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ESPE 2021
The 59th Annual Meeting Online
22-26 September 2021

A CASE OF A FLOATING-HARBOR SYNDROME IN A CHILD WITH SEVERE SHORT STATURE

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INTRODUCTION

Floating-Harbor syndrome is a rare autosomal dominant genetic disorder caused by mutations in SRCAP gene. This condition is characterized by dysmorphic facial features, short stature and expressive language delay. Here we present a clinical case of a child with Floating-Harbor syndrome.

GENETIS ANALYSIS

A known denovo heterozygous c.7330C>T:p.R2444X mutation in SRCAP gene was identified using whole exome sequencing

TREATMENT

The girl was started on recombinant growth hormone (rGH) therapy with a dose of 0.033 mg/kg/day.

CLINICAL CASE

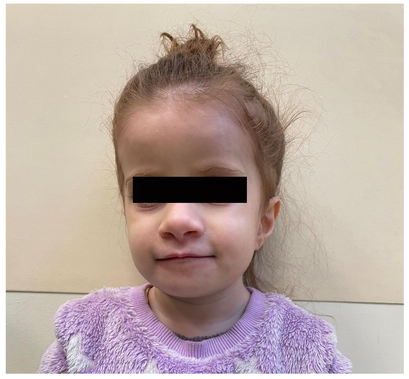
A girl to consanguineous healthy parents was born at 37 gestation weeks with height 47 cm (SDS=-0,46) and weight 2240 g (SDS=-1,6). Poor growth has been noticed since birth.

The girl has *distinctive dysmorphic features such as:* bluish sclera, low nasal bridge, frontal bossing, long eyelashes, wide columella, short filter and broad mouth with thin upper lip, teeth hypoplasia (Pic.1) and brachydactyl (Pic.2).

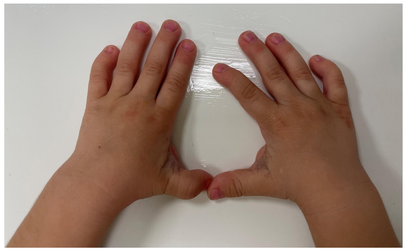
The examination at 3,5 years: height SDS =-3,6, bone age (15 months), normal IGF- 1, normal pituitary hormones and a peak growth hormone response of 18,7 ng/ml on standard glucagon stimulation test (Tab.1).

Follow up examination at the age of 4 years: height SDS = -4,11 (Pic.3).

Picture 1. The patient at the age of 4 years



Picture 2. Patient's hands



Picture 3. Growth data

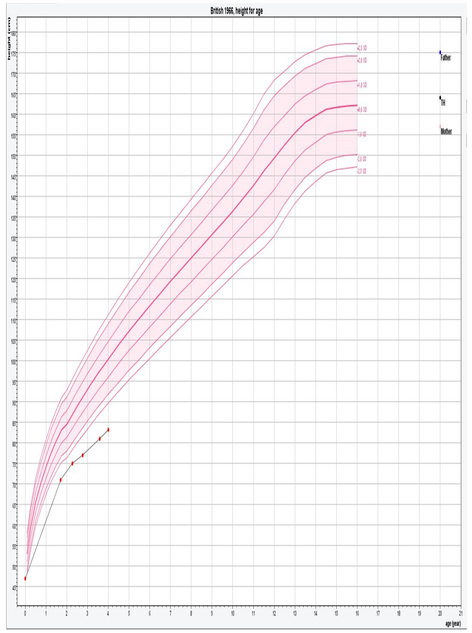


Table 1. Hormonal assessment

HORMONES	PATIENT'S VALUE	RANGE
TSH (ulU/ml)	3.7	0.64-5.76
fT4 (pmol/l)	14.69	11.5-20.4
PRL (mlU/l)	108.7	
Cortisol (nmol/l)	579.1	77-630
IGF-1 (ng/ml)	108.7	8-251
GH peak (ng/ml)	18.7	(>7 ng/ml)

CONCLUSIONS

Floating-Harbor syndrome is a rare cause of severe short stature in children. Further studies are necessary to evaluate the effectiveness and safety of rhGH therapy in these patients.

CONTACT INFORMATION

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