Ebstein's Anomaly and Left Ventricular Non Compaction Cardiomyopathy: A Not So Unusual Association

Daniele Andreini[1], Gianluca Pontone[1], Saima Mushtaq[1], Manuela Muratori[1], Marco Guglielmo,[1] Edoardo Conte[1], Patrizia Carità[2], Mauro Pepi[1]

1. Centro Cardiologico Monzino, IRCCS, Milan, Italy.
2. University Hospital Paolo Giaccone, Palermo, Italy.

Corresponding author:
Patrizia Carità,
Via del Vespro, 129
Palermo, Italy
Email: patcar@hotmail.it

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Left ventricular non-compaction (LVNC) is a rare congenital cardiomyopathy characterized by the presence of an extremely thickened endocardial layer with prominent trabeculations and deep recesses in communication with ventricular chamber and determining a typical spongeous aspect [1]. Non-compaction of the ventricular myocardium is thought to be a congenital arrest of endomyocardial embryogenesis in utero [2]. It has been reported a strong association with uncommon neuro-muscular disorders, such as Barth syndrome, Emery-Dreifuss muscular dystrophy, myotubular myopathy, as well as mitochondrial abnormalities [3-5]. LVNC has been also associated with other cardiac abnormalities such as Ebstein's anomaly [6]. Ebstein's anomaly is a rare congenital cardiac disease, affecting about 1 in 200.000 live births [7]. It is characterized by apical displacement of the septal leaflet of the tricuspid valve causing significant tricuspid regurgitation and reduction of the functional right ventricle, right atrial and right ventricular dilatation and arrhythmias [8]. Recent studies have identified a genetic mutation (MYH7) as a cause of Ebstein's anomaly associated with LVNC and other congenital heart disease [7].

We presented two cases evaluated in our Institution. The first case is a 55-year-old man, who was evaluated because of mild ST-segment anomalies on ECG. The echocardiography showed a right ventricle (RV) dilation and a septal tricuspid valve leaflet apical displacement, with a suspicion of Ebstein's anomaly and/or arrhythmogenic right ventricular dysplasia. In order to evaluate these findings, the patient was referred to our Institute and underwent contrast-enhanced cardiovascular magnetic resonance (CMR, Discovery 450, GE Healthcare, 1,5 T). Cine imaging by steady-state free-precession sequence showed a moderate dilation of the left ventricle (LV), with normal regional kinesis and systolic function and presence of an extensive non-compacted myocardial layer lining the cavity of the middle-apical portions of the LV, with ratio of noncompacted to compacted myocardium >2.3 in diastole in all these segments (Figure 1, Panels A and B). The black-blood T2-weighted short inversion-time inversion-recovery sequence in short-axis confirmed the "sponge-like" appearance of LV myocardium (Figure 1, Panel C). Cine imaging also showed a mild dilation of the RV, with normal kinesis and systolic function and confirmed the presence of septal tricuspid valve leaflet apical displacement, with a distance between annulus plane and septal leaflet of 21 mm, even in the absence of severe RV atrialization. These findings were consistent with diagnosis of LV Non-Compaction and Ebstein anomaly. Patient also underwent transthoracic two-dimensional and three-dimensional echocardiography, confirming the LV hypertrabeculation, the septal tricuspid valve leaflet apical displacement (Figure 1, Panel D) and the spongy myocardium of the LV apex.

The second case refers to a 46-year-old asymptomatic man who was referred to our Institution for the CMR after the percutaneous closure of an interatrial septum defect for the echocardiographic suspicion of Ebstein's anomaly with right ventricle dilation. He previously underwent to ablation of accessory pathway for Wolf-Parkinson-White syndrome. The CMR images confirmed the tricuspid anomaly, and interestingly showed the association of left ventricular non compaction and supra valvular pulmonary stenosis with post stenotic mild dilation (36 mm) of pulmonary artery. (Figure 2, Panels A-C).

* Corresponding author. E-mail: patcar@hotmail.it
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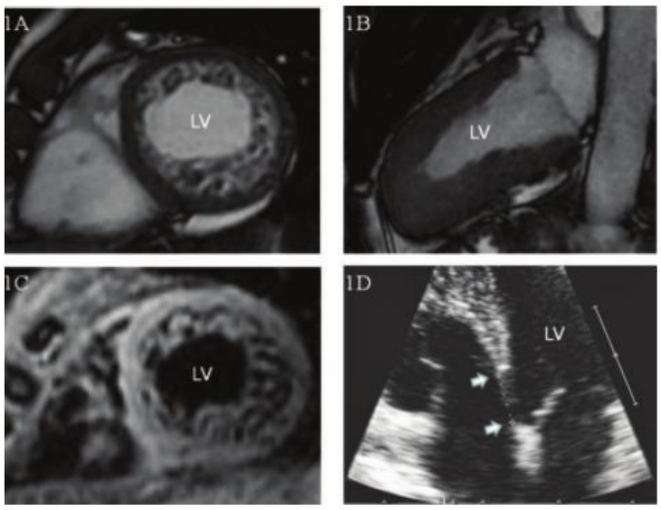

Figure 1. Panel A. Cardiovascular magnetic resonance steady-state free-precession sequence, short axis, showing the presence of an extensive non-compacted myocardial layer lining the cavity of the middle-apical portions of the left ventricle. Panel B. Cardiovascular magnetic resonance steady-state free-precession sequence, long axis, confirming the hypertrabeculation of the middle-apical portions of the left ventricle. Panel C. Cardiovascular magnetic resonance black-blood T2-weighted short inversion-time inversion-recovery, short-axis, confirmed the "sponge-like" appearance of left ventricle myocardium. Panel D. Two-dimensional echocardiography, 4-chambers view, showing the apical displacement of the septal tricuspid valve leaflet (headarrows: distance of 21 mm from the annulus to the septal leaflet inserction). LV=left ventricle.

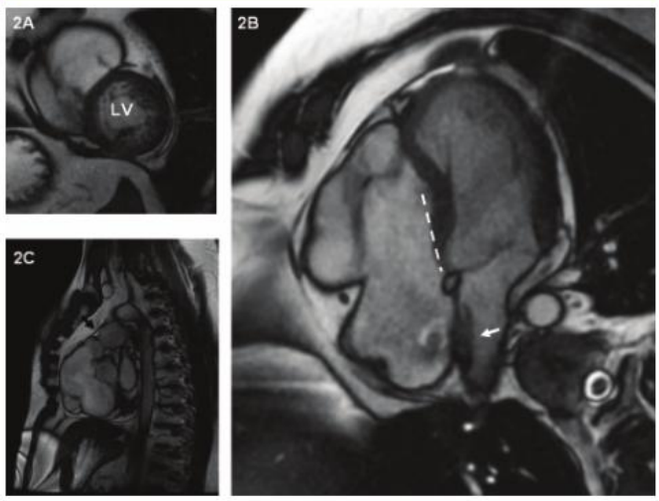

Figure 2. Panel A. Cardiovascular magnetic resonance steady-state free-precession sequence, short axis, showing the presence of an extensive non-compacted myocardial layer lining the cavity of the middle-apical portions of the left ventricle. Panel B. Cardiovascular magnetic resonance steady-state free-precession sequence, long axis 4 chambers, showing the apical displacement of the septal leaflet of the tricuspid valve leaflet and the occluder of interatrial septal defect (white headarrow). Panel C. supra-valvular pulmonary stenosis (black headarrow)

The review of literature and our findings suggest that Ebstein's anomaly is a cardiac disorder that is not confined to the right heart [9-10]. The association with LVNC is not so uncommon and all patients with Ebstein's anomaly should be carefully evaluated for LVNC. The prompt identification of myocardial non compaction is important because of its high mortality due to progressive heart failure, tromboembolism and malignant arrhythmias. The CMR allows a comprehensive evaluation of associated findings.

Declarations of Interest

The authors declare no conflicts of interest.

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The authors state that they abide by the "Requirements for Ethical Publishing in Biomedical Journals [11].

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