P2-948

Kocher-Debre-Semelaigne syndrome: hypothyroidism with muscle pseudohypertrophy.

Elena Bogova[1], Deryagina Alesya[1], Shyryaeva Tatiana[1], Anatoly Tulpakov[1]

Institute of Pediatric Endocrinology, Endocrinology Research Centre, Moscow, Russia

The authors declare that there are no conflicts of interests associated with this manuscript

Background

Kocher-Debre Semelaigne syndrome (KDSS) is a rare form of myopathy in patients with longstanding moderate-to-severe hypothyroidism.

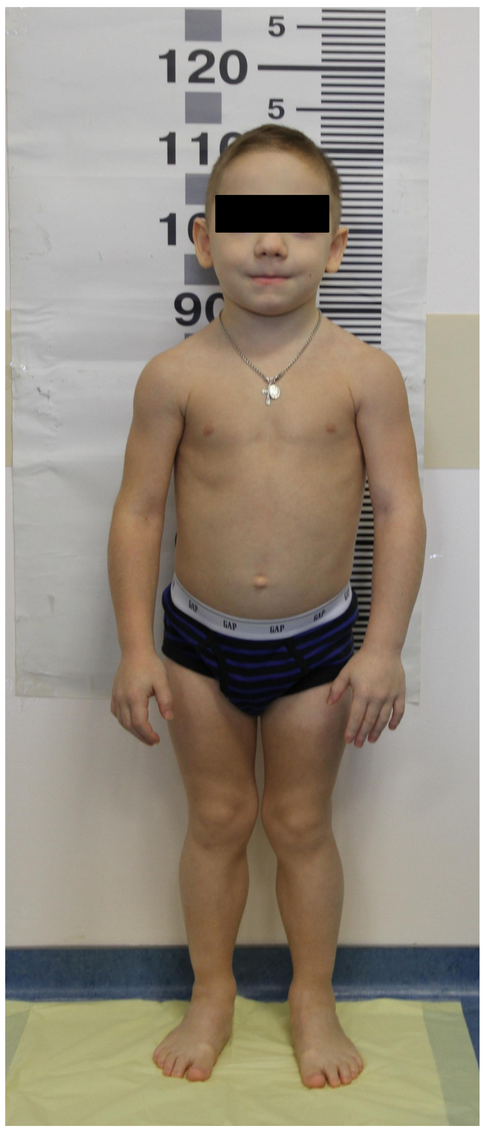
Objective

We present the case of 7-year-old boy who developed muscular pseudohypertrophy, associated with long-term untreated hypothyroidism.

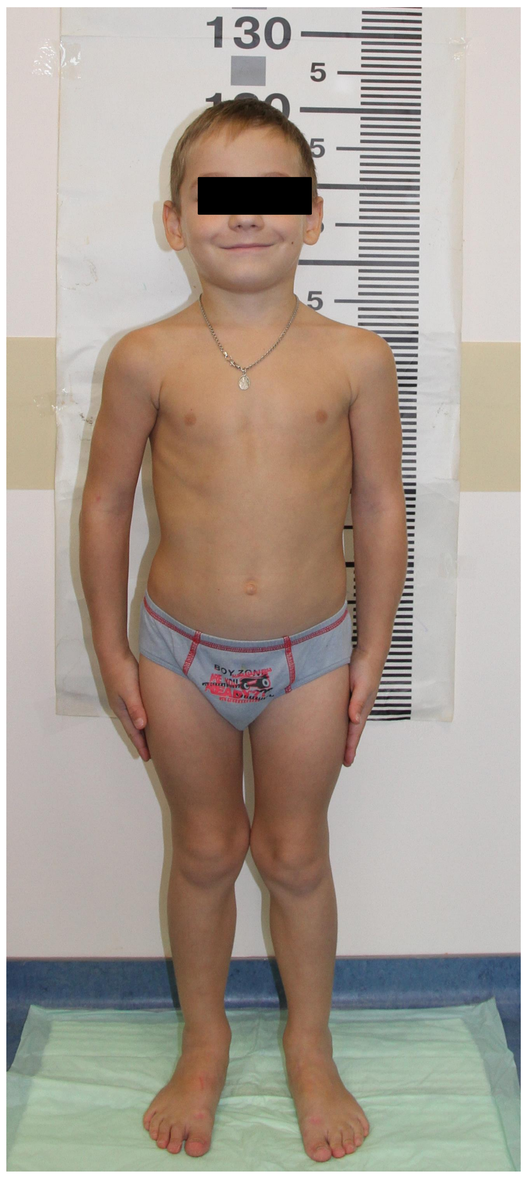
RESULTS

A 7-year-old boy presented with growth failure, lassitude and lethargy. He was born small for gestational age at 28 weeks gestation from twin pregnancy. His developmental assessment showed normal gross motor milestones during the first year of life. The child was apparently asymptomatic till 3-4 years of age, when the parents started noticing progressive weakness and lethargy along with deteriorating academic performance and growth retardation. The patient gave a history of chronic constipation. On examination he was found to be normal statured (height SDS=-1,67; height velocity SDS=-5,5) with prominent muscular build. He showed muscle hypertrophy with enlargement of the ***calf muscles*****.** He had hoarseness of voice, a dry texture of skin, a macroglossia and an odematous face (Pic. 1).

B*rain imaging* findings *showed pituitary enlargement*. The boy was found to have hypothyroidism [TSH=807 μIU/ml (0.5–4.8), free T4=5.15 pmol/l (11-18,6)]. Primary autoimmune etiology was confirmed by raised anti-TPO antibody titre >1000 mU/ml and ultrasound features of *autoimmune thyroiditis*. The serum creatinine phosphokinase [447 U/l (30-200], aspartate transaminase [113 U/l (5-34)], alanine aminotransferase [176 U/l (0-55)] and the lactate dehydrogenase [256 U/l (125-220)] levels were elevated. A diagnosis of KDSS was made on the basis of the above mentioned findings, and the child was started on levothyroxine supplementation 50μg/day. On follow-up after 3 months the child was found to be euthyroid, he showed significant improvements in his symptoms and regression in volume of calf muscles was noted. After 8 months of levothyroxine treatment growth parameters improved significantly (Pic. 2).



Picture 1. A 7-year-old boy with pseudomuscular hypertrophy in Kocher-Debre Semelaigne syndrome



Picture 2. A 7-year-old boy with Kocher-Debre Semelaigne syndrome after 8 months of levothyroxine treatment

CONCLUSION

This report is to increase awareness of the uncommon presentation of hypothyroidism in the form of Kocher-Debre-Semelaigne syndrome, which is a rare and a reversible condition.

DOI: 10.3252/pso.eu.55ESPE.2016

Poster presented at: ESPE 2016 PARIS