Neural Networks in Phenotype Mapping

The applications of machine learning in genetic diagnostics

The premise of machine learning can bring great benefits and opportunities for the field of rare genetic disorders. From accounting for thousands of features to unsupervised anomaly detection for novel disorders, machine learning leverages pattern identification to make conclusions amongst different phenotypic information and map it back to genotypes. For the 8,000 rare multigenic disorders and the thousands of data points, machine learning can make conclusions and find interesting trends that can help us learn and better treat patients.

Neural Networks & Analysis

Utilizing a wide variety of phenotypic information to map genotypic information. Improving a pre-trained database with analysis and data-driven insights from model predictions.

Generated data analysis -> correspond patient information from a variety of phenotypic information
Because machine learning is able to find trends over a given period of time, models are more accurate at identifying genetic complications by taking into account rules the model learned from experience.

Supervised with unsupervised -> evolving from predictions to anomaly detection for novel disorders
Supervised learning can be a powerful tool in diagnosis and can be paired with unsupervised methods to identify a wider range of genetic disorders. This can be especially critical in identifying unique genotypic information all from just phenotypic inferences.

Research and Pattern Identification -> discovering new trends over more complex feature sets
Machine learning algorithms can help drive research progress in identifying and understand the complex interactions between genotype mutations and phenotypic responses. For cystic fibrosis, data-driven analysis has revealed an undiscovered correlation between ferric iron, blood cell concentration and the CFTR gene.

Sensitivity to change and mutations -> future proofing and opportunity in personalized medicine
Only 5% of rare genetic disorders have cures and current DNA sequencing methods are not sufficient for developing directed medicine. AI can help understand the complex interactions of genes and drive research to identify other phenotypic medicine possibilities targeted to the individual's genotype info.

PROJECT PHENO