

Newborn Screening, Inborn Errors of Metabolism

AB073. Classic infantile-onset Pompe disease: phenotypes and outcomes of 5 Vietnamese patients receiving enzyme replacement therapy

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Background: Pompe disease (PD) or glycogen storage disease type II is a lysosomal storage disorder, caused by mutations of *GAA* gene which results in deficiency of acid alpha-glucosidase (GAA) enzyme that involves in metabolism of glycogen in the lysosomes. Its incidence is 1/14,000–1/100,000. PD is divided into three types: classic infantile onset, non-classic infantile onset, and late onset. Early enzyme replacement therapy (ERT) before developing respiratory distress may lead to good outcome. Since 2013, we have identified 16 cases with classic infantile-onset and 5 cases were treated with ERT. Herein, we describe phenotypes and outcomes of five infantile-onset PD patients who received ERT.

Methods: GAA enzyme assay was done at National Taiwan University Hospital.

Results: Ages of diagnosis were 12, 38 and 70 days, 5 and 9 months of age. Clinical presentations included macroglossia (5/5), hypertrophic cardiomyopathy (5/5), failure to thrive (5/5), facial weakness and hypotonia (3 patients diagnosed after 70 days of age), respiratory failure (1 patient diagnosed at 9 months of age). All patients had mildly elevated plasma CK (270–380 UI/L) and transaminase (60–260 UI/L). Ages at starting ERT were 28 and 58 days, 3, 6 and 10 months. The time intervals from diagnosis to starting ERT were between 14 days and 1 month. The durations of ERT were 4–22 months. The outcomes were good. All patients had improvement of cardiac functions shown on echocardiography, respiratory status, and motor development. The patient who first received ERT at 10 months of age was reportedly dead at home due to food obstruction at 18 months of age. Current ages of the survivors were 5–24 months.

Conclusions: Patients with classic infantile-onset PD will have good outcomes if ERT is started early. Newborn screening for this disease is necessary to yield an early diagnosis.

Keywords: Pompe disease (PD); glycogen storage disease type II; enzyme replacement therapy (ERT)

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