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Romanian Prader-Willi Association

ASOCIACIÓN MADRILEÑA
PARA EL SÍNDROME DE
PRADER-WILLI



CENTRAL HYPOTHYROIDISM (CH) IN PATIENTS WITH PRADER WILLI SYNDROME DURING THE FIRST 2 YEARS OF POSTNATAL LIFE

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INTRODUCTION: Prader Willi syndrome (PWS) is a genetic disorder with a well characterized phenotype, mostly related to hypothalamic dysfunction. Features during the first 2 years (y) of life are a moderate intra-uterine and post-natal growth delay, general muscular hypotonia, feeding difficulties, and delayed psychomotor development. Endocrine disorders such as growth hormone deficiency and hypogonadism have been described. However, in infant affected patients, thyroid function has not been well studied. The **aim** of this study was to analyze hypothalamo-pituitary-thyroid function during the first 2 years of life in PWS patients.

METHODS AND DESIGN: 11 patients (9 males, 2 females) with fulfilled clinical criteria for PWS diagnosis and confirmed molecular analysis were included. Birth weight (BW) and TSH neonatal screening (NS) was evaluated. At PWS diagnosis, auxological parameters, bone age (BA), basal serum thyroid stimulating hormone (TSH), total (t) thyroxine (T4) and free (f) T4 levels were determined. Mean (\pm SD) hormone values were compared with age-and gender-matched healthy control subjects.

RESULTS: 10/11 had normal BW for gestational age .TSH NS was normal in 11/11. At PWS diagnosis, all patients had adequate weight for height. Mean chronological age (CA): $0.66 \pm 0.51y$ (range 0.08-1.56 y), mean BA delayed (5/11) CA-BA: $0.52 \pm 0.3y$, mean height SDS: -1.62 ± 1.11 were found. TSH levels were within normal range: $2.39 \pm 1.6 \mu g UI/ml$. tT4: $7.23 \pm 1.15 \mu g/ml$ and fT4: $0.72 \pm 0.08 ng /dl$ were significantly lower than age match control and below the lower limit of 95 % confidence interval of the normal population reference.

WE CONCLUDE that central hypothyroidism is a common feature in PWS patients during infancy. TSH NS alone is not an accurate tool to be applied in PWS. Pediatricians should be aware of the need to evaluate central hypothyroidism in PWS in this critical period of thyroid action on neurological development. In the presence of CH we might propose that early thyroxine replacement therapy could improve severe early features and morbidity.