Hereditary transthyretin cardiac amyloidosis with phenotypic features of non-compaction cardiomyopathy presenting as ventricular tachycardia – a case report

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Introduction: Hereditary transthyretin cardiac amyloidosis (ATTR-CA) is an infiltrative cardiomyopathy caused by mutation of the transthyretin (TTR) gene[1]. We present our center's first experience in diagnosing this rare disease, which is associated with a high mortality.

Case report: 59-year-old male was admitted to the Coronary Care Unit due to hemodynamically unstable monomorphic ventricular tachycardia with left bundle branch block morphology **(Figure 1).** Urgent electrocardioversion was performed with successful hemodynamic stabilization. He denied dyspnea, chest pain and previous heart disease. His sister died suddenly at age 53. Serial electrocardiograms and laboratory parameters did not show any definite signs of acute coronary syndrome. Urgent coronary angiography was performed to rule out underlying ischemic injury, which revealed subtotal stenosis of the mid left anterior descending artery (LAD), with normal findings of the remaining epicardial arteries. Successful percutaneous coronary intervention of LAD was performed. An echocardiogram revealed left ventricular hypertrophy with diffuse myocardial fibrosis and apico-posterior-lateral hypertrabeculation with decreased systolic and restrictive diastolic function **(Figure 2).** Due to the discrepancies between the ultrasound, electrocardiographic and angiography findings, further

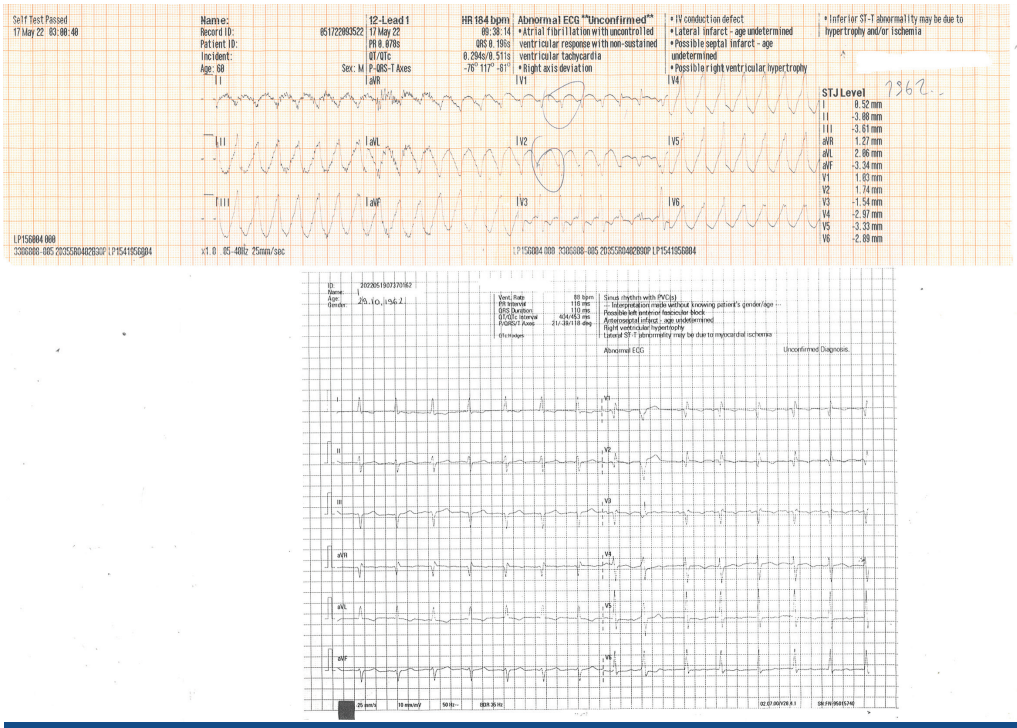

FIGURE 1. Ventricular tachycardia with left bundle branch block morphology.

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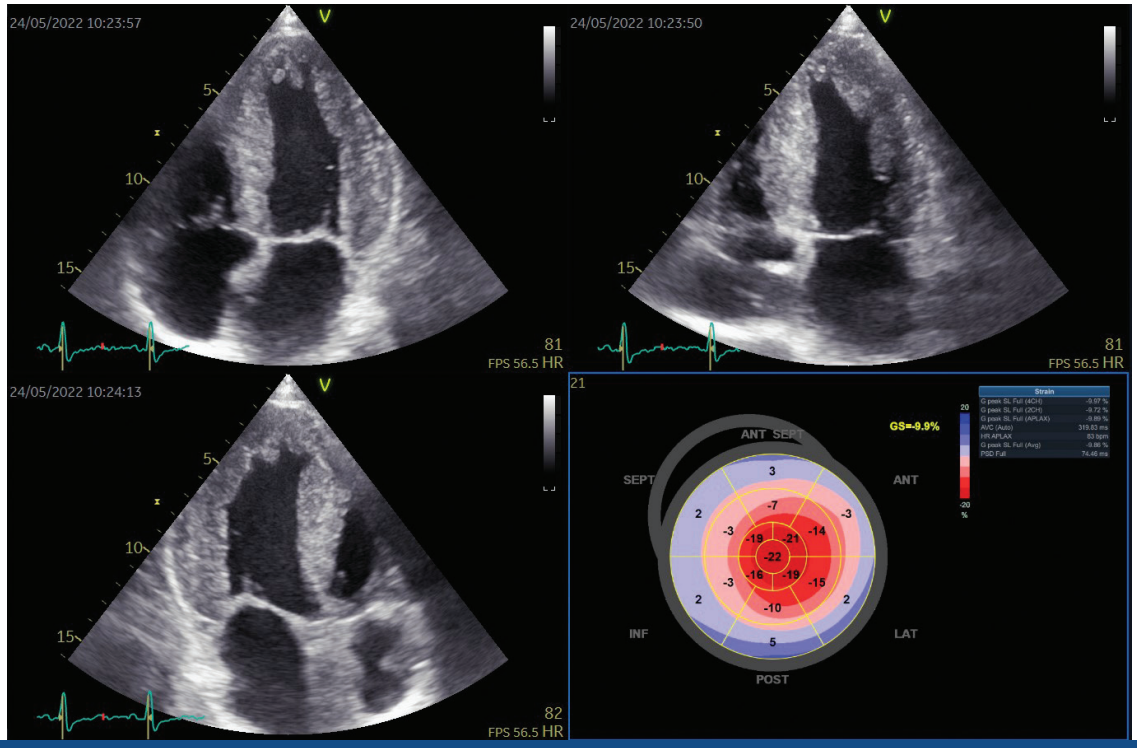

FIGURE 2. Left ventricular hypertrabeculation with apical sparing pattern.

work-up of hypertrophic cardiomyopathy was performed. Cardiac magnetic resonance imaging showed morphologic features of "non-compaction" cardiomyopathy (NCC). Also, nuclear imaging using technetium pyrophosphate (Tc-99 PYP) was performed, which showed diffusely increased uptake by the myocardium, highly suggestive of TTR amyloidosis. A cardioverter-defibrillator was implanted, and genetic testing for hereditary TTR amyloidosis was carried out. The patient was then discharged. Genetic testing confirmed our suspicion, with a mutation of pathogenic clinical significance. Genetic counseling was provided to the patient and his family members, and the patient was informed about available specific treatment options. The patient was then referred to a heart failure center of excellence for further treatment.

Conclusion: The aim of this case presentation was to increase clinical awareness of ATTR-CA as a cause of hypertrophic cardiomyopathy. Furthermore, only a few cases of ATTR-CA with morphologic characteristics of NCC have been reported in the literature.

LITERATURE

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