Case Report

Aortic Valve Replacement in a Woman with Osteogenesis Imperfecta

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The mortality rate in cardiac surgery patients with heritable generalized connective tissue disorders, such as Marfan's syndrome and osteogenesis imperfecta, is high due to tissue friability. We describe a successful open heart surgery for repair of aortic regurgitation in a woman with osteogenesis imperfecta (OI). Although tissue friability caused no problems during surgery in this case, it should be kept in mind when operating on patients with OI. (Ann Thorac Cardiovasc Surg 2002; 8: 51–53)

Key words: osteogenesis imperfecta, aortic valve replacement, tissue friability

Introduction

Osteogenesis imperfecta (OI) is a heritable disorder of the generalized connective tissue and occurs when recognized type I collagen is changed to type III collagen during biosynthesis. OI has been widely recognized to comprise bone brittleness and tissue friability, including that of the sclera, ligaments, and inner ear. Cardiovascular involvement in OI is rare compared with that of other connective tissue disorders such as Marfan's syndrome.[1)] Morbidity and mortality rates in OI patients undergoing cardiac surgery have been shown to be high.[2,3)] We describe a successful cardiac surgery for aortic regurgitation in a woman with OI.

Case

A 58-year-old woman was referred to our hospital for surgical correction of aortic regurgitation. Twenty years earlier, bilateral ossicular implantation was performed for deafness. She was first diagnosed with OI at this time. Her history was marked by multiple childhood bone fractures, but neither her parents nor other relatives showed characteristics of OI. Six years prior, she had been diagnosed with aortic regurgitation by transthoracic echocardiography. At admission, the patient's height was 142 cm and weight was 42 kg, and blue sclera was noted (Fig. 1). Her blood pressure was 170/0 mmHg and her pulse rate was 72/min. A diastolic cardiac murmur was detected clearly over a broad area of the anterior chest wall.

Chest radiography showed moderate cardiomegaly with a cardiothoracic ratio of 60%. Blood chemistry and coagulation studies were normal. Aortography and transthoracic echocardiography showed moderate aortic regurgitation but no dilatation of the aortic annulus (Fig. 2), ventricular angiography showed severe dilatation of the left ventricle (end-systolic volume index: 116 ml/m^2).

A standard median sternotomy was performed; the sternum was found to be thin and brittle. Cardiopulmonary bypass was established and aortotomy was done. Macroscopically, all three aortic valve leaflets were thickened but there was no evidence of dilatation of the aortic annulus. The aortic valve was replaced using a 21-mm St. Jude Medical prosthetic valve. Following the general practice at our institution, the prosthetic valve was buttressed with mattress sutures and pledgets using 2-0 braided polyester fiber. The patient was weaned easily from cardiopulmonary bypass, the sternum was closed with stainless steel wires. No blood products were given during the operation, but postoperative bleeding through the poststernal drainage tube occurred and was considered to be related to friable of the sternum. The patient was extubated six hours postoperatively. Postoperative blood loss was excessive (500 ml for the first eight hours). Blood products

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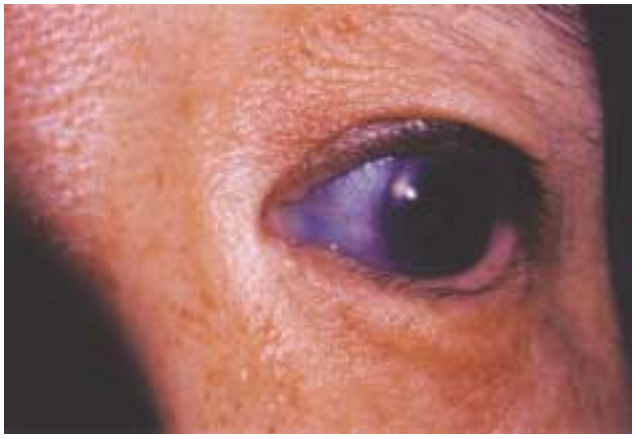

Fig. 1. Clinical photograph of blue sclera.

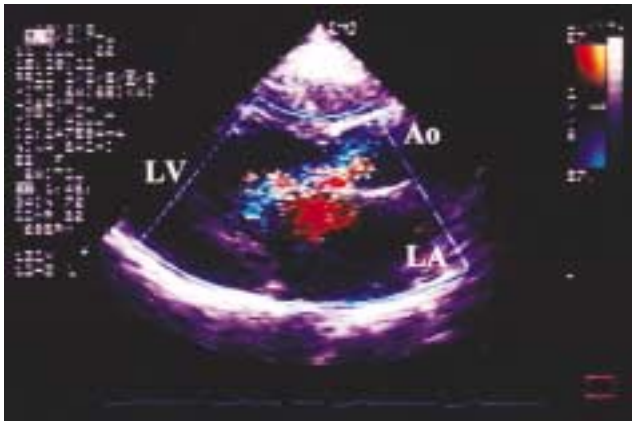

Fig. 2. Long-axis view of transthoracic echocardiography. Aortic regurgitant flow was observed in the left ventricle.

were transfused at the intensive care unit. She was discharged 14 days after surgery. Microscopic examination of the resected aortic valve showed myxoid degeneration without calcification or rheumatic degeneration (Fig. 3). One year postoperatively, she is able to carry out a normal life without dehiscence of the sternum.

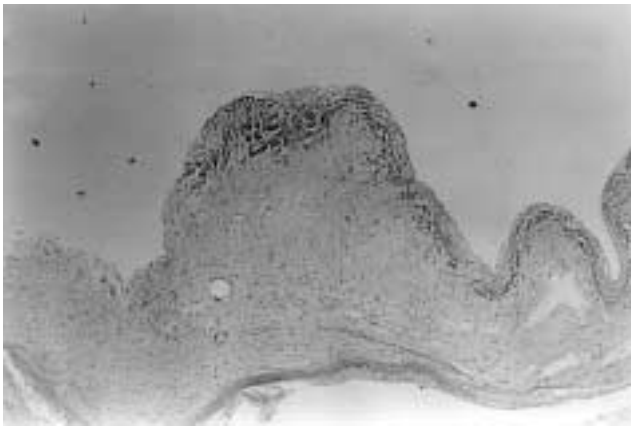

Fig. 3. Microscopic examination of the resected aortic valve showed myxoid degeneration without calcification or rheumatic degeneration.

Comment

OI is a heritable disorder of generalized connective tissues and the prevalence of OI is similar to that of Marfan's syndrome. Cardiovascular involvement in OI is rare compared with that of Marfan's syndrome.[1)] Cardiovascular surgery has been reported in 31 patients with OI including our case, according to our survey of the English-language literature.[2-7)]

In Japan seven cases have been reported (Table). The number of male OI patient who underwent cardiac surgery is 28 patients. Including the present patient, only three female OI patients who have undergone cardiac surgery have been reported worldwide.[2,3)] To our knowledge, our case is the first reported of open heart surgery for a woman with OI in Japan (Table). There should be no difference in the prevalence of OI between men and women; inheritance in patients with nonlethal OI is autosomal dominant. Pyritz and Levin have suggested that aortic dilatation is frequent in male OI patients and that it might be associated with aortic regurgitation.[8)] Hortop et al., however, reported no difference in the frequency of aortic dilatation between male and female OI patients.[1)] If there is a difference, in the number of males who undergo cardiac surgery versus the number of females, the reason is unclear.

The mortality rate in cardiac surgery patients with heritable generalized connective tissue disorders, such as Marfan's syndrome and osteogenesis imperfecta, is high. Six of the 31 OI patients, three of the seven cases in Japan, who underwent cardiac surgery died postoperatively from hemorrhagic complications considered to be related to tissue friability.[2-7)] However, in an OI patient with tissue friability who underwent ascending aorta replacement due to aortic dissection, hemostasis was achieved normally.[2)] Although coagulation studies offer no evidence that all OI patients are prone to hemorrhagic complications, prevention of tissue injury is nevertheless essential to avoid excessive bleeding during cardiovascular surgery. A patient reported by Wong et al. who underwent valve replacement required no perioperative blood transfusion, because the tissue was treated gently during surgery.[3)] Closure of the sternum with sternal bands[5)] and cardiac surgery via ministernotomy[7)] to prevent chest trauma have been reported recently. Paravalvular leakage developing

Table. Reported cases of cardiovascular operation in patients with osteogenesis imperfecta in Japan

Author (year)	Age	Sex	Coagulation study	Diagnosis	Surgical procedure	Tissue friability	Hemostasis	Result
Nakamura[8] (1987)	49	M	Normal	AR MR	AVR MVR	N.S.	Difficult	Dead (3POD)
Uemura[9] (1991)	34	M	Normal	MR	MVR	N.S.	Normal	Alive
Ohteki[10] (1991)	23	M	N.S.	AAE	Aortic root reconstruction	Yes	Difficult	Dead (8POD)
Moriyama[2] (1995)	32	M	N.S.	Aortic dissection	Ascending ao replacement	Yes	Normal	Alive
Ichikawa[4] (1996)	62	M	Normal	AR MR	AVR MVR	No	N.S.	Dead (3POD)
Maekawa[6] (1997)	54	M	Normal	AAE MR	Bentall MVR	N.S.	Normal	Alive
Our case	58	F	Normal	AR	AVR	Yes	Normal	Alive

M: male, F: female, AR: aortic regurgitation, MR: mitral regurgitation, AVR: aortic valve replacement, MVR: mitral valve replacement, AAE: annulo-aortic ectasia, N.S.: not stated.

after double valve replacement has been reported; therefore we consider it necessary to reinforce mattress suture lines with pledgets so that valvular dehiscence does not occur after cardiac valve replacement.[3] Mortality after cardiac surgery in OI patient may decrease if surgical treatment is done gently and surely, and tissue friability is kept in mind.

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