A 17-year old female presented with atypical chest pain. Clinical examination and ECG was unremarkable but an echocardiogram demonstrated ‘unusual ventricular foreshortening’.

Subsequent cardiac magnetic resonance imaging demonstrated all four characteristics of a rare primary cardiomyopathy. A truncated left ventricle (Panels A and B), spherical remodelling, and bulging of the inter-ventricular septum towards the right ventricle (Panels B, D, and E). RV apex formation with wrapping around of the RV apex around the deficient left ventricular apex (Panels A and B) and fatty replacement of the LV apex (Panels D, E and H). In this case, apical fat pad and additional apical fatty infiltration of the LV apex were clearly delineated by T1 mapping (Panel F) which to our knowledge has not been applied in this pathology before. Late gadolinium enhancement revealed mid-wall hyper-enhancement in the mid-ventricular septum (Panels D and E). Taken together these findings were diagnostic of left ventricular apical hypoplasia (LVAH) (Supplementary material online, Video 1).

Left ventricular apical hypoplasia was first identified in 2004 and is postulated to be a congenital abnormality due to inadequate dilatation of chambers during partitioning. This is the third case to identify mid-wall fibrosis in a case of LVAH. This finding has not consistently been described, as a characteristic of LVAH suggesting the possible role of CMR in risk stratifying patients with this poorly understood condition. Furthermore, this may provide evidence for an overlap with other inherited idiopathic cardiomyopathies of the DCM spectrum.

Patients usually present with mild, non-specific symptoms although arrhythmic presentations have been reported. Most respond well to standard cardiac failure therapy.

Supplementary material is available at European Heart Journal online.