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AORTIC VALVE LESIONS ASSOCIATED WITH OSTEOGENESIS IMPERFECTA

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This case report concerns aortic stenosis and insufficiency in a young woman with osteogenesis imperfecta. A recent article by Criscitiello *et al* brought attention to the possibility that these lesions might be more than coincidentally related. That the Marfan syndrome, the Ehlers-Danlos syndrome, the Hurler syndrome and pseudo-xanthoma elasticum are known to involve significant cardiovascular abnormalities has been reasonably established. Osteogenesis imperfecta is the fifth condition included by McKusick under the classification of "heritable disorders of connective tissue" and does not seem to be frequently associated with cardiovascular abnormalities. Therefore, a review of the cases of osteogenesis imperfecta at Henry Ford Hospital was made in a search for any significant cardiovascular abnormalities. The only case of an aortic valve lesion in the 24 patients with osteogenesis imperfecta, over a 26-year span from 1939 to 1965, is described below.

CASE REPORT

L. D. (HFH 1055067), a 27-year old white female, was referred to Henry Ford Hospital in February 1962 for evaluation of heart murmurs. A heart murmur had first been heard during her first pregnancy at age 21. No symptoms occurred until her fifth pregnancy when she developed shortness of breath and pedal edema requiring digitalis therapy. At this time, a premature child was born at seven months of gestation and subsequently died. L. D. is gravida 6, para 5, ab 2, premature 1. One term-child died of congenital heart disease. Her latest symptoms just prior to her visit to Henry Ford Hospital included several episodes of exertional faintness and vertigo without syncope or chest pain. There was no history of rheumatic fever, sore throats, growing pains, epistaxis or hearing difficulty. She had scarlet fever before the age of five. In her childhood she sustained five fractures, one of each elbow and each ankle as well as one vertebra. The elbow fractures required open reduction at age 11. She has always had blue sclerae. Two of her living children have osteogenesis imperfecta and one is suspect. Neither of her two deceased children was known to have the disease.

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Her father, two paternal aunts and paternal grandmother all had osteogenesis imperfecta. The patient has no siblings.

Examination indicated her to be a well-developed, well-nourished white female, 66½" tall, weighing 170 pounds, with a blood pressure of 100/60 mm Hg and a regular pulse rate of 76 per minute, Corrigan in nature. She was a normocephalic, had blue sclerae and no evidence of doliochostenomelia. A systolic thrill was felt over both carotids, the suprasternal notch and the aortic area. The PMI was 10 cm to the left of the midsternal line in the fifth intercostal space. There was a harsh Grade III ejection-type systolic murmur at the aortic focus radiating into the neck. A loud early diastolic blowing murmur was heard best at the lower left sternal border. The second aortic sound was diminished but present. No signs of congestive failure were present.

The VDRL was nonreactive. Chest fluoroscopy revealed calcification in the aortic valve, slight left ventricular enlargement and prominence of the ascending aorta. Lung fields were clear. Electrocardiograms indicated early left ventricular hypertrophy. Right and left cardiac catheterization indicated mild aortic stenosis and high end-diastolic left ventricular pressure. Aortic insufficiency was suggested by clinical evaluation and left ventricular end-diastolic pressure of 17 mm Hg.

Clinically, electrocardiographically and radiographically she has been stable in the past three years.

DISCUSSION

To assume that the aortic valve lesion in this case is a reflection of the connective tissue defect due to osteogenesis imperfecta would be pure speculation. Calcification of the valve combined with a history of scarlet fever would make one suspect rheumatic etiology.

Criscitiello *et al* recently presented three cases of cardiac abnormalities with osteogenesis imperfecta. Two had aortic insufficiency with no other suggestion of etiology than a heritable connective tissue disorder. The third had several lesions, including a bicuspid aortic valve with a torn cusp and a fenestration of the non-coronary cusp thought to have produced the aortic insufficiency. Other lesions were fenestrations of the pulmonic valve and an aneurysmal deformity of the mitral valve. The aortic valve required surgical replacement, but the patient died. At autopsy, focal degenerative changes were seen between the reduced number of elastic lamellae of the aorta and pulmonary artery. Accumulations of acid mucopolysaccharide were detected in these areas. Changes were consistent with cystic medionecrosis. The aortic valve leaflets were consistent with a healed or healing valvulitis. (There had

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been a previous bout of SBE). No stigmata of rheumatic heart disease were seen in the myocardium.

Criscitiello's speculation was that it would be highly likely that lesions basically related to the underlying tissue defect of osteogenesis imperfecta had gone unrecognized and had been classified under other categories.

Between 1939 and 1965 twenty-four cases of osteogenesis imperfecta have been indexed at the Henry Ford Hospital. These were reviewed. The only significant cardiac abnormality found was in the present case presentation.

RESULTS OF RIGHT AND LEFT ATRIAL PERCUTANEOUS CARDIAC CATHETERIZATION

	Pressure		Vol. %	% Saturation
	Syst./Diast.	Mean		
Left Atrium		13 mm Hg		
Left Ventricle	145/8-11-17			
Aorta	135/70			
Right Atrium		5		
Right Ventricle	25/4-6			
Pulmonary Artery	24/13		9.7	71
Femoral Artery			12/7	93
Cardiac Output	7.8 L/min			
Cardiac Index	4.0 L/min			

REFERENCES

1. McKusick, V. A.: *Heritable Disorders of Connective Tissue*. 2nd ed. St. Louis, C. V. Mosby Co., 1960.
2. Criscitiello, M. G., Ronan, J. A., Besterman, E. M. M., and Schoenwetter, W.: Cardiovascular abnormalities in osteogenesis imperfecta. *Circulation* 21:255-262, Feb. 1965.

