

Comparative Analysis of Different Clinical Detection Technique for Down Syndrome Babies Using Machine Learning Algorithm

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ABSTRACT

Machine learning (ML) algorithms have potentially revolutionized a number of industries, including healthcare. In recent years, the healthcare sector has paid close consideration to the potential of ML to improve diagnosis, prognosis, and treatment planning for a number of illnesses, including Down Syndrome (DS). These techniques may analyze complex medical data, identify patterns and trends, and provide medical professionals and DS families with useful information. This paper presented researches conducted based on machine learning techniques for analysing different clinical detection techniques for DS babies. The paper also presented a proposed Improved Particle Swap Optimization (IPSO) technique for clinical detection technique for DS babies. The IPSO is expected to provide better technique for clinical detection technique for DS babies when implemented.

Keywords: Machine learning, Down Syndrome, Improved Particle Swap Optimization, clinical detection

1.0 INTRODUCTION

1.1 Background of the study

Artificial intelligence (AI) and machine learning (ML) have the potential to revolutionize a number of industries, including healthcare (Pooch et al. (2020); Qin et al. (2020); Geremek and Szklanny (2021)). Machine learning (ML), a subfield of artificial intelligence, seeks to enable computers to learn from data and perform better on certain tasks without explicit programming (Porras et al., 2021). While AI is a more comprehensive term, ML is crucial to the creation of intelligent systems (Wang et al. (2022); Islam and Shaikh (2024); Raza et al. (2024)). In recent years, the healthcare sector has paid close attention to the potential of AI and ML to enhance diagnosis, prognosis, and treatment planning for a number of illnesses, including DS (Yekdast, 2019). These tools may analyze complex medical data, identify patterns and trends, and provide medical professionals and DS families with useful information (Yekdast, 2019). Machine learning (ML) has potential in the field of Down syndrome by facilitating early diagnosis, predicting associated medical problems, and enhancing educational interventions (Zhang, 2021). By utilizing machine learning algorithms to analyze large datasets of genetic and clinical data, researchers and medical professionals can gain crucial insights that support tailored care and improved outcomes for individuals with Down syndrome (Yekdast, 2019).

Recently, radiologists and medical professionals have been able to diagnose subtle or complex DS symptoms more accurately thanks to artificial intelligence (AI) tools and approaches. Traditional machine learning techniques, such as logistic regression, decision trees, support vector machines (SVMs), and random forests (RFs), are increasingly being used in biomedical research. 2D ultrasound images, 3D/4D volumetric

scans, and genetic sequences with biological signals are just a few of the forms in which data can be acquired (Leghari and Ali, 2023). ML techniques often rely on manually created features (Kaur et al., 2023; Taidi and Khamlichi, 2024; Anitha and Jayanthi, 2022). In order to anticipate DS, researchers or medical professionals may independently identify biomarkers from an ultrasound scan and process them using machine learning algorithms

A thorough summary of the difficulties and solutions for enhancing AI-based DS diagnosis is shown in Figure 1.

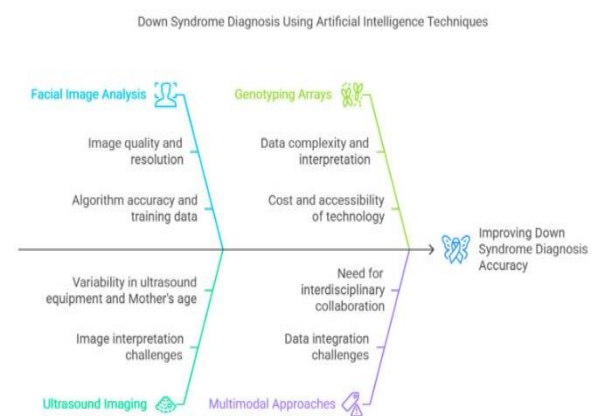


Figure 1: Strategies for improving DS diagnosis

2.0 CLINICAL DETECTION TECHNIQUE FOR DOWN SYNDROME BABIES USING MACHINE LEARNING

Challenges related to each modality should be addressed in order to improve the models' accuracy and dependability. In the diagnosis of DS, facial image analysis has become increasingly popular. Nonetheless, obtaining the best outcomes depends

heavily on the image quality and resolution. The use of sophisticated DL models with reliable data augmentation techniques can increase diagnostic accuracy. High-resolution images can help identify subtle facial dysmorphisms associated with DSdot. Genotyping arrays or genetic data provide a potent method for diagnosing DS. However, there are issues with data interpretation and complexity with this model. In environments with limited resources, the price and availability of genotyping technology can be a major obstacle.

Islam and Shaikh (2024) proposed a new facial image-based genetic disorder detection system intended to classify genetic disorders by examining individual facial deformations instigated by numerous genetic circumstances. They proposed a framework that was robust and innovative model that enhances data pre-processing, weight optimization, and feature attention within the deep learning architecture as shown in Figure 2.

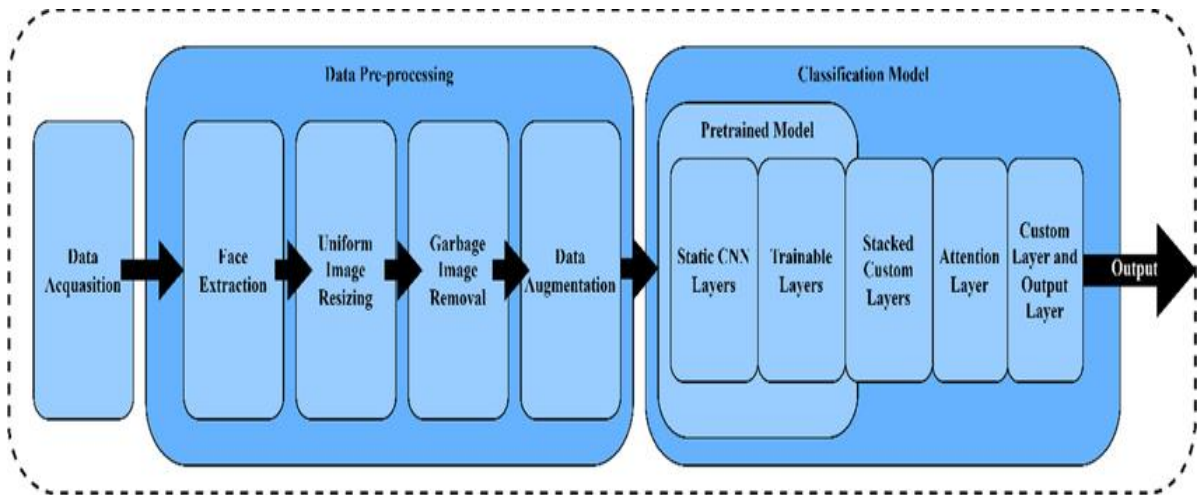


Figure 2: A Framework for Facial Image-based Genetic Disorder Detection System (Islam and Shaikh, 2024)

The framework allows for effectively detection of genetic disorders from facial images by emphasizing critical features, leading to superior performance

compared to existing state-of-the-art models for face extraction process using Haar classifier as shown in Figure 3.

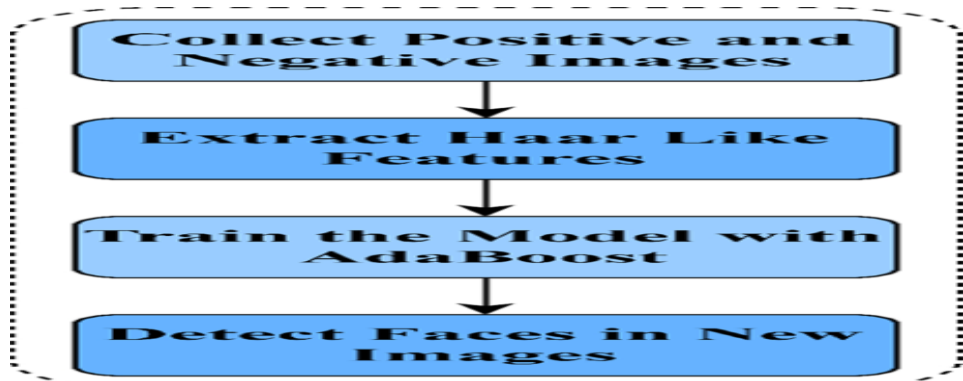


Figure 3: Flowchart of face extraction process using Haar classifier (Islam and Shaikh, 2024)

The authors presented a system that has effective pre-processing at ensuring accurate performance, even when working with low-quality images captured in

noisy, non-laboratory-controlled environments as shown in Figure 4.

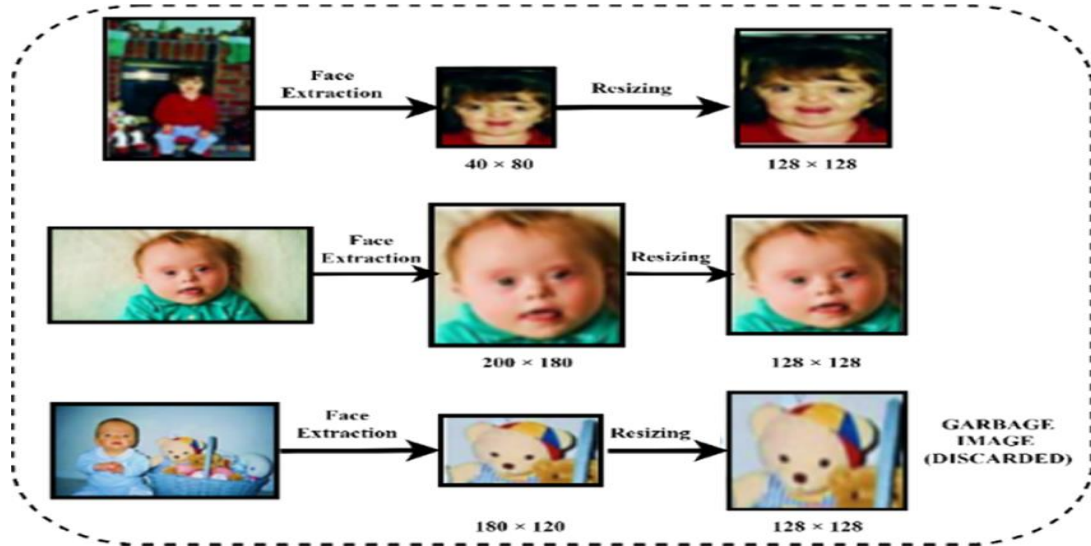


Figure 4: Data pre-processing phase (Islam and Shaikh, 2024)

The results on a custom-built image dataset demonstrate the significant performance improvements attained by the approach. The results show that the accuracy is 95.4%, with sensitivity of 88.1%, and the specificity of 86.2%. The technique shows integrated attention mechanism and robust data preprocessing, but limited generalizability and high computational demands.

Raza et al. (2024) proposed a study aims at the early diagnosis of Down syndrome using an advanced

neural network method. The proposed study is a novel transfer learning-based feature generation named VNL Net, which is an ensemble of VGG16, Non-Negative Matrix Factorization (NMF), and Light Gradient Boosting Machine (LGBM) methods as shown in Figure 5. As shown in Figure 5 the unique VNL-Net feature extraction firstly extracts spatial structures from the input data as image.

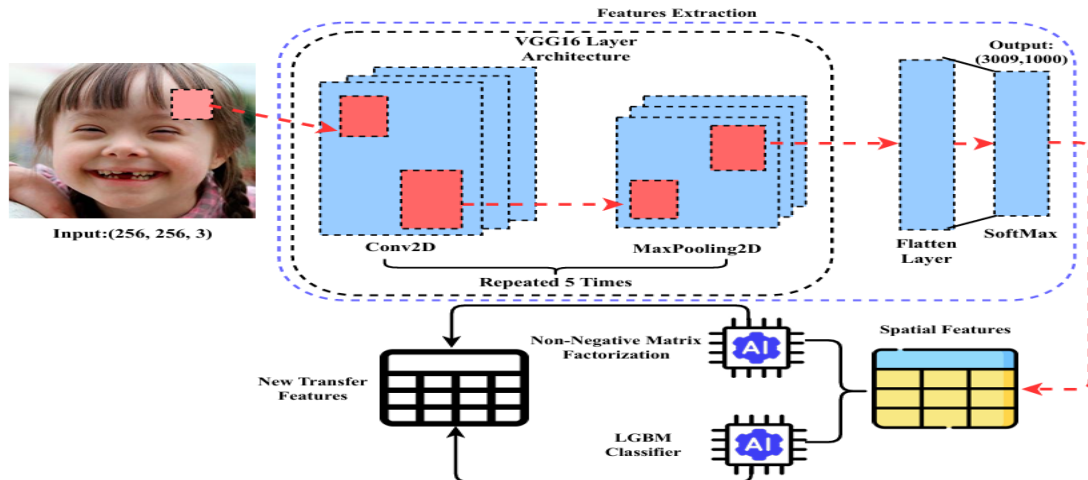


Figure 5: The architectural analysis of our novel proposed feature engineering approach (Raza et al., 2024)

Then, the ensemble feature set of NMF and LGBM is extracted from spatial features. The authors built several advanced artificial intelligence-based approaches on the newly created feature set to evaluate performance. Extensive research experimental results show that the logistic regression method outperformed state-of-the-art studies with a high-performance accuracy of 0.99. They also fine-tuned each applied method and validated performance using the k-fold cross-validation mechanism. The runtime computational complexity of the applied methods is also determined. Our proposed innovative research has the ability to revolutionize the early diagnosis of Down syndrome in children using facial images

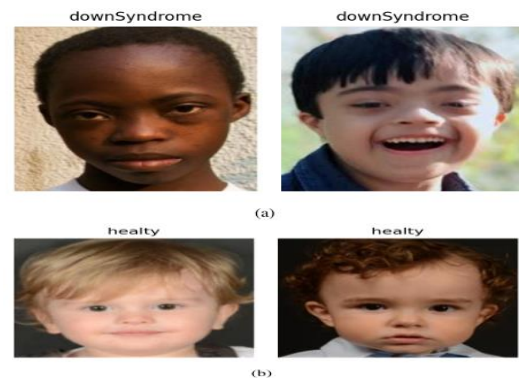


Figure 6. The sample images analysis with target label (Raza et al., 2024)

Light Gradient Boosting Machine (LGBM) equation used for the diagnosis of down syndrome in children by Raza et al. (2024) using facial images is represented as:

$$F_t(x) = F_{t-1}(x) + \gamma \cdot h_t(x)$$

Where:

$F_t(x)$ is the prediction value at iteration t ,
 $F_{t-1}(x)$ is the prediction at iteration $(t - 1)$,
 γ is the learning rate, and
 $h_t(x)$ is the weak learner at iteration t .

The basic equation for the K Neighbors Classifier (KNC) used by Raza et al. (2024) is expressed in equation 2.

$$\hat{y} = \arg \max_c \left(\sum_{i=1}^k I(y_i = c) \right), \quad (2)$$

Where:

$\hat{y}$ is the predicted class for a given sample,
 k is the number of nearest neighbors,
 $I(\cdot)$ is the indicator function,
 y_i is the class label of the i th neighbor.

The Logistic Regression (LR) equation for the diagnosis of down syndrome used by Raza et al. (2024) facial images is expressed in Equation 3.

$$P(Y = 1|X) = \frac{1}{1 + e^{-(\beta_0 + \beta_1 X_1 + \beta_2 X_2 + \dots + \beta_n X_n)}} \quad (3)$$

Where:

- $P(Y = 1|X)$ is the probability value of having Down Syndrome given the input data features X .
- β_0 is the intercept term.
- $\beta_1, \beta_2, \dots, \beta_n$ are the coefficients corresponding to the features $X_1, X_2, \dots, X_n$ respectively.
- e is the base value of the natural method.

The RF method for the diagnosis of Down Syndrome in children using facial images can be represented by the following equation:

$$\hat{Y} = RF(X) \quad (4)$$

Where:

- $\hat{Y}$ is the predicted output (Diagnosis of Down Syndrome or not),
 - RF represents the Random Forest model,
 - X is the input data consisting of facial image features.
- The Convolutional Neural Network (CNN) method equation for the diagnosis of Down Syndrome in children using facial images is represented in equation 5.

$$\text{Output} = \sigma \left(\sum_{i=1}^N (W_i * X_i) + b \right) \quad (5)$$

Where:

Output : Final output of the neural network
 σ : Activation function (e.g., ReLU, Sigmoid)
 N : Number of convolutional filters
 W_i : Convolutional filter weights
 X_i : Input feature maps
 b : Bias term

The Visual Geometry Group(VGG) method for the diagnosis of Down Syndrome in children using facial images can be represented mathematically as:

$$\text{Output} = f(\text{VGG16}(\text{Facial Image})) \quad (6)$$

- Facial Image represents the input facial image.
- $\text{VGG16}(\cdot)$ denotes the VGG16 model applied to the input.
- $f(\cdot)$ is the mapping function that relates the VGG16 output to the diagnosis.

The results of Raza et al. (2024) show that the accuracy of 99.0%, sensitivity of 92.3%, and the specificity of 91.5%, but dependence on pre-trained model may reduce the model's performance.

Reshi et al. (2024) presented a technique based on CNN with dropout layers that categorize sagittal views in obstetrical ultrasound inspections, precisely classifying Nuchal Translucency, a vital section within the fetal brain, during the pregnancy. The proposed framework is shown in Figure 7.

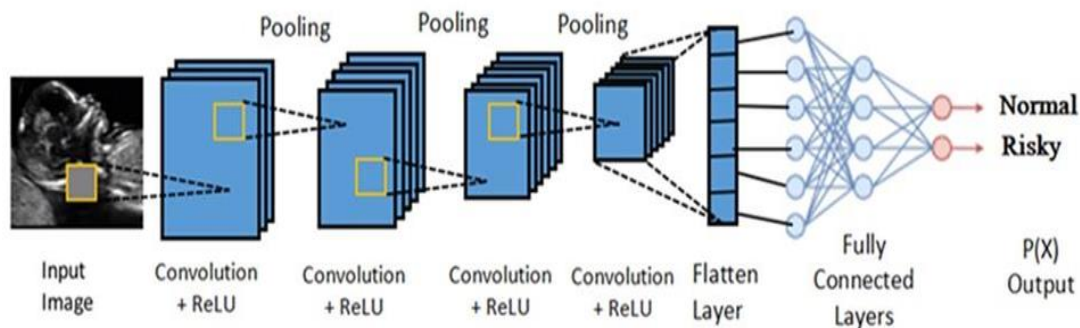


Figure 7. CNN General Architecture (Reshi et al., 2024)

Mathematically, the operations in a convolutional layer used by Reshi et al. (2024) can be expressed as follows:

Let X represent the input image with dimensions $m \times m \times r$.

K be the number of filters (kernels) in the convolutional layer.

n be the size of each filter ($n \times n \times q$), where

q is either equal to or less than r .

P denotes the pooling size ($p \times p$).

$$F_i = \sigma \left(\sum_{j=1}^q \sum_{u=1}^n \sum_{v=1}^n W_{i,j,u,v} * X_{j,u+x-1,v+y-1} \right) + b_i$$

Where:

F_i is the i th feature map,

σ is the activation function, (Rectified Linear Unit activation function (RELU in this case))

$W_{i,j,u,v}$ are the weights

$X_{j,u,v}$ is the input image patch and b_i is the bias.

Pooling operation used by Reshi et al. (2024) can be expressed mathematically as:

$$F'_i = F_i \downarrow P$$

Where:

F'_i is the subsampled feature map and

$\downarrow P$ represents mean or maximum pooling over $p \times p$ contiguous portions.

The operations of the layer can be represented as:

$$Y_k = f \left(\sum_{i=1}^K W_{k,i} \cdot F'_i \right)$$

Where:

Y_k is the output of the k th neuron in the fully connected layer,

$W_{k,i}$ are the weights, and

$f(\cdot)$ is the activation function.

The results for Reshi et al. (2024) show the accuracy of 94.5%, the sensitivity of 91.8%, and the specificity of 94.2%. It focuses on early pregnancy and

comprehensive framework, but has high computational requirements.

A technique based on RF and GBM-based classification was presented by Do et al. (2024) that uses 1035 individuals; with ethnicity of Vietnamese; and the location is Vietnam as shown in Figure 8. The most widely used learning metric in classification problems is accuracy. In order to minimize overfitting while preserving performance, a grid search approach was employed to minimize the accuracy difference between testing and validating results, keeping both at the greatest value.

The model presented by Do et al. (2024) was fine-tuned using the following hyper-parameters:

K-Nearest Neighbours: When creating a prediction or classification, k is the number of nearest neighbours to take into account.

SVM: C regulates the penalty for data points that fall inside the margin or on the incorrect side of the decision boundary.

RF:

m try regulates how many features (variables or predictors) are chosen at random as potential splitting candidates at each tree node as the tree grows.

The number of trees in the model is denoted by n_{tree} .

max_depth (depth) is the tree's maximum depth.

XGBoost

The number of decision trees (weak learners) that are successively added to the ensemble is determined by n_{rounds} .

min_child_weight (mcw) determines the minimal total instance weight required for a child.

The percentage of training data that is randomly sampled during each boosting round is managed by the $subsample$ (ssp).

When constructing each tree, the fraction of variables to be randomly sampled is determined by $colsample_bytree$ (csb).

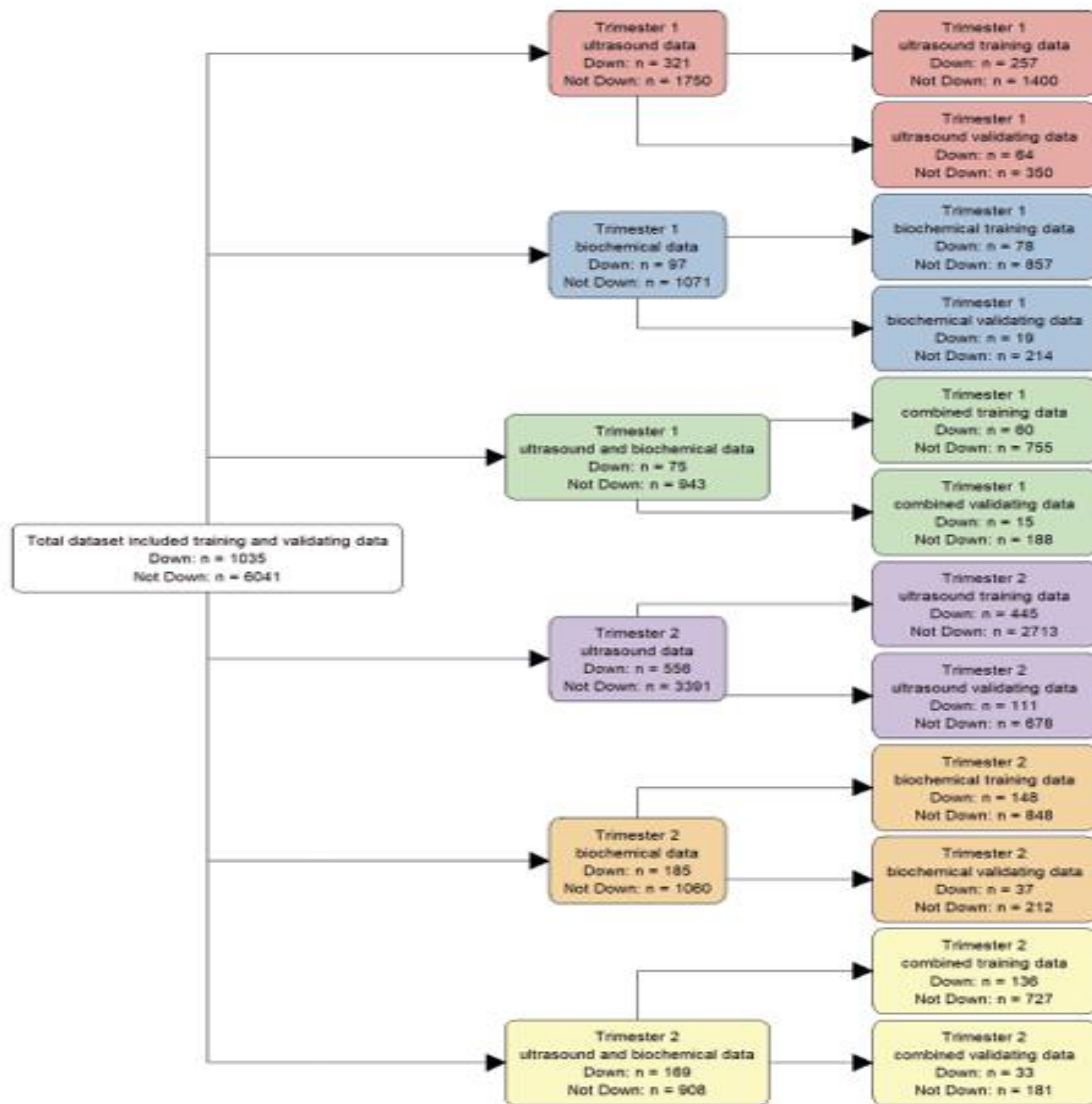


Figure 8: Overview of data Subsets Constructed for Testing and Validation, Vietnam National Hospital of Obstetrics and Gynecology, 2012 – 2022 (Do et al., 2024)

max_depth (xgbdepth) determines each tree's maximum depth inside the ensemble. The step size is determined by the eta (learning rate) or learning rate used in the slope boosting procedure.

Figure 9 shows the training and evaluating machine learning models for each of six data subsets, Vietnam National Hospital of obstetrics and gynecology for 2012 – 2022

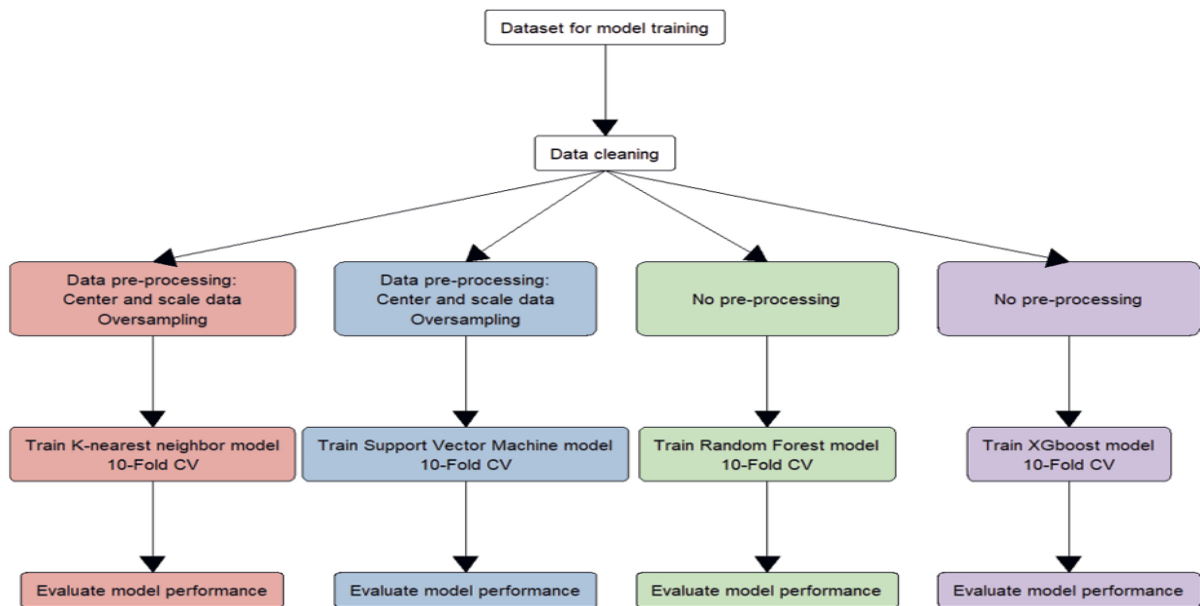


Figure 9: Training and Evaluating Machine Learning Models for each of Six Data subsets, Vietnam National Hospital of Obstetrics and Gynecology, 2012 – 2022 (Do et al., 2024)

As shown in Figure 10, Mavaluru et al. (2024) presented a research work that explores the incorporation of medical and information technology, mainly concentrating on the use of data analytics and deep learning methods in medical image processing. The research work focused specifically to addresses the diagnosis and prediction of fetal situations, including Down Syndrome (DS), through the investigation of ultrasound images. It proposes

progressive practices, integrating erudite deep learning designs and data augmentation methods to advance fetal diagnosis. The results for Do et al. (2024) show that the accuracy is 90%, the sensitivity is 83.2%, and the specificity is 81.5%. the technique has integration of multi-modality data and adaptations to diverse populations, but high computational resources and dependence on data quality.



Figure 10: algorithmic flow outlines a typical sequence for fetal growth detection (Mavaluru et al., 2024)

Mavaluru et al. (2024) used 1075 images; the mother's ages range: 28–35 years; the ethnicity is Indian; and location: India Feature extraction and segmentation approaches with SVM, Accuracy = 98.2%, sensitivity = 90.7%, and specificity = 92.8%, Integration of fuzzy gradient vector flow and wavelet-based segmentation, but limited generalization and segmentation challenges.

Mavaluru et al. (2024) presented two major algorithms as shown in Algorithms 1 and 2. Algorithm 1 is for pseudo code of fetal health classification from a medical image, while Algorithm 2 presented a high-level pseudo-code algorithm outlining the key steps involved.

Algorithm 1

Proposed model.

// Pseudo code for Fetal Health Classification from a Medical Image

// Start

START

 // Load the acquired medical image

 image = LOAD MEDICAL IMAGE("path to image")

 // Denoise the image

 denoised_image = DENOISE IMAGE(image)

 // Binarize the image for further analysis

 binary_image = BINARIZE IMAGE(denoised_image)

 // Segment the fetal parameters from the binarized image

 segmented_fetal_parameters = SEGMENT FETAL PARAMETERS(binary_image)

 // Measure the fetal parameters

 measured_parameters = MEASURE FETAL PARAMETERS(segmented_fetal_parameters)

 // Extract features from the measured parameters

 features = EXTRACT FEATURES(measured_parameters)

 // Load the Artificial Neural Network (ANN) model

 ann_model = LOAD ANN MODEL("path to model")

 // Predict the condition of the fetus using the ANN model

 prediction = ann_model.PREDICT(features)

 // Determine if the fetus is normal or abnormal based on the prediction

 IF prediction == "Normal" THEN

 OUTPUT "Normal fetus"

 ELSE IF prediction == "Abnormal" THEN

 OUTPUT "Abnormal fetus"

 END IF

// End

END

Algorithm 2

High-level pseudo-code algorithm outlining the key steps involved.

import NumPy as np

import TensorFlow as tf

from sklearn.model_selection import train_test_split

from data_preprocessing import preprocess_data

from deep_learning_model import build_model

from evaluation_metrics import calculate_metrics

 # Load and preprocess the fetal ultrasound images and associated labels

 X, y = preprocess_data('data_directory')

 # Split the dataset into training, validation, and test sets

 X_train, X_val, X_test, y_train, y_val, y_test = train_test_split(X, y, test_size=0.2, random_state=42)

 # Create a deep learning model for fetal diagnosis

 model = build_model(input_shape=X_train[0].shape)

 # Train the deep learning model on the training data

 model.fit(X_train, y_train, epochs=10, batch_size=32, validation_data=(X_val, y_val))

 # Evaluate the trained model on the test set

 loss, accuracy = model.evaluate(X_test, y_test)

 # Make predictions on new fetal ultrasound images

 new_image = load_and_preprocess_new_data('new_image_path')

 prediction = model.predict(new_image)

 # Calculate metrics such as accuracy, precision, recall, and F1-score

 metrics = calculate_metrics(y_true, y_pred)

 # Visualize the results, such as segmented images or diagnostic reports

 # Deploy the model in a clinical setting for real-world fetal diagnosis

 # Summarize the findings and the potential impact of the algorithm on fetal diagnosis

Alonso et al. (2025) presented a novel approach for screening women in their first trimester of pregnancy to identify those at high risk of having a child with Down syndrome (DS), using machine learning algorithms. The models incorporate a set of input variables, including demographic variables such as

maternal age, weight, ethnicity, smoking status, and diabetes status, as well as laboratory variables like nuchal translucency (NT), pregnancy-associated plasma protein-A (PAPP-A), and beta-human chorionic gonadotropin hormone (B-HCG) levels.

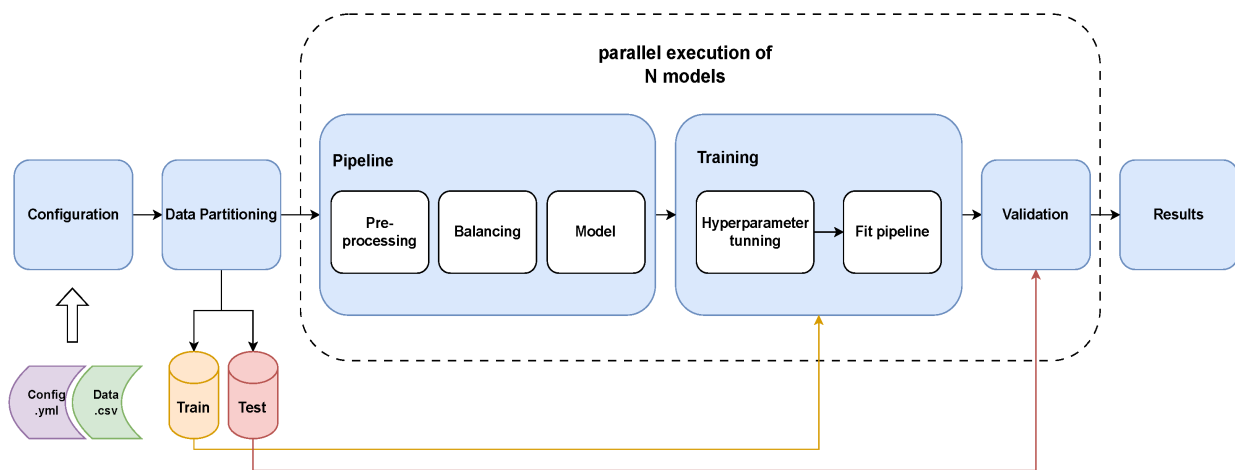


Figure 11. Machine learning model training workflow (Alonso et al., 2025)

Figure 11 depicts the complete process for training and validating the machine learning models. The workflow begins with the input of the configuration file and the dataset. The data are then split into training and test sets. The training set undergoes a series of pipeline steps: pre-processing (including scaling and encoding), balancing (using SMOTE and under sampling), and model fitting. Hyperparameter optimization is performed to tune the model parameters. The trained model's performance is then evaluated using the held-out test set, and the final performance metrics are reported. As shown in Figure 11, the components are:

Splitting

The primary purpose of the splitting step used by Alonso et al. (2025) is to prevent overfitting, a phenomenon where a model performs well on the data it has already seen but poorly generalizes to unseen data. In this process, the dataset is divided into two subsets: a “training data” subset, comprising 80% of the original samples, and a “test data” subset, containing the remaining 20% of the original dataset. The test data are put aside to validate the resulting models with unseen data at the end of the training phase. The splitting process involves (1) shuffling all samples to eliminate any potential bias caused by the original order and (2) maintaining the original proportion of the outcome variable in both subsets using stratified splits.

Pipeline Building

Prior to the training stage, a pipeline of transformers and a final estimator is constructed (Alonso et al., 2025). This pipeline is used in both the training and validation steps, ensuring that all transformations are consistently applied to the training, validation, and prediction data. As shown in Figure 11, the pipeline comprises three steps (Alonso et al., 2025): pre-processing, balancing, and modeling.

Pre-processing: This pipeline step distinguishes between categorical and numerical columns. Numerical columns are standardized by scaling them to have a mean of 0 and a standard deviation of 1. Categorical columns, on the other hand, are encoded using the One-Hot Encoding technique. This technique creates new categorical columns for each unique value and assigns a binary value of 1 or 0 based on the presence or absence of the corresponding value.

Balancing: Classification models typically perform poorly on such datasets because they tend to over-predict the most prevalent class. To address this issue, balancing techniques are applied within the pipeline. SMOTE (Synthetic Minority Oversampling Technique) is used to increase the number of positive samples, while random under-sampling is used to balance the dataset. To achieve a balanced sample, SMOTE is systematically applied to each training set until the positive samples represent 5% of the total, followed by a random reduction in negative samples.

Model: The final step in the pipeline is the insertion of the classification model, which is used for training or predicting new samples after the pipeline is fitted.

Model Training

Once the pipeline is constructed, the model is ready for training. K-fold cross-validation, a robust statistical technique, is employed to evaluate the performance of machine learning models and to assess how well they are expected to generalize to unseen data. In this method, the training data are partitioned into k equally sized subsets or folds. The model is trained k times, each time using $k-1$ of the folds as the training set and the remaining fold as a validation set. This process provides k different estimates of the model's performance on independent data, which are then averaged to obtain a more reliable overall evaluation. The pipeline ensures that data preparation before model fitting is performed exclusively on the training data within each fold, preventing data leakage and optimistic model skill estimation. In this study, k is set

to 10, and 10-fold cross-validation is conducted over five repetitions to determine the hyperparameters, providing a more robust approach and mitigating the effects of randomness in the algorithms and data partitions.

Validation

The final step of the training pipeline is to validate the models using the test subset generated in the splitting phase, which remained unseen by any of the models during training. The pipeline created earlier manages the data preparation needed to transform the test set to match the trained set, using the same encoding for categorical columns and standardizing numerical columns with the fitted values from the training phase. The trained classification algorithms achieved ROC-AUC values between 0.970 and 0.982, with sensitivity

and specificity of 0.94. The results indicate that machine learning techniques can effectively predict Down syndrome risk in first-trimester screening programs.

Yalçın et al. (2025) proposed a method that transforms one-dimensional (1D) patient data—including features such as nuchal translucency (NT), human chorionic gonadotropin (hCG), and pregnancy-associated plasma protein A (PAPP-A)—into two-dimensional (2D) Aztec barcode images, enabling advanced feature extraction using transformer-based deep learning models.

Figure 12 illustrates an example of how line data were transformed into Aztec barcodes, while Figure 13 shows sample images categorized by DS risk levels: high, medium, and low.

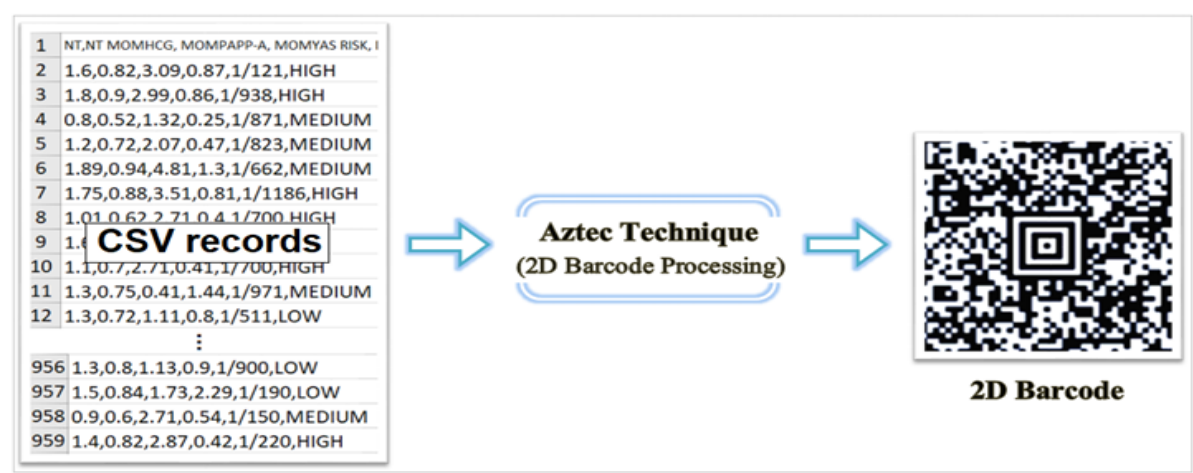


Figure 12. Application of the Aztec barcode technique to raw numerical data from the original dataset (Yalçın et al., 2025)

As shown in Figure 12 the resulting 2D Aztec code visually encodes structured input features (e.g., hCG-MoM, PAPP AMoM,NTMoM,etc.) into a fixed-size image representation. This transformation enables the use of vision-based transformers for classification tasks in prenatal DS risk assessment.

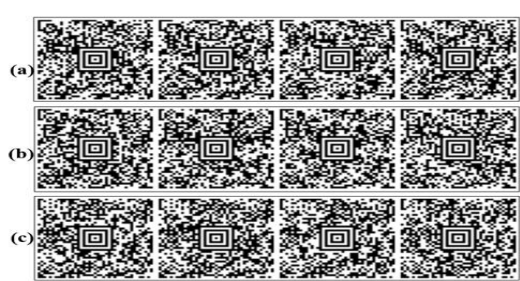


Figure 13. Sample Aztec-coded image set generated after pre-processing raw patient data (Yalçın et al., 2025).

As shown in Figure 14 each image corresponds to a different risk class used in the classification task: (a) high risk, (b) low risk, (c) medium risk. These visual representations are input to the vision-based transformer models for automated classification in the context of prenatal DS risk evaluation.

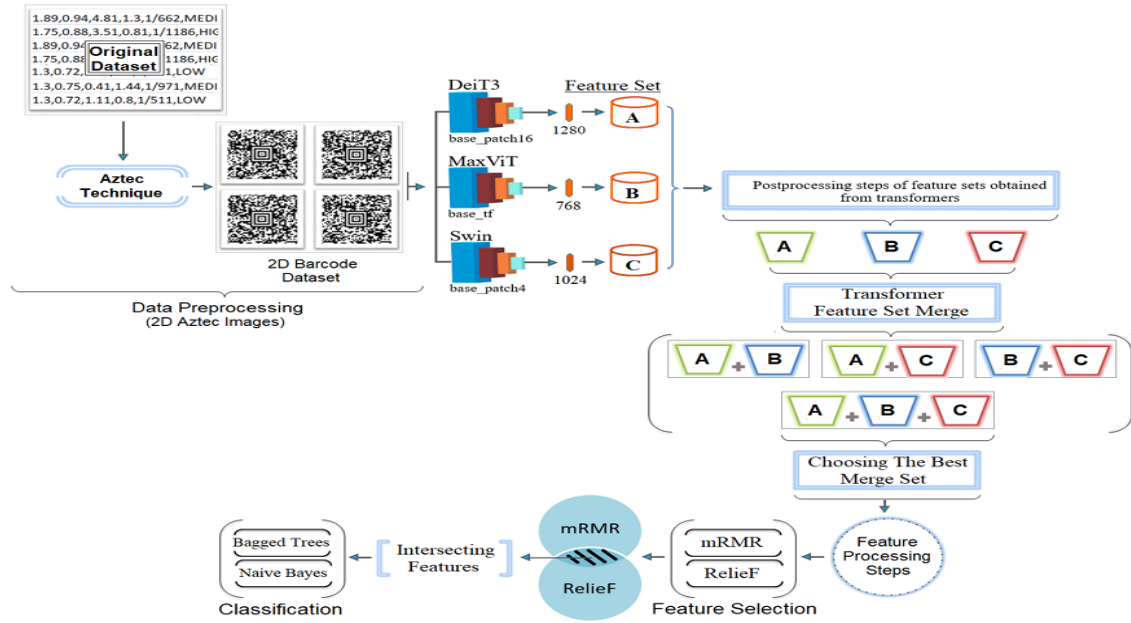


Figure 14. Model design of hybrid approach proposed for this study (Yalçın et al., 2025).

The method by Yalçın et al. (2025) integrates advanced artificial intelligence techniques and consists of five main stages: data preprocessing, transformer model training, feature extraction, feature fusion, feature selection, and classification. In the preprocessing stage, 1D patient data stored in CSV format is transformed into 2D barcode images using the Aztec encoding technique. The overall workflow of the proposed hybrid AI system is illustrated in Figure 4. The results suggest that the proposed hybrid AI method offers a promising and resource-efficient solution for DS risk assessment in first trimester screening. However, further comparative studies are recommended to validate its performance in broader clinical contexts. The approach by Yalçın et al. (2025) achieved up to 100% classification accuracy using the Naive Bayes classifier with 1250 features selected by ReliefF and 527 intersecting features from mRMR. By selecting a smaller but more informative subset of

features, the system significantly reduced hardware and processing demands while maintaining strong predictive performance

Yalçın et al. (2025) proposed a novel diagnostic approach for DS by enhancing the existing bio chemical markers (hCG and PAPP-A) and biophysical measurements (NT) obtained from the combined FTS test, which is performed during the first three months of pregnancy for early detection. Figure 15 illustrates the overall workflow of the proposed architecture. This study proposes an enhanced machine learning (ML) approach for predicting Down Syndrome (DS) risk using first-trimester screening (FTS) data. They presented a predictive ML model was created, and Hyperparameter Tuning (HT) was applied to optimize it for performance.



Figure: 15 The flowchart of the proposed system (Yalçın et al., 2025)

The results of the system presented by Yalçın et al. (2025) demonstrate the model's potential for reliable DS risk prediction, offering a more efficient and less invasive alternative to traditional diagnostic procedures. By enhancing early risk detection, the method could reduce unnecessary referrals for invasive tests like amniocentesis, thereby minimizing patient anxiety and potential complications. Overall, the study contributes to the development of intelligent, data driven solutions for prenatal care.

Chen and Lin (2026) developed a deep learning model as shown in Figure 16 for comparison with the Fetal

Medicine Foundation (FMF) algorithm for first-trimester Down syndrome risk classification in East Asian pregnant women. Using the FMF algorithm-derived risk classification as the reference standard, the original set of FMF input parameters was reduced to the five most informative indicators based on permutation importance analysis and clinical relevance. The five retained indicators—maternal age, age-related background risk of Down syndrome, nuchal translucency thickness, PAPP-A, and free β -hCG—were identified as the most influential features, consistent with established clinical knowledge.

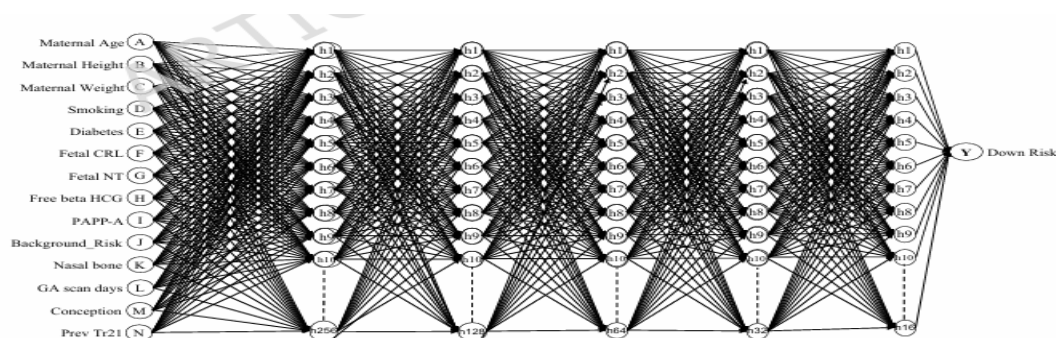


Figure 16. Proposed customized deep learning model based on an artificial neural network (ANN) architecture (Chen and Lin, 2026).

The present findings from Chen and Lin (2026) should be interpreted as an FMF concordance study between the ANN model and standardized FMF-based screening. Overall, as a concordance study, the proposed ANN model using a reduced set of key features can approximate FMF-based risk classification. But further study is required to demonstrate the feasibility of the model in the real-world situation and to evaluate its clinical value using datasets with confirmed trisomy 21 outcomes

Jiang et al, (2026) proposes an interpretable computational framework for prenatal risk assessment and testing-time optimization by integrating ensemble learning, BMI-stratified analysis, and uncertainty evaluation. As shown in Figure 17, for male-fetus samples, Y-chromosome-related measurements were

used as biologically meaningful proxies for fetal signal. Linear regression, polynomial regression, random forest regression, and least-squares boosting were evaluated using cross-validated root mean squared error and coefficient of determination. The estimated optimal sampling ages were approximately 10 weeks, 14 weeks + 5 days, and 23 weeks + 3 days for the low-, medium-, and high-BMI strata, respectively, with greater in stability observed in the high-BMI stratum. For female-fetus samples, aneuploidy screening was formulated as a binary classification task. Random forest substantially outperformed logistic regression, with an ROC-AUC of 0.884 versus 0.538 and an average precision of 0.668 versus 0.070, and supported a decision threshold of 0.1437.

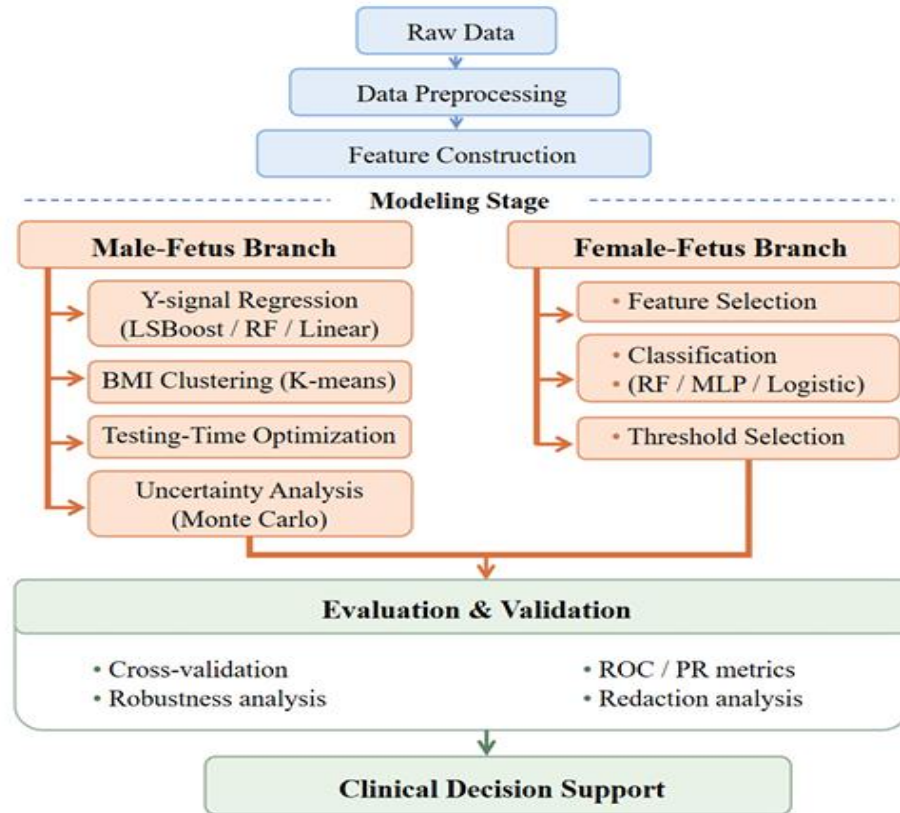


Figure 17. Simplified methodological workflow of the present study (Jiang et al, 2026).

Figure 17 summarizes the main methodological steps. After preprocessing and feature construction, the analysis was organized into two branches. The male-fetus branch included Y-signal regression, BMI stratification, testing-time optimization, and uncertainty assessment. The female-fetus branch included feature selection, abnormality classification, and threshold determination. These findings from **Jiang et al, (2026)** suggest that medical big data-driven methods can improve prenatal risk assessment, testing-time optimization, and uncertainty-aware decision support in prenatal screening.

3.0 PROPOSED IPSO BASED ON IMPROVE CLOUD PARTICLE SWARM OPTIMIZATION (ICPSO) ALGORITHM

The proposed IPSO is based on an Improve Cloud Particle Swarm Optimization (ICPSO) algorithm presented by Cai (2022). As shown in Figure 18, PSO is a novel type of bionic optimization algorithm that is like to that of genetic algorithm (GA) and is a repetitive optimization algorithm. It starts by initiating a set of random solutions like GA and repeatedly keep searches for the best solution. Convergence analysis of particle herd optimization algorithms is the basis of PSO algorithms.

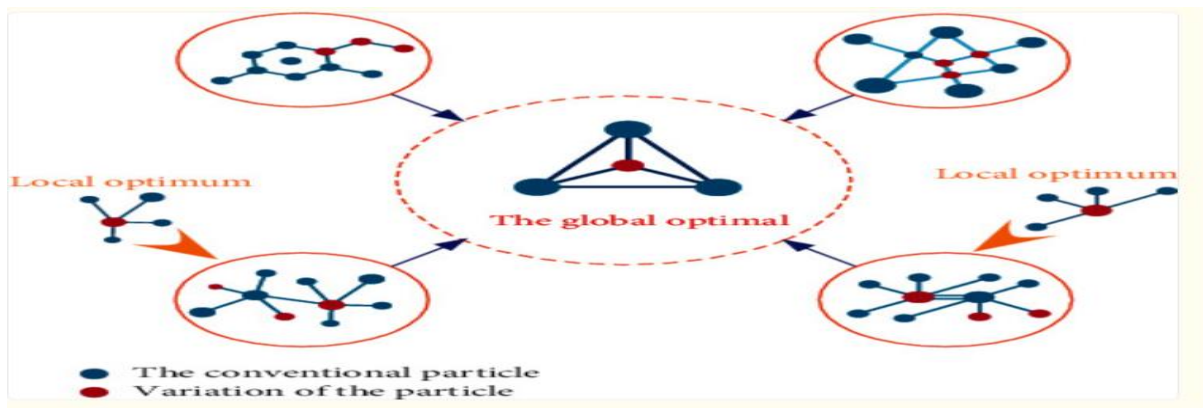


Figure 18: Particle Swarm Optimization (Cai, 2022))
The Proposed IPSO Algorithm

The IPSO algorithm uses two measures to improve the existing PSO. The definitions are:

Definition 1:

(mutation). Given the thresholds N and K in advance, when the global extreme value has not evolved for N consecutive generations or the amplitude of the evolution process is less than k , it is considered that the particles fall into the local optimum, and all particles are mutated through the normal cloud generator according to the global extreme value.

Definition 2:

One-dimensional simple cloud operator $ArForward(C(Ex, En, He))$ is a mapping of π that converts the The IPSO algorithm is:

1. Initialize the population. That is, initialize the position of each particle, individual extreme value $PBEST$, local extreme value $GBEST$, and so on.
2. Calculate the fitness value for each particle and update $Pbest$ and $Gbest$.
3. Judge whether the mutation threshold n is reached. If it is reached, the mutation operation is carried out according to Definition 1. Let the local best (minimum) of all particles be $Gbest$ and make $ex = Gbest$, $en = 2gbest$, $he = en/10$ in normal cloud computing a $(C(ex, en, he))$. According to Definition 2, the normal cloud generator completes the mutation operation of all particles and fails to reach the mutation threshold (4).
4. Evolve each particle. Let the individual minimum of particle I be $Pbest$, let $ex = Pbest$, $en = 2pbest$, $he = en/10$ in normal cloud computing a $(C(ex, en, he))$, generate a new particle J according to the normal cloud generator in Definition 2, and let $I = J$ to complete the evolution operation.
5. If the iteration limit is reached, the $Gbest$ output will end; otherwise, go to (2).

4.0 CONCLUSION

The proposed IPSO is based on an Improve Cloud Particle Swarm Optimization (ICPSO) algorithm presented by Cai (2022). The proposed research includes the analysis of algorithm mechanism, the improvement of the existing algorithm performance and the expansion of algorithm application in predicting down syndrome babies. IPSO algorithm is created based on cloud digital feature coding to improved the active performance of cloud particles. This paper proposes two major improvement measures to improve the existing algorithm search abilities, population variety, and algorithm integration rapidity and correctness. The proposed method will be experimented in order show that the improved method is effective compared with the existing methods.

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References

Alonso, E., Beristain, A., Burgos, J., & Gurrutxaga, I. (2025). Comparison of machine learning algorithms to predict down syndrome during the screening of the first trimester of pregnancy. *Applied Sciences*, 15(10), 5401.

general properties of quality concepts into digital representations. $C \rightarrow \Pi$. The following conditions are met:

$$\Theta = \{t_i | \text{Norm}(En, He), i = 1, \dots, N\}.$$

$$X = \{x_i | x_i, t_i \in \Theta, i = 1, \dots, N\}.$$

$$\prod = \{(x_i, y_i) | x_i \in X, t_i \in \Theta, y_i = \exp(-(x_i - Ex)^2 / (2t_i^2))\}.$$

Anitha Elavarasi, S.; Jayanthi, J. Role of Machine Learning and Deep Learning in Assisting the Special Children's Learning Process. *J. Algebr. Stat.* 2022, 13, 2327–2334.

Cai, M. (2022). An improved particle swarm optimization algorithm and its application to the extreme value optimization problem of multivariable function. *Computational intelligence and neuroscience*, 2022(1), 1935272. <https://doi.org/10.1155/2022/1935272>

Chen, Y. T., & Lin, Y. S. (2026). Deep learning-based first-trimester down syndrome risk prediction in East Asian pregnant women using simplified clinical indicators. *Scientific Reports*.

Chen, Y. T., Chen, G. J., & Lin, Y. S. (2025). Machine learning-based prediction of first trimester down syndrome risk in East Asian populations. *Risk Management and Healthcare Policy*, 1109–1120.

Do, H.D.; Allison, J.J.; Nguyen, H.L.; Phung, H.N.; Tran, C.D.; Le, G.M.; Nguyen, T.T. Applying machine learning in screening for Down Syndrome in both trimesters for diverse healthcare scenarios. *Heliyon* 2024, 10, e34476. [CrossRef]

Geremek, M.; Szklanny, K. Deep learning-based analysis of face images as a screening tool for genetic syndromes. *Sensors* 2021, 21, 6595. [CrossRef]

- Islam, T.U.; Shaikh, T.A. A lightweight attention-based deep learning facial recognition system for multiple genetic syndromes. *Int. J. Data Sci. Anal.* 2024, 29, 1–19.
- Jiang, C., Li, W., Geng, Z., Shang, H., & Li, Y. (2026). Medical Big Data-Driven Prenatal Risk Assessment and Testing-Time Optimization. *Mathematical and Computational Applications*, 31(3), 76.
- Kaur, S., & Singh, J. (2023). Enhancing medical image analysis and disease surveillance in healthcare: A study on pso-aco optimization using swarm intelligence. 2023 2nd international conference on computational modelling, simulation and optimization <https://doi.org/10.1109/ICCMO59960.2023.00058> for (ICCMO) (pp. 269–276). IEEE.
- Leghari, I.M.; Ali, S.A. Artificial Intelligence Techniques to improve cognitive traits of Down Syndrome Individuals: An Analysis. *Int. J. Softw. Sci. Comput. Intell.* 2023, 15, 1–11. [CrossRef]
- Maivaluru, D.; Ravula, S.R.; Auguskani, J.P.L.; Dharmarajlu, S.M.; Chellathurai, A.; Ramakrishnan, J.; Mugaiahgari, B.K.M.; Ravishankar, N. Advancing Fetal Ultrasound Diagnostics: Innovative Methodologies for Improved Accuracy in Detecting Down Syndrome. *Med. Eng. Phys.* 2024, 126, 104132. [CrossRef] [PubMed]
- Pooch, E.H.P.; Alva, T.A.P.; Becker, C.D.L. A Computational Tool for Automated Detection of Genetic Syndrome Using Facial Images. In *Intelligent Systems, Proceedings of the 9th Brazilian Conference, BRACIS 2020, Rio Grande, Brazil, 20–23 October 2020, Proceedings, Part I 9*; Springer International Publishing: Berlin/Heidelberg, Germany, 2020; pp. 361–370.
- Porras, A.R.; Rosenbaum, K.; Tor-Diez, C.; Summar, M.; Linguraru, M.G. Development and evaluation of a machine learning based point-of-care screening tool for genetic syndromes in children: A multinational retrospective study. *Lancet Digit. Health* 2021, 3, e635–e643.
- Qin, B.; Liang, L.; Wu, J.; Quan, Q.; Wang, Z.; Li, D. Automatic identification of down syndrome using facial images with deep convolutional neural network. *Diagnostics* 2020, 10, 487. [CrossRef] [PubMed]
- Raza, A.; Munir, K.; Almutairi, M.S.; Sehar, R. Novel transfer learning based deep features for diagnosis of down syndrome in children using facial images. *IEEE Access* 2024, 12, 16386–16396.
- Reshi, A.A.; Shafi, S.; Qayoom, I.; Wani, M.; Parveen, S.; Ahmad, A. Deep Learning-Based Architecture for Down Syndrome Assessment During Early Pregnancy Using Fetal Ultrasound Images. *Int. J. Exp. Res. Rev.* 2024, 38, 182–193.
- Taidi, L.; El Khamlichi, S. A comprehensive review of artificial intelligence techniques for timely and accurate prediction of Down syndrome. In *Agile Security in the Digital Era*; CRC Press: Boca Raton, FL, USA, 2024; pp. 179–194.
- Wang, J.; Liang, B.; Zhao, L.; Chen, Y.; Fu, W.; Yu, P.; Chen, H.; Wang, H.; Xie, G.; Wu, T.; et al. Multiple genetic syndromes recognition based on a deep learning framework and cross-loss training. *IEEE Access* 2022, 10, 117084–117092.
- Yalçın, E., Aslan, S., Toğaçar, M., & Demir, S. C. (2025). A hybrid artificial intelligence approach for down syndrome risk prediction in first trimester screening.
- Yalçın, E., Koç, T. K., Aslan, S., Demir, S. C., Aykut, S., & Sucu, M. (2025). Hyperparameter optimization based machine learning approach for early diagnosis of fetal genetic disorders. *Discover Computing*, 28(1), 274.
- Yekdast, R. An intelligent method for down syndrome detection in fetuses using ultrasound images and deep learning neural networks. *Comput. Res. Prog. Appl. Sci. Eng.* 2019, 5, 92–97.
- Zhang, H.; Jiang, Y.; Dai, S.; Li, L.; Hu, X.; Liu, R. Application of Intelligent Algorithms in Down Syndrome Screening During Second Trimester Pregnancy. *World J. Clin. Cases* 2021, 9, 4573–4584.