Biventricular hypertrophic cardiomyopathy in a baby diagnosed with Pompe disease

Pompe hastalığı olan bebekte biventriküler hipertrofik kardiyomiyopati

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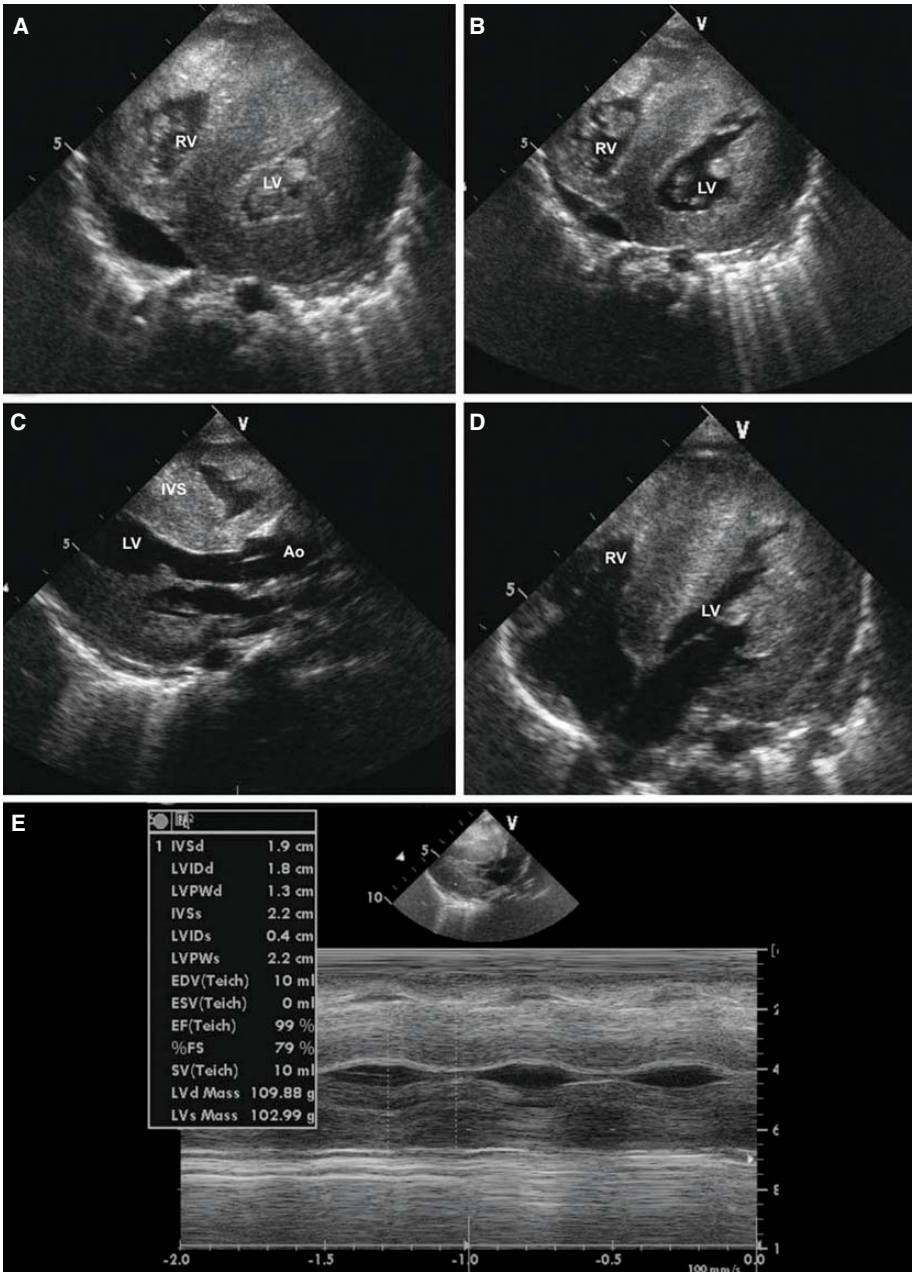
A four-month-old baby was admitted with respiratory distress and difficulty with feeding. She was born at term with a birth weight of 4000 g and had no symptoms after birth. She was the third child of consanguineous parents. She had hypotonia, a grade 2/6 systolic murmur, tachycardia and mild hepatomegaly. Electrocardiography showed a short PR interval, and biventricular hypertrophy. Echocardiography demonstrated a severe form of biventricular hypertrophy, so that both ventricular cavities were nearly obliterated (Figs. and Video*). The left ventricular mass index (389 g/m^2, Reference Range (RR); 70-75 g/m^2) and Z score (+12) were significantly increased. Mild left ventricular outflow tract obstruction was also detected. Serum creatine kinase, creatine kinase-MB, lactate dehydrogenase and alanine aminotranspherase levels were elevated: A pro-B-type natriuretic peptide level was markedly increased (4526 pg/ml RR; 0-100 pg/ml). Dried blood spot analysis revealed no detectable acid alpha glycosidase activity, so the baby was diagnosed with Pompe disease.



Figures– Echocardiography revealed a severe form of biventricular hypertrophic cardiomyopathy. Echocardiographic images obtained at the ventricular-short axis view during the systolic **(A)** and diastolic **(B)** phases of the cardiac cycles, parasternal long axis **(C)** and an apical four chamber **(D)** view. **(E)** M-mode echocardiography also showed marked septal hypertrophy. **Supplementary video files associated with this case can be found in the online version.* Ao: Aorta; IVS: Interventricular septum; LV: Left ventricle; RV: Right ventricle.

The patient was managed with a beta-blocker and enzyme replacement therapy (Myozyme®, Genzyme Corporation, Cambridge, MA, US, 20 mg/kg infusion every 2 weeks). We observed clinical improvement at short term follow up. Despite multidisciplinary and intensive care management, she unfortunately died due to aspiration pneumonia after two months of therapy.