Explainable Detection of Pediatric Congenital Heart Disease Using Prototype Learning on Phonocardiogram Signals

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Aims: This study develops and evaluates an explainable AI framework for detection of pediatric congenital heart disease (CHD) using phonocardiogram (PCG) signals, in settings with limited access to echocardiography. To the best of our knowledge, this is the first study to apply prototype learning to heart sound analysis for CHD detection. The framework provides interpretable predictions and mimics the diagnostic workflow of clinicians by comparing new cases with representative learned patterns.

Methods: PCG recordings from 751 pediatric patients, collected across four auscultation locations in two hospitals in Bangladesh, were preprocessed and standardized into four second fixed-length segments with 50% overlap. A one-dimensional convolutional neural network (CNN) extracted temporal features, followed by a prototype learning module to identify representative patterns of normal and abnormal heart sounds. The model compares input signals with learned prototypes and provides case-based explanations by highlighting clinically relevant temporal regions. Training was conducted at the sample level, whereas evaluation was performed at the patient level, ensuring that recordings from the same patient were not shared between training and testing.

Results: We performed a patient-wise split of 70%, 15%, and 15% for training, validation, and testing, respectively. The framework achieved an accuracy of 91.3%, sensitivity of 98.3%, specificity of 81.4%, precision of 88.1%, and an F1-score of 92.9%. The model demonstrated improved sensitivity compared to existing methods, with a modest trade-off in specificity.

Conclusion: The method provides an accurate and explainable approach for PCG-based CHD detection. By linking predictions to representative heart sound prototypes and highlighting clinically relevant murmur regions, it offers transparent and clinically meaningful decision support. This approach has potential for CHD screening in resource-constrained settings and can help reduce missed CHD cases during early diagnosis.

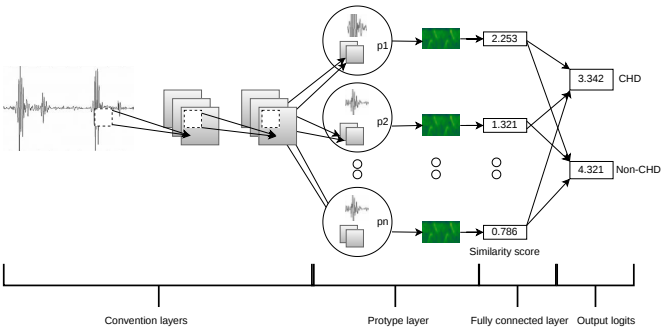

Figure 1. Overview of the proposed prototype learning framework for PCG-based CHD detection.