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Endoped Abstract

Laron Syndrome—Possibilities of Diagnoses, Treatment, and Outcome

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Background

Laron syndrome (growth hormone insensitivity) is a rare autosomal recessive condition characterized by severe short stature with postnatal onset usually [1], particular facial features and hypoglycemia in childhood, and type 2 diabetes mellitus, dyslipidemia, metabolic syndrome, and obesity in adulthood [2], caused by growth hormone receptor (GHR) anomalies (insensitivity). It is estimated that there are 350 people with this condition worldwide [3]. The growth hormone receptor mutation prevents the stimulating effects of growth hormone, inducing insulin-like growth factor-1 (IGF1) deficiency. Treatment with recombinant IGF1, the only treatment option which can improve final height [4], is not available for all patients.

Aim

Present the clinical outcomes of a Laron syndrome patient untreated and on hormonal substitution treatment.

Case Report

A 18-year-old girl with Laron syndrome diagnosed at the age of 2 years and 10 months, based on the clinical feature of severe short stature (height—63 cm; −8.24 SDS, standard deviation score), particular facial features (protruding forehead, saddle nose, large eyes, sparse and thin silky hair, and high-pitched voice), small hands and feet, and sweating and hypotonic episodes, especially in the morning. The endocrine growth axis assessment revealed an elevated growth hormone concentration (>40 ng/mL; 0–8 ng/mL) and very low serum IGF-1 levels (<25 ng/mL; NV = 75–175), respectively, and delayed bone age (9 months—Greulich

and Pyle). The genetic assay showed a homozygous pathological variant (c476T > A L141X) in the 6 exon of the GHR gene. The growth velocity without hormonal substitution was low at 0.23 cm/month (14.5 cm/63 months). After 15 months (between 8 years 1 month and 9 years 4 months) of IGF1 (mecasermin) treatment, the growth velocity increased to 0.53 cm/month, the girl's height increased from 77.5 cm (−8.55 SDS) to 85.6 cm (−7.94 SDS). After IGF1 treatment discontinuation, height velocity fell to 0.22 cm/month. At the age of 17 years and 2 months, hormone replacement therapy with mecasermin was resumed up to 18 years. The patient also had associated pubertal delay; thelarche started at the age of 15 and progressed slowly. The final height of the patient was 117 cm. In addition, she has developed focal epilepsy, obesity, and depression.

Conclusions

Early diagnosis and hormone substitution therapy may reduce the clinical consequences of complications, improving the prognosis.

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