

# Features of somatropin replacement therapy in a patient with Floating Harbor Syndrome

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Growth hormone (GH) deficiency in children, confirmed by stimulation diagnostic tests, in some cases is accompanied by low effectiveness of somatropin replacement therapy, which may be associated with rare genetic syndromes.

## **Aim of our scientific work:**

To study the growth effects of GH therapy in treating a patient with Floating-Harbor Syndrome

## **Methods:**

A GH deficiency was diagnosed in a patient 3 years old using clonidine test (peak stimulated GH 7,2 ng/ml). The patient was observed by a neurologist, cardiologist, ophthalmologist with diagnoses: organic brain disease perinatal genesis, stigma disemбриogenesis, delayed speech development, open oval window, convergent parietic squint.

MRI of the brain and pituitary showed no pathology. Deficiency of other hormones of the adenohypophysis wasn't detected. The bone age significantly lagged behind the passport, was 8-9 months.

GH replacement therapy was prescribed at the age of 4,5 years. SDS growth at the time of observation  $-2,38 + 0,86$  ( $-4.09 \div -1.2$ ). Growth rate  $4,8 + 2,68$  cm/year ( $0,5 \div 10$  cm). SDS growth rate  $0,4 + 2,07$  ( $-4,3 \div 2,55$ ).

## **Results:**

During the molecular genetic survey for the diagnosis of mutations in the genes responsible for the development and functioning of the pituitary-GH-IGF-I system, no mutations were identified. Additional examination revealed mutations in the SCRAPc7466C>G gene, which made it possible to diagnose the presence of Floating Harbor syndrome.

## **Conclusions:**

Diagnosis of GH-deficiency, based only on the determination of GH by stimulation tests, does not allow to verify the pathogenetic diagnosis. Additional carrying out molecular genetic diagnostics in difficult cases while observing a patient over time at low (or insufficiently high) growth rates during treatment with somatropin is necessary to predict treatment.

