

Case Report Double Valve Replacement in a Patient with Maroteaux – Lamy syndrome. An Ultimate Team Challenge.

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Case report

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Abstract

Background

The Maroteaux-Lamy syndrome (Mucopolysaccharidosis type VI) is a rare, inherited metabolic disease that results in progressive tissue accumulation of dermatan-sulfated glycosaminoglycans and inflammatory consequences that almost always affects the heart valves. From the anesthesia point of view, these rare cases are a serious challenge since the features of the syndrome entail difficulties in airway management and ventilation. It's more than possible that the surgical team will perform a non-straightforward procedure.

Case presentation

A 42 year old male with Maroteaux-Lamy syndrome was referred to our department with shortness of breath, due to severe aortic stenosis, and at least moderate mitral valve regurgitation.

The patient was initially scheduled for aortic valve replacement. After multiple attempts with video assisted laryngoscopy, the endotracheal intubation was achieved with the aid of fiberoptic bronchoscopy, while the ventilation succeeded only with laryngeal mask. The somatic features of the syndrome that made the anesthesia induction extremely difficult, also affected the surgical procedure. Suboptimal exposure of the mitral valve, patch enlargement of the aortic root to host the bigger possible prosthesis, and the hard decision to replace the mitral valve even with a marginal indication were the intraoperative difficulties for the surgical team.

Finally, the patient underwent a successful double valve replacement with aortic root enlargement and after 18 months remains improved.

Conclusion

Patients with Maroteaux-Lamy syndrome represent a challenge for both anesthesiologists and cardiac surgeons. The whole team should be well prepared to deal with difficulties in airway management, ventilation and surgical valve exposure. The cardiac surgeon should be ready to offer additional procedures and even replace "prematurely" a moderately diseased valve in order to avoid a dangerous reoperation. The limited knowledges on the natural history and progression of the Maroteaux-Lamy syndrome valvulopathy and the difficulties in anesthesia induction support this approach.

Background

Maroteaux-Lamy syndrome, first described by Maroteaux in 1963, is a rare, inherited metabolic disease, caused by mutations in the gene coding for N-acetylgalactosamin-4-sulfatase, a lysosomal enzyme involved in the degradation of dermatan sulfate [1]. Dermatan-sulfated glycosaminoglycans are a prominent component of normal cardiac valve tissue, so their progressive accumulation and inflammatory consequences result in left heart valve degeneration. Nearly all patients with Maroteaux-

Lamy syndrome will develop cardiac valve thickening, and dysfunction requiring valve replacement during their life course [2]. It is more than probable, that the procedure will not be straightforward due to the physical characteristics of these patients. In the literature, there are only few reports of double valve replacement in patients with this syndrome. We present our experience highlighting the intraoperative management, and decision making.

Case Presentation

A 42 year old male with diagnosed MLS since childhood was referred to us due to symptomatic severe aortic stenosis, along with at least moderate mitral valve regurgitation. The patient was presented in sinus rhythm, with short trunk (H 135cm, W 40kg), stiff neck, limited joints mobility, large tongue, limited mouth opening, forehead bossing, and umbilical hernia [figure 1A]. The echocardiogram showed severe aortic valve stenosis (mean gradient 69 mm Hg, peak velocity 4,16 m/s), at least moderate mitral regurgitation (regurgitant orifice area 0.37 cm², regurgitant fraction 50%), left ventricle hypertrophy (mass index 120 g/m²), left ventricle ejection fraction 55%, indexed left atrial volume 35ml/m², and mean pulmonary artery systolic pressure at rest 28mmHg. The angiogram revealed normal coronary arteries.

The oropharyngeal features such as neck stiffness, small mouth opening, with large tongue of the patient, made the airway management a very difficult and life-threatening procedure. Specifically, the routine endotracheal intubation was impossible, despite the multiple attempts even with video assisted laryngoscopy. Meanwhile, the ventilation was possible only with the use of laryngeal mask. Additionally, the restricted compliance of the thoracic cage due to the thoracic joints degeneration and stiffness, made more difficult the patient's ventilation. Finally, the intubation was achieved with fiberoptic bronchoscope. The insertion of a transesophageal ultrasound probe was considered dangerous and therefore was avoided.

The operation was performed with midline sternotomy, aorto-bicaval cannulation, moderate hypothermic cardiopulmonary bypass and Bretschneider cardioplegic solution. The intraoperative findings included fibrotic and stenotic aortic valve [figure 1B] with a small aortic root and macroscopic degeneration of mitral leaflets. An aortic and mitral valve replacement with Sorin Bicarbon Slimline 17 and Sorin Bicarbon Fitline 23 (Sorin Group Italia S.r.l., Saluggia VC Italy) respectively was performed. The macroscopically diseased mitral valve with at least moderate regurgitation, the uncertain natural progress of the valve dysfunction and also the need to avoid another dangerous anesthesia induction in the future forced the decision to replace the mitral valve. The small aortic ring and aortic root were addressed with a modified Nick's procedure using a polyester vascular patch (LeMaitre Vascular, Burlington, MA USA).

The patient experienced an uneventful in hospital course, and he was discharged the 8th postoperative day. No patient-prosthesis mismatch signs were presented. The histological examination of the mitral valve revealed leaflets myxoid degeneration and clear cells accumulation [figure 2]. Eighteen months postoperatively, the patient remains asymptomatic in sinus rhythm [figure 1A]. The recent echocardiogram showed normal left ventricular function, normal function of the prosthetic valves without

leak. The aortic prosthesis mean and peak gradients were 11, 8 and 22, 8 mmHg respectively, and the aortic peak velocity 2, 39 m/sec. The mitral prosthesis peak gradient was 6, 63 m/sec.

Discussion And Conclusions

The Maroteaux-Lamy syndrome prevalence varies among populations from 0.0132 to 20.0/100000 newborns [1]. Despite the heterogeneity of the syndrome phenotype, mental development is usually normal, and cardiac valve pathology is present in almost all individuals. Somatic features of the syndrome are growth retardation, dwarfism, coarse facial features, joint stiffness, dysostosis multiplex, hepatosplenomegaly, corneal clouding, macrocephaly, hearing loss, and umbilical, or inguinal hernias [2]. Specific features of the syndrome constitute a great challenge for both anesthesiologists and cardiac surgeons. Neck stiffness, large tongue, small mouth opening, tracheobronchial narrowing, chest deformities and poor ribs mobility, all represent challenges for airway management and ventilation. Fatal events during anesthesia induction have been described in the literature [3].

Difficult sternal retraction due to skeletal abnormalities and consequently problematic mitral valve exposure, along with the inconveniently small aortic and mitral annulus with increased patient-prosthesis mismatch possibility, constitute the intraoperative challenges for the cardiac surgeon.

In the literature, bail out maneuvers like implanting a reversed aortic prosthesis in the mitral position or aortic root enlargement, such as the modified Nick's we offered to our patient are extremely rare [4].

In addition to the technical difficulties, hard intraoperative decisions will be taken, especially in cases of a second moderately diseased valve, such as the mitral valve of our patient. Performing a double valve replacement definitely increases the operative risk. Leaving unaddressed the second diseased valve exposes the patient to an increased risk of a difficult reoperation. Previous papers proposed a non-aggressive approach with the second diseased valve, expecting stabilization with the enzyme replacement therapy [5]. Unfortunately, recent scientific data suggest that enzyme replacement therapy has no effect on the diseased valve [6].

The need to avoid a dangerous reoperation should influence the decision regarding the second diseased valve.

In conclusion, patients with Maroteaux-Lamy syndrome represent a heart team challenge. Proper cardiac anesthesia and cardiac surgery preparation are mandatory for the management of such a rare and vulnerable patients. An additional "premature" replacement of a second moderately diseased valve might be a useful option.

Abbreviations

No abbreviations used.

Declarations

Ethical declarations and consent. Approval was obtained from the Athens Medical Center Ethics Committee with reference number: KM140721, which confirms that the study was performed in accordance with the Declaration of Helsinki, and hasn't been granted an exemption from requiring ethics approval.

The written patient's consent for treatment, participation and publication was obtained.

Availability of data and materials. The used datasets of the current study are available from the corresponding author on reasonable request.

Competing interests. The authors declare that they have no competing interests.

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Figures

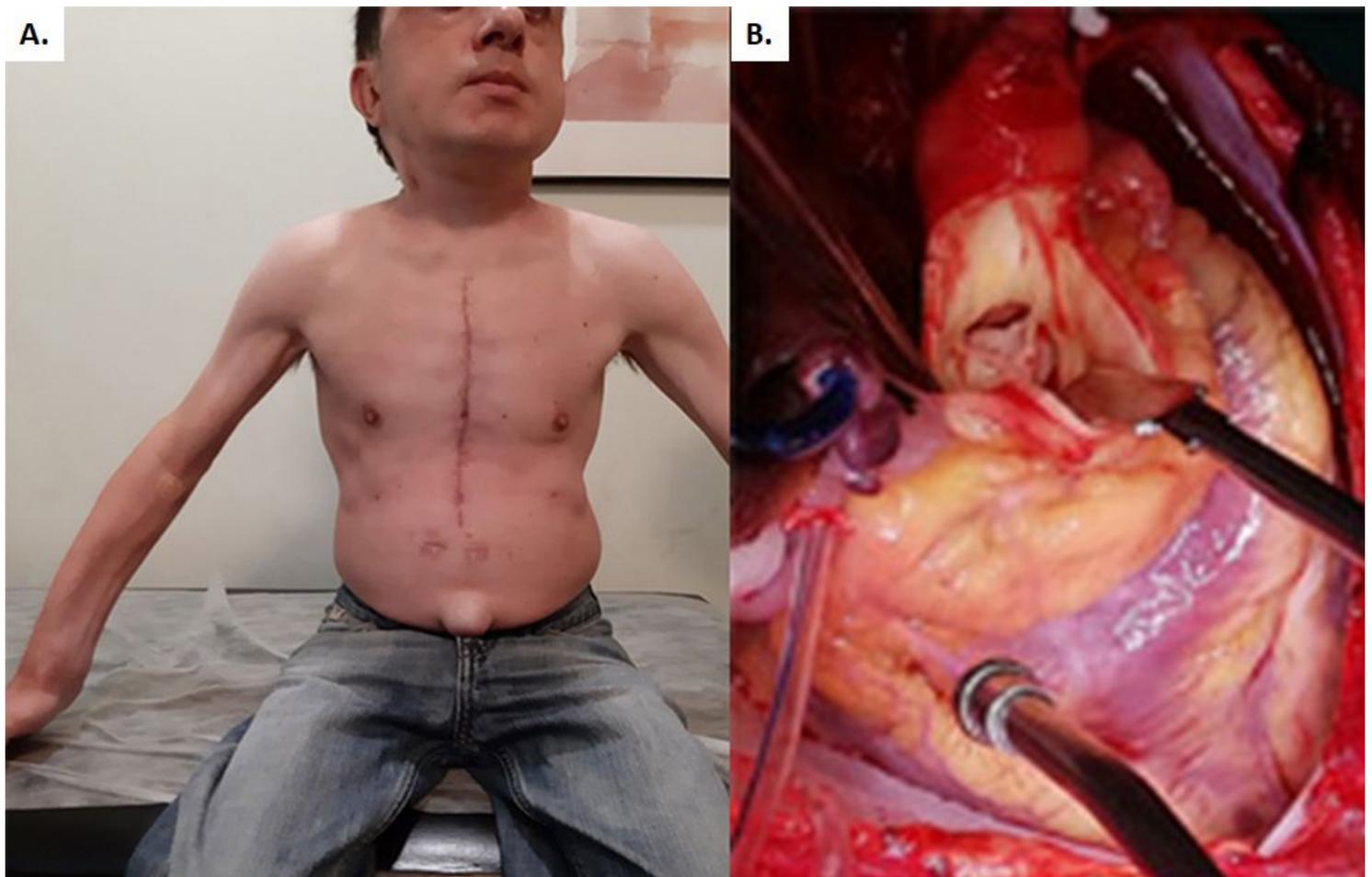


Figure 1

A. Fibrotic and stenotic aortic valve. B. The postoperative photo of the patient shows his phenotype.

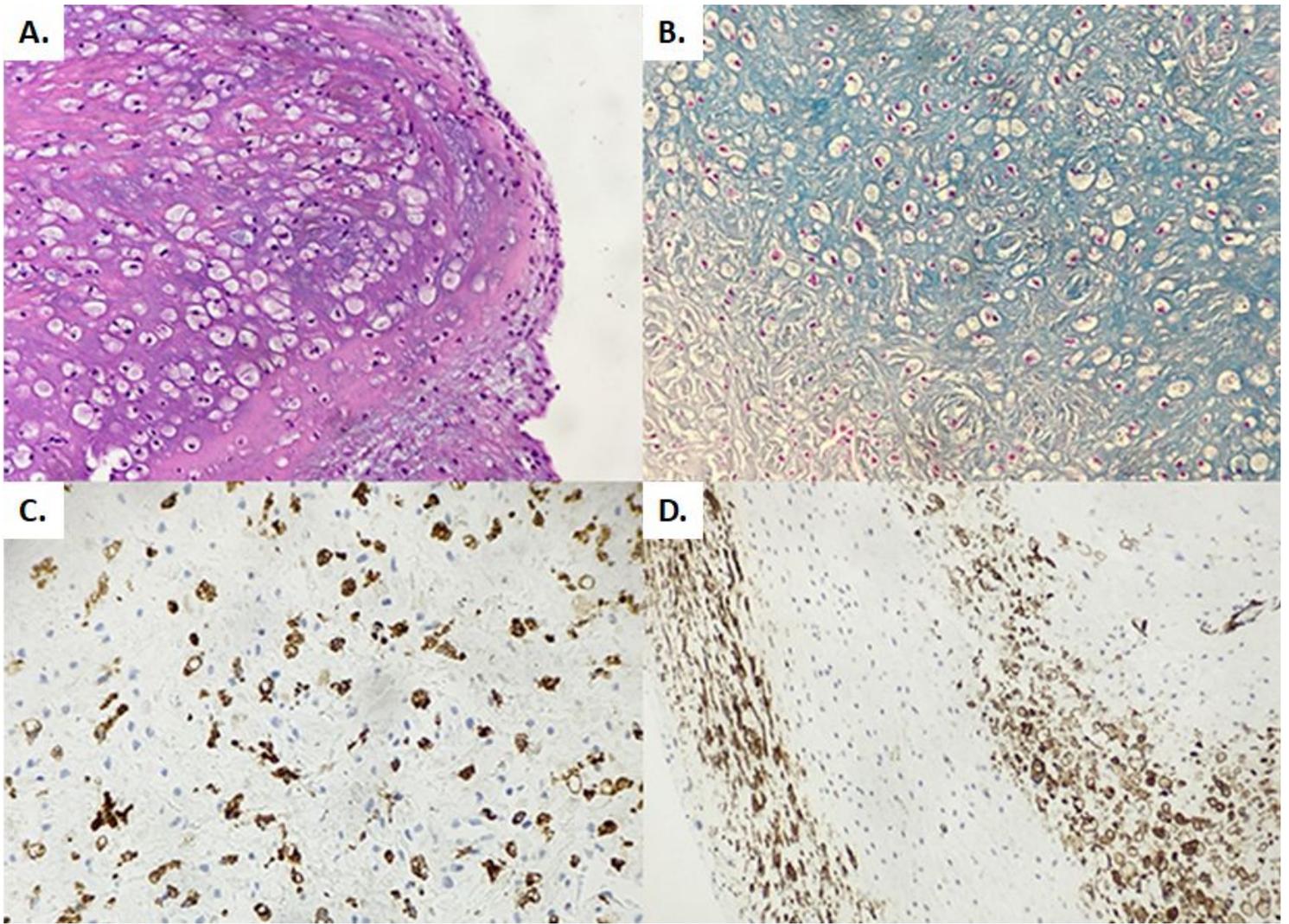


Figure 2

Mitral valve histological examination. A. Valve stromal infiltration from clear cells, polysaccharides and mucopolysaccharides, B. Alcian Blue 2,5 positive in stromal glycosaminoglycans, C. CD68 (+) in clear cells, D. Actin (+) in myofibroblasts and clear cells.