

Poster Abstract: Clinical

Motor Development in Infantile Pompe Disease

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BACKGROUND

Since 2006, ERT with alglucosidase alfa is available for infantile Pompe disease. The standard dosage for infantile patients is 20 mg/kg every other week. As the heart responds rapidly to treatment, the progress of motor development determines the response to treatment. Six infantile Pompe patients with different points of diagnosis and therapy initiation were treated in our centre. The assessment of milestones and motor skills have been evaluated using the Alberta Infant Motor Scale (AIMS).

CASE REPORTS

Patient 1: CRIM-positive. First infusion at 21 weeks postnatally (within the phase III study). At 9 months of age: 9 points of AIMS. Motor development improved with ERT. At the age of 4 years, she remained at 39 points.

Patient 2: CRIM-negative. Second child of consanguineous parents with Turkish ancestry (first child passed away aged 10 months due to infantile Pompe disease).

First infusion at the 5th day of life: AIMS remained age appropriate from birth until the age of 3 years.

Patient 3: CRIM-negative. First child of consanguineous parents with Turkish ancestry. First symptoms occurred at 2 months of age, with muscular hypotonia, dyspnea, failure-to-thrive, dysphagia due to weakness and cardiomegaly. First infusion at 10 weeks postnatally. We administered 20 mg/kg/very week until 24 weeks postnatally to gain a better response of cardiac muscle; thereafter, we switched to biweekly applications of the enzyme. At 2 months of

age: 1 point; thereafter, fast catch-up of motor function. At the age of 16 months, she gained 58 points.

Patient 4: CRIM-negative. First child of non-consanguineous parents. First infusion at 6 months of age; thereafter, fast catch-up of motor function, but with high antibody titers. She rapidly lost motor function, became ventilator dependent and lived until 2 years of age.

Patient 5: CRIM-positive. Second child of non-consanguineous parents. First infusion at 5 months of age; thereafter, fast catch-up of motor function, but not yet walking.

Patient 6: CRIM-negative. Second child of consanguineous parents with Turkish ancestry. First infusion at 10 months of age, slight catch-up of motor function for 5 weeks, but thereafter, with high antibody titers, rapid deterioration and death due to cardiac arrest at 14 months of age.

RESULTS

Patients 1 and 5 are wheelchair-dependent and have not reached all motor milestones. Patient 2 achieved all age-appropriate motor milestones. Patient 3 caught up with the motor development of unaffected coeval infants within 1 year.

CONCLUSIONS

The reported cases clearly illustrate the different progress of the classic infantile Pompe disease, depending on the time of initiation of ERT, pre-existing irreversible pathology, and high antibody titers. For the best therapeutic outcome, it is crucial that the disease is diagnosed as early as possible before irreversible tissue damage occurs. Further studies are needed to determine the impact of antibody formation and CRIM status on the clinical efficacy.

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