Functionally uni-leaflet mitral valves in a family: a case series

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Functionally uni-leaflet mitral valves are extremely rare. In severe cases, they are considered incompatible with life beyond the neonatal period. We present a case series of three adults with almost complete absence of the posterior mitral valve leaflet and who are asymptomatic, with no more than mild mitral regurgitation. We believe that this is the first reported instance of such findings in a single family.

Keywords
- Echocardiography
- Mitral valve
- Uni-leaflet valve
- Congenital heart disease
- Family

Case series

Patient A
An 18-year-old asymptomatic female was noted to have a pan-systolic murmur during a routine examination and was referred for a screening transthoracic echocardiogram. This demonstrated an elongated and thickened anterior mitral valve leaflet with a mild degree of prolapse (see Supplementary data online, Video S1). The posterior leaflet was extremely small and redundant with just a short stump visible. The resultant coaptation line was eccentrically displaced with accompanying mild mitral regurgitation, directed posteriorly (see Supplementary data online, Figure S1 and Video S2).

The subvalvular apparatus was anatomically normal with appropriate chordal attachments and papillary muscles (see Supplementary data online, Video S3). There was no evidence of chamber enlargement and no other valvular dysfunction was noted. Subsequent transoesophageal echocardiography confirmed the findings of an elongated anterior mitral valve leaflet and an extremely small (almost absent) posterior leaflet which contributes little to valvular function (Figure 1).

Patient B
Patient B (aged 17, the sister of patient A and her only sibling) was also asymptomatic with similar auscultatory findings and was referred for a transthoracic echocardiogram. This is again demonstrated a thickened, elongated anterior mitral valve leaflet with an under-developed posterior leaflet (see Supplementary data online, Video S4). Once more, posteriorly directed mild mitral regurgitation was noted.

Patient C
Patient C, the 46-year-old mother of patients A and B, underwent screening transthoracic echocardiography in view of her daughters’ echocardiographic findings. Patient C was also asymptomatic. Her echocardiogram had similar appearances to those of her daughters (see Supplementary data online, Video S5).

Figure 1 Transoesophageal echocardiography of the left atrium (LA) and left ventricle (LV) showing the elongated anterior mitral valve leaflet (solid arrow) and extremely small posterior leaflet (dotted arrow) of patient A.
Discussion

All three cases described in this report are asymptomatic with functionally uni-leaflet mitral valves. The posterior mitral valve leaflet in each case is almost absent, with no worse than mild mitral regurgitation present.

Congenital anomalies of the mitral valve apparatus are rare. Of such cases, congenital mitral stenosis, atresia, accessory valvular tissue, and cleft mitral valve are more common. Descriptions of functionally uni-leaflet mitral valves (either partial or complete leaflet agenesis/hypoplasia) are extremely rare and largely limited to a few case reports. To the best of our knowledge, this case series is the first reported instance in a family.

In the most severe form (complete leaflet absence), cases are usually considered to be incompatible with life beyond the neonatal period. Asymptomatic patients, however, do exist and a prevalence of 1:8800 has previously been documented in pre-selected patient cohorts. The prognosis in these asymptomatic patients is uncertain. The potential for worsening mitral regurgitation (and hence potential morbidity and mortality), primarily as a consequence of annular dilation, has also been postulated. Therefore, long-term monitoring with serial echocardiography seems appropriate.

The family members have been placed under annual review with transthoracic echocardiography to monitor progression of the mitral regurgitation. The genetic basis for this abnormality is unknown and, at the time of writing, a formal genetics review has not been undertaken.

Supplementary data

Supplementary data are available at European Journal of Echocardiography online.

Conflict of interest: none declared.

References