



EVALUATION OF OSTEOPOROSIS IN WOMEN WITH PRADER-WILLI SYNDROME

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INTRODUCTION: In Prader-Willi Syndrome (PWS) hypothalamic dysfunction is the cause of some clinical findings like growth hormone deficiency (GHD), hypogonadism. Both hypogonadism and GHD lead to an alteration of body composition with increased fat mass, reduction of lean mass and increased risk of osteoporosis. The aim of this study is to investigate the presence of osteoporosis in PWS women.

PATIENTS and METHODS: We studied 15 women, age 26.49±4.47 (range 20-35 yrs), BMI 51.67±13 kg/m² (range 30.8-71.4) with genetically confirmed PWS. A deletion of chromosome 15q (FISH) was found in 9/15 (60%). Nine pts had primary amenorrhea (group A) and 6 were menstruated (group B). Seven patients were treated with GH during childhood, 4/9 (44%) in group A and 3/5 (60%) in group B. None had history of treatment with estrogens or other therapy that could affect bone metabolism. Dual Energy X-ray Absorptiometry (DXA) (Hologic Delphi 70400) was performed in all patients and bone mineral density (BMD) of lumbar spine (expressed also as T score and Z score), Fat %, Fat mass (kg), Lean mass (kg) were calculated. Basal FSH, LH and 17β-estradiol were measured. Data are expressed as mean±SEM.

RESULTS: Basal LH (0.72±0.21UI/L) and FSH (2.22±0.56 UI/L) revealed a hypogonadotropic hypogonadism in all patients. In group A BMD of lumbar spine was 0.86±0.04 g/cm², T score -1.69±0.32, Z score -1.62±0.31, fat % 50.5±1.2, fat mass 62.6±7.9 kg, lean mass 52.3±3.4 kg. Two patients out of 9 (22%) had osteoporosis (T and Z score<-2.5 SD); 5/9 patients (56%) had osteopenia (T and Z score <-1 and >-2.5 SD), 2/9 patients (22%) had normal BMD. In group B BMD of lumbar spine was 0.90±0.06 g/cm², T score -1.3±0.5, Z score -1.2±0.5, fat % 48.5±1.1, fat mass 53.3±6.5 kg, lean mass 54±5.2 kg. One patient out of 6 (16%) had osteoporosis, 3/6 patients (50%) had osteopenia, 2/6 patients (34%) had normal BMD. No significant difference was found between group A and B in BMD, fat %, fat mass, lean mass, BMI, age and gonadotropin levels. Meanwhile group B showed higher 17β-estradiol levels than group A (63.37±11.65 vs 29.16±5.3, p=0.0128). The linear regression analysis showed a significant positive correlation between 17β-estradiol levels and BMD in group A (r²=0.531, r=0.729, p=0.0402). No significant correlation was found in group B. Furthermore, in both groups BMD was similar in GH treated subjects in comparison to patients without GH therapy.

CONCLUSION: In our study a high prevalence of osteoporosis (22 % in amenorrheic and 16% in women with menses) and osteopenia (56% and 50% respectively) was found independently from the presence or absence of menses. Furthermore, BMD doesn't seem to be preserved by GH therapy. However, wider studies on amenorrheic and/or menstruated PWS woman as well as a comparison with a group of amenorrheic PWS treated with oestrogen are needed.