CLINICAL THYROIDOLOGY **FOR PATIENTS**

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HYPOTHYROIDISM

Children with Prader–Willi Syndrome frequently have central hypothyroidism

BACKGROUND

Prader–Willi syndrome is a rare genetic disease with an incidence of 1 in 10,000 to 1 in 16,000 of newborns. Patients with this disorder have a variety of problems including moderate to severe mental retardation, short stature and severe obesity that is a result of a marked increase in appetite. There are multiple endocrine abnormalities that affect pituitary function. The pituitary produces TSH that regulates thyroid function. The vast majority of hypothyroidism is cause by thyroid gland failure and is associated with increased TSH levels. Hypothyroidism due to pituitary problems (central hypothyroidism) is associated with normal or low TSH levels in the setting of low thyroid hormone levels. Central hypothyroidism has been reported in 20% to 30% of patients. This study examined thyroid function in all infants diagnosed at the Hospital de Pediatria Garraham in Buenos Aires over a period of 5 years.

THE FULL ARTICLE TITLE:

Vaiani E et al. Thyroid axis dysfunction in patients with Prader–Willi syndrome during the first 2 years of life. Clin Endocrinol 2010;73:546-50.

SUMMARY OF THE STUDY

Eighteen patients (11 boys and 7 girls) up to 2 years of age were included in this study. The diagnosis was documented by genetic testing. The diagnosis of central hypothyroidism was considered if serum T_4 was less than the 2.5th percentile of the normal range and if there was absence of the expected increase in serum TSH.
In 61% of patients (11 of 18), T_4 values were below the 2.5 percentile and in 2 additional cases the value was just borderline. With the exception of one case, all patients had normal serum T_3 levels. Interestingly, body length was significantly shorter in the hypothyroid patients than in the small group of patients with normal T_4 levels.

WHAT ARE THE IMPLICATIONS OF THIS STUDY?

This small study suggests that central hypothyroidism is common in Prader–Willi syndrome and is seen at a very early stage of life. At present, it is not clear whether this hypothyroidism is short-lived or permanent. While these findings do need to be confirmed, this study suggests that pediatricians treating patients with Prader–Willi syndrome should have a low threshold for diagnosing and treating central hypothyroidism.

— Alan P. Farwell, MD

ATA THYROID BROCHURE LINKS

Hypothyroidism: http://thyroid.org/patients/patient_brochures/hypothyroidism.html

ABBREVIATIONS & DEFINITIONS

Prader–Willi syndrome: a rare genetic disorder associated with moderate to severe mental retardation, short stature and obesity due to over-eating.

Pituitary gland: this endocrine sits at the base of the brain and secretes hormones that control thyroid and adrenal function, growth and reproduction. The pituitary gland secretes TSH to control thyroid function.

Hypothyroidism: a condition where the thyroid gland is underactive and doesn't produce enough thyroid hormone. Treatment requires taking thyroid hormone pills.

TSH: Thyroid stimulating hormone – produced by the pituitary gland that regulates thyroid function; also the best screening test to determine if the thyroid is functioning normally.

Thyroxine (T_4): the major hormone secreted by the thyroid gland. Thyroxine is broken down to produce Triiodothyronine which causes most of the effects of the thyroid hormones.