



HYPOTHYROIDISM

Delaying ultrasonography in congenital hypothyroidism may give misleading results.

BACKGROUND

Congenital hypothyroidism is a disorder in which babies are born with low thyroid hormone levels, either because the thyroid did not develop properly (thyroid dysgenesis) or because the thyroid has problems in one of the needed steps to make thyroid hormones (thyroid dyshormonogenesis). Congenital hypothyroidism is estimated to occur in 1:1700 newborns in the most recent literature and, if left untreated or if treatment is delayed, it irreversibly affects brain development. Thyroid scintigraphy is a procedure in which a radioactive compound which is taken up by the thyroid gland is used to determine the location of the gland or to confirm the absence of thyroid tissue. This procedure needs to be done when the child is not on thyroid hormone, either prior to starting treatment or after holding treatment for some weeks.

Thyroid ultrasound and thyroid scintigraphy have been used to determine the cause of congenital hypothyroidism, whether due to dyshormonogenesis or dysgenesis. In dysgenesis, the gland may be absent, smaller, or in an abnormal position. In dyshormonogenesis, the gland is usually normal or larger in the absence of thyroid hormone replacement. Thyroid dyshormonogenesis may be inherited in 25% of the children in a family, so it is important to make the right diagnosis of this condition for genetic counseling. Since the outcomes of congenital hypothyroidism depend on starting treatment as soon as possible after diagnosis, diagnostic studies to determine the etiology of congenital hypothyroidism are usually delayed after the age of three years, or not done at all, which may cause uncertainty in the patient and lack of adequate genetic counseling. This study was done to determine whether ultrasound of the thyroid could have a role in the early diagnosis of congenital hypothyroidism and to determine whether delaying ultrasound could provide misleading information.

THE FULL ARTICLE TITLE

Borges MF et al. Timing of thyroid ultrasonography in the etiological investigation of congenital hypothyroidism. *Arch Endocrinol Metab*. February 13, 2017 [Epub ahead of print]. doi:10.1590/2359-3997000000239

SUMMARY OF THE STUDY

A total of 44 patients with a diagnosis of congenital hypothyroidism from the state of Minas Gerais, Brazil, were invited to have thyroid US at the Universidade Federal do Triângulo Mineiro in Uberaba, Brazil. All except three accepted the invitation and participated in the study (23 females and 18 males), ranging in age from 0.2 to 45 years. All were receiving treatment and were considered to have congenital hypothyroidism, except for 1 patient, whose elevation in TSH was transient and had resolved. Patients were divided in two groups: Group 1 (23 patients) included patients diagnosed by the State Neonatal Screening program and Group 2 (21 patients) included patients who had been followed in the city of Uberaba's Municipal Health Unit. In Group 1, 15 patients had already undergone ultrasound and scintigraphy between ages 3 and 4. In Group 2, 15 of the patients had previously undergone ultrasound, but only two had undergone thyroid scintigraphy. Information related to these prior studies was obtained from the medical records. The second ultrasound was compared to the initial one, when available. When a thyroid was found, measurements were obtained to calculate the thyroid volume. The volumes were compared with reference ranges from medical references or from normal children to determine whether glands were bigger, normal or smaller than normal.

The major results of the study were that 24.5% of patients (10 patients) did not have a visible thyroid on ultrasound. All of these patients were diagnosed with thyroid dysgenesis. A total of 4 of these patients had previously undergone thyroid scintigraphy and 8 had undergone another US. In only 5/8 patients, there was complete agreement between the two ultrasounds, however, the second ultrasound identified all cases of dysgenesis. In 31 patients, the thyroid was noted in its normal location. In 18 patients, the thyroid was normal size, but 1 of these patients had only one lobe (half the thyroid) and another had transient elevation of TSH level, which resolved. Therefore, 16 patients were given a diagnosis of dyshormonogenesis. The thyroid was smaller than normal in 13 patients; in 6 of these patients, the initial ultrasound

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showed the thyroid size to be normal and in 4 the thyroid volume in the second ultrasound was less than in the initial US. Therefore, these 6 patients were also assigned a diagnosis of dyshormonogenesis. In the remaining 7 of these 13 patients, 1 was diagnosed as having central hypothyroidism and the other 6 were variously referred to as having small thyroids (hypoplasia or thyroid dysgenesis). Half of the patients with small thyroid glands on the second US may have been diagnosed with dysgenesis instead of dyshormonogenesis if they had not one an initial US earlier in life.

WHAT ARE THE IMPLICATIONS OF THIS STUDY?

Thyroid ultrasound is a test that can be obtained in early infancy without delaying treatment for congenital hypothyroidism. Ultrasound can give valuable information in making an accurate diagnosis for the cause of congenital

hypothyroidism, especially when done in early infancy when delaying thyroid ultrasound may lead to the wrong diagnosis of thyroid dysgenesis in the patients in which thyroid hormone replacement can decrease the size of the thyroid. As thyroid dyshormonogenesis may be inherited in 25% of the children in a family, it is important to make the right diagnosis of this condition for genetic counseling.

— Liuska Pesce, MD

ATA THYROID BROCHURE LINKS

Hypothyroidism (Underactive): <https://www.thyroid.org/hypothyroidism/>

Thyroid Function Tests: <https://www.thyroid.org/thyroid-function-tests/>

Thyroid Hormone Treatment: <https://www.thyroid.org/thyroid-hormone-treatment/>

ABBREVIATIONS & DEFINITIONS

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

Congenital hypothyroidism: hypothyroidism that exists at birth either because the thyroid did not develop properly (thyroid dysgenesis) or because the thyroid has problems in one of the needed steps to make thyroid hormones (thyroid dyshormonogenesis). Congenital hypothyroidism is estimated to occur in 1:1700 newborns.

Thyroid dysgenesis: a cause of congenital hypothyroidism where the thyroid did not develop properly

Thyroid dyshormonogenesis: a cause of congenital hypothyroidism where the thyroid has problems in one of the needed steps to make thyroid hormones. Thyroid dyshormonogenesis may be inherited in 25% of the children in a family

Central hypothyroidism: a rare cause of hypothyroidism where the thyroid gland is normal and the problem is inadequate TSH secretion from the pituitary gland.

Thyroid hormone therapy: patients with hypothyroidism are most often treated with Levothyroxine in order to return their thyroid hormone levels to normal. Replacement therapy means the goal is a TSH in the normal range and is the usual therapy.

TSH: Thyroid Stimulating Hormone — the hormone that stimulates the thyroid gland to make thyroid hormones. High TSH correlates with primary hypothyroidism

Thyroid ultrasound: a common imaging test used to evaluate the structure of the thyroid gland. Ultrasound uses soundwaves to create a picture of the structure of the thyroid gland and accurately identify and characterize nodules within the thyroid. Ultrasound is also frequently used to guide the needle into a nodule during a thyroid nodule biopsy.

Thyroid scintigraphy: this imaging test uses a small amount of a radioactive substance, usually radioactive iodine or technetium 99, to obtain a picture of the thyroid gland.