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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

Mirosław 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.

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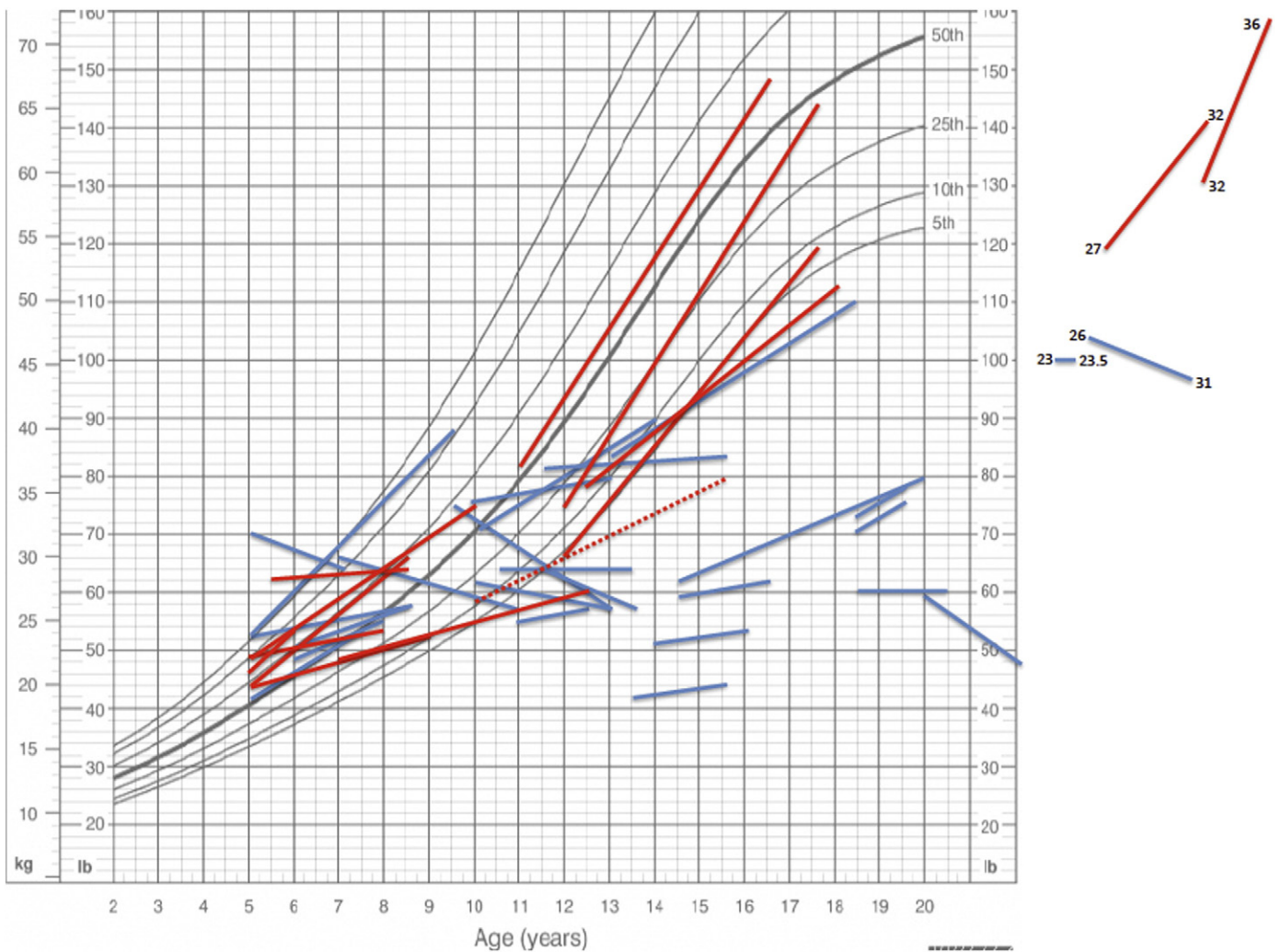


Fig. 1. Body weight and growth changes in studied patients with MPS II. A single female patient was marked with a dotted line. (CDC growth charts for boys; developed by the National Center for Health Statistics in collaboration with the National Center for Chronic Disease Prevention and Health Promotion,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

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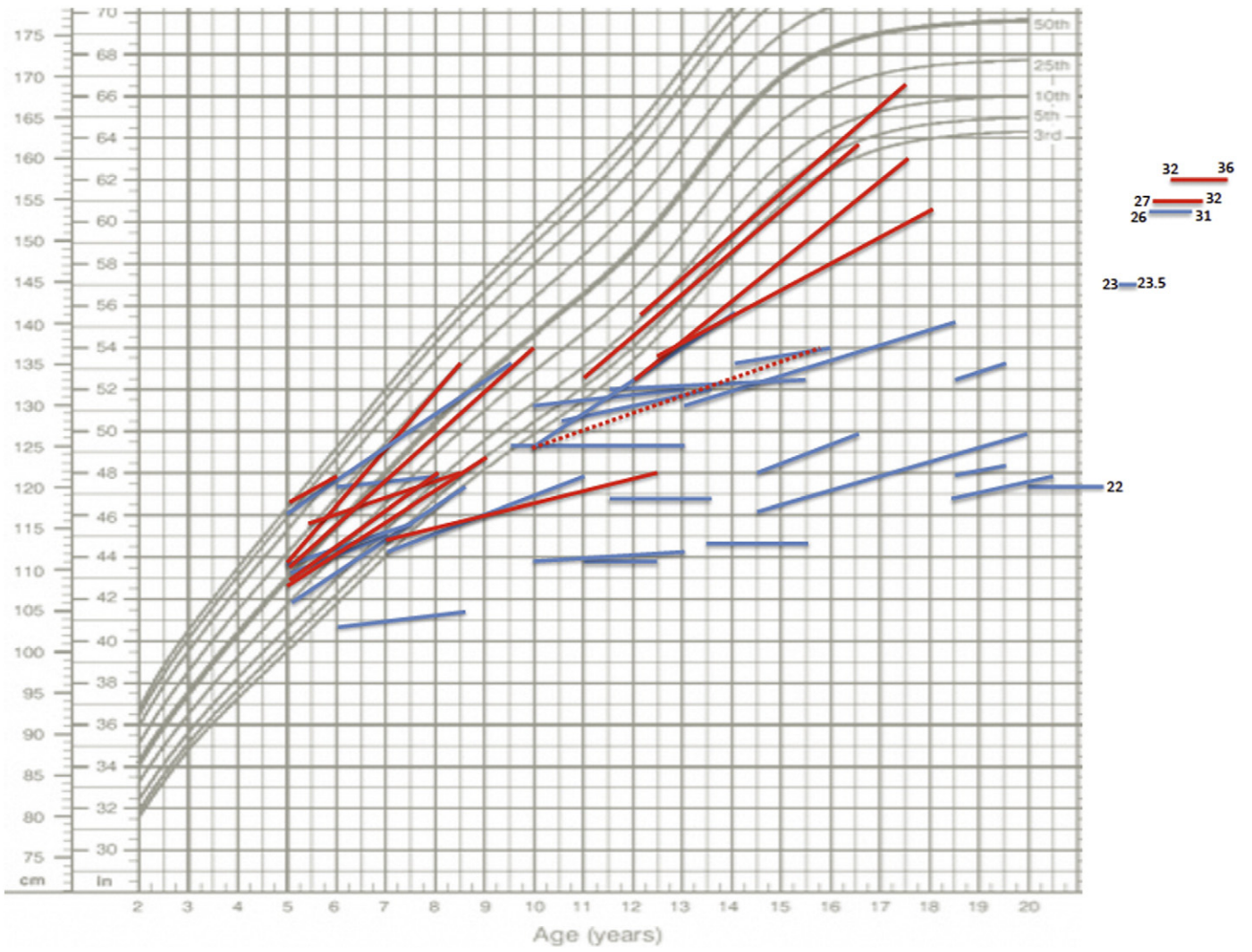


Fig. 1 (continued).