AMERICAN THYROID ASSOCIATION
Optimal Thyroid Health for All

Volume 14 | Issue 3 | March 2021

Clinical Thyroidology® for the Public

HYPOTHYROIDISM

A review of the 2020 guidelines for congenital hypothyroidism

BACKGROUND

Congenital hypothyroidism is a type of hypothyroidism that is present at birth either because the thyroid did not develop properly or because the thyroid has problems in making thyroid hormones. It occurs in 1:1700 newborns. Thyroid hormones play an important role to growth and development. In particular, normal brain development is dependent on thyroid hormone and the absence of thyroid hormone during the first 3 months of life results in marked decrease in intelligence. Newborn screening is helpful in identifying patients with congenital hypothyroidism. Screening allows for early diagnosis and treatment of congenital hypothyroidism to avoid severe developmental delay. Unfortunately, 70% of infants in the world are born in regions without newborn screening programs. The best test for detecting congenital hypothyroidism is measuring the thyroid stimulating hormone (TSH) level. TSH is a hormone that regulates thyroid function and the TSH level can help determine if the thyroid is working normally. Early treatment of congenital hypothyroidism and close monitoring to normalize the TSH level is important. The purpose of this study is to update the guidelines for the diagnosis and treatment of congenital hypothyroidism.

THE FULL ARTICLE TITLE

van Trotsenburg AS et al. 2020 Congenital hypothyroidism: A 2020 consensus guidelines update An ENDO-European Reference Network (ERN) initiative endorsed by the European Society for Pediatric Endocrinology and the European Society for Endocrinology. Thyroid. Epub 2020 Dec 3. PMID: 33272083.

SUMMARY OF THE STUDY

A total of 22 participants from the Endo-European Reference Network (ERN) and the European Society for Pediatric Endocrinology and the European Society for Endocrinology aimed to update the guidelines for the diagnosis and management of congenital hypothyroidism. A review of the literature was completed to identify important articles on neonatal screening, diagnosis and management of congenital hypothyroidism. The guidelines were based on evidence (evidence-based) and were graded on the strength of the recommendations and quality of evidence. If evidence was lacking, recommendations were made based on expert opinion.

For abnormal congenital hypothyroidism screenings, the guidelines recommend measuring serum free thyroxine (FT_4) and TSH levels. Thyroxine is the major hormone made by the thyroid gland. Congenital hypothyroidism is categorized as mild (FT_4 levels of 10 – 15 pmol/L), moderate (FT_4 levels of 5 – 10 pmol/L), or severe (FT_4 levels < 5 pmol/L). Management of congenital hypothyroidism includes replacing the missing thyroid hormone with levothyroxine. The dose of levothyroxine is based on body weight per day and adjusted to keep thyroid hormone levels and TSH within normal. Treatment with levothyroxine should begin if the FT_4 is low and the TSH is elevated or if the TSH is > 20 mU/L even if FT_4 is normal. Treatment with levothyroxine should also begin if the TSH > 6 mU/L beyond 21 days of age. Genetic testing can help with make the diagnosis and can guide genetic counseling. All newborns with congenital hypothyroidism should be evaluated for birth defects such as cardiac defects and hearing loss. Preterm, low-birth-weight or very-low-birth weight infants, same-sex twin of an affected baby, and patients with Down syndrome have an increased risk for false-negative congenital hypothyroidism screening results (when screen results are negative, but patient actually has congenital hypothyroidism). For these newborns, a second screening test is recommended at approximately 10 to 14 days of age. In patients with congenital hypothyroidism, brain development progress and hearing should be regularly evaluated.

WHAT ARE THE IMPLICATIONS OF THIS STUDY?

Early identification and appropriate treatment of congenital hypothyroidism is important in optimizing neurodevelopmental outcomes in these patients. Further studies are needed to understand the genetic causes and the increasing number of individuals who have congenital hypothyroidism.

— Priya Mahajan, MD

HYPOTHYROIDISM, continued

ATA THYROID BROCHURE LINKS

Congenital Hypothyroidism: https://www.thyroid.org/congenital-hypothyroidism/
Thyroid Function Tests: https://www.thyroid.org/thyroid-function-tests/
Thyroid Hormone Treatment: https://www.thyroid.org/thyroid-hormone-treatment/

ABBREVIATIONS & DEFINITIONS

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.

Congenital: Condition that exists at birth.

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

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.

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 produced by the thyroid gland. T_4 gets converted to the active hormone T_3 in various tissues in the body.

Levothyroxine (T_4): the major hormone produced by the thyroid gland and available in pill form as Synthroid™, Levoxyl™, Tirosint™ and generic preparations