ESPE Abstracts (2014) **82** P-D-3-3-839

A Rare Syndrome Benefits from GH Therapy: Hypotonia–Cystinuria Syndrome

Onur Akin, Bulent Hacihamdioğlu & Ediz Yeşilkaya

Gulhane Military Medical Academy, Ankara, Turkey

Background: Hypotonia–Cystinuria syndrome (HCS), characterized by hypotonia at birth, poor feeding and growth retardation is an autosomal recessive disorder caused by homozygous microdeletions of PREPL and SCL3A genes. Increase in the urinary amino acids including cystine, lysine, arginine, and ornithine is the main laboratory finding.

Objective and hypotheses: HCS was described in 17 patients so far and previous reports with favorable responses to GH treatment are present. Herein, we report a case with HCS who were successfully treated with GH.

Method: A 8-year-old girl was referred to Pediatric Endocrinology outpatient department because of short stature. She was born at 38 weeks after a normal pregnancy and her birth weight was 2400 g. She was evaluated by Department of Pediatric Metabolism for poor feeding and hypotonia when she was 2 months old and her urinary levels of cystine and dibasic amino acids were found to be increased. Diagnosis of HCS was confirmed by detection of SLC3A/PREPL deletions. Hypotonia and poor feeding has improved by age. Physical examination revealed mild facial dysmorphism, proximal weakness, nasal speech, and mild learning disability along with short stature. Her height was 112.5 cm (SDS −3.47). Her serum IGF1 level was 37 ng/ml (SDS −2.9). Peak GH levels after provocation tests were 1.1 and 1.7 ng/dl.

Results: GH treatment (0.2 mg/kg per week) was initiated. Her height reached to 125.9 cm (SDS −1.72, growth velocity: 13.4 cm/year) after 1-year treatment.

Conclusion: The clinical findings of HCS may resemble to Prader–Willi Syndrome (PWS) in terms of poor feeding, hypotonia at birth improving by age and gradually increasing risk of obesity. It should be considered in the differential diagnosis of patients who present symptoms similar to PWS. GH replacement therapy for short stature is a well established treatment in patients with HCS.