Molecular Genetics and Metabolism Reports 6 (2016) 5–7

Contents lists available at ScienceDirect

Molecular Genetics and Metabolism Reports

journal homepage: www.elsevier.com/locate/ymgmr

Correspondence

Patient's weight can decide about spending millions on enzyme replacement therapy in MPS II

Abbreviation:
MPS
Topic:
mucopolysaccharidosis

Keywords:
Pediatric orphan disease
Enzyme replacement
Public health
Expensive therapies
Walking test

Enzyme replacement therapy in mucopolysaccharidosis type II (MPS II, Hunter disease) is extremely expensive and treatment effects are hardly predictable due to phenotypic variability [1]. We would like to address an additional problem — the paucity of robust clinical tests allowing for monitoring of effectiveness of enzymatic treatment.

Since 2009 enzyme replacement therapy with idursulfase has been offered to all Polish patients with MPS II aged >5 years, independently on disease severity. A total of 40 patients have started treatment up to date. In 26 persons treatment was eventually discontinued due to disease progression, death of the patient (two cases), or anaphylaxis (two cases). Unfortunately, reaching the consensus on treatment failure required sometimes several years of observation, as the classic clinical monitoring parameters [2,3] were not always informative. Specifically: the distance covered in the six-minute walking test could not be applied in 20 non-ambulant patients, pulmonary function tests were performed only in older patients without intellectual disability and monitoring of liver size or of heart involvement was helpful only in those patients with significant hepatomegaly or cardiac dysfunction.

Surprisingly, basic anthropometric measurements proved to be very useful for assessment of treatment effectiveness in some doubtful cases. No weight gain or even weight loss was observed in severely affected patients, contrary to those with milder disease in whom an overall anabolic effect was observed (Fig. 1). Our findings are consistent with previously published observations [4,5] reporting decline of the linear growth in untreated children with MPS II, which started at the age of four to six years.

In conclusion, we recommend thoughtful use of growth charts, as it can help to avoid wasting millions for prolonged enzymatic therapy in some cases of MPS II.

Contributors' statements

Miroslaw Bik-Multanowski analyzed data and drafted the manuscript. All authors contributed to data collection, reviewed and revised the manuscript, and approved the final manuscript as submitted.

Conflicts of interest

The authors declare no conflicts of interests.

Financial disclosure statement

The authors state that they have no financial relationships that could be relevant for the work.

References

[1] M. Scarpa, Z. Almassy, M. Beck, et al., Mucopolysaccharidosis type II: European recommendations for the diagnosis and multidisciplinary management of a rare disease, Orphanet J. Rare Dis. 6 (2011) 72.
[2] J. Muenzer, J.E. Wraith, M. Beck, et al., A phase II/III clinical study of enzyme replacement therapy with idursulfase in mucopolysaccharidosis II (Hunter syndrome), Genet. Med. 8 (2006) 465–473.
[3] R. Tomanin, A. Zanetti, F. D'Avanzo, et al., Clinical efficacy of Enzyme Replacement Therapy in paediatric Hunter patients, an independent study of 3.5 years, Orphanet J. Rare Dis. 9 (2014) 129.
[4] I.D. Young, P.S. Harper, R.G. Newcombe, I.M. Archer, A clinical and genetic study of Hunter's syndrome. 2 Differences between the mild and severe forms, J. Med. Genet. 19 (1982) 408–411.
[5] P. Patel, Y. Suzuki, M. Maeda, et al., Growth charts for patients with Hunter syndrome, Mol. Genet. Metab. Rep. 1 (2014) 5–18.

Miroslaw Bik-Multanowski*
Department of Medical Genetics, Jagiellonian University, Medical College, Krakow, Poland
Corresponding author at: Department of Medical Genetics, Jagiellonian University, Medical College, Ul. Wielicka 265, 30-663 Krakow, Poland.
E-mail address: miroslaw.bik-multanowski@uj.edu.pl

Ewa Jamroz
School of Medicine in Katowice, Department of Paediatrics and Developmental Age Neurology, Medical University of Silesia, Katowice, Poland

Lukasz Kaluzny
Department of Pediatric Gastroenterology and Metabolism, Medical University, Poznan, Poland

http://dx.doi.org/10.1016/j.ymgmr.2015.12.002

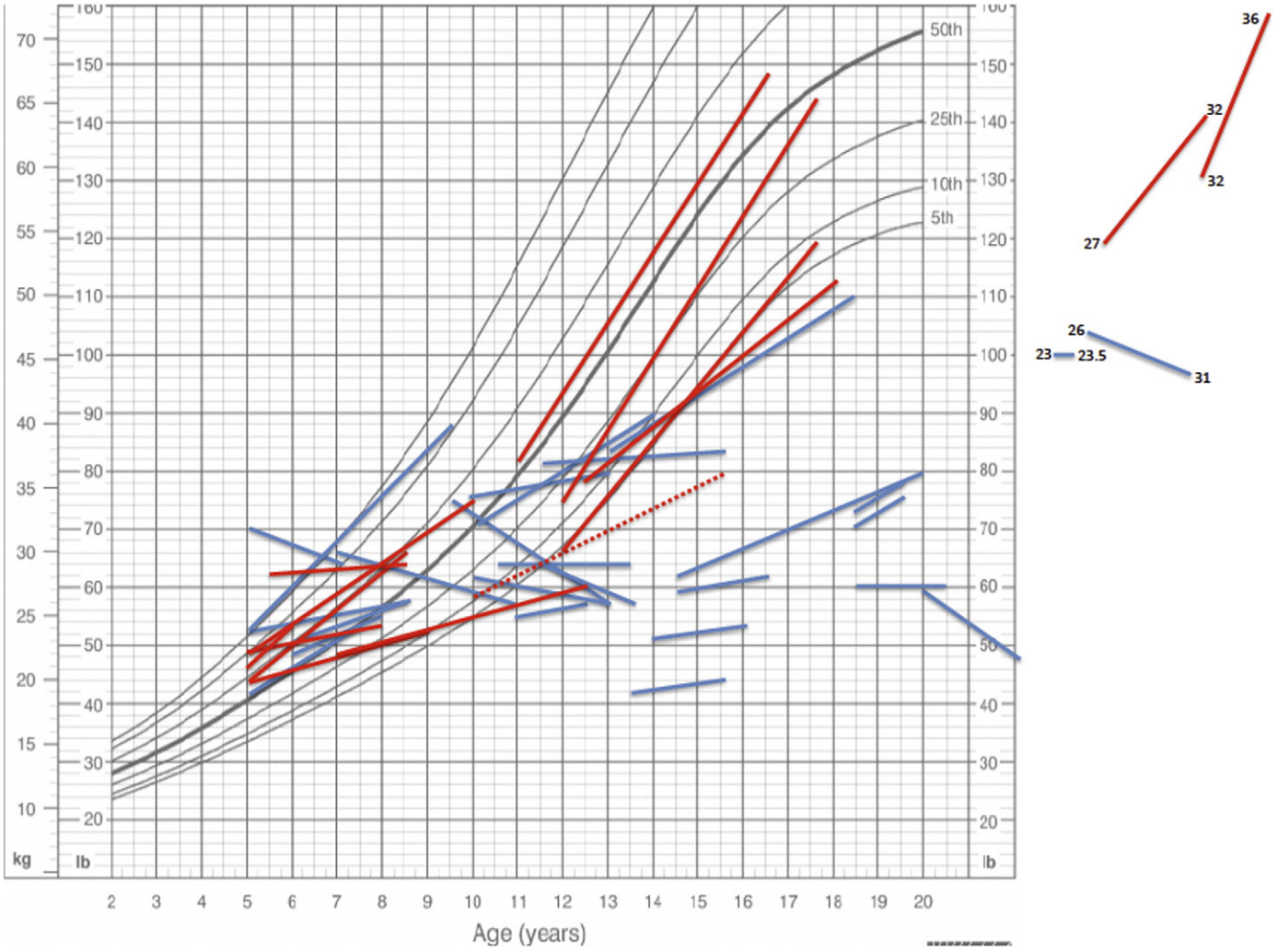

Fig. 1. Body weight and growth changes in studied patients with MPS II. A single female patient was marked with a dotted line. (CDC growth charts for boys; developed by the National Center for Health Statistics in collaboration with the National Center for Chronic Disease Prevention and Health Promotion, 2000).

Ewa Pronicka
Dariusz Rokicki
Department of Pediatrics, Nutrition and Metabolic Diseases, The Children's Memorial Health Institute, Warszawa, Poland

Ewa Starostecka
Metabolic Department, Polish Mother's Health Memorial Institute, Lodz, Poland

Jolanta Sykut-Cegielska
Screening Department, Institute of Mother and Child, Warszawa, Poland

Mieczyslaw Walczak
Department of Paediatric Endocrinology and Diabetology, Pomeranian Medical University in Szczecin, Poland

6 December 2015

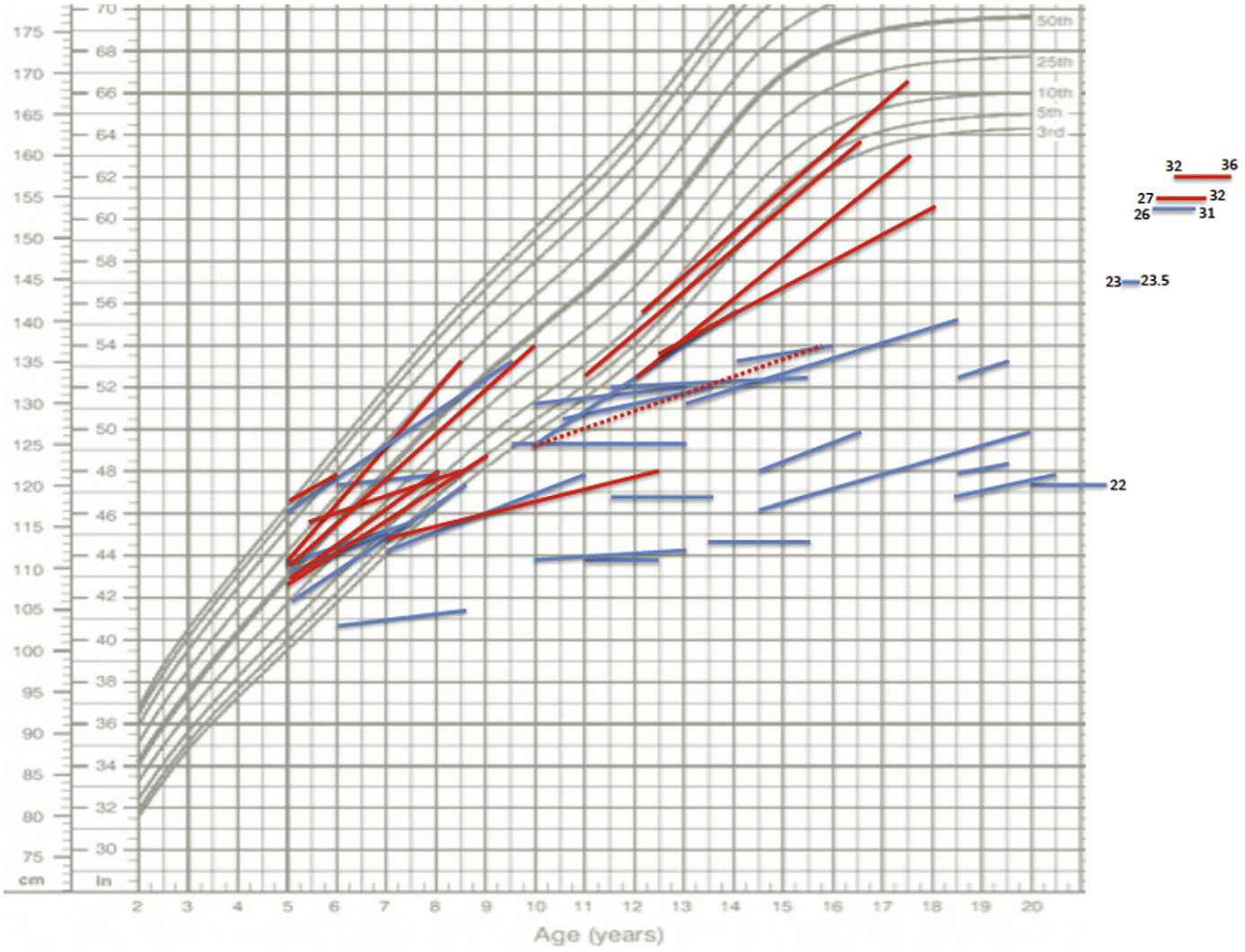

Fig. 1 (*continued*).