

## Newborn Screening, Inborn Errors of Metabolism

## AB028. Very early treatment for infantile-onset Pompe disease contributes to better outcomes: 10-year experience in one institute

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**Background:** To evaluate the outcomes of very early treatment in our patients and compare these data with other infantile-onset Pompe disease (IOPD) cohort studies.

**Methods:** In this nationwide program, one million newborns were screened for Pompe disease. We diagnosed IOPD in 21 of these newborns, and all were treated and followed in our hospital.

**Results:** After 2010, the mean age at first enzyme-replacement therapy (ERT) was 10 days. Our patients had better biological, physical, and developmental outcomes and lower anti-rh acid alpha-glucosidase antibodies after 2 years of treatment, even compared with one group that began ERT just 10 days later than our cohort. No patient had a hearing disorder or abnormal vision. The mean age for independent walking was 11.6±1.3 months, the same age as normal children.

**Conclusions:** ERT for patients with IOPD should be initiated as early as possible before irreversible damage occurs. Our results indicate that early identification of patients with IOPD allows for the very early initiation of ERT. Starting ERT even a few days earlier may lead to better patient outcomes.

**Keywords:** Infantile-onset Pompe disease (IOPD); enzyme-replacement therapy (ERT); newborn Screening

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