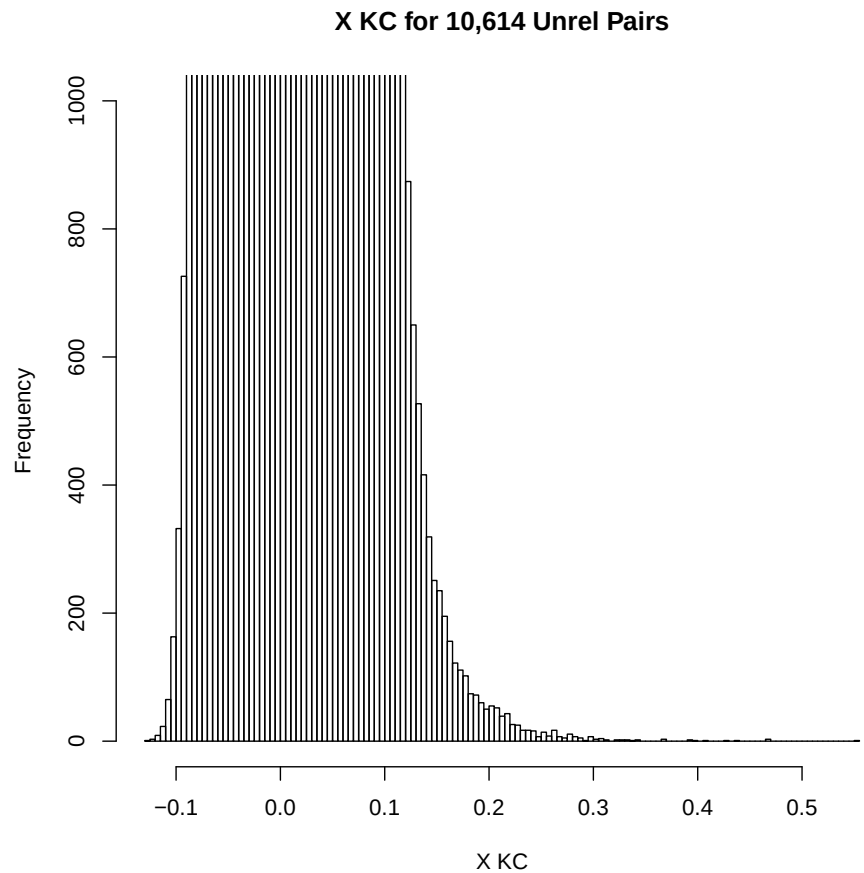




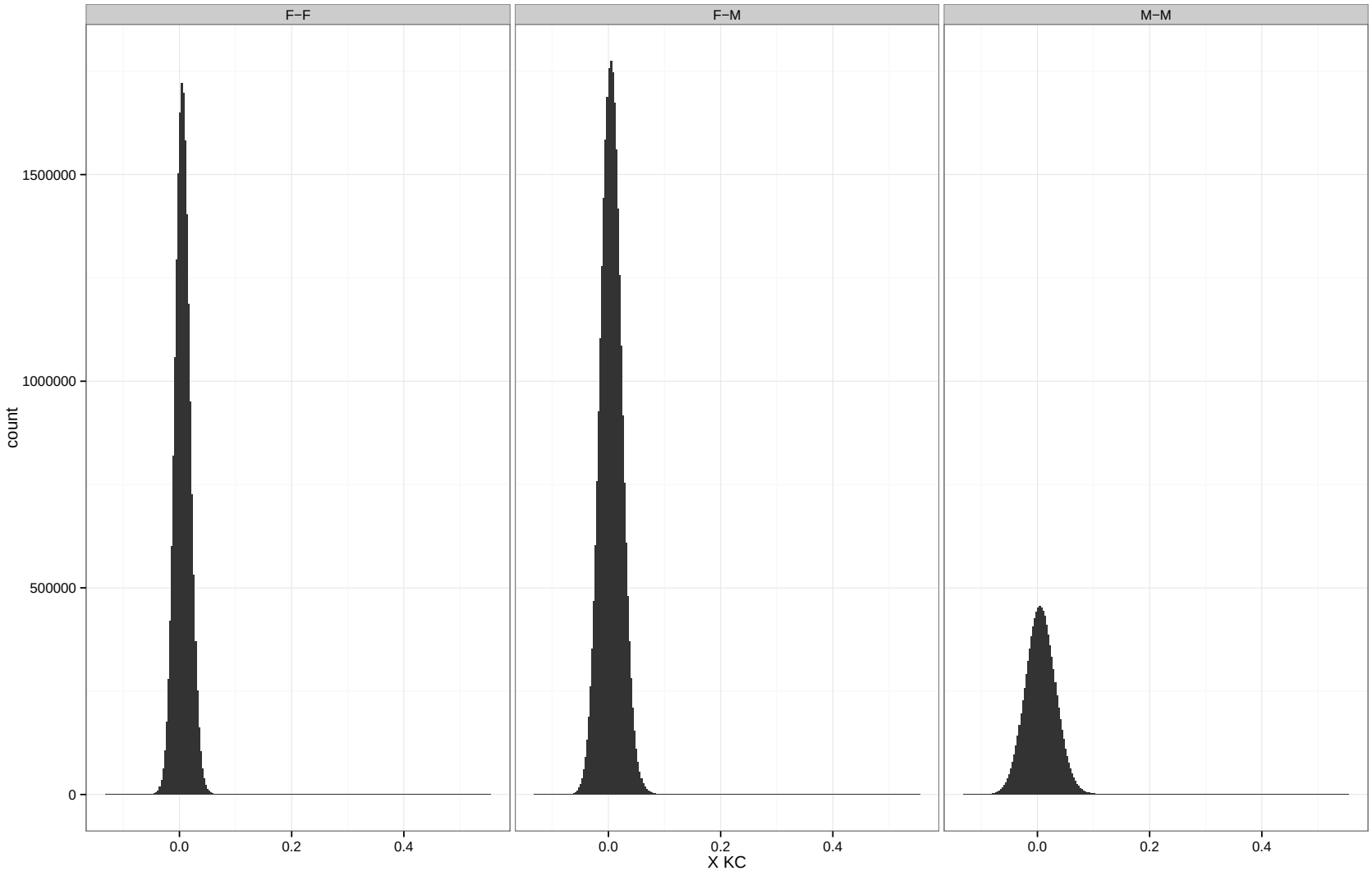
Recall: on the X

X chromosome KC for autosomally unrelated samples.

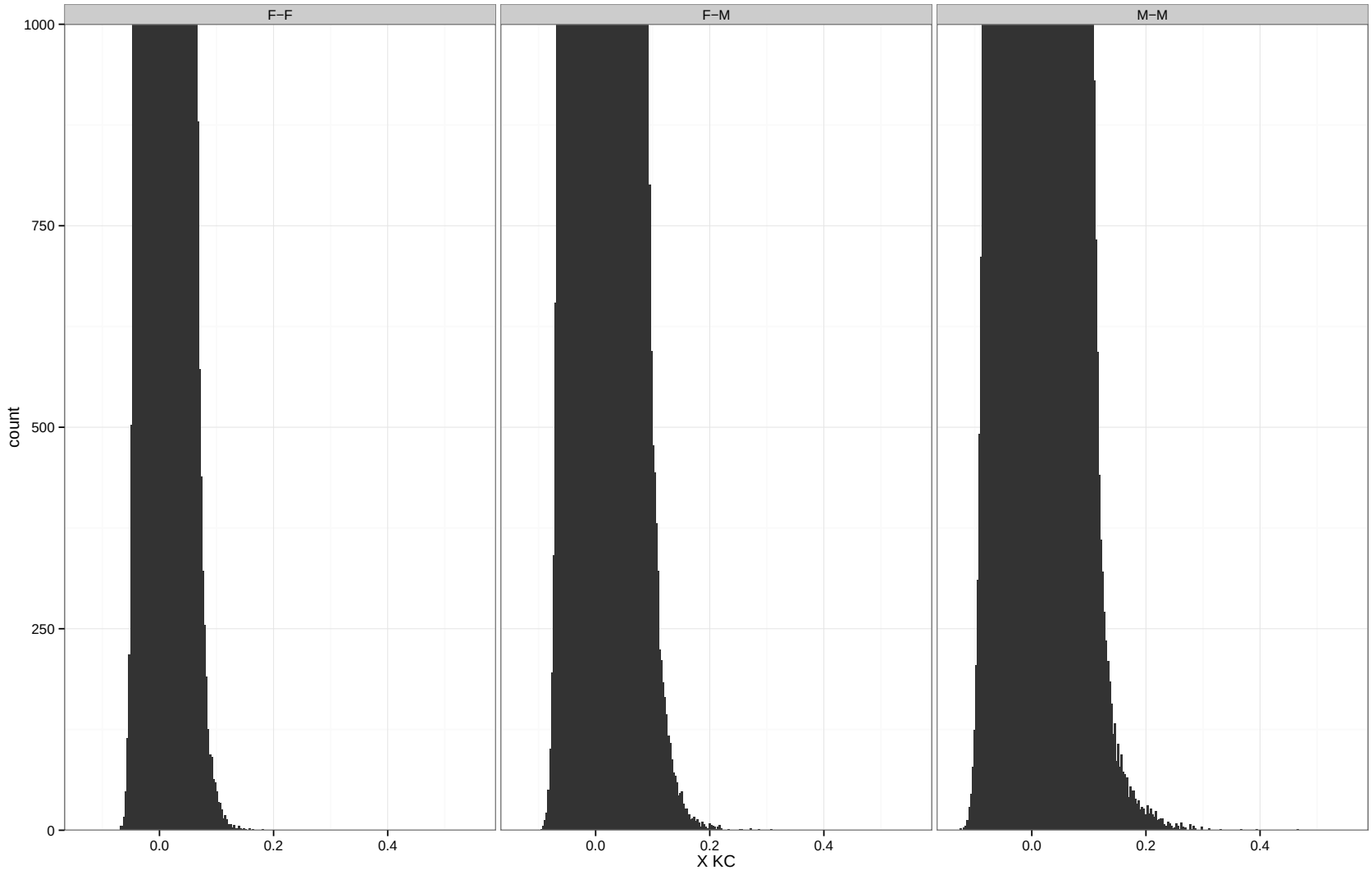




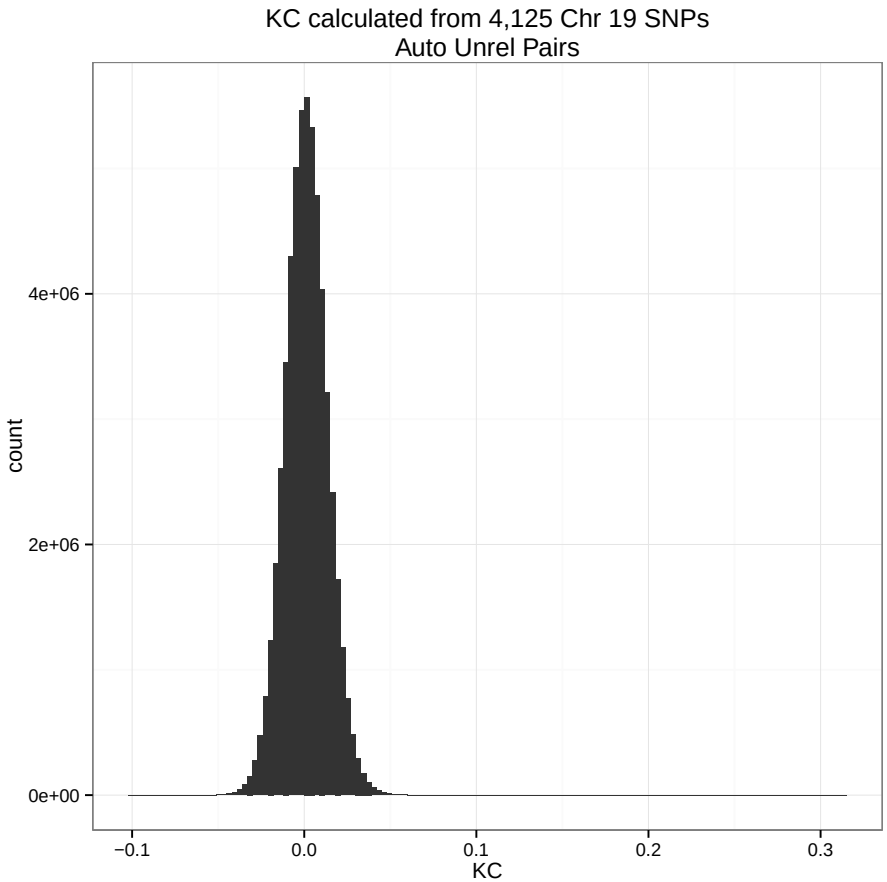
X KC by sex

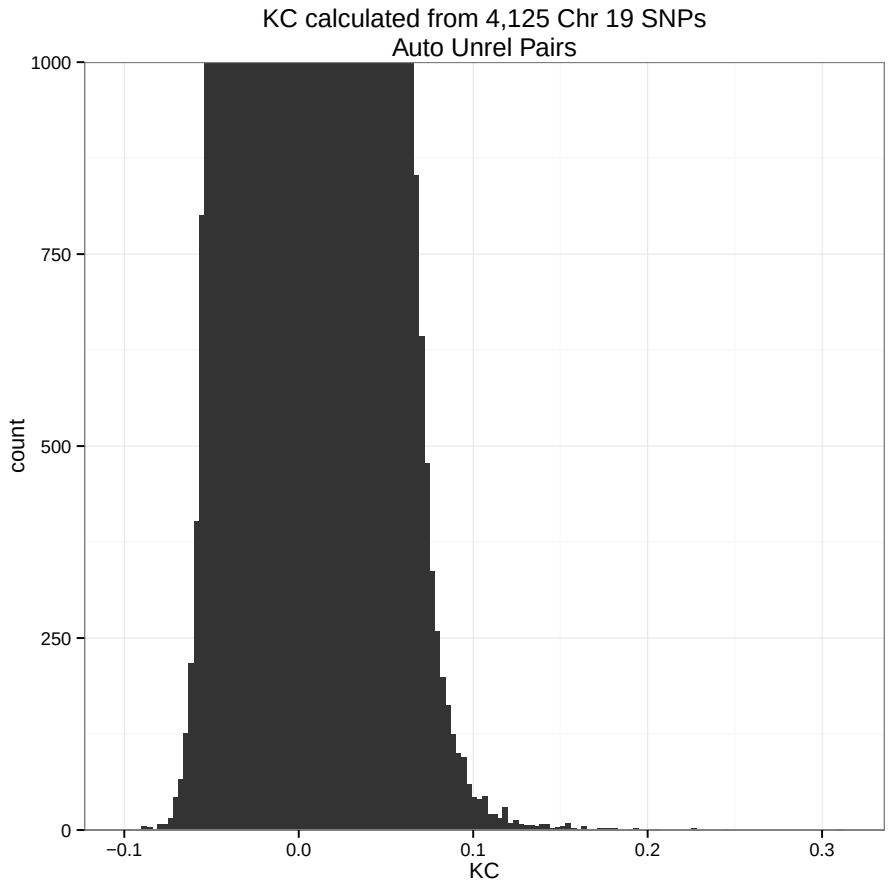


X KC by sex, truncated

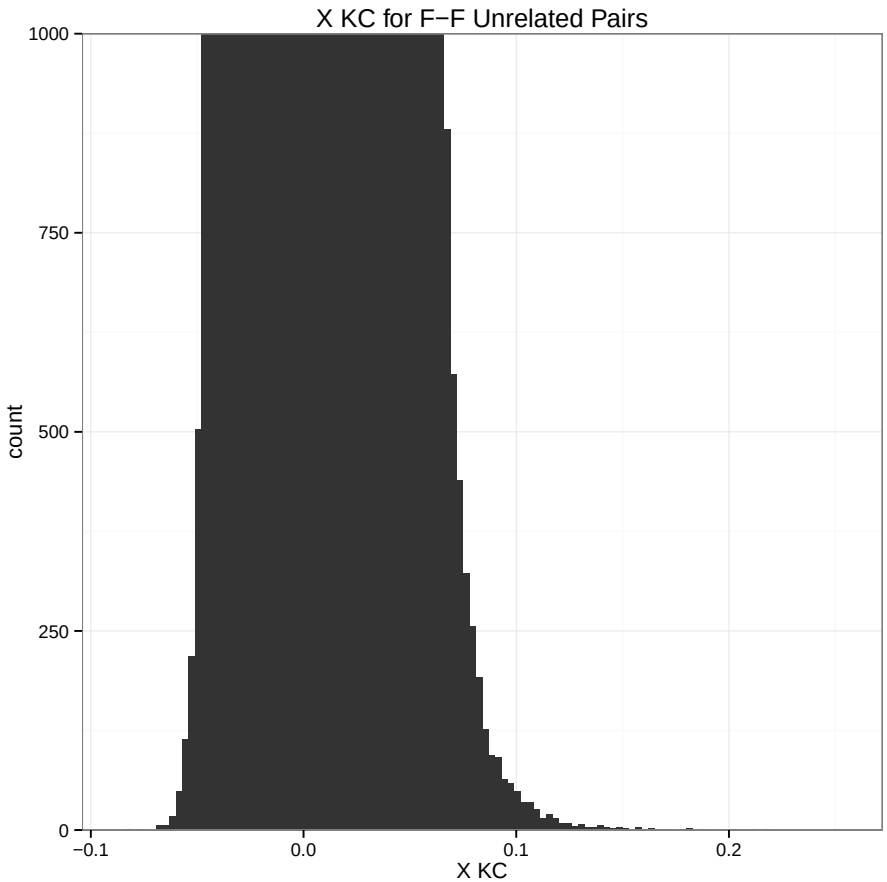


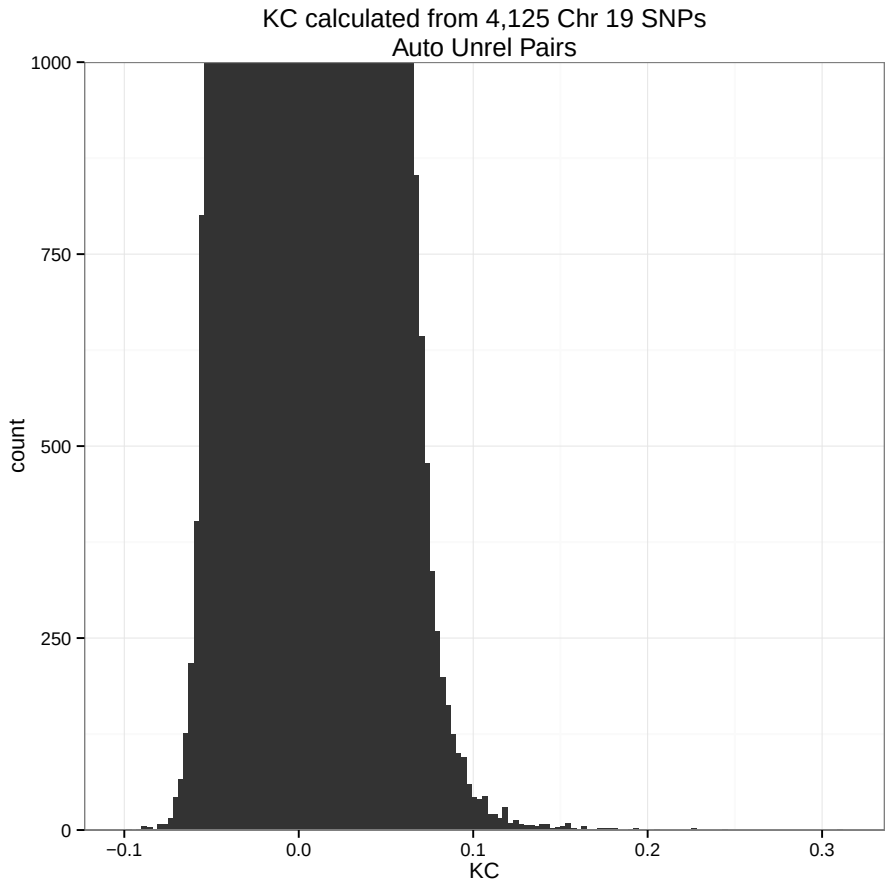
Chr 19 KC for autosomally unrelated samples





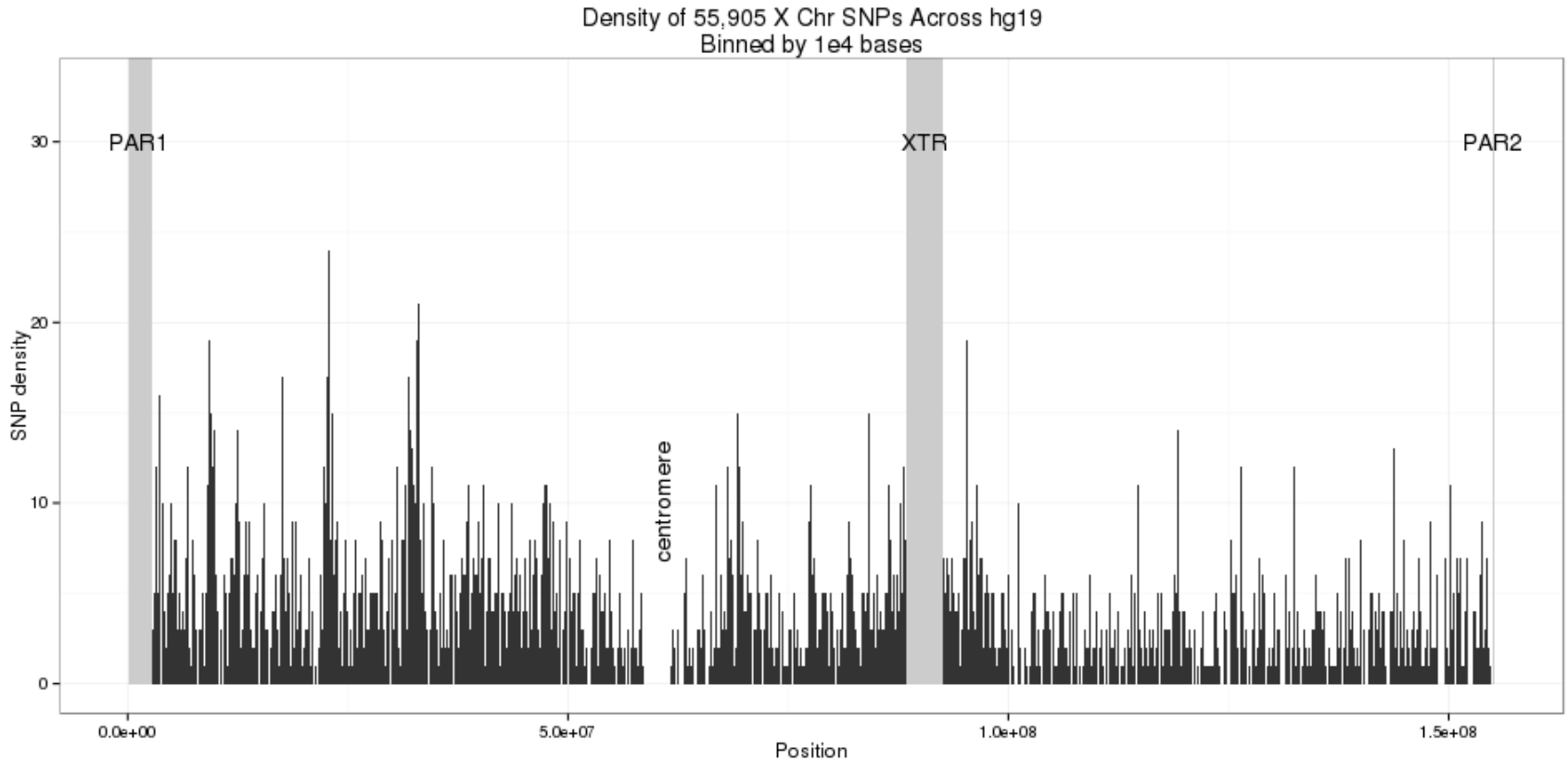
Chr 19 & X chr F-F KC for autosomally unrelated samples





X chromosome SNP density

For running Beagle IBD on the X.



Chromosome 19 SNP density

For comparison to the X.