Endocrinological evaluation of male patient with Floating-Harbor syndrome –case report

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Introduction

Patients with Floating-Harbor syndrome have broad spectrum of clinical presentation, but most of them have short stature, low birth weight, delayed bone age, delayed speech development, typical craniofacial features, anomaly of fingers and toes, cryptorchidism in males, renal anomalies, strabismus.
It is a rare disorder –about 50 cases in literature

Patient's characteristic

- 4 year old male patient
- mutation in SRCAP gene Gln2622Ter/-
- followed up in Pediatric Endocrinology Outpatient Clinic since he was one year of age
- born small for gestation age, with birth weight 2700g in 40Hbd.
- Parents without chronic diseases
- Father's height 183cm +0,33SD
- Mother's height 173cm +1,45 SD

Anthropometry

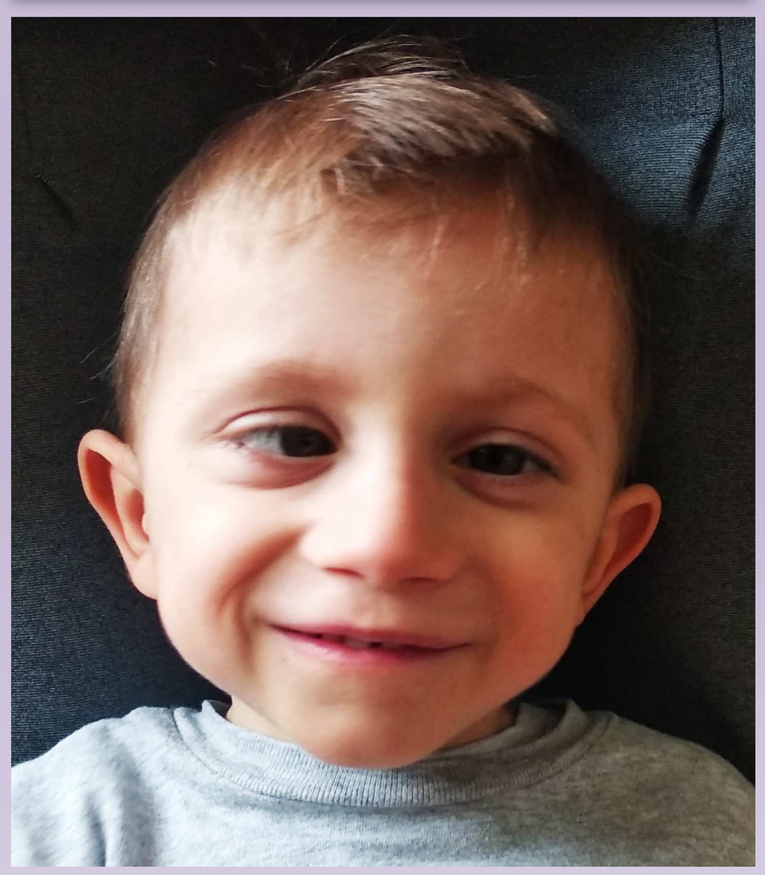
Age (years)	40 Hbd	0,5	1	2	3	4
Length/ Height (cm)	46 <-3SD Niklasson BMC Pediatrics 2007	63 z-score -2,76	68 z-score -3,63	76 z-score -4,65	82 z-score -4,38	87 z-score -4,31
Weight (kg)	2,7 -2,5SD Niklasson BMC Pediatrics 2007	5 z-score -4,11	6 z-score -3,72	7 z-score -3,87	9 z-score -3,52	10 z-score -3,14

Growth hormone, IGF-1, BP-3, bone age, cortisol evaluation

- Spontaneus (nocturnal test) –GH peak 13,3 ng/ml
- Glucagon stimulation test – GH peak 11,9 ng/ml; cortisol peak 601 nmol/l
- clonidine stimulation test – GH peak 13,7 ng/ml
- IGF-1 91,5 ng/ml
- BP-3 2,58 ug/ml
- Bone age -6 months at age 3,8.

Clinical fenotype

- some typical craniofacial features, like triangular face, short philtrum, wide mouth with a thin vermilion border of the upper lip, low-set ears, long nose
- strabismus, defect of vision;
- brachydactyly, clinodactyly, broad finger tips
- short stature
- low body mass
- speech development delay especially in verbal communication, but his speech understanding and general development is in quite good level
- testis 2ml in scrutum, G1, P1, Ax1, Tanner 1
- kidney defect not found



Thyroid function

- Euthyroidism TSH 1,1uU/ml; fT4 15,6 pmol/l
- thyroid antibodies were negative

- Thyroid USG – normal echogenicity and echostructure; bilaterally focal lesion up to 3,7x5,5x3,2mm and 4,9x5,2x2,6mm; total volume 1,43ml.

Other laboratory tests

- Creatinine, ALT, morfology, CK, Na, K, Ca, Pi, Mg in normal range
- 25OHD below lower limit despite suplementation

HbA1c and antibodies

- HbA1c in normal range 5%
- TGA IgA negative; antibodies GAD, IA2, ZnT8 negative

USG

- Abdominal USG revealed normal liver, splane and kidneys;
- ECHO -normal structure of heart

MRI

- MRI revealed normal hypophisis; normal structure of cerebri

OGTT – glucose, insulin

Fasting glucose 55mg%; 87mg% in 120' of OGTT.
Fasting insulin <1,6 uU/ml; 3,3uU/ml in 120' of OGTT.
After OGTT urine test revealed massive glucosuria

Lipids

Elevated total cholesterol: 231 mg/dl;
LDL-cholesterol: 131mg/dl;
Triglicerides 77mg/dl;
HDL 85mg/dl.

Growth hormone therapy

rGH treatment was started for SGA patient one month ago.

Conclusions

- The patient with Floating-Harbor syndrome didn't revealed endocrinopaties, but SGA, short stature, bone age delay and glucosuria

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