

compared to standard workflows.

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Identification of Hypertrophic Cardiomyopathy in Children by Machine Learning Applied to Echocardiography

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Background: Hypertrophic cardiomyopathy (HCM) is a significant cause of sudden cardiac death in children, necessitating for precise and early detection. While supervised learning is increasingly utilized in predicting heart conditions, current algorithms relying on billing codes struggle to utilize electronic health record (EHR) data to identify HCM cases. This study aims to develop machine learning (ML) models capable of accurately identifying HCM cases through analysis of measurements from transthoracic echocardiography (TTE) reports. Method: Four supervised-learning ML models (classification and regression trees, support vector machines, random forest, and extreme gradient boosting) were trained and validated using a case-control, retrospective study involving 1,088 pediatric patients undergoing TTE. Among these, 272 HCM patients were matched with 816 control patients without HCM based on sex, age, and year of TTE. The dataset was randomly split, with 80% used for training and validation, and the remaining 20% for testing. Ten-fold cross-validation was employed to assess the performance of each model. For an accurate, echo diagnosis of HCM in the pediatric population, age, sex, and BSA were included in the models. Results: All ML models demonstrated strong performance range between 86-95% in the test set with the random forest model showing the best performance metrics (AUC 0.95, PPV 0.98, Sens 0.95, Spec 0.97) and predicting 10 out of 12 characteristics. Key predictors for HCM included mitral valve medial E/e' ratio, interventricular septum diastolic thickness, left ventricular (LV) mass, ejection fraction, LV outflow tract obstruction systolic peak velocity, LV posterior wall diastolic thickness, mitral valve lateral annulus e' velocity, LV global longitudinal strain, body surface area, left atrial maximum volume, and mitral valve medial annulus e' velocity. Conclusion: This study highlights the effectiveness of ML models utilizing echocardiographic measurements for accurately identifying pediatric HCM cases. These models offer versatility for clinical decision support, EHR-based cohort studies, and quality improvement initiatives. Prioritizing meticulous interpretation and precise diagnosis, the use of ML models reinforces the foundation for safe and proficient clinical.

Table 1. Echocardiographic Measurements Used as Input for the Machine Learning Model

Measurement	Median (IQR)		
	HCM cases (n=272)	Controls (n=816)	P value
Ventricular septal diastolic thickness, mm	17 (13 to 26)	8 (7 to 9)	<.001
Left ventricular posterior wall diastolic thickness,	11 (8 to 14)	8 (7 to 9)	<.001
mm			
Left ventricular mass, g/m2	229 (130.5 to 358.5)	109 (68 to 148)	<.001
Left ventricular ejection fraction, %	71 (66 to 75)	62 (59 to 65)	<.001
Left ventricular global longitudinal systolic strain,	-15 (-18.3 to -11)	-20 (-22 to -19)	<.001
96			
Mitral E velocity, m/sec	0.9 (0.8 to 1.1)	1 (0.8 to 1)	.094
Mitral A velocity, m/sec	0.5 (0.4 to 0.7)	0.5 (0.4 to 0.6)	<.001
Mitral E/A ratio	1.7 (1.3 to 2.2)	2 (1.6 to 2.5)	<.001
Mitral valve deceleration time, ms	177 (152 to 211)	164 (148 to 184)	<.001
Mitral medial annulus e' velocity, m/sec	0.08 (0.06 to 0.1)	0.13 (0.12 to 0.16)	<.001
Mitral medial E/e' ratio	11.7 (10.0 to 16.0)	6.9 (6.0 to 8.3)	<.001
Mitral lateral annulus e' velocity, m/sec	0.1 (0.07 to 0.13)	0.18 (0.15 to 0.2)	<.001
Mitral lateral E/e' ratio	9.1 (6.8 to 12.9)	5.3 (4.5 to 6.5)	<.001
Tricuspid regurgitation systolic peak velocity, m/sec	2.5 (2.3 to 2.8)	2.3 (2.1 to 2.4)	<.001
Left atrial volume index, mL/m2	35 (24.2 to 42.5)	21.5 (19 to 25)	<.001
Resting left ventricular outflow tract peak velocity,	3.9 (2.1 to 4.95)	NA	NA
m/sec			
Left ventricular intracavitary peak velocity, m/sec	2.8 (2.2 to 3.5)	NA	NA

diastolic medial annular velocity; HCM, hypertrophic cardiomyopathy; NA, not available

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Novel Deep Learning Model for the Detection of Cardiac Amyloidosis: A Multicenter, International Study

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Background: Although echocardiography plays a foundational role in the evaluation of individuals suspected to have cardiac amyloidosis (CA), current methods lack accuracy for CA detection, which may delay diagnosis and treatment. We therefore aimed to develop and validate a deep learning (DL) model to detect CA from an apical 4chamber view on echocardiography. **Methods**: Utilizing a global, multicenter CA regis-try, we trained and validated (75%/25% split) a 3D convolutional neural network (Figure 1A) to detect CA using 2757 apical 4-chamber (A4C) images derived from confirmed 279 CA patients-56% with transthyretin (ATTR) and 44% light chain (AL)-and 406 controls (Figure 2B). Given the role of echocardiography as a screening tool, the model was optimized for sensitivity. We subsequently tested the DL model using an independent test dataset with 1120 A4C images of 124 CA patients (50% ATTR and 43% AL) and phenotypic controls with LVH (n=131) in whom CA was suspected and excluded. Results: Of the 255 patients in the test cohort (age 70.5±11.1 years), 63% were male, 30% white, and 45% black. The overall AUC of the DL model in the test cohort was 0.86. The sensitivity, specificity, positive, and negative predictive values in the test cohort were 91%, 53%, 66%, and 85% respectively (Figure 3). Conclusions: Our DL model demonstrated excellent sensitivity and negative predictive value for differentiating CA from phenotypic controls. DL may augment the screening accuracy of echocardiography, preventing diagnostic delays and helping to better identify those at highest risk of CA who require further confirmatory testing.