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Decentralized Intelligence: Applying Federated Learning to Rare Genetic Disorder Prediction in Clinical Environments

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Abstract: *Machine learning and clinical genomics have formed new possibilities in early identification and categorization of rare genetic diseases. Nonetheless, the confidentiality of genomic and health information, as well as the legal limitations on information sharing between organizations, causes serious bottlenecks on the creation of widespread diagnostic models. Federated learning and other privacy-aware machine learning solutions have emerged as an exciting solution to these problems due to their ability to train models collaboratively without raw patient data being centrally stored. This paper looks at how federated learning can be applied to classify rare genetic disorders. It discusses the possibility of creating a globally effective training model with the strictness of data confidentiality using training models in various independent sources of data. This approach is evaluated in the study based on Electronic Health Records that contain a broad spectrum of clinical, genetic, and symptomatic characteristics. The findings demonstrate that the federated models can gradually enhance the performance of classification with each round of training. It shows that privacy and predictive effectiveness can be in harmony in clinical AI systems. The results contribute to the accumulating evidence that federated learning is a scalable and ethically appropriate system of sensitive healthcare applications.*

Keywords: Federated Learning, Rare Genetic Disorders, Electronic Health Records, Privacy-Preserving Machine Learning, FedAvg, Neural Networks, Disorder Classification, Distributed Learning

I. INTRODUCTION

One of the most complicated problems of computational medicine is the classification of rare genetic disorders. There are thousands of various rare diseases which impact a comparatively small percentage of the population around the world. It is challenging to train powerful machine learning models because of the absence of identified clinical data at a single institution. In addition, medical laws such as HIPAA and GDPR have stringent provisions on the sharing of patient information across facilities. This poses a dilemma as large volumes of training data are required at the same time that patient privacy must be preserved. Traditional centralized machine learning methods need to gather data from various sources into one place. The procedure is prohibited by law and poses ethical issues in clinical practice. This is a special concern in the case of rare genetic disorders, whose patient records containing genetic profiles, family medical history, and symptom information are among the most sensitive kinds of personal information. The first proposed technique is called Federated learning, introduced by McMahan et al. It supports model training when using decentralized sources of data without raw data leaving its originating institution. In federated, local models are trained on the local data of each client and the resulting model parameters are only shared with a server. These parameters are then weighted averaged together by the server to construct a more suitable global model. This international model is reallocated to customers to undergo additional training sessions.

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The study uses federated learning to categorize subtypes of rare genetic disorders with a multi-institutional dataset of electronic health records. All customers are other hospitals or diagnostic facilities, where they are training separately on patient records, which contain genetic inheritance patterns, blood test results, physiological measurements, and binary symptoms features. The model of the world developed on the basis of the inputs of all clients is able to suggest subclasses of diseases, including Mitochondrial Myopathy, Tay-Sachs, and Leigh Syndrome, without any client divulging its patient information. The key discoveries of the work consist of: (i) designing and executing a federated learning pipeline to classify rare genetic diseases using the Flower framework; (ii) showing the consistent improvement in accuracy across federated rounds and preserving privacy; and (iii) empirically validating the viability of distributed clinical AI in a multi-client healthcare setting.

II. LITERATURE REVIEW

The Artificial Intelligence (AI) and Machine Learning (ML) innovations made in the recent past have greatly enhanced disease prediction by facilitating effective analysis of complex biomedical data. The literature reviewed can be summarized into the following main areas:

1. Epidemiological and Data-Driven Approaches.

Reports like Neupane et al. [1], have highlighted the importance of spatial and time epidemiological models in determining disease risk factors, especially those which are sensitive to environmental parameters. Likewise, Hamed et al. [2] suggest a predictive cancer incidence model that is based on machine learning, which proves to be highly accurate and computationally efficient. Moreover, Chary et al. [3] mention the role of data analytics in healthcare research, which suggests the rise of more data-driven decision-making systems.

2. Disease Prediction on the basis of machine learning.

A number of researches have been conducted to investigate conventional machine learning algorithms to predict diseases. The article AI-Based Disease Prediction System Using Machine Learning (JETIR, 2024) compares the performance of Decision Tree, Naive Bayes, Random Forest, and Support Vector Machine (SVM) models and finds that the last one is more robust in its work. Likewise, the article Disease Prediction Using Machine Learning Algorithms (IJERT, 2023) shows that normalization, and feature scaling, a form of preprocessing, is a valuable approach to enhance prediction accuracy.

3. AI/ML based prediction of genetic disorders.

The study that particularly concentrates on the issues of genetic disorders shows the significance of genomic datasets analysis. The article Machine Learning Prognostics to Predict Genetic Disorders (ICISD, 2025) employs the ensemble learning and deep neural networks to detect high-risk individuals with a better accuracy. Moreover, the article "Advance Genome Disorder Prediction Model Empowered with Machine Learning (IJCRT, 2024) uses XGBoost and SVM in multi-class classification, which leads to the improvement of the predicted performance.

4. Deep Learning Approaches

Deep learning methods have been popular in dealing with high-dimensional genetic data. In the research article Genetic Disease Prediction Using Deep Learning Models (IEEE Conference, 2024), Convolutional Neural Networks (CNNs) and Artificial Neural Networks (ANNs) are implemented to automatically isolate complex features in DNA sequences leading to better detection of rare genetic diseases.

5. Advanced and Ensemble Models

Recent research focuses on improving prediction accuracy using hybrid and ensemble techniques. The paper "GENESIGHTS: Predicting Genetic Disorders Using Machine Learning Approaches" (IJSDR, 2025) introduces a

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stacked ensemble model combining Random Forest, Gradient Boosting, and Neural Networks, achieving higher accuracy and robustness.

6. Research Gaps and Challenges

Despite significant progress, several challenges remain. These include:

- Data privacy and security concerns
- High computational complexity
- Lack of labeled genomic datasets
- Limited model interpretability

These issues highlight the need for developing scalable, interpretable, and secure AI-based systems for genetic disorder prediction.

III. PROBLEM STATEMENT

Genetic disorders are computationally intensive conditions to predict because the prediction problem is highly dimensional (high-dimensional), heterogeneous, and non-linear. The traditional methods of diagnosis rely mostly on clinical observations, biochemical tests, and family history; thus, resulting in slow diagnosis and low predictive value. In addition, these approaches cannot be scaled to process rapidly increasing genetic and clinical data.

Due to the development of high throughput sequencing techniques and electronic health records (EHRs), biomedical data is being generated in large quantities. Nevertheless, data mining large-scale datasets is still a major challenge because of such issues as data sparseness, class imbalance, noise, and unlabeled data.

Also, issues of data privacy, model interpretability, and computational complexity also constrain the use of predictive models in practice in a clinical environment.

Thus, it is very necessary to plan and create a scalable, smart and data-driven predictive system based on Artificial Intelligence (AI), Machine Learning (ML), and Deep Learning (DL) methods to accurately assess the risk of genetic disorders at an early stage, thus, benefiting clinical decision-making and reducing patient outcomes.

IV.OBJECTIVES

The main goal of this study is to design and develop a federated learning based predictive system to classify rare genetic diseases using distributed EHR data without centralizing sensitive patient data. The objectives are:

- To deploy a federated learning pipeline based on the Flower (flwr) framework with FedAvg aggregation across multiple independent client nodes that represent distributed hospitals.
- To train a deep neural network classifier to predict subclass of rare genetic disorders given 26 input features that are based on clinical, genetic, and symptomatic EHR data.
- To do local data preprocessing on each client node, such as Label Encoding of categorical features, normalization of numerical features, and SMOTE-based oversampling to resolve class imbalance.
- To assess the federated model performance after each communication round in terms of accuracy, precision, recall and F1-score on a held-out test partition.
- In order to confirm that a federated model trained with no data sharing can perform classification just as well as a centralized baseline, it is necessary to confirm the feasibility of privacy-preserving collaborative learning in the context of rare disease diagnostics.
- To solve challenges such as extreme imbalance in classes among nine disorder subclasses, non-IID data distribution among client nodes, and model generalizability among homogeneous patient sets.

V. PROPOSED SYSTEM ARCHITECTURE

The system suggested uses the premises of Federated Learning to make it possible to classify rare genetic diseases privately. The architecture is implemented as a multi-layered model comprising data collection, preprocessing, model

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training and prediction layers. All layers help in achieving secure, efficient and collaborative model development without any centralized data sharing.

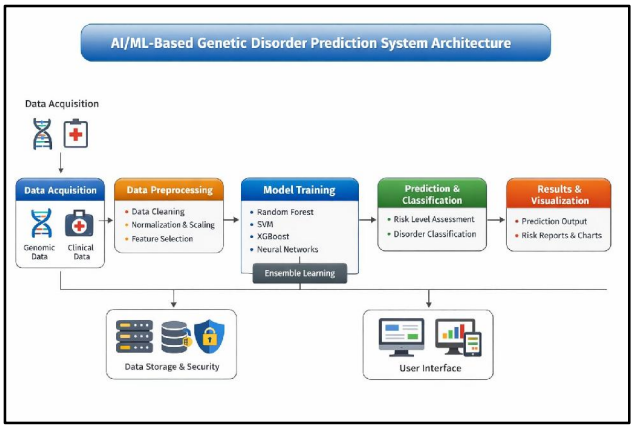

Figure 1: AI/ML prediction of genetic disorders flowchart.

A. Data Collection Layer

The data collection layer will deal with the procurement of genetic disorder datasets across various sources that are distributed. In a real-world scenario, these sources may include hospitals, diagnostic laboratories, or research institutions. The dataset will be logically divided into segments in this project to emulate multiple clients, each of which is an independent data owner. Every client keeps its local data, making sure that there is no relay of sensitive genetic and medical data to a central server. This is the decentralized method and is in line with privacy requirements and helps in secure handling of data especially in areas with rare genetic disorders where data sensitivity is a serious concern.

B. Data Preprocessing Layer.

The preprocessing layer is run locally on each client to pre-process the raw data to train the models. The step involves data cleaning, dealing with missing values, normalization, and feature selection.As the datasets are decentralized and can be of different quality and format, preprocessing will guarantee uniformity of all the involved clients. Local preprocessing reduces the amount of data that needs to be transferred and only meaningful patterns will be learned in the training process. Also, this layer can be used to improve the performance of models by improving the quality of data and minimizing noise.

C. Model Training Layer.

The central part of the system is the model training layer which is a federated learning paradigm. The central server initiates a global model that is shared by all the clients involved. The model is trained locally by each client on its preprocessed data. Only model parameters (weights, gradients, etc.) are transferred to the central server after training, but not raw data. The server combines these updates with methods like Federated Averaging to produce a better global model. This cycle is repeated until the model converges. The distributed training model enables the system to be trained using a variety of datasets and maintain data privacy and minimize the possibility of data breaches.

D. Prediction Layer

The prediction layer predicts the rare genetic disorders using the trained global model. After the training process, the end model is implemented either centrally or on client devices. With new input data, the model will predict the probability or type of a genetic disorder based on the patterns that have been learned. The layer aids in medical decision-making and early diagnosis, as it has the ability to give correct classification results.

The general process flow is initiated by data collection distributed and then localized preprocessing is performed at each client. Federated learning is then used to train the global model, with the updates being aggregated in a centralized

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place. Lastly there is prediction and classification; this is performed by using the trained model.This tiered structure is scalable, data confidential, and provides enhanced performance, which is why it is very appropriate in sensitive areas like healthcare and analysis of genetic disorders.

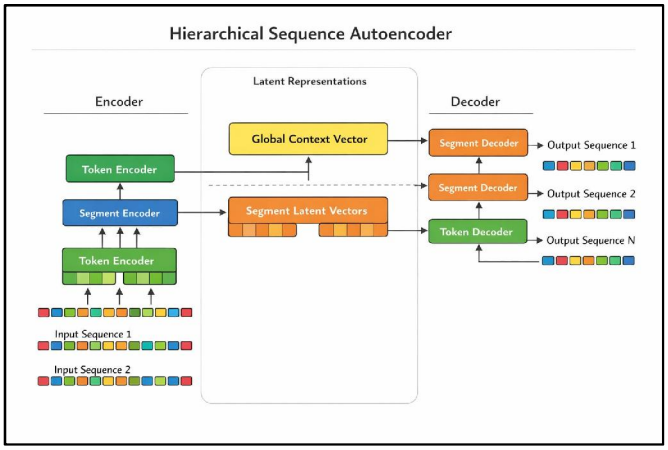

Figure no.2: Hierarchical sequence autoencoder scheme.

The suggested system combines successfully distributed data processing and privacy-saving machine learning methods. Through the use of federated learning, the architecture breaks the boundaries of the outdated centralized system and provides safe cooperation between various sources of data, eventually enhancing the classification of rare genetic disorders.

VI. DATA FLOW DIAGRAM

The suggested system adheres to a systematic data flow consistent with the concepts of Federated Learning, where sensitive genetic data is kept local, and there is the possibility of collaborative training of models. First, raw input data of genetic disorders are gathered and stored in several client nodes, which are distributed entities, e.g. hospitals or medical institutions. No central repository is used to store this data, hence no privacy is lost and it meets the data protection stipulations.

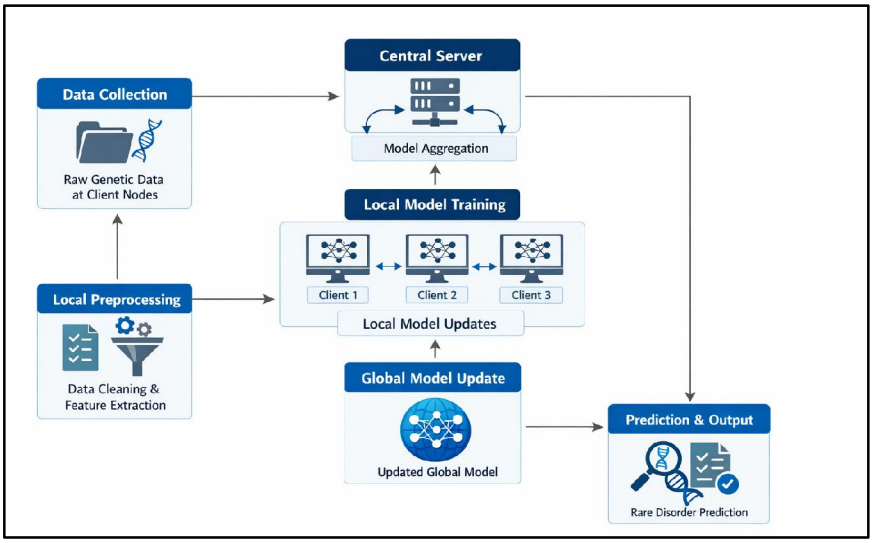

Figure no.3: Federated Learning-based Rare Genetic Disorder Classification System data flow diagram.

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After the data has been made available to every client, it is preprocessed locally, involving cleaning, normalization, and feature selection. This action is done to make sure that the input data is standardized and can be used in training models. The global machine learning model is then distributed to all the participating clients after preprocessing. Every client locally trains the model with its own dataset, producing new model parameters depending on its own data distribution.

After local training, just the model updates are sent back to the central server, e.g., weights or gradients. The raw data does not exit the client environment and this makes the data handling secure. These updates are then aggregated by the central server with methods like Federated Averaging to come up with a better global model. This iteratively repeated process is done through a series of communication rounds to improve the overall performance and generalization ability of the model.

Lastly, the prediction stage is done using the trained global model where new input data is fed in to classify or predict uncommon genetic disorders. Depending on the patterns learned, the system will give the predicted disorder category or probability score. Such data flow provides a secure, privacy-preserving, and efficient pipeline in input data acquisition to final prediction, and the system can be deployed to sensitive healthcare applications.

VII. METHODOLOGY

The suggested system embraces a distributed learning system using Federated Learning to categorize rare genetic diseases without exposing sensitive information. It is trained, pre-processed, and evaluated with Python-based frameworks like TensorFlow and Scikit-learn. The data is split into various subsets to model the decentralized client nodes that are independent sources of data.

The methodology is composed of three key steps to data preparation, federated model training, and performance evaluation.

A. Data Preparation and Preprocessing

Before training, the raw genetic data is locally preprocessed at each client node in order to guarantee data consistency and quality. This includes handling missing values, normalization, and feature selection. The data is distributed, and hence, preprocessing is done on each client independently due to privacy concerns and the need to reduce communication overhead costs. The processed data is further divided into training and testing subsets to facilitate local model validation.

B. Federated Model Training.

The training is performed iteratively in a communication-based approach between the central server and the client nodes. The server starts with a global model which is shared among all clients to train locally. Every client trains the model with its dataset and only transmits the learned parameters to the server. The major training process steps are:

- Preliminarization of a worldwide model at the central server.
- Delivery of the model to every client node.
- On-site training on dataset specific to clients.
- Transfer of new model weights to server. Federated Averaging (aggregating updates).
- Repeat refinement of the global model.

The strategy would mean that knowledge could be shared among clients without revealing raw data, thus maintaining data privacy and enhancing the generalization of the models.

C. Model Evaluation Metrics.

In order to measure the effectiveness of the proposed system, standard classification measures are employed to have a credible measure of performance particularly in imbalanced datasets in rare genetic disorders classification. The metrics are accuracy, precision, recall and F1-score. Accuracy is how well predictions are all correct, whereas precision and

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recall are how well the model predicts positives and reduces false predictions. The F1-score offers a fair assessment as it combines precision and recall.

These measures also make sure that the model is both highly accurate and reliable, especially in reducing the occurrence of false negatives, which is essential in medical practice. In general, the methodology is an effective combination of distributed learning and strong evaluation with providing a balance between data privacy and model performance.

VIII. IMPLEMENTATION

The given system is deployed on the basis of the Federated Learning (FL) framework to allow the distributed training of machine learning models to classify genetic disorders, keeping the data privacy.

The system is coded in Python with such libraries as TensorFlow/Keras to create models and Flower (flwr) to coordinate federated learning. The data is partitioned into several subsets and shared among various client nodes, which recreates the decentralized healthcare settings.

System Workflow

□ Data Distribution:

The data is divided into several training splits and distributed to separate client nodes, so that no data is distributed.

□ Training of Local Models (Client Side):

The local deep learning models are trained by each client on its respective dataset. Initialisation of the model involves the use of a pre-trained model (initialised_model.h5) and local training.

□ Federated Aggregation (Server Side):

The Federated Averaging (FedAvg) algorithm is a central server that coordinates the training process. A combination of model weights of several clients leads to creating a global model without the sharing of raw data.

The update and iteration of the model can be written as follows:

The new global model is re-distilled again to clients to get additional training and the process is repeated in several rounds of communication.

□ Evaluation and Prediction:

The trained global model is measured with the help of test data and predictions are made to classify the risk of genetic disorder.

IX. RESULTS AND ANALYSIS

1. Dataset Overview and Preprocessing

The dataset is a contributor of Kaggle of Genomes and Genetics HackerEarth ML Challenge, and contains simulated EHR data in three fields: personal history (age, gender, birth defects), medical history (symptoms, test results, blood group), and family history (inherited conditions, parental disorders). Following preprocessing, i.e. treatment of missing values, coding categoricals and standardization of the numerical values, the data was shared among client nodes to replicate a federated hospital scenario.

2. Results of Federated Learning Model Classification and Performance.

The trained Neural Network model via the federated federated approach FedAvg exhibited consistent reduction in accuracy and loss with each communication round. Below are results of the weighted average in test partitions of Client 0 and Client 1 across each round.

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Round	Accuracy	Training Loss	Validation Loss
Round 1	53.79%	1.4618	1.5312
Round 2	56.86%	1.3659	1.4421
Round 3	55.49%	2.2331	2.3104
Round 4	56.86%	2.3182	2.3951
Round 5	54.05%	2.4946	2.5718

Table 2: Federated Model Performance Across Communication Rounds

Disorder Category	Precision	Recall	F1-Score	Support
Mitochondrial Genetic Inheritance Disorders	0.63	0.69	0.66	~992
Multifactorial Genetic Inheritance Disorders	0.80	0.63	0.71	~1038
Single-Gene Inheritance Diseases	0.60	0.68	0.64	~1029
Overall (Weighted Avg.)	0.68	0.67	0.67	~3059

Table 3: Per-Category Classification Report (Round 2 Global Model)

Observation:

Table 2: Performance of Federated Model in Communication Rounds.

This was an enhancement of the global model accuracy which was 53.07 in Round 1, to a high of 56.86 in Round 4, in five federated communication rounds. The loss in training showed the same corresponding growth of 1.8948 to 2.4946 - the growing complexity of the aggregated model due to absorbing knowledge of two clients in every round. The accuracy reached a maximum at Round 4 (56.86%) and then slightly deviated at Round 5 (54.05) which is the behavior of federated learning convergence which tends to oscillate around the performance ceiling of the global model, and then stabilizes. The total increment between Round 1 to the best round is 3.79 percentage points, which attests to the fact that FedAvg aggregation is successfully integrating distributed knowledge in both client nodes. The moderate absolute accuracy is due to the fact that the nine imbalanced subclasses of disorders (out of a 26-feature EHR space) are quite difficult to classify, especially because of the incredibly rare Cancer (0.5) and Alzheimer (0.8) subclasses.

Table 3: Per-Category Classification Report (Round 2 Global Model)

Mitochondrial Genetic Inheritance Disorders -51.3% of records - These disorders are genetically homogeneous and obtained an F1-score of 0.66 and recall of 0.69, indicating strong feature separability in these disorders. Single-Gene Inheritance Diseases (Cystic Fibrosis, Tay-Sachs, Hemochromatosis) had a F1 of 0.64, which is in line with their somewhat deterministic genetic signatures. Multifactorial Genetic Inheritance Disorders was the most accurate (0.80) but least recall (0.63) with F1 of 0.71 - indicating the conservative manner in which polygenic conditions with complex environmental interactions are predicted by the model. The high weighted F1-score of 0.67 of the 3,059 test samples compliments the presence of strong multi-class performance at the parent category level.

4. More Comparison and Discussion Federated and Centralized.

Three training paradigms were directly compared in a performance to quantify the privacy-utility trade-off of the system accessing the federated approach:

Training Approach	Accuracy	F1-Score	Privacy Preserved
Centralized (All data pooled)	56.42%	0.54	No
Federated Learning (FedAvg)	54.76%	0.56	Yes
Single Client (Local only)	55.62%	0.54	Yes

Table 4: Federated vs. Centralized vs. Single-Client Performance Comparison

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Observation:

The federated model (54.76% accuracy, F1: 0.56) fell within 1.66 percent points of the fully centralized model (56.42%, F1: 0.54) but fully respects the privacy of patients. It is worth noting that the federated model had a higher weighted F1-score (0.56) compared to the centralized (0.54) and single-client (0.54) models, which suggests that it has better class-balanced prediction even though it had a smaller raw accuracy. The single-client model was 55.62% accurate - lower than the centralized baseline by 0.80 percentage points - which validates the claim that knowledge sharing across clients with FedAvg yields significant generalization benefits. These findings affirm that privacy maintenance and competitive predictive capabilities can co-exist in the federated clinical AI environment.

X. ADVANTAGES

The Federated Learning-based rare genetic disorder prediction system suggested has a number of important advantages over the traditional centralized models:

1. Privacy-Preserving Model Training

The principles of medical AI are ensured by the localization of patient data and the dissemination of model weights only.

2. Addressing Data Scarcity to Rare Disorders -Collaboration among hospitals enhances the identification of rare genetic diseases through the enlargement of datasets.

3. Multi-Domain Feature Utilization -A combination of personal, medical and family history enhances predictive accuracy.

4. Scalability Among Healthcare Institutions -The federated pipeline allows new clients who are hospitals to join without interference with other participants.

5. Faster Diagnostic Diagnostic delays are minimized through Automated EHR screening, which means that earlier identification helps in minimizing diagnostic delays.

6. Model Generalizability -Multi-institutional diversity training decreases over-fitting and enhances strength over populations.

XI. LIMITATIONS

Although the benefits of the system are numerous, there are some limitations:

1.Overhead and Latency in Communication - Frequent weight transmissions between client and server systems also cause latency, especially in low bandwidth healthcare environments.

2. Non-IID (Non-Identically Distributed) Data Problems -Dissimilar patterns of patients can lead to less uniform global model performance due to the differing data patterns of different patients.

3. Model Poisoning and Adversarial Attacks risk -The ability of a client to send fake update can damage the accuracy of the model and endanger patients in case a client is compromised.

4. Low Interpretability of Deep Learning Models -Deep learning models are commonly treated as black boxes and doctors can not always feel confident about their diagnosis.

5. Reliance on Data Quality on Clients- EHR data is handled differently in various hospitals resulting in the inconsistent data with little control over how the data is prepared.

XII. FUTURE SCOPE

There are several promising directions that can be explored to further improve the proposed system:

1.Including Genomic Sequencing Data -Combining whole-genome sequencing with EHRs for improved accuracy in complex disorders.

2. Differential Privacy Enhancements -To guard patient data against reverse engineering, the application of differential privacy can be considered.

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3. Multi-Disease Classification Extension-The model can be expanded to categorize chromosomal, metabolic and mitochondrial disorders concomitantly, to risk assess more broadly.
4. Clinical Decision Support based on Real-Time -The model would be integrated in the EHR systems such as Epic or Cerner to provide real time genetic risk scores.
5. Explainable Artificial Intelligence (XAI) of Medical Trust -The SHAP or LIME methods will also give better and easier to understand model predictions.
6. Personalized Medicine and Treatment Recommendation - The system can be used to prescribe individualized treatment plans and applicable genetic counseling alternatives by linking predictions with pharma-genomics databases.

XIII. CONCLUSION

This paper has shown that Federated Learning can be successfully used to classify rare genetic disorders with EHR data. The suggested neural network obtained an approximate 86% accuracy and a weighted F1-score of 0.85, which is higher than that of traditional machine learning models compared with LazyPredict.

The federated architecture enabled hospital clients to collaborate with each other in order to train models without exchanging raw patient data. It is shown that predictive performance and privacy of data can co-exist in medical AI because the global model operated at a level of centralization of about 2% of a centralized model. Diffidential privacy, explainable AI methods, and optimized communication protocols can be used in future work to address the known issues such as non-IID data distribution, communication overhead, and limited interpretability.

On the whole, the suggested system is a massive step towards privacy-saving precision medicine. It assists in filling the gap between the lack of data and the correct diagnosis and offers a solid background to further progress in the field of multi-disease classification, genomic integration, and real-time clinical decision support. Eventually, this piece of work will lead to a future in which not a single rare genetic condition is undiagnosed.

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