［文章编号］ 1671-587X(2024)05-1426-06 DOI：10. 13481/j. 1671-587X. 20240529

TTE联合TEE诊断主动脉瓣二瓣畸形致二尖瓣前叶瘤1例报告及文献复习

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［摘 要］ **目的：**分析二尖瓣瘤（MVA）的临床表现、影像学特征、治疗措施和疗效，提高临床医师对MVA的认识。**方法：**收集1例主动脉瓣二瓣畸形导致二尖瓣前叶瘤形成患者的临床资料，根据其临床特点和影像学特征明确临床诊断、选择治疗方法并对疗效进行分析，同时进行相关文献复习。**结果：**患者，女性，68岁，因心慌气短13年，加重1个月入院。患者于13年前无诱因出现心慌气短，在当地医院诊断为“心脏瓣膜病”，1个月前上述症状加重而住院治疗。经胸二维超声心动图（TTE）显示左心室肥大，主动脉瓣呈二瓣，瓣叶增厚，回声增强，主动脉瓣前向血流速度增快，瓣口面积2.0 cm^2；二尖瓣前叶瓣缘局部略增厚，回声略增强，呈囊性“蜂窝状”结构，与主动脉瓣反流束密切相关，反流束似进入“囊袋”。术前经食管超声心动图（TEE）显示主动脉瓣二瓣畸形，前方瓣叶舒张期脱向左心室流出道；二尖瓣前叶心房面可探及“囊袋状”结构，随心动周期囊壁形态发生改变，该“囊袋”与左心室血流相交通。超声诊断为主动脉瓣二瓣畸形-横裂式、重度关闭不全伴轻度狭窄和二尖瓣前叶瘤。术中主动脉瓣瓣环扩大，瓣叶呈明显关闭不全，将瓣叶切除，主动瓣位置换1枚23号生物瓣；经主动脉瓣口探查二尖瓣，前叶瓣体可探及 “囊袋样”结构，未进行特殊处理。术后TEE显示主动瓣位生物瓣回声及活动良好，二尖瓣“囊袋状”结构仍然不变。术后10 d和术后4个月随访显示主动瓣位生物瓣回声及活动良好，二尖瓣囊性病变的性质及大小与术前比较无明显变化。**结论：**MVA临床罕见，TTE是目前临床诊断MVA最有价值的影像学诊断方式，特别是TTE联合TEE是最佳诊断方法，且可协助治疗及疗效评价。

［关键词］ 经胸二维超声心动图；经食管超声心动图；主动脉瓣二瓣畸形；二尖瓣瘤；疗效评价
［中图分类号］ R543.1 ［文献标志码］ B

Diagnosis of bicuspid aortic valve malformation resulting in anterior mitral aneurysm by TTE combined with TEE: A case report and literature review

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ABSTRACT Objective：To analyze the clinical manifestations, imaging characteristics, treatment

［收稿日期］ 2023-12-20
［基金项目］ 吉林省科技厅自然科学基金项目（20200201562JC，20200201610JC）
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measures, and efficacy of mitral valve aneurysm (MVA), and to enhance the clinicians' understandings of MVA. **Methods**: The clinical data of one patient with aortic valve bicuspid malformation leading to mitral valve anterior leaflet aneurysm were collected. The clinical diagnosis was confirmed based on the clinical characteristics and imaging features, the treatment methods were selected, and the efficacy was analyzed. The relevant literatures were reviewed. **Results**: The patient, a 68-year-old female, was admitted due to palpitations and shortness of breath for 13 years, and the symptoms worsened one month ago. Thirteen years ago, the patient experienced palpitations and shortness of breath without any inducement and was diagnosed with "heart valve disease" in the local hospital. The symptoms worsened one month ago, leading to hospitalization. The transthoracic two-dimensional echocardiography (TTE) results showed the left ventricular hypertrophy, bicuspid aortic valve with thickened and echogenic leaflets, the forward flow velocity was increased, and the aortic valve orifice area was 2.0 cm^2; the mitral valve anterior leaflet margin was slightly thickened and echogenic, presenting a cystic "honeycomb-like" structure closely related to the aortic valve regurgitation jet, which appeared to enter the "sac". The preoperative transesophageal echocardiography (TEE) results showed the bicuspid aortic valve malformation with the anterior leaflet prolapsing into the left ventricular outflow tract during diastole; a "sac-like" structure was detected on the atrial surface of the mitral valve anterior leaflet, changing shape with the cardiac cycle and communicating with left ventricular blood flow. The ultrasound diagnosis was bicuspid aortic valve malformation with transverse fissure, severe regurgitation with mild stenosis, and mitral valve anterior leaflet aneurysm. Intraoperatively, the aortic valve annulus was enlarged with the significant leaflet regurgitation. The leaflets were excised, and a 23 mm bioprosthetic valve was implanted in the aortic position. Upon exploration through the aortic valve orifice, a "sac-like" structure was found on the mitral valve anterior leaflet, which was not specially treated. The postoperative TEE results showed good echo and activity of the aortic bioprosthetic valve, and the "sac-like" structure on the mitral valve remained unchanged. The follow-up results at 10 d and 4 months after operation showed good echo and activity of the aortic bioprosthetic valve, and compared with before operation, there was no significant change in the nature and size of the mitral valve cystic lesion. **Conclusion**: MVA is clinically rare. TTE is currently the most valuable imaging diagnostic method for MVA, especially when combined with TEE, which is the best diagnostic method and can assist in the treatment and efficacy evaluation.

KEYWORDS Transthoracic echocardiography; Transesophageal echocardiography; Bicuspid aortic valve; Mitral valve aneurysm; Efficacy evaluation

二尖瓣瘤（mitral valve aneurysm，MVA）是一种罕见的疾病，在接受经食管超声心动图（transesophageal echocardiography，TEE）检查的患者中，其发病率仅为0.20%～0.29%。MVA的病因和形成机制不明，主要病因为感染性心内膜炎的并发症，其他病因有类风湿性心脏病、马凡综合征、主动脉返流及肥厚性心脏病等，常与主动脉瓣的感染性心内膜炎合并出现，可伴有二尖瓣关闭不全，因主动脉瓣偏心反流束冲击二尖瓣前叶而形成者较少。MVA患者的临床表现与病变严重程度有关，部分患者无明显体征或症状，其严重的并发症为MVA破裂和全身栓塞，MVA破裂后导致左心的前负荷明显增加，甚至急性左心功能不全。其最适宜的诊断方法为经胸二维超声心动图（transthoracic echocardiography，TTE）联合经食管三维超声心动图（three-dimension transesophageal echocardiography，3D-TEE），且可指导诊疗方案的制订。现收集本院收治的1例MVA患者的临床资料，并进行相关文献复习，探讨其临床表现、影像学特征、诊疗方案和预后随访效果，提高临床医师对该病的认识。

1　临床资料

1.1　一般资料　患者，女性，68岁，因心慌和气短13年，加重伴腹胀1个月入院。患者缘于13年前无诱因出现心慌伴胸闷和气短，经休息后可缓解，此后上述症状反复发作。于当地医院诊断为“心脏瓣膜病”，并进行药物治疗，症状可缓解。1个月

前上述症状加重且伴有腹胀，药物治疗效果欠佳。于2021年6月29日住院治疗。5年前于当地医院诊断为布鲁氏菌病，经治疗后好转。专科检查：心音有力，主动脉瓣听诊区可闻及舒张期叹气样音。TTE结果显示：左心室（left ventricle，LV）肥大，LV厚度为55.8 mm，室间隔（interventricular septum，IVS）厚度16.4 mm，左心室后壁（left ventricular posterior wall，LVPW）厚度15.0 mm，射血分数（ejection fraction，EF）65%。主动脉瓣呈二瓣，瓣叶增厚、回声增强，反流颈9 mm，反流面积12.2 cm^2，反流束沿二尖瓣前叶偏心冲击二尖瓣前叶心室面；主动脉瓣前向血流速度（3.3 $m\cdot s^{-1}$）增快，峰值梯度（peak gradient，PG）45 mm，平均压差（mean pressure gradient，MPG）22 mmHg，瓣口面积2.0 cm^2。二尖瓣前叶瓣缘局部略增厚、回声略增强，前叶瓣体局部回声紊乱，呈囊性“蜂窝状”结构，大小为22.1 mm×20.3 mm（图1），与主动脉瓣反流束密切相关，反流束似进入“囊袋”，二尖瓣后叶回声未见异常，瓣口前向血流未见异常，未见明确反流。术前3D-TEE显示：主动脉瓣二瓣畸形，呈前后排列，瓣叶增厚，回声增强（图2），前方瓣叶舒张期脱向左心室流出道（图3）。二尖瓣前叶心房面可探及“囊袋状”结构，大小为16.3 mm×12.9 mm，随心动周期囊壁形态发生改变，收缩期“囊袋”充盈凸向左心房腔，舒张期囊袋塌陷，囊袋颈5.2 mm；彩色多普勒血流成像（color doppler flow imaging，CDFI）显示：主动脉瓣反流束冲击二尖瓣前叶“囊袋”，该“囊袋”与左心室血流相交通，二尖瓣瓣口未见明确反流（图4）。超声诊断为主动脉瓣二瓣畸形-横裂式、粘液液样变性、前方瓣叶脱垂、重度关闭不全伴轻度狭窄；二尖瓣前叶“囊袋”形成、考虑MVA。

1.2　诊疗经过　患者主动脉瓣瓣环扩大，瓣叶呈明显关闭不全，无赘生物附着，将瓣叶切除，进行病理学检测，主动瓣位置换1枚23号生物瓣；经主动脉瓣口探查二尖瓣，未见明确囊肿和赘生物等，前叶瓣体可探及1个大小18 mm×12 mm“囊袋样”结构，“囊袋”颈约5 mm，该结构不影响二尖瓣关闭，故二尖瓣未进行特殊处理。病理结果：瓣膜（主动脉瓣）纤维结缔组织增生，玻变伴黏液变和钙化。

1.3　术后随访　术后即刻TEE显示：主动瓣位生物瓣回声和活动良好，无明确反流及瓣周瘘，二尖

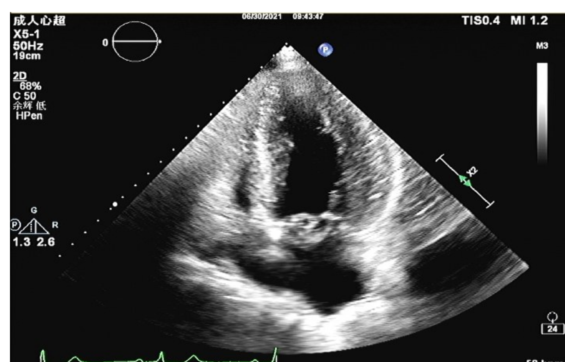

图1　TTE检测MVA患者二尖瓣前叶心室面呈囊性“蜂窝状”声像图

Fig. 1　Cystic “honeycomb” acoustic image of anterior mitral valve ventricle of MVA patient detected by TTE

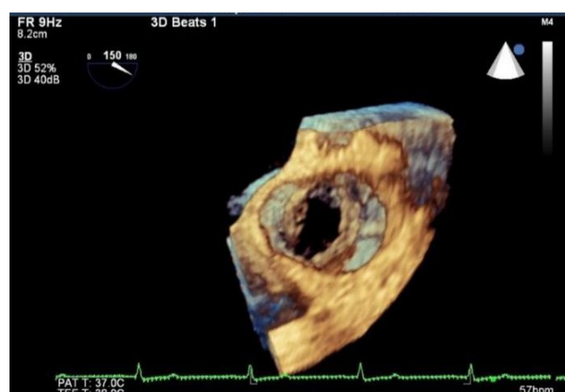

图2　3D-TEE检测MVA患者术前主动脉瓣二瓣畸形声像图

Fig. 2　Acoustic image of bicuspid aortic valve of MVA patient before operation detected by 3D-TEE

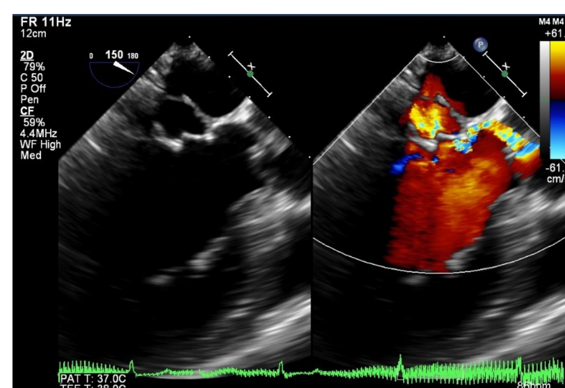

图3　3D-TEE检测MVA患者术前主动脉瓣病变及瓣叶脱垂声像图

Fig. 3　Acoustic image of aortic valve lesions and valve prolapse of MVA pateient before operaton detected by 3D-TEE

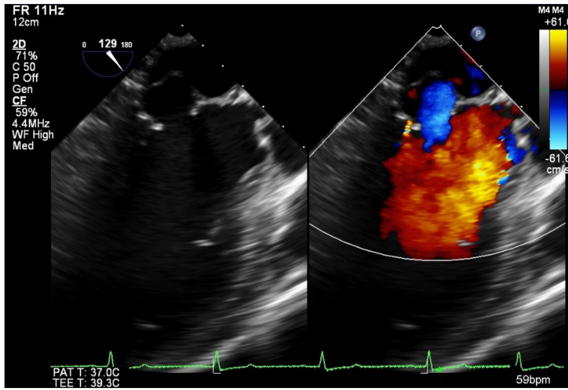

图4　TEE检测MVA患者术前二尖瓣前叶心室面“囊袋状”结构声像图

Fig. 4　Acoustic image of “sac-like” structure of anterior mitral valve surface of MVA patient before operation detected by TEE

瓣“囊袋状”结构仍然存在、大小不变，二尖瓣口探及微量反流。术后10 d TTE显示：主动瓣位生物瓣回声及活动良好，未见明确反流及瓣周瘘，二尖瓣探及大小为22.6 mm×13.7 mm囊性病变，性质及大小较术前无明显变化。术后4个月胸TTE显示：主动瓣位生物瓣回声及活动良好，未见明确反流及瓣周瘘，二尖瓣探及大小21.3 mm×13.3 mm囊性病变，性质及大小较术前无明显变化。

2　讨　论

MVA是1729年首次提出的一种罕见的疾病[1]。在接受TEE的患者中，MVA发病率仅为0.20%～0.29%[2]。表现为二尖瓣瓣体局部向左心房的囊状隆起，收缩期像囊袋一样突入左心房腔，舒张期囊袋塌陷[3]。前叶比后叶更易受累[4]。另有报道[5]将该类病变命名为二尖瓣憩室。MVA通常发生在感染性心内膜炎，特别是主动脉瓣受累并引起严重的主动脉瓣返流时[6]，通常为孤立病变，但多发性动脉瘤也有报道[7]。MVA发病机制尚不明确，绝大多数MVA患者均观察到活动性心内膜炎或慢性炎症改变累及主动脉瓣[8-9]，推测其形成机制包括主动脉瓣心内膜沿二尖瓣主动脉瓣间纤维直接延伸至二尖瓣前叶，随后通过主动脉瓣的反流撞击二尖瓣前叶的心室表面[10]，导致瓣膜组织局部弱化、剥离和结构损伤，随后扩张。感染过程和随后的愈合可能削弱局部瓣膜组织，在心内压的作用下，可能形成真性动脉瘤[11]。其他易感因素包括结缔组织疾病、利布曼-萨克斯（Libman-Sacks）心内膜炎、瓣膜退行性改变和肥厚性梗阻性心肌病[12-15]。上述因素可导致瓣膜变弱，囊状外翻，直径1～4 cm，显微镜下可见疤痕和肉芽组织，亦可能伴有血栓。本患者特殊之处在于无感染性心内膜炎病史，仅存在主动脉瓣先天性二瓣畸形导致主动脉瓣偏心反流束冲击二尖瓣前叶这一诱因，而该种病变更为罕见。

MVA易与二尖瓣血性囊肿相混淆，后者是一种先天性良性心脏肿瘤，主要在胎儿和6个月以内婴幼儿的尸体解剖中发现，多数囊肿可自行消退，且在成人中发病罕见[16]。其形成原因考虑为瓣膜形成过程中血流冲击瓣膜表面的裂隙内陷，后期裂隙未愈合形成，囊肿内部多为血栓样物质[17]。TTE可发现MVA与左心室之间的血流交通，在疾病危险分层、评估、诊断和术后随访中起核心作用。但由于图像清晰度和扫查角度的限制，TTE对疾病的细化诊断有一定的局限性。TEE能够更清晰地显示MVA和左心室之间的细微结构关系，清晰显示血流流入及流出“囊袋”的过程，可将MVA与其他异常区分开来。因此与TTE比较，TEE是一种更可靠、更灵敏和更准确的方法。

MVA患者的临床表现与病变严重程度有关，部分患者无任何体征或症状。MVA破裂可导致血流动力学迅速恶化，导致急性严重二尖瓣反流和急性肺水肿。有文献[18-22]报道MVA并发穿孔后自然愈合的罕见案例。MVA的最佳治疗方式仍有争议[19-23]。保守治疗适用于小而不复杂的MVA，但需要密切随访和监测[20-24]。当MVA破裂或未破裂的MVA较大或伴有严重反流时，需要进行二尖瓣修复或置换手术[21]。该患者MVA大小尚可，未见穿孔及血栓，且二尖瓣瓣口未见反流，因此该患者仅接受主动脉瓣位生物瓣置换手术。术后主动脉瓣位生物瓣未见明确反流及瓣周瘘，原主动脉瓣偏心反流束对二尖瓣前叶瓣体的冲击消失，MVA进一步加重的可能性较小，但亦需要进行长期随访[25]。

综上所述，MVA病因和发病机制不明，常为感染性心内膜炎的并发症，临床表现无特异性，该患者因无感染性心内膜炎病史而罕见。超声心动图等影像学检查有助于MVA术前诊断和鉴别诊断，二维超声心动图是目前临床诊断MVA最有价值的影像学诊断方式，TTE联合TEE是最佳诊断

方案，且可协助MVA患者的诊疗、随访及疗效评价。

利益冲突声明：

所有作者声明不存在利益冲突。

作者贡献声明：

闫岩参与选题和论文撰写，郭锋参与数据收集整理和论文审校，杨四宝和石少敏参与论文审校。

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