Advancing Cleidocranial Dysplasia Diagnosis: A Novel Deep Learning Tool Utilizing Thoracic Radiograph Images

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INTRODUCTION

Cleidocranial dysplasia (CCD) is a rare genetic autosomal dominant disorder caused by mutations on the **RUNX2 Gene**, a transcription factor involved with osteoblast differentiation. CCD is characterized by a wide range of skeletal abnormalities including widened fontanelles, supernumerary teeth and ribs, sloping shoulders, conductive hearing loss, and hypoplasia/absence of the lateral clavicle. **Early diagnosis is critical** for family planning and implementation of minimally invasive interventions for symptoms such as overcrowding, chronic pain, spinal curvature, and risk for osteosarcomas and hearing loss.

OBJECTIVES

The creation of **a deep learning-based convolution neural network model** and **user-interface called CCD Detection System (CCDDS)** for CCD diagnosis, utilizing radiograph images from publicly available sources (under fair use) of early childhood to early adulthood will provide clinicians with a valuable tool to aid in the **accurate and timely identification** of CCD cases based on, **enabling personalized treatment plans** and **improving patient** care and outcomes. The project aims to create a robust tool despite limited data due to rarity, with room for future growth with data availability and architectural improvements.

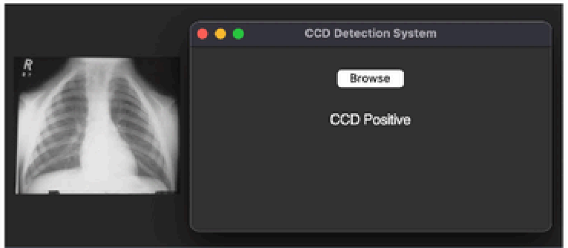


Figure 6. The CCDDS user interface contains a functionality to browse for a thoracic radiograph (as shown in the left of the image), and select for testing. It will then display the result of "CCD Positive" or "CCD Negative"

METHODS

Data Collection: A dataset of de-identified CCD and non-CCD thoracic radiograph images (of ages 3-30) were collected from publicly accessible platforms, ensuring compliance with ethical guidelines and participant privacy. **Data Preprocessing:** the dataset was grayscale, normalized, resized, and augmented. Through preprocessing techniques, the dataset is effectively grown to 8 times larger. **Model Architecture:** A convolutional neural network model was designed and trained using a transfer learning approach with a pre-trained ResNet50V2. **Model Training:** The dataset was split in half for training and validation, each with 50% CCD positive and 50% CCD negative radiographs. The training went through 50 epochs (iterations of training) in which the ResNet50V2 model is iteratively updated to improve predictive performance. **Model Testing:** The model was tested using 7 positive and 7 negative radiographs and evaluated using appropriate metrics.

Class Name	Precision	1-Precision	Recall	1-Recall	f1-score
Class0	0.8571	0.1429	0.7500	0.2500	0.8000
Class1	0.7143	0.2857	0.8333	0.1667	0.7692
Accuracy	0.7857				
Misclassification Rate	0.2143				
Macro-F1	0.7846				
Weighted-F1	0.7868				

Figure 3. Confusion Matrix displays test outcomes and relevant statistics

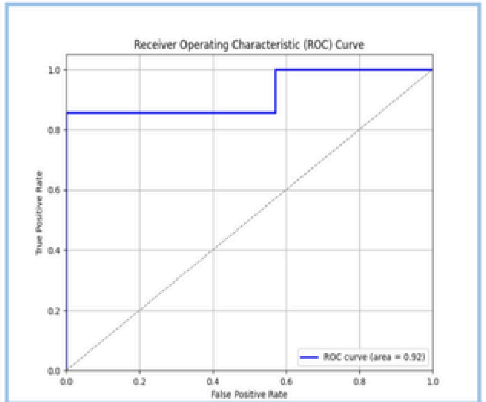
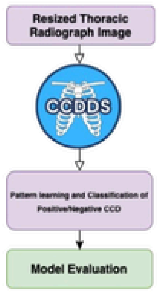


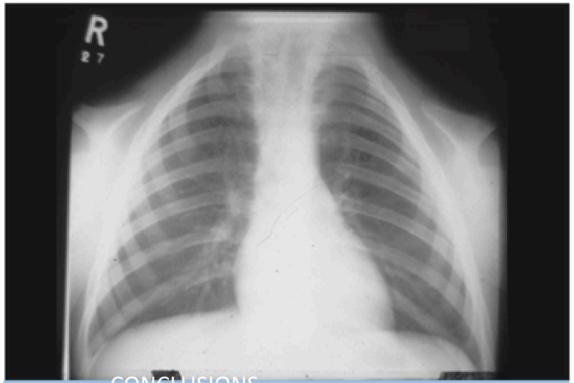
Figure 4: The ROC Curve graph generated based on test results highlights the specificity and sensitivity tradeoff

RESULTS

After 50 epochs of training, and testing of 7 positive and 7 negative radiographs, the CCDDS tool's ResNet50v2 architecture results displayed:

- **High Precision: 0.8571.**
- **High Specificity: 0.8333.**
- **Excellent ROC Curve (Figure 4).**
- **Excellent Area Under Curve Statistic (AUC): 0.92.**
- **Limited sensitivity (0.75).**
- **Limited negative predictive value (0.7143)**





CONCLUSIONS
Figure 1. Thoracic radiograph shows down-sloping shoulders, absent left/right clavicle (red), and wide oblique ribs (blue)

CONCLUSIONS

The **CCDDS diagnostic tool** shows promising preliminary results in accurately identifying CCD-positive cases trained from publicly available radiograph images as a work in progress. **Ongoing work** is focused on developing a custom U-Net architecture to improve accuracy and handling of subtle skeletal abnormalities, as well seeking strategic collaborations and approvals to obtain CCD radiographs and real time imaging datasets from clinicians, as the current dataset is limited due to the rarity. The improvements will increase diagnostic accuracy. Future plans include deployment of the CCDDS tool as a real-time diagnostic tool.

REFERENCES

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