EVALUATION OF THE EFFECT OF X CHROMOSOME ABNORMALITIES ON THE RESPONSE TO GROWTH HORMONE THERAPY IN CHILDREN WITH TURNER SYNDROME

INTRODUCTION

Turner syndrome (TS) is a condition caused by structured or numeric abnormalities of X chromosome. Growth deficiency characterizes all patients with TS and may be associated with haploinsufficiency of the SHOX gene. The aim of the study was to determine if the response to the human growth hormone (hGH) therapy might be influenced by the type of karyotype abnormalities.

MATERIALS AND METHODS

57 TS patients treated with hGH therapy with a 3-year follow-up were enrolled in the study. Genetic analyses in order to evaluate the X chromosome structured or numeric abnormalities were performed and patients were categorized according to their karyotype type 1 = X monosomy (n=35), 2 = isochromosome (n=11), 3 = marker chromosome or Y chromosome (n=5), 4 = X-mosaicism (n=6) [Fig.1]. The groups did not vary significantly in the dosage and age of the patients. Anthropometric parameters and height velocity (HV) were evaluated annually. Height and HV were expressed as standard deviations scores (SDS) and deltaHSDS were calculated to show the progress in growth. Target height and target height SDS (TH SDS) were used to show the results of the 3-year long therapy.

RESULTS

During the first year of the therapy all groups responded well to the treatment with hGH. The only statistical difference occurred between patients with isochromosome, which tended to have a poorer response, and X-mosaicism, with the best response (0.579±0.196 vs. 0.851±0.171; p=0.012). [Fig.2]

The second and the third year showed the biggest difference in the effectiveness of the therapy. The response in patients with X-monosomy and isochromosome deteriorated significantly in comparison to other two groups (patients with marker chromosome or Y chromosome and patients with X-mosaicism). [Fig.3,4]

After 3 years of therapy all patients improved the score in comparison to their target height but better results were shown in patients with marker chromosome or Y chromosome and patients with X-mosaicism, which had the best final result. [Fig.5,6]

CONCLUSIONS

X-monosomy or the presence of isochromosome determines a poorer response during the second and third year of hGH therapy in TS. The best response to hGH therapy during the second and third year was observed in TS patients with X-mosaicism and marker chromosome or Y chromosome. The results of the study might prove the need of a more individualized dosage of hGH during the second and third year of therapy in patients with X-monosomy and isochromosomes.