



HYPOTHYROIDISM IN CHILDREN

U.S. newborn screening of congenital hypothyroidism

BACKGROUND

Congenital hypothyroidism is a condition in newborns in which the thyroid gland is underactive and does not produce sufficient thyroid hormone starting at birth. It occurs in 1 in every 2,000-4,000 live births, and approximately 1,400 newborns in the U.S. are diagnosed each year. It is an important disease to detect, since not having enough thyroid hormone as a newborn and during infancy can result in poor growth and abnormal brain development. When congenital hypothyroidism is found in a newborn, thyroid hormone medication is started immediately to prevent these severe problems and most children who are treated have an excellent prognosis.

Most infants at birth will show no clinical symptoms of low thyroid hormone levels. Thus, newborn screening of congenital hypothyroidism began in most developed countries during the early 1970s. In the United States, thyroid blood tests are done 1-3 days after birth at the hospital as part of a national newborn screening program to detect congenital hypothyroidism. However, there are many types of thyroid blood tests, and the specific type of that is done depends on each individual state's newborn screening practices. Each state also sets their own cutoffs for determining when the result should be interpreted as abnormal. For blood TSH, levels should normally be high in the first few days after birth, then fall to the adult range by two weeks of age. This paper examines the differences in thyroid screening programs in newborns.

THE FULL ARTICLE TITLE

Kilberg MJ et al 2018 Newborn Screening in the U.S. May Miss Mild Persistent Hypothyroidism. *J Pediatr* 192:204-8.

SUMMARY OF THE STUDY

In this study, the researchers contacted each of the newborn screening programs in all U.S. states and

territories to gather information on what type of thyroid blood testing is used to detect congenital hypothyroidism and how results are interpreted and followed up. They found that all programs test for congenital hypothyroidism within 1-3 days of birth. Some states would still run samples if collected after day 3 and, although older babies should have a lower TSH level, many of these programs did not adjust the result for the age of the baby.

Some programs test babies with blood TSH tests, others with T₄ tests followed by TSH tests, and still others with both of these. Among all programs, there were differences of when a TSH value is considered abnormal. The authors concluded that state newborn screening programs for congenital hypothyroidism vary widely in the U.S. In many of the programs, mild cases of congenital hypothyroidism may be missed.

WHAT ARE THE IMPLICATIONS OF THIS STUDY?

Newborn screening of congenital hypothyroidism is essential for diagnosing, treating and preventing the serious consequences of untreated congenital hypothyroidism. However, there are no national newborn screening standards in the United States and testing is done by each state. This study shows that states' newborn screening programs for congenital hypothyroidism in the United States vary widely and the testing approaches of some programs may potentially miss some cases of mild congenital hypothyroidism. The authors suggest that adopting the same approach in all congenital hypothyroidism screening programs across the United States would allow greater consistency of how babies with congenital hypothyroidism are diagnosed and treated.

— Angela M. Leung, MD, MSc





HYPOTHYROIDISM IN CHILDREN, continued

ATA THYROID BROCHURE LINKS

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

Congenital Hypothyroidism: <https://www.thyroid.org/congenital-hypothyroidism/>

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

ABBREVIATIONS & DEFINITIONS

Congenital: A term that refers to diseases present at birth.

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.

MARCH Medullary Thyroid Cancer *Awareness Month*



AMERICAN THYROID ASSOCIATION®
ATA | www.thyroid.org

