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Romanian Prader-Willi Association

ASOCIACIÓN MADRILEÑA
PARA EL SÍNDROME DE
PRADER-WILLI



HORMONE REPLACEMENT IN ADOLESCENTS AND ADULTS WITH PRADER-WILLI SYNDROME

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INTRODUCTION: Hypogonadism is present almost universally in males with Prader-Willi syndrome (PWS) and in the majority of females. Anecdotally testosterone replacement in males and oestrogen replacement in females are under-prescribed due to the concerns about behavioural change and interactions with other co-morbidities. Hormone replacement therapy (HRT) has a positive effect on bone mineral density (BMD), may be cardio-protective and may produce a physical appearance closer to peers. The lowered BMD seen in males and females with PWS demands attention, due to increasing longevity and fracture risk in this population.

AIMS & METHODS: The PWS clinic for adolescents and adults, at a major public teaching hospital in Sydney, Australia, has enrolled 64 clients over the past 16 years. Thirty eight people attend regularly, from within and outside the metropolitan area. The aim of this retrospective study was to look at endocrine function, BMD and HRT use in this clinical cohort and compare the difference between those on and those not on HRT and the reasons behind decisions regarding the use of HRT.

RESULTS & DISCUSSION: Fifteen clients, 8 male, 5 female receive HRT and have had BMD studies. The genotype split is 10 with deletion and 4 with UPD, and 1 non-deleted. Mean age of the treated cohort at their first BMD was 21 years and baseline BMD T scores range from -0.8 to

- 3.0 standard deviations. There was no difference in age at baseline between the treated group and non treatment group of 12 clients (6 male, 6 female), with similar baseline data available. The T scores for the non treated group ranged from 0.9 to -3.8 standard deviations. Baseline testosterone and oestrogen levels for both groups were in the hypogonadal range.

Clinically significant improvements in spinal BMD were seen in all but 2 of the HRT patients after treatment but clinically significant improvements in hip BMD were only seen in 3 HRT clients.

One client has demonstrated adverse affects to testosterone after 2 separate attempts to treat, each resulting in cessation of treatment. HRT treatment schedules appear to be influenced by attendance rates, parent and carer attitudes and the geographical distance from our service. Regular adult clinical care of patients with PWS is continually encouraged to ensure consistent use of HRT and prolonged improvement in BMD.