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RESEARCH ARTICLE

Novel Transfer Learning Based Deep Features for Diagnosis of Down Syndrome in Children Using Facial Images

ALI RAZA[1], KASHIF MUNIR[2], MUBARAK S. ALMUTAIRI[3], AND RUKHSHANDA SEHAR[1]

[1]Institute of Computer Science, Khwaja Fareed University of Engineering and Information Technology, Rahim Yar Khan 64200, Pakistan
[2]Institute of Information Technology, Khwaja Fareed University of Engineering and Information Technology, Rahim Yar Khan 64200, Pakistan
[3]College of Computer Science and Engineering, University of Hafr Al Batin, Hafr Al Batin 31991, Saudi Arabia

Corresponding authors: Ali Raza (ali.raza.scholarly@gmail.com) and Kashif Munir (kashif.munir@kfueit.edu.pk)

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ABSTRACT Down syndrome is a chromosomal condition characterized by the existence of an additional copy of chromosome 21. This genetic anomaly leads to a range of developmental challenges and distinct physical characteristics in affected children. Children with Down syndrome often exhibit specific craniofacial proportions, such as a relatively shorter midface and broader facial width. These distinct facial features, including a flat nasal bridge, almond-shaped eyes, and a small and somewhat flattened head, can serve as valuable indicators for early diagnosis and intervention. This study aims at the early diagnosis of Down syndrome using an advanced neural network approach. We used 3,009 facial images of children with Down syndrome and healthy children taken from the age group range of 0 to 15 for conducting our research experiments. We proposed 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. This unique VNL-Net feature extraction initially extracts spatial features from input image data. Then, the ensemble feature set of NMF and LGBM is extracted from spatial features. We 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. We 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.

INDEX TERMS Down syndrome, machine learning, transfer learning, ensemble features, deep learning.

I. INTRODUCTION

Down syndrome, also known as trisomy 21, is one of the most prevalent chromosomal disorders affecting children worldwide [1]. Down syndrome is characterized by an extra copy of chromosome 21, which gives rise to a distinct set of physical and cognitive traits [2]. The physical traits include distinctive facial features, such as almond-shaped eyes, a flattened nasal bridge, and a protruding tongue, which serve as critical diagnostic indicators. This condition has been the subject of extensive research aimed at unraveling its underlying genetic mechanisms [3] and comprehending its multifaceted impact on child development.

Down syndrome in children is the main cause of growth delay and is increasing day by day. A recent study reported that in 12–14 per 10,000 live births in Sweden [4]. In the United States (US), it is reported that there are 8.27 individuals with Down syndrome per 10,000 of the population [5]. This represents 270,600 individuals with Down syndrome living in the US, based on the 2018 US population size. There

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is a need for an automated early diagnosis of Down syndrome to overcome these mortality rates.

In recent years, there has been a burgeoning interest in leveraging advanced computational approaches for the early diagnosis of Down syndrome, a chromosomal abnormality with significant medical implications. Among these techniques, machine learning, deep learning, and transfer learning [6] have come up as powerful tools in the domain of medical image analysis. Specifically, researchers have turned to neural networks, a class of computational models inspired by the structure and functioning of the human brain [7], to analyze facial image data for Down syndrome detection. Machine learning algorithms enable the extraction of intricate patterns and features from these images [8]. In contrast, deep learning architectures, characterized by multiple layers of interconnected nodes, have demonstrated remarkable capabilities in discerning subtle facial phenotypic traits associated with Down syndrome.

Transfer learning-based deep neural networks are built for the detection tasks [9]. This research proposes a novel transfer learning-based feature engineering mechanism for the effective diagnosis of Down syndrome in children. Classical approaches use facial image data for the detection of Down syndrome. Our research presents an effective transfer learning mechanism that shares the experience of a learning approach for training. Our research findings show that transfer learning mechanisms are much better than classical approaches.

Our main research contributions toward the diagnosis of Down syndrome in children are as follows:

- A novel transfer learning-based VNL-Net approach is proposed for effective feature engineering from image data. The VNL-Net method initially extracts spatial features from the image data. Subsequently, a new ensemble feature set is formed, incorporating Non-Negative Matrix Factorization and features derived from the LGBM classifier.
- We have developed two advanced deep learning models and four machine learning models using facial image data and newly created features. The performance of the applied methods is validated through K-fold cross-validation. Additionally, we have assessed the computational complexity of each method.

The remaining sections of this study are structured as follows: In Section II, an analysis of the existing literature is presented. Our proposed methodology is outlined in Section III. The outcomes of the implemented advanced methods are evaluated and compared in Section IV. The key discoveries from our investigation are summarized in Section V.

II. LITERATURE ANALYSIS

This literature analysis section aims to establish a contextual framework for the application of deep neural networks in pediatric Down syndrome diagnosis. This involves a thorough exploration of the historical evolution of image-based diagnostic techniques, emphasizing key milestones, methodologies, and technological advancements that have facilitated the integration of deep learning algorithms.

In this research [10], the authors proposed a novel experimental finding for detecting obstructive sleep apnea in subjects with Down syndrome. The experiment included 64 participants aged between 3 and 35 years. The down syndrome dataset comprised medical histories, vital signs, and physical exams. For an apnea-hypopnea index (AHI) > 1/hTST and AHI > 5/hTST, the proposed model showed positive and negative predicted values of 73.7% and 60%, respectively. The performance scores of this research were found to be low.

In this review research [11], the authors analyzed different machine learning algorithms to identify Obstructive Sleep Apnea (OSA). The data used in this experiment was collected by two researchers from the Web of Science and Scopus, employing various electronic research methods. Finally, 19 studies with 4767 separate pediatric sleep investigations were conducted. Machine learning demonstrated improved diagnostic performance, achieving its best results for AHI = 10 e/h, with 0.652 sensitivity, 0.931 specificity, and a 0.940 area under the SROC curve.

In this research [12], an ultrasound (US) imaging system approach is proposed for detecting Down syndrome due to its safety, cost-effectiveness, and non-invasiveness. The Mediscan Fetalcare Research Foundation from India provides the dataset used in this research. The US fetal image dataset contains 100 fetuses, with 50 healthy and 50 Down syndrome fetuses between 11-14 weeks of gestation. By employing different Deep Learning models, including a Convolutional Neural Network (CNN) and Visual Geometry Group (VGG-16), it was found that the most effective algorithm achieving the highest accuracy is VGG-16, with a 91% accuracy.

In this research [13], the authors identify the behaviors of children and parents with Down syndrome using human activity recognition techniques. They utilized advanced technologies and video-based data to analyze the activities and behavior of individuals with Down syndrome. The experimental findings from the video dataset, employing the deep learning model C3D, demonstrated an accuracy of 85%. The physical interventions were also detected.

In this research [14], a prenatal screening method for predicting Down syndrome was proposed, aiming to enhance the accuracy of diagnosis. This was achieved by employing a multi-branch CNN model integrated with a feature rearrangement technique [19], [20]. To configure the CNN model, the suggested feature rearrangement strategy utilized feature grouping and Pearson correlation testing. The experiment yielded promising results, with a Recall of 0.9023, an F1-score of 0.8969, and a balanced accuracy of 0.9314 when utilizing the CNN model.

In this research [15], the authors proposed a face recognition method to diagnose facial diseases. They used a limited dataset to perform computer-aided face diagnosis

TABLE 1. The previously published studies literature summary is comparatively analyzed.

Ref	Year	Dataset	Proposed Technique	Performance Score	Limitations
[10]	2022	Down syndrome dataset	Ahi	73%	The poor generalization abilities.
[11]	2021	Different datasets analysis	ML	94%	The classical Machine learning methods were used.
[12]	2022	Us fetal images	DL , VGG16b	91%	The classical Deep learning methods were utilized.
[13]	2022	Video dataset	DL model C3D	85%	The poor generalization abilities.
[14]	2023	Prenatal screening data	CNN	93%	Again the classical Deep learning methods were used.
[15]	2020	Disease-Specific Face (DSF)	DL, CNN	90%	The authors used classical Deep learning methods.
[16]	2020	CASIA-WebFace datasets	DL, CNN	95%	The classical Deep learning methods were used.
[17]	2013	Image base Dataset	ML, SVM	94%	The classical Deep learning methods were used.
[17]	2013	Down syndrome dataset	ML, SVM	97%	The research employed Classical Feature engineering approaches.
[18]	2023	Down syndrome students dataset	DL, CNN	85%	The poor generalization abilities.

for both single (beta-thalassemia) and multiple diseases (beta-thalassemia, hyperthyroidism, Down syndrome, and leprosy). In this experiment, the DSF dataset was utilized, specifically designed for special facial features, containing 350 unique image features. The results of the further analysis demonstrated that the CNN (Convolutional Neural Network) was the most effective Deep Learning Model algorithm, achieving 90% accuracy.

In this research [16], Down syndrome identification using facial features is proposed. The data is divided into two parts: general face features and detailed face features. The data used in this experiment is the CASIA-Web FACE dataset, which is a publicly available dataset including 493,750 images with 10,562 individuals. A deep learning approach, named Convolutional Neural Network (CNN), was employed in this experiment. In Down Syndrome identification, the deep convolutional neural network achieved an accuracy of 95.87%, a recall of 93.18%, and a precision of 97.40%.

In this research [17], the authors proposed a policy based on machine learning algorithms to diagnose Down Syndrome. The authors used a modified model to detect facial landmarks. From these landmarks, geometric and texture features were extracted. The experiment utilized an image-based dataset. In this study, Down Syndrome was detected using multiple classifiers. The best performance was achieved using the Support Vector Machine (SVM) machine learning model, with an accuracy of 94.6%, precision of 93.3%, and recall of 95.5%.

In this research [17], a method is proposed to detect Down syndrome using facial recognition. In this experiment, two datasets were collected, each containing forty-eight pictures of patients—twenty-four normal and twenty-four abnormal patients. The features extracted in this study include both local and geometric features. This technique demonstrated efficient results with respect to geometric and texture features, achieving an accuracy of 97.92% along with good precision and recall for the combined features.

In this research [18], the authors proposed a system that comprises three CNN networks. Initially, facial expressions are recognized, after which DS patient photographs are used to validate the data by employing transfer learning based on the mini-Xception architecture. In the final stage, these two networks are combined to yield efficient results. For this experiment, a Down Syndrome dataset consisting of 1200 images with ages between 8 and 12 years was utilized. The CNN model was employed to achieve an accuracy of 85%. The accuracy was further increased to 91%, particularly in Down Syndrome and emotion recognition, by using an algorithm optimized through the tuning of specific hyperparameters of the network.

A. RESEARCH GAP

After conducting an extensive review of existing literature, the subsequent analysis has revealed certain areas within the research that lack sufficient investigation:

- Previously, researchers utilized classical machine learning approaches for the detection of Down syndrome in children. There is a need for advanced transfer learning approaches. Additionally, the performance scores were low for diagnosis in state-of-the-art studies.

III. PROPOSED METHODOLOGY

Our proposed innovative research methodology for the detection of Down syndrome in children is illustrated in Figure 1. Initially, we collected standard image data of children with both healthy and Down syndrome faces. Subsequently, basic image processing steps were applied to the facial image dataset. A novel feature set, based on transfer learning, was extracted from the preprocessed images. The newly obtained transfer features were then divided into training and testing sets. The training process involved using 80% of the data to train machine learning and deep learning models. Following training, the models were evaluated using the remaining 20% of the data. The model demonstrating superior performance was selected for the accurate detection of Down syndrome in children.

A. FACIAL IMAGE DATASET OF CHILDREN

This research utilized a multi-featured dataset [21] comprising 3,009 facial images of children with Down syndrome and healthy children. The facial images of children were collected from the age group ranging from 0 to 15. Sample images are illustrated in Figure 2. We also conducted a distribution analysis of images within the target classes, demonstrating that the dataset is balanced. The analysis of distributions revealed that the healthy class comprises

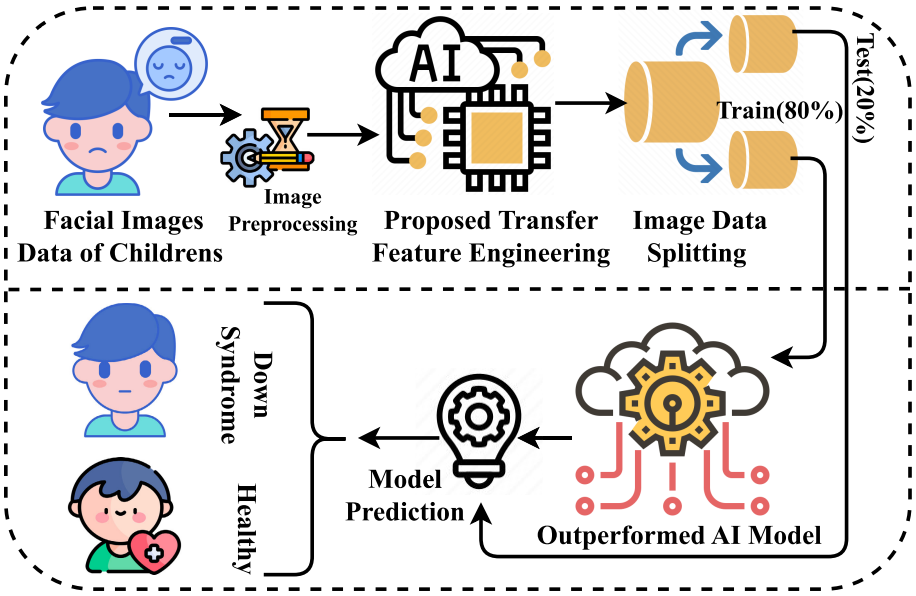


FIGURE 1. The architectural analysis of our novel proposed research methodology.

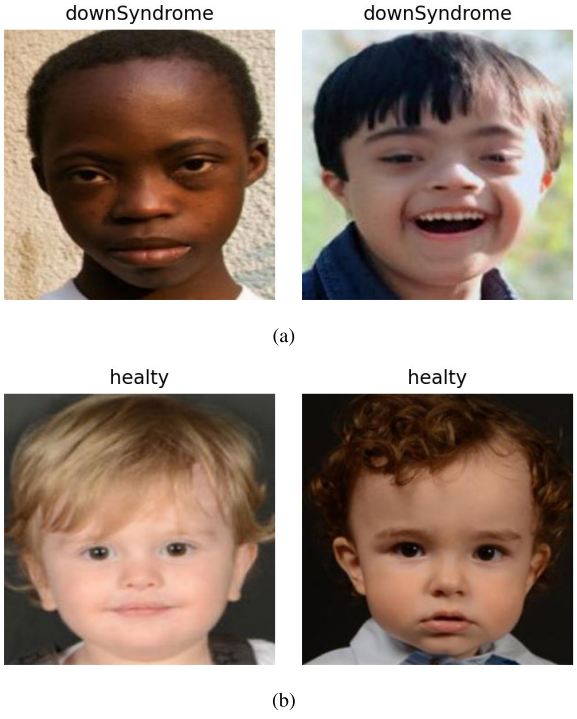


FIGURE 2. The sample images analysis with target label.

1509 image samples, while the Down Syndrome class contains 1500 image samples.

B. IMAGE PREPOSSESSING

We performed the basic image processing steps to facilitate further research experiments. Initially, we loaded facial color image data and iterated through the images in the label folder. We resized all loaded images to a uniform size during the data iteration. Subsequently, we converted the image pixel data into NumPy arrays and transformed them into tensors, ensuring consistency in image sizes. Following this, we mapped the image target labels to numerical values, assigning 'downSyndrome' as 0 and 'healthy' as 1. After completing this step, we divided the data into testing and training sets using an 80%-20% ratio.

C. NOVEL PROPOSED APPROACH

The workflow architecture of our novel proposed transfer learning-based feature engineering VNL-Net is described stepwise in Figure 3. Initially, the facial image data of children is inputted into the VGG16 approach for spatial feature extraction. A new set of 1000 spatial features is formed using VGG16. Then, features are extracted using Non-Negative Matrix Factorization (NMF) [22], [23], which identifies two non-negative matrices in the feature space. This factorization in NMF helps to select features with high variance values. Again, the newly created spatial features are input to the LGBM classifier, which extracts transfer features. Finally, the NMF features and LGBM classifier-based features are combined as an ensemble to build the applied machine learning techniques. In this research, our proposed transfer learning mechanisms help improve performance accuracy for the detection of Down syndrome in children.

D. APPLIED ARTIFICIAL INTELLIGENCE APPROACHES

Artificial Intelligence (AI) techniques for diagnosing Down Syndrome in children leverage advanced algorithms to analyze facial images precisely. These techniques employ machine learning approaches, such as convolutional neural networks (CNNs), to extract intricate features indicative of Down Syndrome. By autonomously learning and recognizing subtle patterns in facial characteristics [24], AI contributes

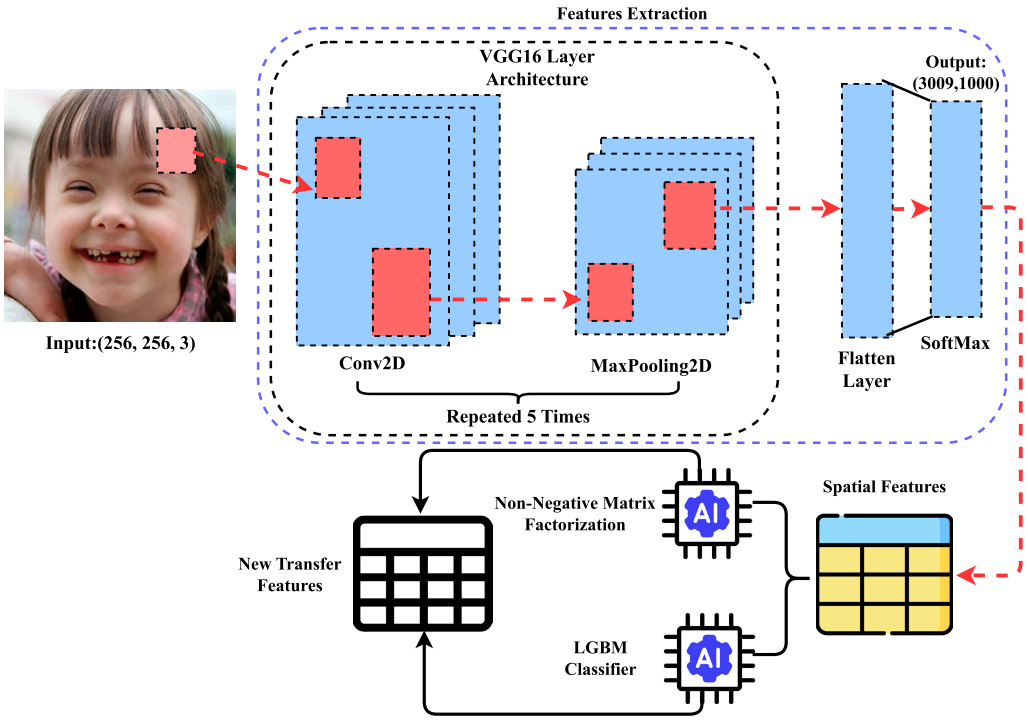


FIGURE 3. The architectural analysis of our novel proposed feature engineering approach.

to accurate and early diagnosis, enhancing the potential for timely intervention and care for affected children.

E. LIGHT GRADIENT BOOSTING MACHINE

The Light Gradient Boosting Machine (LGBM) approach [25] operates by iteratively combining weak learners, typically decision trees, to create a robust predictive model for diagnosing Down Syndrome in children using facial images. LGBM excels in handling large datasets and implements a gradient-based learning approach, optimizing the model's performance [26]. By efficiently boosting the model's accuracy through multiple iterations, LGBM enhances the classification of facial features, contributing to a more effective and reliable diagnostic tool for identifying Down Syndrome in pediatric patients. The LGBM equation for the Diagnosis of Down Syndrome in Children Using Facial Images can be represented as:

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

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.

F. K NEIGHBORS CLASSIFIER

The K Neighbors Classifier (KNC) method [27], [28] operates by identifying the proximity of data points in a feature space, classifying each point based on the labels of its k-nearest neighbors. In diagnosing Down Syndrome in children using facial images, the KNC leverages the facial features extracted from images to create a multidimensional representation. By comparing these features with those of neighboring data points, the algorithm assigns a classification label, aiding in accurately identifying individuals with Down Syndrome. This mechanism enhances the diagnostic process by leveraging the spatial relationships among facial features for effective pattern recognition. The basic equation for the KNC method can be expressed as:

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

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 ith neighbor.

G. LOGISTIC REGRESSION

Logistic Regression (LR) [29] operates as a binary classification method for diagnosing Down Syndrome in children based on facial images. By analyzing features extracted from these images, LR calculates the probability of a given image belonging to either the 'Down Syndrome' or 'Healthy' category. The model optimizes its parameters through a training process, adjusting the weights assigned to different

features to achieve accurate predictions and providing a straightforward and interpretable tool for diagnostic purposes. The LR equation for the diagnosis of Down Syndrome using facial images can be expressed as follows:

$$P(Y = 1|X) = \frac{1}{1 + e^{-(\beta_0+\beta_1X_1+\beta_2X_2+...+\beta_nX_n)}} \quad (3)$$

Here:

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

H. RANDOM FOREST

The Random Forest (RF) method [30] operates by constructing many decision trees during training, each being built on a random subset of the dataset's features. In diagnosing Down Syndrome in children using facial images, RF leverages these diverse trees to make predictions based on facial features collectively [31]. By aggregating the results from multiple trees, the RF algorithm enhances accuracy and robustness, providing a reliable mechanism for identifying patterns in facial images associated with Down Syndrome in a diagnostic context. 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.

I. CONVOLUTIONAL NEURAL NETWORK

The Convolutional Neural Network (CNN) method [32] employs a hierarchical approach to learn intricate patterns and features from facial images automatically. In diagnosing Down Syndrome in children, CNNs analyze facial features through convolutional layers, capturing low-level details and high-level representations. The network's ability to discern complex patterns within facial data enables it to extract relevant information crucial for accurate diagnosis [33]. It makes CNNs a powerful tool in medical image analysis for identifying Down Syndrome in children based on facial images. The following equation represents a basic CNN method for the diagnosis of Down Syndrome in children using facial images:

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

TABLE 2. Hyperparameter tuning analysis of applied advanced models for image analysis.

Method	Hyperparameters Tuning
LGBM	n_estimators=300, boosting_type='gbdt', num_leaves=31, learning_rate=0.1
RF	max_depth=300, criterion="gini", n_estimators=300, splitter="best"
LR	penalty='l2', tol=1e-4, max_iter=100, solver='lbfgs'
KNC	weights='uniform', n_neighbors=2, leaf_size=30, metric='minkowski'
CNN	input_shape = (256, 256,3), Dense(1), activation= 'sigmoid', optimizer='adam', loss='binary_crossentropy', metrics=['accuracy', 'Precision', 'Recall', 'AUC'], Dropout(0.6)
VGG16	input_shape = (256, 256,3), Dense(1), activation= 'sigmoid', optimizer='adam', loss='binary_crossentropy', metrics=['accuracy', 'Precision', 'Recall', 'AUC'], Dropout(0.6)

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

J. VISUAL GEOMETRY GROUP

The Visual Geometry Group(VGG)16 method [34] for diagnosing Down Syndrome in children using facial images operates by leveraging a deep convolutional neural network architecture. This model can extract intricate features from facial images through multiple convolutional layers, capturing hierarchical patterns crucial for accurate diagnosis. The method utilizes the VGG16 architecture's ability to learn complex representations [35], allowing it to discern subtle facial features indicative of Down Syndrome, ultimately enhancing the precision and reliability of the diagnostic process. The VGG16 method for the diagnosis of Down Syndrome in Children Using facial images can be represented mathematically as:

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

where:

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

K. HYPERPARAMETER SETTINGS

The best-fit hyperparameters [36] for the applied machine learning and deep learning models are described in Table 2. We fine-tuned each applied approach using k-fold cross-validation mechanisms and dynamic testing and training. The results of our research show that through the fine-tuning of hyperparameters, we achieved high-performance scores for the diagnosis of Down Syndrome in children using facial images.

TABLE 3. The experimental setup parameter with the specification.

Specification	Value
Environment model	Xeon(R) Intel(R) CPU @ 2.20GHz
Programming language	Python 3.0
CPU MHz	2200.21
Cache size	5632 KB
RAM	13 GB
CPU cores	1
Address sizes	48 bits virtual, 46 bits physical
Bogomips	4399.9

IV. RESULTS AND DISCUSSIONS

Our study's results and discussions present the outcomes of applying a novel approach to leveraging transfer learning for ensemble feature extraction in the context of Down Syndrome diagnosis. This section critically analyzes the performance metrics to evaluate the proposed method's effectiveness compared to existing approaches.

A. EXPERIMENTAL SETTING

The experimental configuration for our research is established through the utilization of a cloud-based implementation of Jupyter Notebook, namely Google Colab. Performance evaluation of the employed machine learning methods is conducted using metric scores such as accuracy, F1, precision, and recall. The details of the environment employed can be found in Table 3.

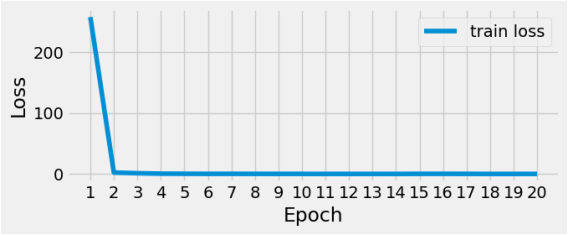
B. CLASSICAL NEURAL NETWORK RESULTS

We have initiated our research by comparing results obtained through classical neural approaches. The facial images of children are initially input into classical methods such as CNN and VGG16 for performance evaluations. The findings of these classical approaches are presented in this section.

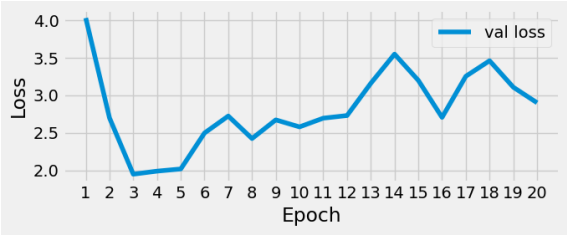
1) RESULTS OF CNN

The performance results of the applied CNN approach during training on images are illustrated in Figure 4. The CNN model is built on image data for 20 epochs. The time series analysis reveals that the training loss scores are high during the initial two epochs of training. Similarly, the validation loss is also elevated during the first two epochs. However, both the training and validation accuracy scores of the CNN gradually increased. Initially, accuracy scores are low for both training and validation sets, but they improve as the epochs progress. The results indicate that the validation accuracy of the CNN during training consistently remains in the range of 60% to 70%.

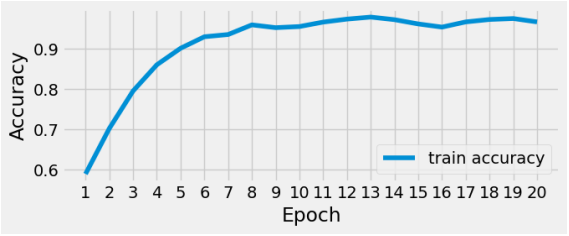
The performance analysis of the applied CNN model for unseen testing is described in Table 4. The analysis revealed that the loss scores are very high using the CNN approach, reaching 3.5187. Due to the elevated loss error, the performance accuracy is also low, measuring 0.66 for the diagnosis of Down syndrome. The analysis indicates that the classical CNN failed to achieve acceptable performance scores for unseen testing data.



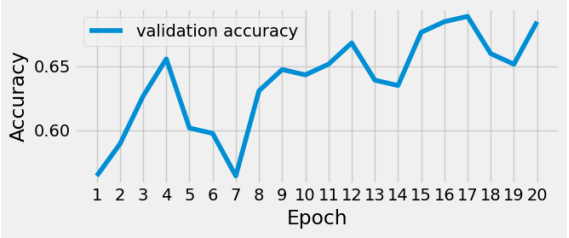
(a) Loss



(b) Loss



(c) Accuracy



(d) Accuracy

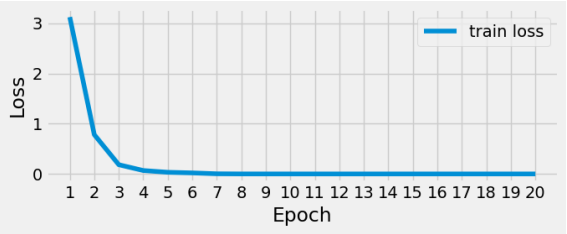
FIGURE 4. The time series analysis of applied CNN neural network.

TABLE 4. The performance analysis of applied CNN model using original images data.

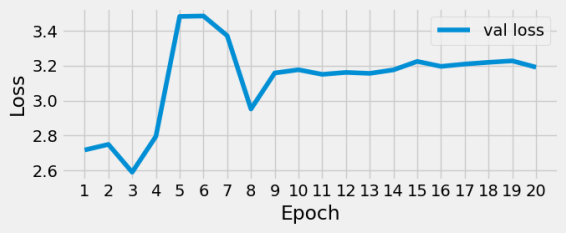
Accuracy	Loss	Target	Precision	Recall	F1
0.66	3.5187	DownSyndrome	0.68	0.69	0.68
		Healty	0.63	0.62	0.63
		Average	0.66	0.66	0.66

2) RESULTS OF VGG16

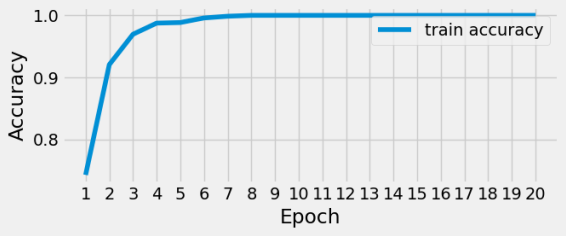
The performance results of the applied VGG16 approach during training on images are illustrated in Figure 5. The VGG16 model is built on image data for 20 epochs. The time series analysis reveals that the training loss scores are high during the initial four epochs of training. Similarly, the validation loss is also elevated during the first two epochs. However, both the training accuracy and validation accuracy scores of the VGG16 gradually increased. Initially, accuracy scores are low for both training and validation sets, but they improve as the epochs progress. The results indicate that the validation accuracy of the VGG16 during training



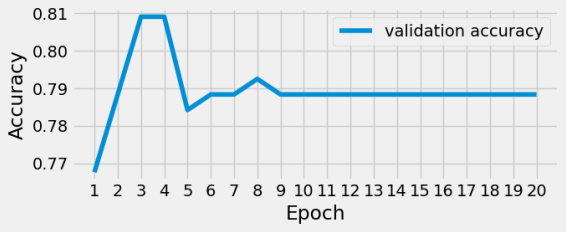
(a) Loss



(b) Loss



(c) Accuracy



(d) Accuracy

FIGURE 5. The time series analysis of applied VGG16 neural network.

consistently remains in the range of 80% to 90%. The training accuracy is also above 90% during analysis.

The performance analysis of the applied VGG16 model for unseen testing is described in Table 5. The analysis infers that the VGG16 approach achieved acceptable accuracy scores of 84% for the diagnosis of Down syndrome in children. The loss scores are also lower compared to the CNN approach. The classical VGG16 approach performed better than CNN. This analysis concludes that the VGG16 approach can be used for further research analysis as it achieved good performance; however, it is not up to the mark.

C. PERFORMANCE ANALYSIS USING SPATIAL FEATURES

During the results analysis of VGG16, we infer that it can help increase accuracy scores. The spatial data features from facial images are then extracted using VGG16, and performance is demonstrated in Table 6. The applied advanced machine learning approaches are trained on the spatial features,

TABLE 5. The performance analysis of applied VGG16 model using original images data.

Accuracy	Loss	Target	Precision	Recall	F1
0.84	2.0685	DownSyndrome	0.84	0.87	0.85
		Healty	0.84	0.81	0.82
		Average	0.84	0.84	0.84

TABLE 6. The performance analysis of applied approaches using spatial features.

Method	Accuracy	Target	Precision	Recall	F1
LGBM	0.81	DownSyndrome	0.82	0.82	0.82
		Healthy	0.79	0.80	0.80
		Average	0.81	0.81	0.81
KNC	0.53	DownSyndrome	0.55	0.69	0.61
		Healthy	0.49	0.35	0.41
		Average	0.52	0.52	0.51
LR	0.59	DownSyndrome	0.61	0.62	0.62
		Healthy	0.56	0.55	0.55
		Average	0.58	0.58	0.58
RF	0.69	DownSyndrome	0.77	0.61	0.68
		Healthy	0.64	0.79	0.70
		Average	0.70	0.70	0.69

TABLE 7. The performance analysis of applied methods using the novel proposed approach.

Method	Accuracy	Target	Precision	Recall	F1
LGBM	0.98	DownSyndrome	0.99	0.98	0.98
		Healthy	0.98	0.99	0.98
		Average	0.98	0.98	0.98
KNC	0.98	DownSyndrome	0.98	0.99	0.98
		Healthy	0.99	0.97	0.98
		Average	0.98	0.98	0.98
LR	**0.99**	**DownSyndrome**	**0.99**	**0.98**	**0.99**
		Healty	**0.98**	**0.99**	**0.99**
		Average	**0.99**	**0.99**	**0.99**
RF	0.98	DownSyndrome	0.98	0.98	0.98
		Healthy	0.98	0.98	0.98
		Average	0.98	0.98	0.98

and the results are reported here. The results present that the LGBM approach achieved good accuracy scores in comparison. Only the KNC method achieved lower scores during the comparisons. This analysis reveals that results are improved; however, the performance is not up to the mark for the critical diagnosis of Down syndrome. There is still a need for advanced transfer learning mechanisms to enhance performance scores.

D. PERFORMANCE ANALYSIS USING PROPOSED APPROACH

The performance comparisons of applied machine learning models using the proposed VNL-Net-based features are described in Table 7. Surprisingly, the results of the applied methods improved using the proposed innovative strategy in this research. All applied methods achieved accuracy scores above 98%. The applied LR method outperformed compared to others with high accuracy scores of 99%. This concludes that, with our proposed approach, applied methods achieved high-performance scores for the diagnosis of Down syndrome using facial images of children.

The radar chart-based performance comparison of classical spatial and newly proposed features is illustrated in Figure 6. This radar analysis shows that our proposed method helps

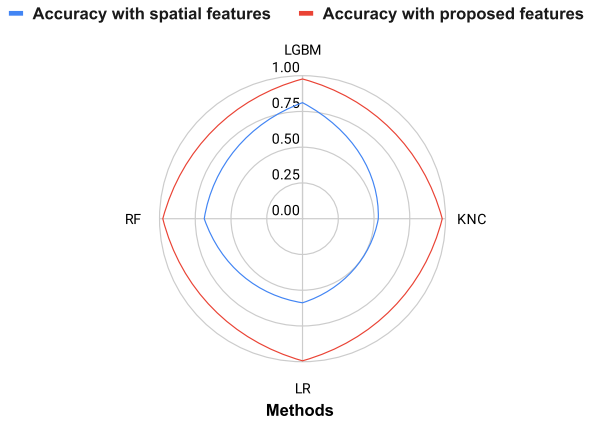


FIGURE 6. The radar chart-based performance compression of classical spatial and novel proposed features.

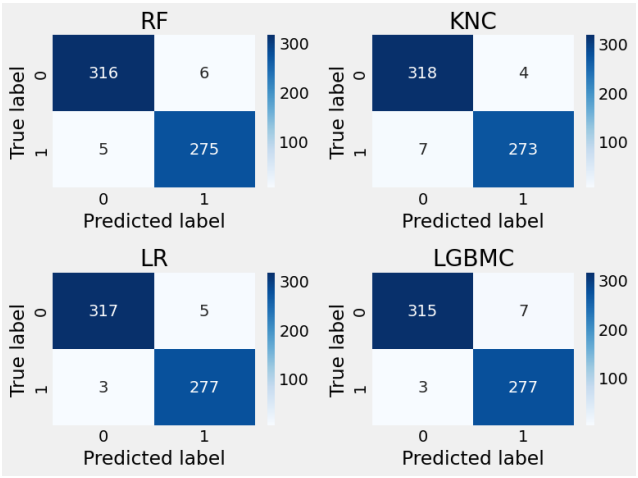


FIGURE 7. The confusion matrix analysis of applied methods using the novel proposed approach.

achieve high-performance outcome for all applied methods. The graph depicts a broader coverage of performance accuracy using the proposed approach. In this research study, our novel proposed approach achieved high accuracy scores for all applied methods.

The performance evaluation, conducted through confusion matrix analysis of the applied methods utilizing the newly proposed features, is presented in Figure 7. The analysis reveals that all applied techniques achieved lower error rates, minimizing misclassifications during the unseen testing phase. The results validate the superior performance of the proposed LR approach, demonstrating a high correct classification rate and a low error rate compared to other methods.

E. K-FOLD CROSS VALIDATIONS ANALYSIS

We then validate the performance scores of the applied machine learning method using K-fold cross-validation mechanisms, and validation results are reported in Table 8. We have used 10 folds of data for result validations. The analysis results show that all applied methods achieved high K-fold accuracy, which is above 98%, with minimal standard

TABLE 8. The performance valuations based on kfold cross variations mechanism.

Method	Kfold	Accuracy	Standard Deviations (+/-)
LGBM	10	0.9818	0.0058
KNC	10	0.9737	0.0107
LR	10	0.9807	0.0072
RF	10	0.9821	0.0054

TABLE 9. The run-time computations complexity analysis of applied approaches.

Method	Runtime Computations (seconds)
LGBM	0.04302
KNC	0.0049
LR	0.1213
RF	0.8936

deviation scores. This analysis confirms the generalization of the proposed LR model for the diagnosis of Down syndrome in children.

F. COMPUTATIONS COMPLEXITY ANALYSIS

The runtime computational complexity results of the applied approaches is described in Table 9. The analysis revealed that the applied machine learning approaches achieved lower scores in time computational complexity. The RF approach obtained significantly high time computational complexity scores of 0.8936. The proposed LR method detects Down syndrome in children using a time computational complexity of 0.1213 seconds.

G. FEATURE SPACE ANALYSIS OF PROPOSED FEATURES

The provided features space analysis is employed to visualize and analyze the spatial distribution of facial features. The 3D graph plot presumably contains data representing facial features, and the features space function is utilized to generate a three-dimensional scatter plot. The two different-colored dots specify the marker style for each data point and differentiate between labels associated with Down syndrome and healthy individuals. The analysis indicates the number of clusters to identify, aligning with the binary classification of Down syndrome and healthy labels. This research approach integrates new spatial features analysis to facilitate the discrimination between healthy and Down syndrome-affected children based on their facial features.

The analysis of feature space for the recently generated transfer feature set is presented in Figure 3. This examination reveals that features generated through the VNL-Net method exhibit a high degree of linear separability. The pronounced linearity in the separability of these features contributes significantly to improving the performance scores for diagnosing Down syndrome in children.

H. STATE OF THE ART STUDIES COMPARISON

For a fair comparison, we have evaluated the performance scores of our novel proposal against state-of-the-art methods. The analysis reveals that classical machine learning and deep learning methods were predominantly used in previous

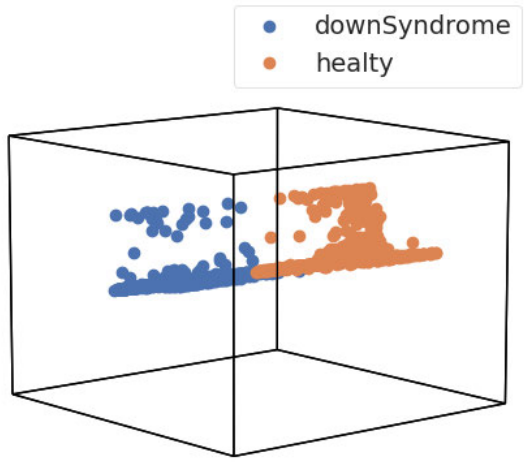


FIGURE 8. The features space analysis of extracted new features from facial images.

TABLE 10. Performance comparison of the proposed approach with State of the art methods.

Ref	Learning Type	Proposed Technique	Accuracy	Precision
[10]	Machine learning	Ahi Method	73%	60%
[18]	Deep learning	CNN Method	85%	90%
[17]	Machine learning	SVM Method	97%	93%
Our	**Transfer learning**	**Novel VNL-Net LR**	**99%**	**99%**

research. In this study, we employed advanced transfer learning mechanisms with a novel approach. The highest accuracy achieved previously was 97%. This analysis concludes that our proposed approach outperformed the state-of-the-art studies, achieving high-performance scores of 99%.

V. CONCLUSION AND FUTURE WORK

This study proposes an innovative methodology for the early diagnosis of Down syndrome using an advanced deep neural network approach. We utilized 3,009 facial images of children with Down syndrome and healthy children taken from the age group range of 0 to 15 for conducting our research experiments. We introduce a novel transfer learning-based feature generation named VNL-Net for feature extraction, which initially extracts spatial features from input image data. Subsequently, the ensemble feature set of NMF and LGBM is derived from these spatial features. We constructed several advanced machine learning and deep learning models on the newly created feature set to evaluate performance. Two advanced deep learning models and four machine learning models were developed using facial image data and the newly created features. The performance of the applied methods is validated through K-fold cross-validation. Additionally, we assessed the computational complexity of each method. Extensive research results demonstrate that the logistic regression method outperformed state-of-the-art studies with a high accuracy of 0.99. Each applied method was fine-tuned, and performance was validated using the K-fold cross-validation mechanism. The runtime computational complexity of the applied methods is also determined.

A. FUTURE WORK

In the future, we intend to develop an interactive Graphical User Interface (GUI) wherein parents can assess whether their children have Down syndrome. This assessment can be conducted simply by utilizing a facial picture or a live video of the child. Additionally, we aim to enhance the performance of the interface further by incorporating advanced neural network approaches.

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ALI RAZA received the Bachelor of Science degree in computer science from the Department of Computer Science, Khwaja Fareed University of Engineering and Information Technology (KFUEIT), Rahim Yar Khan, Pakistan, in 2021, where he is currently pursuing the M.S. degree in computer science. His current research interests include data science, artificial intelligence, data mining, natural language processing, machine learning, deep learning, and image processing.

KASHIF MUNIR received the B.Sc. degree in mathematics and physics from The Islamia University of Bahawalpur, Pakistan, in 1999, the first M.Sc. degree in information technology from Universiti Sains Malaysia, in 2001, the second M.S. degree in software engineering from the University of Malaya, Malaysia, in 2005, and the Ph.D. degree in informatics from the Malaysia University of Science and Technology, Malaysia, in 2015. He has been in the field of higher education, since 2002. After an initial teaching experience in courses with the Binary College, Malaysia, for one semester, and the Stamford College, Malaysia, for around four years. Later, he relocated to Saudi Arabia. He was with the King Fahd University of Petroleum and Minerals, Saudi Arabia, from September 2006 to December 2014. He moved to the University of Hafr Al Batin, Saudi Arabia, in January 2015. In July 2021, he joined the Khwaja Fareed University of Engineering and Information Technology, Rahim Yar Khan, as an Assistant Professor with the IT Department. He has published journal articles, conference papers, book, and book chapters. His research interests include cloud computing security, software engineering, and project management. He has been in the technical program committee of many peer-reviewed conferences and journals, where he has reviewed many research articles.

MUBARAK S. ALMUTAIRI received the B.Sc. degree in systems engineering from the King Fahd University of Petroleum and Minerals, Dhahran, Saudi Arabia, in 1997, the M.Sc. degree in industrial and systems engineering from the University of Florida, Gainesville, FL, USA, in 2003, and the Ph.D. degree in systems design engineering from the University of Waterloo, Waterloo, Canada, in 2007. From 1997 to 2000, he was an Industrial Engineer with Saudi Arabia Oil Company (Aramco). He is currently the Dean of the Applied College, University of Hafr Al Batin (UHB), Hafr Al Batin, Saudi Arabia, where he is also an Associate Professor with the Computer Science and Engineering Department. His research interests include decision analysis, expert systems, risk assessment, information security, fuzzy logic, and mobile government application.

RUKHSHANDA SEHAR received the B.Sc. degree in computer science and the M.C.S. degree from The Islamia University of Bahawalpur, in 2017 and 2021, respectively, and the M.S. degree in computer science from the Khawaja Fareed University of Engineering and Technology (KFUEIT). Her area of interests include machine learning and deep learning.