# Research Approach: Evolutionary genetics of maize

## Maize as a model for plant evolutionary genetics

Genome size variation in plants, humans. Differences in large complex genomes – Fraser (Fraser 2013) vs. (Pyhäjärvi et al. 2013) or (Hancock et al. 2011) and gene expression (Hufford et al. 2012)

### Domestication

## Local adaptation

## Experimental evolution

### Genome evolution

TE insertions (Studer et al. 2011, Makarevitch et al. 2015) and regulation. genome size too (Tenaillon et al. 2011)

michelle stats. 344K insertions TEs, CNVs, genome size evolution missing data vs. allele frequencies. TEs

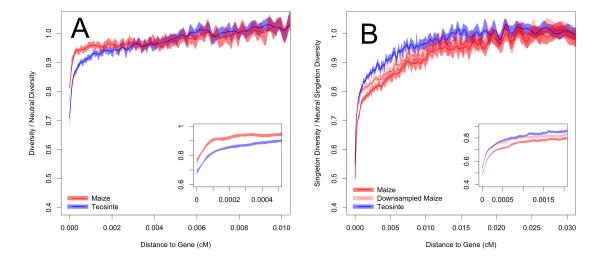


Figure 1: Relative level of diversity versus distance to the nearest gene, in maize and teosinte. Two measures of diversity are shown. A Pairwise neuleotide diversity  $\pi$  is most influenced by intermediate frequency alleles and reflective of long-term evolutionary patterns. The weaker effect of purifying selection seen in maize is consistent with a domestication bottleneck that lowered long-term effective population size. B Number of singletons polymorphisms  $\eta_1$ . These lowest frequency alleles are young, reflecting very recent evolutionary patterns. Here the impact of purifying selection is stronger in maize, consistent with exponential growth following domestication.

#### References

- H. B. Fraser. Gene expression drives local adaptation in humans. *Genome research*, 23(7):1089–1096, 2013.
- A. M. Hancock, B. Brachi, N. Faure, M. W. Horton, L. B. Jarymowycz, F. G. Sperone, C. Toomajian, F. Roux, and J. Bergelson. Adaptation to climate across the arabidopsis thaliana genome. *Science*, 334(6052):83–86, 2011.
- C. N. Hirsch, J. M. Foerster, J. M. Johnson, R. S. Sekhon, G. Muttoni, B. Vaillancourt, F. Peñagaricano, E. Lindquist, M. A. Pedraza, K. Barry, et al. Insights into the maize pangenome and pan-transcriptome. *The Plant Cell Online*, 26(1):121–135, 2014.
- M. B. Hufford, X. Xu, J. Van Heerwaarden, T. Pyhäjärvi, J.-M. Chia, R. A. Cartwright, R. J. Elshire, J. C. Glaubitz, K. E. Guill, S. M. Kaeppler, et al. Comparative population genomics of maize domestication and improvement. *Nature Genetics*, 44(7):808–811, 2012.
- I. Makarevitch, A. J. Waters, P. T. West, M. Stitzer, C. N. Hirsch, J. Ross-Ibarra, and N. M. Springer. Transposable elements contribute to activation of maize genes in response to abiotic stress. *PLoS genetics*, 11(1):e1004915, 2015.
- T. Pyhäjärvi, M. B. Hufford, S. Mezmouk, and J. Ross-Ibarra. Complex patterns of local adaptation in teosinte. *Genome biology and evolution*, 5(9):1594–1609, 2013.
- A. Studer, Q. Zhao, J. Ross-Ibarra, and J. Doebley. Identification of a functional transposon insertion in the maize domestication gene tb1. *Nature Genetics*, 43(11):1160–1163, 2011.

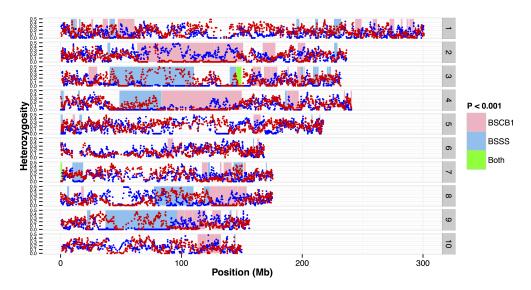


Figure 2: Heterozygosity across all 10 chromosomes of maize from cycle 16 of the Iowa Reciprocal Recurrent Selection experiment. Heterozygosity values in the BSSS (blue dots) and BSCB1 (red dots) populations are superimposed in one panel. 2cM windows of heterozygosity lower than 10 of 10,000 simulations (P < 0.001) are shaded in light blue (BSSS) or pink (BSCB1). Two regions genome-wide show significantly low heterozygosity in both populations and are shaded green.

M. I. Tenaillon, M. B. Hufford, B. S. Gaut, and J. Ross-Ibarra. Genome size and transposable element content as determined by high-throughput sequencing in maize and zea luxurians. *Genome biology and evolution*, 3:219–229, 2011.

Figure 3: The effect of de novo non-reference transposable element insertion on gene expression. Panels show the impact of insertions identified in 1 (far left) to > 10 (far right) inbred lines in a panel of 23 maize inbreds. Shown in each panel is the relative expression of genes in inbred lines with the insertion within the annotated gene region to the mean expression of that gene across 500 maize lines (Hirsch et al. 2014). Recent, low frequency insertions tend to decrease gene expression compared to older, high-frequency insertions.

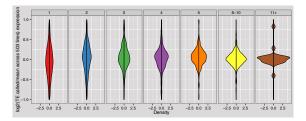


Figure 4: The effect of copy number variation (CNV) on estimates of Tajima's D, a measure of the allele frequency spectrum. In regions with no CNV, Tajima's D is negative consistent with population expansion. Tajima's D is inferred to be strongly postiive in regions with high-frequency CNVs, however, which could be (erroneously) interpreted as population decline or balancing selection.

