Genotype to Phenotype Mapping via Deep Learning

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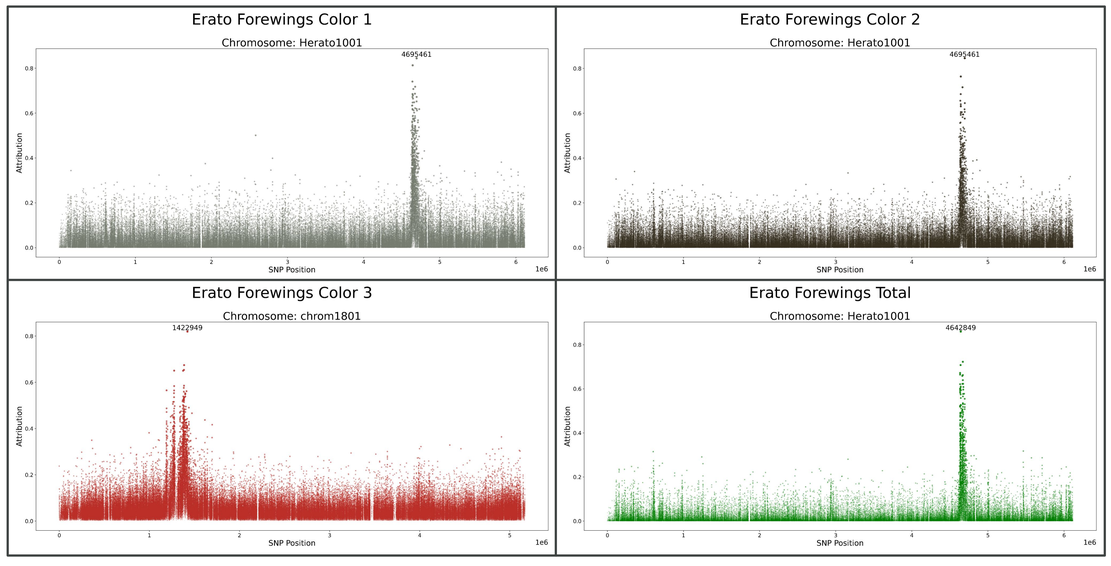
Abstract

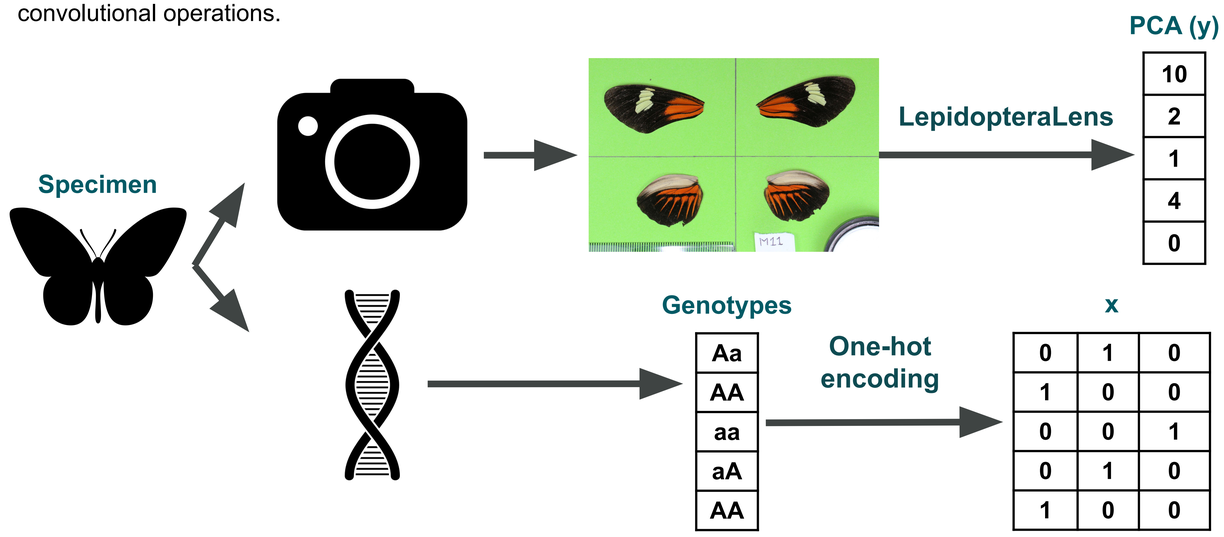
- We aim to **identify genes and mutations associated with phenotypic variation** measured in Heliconius butterfly images.
- We **train a convolutional neural network** to predict phenotypic variation from genotypes.
- Given the trained network **we apply saliency methods** to highlight which genes the neural network used to make its prediction.
- Our findings **identify existing genes** responsible for color pattern variations previously discovered by GWAS.
- Our future work aims to improve modeling, use deep learning features as phenotypes, address missing heritability, and uncover epistasis behavior.

Results



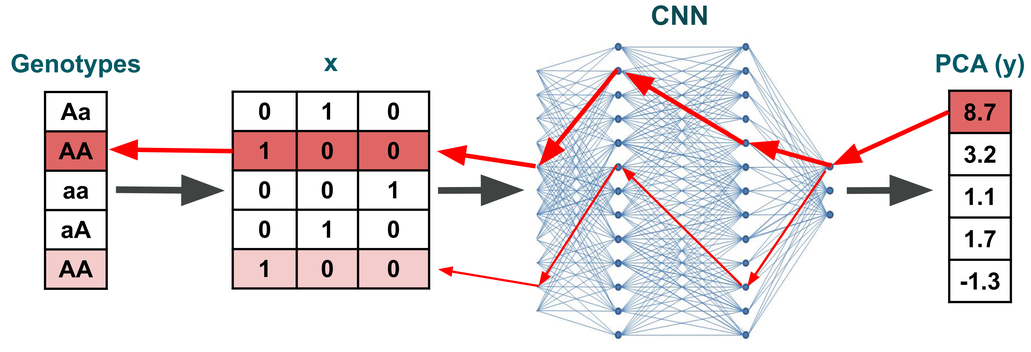
Methods (Data)

- Each butterfly specimen is **imaged and sequenced** into a format compatible for machine learning training.
- **Images** are sent through **Patternize** to obtain PCA values associated with a targeted phenotype (e.g. red color variation).
- The **DNA sequence** of each butterfly is processed to obtain genotypes and then **one-hot encoded** to allow for convolutional operations.



Methods (Modeling)

- A **convolutional neural network** based on **SoybeanNet** is trained. Early stopping with a held out validation set is incorporated to prevent overfitting.
- We then apply **Guided Grad-CAM** to the predicted output across a held out testing set and visualize the aggregated importance of each gene.



References

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