

Translational Bioinformatics Frameworks and Al Solutions for Multiomics Research



AgBioData Webinar Aug 6, 2025

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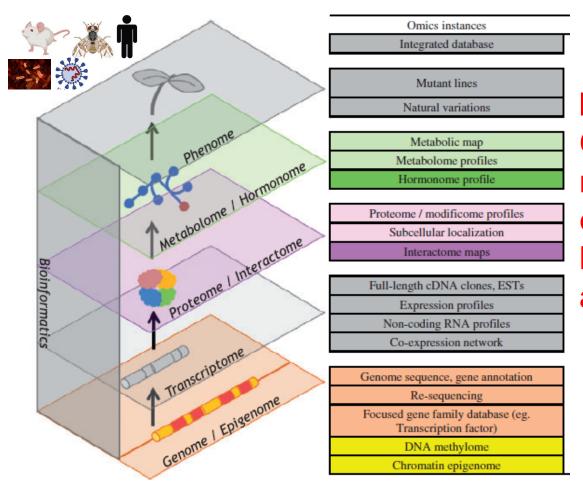
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Outline

- Introduction
- Translational Bioinformatics Frameworks and Al Solutions
 - Multiomics Data Framework Development
 - Multiomics Data Integration Tool Development
 - ➤ Multiomics Informatics Method Development



Genomics and Multiomics Datasets



Informatics and
Computational methods have
rapidly become an essential
component for all biological,
biomedical, animal science
and plant science studies!





The Model Has Changed!!

The Model of Generating and Consuming Data has Changed!!

Old Model: Few organizations are generating data, all others are consuming data



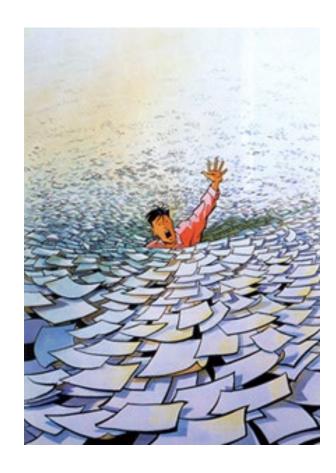
New Model: All of us are generating data, and all of us are consuming data





What's Challenging?

- It is the diversity of high-throughput technologies and the amount of generated data that need to be integrated together, to convert data into meaningful knowledge and making novel discoveries!
- Multiomics data are often individually scattered across different databases and repositories, making it difficult for its users to access them efficiently.
- Getting a comprehensive view from data integration needs advanced techniques.

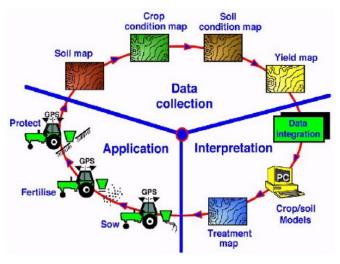




Precision Agriculture

- Precision agriculture is one of many modern farming practices that make production more efficient.
- Decisions based on many Agronomic Data
 - Hybrid selections, Plant populations
 - Yield data , Soil data
 - Pesticide, Fungicides
 - Genotypes, Environments
 - Economics, Market Prices
- Data generated can be utilized and applied for selected crop breeding, improving yield, pest management, crop disease analytics, spraying decisions and many more.





Comparetti et al. 2015





Modern Farming







BIG Data Challenges





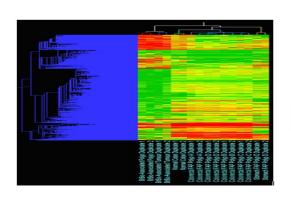


Emerging Trends

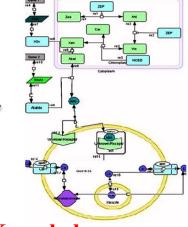




Joshi Lab **Enabling Translational Research**















Data: Bulk & Single Cell

Genomics & Epigenomics

Transcriptomics

Proteomics

Metabolomics

Phenomics and Others

Data volume: huge **Data types: diverse** Data access: spread Data complexity: high



Knowledge:

Functions

Pathways

Regulation

Networks

Homology

Phenotypes:

Plants & Crops

Animals & Pets

Human & Diseases

Microbes & Viruses



Precision Agriculture Precision Medicine Food Security Genomic Epidemiology





Joshi Lab - 3 Essential Research Pillars

Translational Bioinformatics

Multiomics
Informatics
Data Analytics
&
Framework

☐ High Throughput Analytics

Development

- Database and Web-BasedPortals Development
- Downstream Analysis andInference Tools Development
- ☐ Training and Feedback

Multiomics
Informatics
Data Integration
&

Web-based Tool Development

- Multiomics Data IntegrationTechniques
- □ Web-based Tool Development for discovery
- □ Public Accessibility and User Base Support

Multiomics Informatics Method &

Algorithm Development

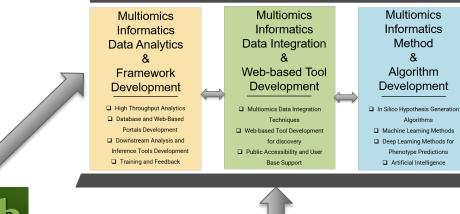
- ☐ In Silico Hypothesis Generation
 Algorithms
 - Machine Learning Methods
 - Deep Learning Methods for Phenotype Predictions
 - ☐ Artificial Intelligence (AI)





Joshi Lab

Enabling Translational Bioinformatics Research



Soybean Knowledge Base

https://soykb.org/



KBCommons Knowledge Base

https://kbcommons.org/



Multiomics Data Integration

Allele Catalog Tool

GenVarX Tool

SNPViz Tool

AccuTool

MADis Tool



G2PDeep

https://g2pdeep.org/

IMPRes

https://impres.missouri.edu/impres

scPlantAnnotate

https://scplantannotate.missouri.edu/

scPlantAnnotate





Translational Bioinformatics Resources and Al Solutions

- Multiomics Data Framework Development
 - SoyKB and KBCommons
 - ❖3D Omics Suite
 - Cross-species and Comparative Multiomics Translation (CCMT) Tool



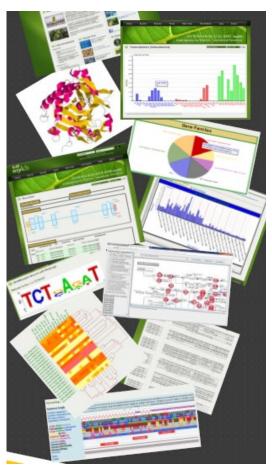
Soybean Knowledge Base (SoyKB)

- SoyKB is a comprehensive all-inclusive, "one-stop-shop" web resource bridging soybean translational genomics and molecular breeding
- SoyKB provides integration of multi-omics data types including
 - Epigenomics
- Genomics
- Transcriptomics
- Proteomics
- Metabolomics
- Phenomics
- SoyKB provides access to analytics linked to XSEDE HPC and cloud data storage on Cyverse
- Has 1000+ registered users across academic and Industry all over the world



https://soykb.org





Joshi et al. Plant Genomics Databases 2016; NAR 2014; BMC Genomics 2012.





KBCommons Informatics Framework

- Knowledge Base Commons (KBCommons) comprehensive framework providing web-based access for
 - Storage, management, sharing, analysis, integration and visualization of genomics and multi-omics data.
 - Adopts, expands and customizes our SoyKB tools for other model organisms, crops, animals and biomedical diseases.
 - Provides seamless links to HPC and cloud resources and integration of in-house developed suites of graphical visualization tools.



Available for various important species including:

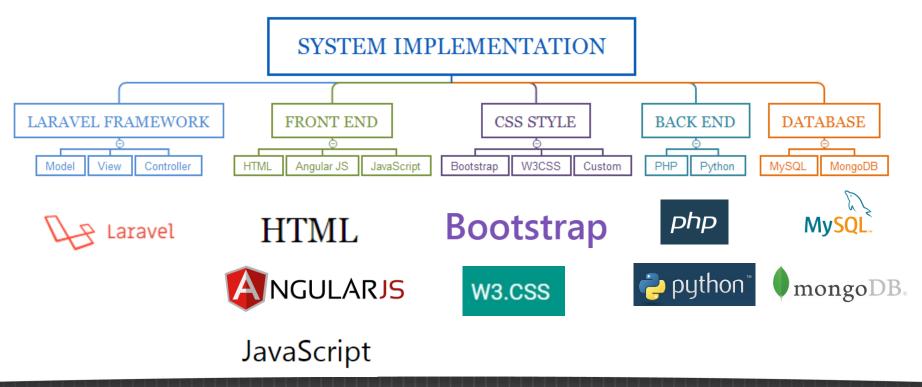
- Plant & Crops (Maize, Arabidopsis, Rice, Camelina, Cuphea, Pennycress)
- Animal & Pets (Mouse, Rat, Dog, Worm)
- Human & Diseases (Cancer, Endometriosis)
- Microbes & Viruses (HIV, B. japonicum, SARS-CoV-2)





KBCommons Architecture

 KBCommons is implemented using Laravel-PHP framework, Javascript, PHP, MySQL and MongoDB databases.







KBCommons - V1 Website redesign currently ongoing !! - V2



KNOWLEDGE BASE COMMONS



BROWSE KB

Below is the list of Organism we have. Please select to proceed using the tools.



CONTRIBUTE TO KB

Got new data to update the existing organism with? Click below to add. Please login first!!



ADD VERSION TO KB

Did not find the specific version of Organism you are looking for?? Click below to add.



CREATE NEW KB

Did not find the KB of Organism you are looking for?? Click below to add.

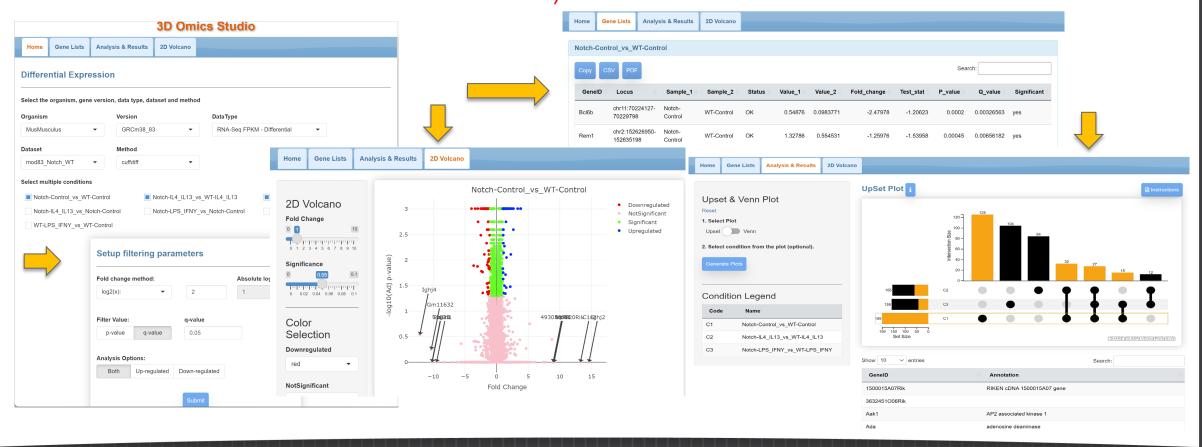








Differential Expression Suite being upgraded to 3D Omics Studio to support diverse multiomics data and now incorporate Upset Plots, Function and Pathways Enrichment Analysis (GO, KEGG, Reactome) and PPI Networks





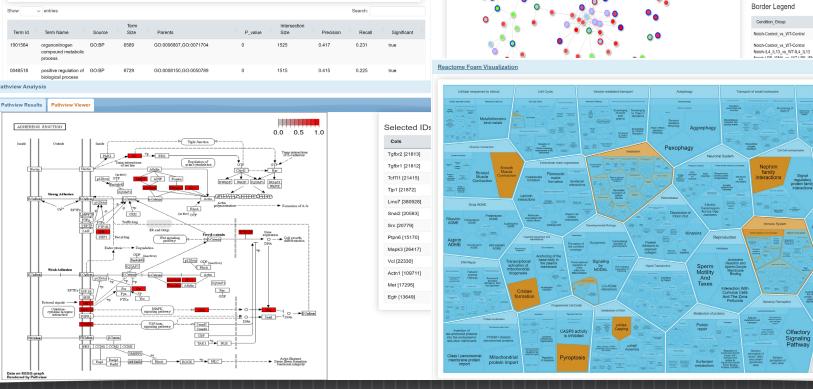


3D Omics Studio

Enrichment Analysis



KEGG Pathways



PPI Network

Reactome Foam View





Select by Group V

Cross-species and Comparative Multiomics Translation (CCMT) Tool



Tool in KBCommons for comparing multiomics data between species and/or between various multiomics data types within species and generating gene regulatory networks



Cross-Species Tool

Comparative Tool

GENIE3 Regulatory Network





CCMT Tool - Cross-Species



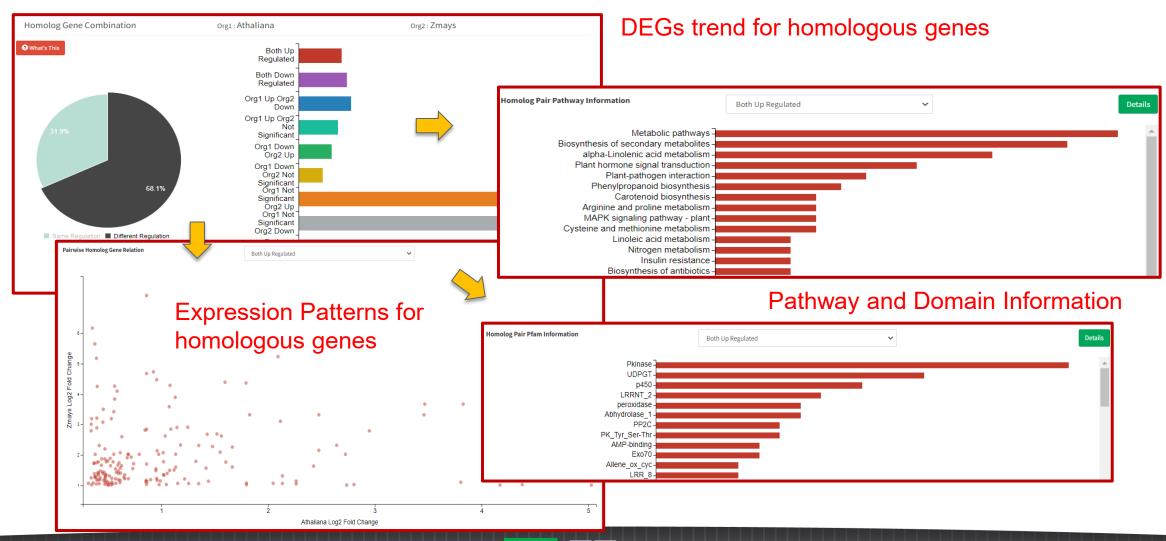






CCMT Tool – Cross-Species



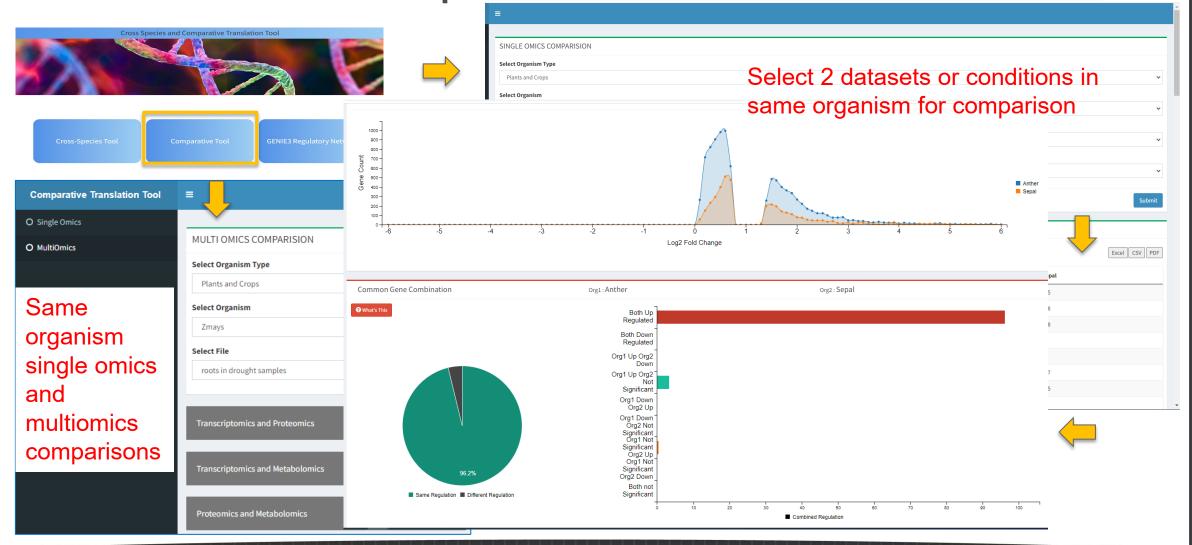






CCMT Tool - Comparative

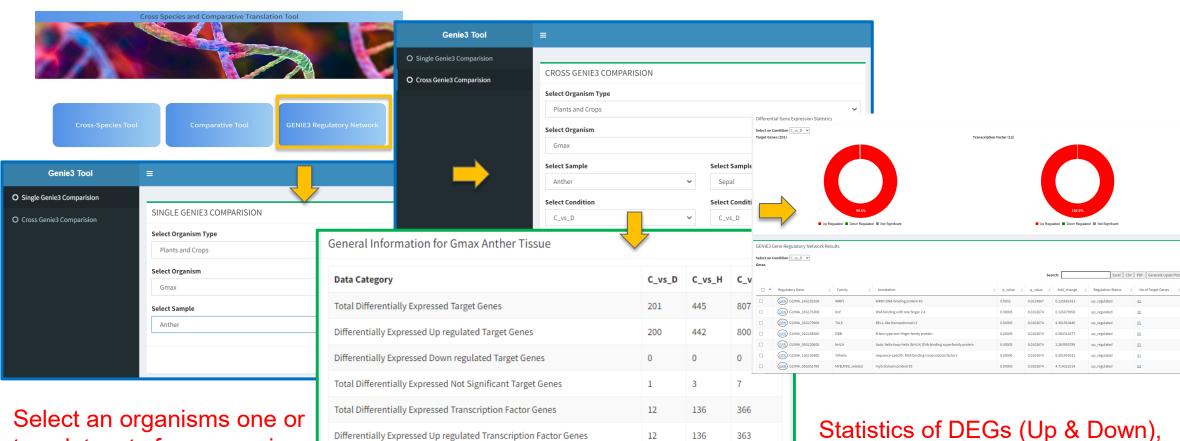






CCMT Tool – Regulatory Network





two datasets for comparison for gene regulatory analysis

TFs and Regulated Target genes with fold changes in condiitons



Differentially Expressed Down regulated Transcription Factor Genes

Total Differentially Expressed Not Significant Transcription Factor Genes



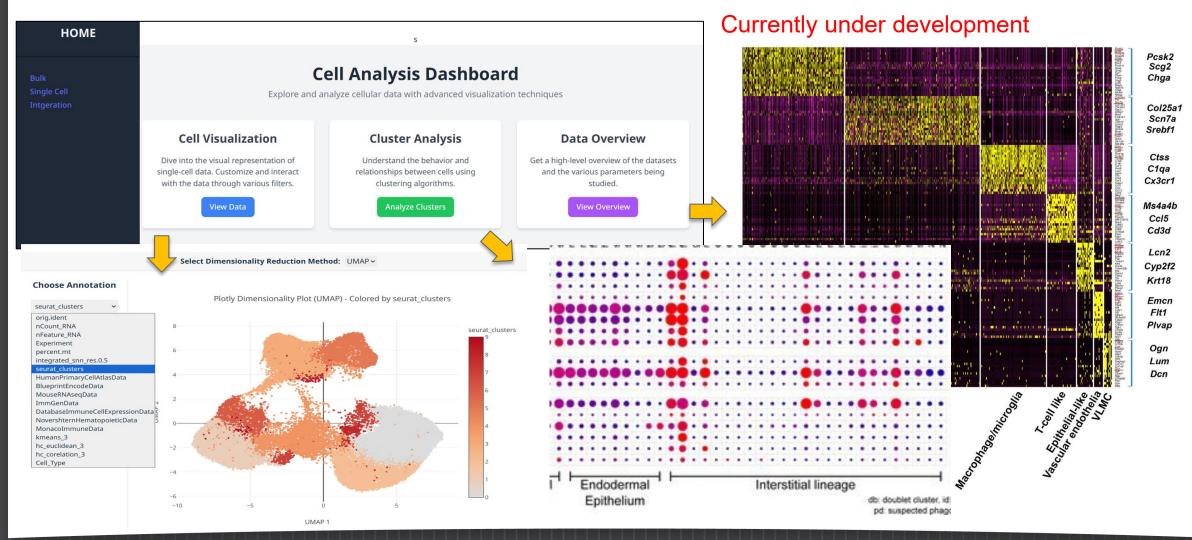
CCMT Tool – Regulatory Network







Single Cell Data Visualization in KBCommons







Translational Bioinformatics Resources and Al Solutions

- ➤ Multiomics Data Integration Tool Development
 - Web Based Tools in SoyKB and KBCommons
 - SNPViz v2.0
 - AccuTool
 - Allele Catalog
 - GenVarX

- Phenotype Distribution Tool
- Protein Sequence Logo Tool
- MADis



NGS Resequencing Data in Soybean

Soybean Curated Panels	Soy775	Soy1066	Soy2939
Number of Positions	37,207,330	38,302,167	44,301,648
Number of Positions (SNPs)	31,694,543	32,524,427	37,351,027
Number of Positions (Indels)	5,738,668	5,523,044	6,691,310
Number of Samples	770	1,066	2,939



Utilizes the NGS resequencing data generated over a decade ago from prior USB investments and enables newer discoveries by making it more accessible and interpretable for the community

Curated panels upgraded with newer accessions data from our collaborators

Community can submit their accessions of interest for yearly upgrades in future





SoyHub in SoyKB

https://soykb.org/soyhub.php/

A "HUB" for collection of soybean applied genomics prediction tools based on the soybean curated panels (Soy775, Soy1066 and Soy2939) including diverse resequenced accessions.



SOYBEAN KNOWLEDGE BASE (SoyKB) A web resource for Soybean Translational Genomics





Welcome to SoyHUB

A hub for soybean-applied genomics predictions based on a curated panel of diverse soybean resequenced accessions (Soy1066).

Explore variation:

Allele Catalog

- Find accessions with certain allele
- Find new alleles in known genes

GenVarX

- Explore variation in promoters
- Search TFs
- Explore CNV

MADis

- Mutative allele discovery
- Mutative allele position combinative calculations

Protein Sequence Web Logo

Generates sequence logos for protein based on multiple organisms

Predict new causal mutations:

AccuTool

- Use GWAS results for prediction
- Calculate Accuracy for your markers or candidate causative mutations (CM) based on Soy775 35.7M variant positions

SNPViz

- Check genomic context of your variant positions in empowered haplotype viewer on various resequenced data sets

Reference Interassembly Gene Browser

Search between reference genotypes, genome assemblies, or

Survey for SoyHUB

We would love to hear from you about your experience with SoyHUB and our genomic analysis tools. Your opinion is valuable to us as it will help us improve our existing tools and develop new features. The survey should only take you a few minutes.

Your responses will be completely anonymous unless you leave your email address to get in touch with us. We appreciate your input!

Click here for Feedback Form





Tools and Methods Developed for Soybean (and beyond)

- SNPViz http://soykb.org/SNPViz2/
- AccuTool https://soykb.org/AccuTool/
- Soybean Allele Catalog
 https://soykb.org/SoybeanAlleleCatalog
 Tool/
- Soybean Genomic Variation Explorer (GenVarX)

https://soykb.org/SoybeanGenVarX/

- Soybean Phenotype Distribution Tool
 https://soykb.org/SoybeanPhenoDistTool
- Soy Protein Sequence Logo Tool https://soykb.org/SoybeanProteinSequenceLogos/
- Soybean Multiple Alleles Discovery (MADis) Tool

https://soykb.org/SoybeanMADisTool/





SNPViz v2.0 for Haplotype Analysis and Allele Discovery



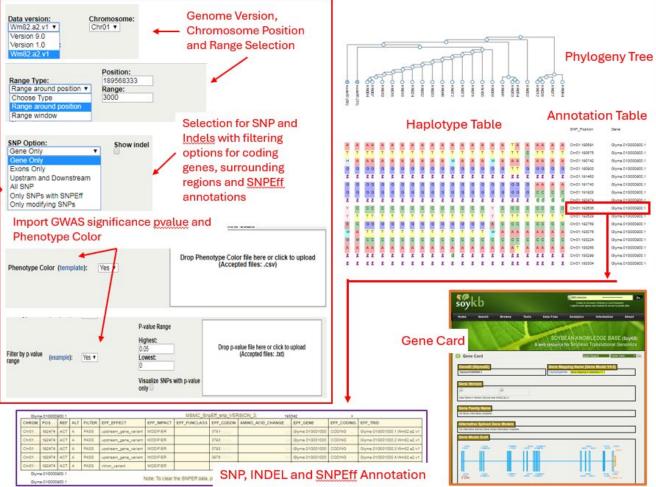
SNPViz v2.0 for haplotype analysis and visualization of phylogeny and haplotype to discovery allelic variations.

Langewisch et al. Plos One

Zeng et al. BIBM 2020,

IJDMB 2021

2014

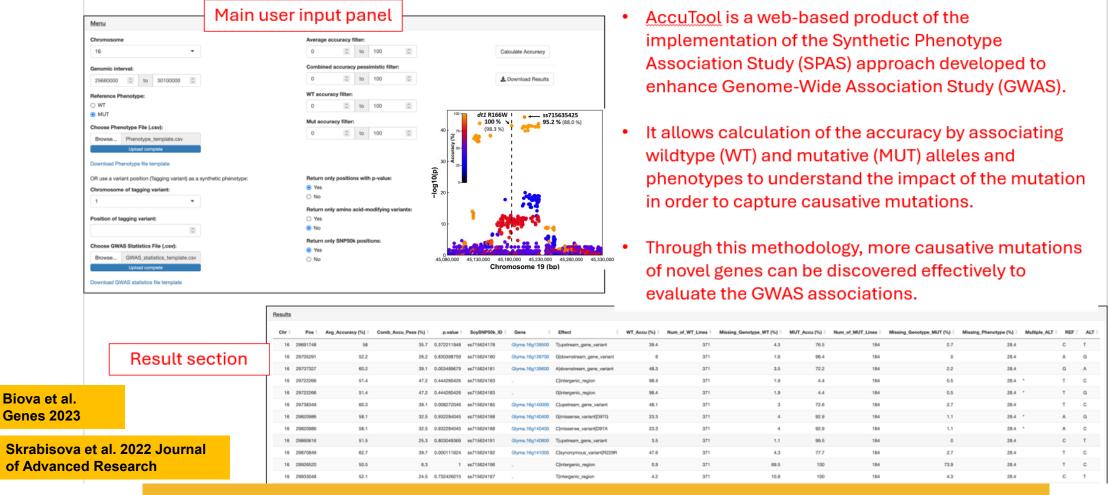


Tool is designed to utilize Soy Curated Panels to enable efficient identification of soybean genes that control phenotypes by combining GWAS results and haplotype analysis





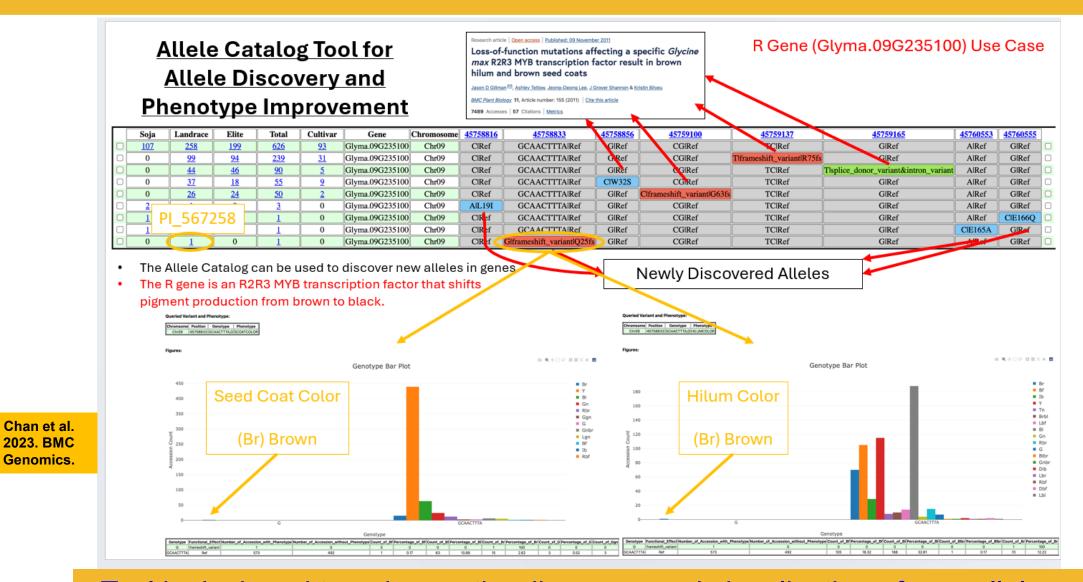
Soybean Accuracy Tool (AccuTool) for Synthetic Phenotype to Causative Mutation Analysis



AccuTool is designed to identity causative mutations by effectively evaluating GWAS hits by utilizing Soy Curated Panels







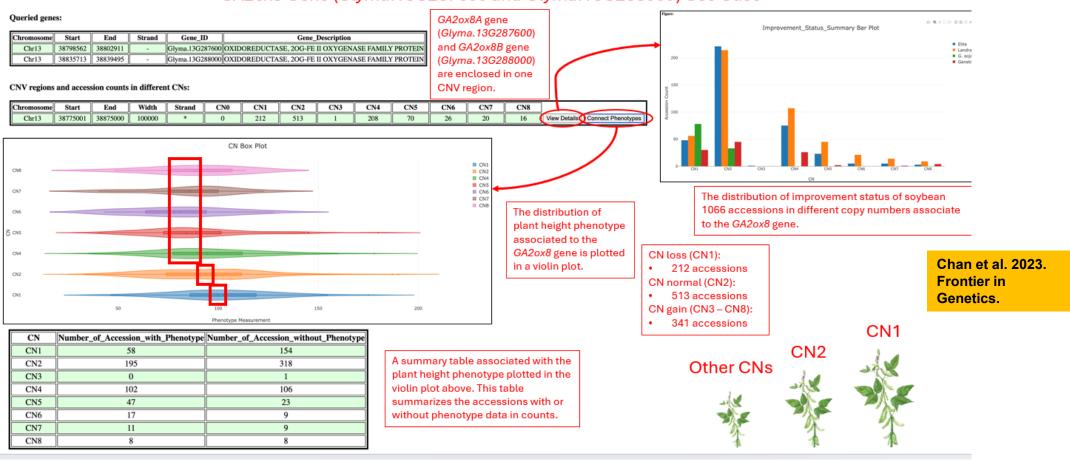
Tool is designed to enhance the discovery and visualization of new alleles and connect causative alleles to phenotypes by utilizing Soy Curated Panels





Genomic Variations Explorer (GenVarX) Use Case to Target Copy Number Variations Affecting Soybean Plant Height

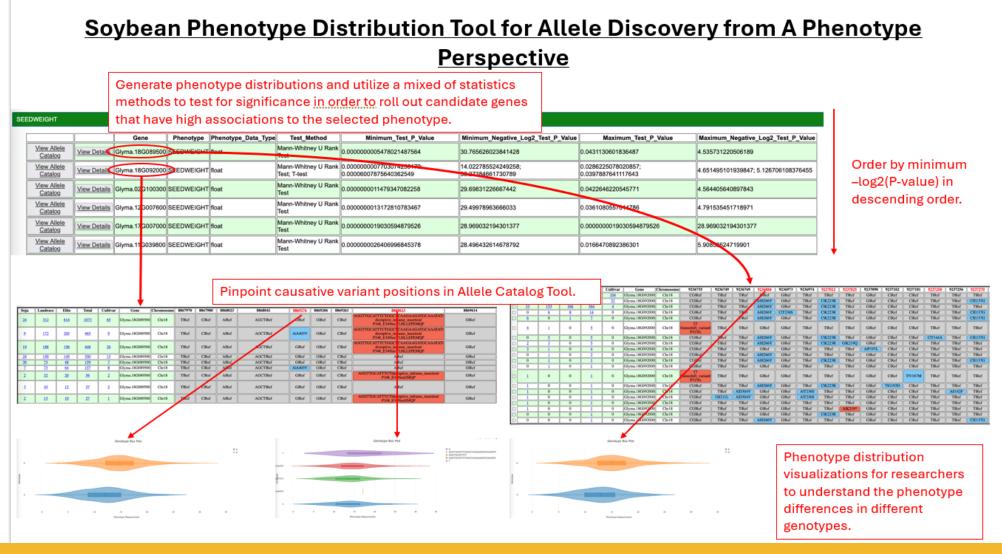
GA2ox8 Gene (Glyma.13G287600 and Glyma.13G288000) Use Case



Tool is designed to utilize Soy Curated Panels for discovery of variations in promoter regions and copy number variations (CNV) impact on phenotypes





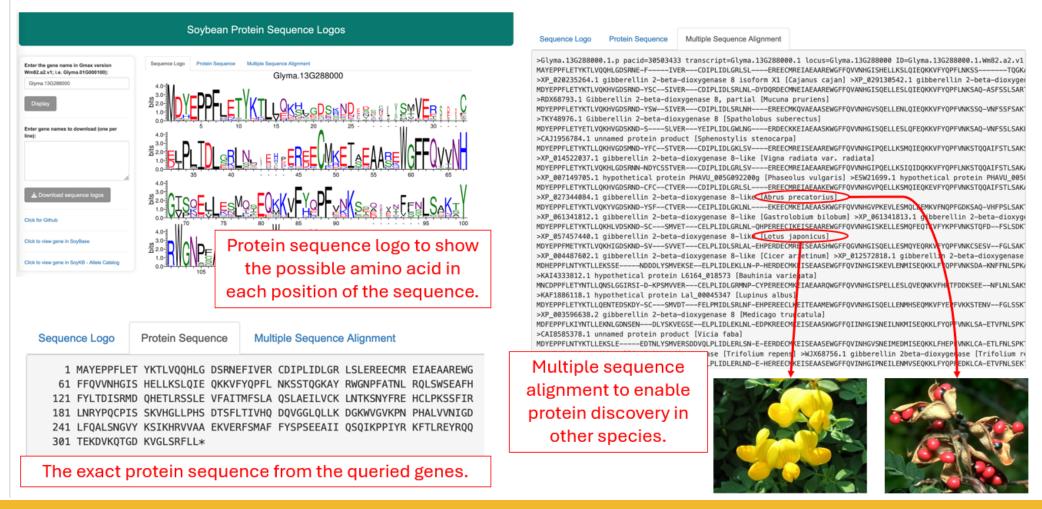


Tool is designed to easily identify the most important alleles for any given phenotype by ranking all identified novel alleles by using the Allele Catalog Tool





Soybean Protein Sequence Logos Tool for Cross Species Protein Discovery



Tool is designed to support identification and exploration of amino acid diversity in a protein across species to support biological inference of Allele Catalog results





Multiple Allele Discovery (MADis) Tool Use Case to Understand Pod Colors Variant positions in Number of variant MADis Tool Results Brown and tan positions in the Allele Catalog Tool Results 37,806,091 704 594 98.1 37,806,119 Position on Chromosome 19 (Glyma.19G120400) 37,806,160 37806091 37806119 37819253 37819382 37819390 Soja Landrace Elite Total Cultivar 37806160 37,819,253 272 288 664 117 **G|Ref** CIRef Ref_Ins GAIRef C|Ref AlRef 7 89 62 161 C|Ref **G|Ref** CTCCGGA|RI_indel Ref_Ins GAIRef **A**|Ref 37,819,382 CTCCGGA|RI_indel Ref_Ins G|K405fs 65 45 111 20 CIRef **G|Ref** AIRef 37,819,390 35 35 71 **G|Ref** CTCCGGA|RI_indel Ref_Ins **GA|Ref** TIR31C AIRef 17 18 C|Ref A|G40D CTCCGGA|RI_indel Ref_Ins **GA|Ref** A|Ref CTCCGGA|RI_indel AIL381fs **GA**|Ref GIRef C|Ref AIRef CIRef **G|Ref** CTCCGGAIRI_indel Ref_Ins **GA|Ref** AGIE402fs 264 121 **R31C** G40D L381fs K405fs E402fs G. soja REF RI_indel RI_indel + other Skrabisova et al. 2024 Frontiers in Genetics G. max

Allele distribution in Glyma. 19G120400 in Soy1066 and its correlation with different pod-colored phenotypes.

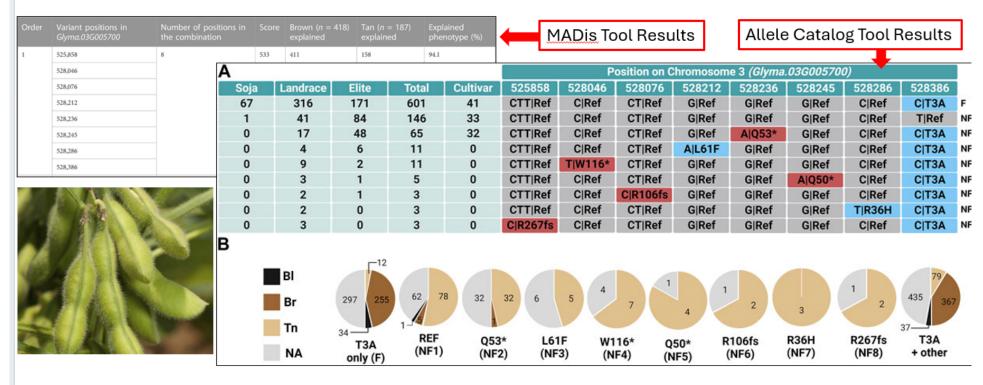
- · The RI_indel-only allele has higher chance in leading to black pod colors.
- The RI indel-only allele is crucial for the production and accumulation of pigments responsible for the black color of pods.

Tool is designed to identify multiple alleles and more importantly causative mutations using Soy Curated Panels for phenotypes of interest





Multiple Allele Discovery (MADis) Tool Use Case to Understand Pod Colors



Allele distribution in Glyma.03G005700 in Soy1066 and its correlation with different pod-colored phenotypes.

Skrabisova et al. 2024 Frontiers in Genetics

 The T3A missense mutation has higher chance leading to loss of pigment in soybean pods.

Tool computes a score for a combination of variant positions in a single candidate gene and based on the highest score, identifies the best number and combination of causative mutations.





Tools Expanded to Other Plants & Crops

- KBCommons and 3D Omics and CCMT -> All organisms
- SNPViz -> Arabidopsis, Maize, Mouse
- Allele Catalog -> Maize, Arabidopsis, Rice, Poplar
- GenVarX -> Arabidopsis, Rice
- Phenotype Distribution Tool -> Arabidopsis, Poplar
- Protein Sequence Logo Tool -> Arabidopsis, Sorghum
- MADis -> Maize





Translational Bioinformatics Resources and Al Solutions

- Multiomics Informatics Method Development
 - Al Solutions
 - G2PDeep Phenotype Prediction using Deep Learning
 - scPlantAnnotate scRNAseq annotation using LLMs



Genotype to Phenotype Deep Learning Framework (G2PDeep) for Genotype to Phenotype Prediction

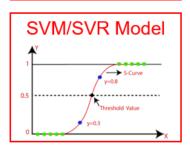
G2PDeep

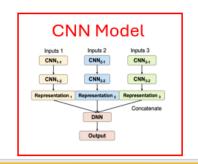
Enable Al through G2PDeep

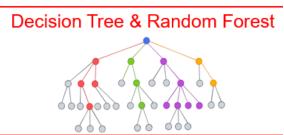
Logistic Regression

https://g2pdeep.org/

Enable AI through G2PDeep framework for training multiomics data to create different variant of models for crop phenotype predictions and discover causative mutations.







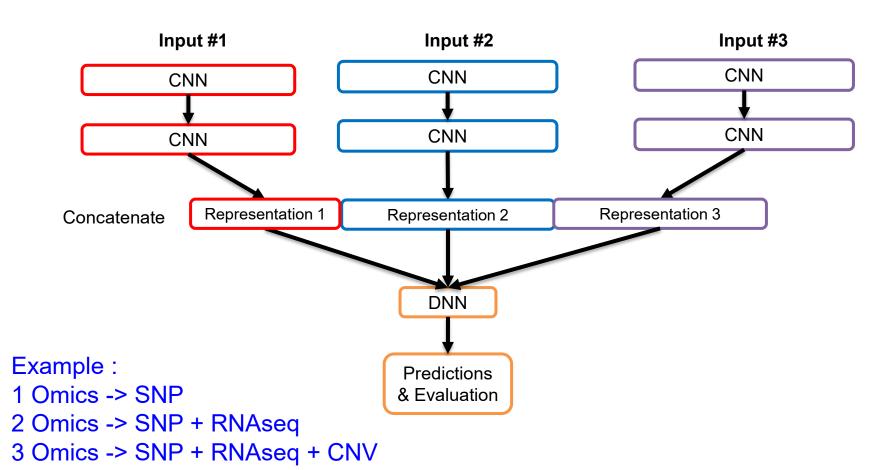
miRNA expression Gene expression **SNP** DNA methylation **Protein expression** CNV **Multi-omics data** SVM / SVR **Logistic Regression** CNN based model **Decision Tree** TensorFlow Random Forest Modeling **Basic options** Advance option **Automate Default hyperparameters Custom hyperparameters** hyperparameter tuning **Training strategy** Metrics **Training information** Zeng et al. 2021 Classification **NAR Web Server** Model details Accuracy / AUC score Learning curve Regression Zeng et al. 2024 Estimated training time MAE / R² / PCC score bioRxiv; 2025 Model analysis Bioinformatics. **Under Review** Prediction Discovery Significant biomarkers Categorical phenotype prediction Inference Quantitative phenotype prediction • **Gene Set Enrichment Analysis**

G2PDeep is a comprehensive web-based platform to do deep learning based phenotype prediction and markers discovery using any (1-3) combinations of 6 diverse multiomics data types



G2PDeep v2 – Omics Combinations

Multi CNN-based model



NAR Web Server
Zeng et al. 2024
bioRxiv; 2025
Bioinformatics,

Under Review

Zeng et al. 2021





G2PDeep Application to Soybean

- Soybean Soy1066 curated panel (32M SNPs) and GRIN Phenotypic data overlap
- Bulk transcriptomics data available from Genome Sequence Archive (GSA) database in BIG Data Center (CRA009979) for 600+ accessions
- Together can be utilized for phenotypic predictions for diverse traits

PhenoTyp ▼	Samples	
ACIMPT	586	
HILUMCOLOR	574	
MATGROUP	574	
SCOATCOLOR	574	
FLWRCOLOR	572	
PUBCOLOR	572	
<u>PUBDENSITY</u>	571	
PODCOLOR	568	
<u>SCOATLUST</u>	567	
STEMTERM	567	
<u>PUBFORM</u>	565	
LINOLEIC	480	
LINOLENIC	480	
<u>OIL</u>	480	
PROTEIN	480	
<u>OLEIC</u>	465	
PALMITIC	465	
STEARIC	465	
<u>FLOWERDATE</u>	433	
<u>SEEDQUAL</u>	433	
<u>SEEDWEIGHT</u>	433	
<u>HEIGHT</u>	438	
MATDATE	438	
LODGING	435	
YIELD	435	
<u>SHATEARLY</u>	403	
TERMSCORE	353	
NEMATOYST 03	358	
MOTTLING	345	

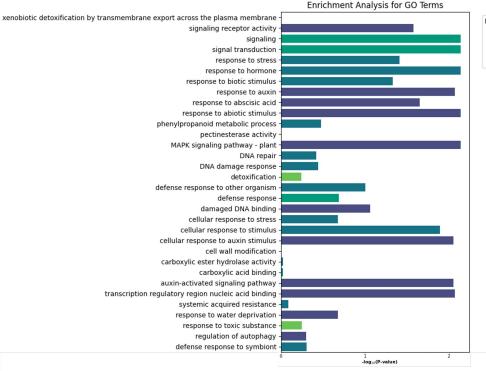




Soybean Cyst Nematode (SCN) Resistance Prediction in Soybean

Omic	Accessions	Features	•
oSNPs	268	4162197	0
CNV	268	1092	

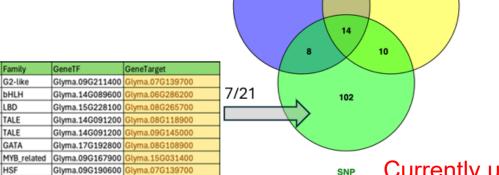
Omics	F1 (Score)	Accuracy
SNP	0.7127	0.89
CNV	0.6845	0.86
SNP CNV	0.7693	0.79





$$F_1 = rac{2}{ ext{recall}^{-1} + ext{precision}^{-1}} = 2rac{ ext{precision} \cdot ext{recall}}{ ext{precision} + ext{recall}} = rac{2 ext{tp}}{2 ext{tp} + ext{fp} + ext{fn}}$$

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Currently under evaluation

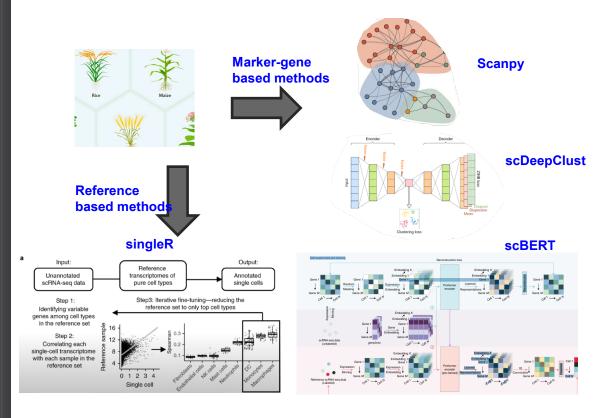
New candidate gene



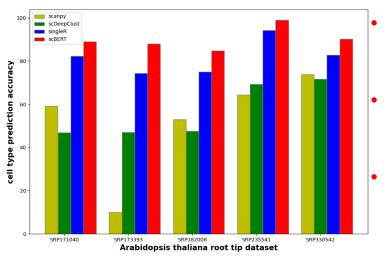


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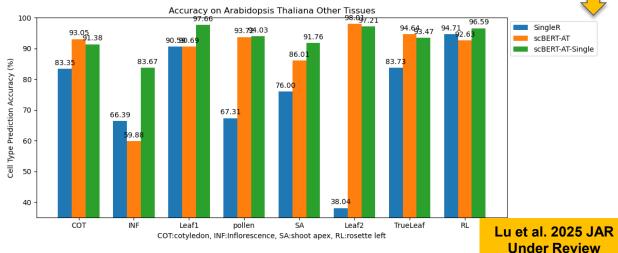
scPLantAnnotate - scRNAseq annotations using LLMs



 Currently predictions being done using scRNAseq datasets from maize, rice, soybean available in scPlantDB and from our collaborators.



- Use large language models (LLMs) for predictions.
- scBERT-AT-Single is ONE single model for Arabidopsis.
- Trained on 28 datasets across all tissues, 1.2M cells. 50 cell types.





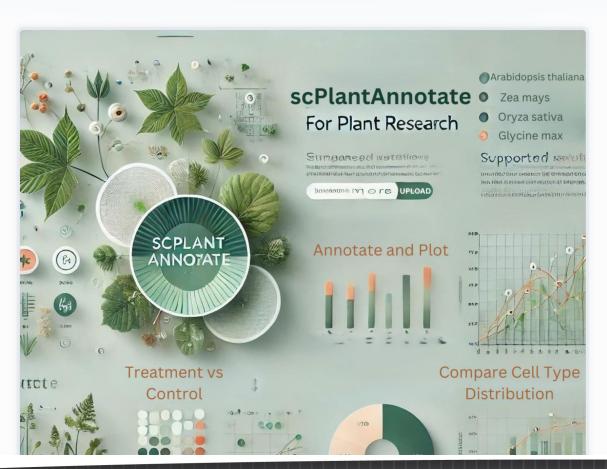




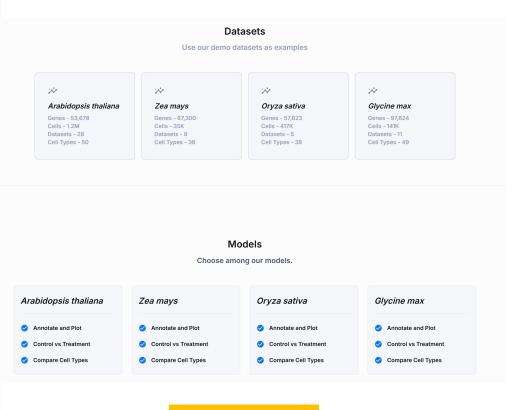
scPlant Annotate

Enhancing Plant Cell Type Annotation Through Advanced Deep Learning Approaches in scRNA-seq Data.

Learn More



https://scplantannotate.missouri.edu/



Lu et al. 2025 JAR Under Review



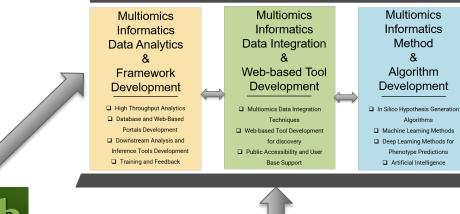
Summary

- Translational Bioinformatics resources and AI solutions provide great opportunities to incorporate genomics and multiomics information into advancing research for a diverse set of applications including crop breeding.
- As the technology and computational fields advance, it is important to adopt and incorporate these advanced techniques and build applications to facilitate easy translation.
- Making the advanced AI solutions more accessible to the users, will facilitate easier adoption and biggest impacts!



Joshi Lab

Enabling Translational Bioinformatics Research



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https://soykb.org/



KBCommons Knowledge Base

https://kbcommons.org/



Multiomics Data Integration

Allele Catalog Tool

GenVarX Tool

SNPViz Tool

AccuTool

MADis Tool



G2PDeep

https://g2pdeep.org/

IMPRes

https://impres.missouri.edu/impres

scPlantAnnotate

https://scplantannotate.missouri.edu/

scPlantAnnotate





Acknowledgements









SoyKB - KBCommons Team

- Yen On Chan (MUIDSI)
- Manish Immadi (CS)
- Sabin Dahal (CS)

Bioinformatics Analytics Team

- Mohit Verma (Postdoc)
- Sameep Dhakal (MUIDSI)
- Preethi Induri (Senior Research Specialist II)

Method Development Team

- Mohammad Vahed (Scientist)
- Zhen Lyu (CS)
- Chunyang Lu (CS)



Shuai Zeng

Key Collaborators

Dr. Dong Xu (EECS)

USB Soybean Project Collaborators

- Dr. Kristin Bilyeu
- Dr. Maria Skrabisova
- Jana Biova
- Anser Mahmood (Plant Sci)

Multiomics Plant Project Collaborators

- Dr. Ron Mittler
- Dr. Ed Cahoon
- Dr. Marc Libault





Acknowledgements





















https://soykb.org/

KBCommons Knowledge Base



https://kbcommons.org/





https://g2pdeep.org/

scPlantAnnotate

https://scplantannotate.missouri.edu/





G2PDeep

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