CLINIC, LABORATORY AND PROGNOSTIC FINDINGS IN PATIENTS WHO ARE DIAGNOSED BY KOWARSKİ SYNDROME (BIOLOGICALLY INACTIVE GROWTH HORMONE)

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INTRODUCTION

Short stature and low insulin-like growth factor-I (IGF-I) concentration despite normal to high GH concentration suggest impaired GH effect. The prototypical GH insensitivity syndrome was described and characterized by the absent or defective GH receptors. Growth retardation resulting from biologically inactive GH was also described, but the molecular basis of biologically inactive GH has remained unclear.

AIM

The clinical, laboratory and prognostic findings of the patients diagnosed with Kowarski Syndrome (biologically inactive growth hormone) were evaluated between February 2009 and February 2019 in order to contribute to literature.

METHOD

110 cases diagnosed with Kowarski syndrome between February 2009 and February 2019 were evaluated retrospectively. 13 cases were excluded because their data could not be reached. 97 cases continued to work. 92 cases at least one year, 62 cases at least two years, 30 cases at least three years, 9 cases at least four years and two cases at least five years were followed.

RESULTS

Age at the beginning of medical therapy, body height, body weight, SDS of height, SDS of weight, body surface area, BMI, SDS of BMI, bone age, height age, status of puberty, pituitary MRG, parents' height, midparental height, SDS of midparental height, pretreatment length growth rate, thyroid function tests, IGF-1 level, responses to the growth hormone stimulation tests, responses to the IGF-1 generation test, GH treatment doses, adverse effects after GH treatment, gaining height and SDS of height were recorded in certain periods with GH treatment. Dose of rhGH was applied between 0.2-0.25 mg/kg/week in our department. The average age of the patients was 9,44 ± 3,34 year (48,5% male and 51,5% female). Pre-treatment SDS of height -3,21 ± 1,03, midparental height 162,51 ± 8,92, SDS of midparental height -1,04 ± 0,74, bone age 7,04 ± 3,26 year, height age 6,78 ±3 year and BMI 15,55 ± 2,42 kg/m², pretreatment length growth rate 3,37 ± 0,92 cm/year were found. Height gaining was 9,04 ± 2,58cm/year at the end of the first year, 7,3±1,66cm/year at the end of the second year, 6,48 ± 1,58 cm/year at the end of the third year, 6,96 ± 0,97 cm/year at the end of the fourth year, 5,34 ±2,58 cm/year at the end of the fifth year of the medical therapy. SDS of height gaining was 0,54 ± 0,48 after the first year, 0,31 ± 0,31 after the second year , 0,28 ± 0,31 after the third year of the medical therapy. Hypothyroidism was detected in 4 patients and scoliosis was detected in 4 patients during the follow-up of patients receiving GH treatment. It was found that height and height SDS gain were highest in the first year of treatment and gradually decreased in the following years.

CONCLUSIONS

Kowarski syndrome has a very good response with low side effects to GH treatment similar to GH deficiency. Therefore, GH treatment should be started in these cases. This study is unique presentation because of the most numbered patients with kowarski syndrome (n=97) is included. The cases treated with the diagnosis of Kowarski syndrome were investigated with multiple parameters.

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