

A rare and challenging mitral valve replacement in a child with Hurler syndrome

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ABSTRACT

Mucopolysaccharidoses are progressive inherited lysosomal storage disorders, and early cardiac involvement is common. Valvular involvement of mucopolysaccharidoses usually causes regurgitation, most commonly affecting the left heart and mitral valve. In this case, we discuss the treatment and perioperative management of mitral valve degeneration in a child with Hurler syndrome by performing mitral valve replacement.

Keywords: Hurler syndrome, valvular disease, mucopolysaccharidosis.

Mucopolysaccharidoses (MPSs) are progressive inherited lysosomal storage disorders caused by the absence of enzymes involved in glycosaminoglycans' (GAGs) degradation. Mucopolysaccharidosis causes multiorgan system dysfunction and clinical implications that vary according to the specific GAG accumulated and the specific enzyme mutations present.^[1] There are seven subtypes of MPS defined in the current literature. Early cardiac involvement is common, particularly in patients with MPS type 1 (Hurler syndrome).^[2] There are very rare pediatric cases published on valvular surgery in patients with Hurler syndrome in the current literature. In this case, we discuss the treatment and perioperative management of mitral valve degeneration in a child with Hurler syndrome by performing mitral valve replacement.

CASE REPORT

An 11-year-old female diagnosed with MPS type 1 when she was seven-year-old presented to our clinic. Phenotypically short stature, thickening of the face, short and stiff neck, claw hand, and wide abdomen were observed (Figure 1). The patient had a history of frequent respiratory tract infections and multiple intensive care unit hospitalization, and a tracheostomy was performed two years ago. The patient had been undergoing enzyme replacement therapy (Laronidase; Genzyme Europe B.V, Amsterdam, Netherlands) since

the age of eight. The patient, who was admitted to the emergency department with a pneumonia attack 15 days ago, received intensive antibiotherapy treatment in the intensive care unit. On physical examination of the patient, a holosystolic murmur extending to the axilla was heard upon listening. Crepitant rales were heard in the bilateral lower zones of her lungs. The patient had hepatomegaly and 2+ pitting edema in both lower extremities. Transthoracic echocardiography revealed moderate mitral stenosis and severe mitral regurgitation. The mitral valve area was 0.95 cm², the peak gradient was 24 mmHg, the mean gradient was 10 mmHg, and the regurgitant fraction was 46% (Figure 2). Subvalvular involvement, thickened leaflets, and commissural fusion were detected. The left atrium bulged to the right, and marked left atrium dilatation (4.6×5 cm) was observed. The patient was discussed in the pediatric cardiology and cardiac surgery council, and a decision for mitral valve replacement was made.

The patient was taken to the operating room. A median sternotomy was performed. The patient had a

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Figure 1. Phenotypic appearance of the patient with short stature, prominent smallness of the jaws, cervical shortness, rough facial features, claw-like hands, and umbilical hernia.

large thymus tissue covering the entire mediastinum. The thymus was completely removed. It was observed that the mediastinum was quite narrow. Aortic bicaval cannulation was performed. The procedure was initiated under moderate hypothermia. Antegrade blood cardioplegia was used for diastolic arrest. Left

atriotomy was performed through the Sondergaard groove. Mitral valve leaflets were in dysmorphic appearance, and both leaflets were markedly thickened. Chordae tendineae were shortened, and papillary muscles were thickened. Diffuse involvement of the subvalvular apparatus impaired the mobility of the leaflets. There was thickening in the mitral annulus due to dense deposit accumulation, and the tissue was fragile (Figure 3).

Mitral valve leaflets were resected. It was not possible to preserve the subvalvular apparatus structures in this patient as it was obvious that it would adversely affect the opening of the mechanical valve leaflets. The annulus was tight and flimsy. Therefore, the annular structure was strengthened by crossing pledget-supported single sutures. Bileaflet size 20 supra-annular mechanical mitral valve (Open Pivot AP; Medtronic, Minneapolis, MN, USA) was used for replacement. Mechanical valve opening was checked with intraoperative transthoracic echocardiography. Cross-clamp time was 112 min. The patient was weaned from cardiopulmonary bypass with a low-dose dopamine infusion. In the third postoperative hour, the intubation tube was replaced with a tracheostomy cannula. The patient's need for positive pressure ventilation continued for six days due to early pulmonary atelectasis. The patient, who was applied respiratory physiotherapy during the intensive care period, was taken to the clinical service on the 15th day. During this whole process, enzyme replacement therapy treatment was continued without interruption. The patient was discharged on the 19th postoperative day without any complications.

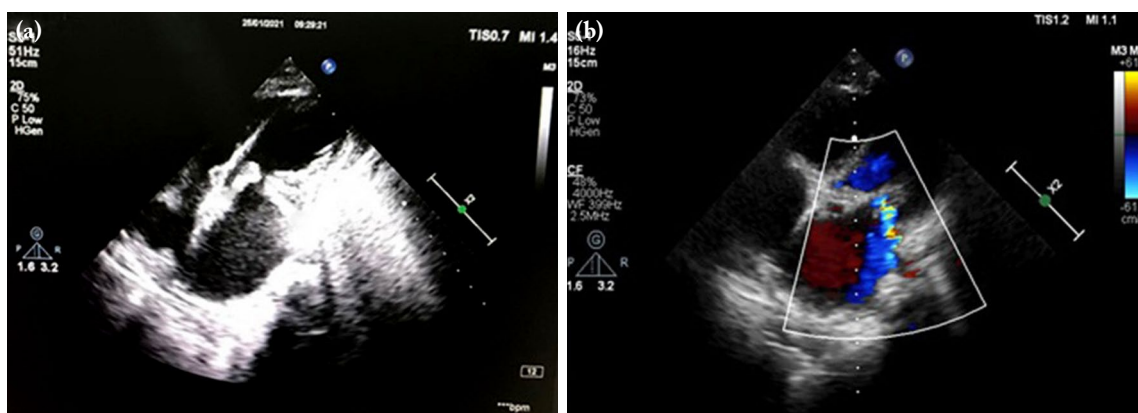


Figure 2. Transthoracic echocardiography demonstrates thickened mitral valve leaflets, (a) thickened and fused subvalvular apparatus, and (b) eccentric failure jet.

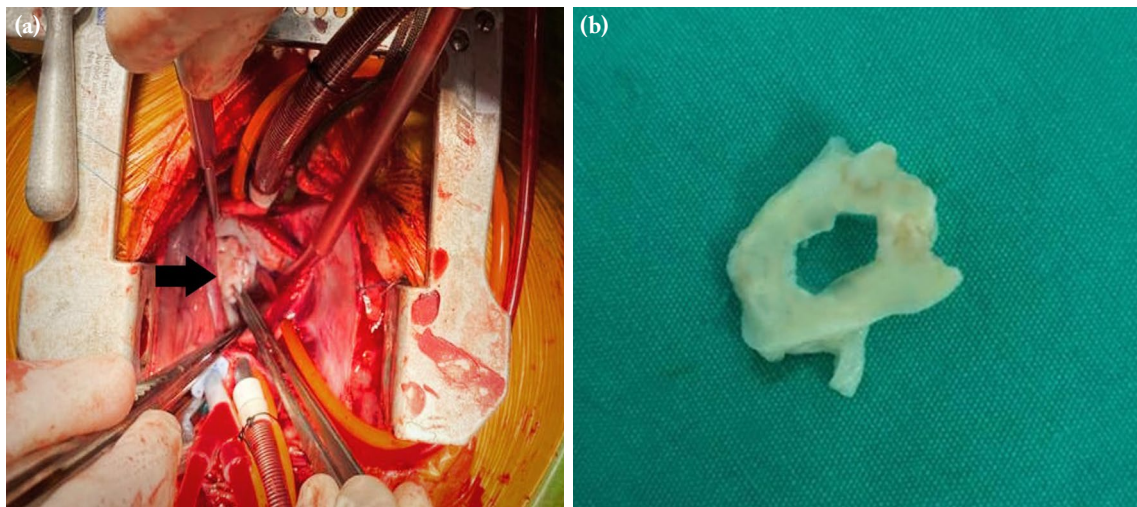


Figure 3. (a) The intraoperative view of the patient (the mitral valve is seen through the left atriotomy) and (b) the mitral valve removed in a single piece.

DISCUSSION

Mucopolysaccharidosis can affect all tissues that develop due to genetic mutation of lysosomal enzymes and where deposits accumulate. Phenotypes such as growth retardation, skeletal and joint deformities, vision and hearing problems, spinal cord compression symptoms, and hepatomegaly may be encountered. Cardiac involvement is observed at a high rate in this patient group. Previous studies have shown that the causative subtypes (MPS types 1, 2, and 4) are more commonly associated with cardiovascular pathologies, particularly with dermatan sulfate catabolism defects.^[1,3] In the current literature, it is seen that the valvular involvement of MPS usually causes regurgitation, most commonly affects the left heart, and the most frequently affected valve is the mitral valve. The positive effects of medical treatment on valvular involvement in patients with an early diagnosis are controversial. After the administration of enzyme replacement therapy, myocardial and ventricular function improved in patients with MPS, myocardial deposit accumulation decreased, and progression slowed. However, poor vascularity of heart valves limits enzyme replacement therapy's effect on valvular degeneration.^[4]

There are procedural difficulties in valvular replacement surgery in patients with MPS. As the mediastinum and heart chambers are tight, this situation makes surgical manipulation and retraction difficult in patients with MPS. Besides, periprosthetic

annular tissue has a narrow and tight structure due to deposit accumulation. However, the annular tissue is not structurally durable enough. The use of pericardial pledget and felt pledget has been reported in the literature.^[5,6] We preferred to use Teflon felt to increase durability. Additionally, due to annular stenosis, we preferred a supra-annular prosthetic valve. Placing the prosthesis in a supra-annular position appears to be a good alternative in patients with MPS, where traditional annular implantation is often not possible.

The positive effects of preserving the subvalvular apparatus on postoperative myocardial functions in mitral valve replacement are already known. However, in the patient we operated on, there was a mass effect due to the severe thickening of the papillary muscle, and the chordae tendineae were severely shortened. We think that aggressive excision of papillary muscles and chordae tendineae will prevent the need for reintervention due to complications such as a stuck prosthetic valve. Kitabayashi et al.^[5] reported in their case report in 2007 that they resected the chords and papillary muscles.

We think that myocardial protection during cardiopulmonary bypass is essential in these patients. Since diffuse intimal proliferation and coronary arterial stenosis/occlusion in epicardial coronary arteries due to GAG storage were detected in two studies.^[7,8] In addition, subvalvular apparatus excision for valve replacement in these patients may cause myocardial dysfunction. Therefore, we tried to overcome this

problem by adhering to the full-dose cardioplegia protocol, applying topical cold saline, and venting the left heart chambers.

In the postoperative period, a restrictive lung disease with reduced compliance, predisposition to upper and lower respiratory tract infections, short tracheal length, macroglossia, and neck deformities make these patients' extubation process difficult. We applied intensive respiratory physiotherapy to our patient during the preoperative period. Besides, since the patient had a tracheostomy, we performed intraoperative intubation procedures and postoperative respiratory management more efficiently. However, our patient had a prolonged intensive care period due to the development of atelectasis. The literature observed that patients with MPS who underwent valve surgery had an extended intensive care period, and pulmonary complications were frequent. Therefore, when the decision for valve replacement is made for these patients, it is necessary to be prepared for lung complications during the intensive care unit stay.

In conclusion, cardiac involvement in patients with Hurler syndrome may further impair already poor pulmonary function. The elimination of valve pathology prolongs the patient's life. Good perioperative preparation is vital in this patient group. The surgeon should know that they will encounter a narrow and structurally challenging annulus in the intraoperative period and take precautions against this situation.

Patient Consent for Publication: A written informed consent was obtained from the patient's parents for the publication of this case report and accompanying images.

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Author Contributions: Idea/concept, data collection and/or processing, analysis and/or interpretation, writing

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