

Genomic evidence for a complex emergence of early modern humans

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Background and motivation

- The emergence of early modern humans several hundred thousand years ago was marked by distinctive morphological and behavioral changes that are evident from skeletal and archaeological data.
- Changes in fossil morphology around 300Kya suggest there was a change in the breeding system towards a more equal distribution of reproductive success (1, 2)
- If such a change did occur, we should see a signal of this in the ratio of X to autosome effective population size.
- The X:A ratio is affected by several evolutionary forces, including demographic, life history, and selective effects
- Here, we set out to test this hypothesis by reconstructing the effective population size history on the X chromosome and autosomes.

Methods

- Data: 1000 Genomes High Coverage genome sequences 81 female YRI individuals (3)
- We ran PSMC (4) on autosomes and X chromosome separately
- Compute ratio of X chromosome to autosome population size through time, correcting for differences in mutation rate between autosomes and X
- Compute variance via jackknife over chromosomes (for autosome analysis) or over 5MB windows (for X chromosome analysis)

Results (continued)

- While the signal appears diffuse across the X chromosome, we do observe a pileup of coalescences at X:66.9-67.5MB and X:77.45-78MB
- Both are unlikely under neutrality ($Z=10, 6.6$)

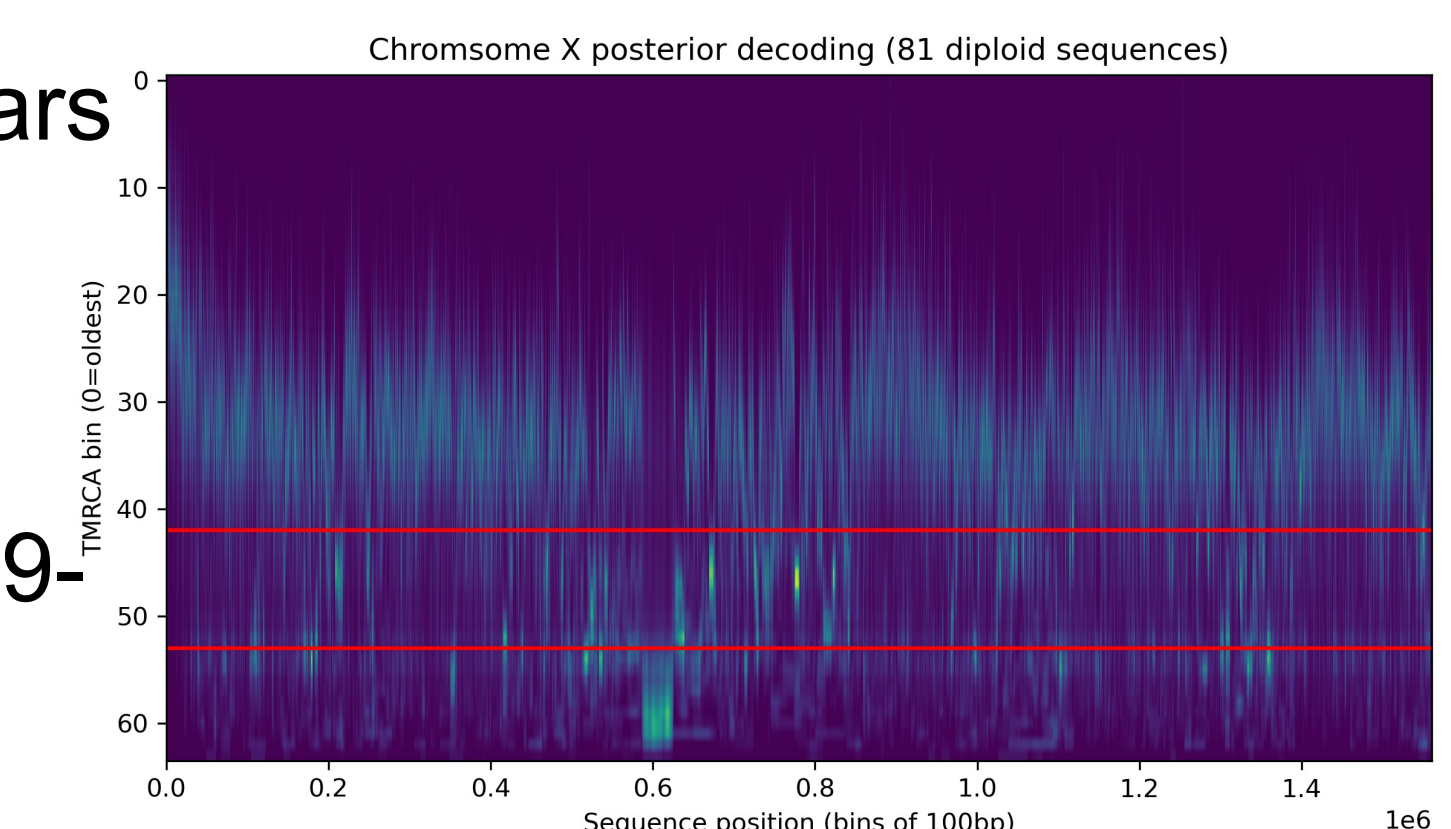


Fig 3. PSMC posterior decoding on chrX averaged across individuals

Results

- For most of human history, X:A ratio is >0.75
- Around 500-300Kya, the X:A ratio drops to the null expectation
- More recently, X:A ratio increases again

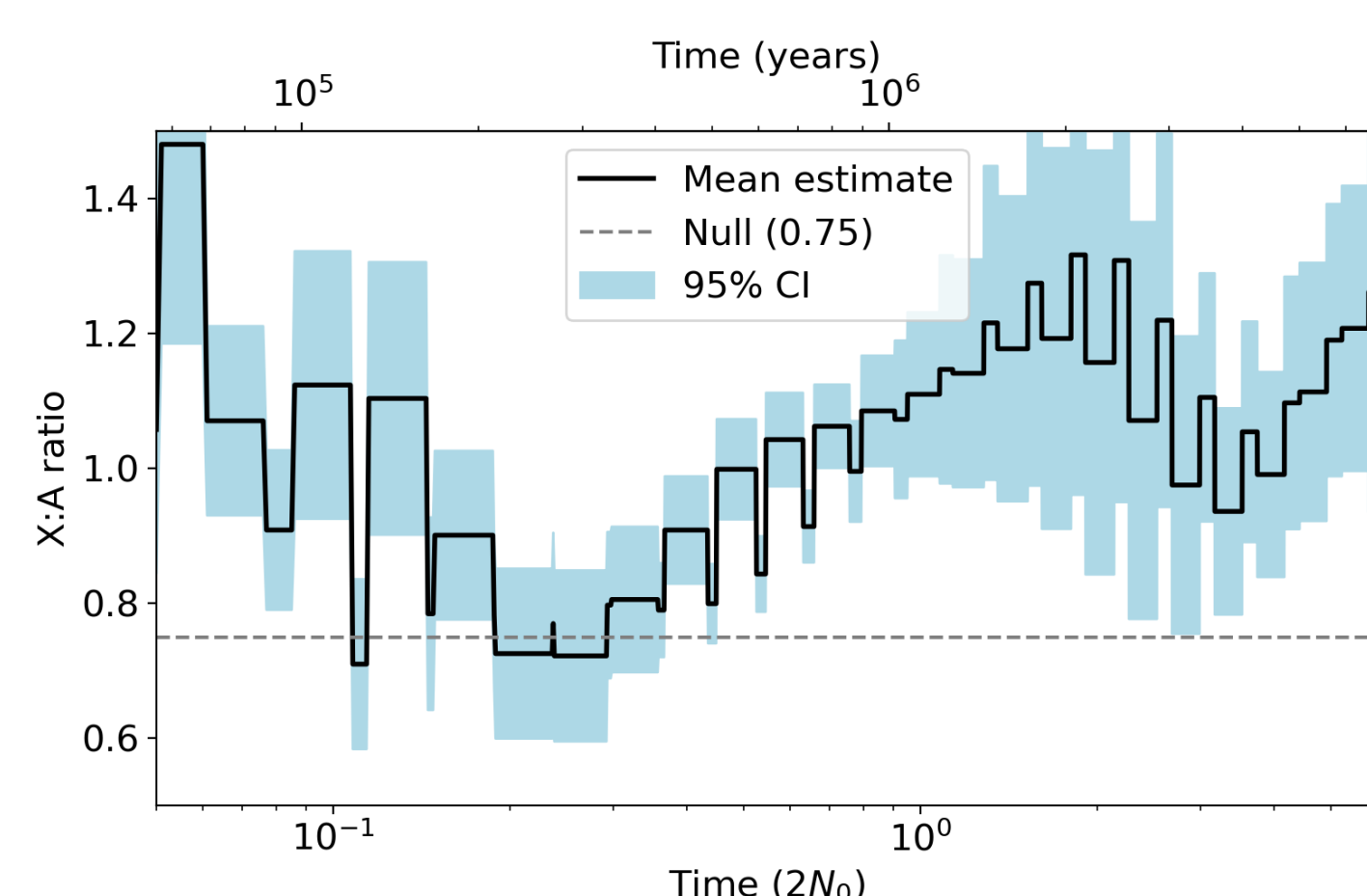


Fig 1. X:A ratio through time.

- A decrease in the ratio could be consistent with selection reducing the coalescent rate on the X chromosome
- Selection tends to act locally, so we tested whether any regions contributed disproportionately to the signal.
- We find that the signal is spread across chromosome X, consistent with demographic effects driving the signal (Fig 2)

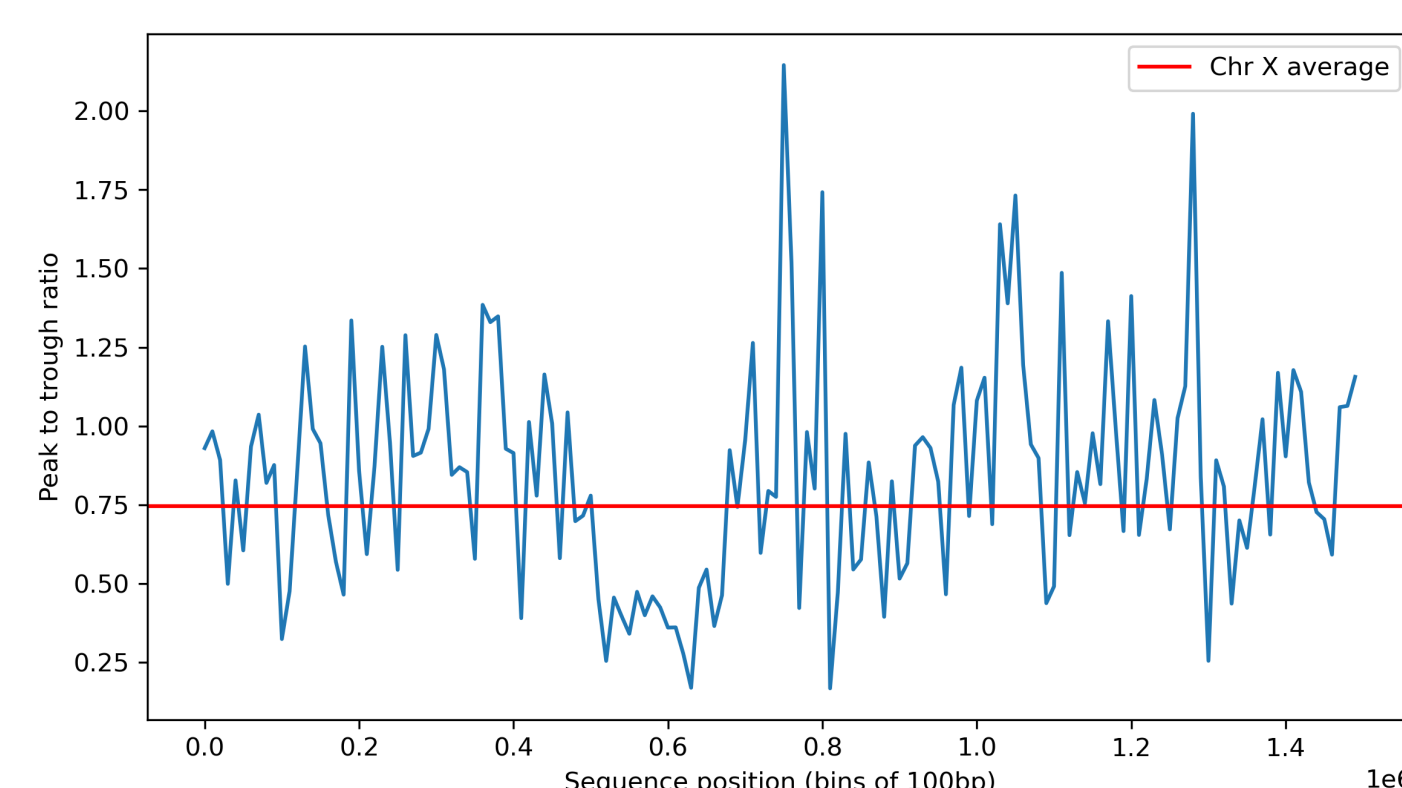


Fig 2. Peak to trough ratio across chromosome X

Conclusions and future directions

- PSMC results are consistent with a change in the variance of male reproductive success between 500-300Kya, around the emergence of early modern humans
- Selection does not appear to drive the signal
- Recent increase in ratio is consistent with a population-specific signal of polygyny in YRI (5)
- Additional modeling may help enumerate other models (including changes in life history traits) that could produce this pattern
- Comparisons to other primates and high-coverage archaic individuals would help to see how widespread this phenomenon is

References

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