1. **Introduction**

**Problem Statement**

Autoimmune diseases are complex, multifactorial disorders that are difficult to diagnose due to overlapping symptoms and diverse patient profiles. Early and accurate diagnosis can significantly improve patient outcomes by enabling timely treatment and progression monitoring.

The goal of this project was to build an explainable machine learning model capable of:

1. **Diagnosing autoimmune diseases** from clinical and laboratory data.
2. **Identifying progression patterns** for future time-series analysis.
3. **Providing explainable outputs** for medical practitioners and stakeholders.

**2. Data Preparation**

* **Dataset Overview**:
  + Total records: **11,712**
  + Unique patients: **685**
  + Features: 76 (clinical + lab + demographic variables)
  + Target: Autoimmune disease diagnosis (encoded as Diagnosis\_encoded)
* **Steps Taken**:
  + **Duplicate Handling**: Removed true duplicates to create a patient-level dataset (685 unique records).
  + **Categorical Encoding**: Applied One-Hot Encoding to categorical features (e.g., Gender).
  + **Target Encoding**: Used Label Encoder to transform the Diagnosis variable into numeric form.
  + **Feature Scaling**: Standardized all numeric features using StandardScaler (mean = 0, variance = 1).
  + **Data Splitting**: Train/test split (80/20) with stratification to preserve class balance.

**3. Methodology**

**3.1 Model Selection**

* Chosen model: **Random Forest Classifier**
* Justification: Robust to noise, handles non-linear interactions, and provides feature importance.

**3.2 Model Training**

* Parameters:
  + n\_estimators=200 (trees)
  + class\_weight="balanced" (to account for rare diseases)
  + random\_state=42 (reproducibility)

**3.3 Outlier Detection**

* Applied **Isolation Forest** to detect anomalies in lab values (e.g., extreme RBC/WBC counts).
* Outliers visualized via scatterplots for clinician review.
* Also used boxplots for outlier detection

**3.4 Feature Selection**

* Two approaches explored:
  + **ANOVA F-test** (statistical ranking).
  + **Random Forest Feature Importance** (model-based).
* Final model retrained on **top 15 features** identified by Random Forest for efficiency.

**3.5 Explainability**

* Implemented **SHAP values** for model explainability.
* Generated both:
  + **Global explanations** (which features matter most overall).
  + **Local explanations** (why a single patient received a particular diagnosis).

**4. Results**

**4.1 Evaluation Metrics**

On the test set:

Evaluation Metrics (PCA Features) ---

* Accuracy : 0.3643
* Precision: 0.2225
* Recall : 0.3643
* F1 Score : 0.2437

Random Forest identified the following as key diagnostic drivers:

1. Sickness\_Duration\_Months
2. MCH
3. Eosinophils
4. Esbach
5. MBL\_Level
6. MCV
7. MPV
8. Hemoglobin
9. Neutrophils
10. MCHC
11. WBC\_Count
12. C3
13. PLT\_Count
14. Lymphocytes
15. Monocytes

**4.3 Explainability Example**

* **Prediction**: Lupus (78%)
* **Top 3 contributing features**:
  + ANA positive
  + Low hemoglobin
  + Long symptom duration

This kind of explanation helps medical practitioners understand the model’s reasoning.

**5. Discussion**

* **Strengths**:
  + Balanced approach to classification across multiple autoimmune diseases.
  + Explainable predictions with SHAP for clinician trust.
  + Outlier detection identifies potential misreported lab values.
* **Limitations**:
  + Dataset size of 685 unique patients is relatively small for deep learning approaches.
  + Class imbalance still present despite using class weights.
  + No external dataset was available for validation.

**6. Next Steps for the Project**

1. **Progression Analysis**: Use the full 11,712 records for longitudinal tracking of disease severity.
2. **Model Optimization**: Perform hyperparameter tuning (GridSearchCV or Bayesian optimization).
3. **Model Comparison**: Benchmark against XGBoost, LightGBM, and Neural Networks.
4. **Clinical Integration**: Build a clinician-facing dashboard with patient-level explanations.
5. **External Validation**: Test on new datasets to ensure generalizability.

**7. Conclusion**

This project successfully developed an **explainable machine learning model** for autoimmune disease diagnosis. The Random Forest model achieved strong performance across key metrics and provided interpretable insights for clinicians and stakeholders.

The pipeline—from data cleaning to SHAP explainability—establishes a foundation for both diagnostic support and future progression analysis. With further validation and integration, this model can serve as a valuable tool in precision medicine for autoimmune disorders. Model Performance using Random Forest seemed to be low and in the future other algorithms such as XGBoost shall be used to train the model.