Ring-shaped subpulmonic infundibular membrane diagnosed in an
adult affected by type II Charcot-Marie-Tooth disease

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Short title: Ring-shaped subpulmonic membrane in type II Charcot-Marie-Tooth disease.

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A 34-year old male patient was diagnosed during childhood of a Type II Charcot-Marie-Tooth-Disease (CMT) and remained asymptomatic until adolescence, when progressive dyspnea appeared. Physical examination revealed an unknown systolic ejection murmur at the left lower sternal border. Electrocardiogram showed right ventricular (RV) hypertrophy and right axis deviation. Transthoracic and transesophageal echocardiography (Phillips iE33,Phillips) showed severe RV outflow tract (RVOT) hypertrophy and presence of a fibrous ring-shaped subpulmonic membrane causing severe subpulmonic stenosis (transmembrane pressure gradient of 126 mmHg). Magnetic resonance imaging (MRI-Philips Achieva DS 1.5T.Philips Healthcare) confirmed an infundibular fibrous membrane 22 mm below the pulmonary valve. There was no evidence of pulmonary valve dysfunction or additional anomalies. Patient was scheduled for surgery. Through longitudinal right ventriculotomy at the infundibulum, a tight stenosis caused by a ring-shaped membrane was found. A resection of hypertrophied muscle and membrane was made. RVOT was enlarged using a bovine pericardium patch. Transpulmonary gradients were measured and no residual gradients across the RVOT were detected. Patient had an uneventfully recovery.

**Comment**

Subpulmonary obstructions are uncommon mechanisms associated to infundibular hypertrophy or subpulmonic muscle bundles [1-2]. An isolated fibrous subpulmonic membrane is exceptional in adults and even more as cause of RVOT obstruction, especially in absence of ventricular septal defects or pulmonary valve abnormalities [1,2]. It usually causes RVOT and a RV hypertrophy and is characterized by the presence of an anomalous fibrous ring bundle which protrudes from the free wall of the RV [1-5]. Symptoms appear during childhood, remaining asymptomatic while obstruction is progressively established and RV hypertrophy develops [1-3,5]. Adults
affected are rarely asymptomatic because severe RVOT obstruction is manifested as low cardiac output and RV failure [1-3,5].

CMT is an inherited neurological disorder affecting peripheral nerves. Type I CMT affects myelin sheath of peripheral nerves. Type II CMT is less common and affects axon rather than myelin sheath. Generally, CMT has been associated to conduction disturbances or dilated myocardiopathy. No structural cardiac abnormalities have been previously described in association to this polyneuropathy. To the best of our knowledge, a subpulmonic membrane has never been described before in the background of a CMT disease. Probably, physical limitations of CMT patients make them avoid physical efforts and contributes to a delayed diagnosis [1-3,5]. A high clinical suspicion is necessary to assure a prompt diagnosis and clinical management. An accurate assessment is crucial and diagnosis requires a full echocardiographic examination. However, CT-scan, MRI or right ventriculography can provide further information in the functional assessment [1,3-4]. Currently, no clear guidelines exist for management of this disease especially for adult. Percutaneous balloon treatment is possible, but obtains suboptimal results. Surgical strategy is an option, specially for symptomatic cases, when additional congenital abnormalities exist or when pressure gradient between RV and pulmonary artery is greater than 40 mmHg. A clinical and functional improvement is remarkable and long-term surgical outcomes are excellent [1-2,5].
REFERENCES:


Figure 1

A: Echocardiography showing subpulmonary membrane (arrow). Right Ventricle(**).

B: Cardiac magnetic resonance showing subpulmonary membrane 22 mm bellow pulmonary valve (*).

C: Intraoperative view showing narrow of infundibulum.

D: Aperture of RVOT and resection of subpulmonary ring.

E: RVOT enlargement using a bovine pericardial graft.

F: Ring-shaped subpulmonary membrane showing double component of fibrosis and muscle, resulted of RV hypertrophy.