Leveraging VAE to Dissect Inter-individual Variation

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Introduction

Background:

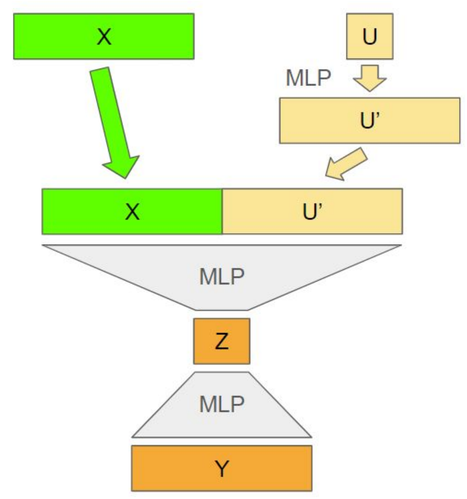
- Brain-related diseases are affected by genetic (G) and environmental (E), which can both be reflected in gene expression
- We aim to identify the latent factors associated with inter-individual variation in gene expression data
- There are, however, many different latent factor analysis frameworks, principal component analysis (PCA), independent component analysis (ICA), variational autoencoders (VAE), etc.

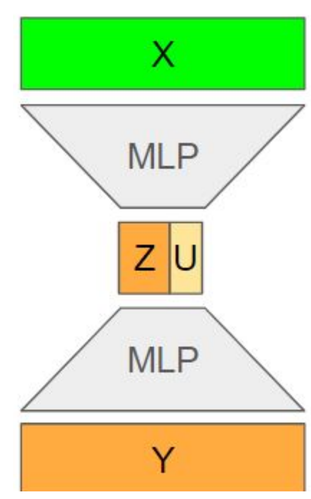
This Study:

- This study focuses on the capabilities of VAE to find latent factors
- We hypothesize that adding prior knowledge, e.g., age, sex, and smoking history, will improve VAE performance, resulting in the identification of biological factors related to brain diseases
- We look into flexible frameworks, such as identifiable VAE (iVAE), that allow the integration of prior knowledge

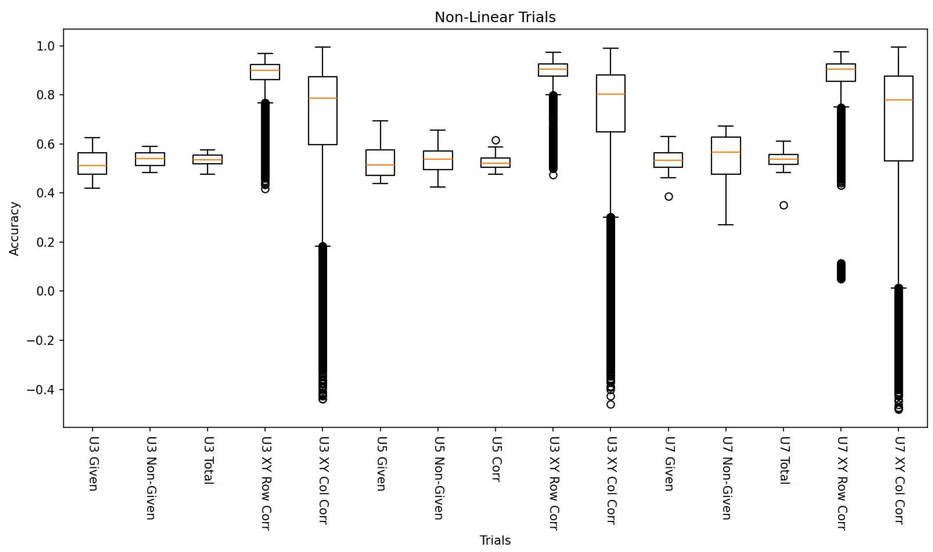
Methodology

- For now, research has mainly been done on simulated data, where the latent factors are already known
- The simulated data was fed through different VAE frameworks that incorporated prior knowledge
- 20 Trials were done for each framework
- Accuracies of latent layer (Z) predictions were compared to the known latent factors
- Additionally, row and column accuracies of the result of the VAE (Y) were compared to the known input data set (X)



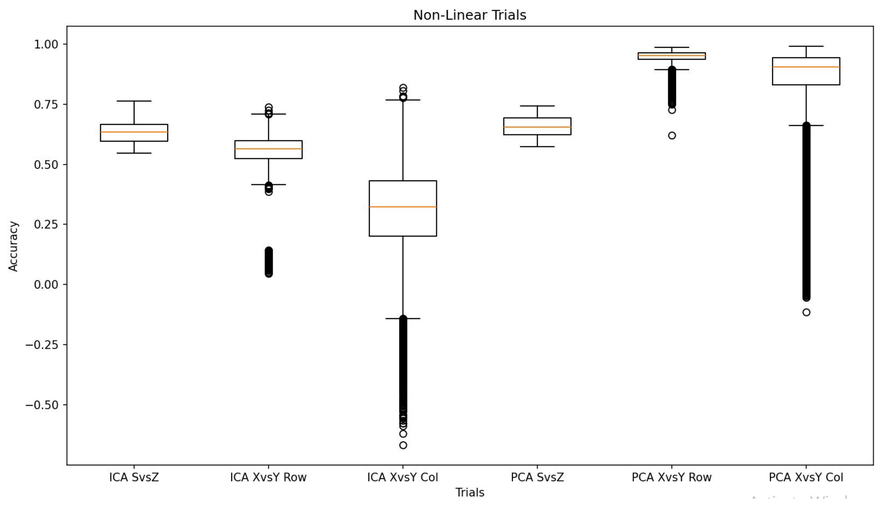


Results



VAE Trials

ICA and PCA Trials



Discussion

- The encoders of the VAE models we tested were found to have similar results to PCA
- But the encoders performed significantly better than ICA
- Unexpectedly, we found giving more prior knowledge did not result in better performance
- This could be due to the simplicity of our simulation data, which was generated through a 3-layer neural network

Future Directions

Further research will be conducted on more complex data to determine the effectiveness of our VAE networks in identifying inter-individual variation in real biological scenarios. Complex data includes simulated data from deeper neural networks and real biological gene expression data.

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