Severe mitral regurgitation due to an extraordinary heart defect

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A previously non-described cause of mitral regurgitation is presented. An asymptomatic 50-year old male who was casually diagnosed of mitral valve Barlow’s disease underwent cardiac surgery due to severe mitral regurgitation. In the operating theatre, a longitudinal fissure of 1.5–2.0 cm length, along the posterior mitral leaflet, was found responsible for the insufficiency. This defect had features of a potential congenital origin and it was successfully repaired with direct suture. Whether it is an atypical mitral cleft, a variation of Barlow’s morphology spectrum or a new congenital heart defect remains unclear.

Keywords: Mitral cleft • Severe mitral insufficiency

CASE PRESENTATION

A 50-year old man, without previous medical history, underwent cardiac surgery due to mitral valve Barlow’s disease (casually diagnosed) and severe mitral regurgitation (Fig. 1, Panel 1A). He had mitral valve diffuse excess tissue with mixomatous changes and prolapse of both anterior and posterior leaflets (Fig. 1, Panel 1B). In the operating room, a longitudinal fissure of 1.5–2.0 cm length was found responsible for the regurgitation. It was rift-like shaped, along the posterior leaflet and parallel to mitral coaptation surface (Fig. 1, Panel 2A, Video 1). Several fibrotic tendinous chords were attached to the edges of the fissure, perfectly epithe- lized without signs of infective endocarditis, and connected to papillary muscles, mimicking primary tendinous cords. Subvalvular apparatus was intact. Although anatomicopathological sample was not obtained, all these data were consistent with a congenital origin.

Mitral valve was successfully repaired with direct fissure suture with prolene (Fig. 1, Panel 2B) and plication to shorten the prolapse. An annuloplasty ring was also implanted with the plication of an elongated chord attached to scallop A2. No mitral regurgitation remained after the procedure. Looking back at 3D-transoesophageal echocardiogram images, the defect was better appreciated (Fig. 1, Panel 3A, Video 2) in mitral defect scheme (Fig. 1, Panel 3B).

DISCUSSION

Mitral valve formation begins during the fourth week of gestation [1]. The first evidence of valvulogenesis is the formation of endocardial cushions in the atrioventricular canal (AVC) and outflow tract of the primitive looped heart tube. During the sixth week, fusion of the endocardial cushions divides the AVC into right and left atrioventricular junctions. Failure of fusion of the superior and inferior cushions is responsible for producing atrioventricular septal defects (AVSDs) [1].

Normally, the anterior (aortic) mitral leaflet is derived from the apposition of the left part of the superior and inferior cushions [1], whereas the posterior (mural) mitral leaflet is derived from mesenchymal cushions that arise laterally in the AVC after cushion fusion [2]. The first visible event in the formation of mural leaflets is the appearance of a small myocardial protrusion from the myocardium of the AVC. The protrusion increases in size and forms a circumferential myocardial funnel-like structure. The underlying ventricular myocardium expands outwards and forms holes and trabeculae, thus excavating the region behind the protrusion and creating a movable leaflet. After embryonic age 14.5, the myocardium of the leaflet will disappear by programmed cell death [3].

A complex network of transcription factors is necessary to promote proper levels of cell proliferation. The process is driven by regulatory proteins that are expressed by genes in local myocytes in a time-dependent manner. Specific mutations altering genetic milieu result in congenital malformations of the atrioventricular valves. Those alterations affecting the development of the lateral protrusion formation might be responsible for the presence of such a defect.

Previous studies have described the existence of mitral valve ‘cleft’ as a congenital malformation. It is a division of one of the leaflets (usually the anterior leaflet) of the mitral valve [1]. The origin is under debate, some authors have considered isolated
cleft to be a 'forme fruste' of AVSD, whereas others have supposed it to be a distinct morphological entity [1].

In AVSD, the positions of both papillary muscles were rotated counterclockwise, whereas in isolated cleft, the position of the papillary muscles was similar to that in normal children. This makes it a good marker of this lesion. Moreover, in AVSD, the cleft points towards the ventricular inlet septum, whereas in isolated cleft, it is usually more directed towards the aortic root.

Isolated cleft is an uncommon cause of mitral insufficiency described for the first time in 1954 with paediatric incidence of 1:1340. Clefts in both leaflets in the same patient have not been described previously [4].
Sometimes, cleft diagnosis is difficult and real-time 3D echocardiography is the gold standard technique. The degree of the mitral cleft was defined as complete or incomplete, according to its extension to the mitral annulus. The degree of mitral regurgitation is not directly proportionate to the size of the cleft. The cleft produces more severe degrees of mitral insufficiency in older patients, suggesting some worsening with age. In younger patients, it may remain undiagnosed for many years.

Mitral valve repair is preferred to mitral valve replacement and usually consists of a direct suture of the cleft and eventually annuloplasty [1]. The prognosis of repaired mitral cleft is usually excellent.

According to the previously reported description of the isolated mitral cleft, it has usually a radial direction from the centre to outwards, indeed, it is defined as a narrow gap that extends at least one half of the way from the free edge towards the hinge line (annulus) [5]. Nevertheless, in this patient, the fissure was parallel to the coaptation mitral valve surface.

To the best of our knowledge, we are presenting a non-described before mitral defect that produces severe mitral regurgitation with potential congenital origin, associated with mitral valve Barlow’s disease. It shares some characteristics with the very well-known mitral clefts, but it is morphologically different in principle. Whether this defect is an atypical mitral cleft, a variation of Barlow’s morphology spectrum or a new congenital heart defect remains unclear.

Conflict of interest: none declared.

REFERENCES