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Cardiac fibroblast activation in Fabry disease on ^{18}F-fibroblast activation protein inhibitor positron emission tomography/computed tomography imaging

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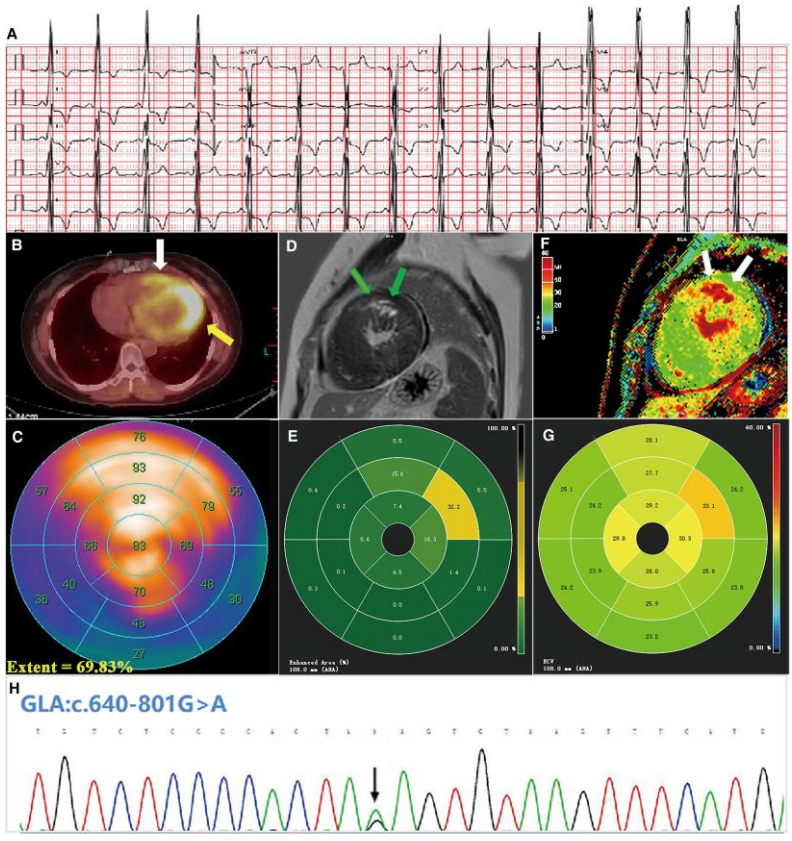

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A 61-year-old woman presented with progressive dyspnoea for 10 years. Electrocardiogram demonstrated a short PR interval (104 ms) and typical left ventricular hypertrophy (LVH; *Panel A*). Left ventricular (LV) outflow tract obstruction (40 mmHg at rest and 101 mmHg during provocative manoeuvres), systolic anterior motion of the mitral valve, and severe LVH (maximum ventricular septum thickness of 27 mm) were shown on transthoracic echocardiography with normal renal function, suggesting hypertrophic cardiomyopathy (HCM).

^{18}F-fibroblast activation protein inhibitor (FAPI) Positron-emission tomography/computed tomography imaging as part of a prospective study for assessing myocardial fibrosis in HCM was performed. Intense and heterogeneous ^{18}F-FAPI uptake was observed in both right (*Panel B*, white arrow) and left ventricles (*Panel B*, yellow arrow). Notably, FAPI uptake involved 69.83% of the LV myocardium (*Panel C*). A small amount of late gadolinium enhancement (LGE) in strip or halo pattern at the insertion part of ventricular septum, mid anterior (*Panel D*, green arrow), and mid anterolateral was identified by cardiac magnetic resonance imaging. Bullseye plot showed the distribution of LGE, which accounted for 7.10% of the LV myocardium (*Panel E*). The global native-T1 times, enhanced-T1 times, and extracellular volume (ECV) were 1254.32 ms, 593.39 ms, and 0.27, respectively. The ECV of the mid anterior was increased (*Panel F*, white arrow). Bullseye plot showed the ECV of each segment using American Heart Association (AHA)-17 segment model (*Panel G*). The patient subsequently underwent genetic testing, which confirmed the final diagnosis of Fabry disease (FD; GLA gene pathogenic mutation: c.640–801G > A het; *Panel H*).

Beyond the LV hypertrophy mimicking HCM, FD in this case demonstrated that cardiac fibroblast activation, which can be tested by FAPI imaging, may also play a crucial role in the progression of FD in addition to glycosphingolipid accumulation and immune activation.[1,2] In this case, we firstly described cardiac FAPI imaging characteristics of heart involvement in a patient with FD, which will help to understand pathophysiologic mechanisms of cardiac FD.

Consent: The authors confirm that written consent for submission and publication of this case report including images and associated text has been obtained from the patient in line with COPE guidance.

Conflict of interest: The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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