



# Rare Disease Diagnosis Assistant

A Machine Learning-based assistant to support doctors in diagnosing **rare diseases** based on patient-reported symptoms.



## Project Overview

Rare diseases often present with vague or overlapping symptoms, making accurate diagnosis a challenge for healthcare providers.

This project aims to **support medical professionals** by building a predictive model that uses **binary symptoms (0/1)** as input and suggests a possible disease class.

The system doesn't replace a doctor — it acts as a **decision-support tool**, highlighting conditions with low confidence and encouraging human verification when needed.



## Technologies Used

- **Python**
- **Pandas / NumPy / Matplotlib / Seaborn**
- **Scikit-learn:** RandomForestClassifier, train\_test\_split, classification\_report
- **Machine Learning:** Supervised classification
- **Performance Metrics:** Accuracy, Precision, Recall, F1-score
- **Explainability Logic:** Warnings for diseases with low data representation



## Dataset

- **Input:** Binary symptom indicators (0 = no, 1 = yes)
- **Output:** Disease label (rare or common)
- **Format:** CSV with features representing symptoms and a target column diseases

- Data set link:  
<https://www.kaggle.com/datasets/dhivyeshrk/diseases-and-symptoms-dataset>



**Note:** The dataset was cleaned and verified to only contain binary values in features.

## Model Workflow

1. **Data Cleaning & Preprocessing**
2. **Train/Test Split**
3. **Model Training** using RandomForestClassifier
4. **Prediction on unseen data**
5. **Overfitting Check** via comparison of training/testing accuracy
6. **Evaluation Metrics**
7. **Custom Logic for Confidence Messages:**
  - a. If **F1-score** < **0.6** or sample count is low → model warns the doctor
  - b. If metrics are reliable → model confirms prediction reliability

## Doctor Guidance Messages

The model prints messages like:

-  "You can rely on this result. The model performed well and was trained on enough data."
-  "Doctor, please verify this result by yourself, due to low sample count."

## Future Enhancements

- Add **Neural Network** alternative model
- Integrate with medical APIs for validation
- Improve explainability via SHAP / LIME
- UI for non-technical medical users



## Disclaimer

It must **not** be used for real-life diagnosis without medical supervision.

Always consult a licensed physician for actual medical decision-making.



## Author

**Basma Sameh**

AI & Data Science Student

GitHub: [basmasameh84]