Association Testing with X Chromosome Data: Adding to Our Pipeline

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27 July 2015

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

    auto genotype $\sim$ auto PC $1 - 5$

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

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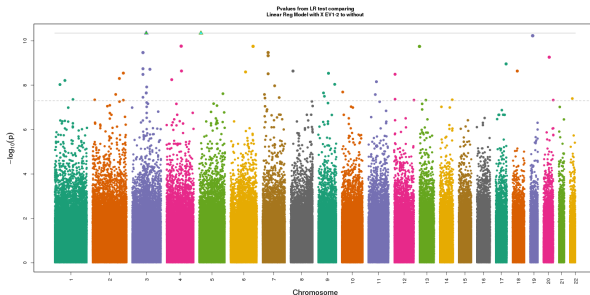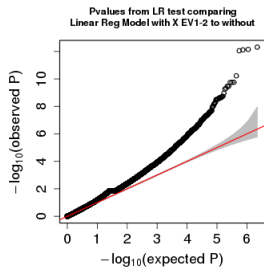- Examine QQ and Manhattan plots.

Are X chromosome effects confounders?

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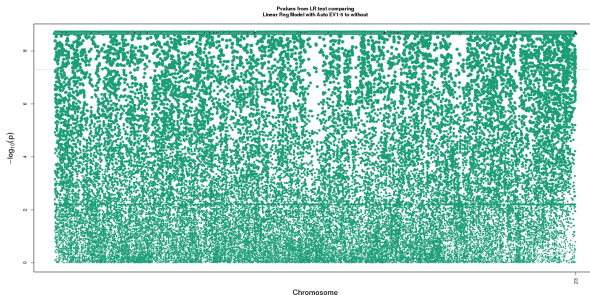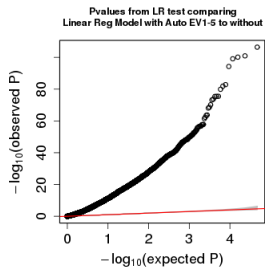
$$\text{X genotype} \sim \text{X chr PC } 1 - 2$$
$$\text{X genotype} \sim \text{X chr PC } 1 - 2 + \text{auto PC } 1 - 5$$

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

Results when testing auto genotypes

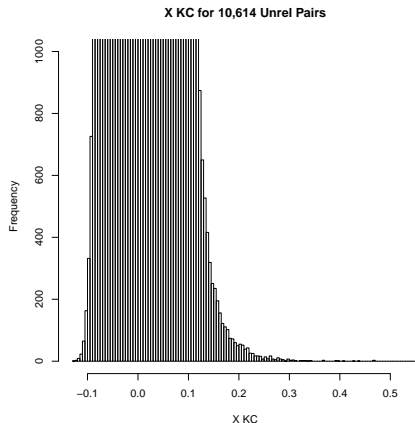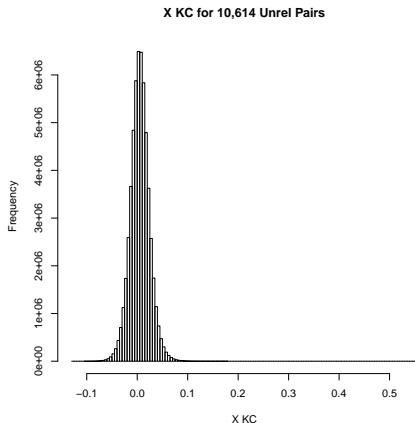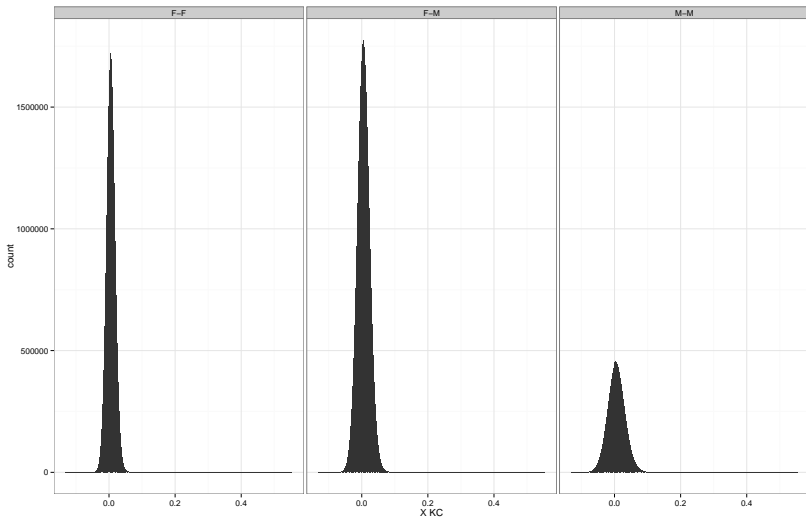# Results when testing X chromosome genotypes

Recall: on the X

X chromosome KC for autosomally unrelated samples.

X KC by sex

X KC by sex, truncated
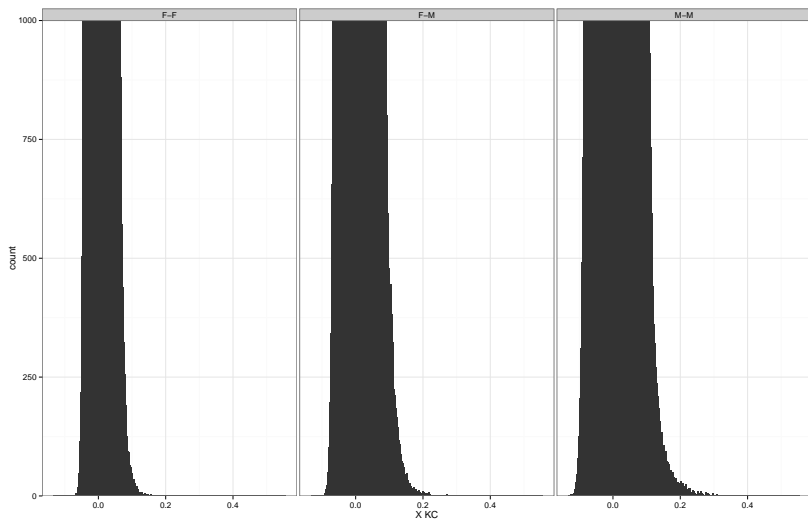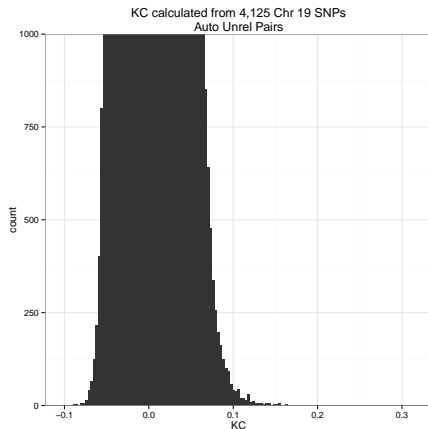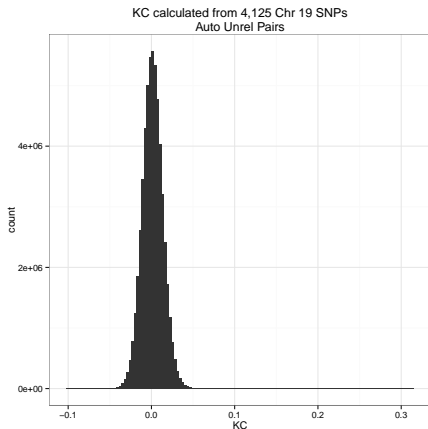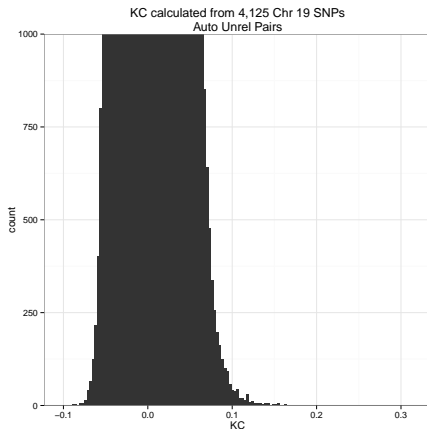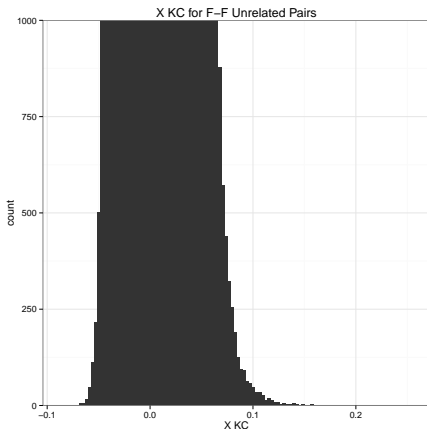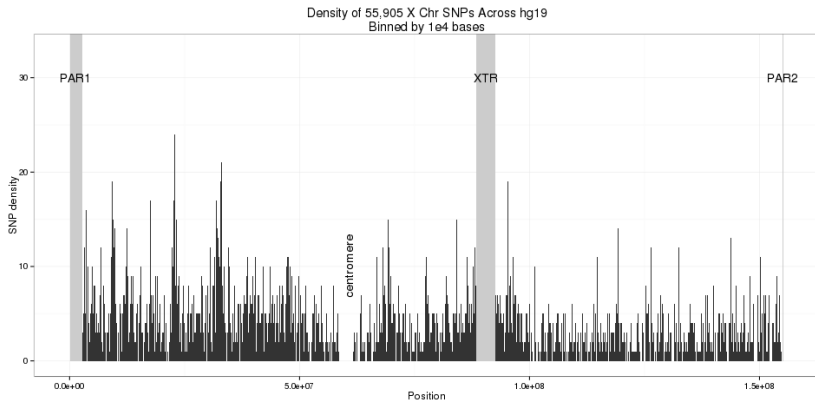
Chr 19 KC for autosomally unrelated samples

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

X chromosome SNP density

For running Beagle IBD on the X.

Density of 55,905 X Chr SNPs Across hg19
Binned by 1e4 bases

Chromosome 19 SNP density

For comparison to the X.

Density of 52,890 Chr 19 SNPs Across hg19
Binned by 1e4 bases