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대동맥판 역류를 동반한 파브리병 환자 1례

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A Case of Fabry's Disease with Aortic Regurgitation

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Fabry's disease is an X- linked glycosphingolipid disorder caused by deficient activity of the enzyme α-galactosidase A, which results in the systemic accumulation of globotriosylceramide (Gb3) in all tissues of the body, especially the skin, cornea, kidney, and heart. Vascular involvement of the heart leads to an early death in males with Fabry's disease, and aortic regurgitation (AR) is common in patients of both sexes having the disease.A 31- year- old man with Fabry's disease who had been treated with recombinant α- galactosidase ERT for 19 months was admitted to evaluate exertional dyspnea. The cardiac workup revealed left ventricular hypertrophy (LVH), increased LV size, and moderate to severe AR. He underwent mechanical valvular replacement and a heart biopsy. The histologic changes showed widespread hypertrophy and vacuolization of cardiac myocytes, and electron microscopy revealed a loss of myelin bodies in the cardiac myocytes. In treating Fabry's disease, enzyme replacement therapy (ERT) has become the standard medical care. ERT has beneficial effects on renal function, heart size and function, but early diagnosis and hormone replacement are essential. ERT should be commenced before organ function is irreversibly impaired. We present a patient with Fabry's disease who underwent aortic valvular replacement operation while on ERT.

Key Words : 파브리 병, 대동맥판 역류, 효소대체 치료
Fabry's disease, Aortic regurgitation, Enzyme replacement