Using a Locally Connected Autoencoder to Identify Candidate Alleles in *Sorghum bicolor*

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Abstract:

Motivation: By isolating Single Nucleotide Polymorphisms (SNPs) thought to be correlated to a phenotype, further research can be done to better target genomes to potentially eradicate diseases or alter certain properties in organisms.
Limitations: **(1)** Allele panels have extremely high dimensionality **(2)** Individuals cluster into sub-populations **(3)** Current approaches suffer from spurious false positives
Objectives: **(1)** Use an unsupervised approach to extract features in the allele panels **(2)** Find encoding dimensions correlated to phenotypes **(3)** Isolate candidate alleles for further investigation

Dataset: *Sorghum Bicolor* Allele Panels

- Allele panels contained 345 accessions with 232,303 SNPs for each accession [1]. Only 125,980 SNPs were used.

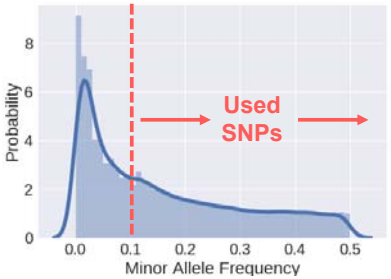


Figure 1: Minor allele frequencies for all 232,303 SNPs

- The *Sorghum bicolor* originated from 37 locations with 49.8% of individuals originating from only 5 locations

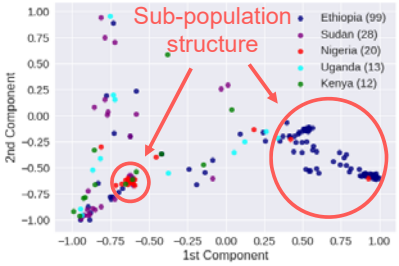
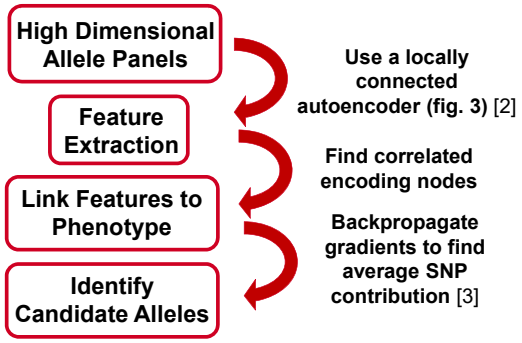


Figure 2: The 1st and 2nd normalized principal components for individuals originating from the 5 most common regions

Methodology:



Locally Connected Autoencoder:

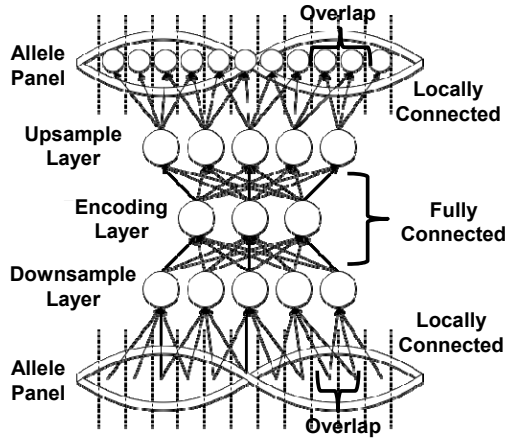


Figure 3: Summarized locally connected autoencoder topology used for feature extraction

Results:

- Correlations were found for fresh weight, stalk height, lignin percent and anthesis date
- **S1_18096891**, **S2_2633689**, **S2_2793224**, **S6_7123769** , and **S6_61098274** were identified as candidate alleles for fresh weight and stalk height

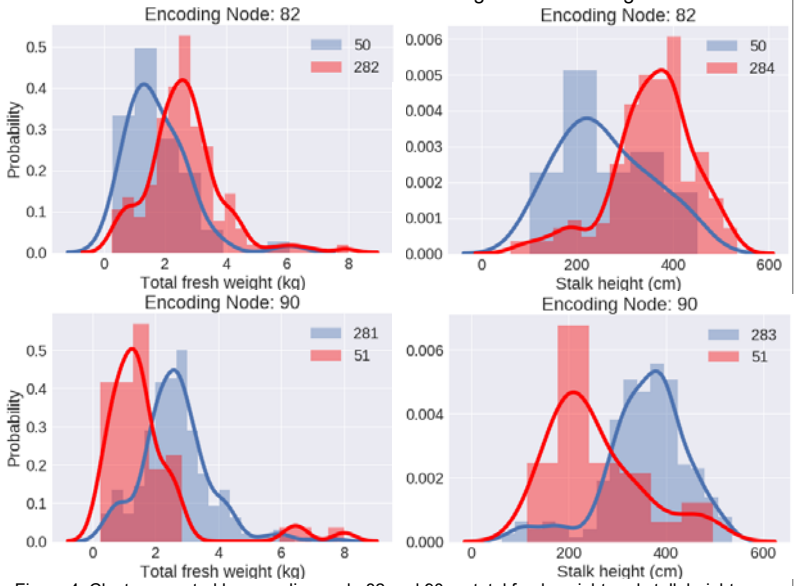


Figure 4: Clusters created by encoding node 82 and 90 on total fresh weight and stalk height data. The individuals in each cluster can be seen in the legend.

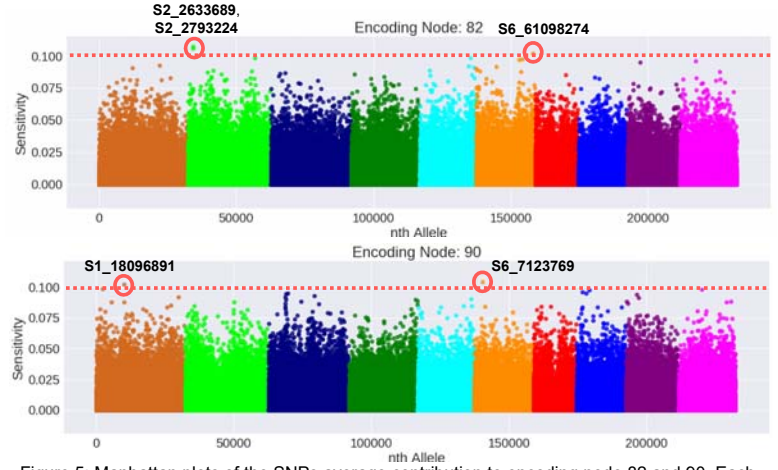


Figure 5: Manhattan plots of the SNPs average contribution to encoding node 82 and 90. Each color corresponds to a new chromosome.

Future Works:

- Test on other allele panels
- Better handle SNPS with highly skewed minor allele frequencies
- Validate candidate alleles through image data
- Confirm candidate alleles through additional biological research

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References:

[1] Z. W. Brenton, "A genomic resource for the development, improvement, and exploitation of sorghum for bioenergy," Genetics, vol. 204, no. 1, pp. 21–33, 2016.
[2] Q. V. Le, "Building high-level features using large scale unsupervised learning," in Acoustics, Speech and Signal Processing (ICASSP), 2013 IEEE International Conference on. IEEE, 2013, pp. 8595–8598.
[3] I. Dimopoulos, "Neural network models to study relationships between lead concentration in grasses and permanent urban descriptors in athens city (greece)," Ecological modelling, vol. 120, no. 2, pp. 157–165, 1999.