SHORT STATURE AND EFFICACY OF GROWTH HORMONE TREATMENT IN A CHILD WITH NAIL-PATELLA SYNDROME. A CASE REPORT.

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CASE REPORT

The patient was born from normal pregnancy; his weight and height were 3.1 kg and 52 cm, respectively. The family history was unremarkable. Mom's height was 163 cm; father's height was 175 cm. A violations of extension in the elbow joints were noted from birth. The hypoplasia of patella was revealed at 1 year old, and the NPS was diagnosed.

The patient presented with nephrotic syndrome at the age of 3 years, he receives ACE inhibitors to the present. A proportionate short stature was detected at 3.5 years old: height 91 cm (SDS = -1.8).

The boy was examined at the hospital at 8 years old: height 113 cm (SDS = -2.3), height velocity (HV) 3 cm/year, IGF-1 - 110 ng/ml, euthyroidism, proteinuria up to 0.3 g/l. His bone age retarded on 4 years from chronological. The organ of vision was without pathology. Hypoplasia of the adenohypophysis was on MRI of the brain. The parents refused to conduct GH-stimulation tests. The diagnosis of NPS was confirmed at 9 years old by detecting of heterozygous pathogenic variant in the LMX1B gene (c.706G>C). This mutation was not identified in his parents.

Growth hormone therapy was started at 9.5 years old at a dose of 0.033 mg/kg/day with height 118 cm (SDS= -2.7) and HV 3 cm/year on start moment. A good growth effect was after 1 year of therapy (at 10.5 years old): height 127 cm (SDS =-1.9), HV 9 cm/year. The normoglycemia and euthyroidism were during therapy, IGF-1 - 173 ng/ml.

No side effects are noted. The therapy was continued.

CONCLUSIONS

The good effect of one year of growth hormone therapy has been described in a boy with the NPS. To determine the effect of somatropin on the final growth and the incidence of side effects in NPS, it is necessary to exchange experience in the treatment of such children.

Authors have nothing to disclose