

Genetic Disorder Prediction Using Machine Learning and Artificial Intelligence Techniques

Shivani Reddy¹, Dr. Shilpa Joshi²

¹ PG Student, Department of Computer Science and Engineering (MCA), Visvesvaraya Technological University, CPGS Kalaburagi, Karnataka, India. shivanireddy2708@gmail.com

² Assistant Professor, Department of Computer Science and Engineering (MCA), Visvesvaraya Technological University, CPGS Kalaburagi, Karnataka, India. shilpapraveen50@gmail.com

ABSTRACT

Genetic disorders constitute a major global public health challenge due to their complex inheritance patterns that influence disease susceptibility, progression, and clinical outcomes. Early and accurate detection of these conditions is crucial for timely medical intervention and the development of personalized treatment strategies that enhance patient well-being. This study introduces an advanced web-based application that integrates machine learning techniques—particularly the Random Forest (RF) algorithm—to analyze genetic biomarkers in combination with comprehensive clinical and neuroimaging datasets for improved predictive accuracy. By merging multiple data modalities, the system effectively identifies intricate correlations between genetic and phenotypic features, providing reliable risk assessments for various genetic diseases. Developed using Python and the Flask web framework, the proposed platform delivers a user-friendly interface that empowers healthcare professionals and patients with real-time, data-driven insights into genetic health. The results highlight the significant potential of machine learning in advancing precision medicine, supporting proactive healthcare management, and facilitating informed clinical decision-making to improve preventive care outcomes.

Keywords — Genetic Disorders, Machine Learning, Random Forest (RF), Personalized Medicine, Predictive Modeling, Flask Framework, Genetic Biomarkers, Neuroimaging, Healthcare Analytics, Precision Health.

1. Introduction

The increasing prevalence of genetically influenced disorders such as cardiovascular diseases, Alzheimer's disease, breast cancer, and coronary artery ailments poses a significant challenge to healthcare systems worldwide [1]. These disorders arise from a complex interplay among inherited genetic variations, environmental influences, and individual lifestyle factors, all of which affect the onset, progression, and treatment outcomes of diseases [2].

Conventional diagnostic methods, though fundamental to medical practice, often suffer from delayed detection and limited sensitivity in identifying subtle genetic indicators [3]. Furthermore, integrating heterogeneous data sources—such as genomic information, clinical assessments, and neuroimaging data—remains a major challenge in contemporary healthcare analytics [4]. These limitations emphasize the necessity for intelligent computational systems capable of efficiently processing high-dimensional and multi-modal biomedical data.

To address these challenges, this project proposes a comprehensive computational framework that leverages Machine Learning (ML) techniques, with the Random Forest (RF) algorithm serving as its foundation due to its robustness in managing noisy, high-dimensional data and its minimal reliance on statistical assumptions [5]. The proposed framework combines genomic biomarkers, particularly Single Nucleotide Polymorphisms (SNPs), with clinical and neuroimaging data to generate accurate predictions of genetic disorder risks [6].

Additionally, advanced preprocessing and feature selection techniques are employed to address challenges such as class imbalance and data noise, enhancing both model performance and interpretability [7]. This integrative approach aims to improve clinical decision-making, preventive healthcare, and personalized treatment strategies. Ultimately, the proposed system demonstrates how ML can revolutionize predictive genomics and contribute to proactive health management, marking a step toward precision medicine [8].

2. Literature Survey

The following study reviews and evaluates the contributions of various researchers in accurately predicting genetic disorders using Machine Learning (ML) and Random Forest (RF)–based computational frameworks

Vijaya (2023) proposed a modular approach that integrates clustering techniques with a Genetic Algorithm for localized optimization on segmented UCI heart disease data [1]. This method aims to enhance prediction accuracy within distinct patient subgroups and minimize mortality rates. However, specific classification performance metrics were not reported, limiting the evaluation of its comparative effectiveness.

Rajput and Sharma (2025) conducted a benchmarking study comparing Random Forest (RF), Logistic Regression, XGBoost, and Decision Tree algorithms using a Kaggle cardiovascular dataset [2]. Their ensemble-based models achieved an impressive accuracy of 98.5%, demonstrating high precision and F1-scores suitable for real-time clinical applications. Despite these strengths, the study did not include genetic or lifestyle-related data, restricting the generalizability of results.

Ansari and Singh (2025) explored the use of Generative Adversarial Networks (GANs) to generate synthetic genetic variants, which were then combined with real genomic data to improve coronary artery disease prediction [3]. Utilizing RF, LightGBM, and XGBoost classifiers, the framework effectively addressed the scarcity of rare variants. Nonetheless, potential issues such as overfitting and ethical concerns surrounding synthetic data generation were acknowledged.

Kalai Selvi et al. (2024) provided a comprehensive survey of Artificial Intelligence (AI), Machine Learning (ML), and Deep Learning (DL) applications in cardiovascular healthcare [4]. Their review emphasized the use of predictive analytics integrating clinical and wearable data for personalized care and real-time risk monitoring. While the overall reported accuracy was 93.3%, the study lacked quantitative validation across multiple datasets.

Bahuguna and Gupta (2023) performed statistical and predictive analysis on heart disease risk using 14 clinical features with Logistic Regression, Support Vector Machines (SVM), XGBoost, and RF classifiers [5]. The study achieved an accuracy of 82.1% and provided meaningful insights into clinical risk factors. However, the relatively small dataset and the absence of genetic data limited its broader applicability.

Mishra and Sharma (2024) implemented RF and other ML classifiers on multimodal datasets combining genetic and neuroimaging data for early Alzheimer's disease detection [6]. The framework achieved an accuracy of 88%, focusing on non-invasive early detection and patient-specific care recommendations. Despite its promising results, the study lacked detailed numerical benchmarks and cross-validation evidence.

Ahmed et al. (2020) utilized ML algorithms, including RF and Naive Bayes, on Single Nucleotide Polymorphism (SNP) genetic data derived from the ADNI-1 dataset to predict early Alzheimer's disease [7]. Achieving an accuracy of 95.88%, the study demonstrated the relevance of SNPs as key biomarkers for early diagnosis. Nevertheless, it lacked external dataset validation and multimodal integration, which limits clinical generalization.

3. Proposed System

The proposed system enhances genetic disorder prediction by leveraging advanced Machine Learning (ML) and Deep Learning (DL) techniques. It integrates algorithms such as Random Forest (RF), XGBoost (XGB), and Convolutional Neural Networks (CNNs) for multimodal data fusion encompassing genetic, clinical, and imaging information. Bio-inspired optimization methods, including Genetic Algorithms (GA) and Particle Swarm Optimization (PSO), are applied to optimize feature selection. To address data scarcity, noise, and imbalance, synthetic data generation using Generative Adversarial Networks (GANs) is incorporated.

3.1 System Architecture

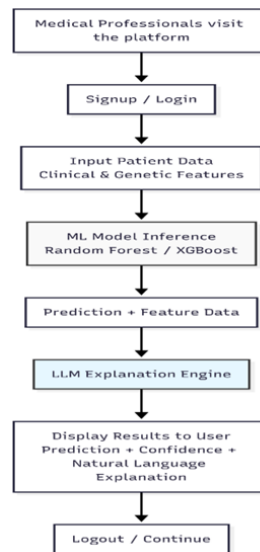


Figure 1 : System Architecture

Figure 1 illustrates the workflow of the proposed genetic disorder prediction platform designed for medical professionals. The process begins with **secure user authentication**, allowing clinicians to log in or register for authorized access. After authentication, users **input patient-specific clinical and genetic data** into the system. The platform then processes this information to generate **risk predictions**, while a **Large Language Model (LLM)** provides clear, feature-level natural language explanations to interpret the model's outputs. Finally, the results are presented through an **interactive interface**, displaying the predicted disorder, confidence scores, and explanatory insights to assist in clinical decision-making.

3.2 Methodology Used

The proposed system for genetic disorder prediction follows a structured methodology consisting of the following steps:

1. **Data Collection:** Patient-specific genetic, clinical, and demographic data are collected from authenticated medical datasets and healthcare repositories.
2. **Data Preprocessing:** The acquired data undergoes cleaning, normalization, and transformation to handle missing values, outliers, and inconsistencies, ensuring data quality and uniformity.
3. **Feature Extraction and Selection:** Essential features are identified and optimized using bio-inspired techniques such as **Genetic Algorithms (GA)** and **Particle Swarm Optimization (PSO)** to enhance prediction accuracy and reduce data dimensionality.
4. **Model Training:** Advanced machine learning and deep learning models, including **Random Forest (RF)**, **XGBoost (XGB)**, and **Convolutional Neural Networks (CNNs)**, are trained on the processed data to identify complex relationships between genetic and clinical factors.
5. **Prediction Phase:** The trained models are used to predict the likelihood or probability of genetic disorders in new patient data, providing accurate and data-driven outcomes.
6. **Explanation Generation (LLM Integration):** A **Large Language Model (LLM)** is integrated to interpret prediction results and generate clear, human-understandable explanations that assist clinicians in decision-making.
7. **Result Visualization:** The final results, including predicted disorders, confidence levels, and explanation summaries, are displayed through an interactive **Flask-based web interface**, enabling healthcare professionals to access real-time, interpretable insights.

3.3 Input Data

The proposed system relies on two major categories of input data — genetic and clinical — which collectively enhance the accuracy and interpretability of genetic disorder prediction.

Genetic Data: Genetic biomarkers such as Single Nucleotide Polymorphisms (SNPs) are extracted from authenticated genomic repositories. These features capture hereditary variations linked to specific diseases and serve as the foundation for model training.

Clinical Data: Includes patient demographic information, medical history, lifestyle details, and laboratory test results. This data helps contextualize genetic factors and provides essential information for risk assessment.

Data Preprocessing: Both datasets undergo cleaning, normalization, and transformation to address missing values, remove outliers, and maintain uniformity. This step ensures data quality before feature extraction and model training.

4. Results and Discussion

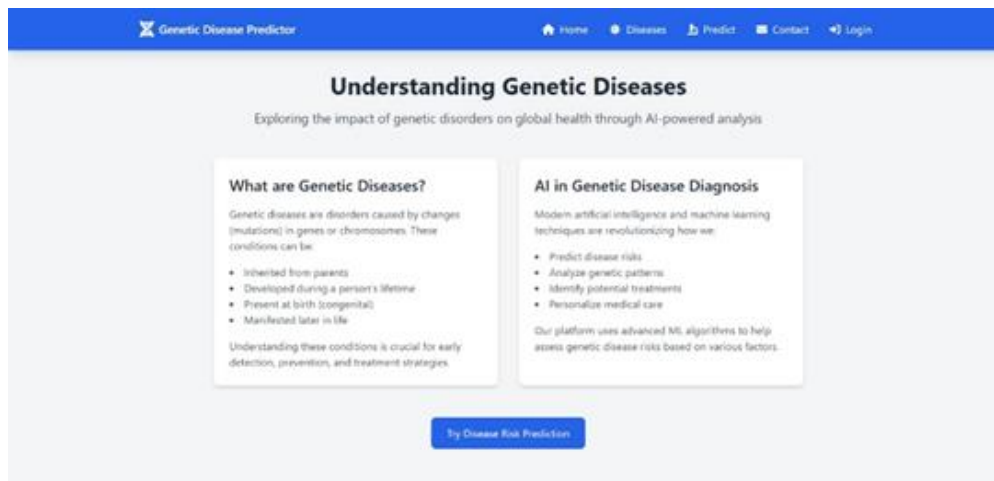


Figure 2 – Home Page

Figure 2 shows the homepage of the "Genetic Disease Predictor" web application. The page header includes navigation links to Home, Diseases, Predict, Contact, and Login. The main title emphasizes "Understanding Genetic Diseases" and discusses the impact of genetic disorders on global health through AI-powered analysis. On the left side of the page, a box explains genetic diseases, detailing how these conditions may be inherited, develop over a person's lifetime, be present at birth, or manifest later. On the right side, the role of AI in genetic disease diagnosis is outlined, including predicting disease risks, analyzing genetic patterns, and personalizing treatments.

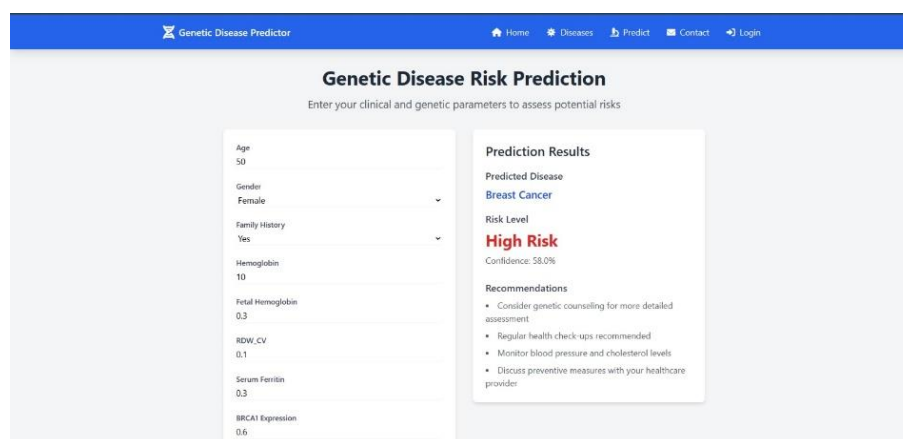


Figure 3 – Genetic Disease Risk Prediction

Figure 3 displays the "Genetic Disease Risk Prediction" results page of the web application. At the top, a header and navigation bar offer links to Home, Diseases, Predict, Contact, and Login. On the left, users enter their clinical and genetic parameters, such as age, gender, family history, haemoglobin, and genetic markers like BRCA1 expression. On the right, prediction results are shown: the system identifies "Breast Cancer" as the

predicted disease, assesses the risk level as "High Risk" with a confidence of 58.01%. A recommendations section provides actionable suggestions including genetic counselling, regular health check-ups, monitoring vital levels, and discussing preventive care with a healthcare provider.

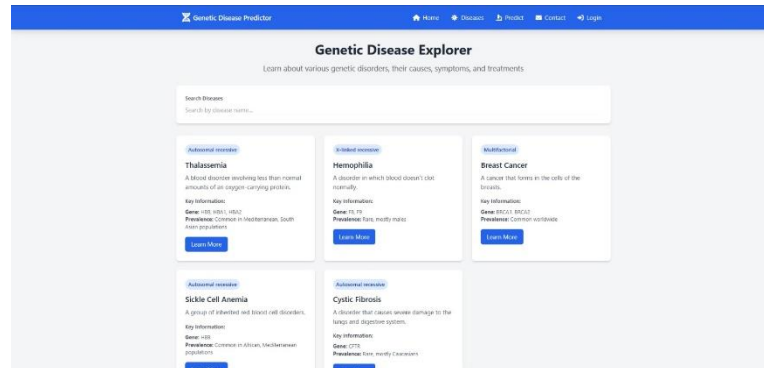


Figure 4: Genetic Disease Explorer

Figure 4 presents the "Genetic Disease Explorer" page of the Genetic Disease Predictor web application. The header and navbar remain at the top, enabling navigation to Home, Diseases, Predict, Contact, and Login. Below the title, a search bar allows users to search for genetic diseases by name. Multiple disease cards are displayed, each summarizing a genetic disorder such as Thalassemia, Haemophilia, Breast Cancer, Sickle Cell Anemia, and Cystic Fibrosis. Each card includes a brief description, key information, and a "Learn More" button for detailed exploration of causes, symptoms, and treatments. This page helps users access curated, foundational knowledge about various genetic diseases—facilitating patient, student, and clinician education through a simple web interface.



Figure 5: Prediction Results

Figure 5 shows the "Prediction Results" panel from the Genetic Disease Predictor application. The system predicts the disease as "Breast Cancer" and clearly states a "High Risk" level, with a confidence score of 58.0%. Under the risk assessment, the panel provides actionable recommendations for the user's healthcare management. Suggestions include seeking genetic counselling, maintaining regular health check-ups, and monitoring blood

pressure and cholesterol levels. Users are also advised to discuss preventive measures with personal healthcare provider for more detailed guidance. This results panel delivers personalized, clinically relevant feedback to support informed decision-making and proactive health management.

5. Conclusion

This project delivers a cutting-edge genetic disorder prediction platform that leverages the power of advanced machine-learning techniques, with the Random Forest algorithm serving as its fundamental predictive engine. Renowned for its effectiveness and robustness in handling intricate genetic datasets, the system analyzes crucial genetic biomarkers—specifically SNPs—in conjunction with pertinent clinical data to generate precise and dependable risk forecasts.

The platform is embodied in an easy-to-use web-application developed using the Flask framework, designed to offer seamless access for healthcare providers as well as patients. Beyond providing raw predictive outcomes, it delivers insightful analyses of genetic risk factors, actionable management guidance, and probabilistic risk assessments that support well-informed clinical decisions. By automating the early identification of genetic disorders and providing clear, interpretable insights, the system strives to progressively improve healthcare outcomes through timely medical interventions. Its scalable, efficient, and accessible architecture represents a significant advancement in the field of genetic risk prediction, aligning closely with the evolving landscape of personalized and precision medicine that demands reliable and streamlined tools for genetic health evaluation.

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