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Non-invasive Detection of Fetal Down Syndrome Using Femur and Intracranial Biomarkers on Ultrasound Images

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***Abstract*—Using 2D sagittal-view fetal ultrasound pictures taken between weeks 11 and 14, a non-invasive, AI-assisted method for the early diagnosis of Down syndrome (Trisomy 21).The conventional image processing techniques with advanced deep learning models to analyze two key biomarkers: femur bone length and intracranial structural morphology.Femur length is extracted through a structured image processing pipeline comprising preprocessing, enhancement, edge detection, segmentation, and measurement. The measured length is then compared against gestational age-specific norms to identify potential deviations indicative of chromosomal abnormalities.In parallel, deep learning-based object detection models—YOLOv5 and Faster R-CNN—are employed to detect and localize critical intracranial structures, including the cavum septum pellucidum (CSP), thalamus, brainstem, cerebellum, and cisterna magna. Accurate identification and morphological analysis of these structures are essential to assessing neurodevelopmental anomalies commonly associated with Down syndrome.The system is trained and evaluated on a dataset of 1,528 annotated sagittal-view ultrasound images obtained from Shenzhen People's Hospital. The results indicate reduced inter-observer variability and improved diagnostic reliability, enabling more consistent and faster clinical decision-making.**

Keywords*—*Down syndrome,Trisomy 21, Femur bone YOLOv5, R-CNN.

I. INTRODUCTION

Down syndrome, also known as trisomy 21, is a genetic disorder resulting from the presence of an extra copy of chromosome 21 in an individual's cells. This condition leads to a range of physical and intellectual challenges that vary in severity. The syndrome is named after John Langdon Down, the British physician who first identified the condition in 1862, although the genetic basis of the disorder was understood later [1]. Typically, a person has 46 chromosomesarranged in 23 pairs within each cell. Down syndrome is brought on by is a full or partial additional chromosome 21 copies, resulting in a total of three copies instead of the usual two. This extra genetic material changesfoetal development and affects an individual's physical and cognitive development.Maternal age is the most significant risk factor. Women over 35 years oldhave a higher probability of giving birth to a child that has Down syndrome due to older eggs have an increased chance of undergoing nondisjunction. Existing Methods to Detect Foetal Down SyndromeSeveral methods exist to detect foetal Down syndrome, ranging from non-invasive screening tests to invasive diagnostic procedures [2]. Screening tests are usually the initial step and help assess the risk of Down syndrome. First-trimester screening combines a blood test measuring PAPP-A and free β-hCG with a nuchal translucency ultrasound, which measures the fluid-filled space at the back of the foetus's neck to assess 32 the probability ofDown syndrome. Second-trimester screening (quadruple screen) measures four substances in the mother's blood—AFP, hCG, estriol, and inhibin A—to estimate the risk, typically performed 15-20 weeks of pregnancy. Cell-free DNA (cfDNA) testing is a more recent, non-invasive screening method that analyses foetal DNA circulating in the mother's blood, providing A high level of precision (over 99%) in detecting Down syndrome as early as 10 weeks. Additionally, ultrasound can be applied toidentify certain markers associated withDown syndrome. In the first trimester [8][9].

II. L TERATURE SURVEY

Thisliteratureanalysissectionaimstoestablishacontextual framework for the application of deep neural networks in pediatric Down syndrome diagnosis. This involves a thoroughexplorationofThe progression of historyofimage-based diagnostictechniques,emphasizingkeymilestones, methodologies, as well as developments in technology that have madethe integration of deep learning algorithms.

In this study[1], the authors proposed a novel experimental finding for detecting obstructive sleep apneain 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.

Inthisreviewresearch[2],theauthorsanalyseddifferent machine learning algorithms to identify Obstructive Sleep Apnea(OSA).The information utilized in thisexperimentwascollected by two researchers from the Web of Science and Scopus, employingvariouselectronicresearchmethods.Finally, 19 studies with 4767 separate pediatric sleep investigations were conducted. Machine learning demonstrated improved

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diagnostic performance, achieving its best results for AHI =10 e/h, with 0.652 sensitivity, 0.931 specificity, and a0.940 area under the SROC curve.

In this research [3],an ultrasound (US) imaging system method is suggested fordetectingDownsyndrome because of its safety, cost-effectiveness, and non-invasiveness. The MediscanFetalCareResearchFoundationfromIndiaprovides the dataset used in this research. The US fetal imagedatasetcontains100fetuses,with50healthyand50 Down syndrome fetuses between 11-14 weeks of gestation. By employing using various Deep Learning models, including a ConvolutionalNeuralNetwork(CNN)andVisualGeometry Group (VGG-16), it was discovered that the most effective algorithmachievingthehighestaccuracyisVGG-16,witha 91% accuracy.

In this research [4], the authors identify the behaviorsof children and parents with Down syndrome using human activity recognition techniques. They utilized advanced technologies and video-based data to analyse the activities and behavior of individuals with Down syndrome. The experimentalfindingsfromthevideodataset,employingthe deeplearningmodelC3D,demonstratedanaccuracyof85%. The physical interventions were also detected.

In this research [5], a prenatal screening method for predictingDownsyndromewasproposed,aimingtoenhance theaccuracyofdiagnosis.Thiswasachievedbyemployinga multi-branchCNNmodelintegratedwithafeaturerearrangement technique [6],[7]. To configure the CNN model,thesuggestedfeaturerearrangementstrategyutilizedfeature grouping and Pearson correlation testing.

Inthisresearch[8],Identification of Down syndrome through facial featuresisproposed.Thedataisdistinguishedintotwoparts: general face features and detailed face features. The information utilized in thisexperiment is the CASIA-Web FACE dataset, which is a publicly available dataset including 493,750 images with 10,562 individuals. A deep learning approach, namedConvolutionalNeuralNetwork(CNN),wasemployed in this experiment. In Down Syndrome identification, the deep convolutional neural network accomplished an accuracy of 95.87%, a recall of 93.18%, and a precision of 97.40%.

In this research [9], a method is proposed to detect Downsyndromeusingfacialrecognition.Inthisexperiment, 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 texturefeatures,achievinganaccuracyof97.92%alongwith good precision and recall for the combined features.

III METHODOLGY

Our proposed innovative research methodology for early, non-invasive detection of fetal Down syndrome using ultrasound images is based on a dual-path approach that integrates both traditional image processing and advanced deep learning techniques. This comprehensive workflow is designed to enhance diagnostic accuracy and support clinicians in making early, reliable diagnoses.

Thebelow flowchart represents thecomplete methodology foran AI-based system aimed at the early, non-invasive detection of fetal Down syndrome using ultrasound images. The procedure begins with the acquisition of sagittal-view ultrasound images taken during 11–14 weeks of gestation. The workflow is divided into two main parallel pipelines:one focused on femur bone analysis and the other on intracranial structure detection.

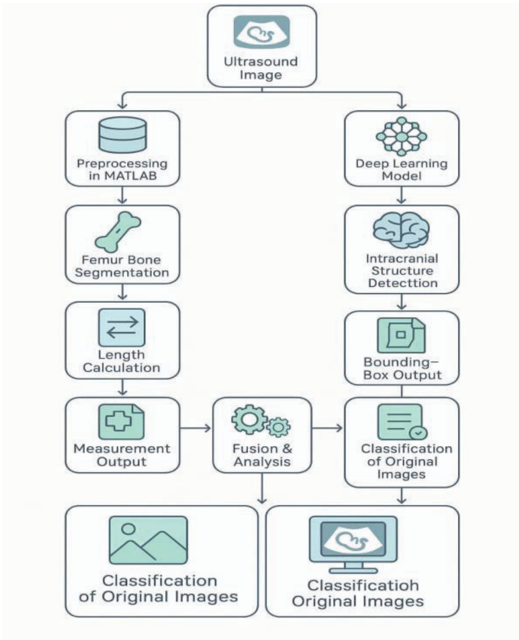

***Fig:1**Flowchart of methodology*

A. Left Branch

Femur Bone Analysis (MATLAB-Based)

- Preprocessing in MATLAB

Ultrasound images are pre-processed using MATLAB to enhance quality Operations like denoising, contrast enhancement, and ROI extraction are applied.

- Femur Bone SegmentationThe femur bone region is segmented using edge detection or thresholding. This is crucial since femur length is a key biomarker in Down syndrome prediction.
- Length CalculationThe segmented femur bone's length is calculated by determining the pixel distance between the two endpoints.
- Measurement Output The computed femur length is converted to gestational age standards and used for clinical reference.

  This metric is forwarded to the fusion module.

B. Right Branch

Intracranial Structure Detection (Deep Learning-Based)

- Deep Learning Model YOLOv5 and Faster R-CNN are applied to detect specific foetal intracranial structures (like nasal bone, midbrain, thalami, etc.).
- Intracranial Structure Detection Models localize key anatomical regions using bounding boxes around each structure.
- Bounding Box Output

  The detected bounding boxes are extracted and stored for analysis.
- Classification of Original Images Based on the spatial arrangement and sizes of detected structures, the system classifies whether the features are typical or abnormal.

C.Fusion and Analysis

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Input Fusion: Outputs from both branches—femur length (measurement output) and bounding boxes (intracranial detection)—are merged.

- Analysis: Based on defined clinical rules and measurements, the system predicts whether Down syndrome markers are present. If anomalies are detected in either or both feature sets(short femur length or abnormal intracranial structure positions), the image is flagged.

IV RESULTS

Our study's results and discussion aimed to make it easier and more accurate to find signs of Down syndrome in unborn babies using ultrasound images. We built a system that uses computer software and artificial intelligence (AI) to look at these images.The software we used is called Skylab, which is free and good for working with images and numbers. The Images and We worked with a large set of ultrasound images-1,528 in total. These images were taken from pregnant women at Shenzhen People's Hospital.

All the images show the baby from the side (sagittal view) and were taken during weeks 11 to 14 of pregnancy. This time is important because it's when doctors can best spot early signs of Down syndrome.Doctors had already marked important parts on each image. These included:

The femur bone (thigh bone)
The nuchal fold (skin at the back of the baby's neck)
Cavum Septi Pellucidum (CSP), a part of the brain
The cerebellum, cisterna magna, brainstem, and thalamus (other brain areas)

These marks helped our computer system know exactly where to look for signs of Down syndrome.

We used two popular AI models: YOLOv5 and Faster R-CNN. These models are excellent at finding and pointing out objects in pictures. We trained the models using 25 images of babies who were known to have Down syndrome. While this is not a lot, each image was carefully labeled, which helped the computer learn better.

The training happened over five rounds. Each round, the computer tried to get better at finding the important parts in the images. We adjusted the model's settings each time to help it learn faster and more accurately.After five rounds of training, the computer system could correctly spot Down syndrome markers in 80% of the images. This means that out of every 10 images, it got 8 right. The "rejection rate"-how often it missed the markers-was 20%.

During training, the AI model learned to tell the difference between normal and Down syndrome fetal ultrasound images by looking for key anatomical features.

After training, the model used its knowledge to map out the most important features (biomarkers) that signal Down syndrome. This mapping is key for the AI to classify images correctly. Table 2 (not shown here) lists these main features and what they look like in Down syndrome cases. Some of the most important

By focusing on these features, the AI can better detect Down syndrome in ultrasound images, helping doctors with early and non-invasive screening.

difference indicates how closely the "measurement criteria" aligns with the "feature" or the extent of their "positive correlation."

Fig 2: *Original image of femur bone*

Fig 3: Enhanced image

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Fig 4: *Segmented image*

Fig 5: ***Femur** length detected*

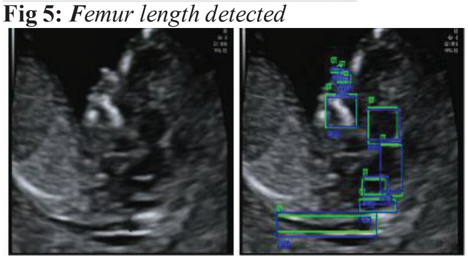

Fig:6 *Results of training phase*

Fig7:*Femur bone detected*

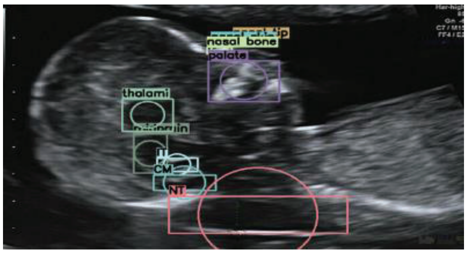

Fig 8:*Fetal down syndrome detected*

a) Performance analysis using evalution metrics

using parameters such as sensitivity,specificity and accuracy. Sensitivity is the number of true positivies that are rightly acknowledged by a diagnostic test. It states hoow well the diagnostic test is identifying a disease.

Sensitivity=T.P/(T.P+F.N) (1)

Specificity is the quantity of the true negatives rightly acknowledged by a diagnostic test. It determines how good the test is identifying normal (negative)condition.

Specificity=T.N/(T.N+F.P) (2)

Accuracy is the quantity of actual outcomes in a population that are either true positives or true negatives. It gauges how accurate a diagnostic test is for a given ailment.

Accuracy=(T.N+T.P)/(T.N+T.P+F.N+F.P) (3)

(Note: T.P stands for true positive,T.N stands for true negative ,F.P stands for fake positive ,F.N stands for false negative)

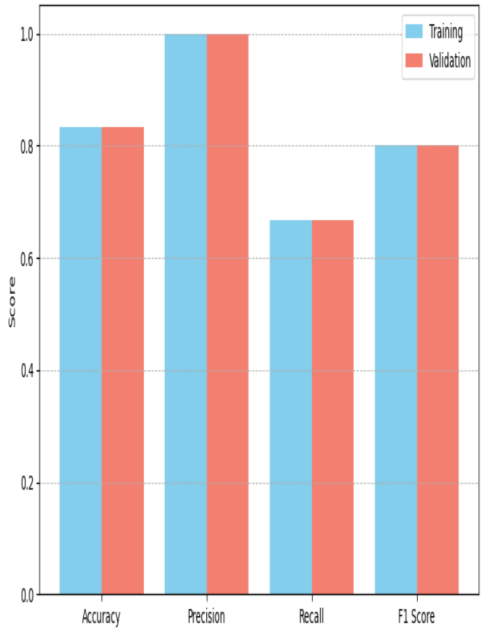

Fig 9:*Comparision of state of art*

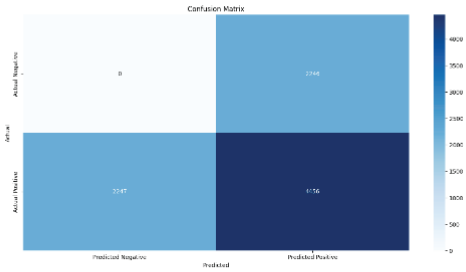

Fig 10:*Confusion matrix of actual image & predicted images*

Sl no	Feature name	Measurement criteria	DS positive correlation
1	10	10	0
2	20	14	6
3	40	34	6
4	80	68	12
5	100	86	14

TABLE I. MODEL TRAINING PARAMETERS AND PERFORMANCE

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The above table represents log of the process evolved over several iterations, detailing the amount of data processed the learning and the data quality control outcomes at each stage.

Iteration No#	No.OfInputImages	LearningRate	RejectionRate
1	50	10	0
2	100	14	6
3	200	34	6
4	30	68	12
5	400	86	14

b) Final Output

Original Image: Saved for reference and comparison.

Foetal Down Syndrome Detected Image: If the fusion analysis indicates high Down syndrome probability, the image is annotated and flagged for review.

I. CONCLUSION

This study proposes an innovative methodology foe the early diagnosis of down syndrome using an ultrasound picture of unborn infants aged eleven to fourteen weeks. The goal is to aid physicians in identifying early indicators ofDown syndrome without needing any risky tests. The proposed methodology focuses on two important things in the ultrasound images: The baby's thigh bone (femur): Doctors know that the length of this bone can sometimes be different in babies with Down syndrome. Certain parts inside the baby's head: Specifically, it looks at the shape and size of a few key structures like the cavum septum pellucidum (CSP), thalamus, and cisterna magna. Changes in these areas can also be linked to Down syndrome. To do this, the project uses powerful computer programs called "deep learning models." Two main models are used: YOLOv5: This model is really good at quickly finding objects in images. It helps to quickly locate thefemur bone and the important parts inside the baby's head. Faster R-CNN: This model is a bit slower but more precise. It's used to carefully examine the details of the structures inside the head, which is important for spotting subtle differences.Normally, doctors have to manually measure the femur bone and carefully look at the brain structures in theultrasound images. This can take time, and different doctors might see things slightly differently.This new AI system can do these tasks automatically, making the process faster and more consistent. It also reduces the chance of human error.Think of it like having a very experienced assistant for the doctor. The AI can highlight the areas the doctor needs to focus on, maybe even put measurement lines on the screen, and suggest if anything looks unusual. This can assist physicians in making choicesmore quickly, especially in places where there aren't many specialists available.A big advantage of this system is that it's non-invasive. This means it doesn't involve any procedures that could harm the baby or the mother, unlike tests like amniocentesis. It just uses standard ultrasound images, which are already commonly used. This makes the system cheaper and easier to use in most hospitals, even in more rural areas.Overall, this project shows that AI can be a powerful tool for improving how we screen for Down syndrome early in pregnancy. It offers a way to make the process more accurate, faster, and safer, ultimately helping to provide better care for both mothers and their babies.

I. FUTURE SCOPE

The first phase of utilizing computers to help identify possible indications of Down syndrome in unborn children is well-explained in this publication. The current system uses regular 2D ultrasound images. But there are also 3D ultrasounds and ultrasounds that show blood flow. If the AI could gain additional knowledge from these detailed images, it could get a much betterunderstanding of the baby. While our current system shows strong potential in helping detect Down syndrome early by looking at femur length and brain features, there are multiple methodswe can improve it in the future: The model is trained on 1,528 images. To make it more accurate and reliable, we need to train it on more images from different populations, using various ultrasound machines and from different stages of pregnancy.Adding support for 3D or even 4D ultrasound scans could help the system see the baby's structures more clearly and detect features more accuratelyespecially inside the brain. We plan to connect the AI system directly to ultrasound machines. This would allow the system to give doctors instant feedback during live scans, helping them make faster and better decisions. In the future, the system could look at more signs of Down syndromelike the nasal bone or signs in the abdomen. This would make the diagnosis more accurate and help detect other genetic conditions too.

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