Cloud-Based Ocular Disease Diagnosis
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Background

Problem
- Around 400 patients are diagnosed with a genetic retinal dystrophy annually.
- The process can become expensive and time consuming due to blood tests. It is very difficult to precisely diagnose rare cases of genetic retinal dystrophies due to hundreds of possible causes.
- Specialized eye hospitals throughout the world have isolated patient data.

Solution
- Create a webapp as a tool to assist with the diagnosis of genetic retinal dystrophies.

Stakeholder Objectives

KEC: Have an accurate cloud-based web app to diagnose retinal diseases. Build a large patient database and maintain patient privacy. Improve machine learning models.

Partner Hospitals: Have access to patient database and maintain patient confidentiality. Conduct further research on retinal diseases.


Engineering Requirements

Machine Learning: Construct predictive model with >70% accuracy for predicting the gene causing a patient’s dystrophy.

Inheritance Pattern: Construct predictive model with >75% accuracy for predicting the inheritance pattern causing a patient’s dystrophy.

Frontend: Build a user-friendly web app that clinicians trust and use.

Backend: Build a web app backend that services user requests for webpages and data in <1 second.

HIPAA: Create a web app that securely manages sensitive patient data in accordance with the Health Insurance Portability and Accountability Act.

Data Analysis: Explore statistical correlations between a patient’s dystrophy and other characteristic features, such as hearing loss.

Machine Learning Prediction Model

Inheritance Pattern Prediction
- We asked a genetic specialist to come up with questions that would help us identify the inheritance pattern causing a patient’s dystrophy. Then we answered those questions based on the patient’s pedigree and used those answers to build our model. Then we used the output as features to feed into the gene prediction model.

Mutated Gene Prediction
- The model is built on top of last year’s model, which was trained on 152 patient data points.

Validation Methodologies

Predictive Model
- 5 - fold cross validation on 50 iterations:
  - Accuracy for Mutated Gene Prediction
    - Model: SVM
    - Naive Guess: 40.09
    - Previous Team: ~67
    - SVM: 80.15

Inference for Accuracy Pattern Prediction
- KEC:
  - Model: SVM
  - Naive Guess: 40.09
  - Previous Team: ~67
  - SVM: 80.15

Web Prototype

Web App
- Database Latency: Database access time = 4s
- Browser Page Load Time: Page load times <= 1s

UI/UX
- Trust of website: Single Ease Question Average = 85%
- Intuitive User Interface: Task Completion Rate = 85%
- Usability testing: Number of Errors/Task = 0.5

Future Work
- Collect more clean data on patients and their family history to better train the machine learning model.
- Deploy a robust web application that is:
  - Secure: The server is safeguarded against common web attacks.
  - Reliable: The patient information form saves entered data at various points of completion.
  - HIPAA-compliant: Stored patient data does not contain PHI, and the database aligns with HIPAA guidelines.

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