Enzyme Replacement Therapy Provides Effective, Long-Term Treatment of Cardiomyopathy in Pompe Disease

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Figure. (A) Echocardiographic features and (B) time course following initiation of enzyme replacement therapy (ERT) in an 8-month-old male patient with Pompe disease. (A) Left ventricular hypertrophy (i) prior to and (ii) showing marked improvement after 10 years of ERT. (B) Time course of echocardiography and laboratory data after initiation of ERT. BNP, brain natriuretic peptide; CK, creatine kinase; LA, left atrium; LV, left ventricle; LVM, left ventricular mass; LVMI, left ventricular mass index.
Pompe disease (PD) is a lethal disorder caused by a deficiency of the lysosomal enzyme acid α-glucosidase. The enzyme deficiency results in hypertrophic cardiomyopathy, respiratory muscle weakness, and subsequently, death, usually within 1 year of birth. Enzyme replacement therapy (ERT) dramatically improves prognosis, and the majority of patients will, therefore, be treated in adulthood within a decade. Previous reports document the efficacy of ERT over a period of 1–2 years, but long-term efficacy data are not yet available.

Here, we report on the clinical course of a male patient, who received ERT for the longest period in Japan, after being diagnosed with PD at 6 months old. Hypertrophic cardiomyopathy was already evident at the time of diagnosis. ERT was initiated when the patient was 8 months old. Remarkable improvements in left ventricular mass (LVM), LVM index (LVMI), and LVMI Z-score were seen in the first 3 years, and gradual improvement continued thereafter for >10 years (Figure). Sequential improvements were also seen in brain natriuretic peptide and creatine kinase.

Given that many pediatric PD patients currently receiving ERT will reach adulthood within a decade, it will be important for cardiologists specializing in adult patients to also understand the time course of this disease.

Disclosures
The authors declare no conflicts of interest.

Reference