CLINICAL VIGNETTE

Ring-shaped subpulmonary membrane in an adult patient with Charcot–Marie–Tooth disease type 2

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A 34-year old male patient was diagnosed with Charcot–Marie–Tooth disease (CMT) type 2 in childhood and remained asymptomatic until adolescence, when he developed progressive dyspnea. Physical examination revealed an unknown systolic ejection murmur at the left lower sternal border. Electrocardiography showed right ventricular (RV) hypertrophy and right axis deviation. Transthoracic and transesophageal echocardiography (Phillips iE33, Phillips Healthcare, Best, the Netherlands) detected severe RV outflow tract (RVOT) hypertrophy and the presence of a fibrous ring-shaped subpulmonary membrane causing severe subpulmonic stenosis (a transmembrane pressure gradient of 126 mm Hg). Magnetic resonance imaging (MRI; Philips Achieva DS 1.5T, Philips Healthcare) confirmed an infundibular fibrous membrane below the pulmonary valve (22 mm from the valve). There was no evidence of pulmonary valve dysfunction or additional abnormalities. The patient was referred for surgery. Using longitudinal right ventriculotomy at the infundibulum, a tight stenosis caused by the ring-shaped membrane was found. A resection of the hypertrophied muscle and membrane was performed. The RVOT was enlarged using a bovine pericardium patch. Transpulmonary gradients were measured and no residual gradients across the RVOT were detected. The patient had an uneventful recovery.

Subpulmonary obstructions are uncommon mechanisms associated with infundibular hypertrophy or subpulmonary muscle bundles.[1,2] An isolated fibrous subpulmonary membrane is rarely observed in adults and was even more rarely reported as a cause of RVOT obstruction, especially in the absence of ventricular septal defects or pulmonary valve abnormalities.[1,2] It usually causes RVOT and RV hypertrophy and is characterized by the presence of an anomalous fibrous ring bundle that protrudes from the free wall of the RV.[1-5] Symptoms appear during childhood. The subsequent course is silent until patients progressively develop obstruction followed by RV hypertrophy.[1-3,5] Adult patients are rarely asymptomatic because severe RVOT obstruction manifests itself as low cardiac output and RV failure.[1-3,5]

Charcot–Marie–Tooth disease is an inherited neurologic disorder affecting peripheral nerves. Type 1 disease affects the myelin sheath of peripheral nerves, while type 2 is less common and affects the axon rather than myelin sheath. Generally, CMT has been associated with conduction disturbances or dilated myocardiopathy. No structural cardiac abnormalities have been previously described in relation to this polyneuropathy. To the best of our knowledge, a subpulmonary membrane has never been described before in the context of CMT disease. It is possible that due to physical limitations patients with CMT disease avoid physical effort, which contributes to a delayed diagnosis.[1-3,5] A high clinical suspicion is necessary to ensure a prompt diagnosis and clinical management. An accurate

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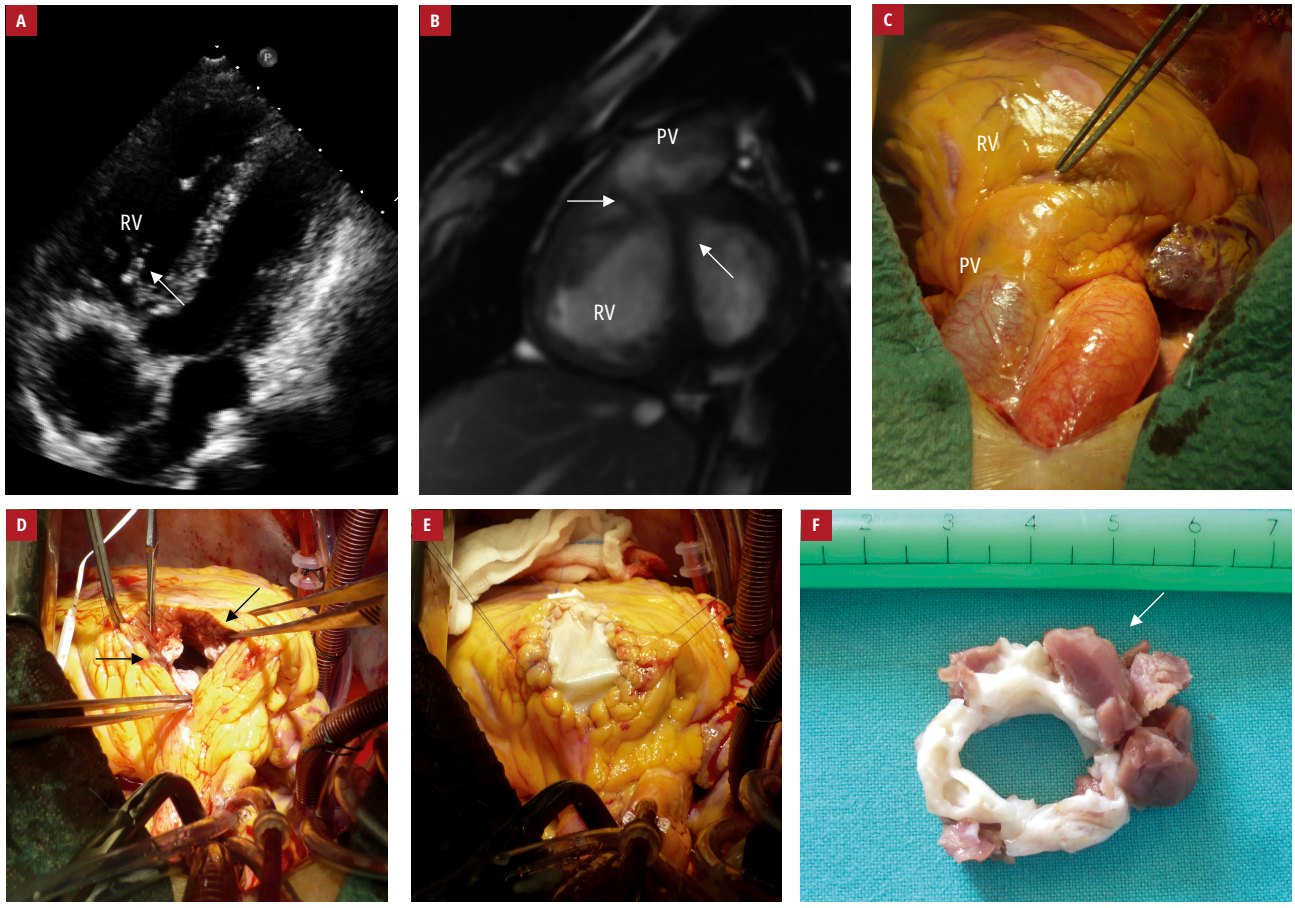

FIGURE 1 **A** – echocardiography showing the subpulmonary membrane (arrow) in the right ventricle (RV); **B** – cardiac magnetic resonance image showing the subpulmonary membrane (arrows) in the RV below the pulmonary valve (PV); **C** – an intraoperative view showing the narrow infundibulum; **D** – aperture of the right ventricular outflow tract (RVOT) and resection of the subpulmonary ring (arrows); **E** – RVOT enlargement using a bovine pericardial graft; **F** – ring-shaped subpulmonary membrane showing the double component of fibrosis and muscle (arrows), result of RV hypertrophy

assessment is crucial, and the diagnosis requires a comprehensive echocardiographic examination. However, computed tomography, MRI, or right ventriculography can provide further details in functional assessment.[1,3-4] Currently, there are no clear guidelines for the management of CMT disease, especially in adults. Percutaneous balloon dilatation is possible but provides suboptimal outcomes. Surgical approach is an option, particularly in symptomatic cases, in the presence of additional congenital abnormalities, or when the pressure gradient between the RV and pulmonary artery is higher than 40 mm Hg. A clinical and functional improvement is remarkable and long-term surgical outcomes are excellent.[1-2,5]

ARTICLE INFORMATION

CONFLICT OF INTEREST None declared.

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