

# **AI-Based Diagnosis and Risk Prediction of Hypertrophic Cardiomyopathy**

## Abstract

Hypertrophic Cardiomyopathy (HCM) is a genetic cardiac disease characterized by abnormal thickening of the heart muscle, often leading to arrhythmias, heart failure, and sudden cardiac death. Early diagnosis and risk prediction are critical for improving patient outcomes. This research aims to develop an Artificial Intelligence (AI) based diagnostic and risk prediction system using multi-modal cardiac data such as electrocardiograms (ECG), echocardiography, and clinical data. The study leverages deep learning and machine learning techniques to enhance early detection accuracy, predict disease progression, and assist clinicians in decision-making.

## 1. Introduction and Background

Hypertrophic Cardiomyopathy (HCM) is one of the most common inherited cardiovascular disorders, affecting approximately 1 in 500 individuals worldwide. It can remain asymptomatic for years and is a leading cause of sudden cardiac death in young adults and athletes. Traditional diagnostic techniques, including ECG and echocardiography, require expert interpretation and are limited by human error and subjectivity. Artificial Intelligence (AI) has emerged as a transformative tool in healthcare, capable of processing large-scale data and identifying subtle patterns beyond human perception. Applying AI to HCM diagnosis could enable early detection and better risk stratification.

## 2. Literature Review

Recent studies have demonstrated the potential of AI and deep learning in detecting HCM from ECG and imaging data. For instance, Siontis et al. (2024) validated an AI-based ECG model achieving an AUC of 0.92 for detecting HCM in international cohorts. Similarly, Peng et al. (2024) developed a deep learning pipeline using 2D echocardiography for differentiating HCM from other cardiac conditions. These works highlight the diagnostic accuracy of AI models; however, challenges remain in generalizability, interpretability, and dataset diversity. This proposal seeks to address these gaps by developing a multi-modal AI model suitable for diverse populations.

## 3. Problem Statement

Despite progress in AI-driven cardiac diagnostics, early detection and personalized risk prediction of HCM remain limited. Current models often rely on single data modalities, reducing their robustness and applicability across different patient populations. There is a need for a unified AI system that integrates ECG, imaging, and clinical data to accurately diagnose HCM, predict risk levels, and provide interpretable insights for clinicians.

## 4. Objectives

The main objectives of this research are:

1. To develop an AI-based model for early diagnosis of Hypertrophic Cardiomyopathy using ECG, imaging, and clinical data.
2. To predict the risk of adverse cardiac events using machine learning techniques.
3. To enhance interpretability and clinical trust through explainable AI methods.
4. To evaluate the system's performance using real-world hospital data.

## 5. Methodology

The research will follow a data-driven approach involving data collection, preprocessing, model development, and evaluation. The data will include ECG signals, echocardiography images, and patient clinical profiles. Deep learning architectures such as Convolutional Neural Networks (CNNs) and Transformers will be employed for feature extraction. A fusion-based model will integrate multi-modal data for diagnosis and risk prediction. Performance will be assessed using metrics like accuracy, AUC, precision, and recall. Explainable AI tools such as SHAP and Grad-CAM will be applied to visualize decision rationale and support clinician understanding.

## 6. Expected Outcomes

The expected outcomes include an AI-based diagnostic system capable of accurately detecting HCM, a predictive model for assessing patient risk levels, and improved clinical decision support through interpretable AI outputs. The study will contribute to healthcare AI by demonstrating the feasibility of multi-modal integration for cardiac disease diagnosis.

## 7. References

1. Siontis, K.C. et al. (2024). Hypertrophic cardiomyopathy detection with artificial intelligence electrocardiography. \*European Heart Journal – Digital Health\*.
2. Peng, B. et al. (2024). Deep Learning Pipeline for Differentiating HCM using 2D Echocardiography. \*arXiv\*.
3. Tison, G.H. et al. (2022). Using AI Electrocardiogram Analysis for Improved Diagnosis of HCM. \*UCSF News\*.
4. Vaid, A. et al. (2022). HeartBEiT: Vision Transformer for ECG Data. \*arXiv\*.