IS GROWTH HORMONE DEFICIENCY A CONTRIBUTOR TO SHORT STATURE IN CUTIS LAXA SYNDROME?

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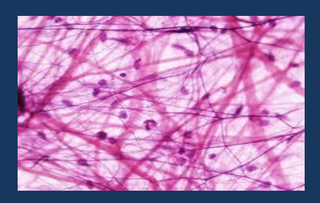


Autosomal dominant cutis laxa type 3 (ADCL3) is a genetic connective tissue disorder characterized by poor pre- and postnatal growth and, rarely, by systemic impairment. The aetiology of short stature is incompletely known, some of these patients reaching normal final height. Less than 50 cases were reported in the literature.

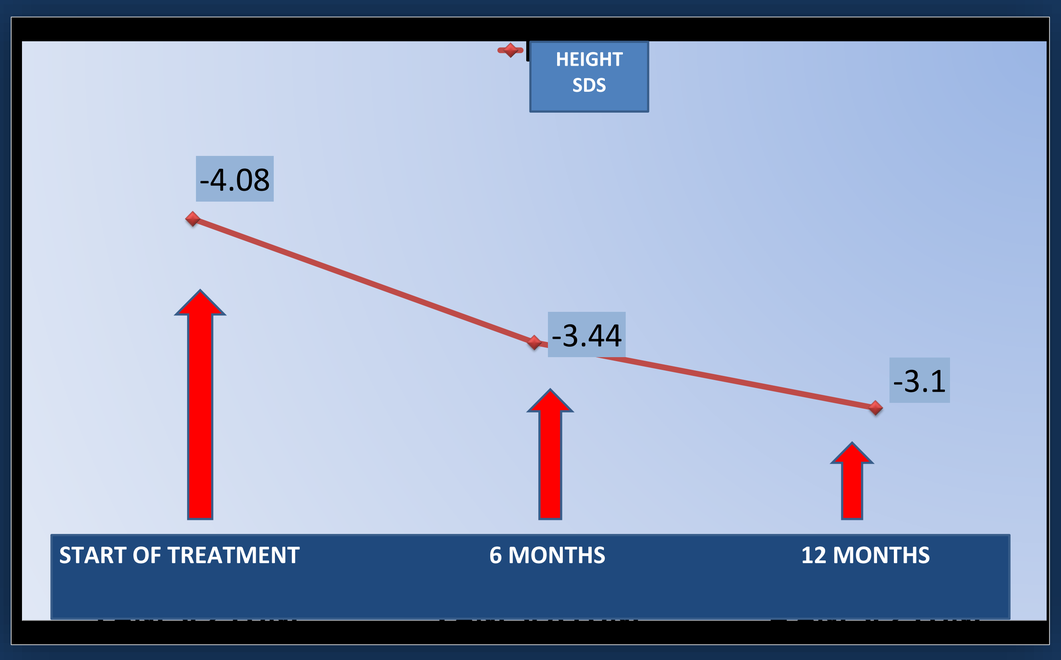
We report the case of a male patient 3.2 years old who presented for endocrinological evaluation of short stature.
Medical history: congenital hypothyroidism treated with levo-thyroxin, congenital cataract, pseudoaphakia, hiatal hernia, micturition disorder. The patient was born small for gestation age (**SGA**) with birth weight of 2080 g and birth length 41 cm at 38 weeks of gestation
Clinical exam: severe short stature (-4,08 standard deviations, SD), facial dysmorphic features, lax, thin skin with vascular markings,, body weight at the upper normal range (BMI at the 80 percentile) and decreased growth velocity (-1,76 SD in the last year).
Paraclinical exams:

- IGF1=111 ng/ml (normal range 49-283),
- peek growth hormone (GH) during two stimulation tests of 8,07 ng/ml and 2,98 ng/ml,
- Adrenal and thyroid tests normal
- bone age: 3 years
- pituitary MRI: pituitary hypoplasia

Diagnosis: short stature due to SGA and growth hormone deficiency
Treatment: somatropinum 0,035 mcg/kg body weight/day
The patient was send for genetic examination and the genetic tests showed ADCL3.

Evolution under treatment with somatropinum

- **at 6 months:** the patient gained 5 cm in height, the height **SDS** was **-3,44 DS**;
- **at 12 months**: the patient gained 5,3 cm in height, SDS for height **-3,1,** IGF 1 = 220 ng/ml (normal range: 49 -283 ng/ml),



This is the first case report of ADCL3 associated with GH deficiency treated with somatropinum. Evolution under treatment will be closely followed in interdisciplinary collaboration to optimize the results.

Referrences:
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2. Mohamed M, Kouwenberg D, Gardeitchik T, et al. Metabolic cutis laxa syndromes. J Inherit Metab Dis. 2011;34:907-916. http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3137780/

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Growth and syndromes (to include Turner syndrome)
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