Journal of Neuromuscular Diseases 1 (2015) S58–S59
DOI 10.3233/JND-159051
IOS Press

Poster Abstract: Therapeutic

Infantile-Onset Pompe Disease: The Care Beyond the Cure

Guja Astrea[1,*], Silvia Perazza[1], Silvia Frosini[1], Elena Moretti[1], Michele Sacchini[2], Eleonora Dati[1], Chiara Pecini[1], Elena Procopio[2], Filippo M. Santorelli[1], Maria A. Donati[2] and Roberta Battini[1]
[1]*Department of Developmental Neuroscience, IRCCS Fondazione Stella Maris, Calambrone, Italy*
[2]*Metabolic and Muscular Unit, Neuroscience Department, Meyer Children's Hospital, Florence, Italy*

BACKGROUND

In classic infantile-onset Pompe disease (IOPD), symptoms start early in life, and there is a fatal outcome if the disease remains untreated.[1] Enzyme replacement therapy (ERT) with recombinant human GAA remains the single treatment favouring a prolonged life expectancy.[2–4] To our knowledge, there have been no studies investigating the role of an early and intensive global rehabilitation programme in IPOD infants treated with ERT. We present an 18-month neurorehabilitation follow-up in a child.

PATIENT AND METHODS

An Italian girl with a mild form of IPOD presented psychomotor delay with tongue protrusion in her first months of life. In consideration of the high serum CK and transaminase levels and a severe hypertrophic cardiomyopathy, a biochemical/molecular diagnosis of GAA deficiency was made at 12 months of age, and the girl was immediately put on ERT treatment.

When we first examined the girl, she presented with facial muscle weakness (she had an expressionless face, open mouth with tongue protrusion and everted lower lip), delayed weaning with feeding difficulties for semi-solid foods, and generalized muscle weakness. She could roll over, could sit, if aided, and could maintain the sitting position for short time, although with progressive dorsal kyphosis and cervical lordosis.

We started the child on a personalized rehabilitation programme to promote gross- and oral-motor abilities and to monitor and prevent early signs of dysphagia. Active and assisted selective movements were facilitated, promoting self-postural changes. Also, we adopted a hip–knee–ankle–foot orthosis with unlock at the hip and knee junctions to facilitate acquisition of vertical skills for a greater autonomy and better quality of life. Meanwhile, we performed indirect work focused on both oral motor abilities and language development.

RESULTS

Despite negative predictors, such as a late ERT treatment and a protracted course of her cardiomyopathy, the patient could achieve several motor milestones, and she progressively improved her muscle power, facial expression, and tongue movements (Figure 1).

Her language remains delayed for her age; however, her expressive vocabulary is increasing, and it is mainly characterized by several homophones. Early combinations of sentences are seldom emerging. The speech intelligibility is reduced due to disordered articulation with hypernasal resonance.

CONCLUSIONS

There are emerging challenges with the prolonged survival of patients with IOPD, with a potential shift from high mortality to high morbidity, given the current limitations in response to ERT. Early access to therapy and personalized rehabilitation strategies be-

*Correspondence to: Guja Astrea, Department of Developmental Neuroscience, IRCCS Fondazione Stella Maris, Calambrone, Italy. E-mail: gastrea@fsm.unipi.it.

ISSN 2214-3599/15/$27.50

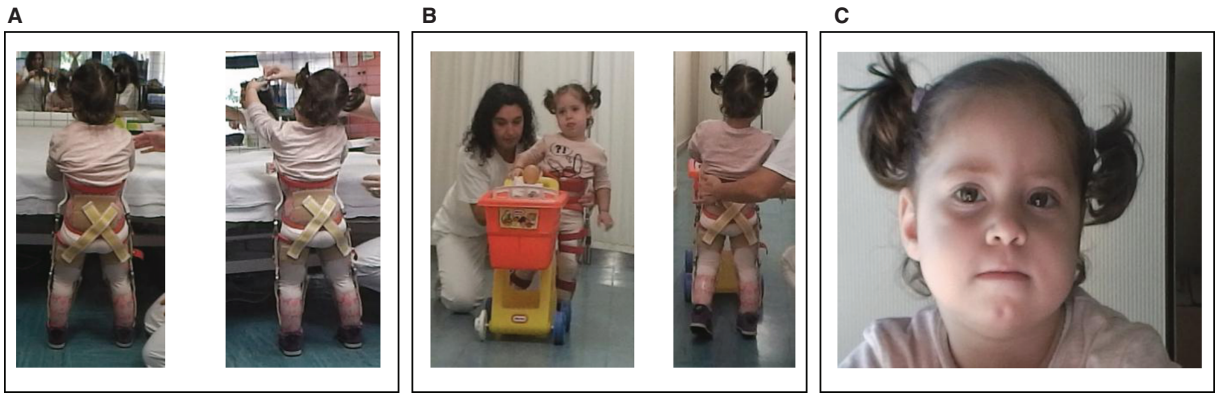

Fig. 1. Best motor performance at age 30 months. The patient had an increased endurance while standing for 40 minutes with the orthosis (A) and could walk indoors for 10 m with assistance (B). Also, her lips could be sealed, though they were still weak and drooping (C).

come the main objective to ensure best outcome and quality of life.

REFERENCES

[1] Kishnani PS, Hwu WL, Mandel H, Nicolino M, Yong F, Corzo D, et al. A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. J Pediatr 2006; 148:671-676.

[2] Van den Hout JM, Kamphoven JH, Winkel LP, Arts WF, De Klerk JB, Loonen MC, et al. Long-term intravenous treatment of Pompe disease with recombinant human alpha-glucosidase from milk. Pediatrics 2004; 113:448-457.

[3] Kishnani PS, Corzo D, Nicolino M, Byrne B, Mandel H, Hwu WL, et al. Recombinant human acid [alpha]-glucosidase: major clinical benefits in infantile-onset Pompe disease. Neurology 2007; 68:99-109.

[4] Case LE, Beckenmeyer AA, Kishnani PS. Infantile Pompe disease on ERT-Update on clinical presentation; musculoskeletal management, and exercise considerations. Am J Med Genet Part C Semin Med Genet 2012; 160C:69-79.