Comparative Study of Prenatal and Postnatal Images for Detecting Down Syndrome in Children

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***Abstract*—Down syndrome is a genetic disorder caused by the presence of an extra copy of chromosome 21, affecting both neurological development and physical features. Early and accurate diagnosis is critical for ensuring timely medical intervention and support. This study presents a comparative analysis of prenatal (ultrasound) and postnatal (facial) imaging modalities for the detection of Down syndrome using deep learning techniques. We employed VGG19, ResNet50, DenseNet121, MobileNetV2, and the Vision Transformer for image classification. An ensemble model integrating four CNN architectures achieved superior performance, with 92% test accuracy on prenatal images and 83% on postnatal images. Among the individual models, ResNet50 outperformed the others across both modalities. Evaluation metrics, including accuracy, precision, recall, and F1-score, confirm the effectiveness of the proposed framework. These results highlight the potential of ensemble learning to enhance the early detection of Down syndrome and improve accessibility to healthcare.**

***Keywords*—Down syndrome; prenatal ultrasound; postnatal facial recognition; CNN; vision transformer; ensemble learning**

I. Introduction

Down syndrome is currently defined as a genetic disorder caused by the presence of an extra chromosome 21, affecting both the physical characteristics and cognitive development of children. However, this disorder was not recognized until 1866 when the English physician John Langdon Down published a detailed description [1] of the condition that would later bear his name. Children affected by Down syndrome often experience academic difficulties, delays in cognitive development, and health complications such as heart disease, hearing loss, and vision impairments. Although there is no cure, early diagnosis, appropriate care, and therapy can significantly improve quality of life. Research into imaging-based detection of Down syndrome is crucial for enabling early diagnosis and enhancing healthcare accessibility. Advances in deep learning and imaging technologies have facilitated the development of cost-effective, noninvasive diagnostic methods. While postnatal imaging [2] captures facial characteristics, prenatal imaging [3]—such as ultrasound and 2D scans—detects structural markers. Combining these two approaches provides a comprehensive diagnostic framework. Motivated by the success of deep learning techniques, we employed transfer learning using models such as ResNet50, VGG19, DenseNet121, MobileNetV2, and the Vision Transformer.

Each of the employed models offers unique advantages; therefore, leveraging an ensemble approach proves beneficial, as it consistently outperforms individual CNN architectures. The key contributions of this study are summarized as follows:

- Evaluation of both ultrasound (prenatal) and facial (postnatal) images to fine-tune models for improved diagnostic accuracy in AI-assisted Down syndrome detection.
- Comparative analysis of two distinct imaging modalities using diverse datasets—1,684 ultrasound images and 2,000 facial images—to enhance generalizability.
- Performance benchmarking of multiple deep learning models across both prenatal and postnatal image datasets.
- Implementation of an ensemble learning strategy that integrates CNN features, resulting in optimized performance and higher accuracy compared to standalone models across both imaging types.

II. Related Works

Several studies have previously explored the prediction and diagnosis of Down syndrome (DS) using a variety of methods, some of which are summarized below:

A study [4] utilized the Random Forest machine learning model to improve the accuracy of DS prediction using second-trimester prenatal images, achieving a detection rate of 66.7% on the training dataset—outperforming traditional approaches. In another study [5], NMR-based metabolomics of maternal serum samples was employed to predict DS. Three key biomarkers were identified, and the inclusion of maternal age as a modifier improved detection efficiency from 48.1% to 51.9%.

Vičić et al. [6] analyzed 157 DS cases out of 6,448 prenatal examinations conducted in Croatia between 2002 and 2014. Key diagnostic markers included maternal age, ultrasound findings, and biochemical screening, while the primary diagnostic procedures were chorionic villus sampling and amniocentesis.

The study [7] introduced CVIFLR, a machine learning model designed for non-invasive DS prediction, and [8] explored deep transfer learning (DTL) in disease diagnosis, achieving a classification accuracy of 93.3%. Thomas et al. [9] used 100 fetal ultrasound images, combining SegNet for nuchal translucency segmentation with AlexNet for classification, resulting in 100% sensitivity, 85.7% specificity, and 91.7% accuracy.

Machine learning models for first-trimester DS screening using clinical data from the UK and Canada were evaluated

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in [10], where deep neural networks (DNNs) outperformed commercial software with an AUC of 0.96 and a 78% detection rate. Similarly, Zhang et al. [11] employed machine learning algorithms for second-trimester prenatal screening, where support vector machines (SVMs) achieved 100% detection with a low false positive rate (FPR).

Another study [12] proposed a 1D convolutional neural network (CNN) model that utilized biomarkers like PAPP-A and hCG along with ultrasound data, reaching a high detection accuracy of 95.17%. Given the ethnic variability of DS features [13], facial recognition tools have shown promise, with one approach achieving 94.3% accuracy. Cornejo et al. [14] developed a novel method using geometric descriptors from facial features, capturing key distances such as intercanthal spacing, nose breadth, and mouth width based on 14 distances from 16 fiducial points.

The Face2Gene AI tool demonstrated strong performance in identifying DS among Thai children [15], with 90% sensitivity for DS cases and 87% for non-DS cases. A convolutional neural network-based method [16] offered a non-invasive solution for DS identification, achieving 99.3% accuracy and outperforming conventional methods like SVM.

In another work [17], extracted image features were used to train a backpropagation neural network (BPNN), which classified nasal bone presence with 86% accuracy. Additionally, a study [18] proposed an expert system framework aimed at enhancing the education of children with DS, while another work [19] introduced a mobile educational platform to support cognitive development through educator-guided learning.

Although not directly related to DS, a study [20] focused on Parkinson's disease—a related neurological disorder—demonstrated the effectiveness of three machine learning approaches for diagnosis. Furthermore, a review [21] provided comprehensive guidelines for the care and development of children with DS. The study [22] emphasized the benefits of early detection and treatment of sleep disorders, such as obstructive sleep apnea (OSA), in DS patients. Finally, [23] demonstrated the use of computer vision and deep learning techniques to analyze parental behaviors, specifically identifying supportive physical interventions during communication with children affected by Down syndrome.

III. Methodology

A systematic method for comparing both prenatal and postnatal images of children with or without Down syndrome is depicted in Fig. 1. This section outlines the methodological pipeline in this study, encompassing data collection, data processing, training various models, model evaluation, and finally the comparison between prenatal and postnatal datasets for detecting Down syndrome in children.

A. Data Collection

Data collection is the initial and most critical step in the proposed methodology. In this study, two distinct datasets are utilized:

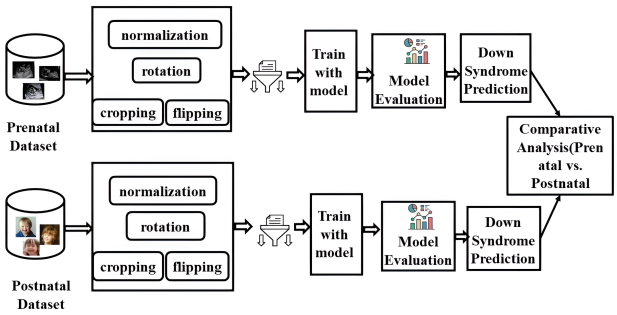

Fig. 1. Proposed methodology for Down Syndrome (DS) detection.

1) Prenatal dataset: This dataset, obtained from Mendeley [https://data.mendeley.com/datasets/n2rbrb9t4f/1], comprises 1,684 ultrasound images of fetuses aged between 11 and 14 weeks. Ultrasound imaging is a standard tool for monitoring fetal development and, in this context, is used to detect potential markers of Down syndrome prior to birth. These markers may include increased nuchal translucency thickness, anomalies in the facial profile, or other structural differences observed in the fetus.

2) Postnatal dataset: Sourced from Kaggle and GitHub [https://github.com/vinayaa1/down-syndrome-detection], this dataset contains 2,000 facial images of infants and children. The majority of samples fall within the 0–15 years age range, with limited representation from 16–18 years. This dataset supports the detection of Down syndrome through facial recognition, leveraging characteristic facial features such as almond-shaped eyes, a flat nasal bridge, and other distinct craniofacial traits. These postnatal images are instrumental in identifying Down syndrome after birth.

Each dataset was split into three subsets: training (80%), validation (10%), and testing (10%). This division ensures unbiased model evaluation and promotes better generalization of the results. It is also important to note that the prenatal and postnatal datasets were sourced independently and do not represent the same children across both stages. The neural network or machine learning model is trained using the training set. Its performance and ability to generalize to new data are then evaluated using the test set. This approach ensures a large and diverse training dataset, a separate set for unbiased evaluation, and the ability to assess the model on unseen samples, simulating potential real-world scenarios.

B. Data Preprocessing

Data processing is a crucial step in data-driven tasks like machine learning, statistical analysis, and decision-making. It transforms raw data into structured, clean, and usable formats, improving data quality and standardizing datasets for machine learning models. Preprocessing steps in Fig. 2 and Fig. 3 include image resizing, normalization, rotation, cropping, flipping, and dataset splitting.

Image resizing ensures uniform input dimensions, while normalization standardizes pixel intensity values across all

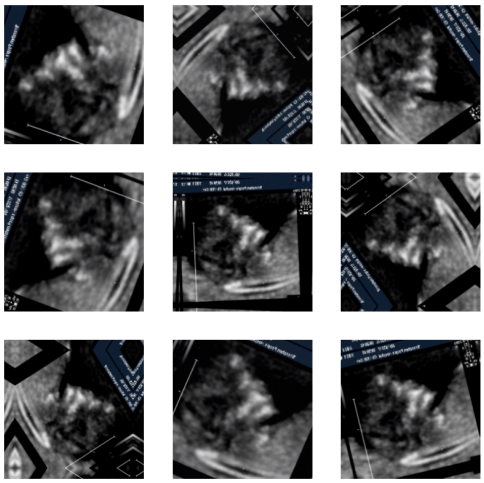

Fig. 2. Data augmentation of prenatal images: Normalized, Rotated, Cropped and Flipped.

Fig. 3. Data augmentation of postnatal images: Normalized, Rotated, Cropped and Flipped.

images. Rotation helps correct misalignments, and cropping reduces background noise, enhancing model efficiency. Flipping augments the dataset by increasing variability, which helps prevent overfitting. Dataset splitting into training, validation, and testing sets enables effective performance evaluation. Collectively, these preprocessing techniques ensure that models learn more effectively from clean and well-prepared input data.

C. Transfer Learning Using Pre-trained CNN Models

This study employs four deep convolutional neural network (DCNN) models—VGG19, ResNet50, DenseNet121, and MobileNetV2—that were pre-trained on the ImageNet dataset and used for feature extraction. Additionally, the Vision Transformer model is utilized to evaluate performance from a transformer-based perspective. These models are applied to both prenatal ultrasound images and postnatal facial images for the detection of Down syndrome in children.

1) VGG19: This 19-layer deep CNN is known for its high accuracy but also for its computational intensity. It uses small 3×3 convolutional filters and ReLU activation. VGG19 is an extended version of VGG16, incorporating three additional layers [24].

2) ResNet50: A 50-layer residual network [25] designed to address the vanishing gradient problem through the use of skip (residual) connections, enabling more effective training of deeper networks.

3) DenseNet121: A densely connected convolutional network with 121 layers, introduced by [26]. In this architecture, each layer receives inputs from all preceding layers, which enhances feature propagation and reduces the number of parameters.

4) MobileNetV2: Designed for mobile and resource-constrained environments, MobileNetV2 uses depthwise separable convolutions to reduce computational cost while maintaining accuracy. It was introduced by [27].

5) Vision Transformer (ViT): Proposed by [28], the Vision Transformer processes images as sequences of patches and utilizes self-attention mechanisms to capture long-range feature dependencies. Unlike CNNs, it leverages transformer-based architectures for image classification.

D. Training and Predicting Models

To obtain the visual Down Syndrome detection results from each model, we extract deep image feature vectors using DenseNet121, VGG19, MobileNetV2, ResNet50, and Vision Transformer. These feature vectors are then passed through the appropriate fully connected dense layers followed by activation layers. The model parameters are optimized using both the Adam optimizer and the stochastic gradient descent (SGD) technique. Training is conducted using the sparse categorical cross-entropy (CE) loss function, shown in (1).

$$CE = -\frac{1}{N}\sum_{i=1}^{N}[y_i \log p(y_i) + (1-y_i)\log(1-p(y_i))] \quad (1)$$

E. Proposed Ensemble Model

In order to enhance accuracy, the ensemble combines the outputs of four CNNs (VGG19, ResNet50, DenseNet121, and MobileNetV2). In Fig. 4, the architecture of the model [29] is described. As illustrated in Fig. 4, each CNN model extracts deep features independently. These features are then concatenated and passed to a classifier for prediction. Soft voting combines the probability outputs, and the final prediction is based on the average likelihood. This method enhances generalization and stability across diverse image modalities. These basic steps include the following:

1) Feature extraction: This involves the removal of the last layers of each model and the extraction of deep feature vectors.

2) Concatenation: Combining these characteristics improves categorization accuracy.

3) Prediction aggregation: Accuracy and resilience are increased by using Yes/No Soft Voting, Hard Voting, Stacking, and Bagging.

4) Optimization: To make the entire training process as efficient as possible, use the Adam Optimizer and the categorical cross-entropy loss function.

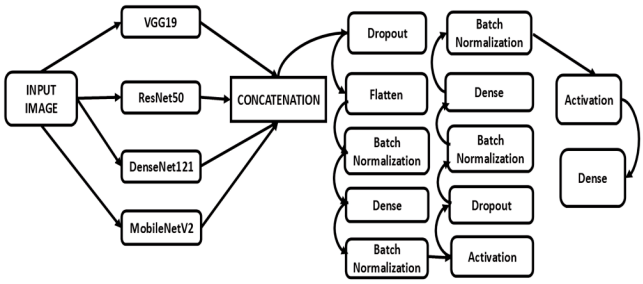

Fig. 4. Architecture of ensemble model.

Model ensembling offers several benefits for improving machine learning performance:

5) Improved generalization: Different models capture diverse features due to their architectural differences. For example, VGG19 is effective for low-level pattern recognition, ResNet50 extracts deep features, DenseNet121 promotes feature reuse and propagation, and MobileNetV2 focuses on efficient feature extraction.

6) Enhanced accuracy: Combining predictions using techniques such as soft voting (averaging probabilities), hard voting (majority voting), or stacking (meta-learning) helps improve overall accuracy.

7) Robustness to data variability: Each model emphasizes different aspects of the data, making the ensemble more robust and better able to generalize across varied datasets.

8) Reduction of overfitting: Using multiple diverse models helps reduce the risk of overfitting by preventing the ensemble from memorizing the training data.

9) Efficient feature learning: The combination of different architectures allows the model to learn features at multiple levels of abstraction, leading to more effective overall feature representation.

In prenatal and postnatal datasets, the ensemble technique ensures robust diagnosis of Down syndrome by enhancing generalization and minimizing overfitting.

F. Performance Measures

The performances are classified into True Positive (TP), True Negative (TN), False Positive (FP), False Negative (FN). Various evaluation metrics are used in this study such as, accuracy, precision, recall, and f1-score.

- Accuracy – Measures the proportion of correct predictions:

$$Accuracy = \frac{TP + TN}{TP + TN + FP + FN} * 100 \quad (2)$$

- Precision – Measures the percentage of correctly predicted positive cases:

$$Precision = \frac{TP}{TP + FP} * 100 \quad (3)$$

- Recall (Sensitivity) – Measures the ability of the model to find all positive cases:

$$Recall = \frac{TP}{TP + FN} * 100 \quad (4)$$

- F1-Score – Harmonic mean of precision and recall, balancing both metrics:

$$F1 = 2 \times \frac{Precision \times Recall}{Precision + Recall} * 100 \quad (5)$$

IV. Experiments and Results

In this study, we utilized both prenatal ultrasound images and postnatal facial images of children to detect Down Syndrome. We fine-tuned five pre-trained models—ResNet50, VGG19, DenseNet121, MobileNetV2, and Vision Transformer—using transfer learning. As represented in Table I and Table II, ResNet50 consistently achieved the highest test accuracy across both imaging modalities. Among the CNN models, the highest test accuracy for prenatal images was 91%, while for postnatal images it was 83%. Although the Vision Transformer demonstrated strong performance, it did not surpass ResNet50. To further improve accuracy, we employed model ensembling, which achieved 92% accuracy for prenatal images and 83% for postnatal images.

TABLE I. Accuracy of Different Models (Prenatal Images)

Model	Validation Accuracy	Test Accuracy
ResNet50	90%	91%
VGG19	86%	85%
DenseNet121	84%	88%
MobileNetV2	83%	86%
Vision Transformer	80%	83%
Ensemble	**91%**	**92%**

TABLE II. Accuracy of Different Models (Postnatal Images)

Model	Validation Accuracy	Test Accuracy
ResNet50	80%	**83%**
VGG19	75%	77%
DenseNet121	70%	72%
MobileNetV2	79%	78%
Vision Transformer	72%	73%
Ensemble	**82%**	**83%**

A. Comparison Between the Models for Prenatal and Postnatal Dataset

For the prenatal dataset, we have the classification report for each model, like ResNet50, DenseNet121, MobileNetV2, VGG19, and Vision Transformer model. From all of the classification matrices, we can see that ResNet50 is beating all other models in all contexts, like accuracy, precision and f1-score in Fig. 5.

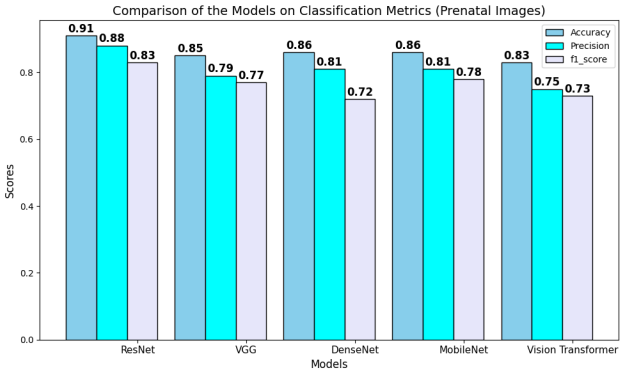

Fig. 5. Comparison chart of the models for prenatal images.

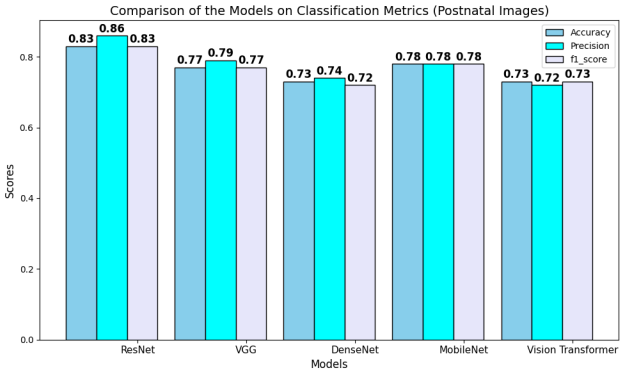

Fig. 6. Comparison chart of the models for postnatal images.

In addition, for the postnatal dataset, we generated classification reports for each model, including ResNet50, DenseNet121, MobileNetV2, VGG19, and the Vision Transformer. Once again, ResNet50 outperforms all other models across all evaluation metrics. Fig. 6 presents a comparative chart of these models based on the prenatal dataset.

B. Performance of the Proposed Ensemble Model for Prenatal Images

1) Accuracy and loss graph: The overall performance based on the training and validation curves is shown in next Fig. 7.

Fig. 7. Performance of the ensemble model (Prenatal images).

It is evident from the graphs that both the training and validation accuracies are close enough, suggesting that the model shows minimal overfitting. Similarly, the loss curve demonstrates a consistent decline for both training and validation sets, with the validation loss remaining low and closely matching the training loss trend. This suggests that the model effectively minimizes error and learns meaningful features from the data with minor overfitting.

2) Confusion matrix: As we know that a confusion matrix helps to evaluate a classification model by presenting true and false predictions for each class. Moreover, this helps to calculate precision, recall and f1-score precisely.

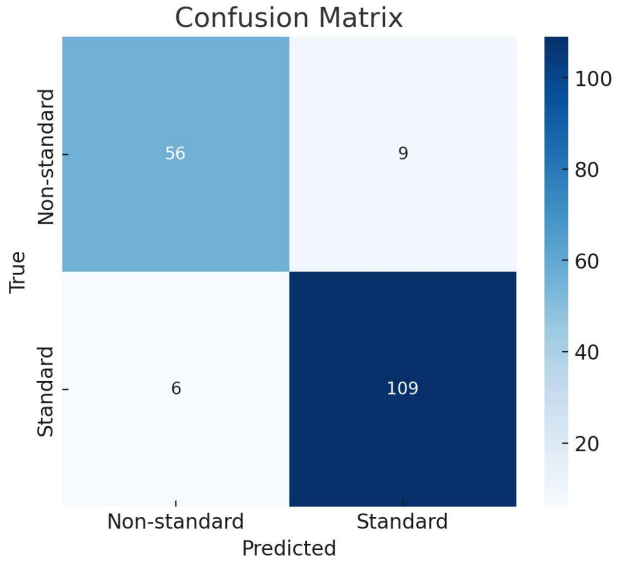

Fig. 8. Confusion matrix of ensemble model (Prenatal images).

Finally, the confusion matrix in Fig. 8 evaluates the performance of the ensemble model for Down Syndrome detection using nuchal translucency (NT) measurements. The model achieves an accuracy of 91.67%, correctly identifying 56 Down Syndrome cases where NT measurements were abnormal. However, it misclassifies nine affected fetuses as normal (false negatives). Additionally, the model correctly identifies 109 normal fetuses with standard NT measurements, while misclassifying six as having Down Syndrome (false positives).

C. Performance of the Proposed Ensemble Model for Postnatal Images

1) Accuracy and loss graph: The line graph in Fig. 9 represents two key plots to evaluate the ensemble model's performance through accuracy and loss curves. The model is shown to perform well with high accuracy in both training and validation with 25 epochs. This also suggests a strong ability to classify with minimal overfitting.

2) Confusion matrix: The effectiveness of an ensemble model for identifying Down syndrome from postnatal images is assessed using this confusion matrix depicted in Fig. 10.

Since there were no false negatives, the model achieved a perfect sensitivity of 100% by properly identifying all 105 newborns with Down syndrome. Nevertheless, it produced a

Fig. 9. Performance of the ensemble model (Postnatal images).

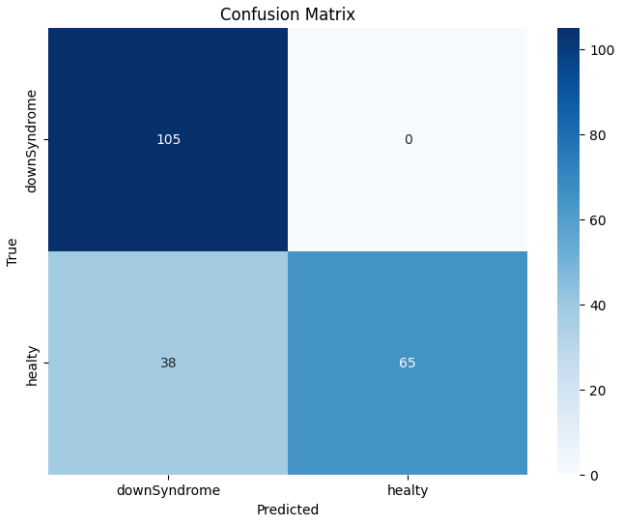

Fig. 10. Confusion matrix of ensemble model (Postnatal images).

false positive rate by incorrectly classifying 28 healthy infants as having Down syndrome. 75 healthy newborns were correctly categorized despite this. Although the model is excellent at identifying cases of Down syndrome without missing any, it may need to be further improved to increase specificity and decrease false alarms because it has a propensity to categorize healthy babies incorrectly.

D. Comparison Between the Ensemble Model for Prenatal and Postnatal Dataset

After doing the ensemble by Feature Fusion Ensemble method with the four models: ResNet50, VGG19, DenseNet121, and MobileNetV2, we have found that the model has increased the performance instantly. The model's accuracy score is almost near to ResNet50 but increased a little bit. Here is a bar graph in Fig. 11, comparing the classification performance between the prenatal and postnatal ensemble models. The prenatal model performs better in all four metrics, indicating its stronger classification capability.

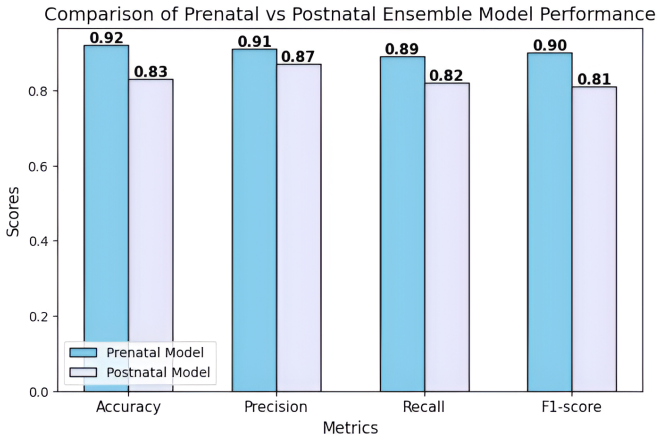

Fig. 11. Comparison chart of the ensemble model (Prenatal vs. Postnatal).

E. Performance Comparison with Existing Research for Down Syndrome Detection

The following Table III represents a comparative analysis of different research studies focused on the detection of Down syndrome using various machine learning models. It includes details on image types, sample sizes, applied models, and their respective accuracies. For example, Reshi, Aijaz Ahmad, et al. [3] used CNN on 1,120 fetus images, achieving an accuracy of 97%. Similarly, Zhao, Qian, et al.[2] applied SVM to facial images (50 DS, 50 healthy) and attained 94% accuracy. The highest reported accuracy (99.3%) was achieved by Feng, Bing, et al. [16], who used a Support Vector Machine (SVM) on SNP maps with a relatively small dataset (315 non-DS, 63 DS cases). This exceptionally high accuracy is likely due to the limited size and controlled nature of the dataset. Unlike some prior studies using smaller or synthetic datasets, our work utilizes publicly available and diverse real-world datasets—namely the Kaggle facial image set and Mendeley ultrasound data—which allows a more robust evaluation. Among those, [3] used the same Mendeley ultrasound dataset, reporting 97% accuracy using CNN, while we achieved 92% through ensemble methods. Moreover, the proposed model in this study processes both ultrasound and facial images, leveraging an ensemble of four CNN models to achieve optimal accuracy—92% for prenatal images and 83% for postnatal images. These results demonstrate that integrating multiple image modalities with deep learning techniques can significantly enhance the accuracy of Down Syndrome (DS) detection.

V. Conclusion

In this study, we explored the effectiveness of different imaging modalities and machine learning techniques for detecting Down Syndrome in children. By analyzing both prenatal (ultrasound) and postnatal (facial) images, our aim was to evaluate the diagnostic potential of each modality individually, as well as their combined efficacy. We investigated the performance of several pre-trained convolutional neural network (CNN) models—ResNet50, VGG19, DenseNet121, and MobileNetV2—on both datasets. In addition, we evaluated the Vision Transformer model for the same task.

TABLE III. COMPARISON OF PREVIOUS STUDIES RELATED TO DOWN SYNDROME

Research	Image Description	Image Samples	Applied Model	Accuracy
[3]	Fetus Image	1120	CNN	97%
[7]	MSS	Non-DownSyndrome: 100,244 DownSyndrome: 108	CVIFLR	95% (AU-CROC)
[2]	Facial Image	DS: 50, Healthy: 50	SVM	94%
[8]	Facial Image	350	DCNN	90%
[17]	Fetus Image	Nasal bone: 50, Without Nasal bone: 50	BPNN	80%
[16]	SNP Maps	Non-DS: 315, DS: 63	SVM	99.3%
[30]	Fetus Image	442	K-means Clustering	87%
Proposed Model	Ultrasound & Facial Images	Ultrasound: 1684, Facial: 2000	CNN & Vision Transformer	Ultrasound: 92%, Facial: 83%

The ensemble of CNN models yielded the highest accuracy, achieving 92% for prenatal ultrasound images and 83% for postnatal facial images, underscoring the advantages of a multimodal approach. Notably, the pre-trained ResNet50 model demonstrated consistently strong performance, achieving a validation accuracy of 89% and test accuracy of 91% on the prenatal dataset, and a validation accuracy of 81.5% and test accuracy of 83% on the postnatal dataset.

Compared to existing studies that primarily focus on either fetal ultrasound or facial imagery using traditional machine learning methods, our approach leverages deep learning to enhance classification performance. Future research should focus on expanding dataset size, incorporating more advanced imaging features, and developing hybrid AI architectures to improve accuracy and generalizability. This includes integrating data from diverse demographics, combining prenatal and postnatal imaging, and utilizing multimodal data fusion. Furthermore, future models could benefit from hybrid AI architectures that combine CNNs, Vision Transformers, and traditional machine learning techniques to enhance feature extraction and ensemble learning.

REFERENCES

[1] A. D. Ataman, E. E. Vatanoğlu-Lutz, and G. Yıldırım, "Medicine in stamps: history of down syndrome through philately," *Journal of the Turkish German Gynecological Association*, vol. 13, no. 4, p. 267, 2012.

[2] Q. Zhao, K. Rosenbaum, K. Okada, D. J. Zand, R. Sze, M. Summar, and M. G. Linguraru, "Automated down syndrome detection using facial photographs," in *2013 35th Annual International Conference of the IEEE Engineering in Medicine and Biology Society (EMBC)*. IEEE, 2013, pp. 3670–3673.

[3] A. A. Reshi, S. Shafi, I. Qayoom, M. Wani, S. Parveen, and A. Ahmad, "Deep learning-based architecture for down syndrome assessment during early pregnancy using fetal ultrasound images," *Int. J. Exp. Res. Rev*, vol. 38, pp. 182–193, 2024.

[4] F. He, B. Lin, K. Mou, L. Jin, and J. Liu, "A machine learning model for the prediction of down syndrome in second trimester antenatal screening," *Clinica Chimica Acta*, vol. 521, pp. 206–211, 2021.

[5] R. O. Bahado-Singh, R. Akolekar, R. Mandal, E. Dong, J. Xia, M. Kruger, D. S. Wishart, and K. Nicolaides, "Metabolomic analysis for first-trimester down syndrome prediction," *American journal of obstetrics and gynecology*, vol. 208, no. 5, pp. 371–e1, 2013.

[6] A. Vičić, T. Hafner, I. B. Vlatković, P. Korać, D. Habek, and F. Stipoljev, "Prenatal diagnosis of down syndrome: A 13-year retrospective study," *Taiwanese Journal of Obstetrics and Gynecology*, vol. 56, no. 6, pp. 731–735, 2017.

[7] L. Li, W. Liu, H. Zhang, Y. Jiang, X. Hu, and R. Liu, "Down syndrome prediction using a cascaded machine learning framework designed for imbalanced and feature-correlated data," *IEEE Access*, vol. 7, pp. 97 582–97 593, 2019.

[8] B. Jin, L. Cruz, and N. Gonçalves, "Deep facial diagnosis: deep transfer learning from face recognition to facial diagnosis," *IEEe Access*, vol. 8, pp. 123 649–123 661, 2020.

[9] M. C. Thomas and S. P. Arjunan, "Deep learning measurement model to segment the nuchal translucency region for the early identification of down syndrome," *Measurement Science Review*, vol. 22, no. 4, pp. 187–192, 2022.

[10] A. Koivu, T. Korpimäki, P. Kivelä, T. Pahikkala, and M. Sairanen, "Evaluation of machine learning algorithms for improved risk assessment for down's syndrome," *Computers in Biology and Medicine*, vol. 98, pp. 1–7, 2018.

[11] H.-G. Zhang, Y.-T. Jiang, S.-D. Dai, L. Li, X.-N. Hu, and R.-Z. Liu, "Application of intelligent algorithms in down syndrome screening during second trimester pregnancy," *World Journal of Clinical Cases*, vol. 9, no. 18, p. 4573, 2021.

[12] D. Wright, I. Bradbury, F. Malone, M. D'Alton, A. Summers, T. Huang, S. Ball, A. Baker, B. Nix, D. Aitken *et al.*, "Cross-trimester repeated measures testing for down's syndrome screening: an assessment." *Health Technol Assess*, vol. 14, no. 33, p. 1, 2010.

[13] P. Kruszka, A. R. Porras, A. K. Sobering, F. A. Ikolo, S. La Qua, V. Shotelersuk, B. H. Chung, G. T. Mok, A. Uwineza, L. Mutesa *et al.*, "Down syndrome in diverse populations," *American Journal of Medical Genetics Part A*, vol. 173, no. 1, pp. 42–53, 2017.

[14] J. Y. R. Cornejo, H. Pedrini, A. Machado-Lima, and F. d. L. d. S. Nunes, "Down syndrome detection based on facial features using a geometric descriptor," *Journal of Medical Imaging*, vol. 4, no. 4, pp. 044 008–044 008, 2017.

[15] N. Vorravanpreecha, T. Lertboonnum, R. Rodjanadit, P. Sriplienchan, and K. Rojnueangnit, "Studying down syndrome recognition probabilities in thai children with de-identified computer-aided facial analysis," *American Journal of Medical Genetics Part A*, vol. 176, no. 9, pp. 1935–1940, 2018.

[16] B. Feng, D. C. Samuels, W. Hoskins, Y. Guo, Y. Zhang, J. Tang, and Z. Meng, "Down syndrome prediction/screening model based on deep learning and illumina genotyping array," in *2017 IEEE International Conference on Bioinformatics and Biomedicine (BIBM)*. IEEE, 2017, pp. 347–352.

[17] T. Anjit and S. Rishidas, "Identification of nasal bone for the early detection of down syndrome using back propagation neural network," in *2011 International conference on communications and signal processing*. IEEE, 2011, pp. 136–140.

[18] C. VENEGAS, R. Cevallos, R. Cordova, D. D. L. CRUZ, J. TOBAR, and P. MEJÍA, "Lukánikas: Educational platform for children with down syndrome manageable by the educator," in *IAENG TRANSACTIONS ON ENGINEERING SCIENCES: Special Issue for the International Association of Engineers Conferences 2016*. World Scientific, 2018, pp. 543–555.

[19] V. Robles-Bykbaev, E. Andrade-Prieto, P. Solorzano-Guerrero, Y. Robles-Bykbaev, F. Pesántez-Avilés, and A. Parra-Astudillo, "An educational support tool based on robotic assistants, mobile apps, and expert systems for children with down syndrome," in *2018 IEEE XXV International Conference on Electronics, Electrical Engineering and Computing (INTERCON)*. IEEE, 2018, pp. 1–4.

[20] R. S. El-Sayed, "A hybrid cnn-lstm deep learning model for classification of the parkinson disease." *IAENG International Journal of Applied Mathematics*, vol. 53, no. 4, 2023.

[21] M. E. Weijerman and J. P. De Winter, "Clinical practice: The care of children with down syndrome," *European journal of pediatrics*, vol. 169, pp. 1445–1452, 2010.

[22] B. Fauroux, S. Sacco, V. Couloigner, A. Amaddeo, A. Ravel, E. Prioux, J. Toulas, C. Cieuta-Walti, H. Walti, R. Luscan *et al.*, "Early detection and treatment of obstructive sleep apnoea in infants with down syndrome: a prospective, non-randomised, controlled, interventional study," *The Lancet Regional Health–Europe*, vol. 45, 2024.

[23] C. R. Galindo-López, J. Beltrán, C. B. Pérez, A. Macias, and L. A. Castro, "Classifying interactions of parents and children with down syndrome in educational environments using deep learning," in *2022 IEEE Mexican International Conference on Computer Science (ENC)*. IEEE, 2022, pp. 1–8.

[24] K. Simonyan and A. Zisserman, "Very deep convolutional networks for large-scale image recognition," *arXiv preprint arXiv:1409.1556*, 2014.

[25] K. He, X. Zhang, S. Ren, and J. Sun, "Deep residual learning for image recognition," in *Proceedings of the IEEE conference on computer vision and pattern recognition*, 2016, pp. 770–778.

[26] G. Huang, Z. Liu, L. Van Der Maaten, and K. Q. Weinberger, "Densely connected convolutional networks," in *Proceedings of the IEEE conference on computer vision and pattern recognition*, 2017, pp. 4700–4708.

[27] A. G. Howard, M. Zhu, B. Chen, D. Kalenichenko, W. Wang, T. Weyand, M. Andreetto, and H. Adam, "Mobilenets: Efficient convolutional neural networks for mobile vision applications," *arXiv preprint arXiv:1704.04861*, 2017.

[28] A. Dosovitskiy, L. Beyer, A. Kolesnikov, D. Weissenborn, X. Zhai, T. Unterthiner, M. Dehghani, M. Minderer, G. Heigold, S. Gelly *et al.*, "An image is worth 16x16 words: Transformers for image recognition at scale," *arXiv preprint arXiv:2010.11929*, 2020.

[29] M. Mujahid, A. Rehman, T. Alam, F. S. Alamri, S. M. Fati, and T. Saba, "An efficient ensemble approach for alzheimer's disease detection using an adaptive synthetic technique and deep learning," *Diagnostics*, vol. 13, no. 15, p. 2489, 2023.

[30] S. Ramanathan, M. Sangeetha, S. Talwai, and S. Natarajan, "Probabilistic determination of down's syndrome using machine learning techniques," in *2018 International Conference on Advances in Computing, Communications and Informatics (ICACCI)*. IEEE, 2018, pp. 126–132.