M19. DOES THE FREQUENCY OF ENZYMATIC REPLACEMENT THERAPY INFLUENCE PATIENTS’ LEVEL OF ADAPTIVE FUNCTIONING?

Stefania Caviglia¹, Ambra Bottari¹, Paola Bazzu¹, Federica Deodato¹, Roberta Taurisano¹, Ilaria Tondo¹
¹Bambino Gesù Children's Hospital

Background Lysosomal storage diseases (LSDs) are a group of rare inherited metabolic disorders that result from defects in lysosomal function. LSDs are generally multisystemic diseases with reduced life expectancy. Enzymatic replacement therapy (ERT) is available for some LSDs and has led to an improvement of many somatic symptoms of some metabolic disorders. To our best knowledge no study has explored the impact of ERT frequency on adaptive functioning in this unique pediatric population. We hereby present a pilot study.

Methods The parents of 27 patients with LSDs, aged between six months and 16 years (mean age 9 years 4 months), were asked to complete the Vineland Adaptive Behavior Scales (VABS) Survey Form interview to measure children’s functional outcomes within four domains: communication, daily living, socialization and motor ability. Disorders present amongst the sample included MPS (N=13; I, I H-S, I H, I S, II, IV-Morquio, VI) Gaucher (N=6; types I and III) and Pompe (N=8; infantile and late onset forms). Data collection took place during patients’ visits to the Bambino Gesu Children’s Hospital in Rome (Italy) for ERT treatment.

Results The total sample obtained significantly lower scores (<85) in the daily living domain compared to scores in other domains and the adaptive behavior composite score. Significant differences were also found for adaptive functioning across age groups, while adaptive functioning did not significantly differ between males and females. No significant result in patients’ adaptive functioning was found by comparing the sample according to diagnostic categories. The adaptive functioning of this sample was not associated with disease characteristics, such as mobility difficulties, neurosensory and neuropsychological outcomes.

When considering only children with an IQ in the normal ranges (>85; N=21), no significant differences were found comparing children’s adaptive functioning by splitting the sample into high and low ERT frequency (high=once a week; low=fortnightly). However, 58% of patients undergoing ERT once a week and 43% of patients undergoing ERT fortnightly presented with scores below the normal ranges (<85) in the adaptive behaviour.

Discussion Despite children with good cognitive capacities who did not differ significantly on level of adaptive functioning based on high or low ERT frequency, results suggest that higher frequency of treatment was associated with a higher occurrence of scores below the normal ranges in the adaptive behavior in this sample. Investigating this tendency could be of crucial importance in understanding whether lower scores in the daily living skills domain of the VABS and, more in general, maladaptive functioning might be associated with ERT frequency. Finally, a clearer understanding of the role of treatment frequency in this population’s adaptive functioning can lead to an improvement of both patients and families’ health outcomes.

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