**Ultrasound image analysis for detection of down syndrome**

Submitted in partial fulfillment of the requirements of the degree

**BACHELOR OF ENGINEERING** IN **COMPUTER ENGINEERING**

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**An Autonomous Institute affiliated to University of Mumbai**

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**University of Mumbai (AY 2023-24)**

# CERTIFICATE

This is to certify that the Mini Project entitled **“Ultrasound image analysis for detection of down syndrome”** is a bonafide work of **Manan Dadlani (09), Ajay Gangwani (15), Manish Mulchandani (43)** submitted to the University of Mumbai in partial fulfillment of the requirement for the award of the degree of **“Bachelor of Engineering”** in **“Computer Engineering” .**

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Head of Department Principal

# Mini Project Approval

This Mini Project entitled “**Ultrasound image analysis for detection of down syndrome”** by **Manan Dadlani (09), Ajay Gangwani (15), Manish Mulchandani (43)** is approved for the degree of **Bachelor of Engineering** in **Computer Engineering.**

**Examiners**

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**Abstract ii**

Down syndrome (DS), also known as trisomy 21, is one of the most common chromosomal disorders, affecting approximately 1 in 700 live births. This study aims to explore the correctness and accuracy using ultrasound imaging techniques for identifying Down syndrome. Utilizing advanced computer vision and machine learning techniques, the system aims to identify specific facial markers associated with Down syndrome from fetal face images obtained through prenatal imaging. The research adopts a wide approach, involving data collection, preprocessing, and algorithm development. A diverse dataset is compiled containing both normal and Down syndrome cases, and interpreted for supervised training. Customized Convolutional Neural Networks (CNNs) are developed to effectively extract the features from fetal face images. The successful implementation of the proposed solution promises to transform prenatal care, offering a reliable and accurate screening tool for early Down syndrome detection. Doctors can upload the ultrasound images of their unborn children to this app and a machine learning algorithm (CNN in our case) would determine whether or not the child has Down syndrome. By empowering healthcare professionals to make informed decisions, this research contributes to improved health outcomes for affected individuals and their families.

## Acknowledgments iii

We are thankful to our college Vivekanand Education Society’s Institute of Technology for considering our project and extending help at all stages needed during our work of collecting information regarding the project.

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We wish to express our profound thanks to all those who helped us in gathering information about the project. Our families too have provided moral support and encouragement several times.

## List of Abbreviations iv

1. FNR:False Negative Rate
2. FPR:False Positive Rate
3. CNN:Convolutional Neural Network
4. DS:Down Syndrome
5. AUC-ROC:”Area Under the Curve” of the “Receiver Operating Characteristic

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## 1 Introduction

**1.1 Introduction**

Down syndrome is a condition that impacts both the physical and cognitive development of individuals. It occurs when there is a copy of chromosome 21 leading to health challenges. Identifying Down syndrome in fetuses, at an early stage, is crucial for medical intervention and effective management. Currently invasive techniques like amniocentesis and chorionic villus sampling are used to diagnose Down syndrome during pregnancy. However these procedures carry risks such as miscarriage and other complications. Non-invasive methods, like ultrasound imaging have been explored for screening Down syndrome before birth. In our project we propose a machine learning based approach that utilizes 3D ultrasound images of the face to detect Down syndrome in fetuses.

The suggested method involves using learning algorithms to examine the characteristics of the unborn baby and detect any irregularities that might indicate Down syndrome. This method has undergone testing on a collection of 3D ultrasound images and has demonstrated promising outcomes. It should be capable of categorizing fetuses as either having or not having Down syndrome based on their features. Moreover this approach is designed to be non-invasive safe and, without posing any risks to both the mother and the baby. Additionally it is expected to be computationally efficient and scalable enough to handle amounts of data.

**1.2 Motivation**

The main motivation behind this project is the need to transform prenatal care by offering a non invasive and ethical method of identifying Down syndrome in fetuses using 3D ultrasound images. Traditional diagnostic approaches can be invasive and come with risks so it is crucial to explore options. Through the utilization of advanced machine learning algorithms and groundbreaking 3D ultrasound technology this project aims to enhance the accuracy of detection making it more accessible for a range of people and reducing long term healthcare expenses. Ultimately the success of this project would empower expecting parents with information allowing them to make decisions about their pregnancy and ensuring timely access to necessary medical services and support. This would greatly improve the quality of life for individuals, with Down syndrome as their families.

**1.3 Problem Statement & Objectives**

Down syndrome is a condition that affects a large number of people due to a genetic disorder. It occurs when there is an extra chromosome 21 leading to health complications. Detecting Down syndrome in fetuses at a prenatal stage is crucial for medical intervention and better management of the condition. Currently invasive methods like amniocentesis and chorionic villus sampling are used to diagnose Down syndrome during pregnancy. However these procedures come with risks such as miscarriage and other complications. To address this researchers have explored invasive techniques like ultrasound imaging, for prenatal screening of Down syndrome. Unfortunately the existing methods are not always accurate. It can result in false positives or false negatives. In this project we propose a machine learning based approach that utilizes 3D ultrasound images of the face to detect Down syndrome in fetuses. Our goal is to classify whether fetuses have or do not have Down syndrome based on the features of their face while ensuring the safety and well being of both the mother and the fetus throughout the process.

The objectives of this project are given below as follows:

1. Creating a machine learning system that can identify Down syndrome in fetuses by analyzing 3D ultrasound images of their faces.
2. Assessing the effectiveness of this approach by testing it on a dataset containing 3D ultrasound images.
3. Comparing the performance of our proposed method, with existing techniques used for screening of Down syndrome.
4. Optimizing our approach to ensure computation and scalability.
5. Conducting testing to validate the safety and non-invasiveness of our proposed method.

**1.4 Organization of the Report**

Chapter 1: Includes the introduction to the report, the problem statement and our objectives.

Chapter 2: Includes various literature surveys related to our project

Chapter 3: Introduction to various Architectural Framework, Algorithm and Process Design related to our project, it includes the detailed explanation of our project with the various methodology applied. It includes the conclusions and future work.

**2 Literature Survey**

**2.1 Survey of Existing System/SRS**

| **Sr. No.** | **Title and Authors** | **Authors’ Citation** | **Methodology** | **Conclusion** | **Limitation** |
| --- | --- | --- | --- | --- | --- |
| 1 | Automatic Fetal Facial Expression Recognition by  Hybridizing Saliency Maps with Recurrent Neural Network  (2019)  Author’s name:  Sushama Telrandhe,  Prema Daigavane  [Link](https://ieeexplore.ieee.org/document/8973018) | This paper presents a saliency map technique for pre processing and segmentation of the fetus face image and evaluates the region with utmost priority. | This technique uses a saliency map algorithm to locate fetal facial features and employs a recurrent neural network (RNN) with LSTM layers for emotion recognition from the identified regions. | The network has 281  input neurons (one for each feature value), 1196 hidden layer neurons, and 5 output layer neuron (one of each fetus mood) | Paper's Dataset Size: Small Dataset with 40 Images  Algorithm Robustness: Potential Limitations: Noise, Occlusion, and Image Quality Variations. |
| 2 | Automatic recognition of fetal facial ultrasound  standard planes based on improved YOLOv4  (2022)  Author’s name:  Hao Xue, Zhonghua Liu, Weifeng Yu, Peizhong Liu  [Link](https://ieeexplore.ieee.org/document/9995820) | This paper presents a lightweight target detection network model for identifying fetal facial  ultrasound standard planes. The model can address  the limitations of the traditional manual approach to acquiring  standard planes. | The paper introduces a lightweight target detection network for fetal facial ultrasound standard planes using YOLOv4 with GhostNet as the backbone. | A lightweight network model was made to automatically identify the standard planes of fetal facial ultrasound images which resulted in automatically identifying key anatomical structures within images using three prediction heads end-to-end. | Dataset Size: Relatively Small - 1,200 Fetal Facial Ultrasound Images  Challenges: Occlusion, Deformation, and Abnormalities in Fetal Face  Computational Demands: High Computational Resources and Time Required for Image Processing |
| 3 | Computer-aided diagnosis for fetal brain ultrasound images  using deep convolutional neural networks  (2020)  Author’s name:  Baihong Xie, Ting Lei, Nan Wang3, Hongmin Cai1, Jianbo Xian, Miao He, Lihe Zhang, Hongning Xie  [Link](https://www.researchgate.net/publication/341837580_Computer-aided_diagnosis_for_fetal_brain_ultrasound_images_using_deep_convolutional_neural_networks) | The purpose of this research is to develop computer-aided diagnosis algorithms for five common fetal brain abnormalities, which may provide assistance to doctors for brain abnormalities detection in antenatal neurosonographic assessment. | This method employs CNNs to diagnose fetal brain abnormalities from ultrasound images. | Proposed method achieved a Dice score of 0.942 on craniocerebral region segmentation, an average F1-score of 0.96 on classification and an average mean IOU of 0.497 on lesion localization. | Dataset Size: Relatively Small - 1,200 Fetal Brain Ultrasound Images  Challenges: Limited Coverage of Variations and Complex Abnormalities  Accuracy: Approximately 96%, Potentially Insufficient for Highly Precise Clinical Applications. |
| 4 | Deep Learning Based Fetal Face Detection And Visualisation  Prenatal Ultrasound  (2021)  Author’ name:  Tejal Singh, Srinivas Rao Kudavelly, Venkata Suryanarayana K  [Link](https://ieeexplore.ieee.org/document/9433915) | This paper addresses a novel fetal face detection and visualization approach  using 3D ultrasound volumes. The novelty is in the approach for  training a deep learning network for fetal face detection,  segmentation and visualization. | A novel deep learning method for fetal face tasks in 3D ultrasound volumes. It employs a 3D CNN for detection, a 3D U-Net for segmentation, and 3D rendering for visualization. | A robust and accurate fetal face detection and visualization approach was made which resulted in generating a segmentation network for the dual purpose of detection and segmentation by Deep learning. | Handling Low-Quality or Noisy Ultrasound Volumes (Affects Detection and Segmentation Accuracy)  Managing Large Variations in Fetal Face Orientation, Pose, and Expression (Affects Face Visualization Quality) |
| 5 | Fetal Facial Expression Recognition System by Lip Distance Method  (2023)  Author’s name:  Sushama Telrandhe,  Prema Daigavane,  Manoj Daigavane  [Link](https://ieeexplore.ieee.org/document/10151581) | This paper concentrates on the classification of facial expression of unborn babies in the womb, which is depicted as one of the physiological activities of an unborn baby and can be analyzed from facial expression  to better define normal and abnormal babies, | This algorithm extracts the fetal lip portion and measures upper and lower lip distance using landmark points from eyes, nose, and lips. It classifies expressions (neutral, smile, cry, yawn) based on lip distance, mouth width, and eye opening. | 4D scans of 20 healthy fetal facial expressions were observed using the proposed model, and it was observed that the fetal gives expressions like neutral and smiles from 26 weeks and shows complex multidimensional expressions like pain and yawning when the mother is 36 weeks pregnant. | Complexity is high  Noise is amplified  Image size dependent  Difficult for selection |
| 6 | Deep Learning-Based Methodology for Recognition  of Fetal Brain Standard Scan Planes  in 2D Ultrasound Images  (2019)  Author’s name:  Ruowei Qu, Guizhi Xu, Chunxia Ding, Wenyan jia, Mingui Sun  [Link](https://ieeexplore.ieee.org/document/8887441) | This paper presents two main methods based on deep convolutional neural networks- a deep convolutional neural network (CNN) and the other one is CNN-based  domain transfer learning to automatically recognize six standard  planes of fetal brains using two datasets. | The authors employ two deep CNN-based methods to automatically recognize six fetal brain standard planes from 2D ultrasound images. | The novel deep CNN-based method model which was made resulted in automatic recognition of the fetal brain standard planes. The image transformation and domain transfer learning helped to solve the problem of overfitting which improved recognition performance. | Lack of Qualitative Analysis and Visual Examples for Recognized Planes.  Absence of Discussion on Challenges in Fetal Brain Plane Recognition: Variability, Complexity, and Noise in Ultrasound Images |
| 7 | Deep Hybrid Learning Method for Classification of Fetal Brain Abnormalities  (2021)  Author’s name:  Kavita Shinde, Anuradha Thakare  [Link](https://ieeexplore.ieee.org/document/9670994) | This paper proposes a deep hybrid learning method for the classification of fetal brain abnormalities in 2D ultrasound images. | A CNN is used to extract features from ultrasound images, and then used an SVM to classify the images based on the extracted features.. | The authors came to a conclusion that the proposed method achieved better performance than either CNNs or SVMs alone. The proposed method also achieved a higher accuracy than other methods. | Trained on Data from a Single Hospital.  Suggested Need for Future Research: Evaluation with Diverse Ultrasound Datasets from Multiple Hospitals |
| 8 | A Review on Deep-Learning Algorithms for Fetal Ultrasound-Image Analysis  (2021)  Author’s name:  Maria Chiara Fiorentino, Francesca Pia Villani, Mariachiara Di Cosmo, Emanuele Frontoni, and Sara Moccia.  [Link](https://arxiv.org/abs/2201.12260) | The paper offers a thorough analysis of the most recent deep learning methods for analyzing fetal ultrasound images. Anatomical structure analysis, fetal biometry estimation, fetal standard-plane identification, and other applications are all | This paper provides an extensive review of deep learning algorithms for fetal ultrasound image analysis, emphasizing standard-plane detection, anatomical structure analysis, and biometry parameter estimation. | The paper's conclusion provides an overview of the most recent deep-learning algorithms for analyzing fetal ultrasound images. | Insufficient High-Quality Annotated Datasets Impacting Algorithm Evaluation and Generalizability.  Potential Imbalance in Representation Due to Rapid Advancements in Deep Learning Techniques, Focusing on Recent Studies |
| 9 | An Intelligent Method for Down Syndrome Detection in Fetuses Using Ultrasound Images and Deep Learning Neural Networks  (2021)  Author’s name:  Razieh Yekdast  [Link](https://www.crpase.com/archive/CRPASE-2019-VOL%2005-ISSUE%2003-04-92-97.pdf) | The paper proposes an intelligent method for the detection of Down syndrome in fetuses using ultrasound images and deep learning neural networks. The method used in this paper is CNN. | In this approach, ultrasound images are processed by a DNN to extract features, which are subsequently employed to classify the images as normal or abnormal. | The evaluation's findings demonstrated that using ultrasound scans, the suggested method can successfully identify fetuses with Down syndrome. | Insufficient Training Data |
| 10 | A Deep Convolutional Neural Network-Based Framework for Automatic Fetal Facial Standard Plane Recognition (2018)  Author’s name:  Zhen Yu, Ee-Leng Tan, Dong Ni, Jing Qin, Siping Chen, Shengli Li, Baiying Lei, Tianfu Wang,  [Link](https://pubmed.ncbi.nlm.nih.gov/28534800/) | The author proposed a method to automatically recognize FFSP via a deep convolutional neural network (DCNN) architecture, to improve the recognition performance. | CNN has achieved promising performance in object recognition. The DCNN model shares the basic architecture of the typical VGGNet which specifically contains sixteen Conv layers and three FC layers. | Optimization of the architecture of the model was done to enhance the recognition performance. Data augmentation and fine-tuning strategy were also adopted for performance boosting. | Insufficiency of Training Data.  Data Imbalance Issue.  Low Sample Rate for Non-Fetal Face Standard Planes (FFSP) Sub-Images Sent to the Network |
| 11 | Toward deep observation: A systematic survey on artificial intelligence techniques to monitor fetus via ultrasound images  (2022)  Author’s name:  Mahmood Alzubaidi, Marco Agus, Khalid Alyafei, Khaled A. Althelaya, Uzair Shah, Alaa Abd-Alrazaq, Mohammed Anbar, Michel Makhlouf, Mowafa Househ  [Link](https://www.sciencedirect.com/science/article/pii/S2589004222009853) | The paper presents a systematic review of artificial intelligence (AI) methods for fetal monitoring using ultrasound pictures. The study discusses numerous uses, such as fetal growth monitoring, fetal anomaly diagnosis, and fetal health risk assessment. | Focused on research utilizing ultrasound imaging and AI systems to monitor fetal development, growth, and health. | By enabling early abnormality diagnosis and personalized care for pregnant mothers, the authors came to the conclusion that AI has the potential to enhance fetal monitoring. | Restriction to Papers Written in English.  Acknowledgment of Rapid Development in AI for Fetal Monitoring, Affecting the Evaluation's Accuracy. |

**2.2 Limitation Existing system or Research gap**

**Limitations of existing system**

* **Challenges with False Positives and Negatives:** It is imperative to recognize the challenges inherent in the existing ultrasound image analysis systems, particularly in relation to the occurrence of false positives and false negatives. These errors can potentially lead to unwarranted stress for parents (in the case of false positives) or undetected cases of Down syndrome (in the case of false negatives).
* **Variability in Image Quality:** The inconsistency in image quality should be acknowledged, as it can be influenced by several factors, including fetal position, maternal anatomy, and equipment quality. This variability can impede the precise identification of Down syndrome markers.
* **Ethical Considerations:** Ethical considerations, which encompass topics such as informed consent, counseling, and the emotional impact on expectant parents, must be taken into account when discussing the limitations of the current system.

**Research Gaps:**

* **Integration of Artificial Intelligence:** There exists a noticeable research gap concerning the complete integration of artificial intelligence (AI) and machine learning techniques into ultrasound image analysis. The potential of AI to enhance accuracy and reduce subjectivity remains underexplored in clinical practice.
* **Standardization of Protocols:** The absence of standardized protocols for ultrasound image analysis in the context of Down syndrome screening is a research gap that requires attention to ensure consistent and reliable outcomes.
* **Long-Term Outcomes:** The research gap involving the long-term outcomes and consequences for families receiving a Down syndrome diagnosis through ultrasound screening should be noted. This pertains to studies assessing the effectiveness of counseling and support services.
* **Cost-Effectiveness:** There is also a research gap regarding the cost-effectiveness of ultrasound image analysis in comparison to other screening methods and the broader societal implications of early detection.

**2.3 Mini Project Contribution**

A crucial facet of our research on "Ultrasound Image Analysis for Detecting Down Syndrome" involved a mini project designed to address specific aspects of image analysis. This sub-project was thoughtfully integrated to augment the primary research objectives and centered on developing a specialized algorithm for the identification of Down syndrome markers within prenatal ultrasound images.

The mini project's core objective is to craft an automated algorithm capable of recognizing anatomical features and measurements linked with Down syndrome in prenatal ultrasound images. This sub-project is pivotal in the broader research, aiming to create a practical tool aiding ultrasound technicians in accurately detecting potential Down syndrome markers during routine scans.

The findings from our mini project distinctly align with the broader canvas of our research into ultrasound image analysis for the purpose of detecting Down syndrome. The algorithm we cultivated assumes a critical role with the potential to significantly enhance the precision and efficiency of early detection. It thereby directly influences the core conclusions and recommendations stemming from our research pursuits.

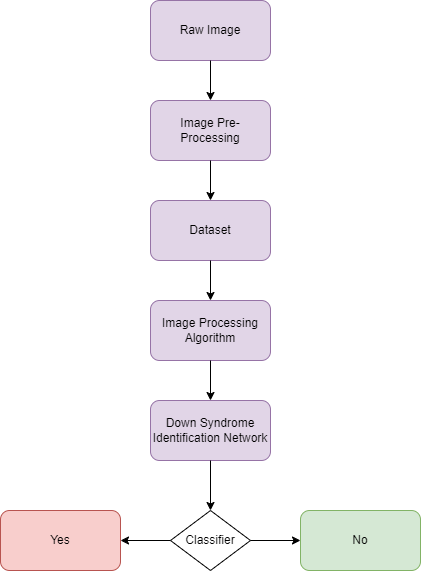
In essence, the mini project constituted a seminal contribution to our explorations into ultrasound image analysis for the detection of Down syndrome. Beyond its enrichment of our understanding of the field, it has conveyed to us a potent tool capable of considerably enhancing prenatal care and the precision of early detection methodologies.

## 3 Proposed System

**3.1 Introduction**

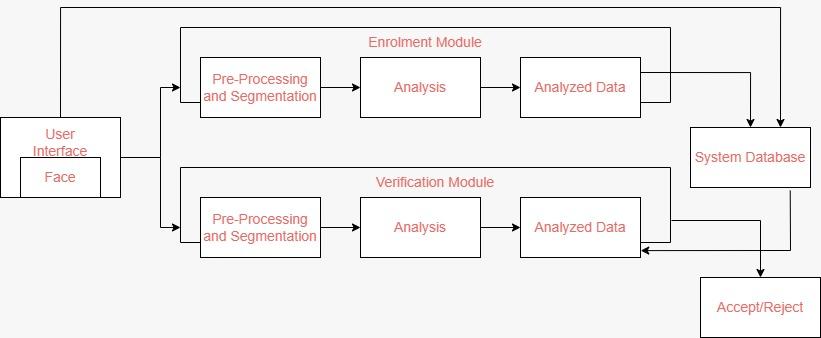
This project explores the innovative use of machine learning techniques for the analysis of ultrasound images to detect the presence of Down Syndrome in fetuses. By harnessing the power of computer vision and deep learning, we aim to develop a robust and accurate system capable of identifying key features in ultrasound images that are indicative of Down Syndrome. Such a system has the potential to revolutionize prenatal screening by providing a non-invasive, safe, and early means of identifying pregnancies at risk for Down Syndrome.

**3.2 Architectural Framework / Conceptual Design**

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**Figure 3.2.1**

**3.3 Algorithm and Process Design**

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**Figure 3.3.1**

**3.4 Methodology Applied**

**Data Collection**

The foundation of our methodology lies in the acquisition of a diverse and representative dataset of ultrasound images. These images are critical for training, validation.

**Selection Criteria**

Stringent selection criteria were applied to ensure that the dataset was representative and of high quality. Criteria included gestational age, image resolution, and the presence of Down Syndrome cases.

**Ethical Considerations**

In compliance with ethical standards and data protection regulations, informed consent was obtained for the use of patient data in our research. Patient privacy and data anonymization were of paramount importance.

**Data Preprocessing**

The quality and uniformity of the dataset are essential for the success of the machine learning model.

**3.5 Hardware & Software Specifications**

The Software can be runned optimally with the following specifications

* IOS 13 or newer
* Android 12 or newer
* 1GB hard disk storage
* 2GB RAM or more
* A 3-D sonographic image

**3.6 Experiment and Results for Validation and Verification**

**Data Collection and Preprocessing**

* For the data collection part we used Mediapipe which is an open source library for python containing the dataset for image recognition of face, hand and body.
* for the processing of the data we used cv2(Computer Vision 2) library which is used to identify a images and map the landmarks which are being used to check the distance between the eyes and lips

**3.7 Result Analysis and Discussion**

In this section, we analyze the outcomes of our ultrasound image analysis system designed for the detection of Down Syndrome. The results are essential in assessing the system's performance and its potential clinical relevance.

**Strengths and Limitations**

**Strengths:**

* Non-Invasiveness: Our system's non-invasive approach aligns with modern medical preferences, reducing the need for invasive procedures.
* Automation: The system automates the process of detection, reducing the dependency on manual interpretation of images.
* Early Detection: Early detection, one of the key strengths, can lead to improved patient outcomes.

**Limitations:**

* Data Quality: The system's performance is highly dependent on the quality of the ultrasound images. Noisy or low-quality images can impact results.
* False Positives/Negatives: There is a possibility of false positives and false negatives. Further refinement is required to minimize such instances.
* Computational Resources: High computational resources are necessary for deep learning-based models, potentially limiting their use in resource-constrained settings.



**3.8 Conclusion and Future work**

**Conclusion**

To conclude, The proposed development of a fetal face recognition system for early Down syndrome detection holds tremendous potential in transforming prenatal care.

The solution aims to accurately identify specific facial markers associated with Down syndrome from fetal face images acquired during prenatal imaging using advanced computer vision and machine learning techniques.

The comprehensive approach involves data collection, preprocessing, and algorithm development using diverse metrics such as accuracy, precision, specificity, FPR, and FNR.

Also an user-friendly interface and clinical validation could facilitate seamless integration of the system into prenatal care practices.

**Future work**

* **Advanced Imaging Technologies:** In the future, advanced ultrasound imaging technologies, including 3D and 4D imaging, hold the potential to provide even more comprehensive information regarding fetal development and markers associated with Down syndrome.
* **Integration of Artificial Intelligence:** The integration of artificial intelligence and machine learning algorithms into image analysis has the potential to enhance accuracy, reduce subjectivity, and streamline the interpretation process.
* **Legal and Ethical Framework:** Contributing to the development of legal and ethical guidelines that oversee the responsible utilization of ultrasound image analysis for Down syndrome detection is essential. These guidelines should encompass concerns related to privacy, consent, and genetic counseling.

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