nature medicine

Article <https://doi.org/10.1038/s41591-024-02971-2>

Screening and diagnosis of cardiovascular disease using artificial intelligence-enabled cardiac magnetic resonance imaging

In the format provided by the authors and unedited

Supplementary Information

Table of Contents

1. Supplementary Table 1. PPV and NPV of the diagnostic model derived from cine and LGE as combined inputs in the primary dataset (n=6650).

Supplementary Table 1 | PPV and NPV of the diagnostic model derived from cine and LGE as combined inputs

*95% confidence interval in the brackets. PPV: positive predictive value; NPV: negative predictive value.

2. Supplementary Table 2. Performance of the screening model in the consecutive testing set (n=961).

AUROC=area under the receiver operating characteristic curve; PPV=positive predictive value (precision); CI=confidence intervals; SAX=short axis; 4CH=four chamber.

3. Supplementary Table 3. The 48 patients from the consecutive testing set, excluded from the reported diagnostic model performance metrics.

Note: it's noteworthy that the AI screening model demonstrated robust performance by correctly classifying all 48 patients into the abnormal class, with a high average confidence score of 0.918. This successful classification, along with the high confidence score, highlights the screening model's robustness in handling a diverse range of cardiovascular diseases, including suspected phenocopies, such as genetic metabolic cardiomyopathy, which extend beyond the commonly recognized 11 CVD classes.

In contrast, the diagnostic model classified these cases with an average extremely low confidence score of 0.585, emphasizing the model's cautious approach when dealing with instances that deviate from the specified 11 CVD classes. Future direction includes the introduction of an additional AI deferral system that could defer cases with low confidence scores, falling below a predefined threshold, for expert human assessment. This collaborative synergy between human clinicians and AI models holds promise for further improving diagnostic accuracy, especially in scenarios beyond the commonly specified 11 CVD classes.

Supplementary Table 3 | The 48 patients from the consecutive testing set, excluded from the reported diagnostic model performance metrics.

tricles, accompanied by a slight thickening of the mid -segment of the ricular septum (maximum approximately 13mm). ricular enlargement with d systolic function is noted, and ation with sinus bradycardia is nsidered. atrial and ventricular ent observed, with no evidence is; consideration of a correlation vthmia. hent of the left atrium is , along with a subtle presence of hals in the left ventricular ium, suggestive of a mild ical condition. ystolic function. a mild thickening of the ricular septum, measuring s includes both CAD and DCM. eals patchy myocardial nent and fibrosis in ardial areas of the basal and rior segments of the left Additionally, an enlarged left ventricular end -diastolic cavity g 60mm is observed. Diagnosis from human experts ricular septal thickening, left ar enlargement with reduced iunction, and multifocal fibrosis bitum and left ventricular lateral noted. Genetic metabolic copathy is under consideration, ting further investigation. left ventricular enlargement nied by reduced systolic interventricular septal ig, and widespread myocardial nent in the left ventricle. metabolic cardiomyopathy is nsideration, necessitating westigation. left ventricular enlargement olic function at the lower limit l, accompanied by a minor fibrosis. Unclear diagnosis nan experts ricular enlargement is nied by a decrease in systolic

4

areas of myocardial fibrosis. Unclear diagnosis from human experts There is left ventricular enlargement accompanied by a decrease in systolic function, along with extensive subendocardial enhancement. Unclear diagnosis from human experts Noticeable thickening of both left and right ventricular walls, coupled with widespread abnormal enhancement of the left ventricular myocardium; genetic metabolic cardiomyopathy remains within diagnostic consideration. Left ventricular enlargement is observed with diminished systolic function and widespread fibrosis within the left ventricular wall. Unclear diagnosis from human experts. Observation of left ventricular enlargement is noted, with preserved systolic function; however, a minor degree of fibrosis in the lateral wall is observed, which does not align with the diagnostic criteria for dilated cardiomyopathy (DCM). Mild left atrial enlargement is noted, accompanied by a mildly thickened left ventricular wall with multifocal fibrotic changes. This presentation is indicative of a cardiomyopathy related to a DES gene mutation. Presence of an occupying lesion in the right ventricular cavity, indicative of a tumor -like pathology, representing a rare condition. Takotsubo syndrome / Stress cardiomyopathy. Constrictive pericarditis, with enlargement of both atria. The mid -segment of the interventricular septum exhibits slight thickening, while maintaining normal left ventricular systolic function. Additionally, there is a suspicion of minor subendocardial fibrosis in the left ventricular inferior wall. Mild left ventricular enlargement is observed alongside normal but reduced

systolic function, indicating asynchronous left ventricular contraction. The possibility of an association with left bundle branch block (LBBB) is under consideration. Aortic valve stenosis accompanied by regurgitation, leading to secondary left ventricular enlargement and interventricular septal thickening. Bicuspid aortic valve malformation leading to secondary left ventricular enlargement, interventricular septal

4. Supplementary Table 4. Performance of the diagnostic model in the consecutive testing set (n=532).

AUROC=area under the receiver operating characteristic curve; CI=confidence intervals. The calculation of the 95% CI was not performed for sample sizes below 50 due to potential limitations in the precision of estimates associated with small sample sizes.

5. Supplementary Table 5. Distribution of demographics and LVEF in the primary dataset.

Supplementary Table 5 | Distribution of demographics and LVEF across 11 CVD classes and the normal control class in the **primary dataset.**

*Q1: the first quartile; Q3: the third quartile; STD: standard deviation; LVEF: left ventricular ejection fraction.

6. Supplementary Table 6. Distribution of demographics and cardiac function in the consecutive testing set.

7. Supplementary Table 7. The typical CMR scan protocol and scanner parameters for the primary and external sets.

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FW: Beijing Fuwai Hospital, Beijing; AZ: Beijing Anzhen Hospital, Beijing; GD: Guangdong Provincial People's Hospital, Guangzhou; HEB: The 2nd Affiliated Hospital of Harbin Medical University, Harbin; LZ: The First Hospital of Lanzhou University, Lanzhou; RJ: Renji Hospital, Shanghai; TJ: Tongji hospital, Wuhan; XH: Peking Union Medical College Hospital, Beijing.

8. Supplementary Figure 1. The distribution of LVEF across the 11 CVD classes and the normal Supplementary Figure 11 COVD control class in the primary dataset

9. Supplementary Figure 2. The clinical prevalence of CVD classes

The effect of modifying the initialized learning-rate (testing in one-fold of the primary cohort with the diagnostic model

