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TOTAL GROWTH HORMONE DEFICIENCY IN TWO OBESE, RED-HAIRED BROTHERS

Z.S. Gucev[1], G.J. Jovanov[2], M. Krstevska-Konstantinova[1]
[1]Pediatric Clinic, Medical Faculty Skopje [2]Health Center Skopje, Macedonia
gucevz@osi.net.mk

IDj and IM are brothers. Two older sisters are healthy and of normal stature. At the age of 28 months IDj was 68 sm (9 SD bellow the mean), his bone age was 6 months. His brother IM at age of 20 months had height of 62 sm (5 SD bellow the mean, bone age 10 months). They both had proportionate short stature, truncal fat.

No signs of malabsorbtion or renal disease were found. Karyotype: male (46, XY). Tests of growth hormone reserve (clonidin, L-dopa, glucagon), GH during sleep were near the detection limit (DELFIA; below 0.03 mU/L; severe GH deficiency). IGF-1, IGFBP-3 values were not available in the beginning, normal after GH treatment. T4 and TSH, prolactin, ACTH, cortisol, were within normal range. Testosteron, 17-OH prrogesteron, DHEA-S and estradiol: results normal for sex and age. CT and MRI of the brain: normal.

GH replacement therapy begun and IDj got 26 sm in the first year. After 6 years of therapy he reached the ~95 percentile. A strong catch-up growth was noticed in the case of IM, too. He is growing around the 97th percentile on the growth curve.

Furthermore IDj developed a clumsy gait, was prone to falling. EMG was non-specifically changed, muscle biopsy is underway. No genetic abnormality in the GH receptor and the Pit-1 factor were detected.