"Jumping" into gene regulation:

Understanding the regulatory adaptation of transposable elements and their effect on gene expression in maize

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SACNAS NDiSTEM 2022
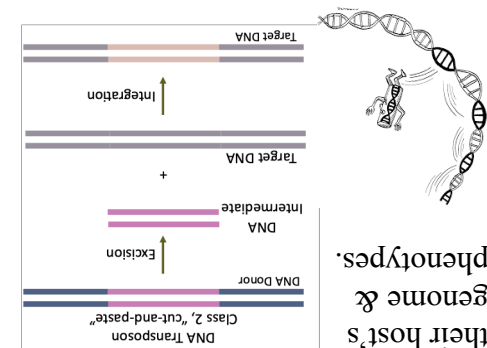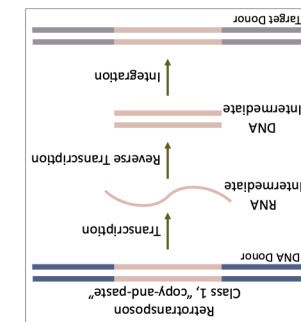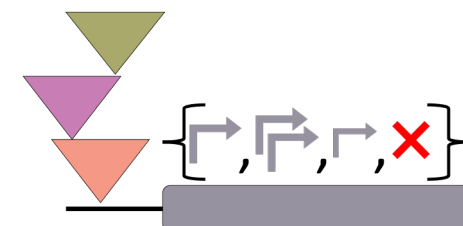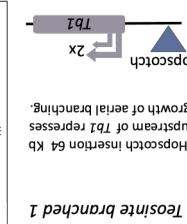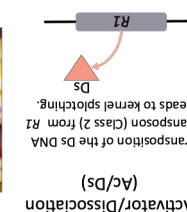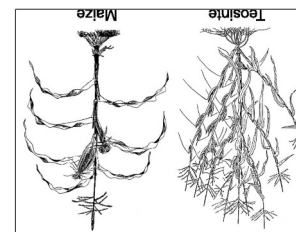Merritt Khaipho-Burch

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Transposable elements (TEs), or "jumping genes," are repetitive mobile genetic sequences that can alter their host's genome & phenotypes.

Class 1, "copy-and-paste" Retrotransposon — DNA Donor, Transcription, RNA Intermediate, Reverse Transcription, DNA Intermediate, Integration, Target Donor

Class 2, "cut-and-paste" DNA Transposon — DNA Donor, Excision, DNA Intermediate, Integration, Target DNA

In maize, 85% of the genomic sequence is made up of transposable elements. These TEs can alter kernel coloration, gene expression, and other phenotypes.

Hopscotch insertion 64 Kb upstream of *Tb1* represses growth of aerial branching.

Teosinte branched 1. Maize / Teosinte. Tb1, Hopscotch 2x

Transposition of the Ds DNA Transposon (Class 2) from *R1* leads to kernel splotching.

Activator/Dissociation (Ac/Ds). R1, Ds

Methods

Collected and sequenced 372 inbred and hybrid maize samples and measured gene expression.

Growing point, Leaf tip, Leaf base, Flag leaf at flowering

Generated a table of when TE insertions were either present or absent across our diverse maize lines.

Taxa	Model 1	Model 2	Model 3
	Gene A TE1	Gene A TE2	Gene B TE3
B73	1	1	0
Mo17	1	0	0
Ky21	1	1	0
Hp301	1	0	0

Created a Kinship matrix to determine how related our maize lines were.

	B73	Mo17	Ky21	Hp301
B73	1.0	0.9	0.5	0.5
Mo17	0.3	1.0	0.7	0.3
Ky21	0.7	0.7	1.0	0.0
Hp301	0.5	0.3	0.0	1.0

Hypothesis

Due to their abundance, transposable elements have become a source of gene regulatory grammar in maize that impacts RNA expression.

No TE — Promoter, Gene
Addition
Disruption
Silencing

This regulatory grammar can be provided through multiple methods.

Our model

RNA Expression ~ TE Presence or Absence + Kinship

Flag Leaf, Growing Point, Leaf Base, Leaf Tip

Count — Mixed Model P-value

Our models suggest that only 0.9% of all TE insertions (11,811) significantly ($p<0.05$) impact the expression of 14% of all maize genes (5,418).

Key Takeaways

TE1 VS. TE2 — Effect on gene — Promoter

Due to high relatedness, we can only study TE effects in a small number of genes (34%).

0.9% ... 14%

In those 34% of genes, 11,811 (0.9%) TE insertions impact 5,418 (14%) genes.

TE1, TE2 — Chromatin status, Methylation status, Promoter, Effect on gene

Future work includes characterizing the regulatory landscape surrounding TEs and their associated genes.

Have any questions, suggestions, or available positions?

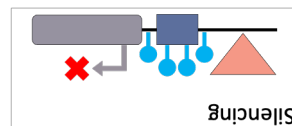Graduating spring 2023

Contact me:

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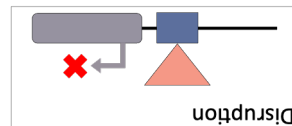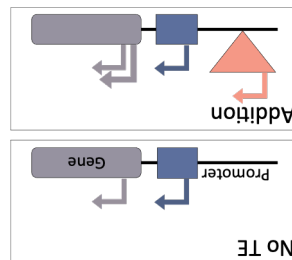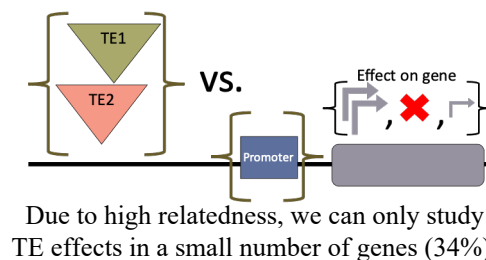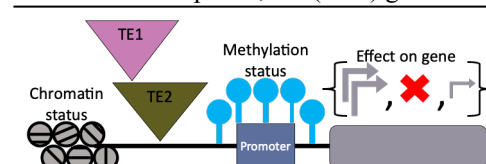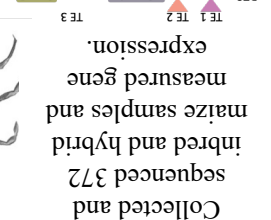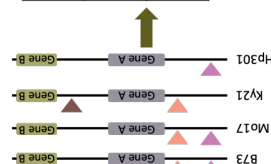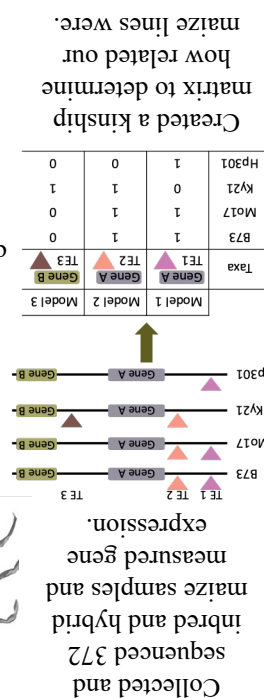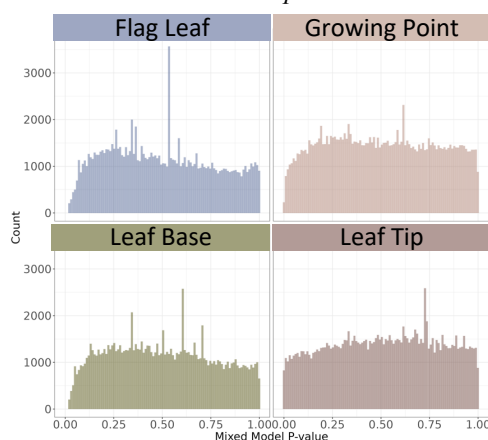

Check out my Google Scholar profile!

Unfold this zine for my CV!

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Ph.D. Candidate

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EDUCATION

May 2023
Cornell University, NY
Doctor of Philosophy,
Plant Genetics, minor Genomics

May 2018
South Dakota State University, SD
Master of Science,
Biological Sciences

May 2016
University of Hawaii at Hilo, HI
Bachelor of Science,
Biology (Cell and Molecular)

RELEVANT SKILLS

R/RStudio

Association Analyses

Genomic Prediction & Selection

Machine Learning

RNAseq Data Analysis

Genome Alignment

Cloud Based Computing (Google CO-Lab, HPC services)

Microsoft Office

Slack

CAREER OBJECTIVE

I am an geneticist with a broad range of experiences in quantitative genetics, classical transmission genetics, molecular biology, plant breeding, and bioinformatics using the model organism *Zea mays*. Skilled in designing and implementing models to understand the genetic basis and regulation of complex quantitative traits.

PROFESSIONAL EXPERIENCE

August 2018–Present
Ph.D. Student
Buckler Lab, Cornell University, Ithaca, NY

- <u>Topics</u>: Elucidating the patterns of pleiotropy and its biological relevance in maize AND Regulatory Adaptation of Transposable Elements and Their Effect on Gene Expression in Maize and the Andropogoneae
- Using association mapping I curated and mapped 120,549 traits across two diverse maize populations to uncover the patterns of pleiotropy and created multiple machine learning models (random forest, gradient boosting) to describe how these pleiotropic loci were biologically regulated.
- Used mixed linear models to understand the impact of transposable elements on gene expression across 372 maize inbreds and hybrids.

HONORS AND AWARDS

2022 - USDA NIFA Predoctoral Fellowship for "*Regulatory Adaptation Of Transposable Elements And Their Effect On Gene Expression In Maize And The Andropogoneae*". Total amount: **$180,000.**

2022 – David L. Call Award from the American Indian and Indigenous Studies Program. Given to one Indigenous graduate or professional student per year who displays outstanding leadership, service, and commitment in pursuit of a graduate degree.

2021 – Best PhD Poster Video at the 6th International Conference of Quantitative Genetics.

2017 – Helen Roberti Native American Scholarship, South Dakota State University

SELECT PUBLICATIONS

*Corresponding author, **Joint Corresponding

Khaipho-Burch, M.*, Ferebee, T., Giri, A., Ramstein, G., Monier, B., Yi, E., Cinta Romay, M., & Buckler, E. S. (2022). Elucidating the patterns of pleiotropy and its biological relevance in maize. In bioRxiv (p. 2022.07.20.500810).

Maharjan, N., **Khaipho-Burch, M.****, Awale, P., Gyawali, A., Shrestha, V., Wu, Y., & Auger, D. L. (2022). Genomic mapping of the modifiers of teosinte crossing barrier 1 (Tcb1). In bioRxiv (p. 2022.07.18.500501).

Gage JL, Mali S, McLoughlin F, **Khaipho-Burch M**, Monier B, Bailey-Serres J, et al. Variation in upstream open reading frames contributes to allelic diversity in maize protein abundance. Proc Natl Acad Sci U S A. 2022;119: e2112516119.

Washburn JD, **Burch MB**, Franco JAV. Predictive breeding for maize: Making use of molecular phenotypes, machine learning, and physiological crop models. Crop Sci. 2020;60: 622–638.