

Do Transposons Drive Early Y Chromosome Degeneration?

Zoë K Humphries, Spencer CH Barrett, Stephen I Wright
Department of Ecology and Evolutionary Biology, University of Toronto

Transposons play a number of important and diverse roles in genome evolution. Could they also be affecting early stages of Y degeneration due to the repressive action of the DNA methylation that is supposed to mitigate their accumulation?

TE-riddled Y chromosomes, are known to degenerate over evolutionary time: could this loss of gene function be due in part to the spreading of DNA methylation?

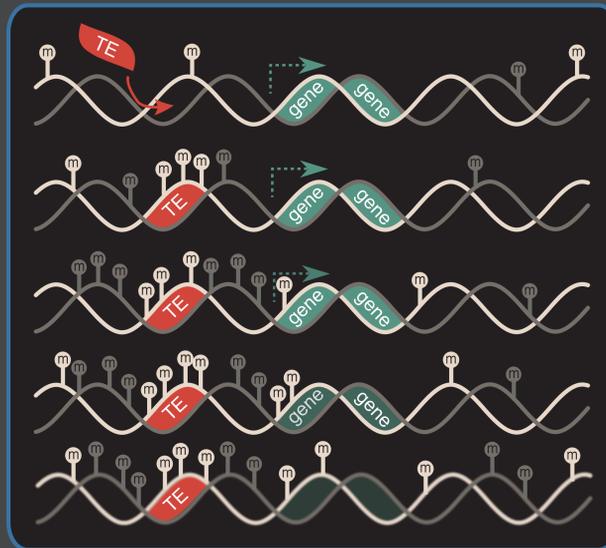
Pathological side effect of genomic immunity

Transposable elements (TEs) aka "selfish genes" make copies of themselves at the expense of their host

DNA methylation may have evolved to mitigate TEs

Methylation has a trade-off: it can spread to nearby genes, lowering expression and even silencing them

Silenced alleles may degenerate even to the point of loss



I hypothesize that the spreading of DNA methylation after TE invasion has played a role in Y chromosome evolution

Examining effects in young sex chromosomes

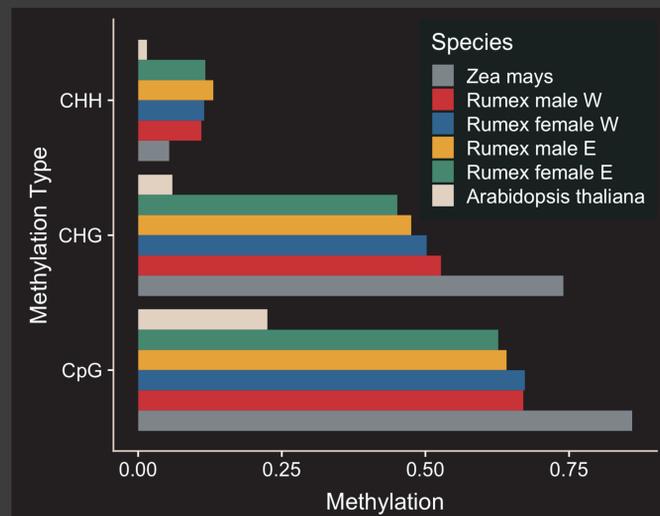
We sampled a male and female pair in two US populations of *Rumex hastatulus*, a species of plant with young sex chromosomes

To measure methylation, we performed whole-genome bisulphite sequencing

These methylome reads were aligned to a reference transcriptome using Bismark (Krueger & Andrews 2011)

To look at the effects of methylation on expression, we incorporated expression data from Hough et al. (2014)

Rumex Has High Levels of Methylation



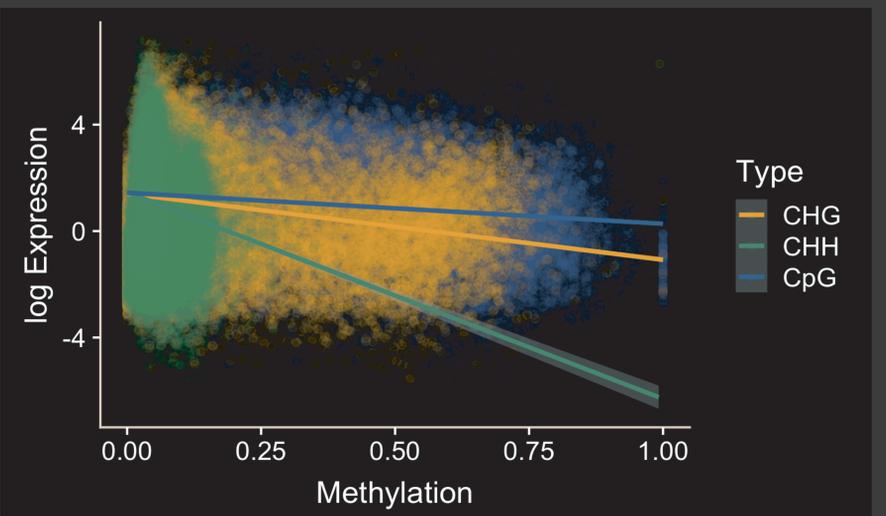
There are three types of methylation: CHH, CHG, and CpG

CHH and CHG are strongly associated with TEs; at low abundance elsewhere

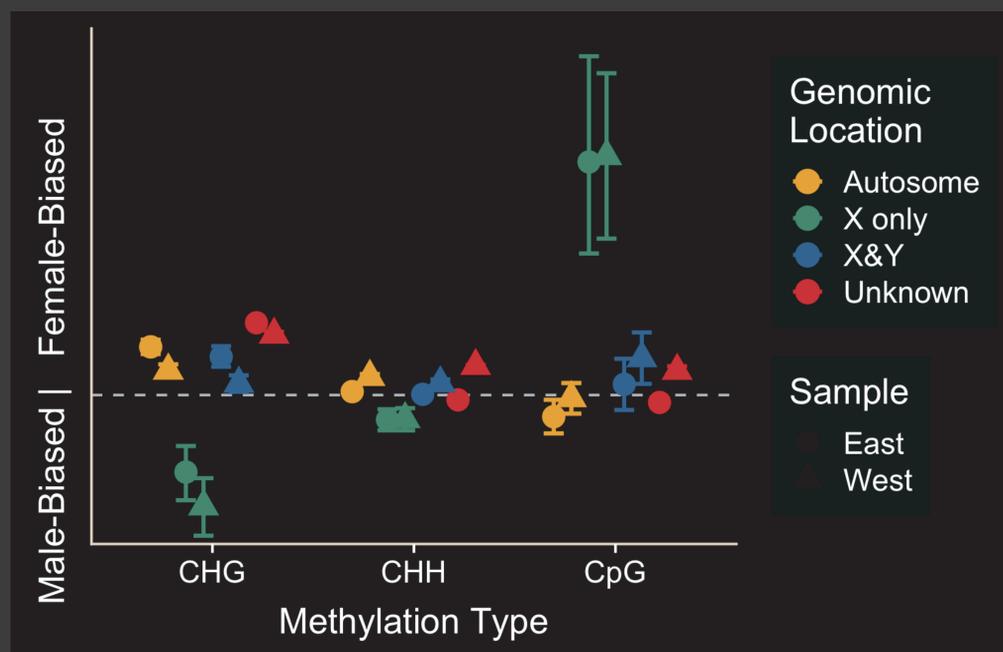
High CHH and CHG also have the greatest tie to lowered expression

Maize (Gent et al. 2013), has a huge genome with lots of TEs, and Arabidopsis (Feng et al. 2010) has a small genome with few TEs

Genes with More Methylation Are Less Expressed



Sex Chromosome Methylation Differences



The category of genes on the X & Y chromosomes contain the Y-linked sequences we currently know about

We expected males to have high TE-associated methylation (CHG, CHH) in these Y-linked sequences

This was not the case: males are methylating their single X instead

These results may be explained by methylome reads mapping to unexpressed, degenerated Y alleles of the "X only" genes

Future Directions

A new reference genome

Increased sample size from four to fourteen

Relationship between TE proximity and methylation of genes

Relationship between level of methylation and gene degeneration

General Relevance

Provide insights into our understanding of sex chromosome evolution, the roles TEs play in genome evolution, and the evolutionary relevance of epigenetics

References

- Feng, S., S. J. Cokus, ...and S. E. Jacobsen. 2010. Conservation and divergence of methylation patterning in plants and animals. *Proc. Natl. Acad. Sci. U. S. A.* 107:8689–94.
Gent, J. I., N. A. Ellis, L. Guo, A. E. Harkess, Y. Yao, X. Zhang, and R. K. Dawe. 2013. CHH islands: de novo DNA methylation in near-gene chromatin regulation in maize. *Genome Res.* 23:628–637.
Hollister, J. D., and B. S. Gaut. 2009. Epigenetic silencing of transposable elements: a trade-off between reduced transposition and deleterious effects on neighboring gene expression. *Genome Res.* 19:1419–28.
Hough, J., J. D. Hollister, W. Wang, S. C. H. Barrett, and S. I. Wright. 2014. Genetic degeneration of old and young Y chromosomes in the flowering plant *Rumex hastatulus*. *Proc. Natl. Acad. Sci. U. S. A.* 111:7713–8.
Krueger, F., and S. R. Andrews. 2011. Bismark: A flexible aligner and methylation caller for Bisulfite-Seq applications. *Bioinformatics* 27:1571–1572.

