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Beyond hypertrophy

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Beyond hypertrophy: unmasking sarcomeric hypertrophic cardiomyopathy in a patient with wild-type ATTR amyloidosis

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A 72-year-old male presented with new-onset heart failure. Initial evaluation showed low-voltage complexes on the electrocardiogram and left ventricular hypertrophy on transthoracic echocardiography (A). Given the suspicion of amyloidosis bone scintigraphy was performed, revealing myocardial tracer uptake (Perugini grade 3)(B). Further testing excluded the presence of monoclonal protein conforming the diagnosis of transthyretin amyloidosis (ATTR).

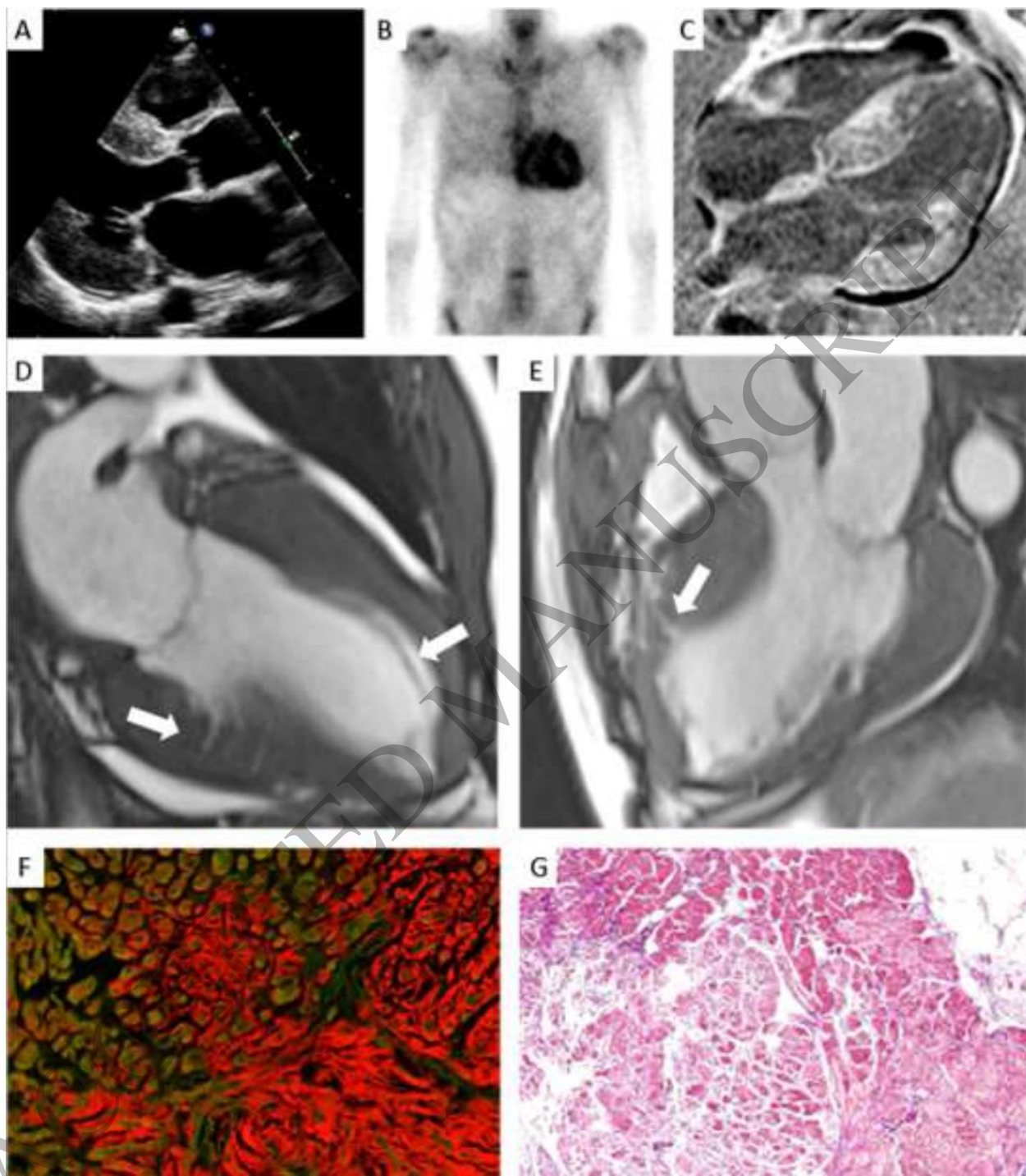
1 Further phenotyping using cardiovascular magnetic resonance (CMR) imaging showed diffuse
2 late gadolinium enhancement of the left and right ventricle and interatrial septum consistent with
3 amyloidosis (C). However, there were also hallmark features of sarcomeric hypertrophic
4 cardiomyopathy (HCM), including reverse septal curvature, myocardial crypts, muscular band and an
5 irregular wall pattern (D, E). Subsequently, DNA analysis was performed including the TTR gene but also
6 the genes involving HCM, and identified a pathogenic variant in the *MYBPC3* gene. No pathogenic
7 variants were found in the TTR gene.

8 The myocardial biopsy revealed amyloid deposits (F), alongside myocardial hypertrophy and
9 fibrosis (G), consistent with a *MYBPC3* variant.

10 This case highlights a rare combination of wild-type ATTR amyloidosis and sarcomeric HCM due
11 to a pathogenic variant in the *MYBPC3* gene. It underscores the importance of comprehensive
12 phenotyping of cardiomyopathies. CMR was essential in identifying features of both amyloidosis and
13 sarcomeric HCM. The discovery of a pathogenic *MYBPC3* variant has significant familial implications,
14 emphasizing the need for genetic counseling and cascade screening. CMR is indispensable in the work-
15 up of left ventricular hypertrophy, not only in achieving diagnostic clarity but also in informing family-
16 based risk evaluation and long-term management strategies.

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18 *Data availability statement: No new data were generated or analysed in support of this*
19 *research.*

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31x35 mm (DPI)