Hypoplasia of the posterior mitral valve leaflet detected in late adulthood

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A 76-year-old male with a pacemaker presented with acute pulmonary oedema requiring invasive mechanical ventilation. Transthoracic echocardiography (TTE) revealed a partially flail anterior mitral leaflet, severe mitral regurgitation (MR), and pulmonary hypertension, not present on previous TTEs. Transoesophageal echocardiography (TEE) demonstrated an elongated anterior mitral valve leaflet (AMVL) with a flail A2 segment due to chordal rupture (Panels 1-A, 1-B, and Supplementary material online, Video S1), a hypoplastic posterior mitral valve leaflet (PMVL, black arrow head in Panels 1-A and 2) with intact chord, and severe, posteriorly directed MR (Panel 1-B) (all mid-oesophageal views, 0°). Three-dimensional TEE short-axis view of the MV, as seen from the left atrium (Panel 3 and Supplementary material online, Video S2), demonstrated the hypoplastic PMVL (black arrow head) and myxomatous AMVL (A1, A2, A3 scallops) with ruptured chordae (white arrows). The patient underwent minimally invasive mitral valve repair, including ring annuloplasty and chordal transfer from the posterior annulus to A2.

The absence of the PMVL is usually fatal in utero, while deficient PMVLs typically present in childhood with symptomatic MR. A few cases have been described in asymptomatic teens and adults. Our case represents the oldest documented patient with a deficient PMVL. It illustrates that a hypoplastic PMVL may be asymptomatic through to late adulthood, with valve competence maintained by an elongated AMVL. In this case, severe MR was caused by chordal rupture of the myxomatous AMVL rather than the deficient PMVL. Hypoplastic PMVLs may be more prevalent in asymptomatic adults than currently recognized.

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