Screening for Congenital Hypothyroidism: U.S. Preventive Services Task Force Reaffirmation Recommendation Statement

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Summary of Recommendation & Evidence

The U. S. Preventive Services Task Force (USPSTF) recommends screening for congenital hypothyroidism (CH) in newborns. (This is a grade "A" recommendation.)

Rationale

Importance

Primary congenital hypothyroidism occurs in approximately 1 of every 3,000-4,000 newborns in the United States. In the absence of prompt diagnosis and treatment, most persons with this disorder will develop various degrees of neurological, motor and growth deficits, including irreversible mental retardation.

Detection

In the U.S., most state-based screening programs utilize serum thyroxine (T_4) and/or thyroid-stimulating hormone (TSH) performed on capillary blood collected from a heel stick and adsorbed onto filter paper.

Benefits of Detection and Early Intervention

Early detection of CH by neonatal screening and appropriate treatment substantially improves neurodevelopmental outcomes for affected persons.

Harms of Detection and Early Treatment

Positive test results, whether true positive or false positive, cause anxiety in parents. For some parents, this anxiety may be considerable.

USPSTF Assessment

The USPSTF concludes that there is high certainty that the net benefit is substantial.

Clinical Considerations

Patient Population under Consideration

This recommendation applies to all infants born in the U.S. Premature, very low birth weight and ill infants may benefit from additional screening because these

conditions are associated with decreased sensitivity and specificity of screening tests.

Screening Tests

Screening for CH is mandated in all 50 states and the District of Columbia, though methods of screening vary. There are two main methods used in the U.S.: Primary TSH with backup T_4; and primary T_4 with backup TSH. A few states use both tests in initial screening (1, 2). Clinicians should become familiar with the tests used in their area and the limitations of the employed screening strategy. For example, a primary TSH method may be falsely negative in low and very low birth weight infants with CH because of delayed elevation in TSH. Additionally, few states currently screen for centrally-mediated congenital hypothyroidism. Families should be provided with appropriate information about newborn screening tests, including the benefits and harms of screening. They should be aware of the potential of a false positive test, and the process required for definitive testing. Nationally, only 1 in 25 positive screening tests are confirmed to be CH (1). Normal newborn screening results for CH should not preclude appropriate evaluation of infants presenting with clinical signs and symptoms suggestive of hypothyroidism.

Timing of Screening. Infants should be tested between 2 and 4 days of age. Infants discharged from hospitals before 48 hours of life should be tested immediately before discharge. Specimens obtained in the first 24-48 hours of age may be falsely elevated for TSH regardless of the screening method used.

Treatment. Primary care clinicians should ensure that infants with abnormal screens receive confirmatory testing and begin appropriate treatment with thyroid hormone replacement within 2 weeks after birth. Children with positive confirmatory testing in whom no permanent cause of CH is found (such as lack of thyroid tissue on thyroid ultrasound or thyroid scan), should, at some time point after the age of 3 years, undergo a 30-day trial of reduced or discontinued thyroid hormone replacement therapy to determine if the hypothyroidism is permanent or transient.

Future Research Needs

Additional research is needed to determine the cost-benefit for different screening strategies, including the use of newer, more accurate TSH measurements, combined TSH-T_4 strategies, and methods designed to identify both primary and central hypothyroidism. Future research attentions should be directed to determining the incremental benefits of routine collection of a second specimen from two week olds. Additional research is also needed on how to ameliorate the affects of false positive results from CH and other newborn screening tests on families, such as improved communication plans for informing parents, better newborn screening informational materials, and reduced time to rule out congenital disorders.

Discussion

In 1996, the USPSTF reviewed the evidence for screening for CH in newborns and recommended screening (3). In 2006, the USPSTF performed a brief literature review and determined the benefits of screening for CH continue to be well established. This update included a search for new and substantial evidence on the benefits and harms of screening (4). The USPSTF found no new substantial evidence on the benefits and harms of screening for CH and therefore, reaffirms its recommendation that all newborns be screened for CH. The 1996 recommendation statement, the 1996 evidence report, and the summary of the updated literature search can be found at http://www.preventiveservices.ahrq.gov.

Recommendations from Other Groups

The American Academy of Pediatrics (AAP) and the American Academy of Family Physicians recommend universal newborn screening for congenital hypothyroidism (2, 5). The AAP, in conjunction with the American Thyroid Association, and the Lawson Wilkins Pediatric Endocrine Society recently published guidelines for screening and treatment for congenital hypothyroidism (2).

References

1. National Newborn Screening Information System. http://www2.uthscsa.edu/nnsis/ Accessed October 30, 2006.
2. American Academy of Pediatrics. Update of Newborn Screening and Therapy for Congenital Hypothyroidism Pediatrics. 2006 June 2006;117(6):2290-303.
3. U.S. Preventive Services Task Force. Screening for Congenital Hypothyroidism. Guide to Clinical Preventive Services, 2nd edition. Alexandria, VA: International Medical Publishing; 1996. p. 503-7.
4. Meyers D, Haering S. Screening for Congenital Hypothyroidism: A Literature Update for the US Preventive Services Task Force. 2006. AHRQ Publication 08-05109-EF-1. Rockville, MD: Agency for Healthcare Research and Quality, 2008. http://www.ahrq.gov/clinic/uspstf/uspscghy.htm.
5. American Academy of Family Physicians. Policy Statement on Newborn Screening. Issue Brief. 2006;2006(October 5):1-12.

U.S. Preventive Services Task Force

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TABLE 1

What the USPSTF Grades Mean and Suggestions for Practice

Grade	Grade Definitions	Suggestions for Practice
A	The USPSTF recommends the service. There is high certainty that the net benefit is substantial.	Offer/provide this service.
B	The USPSTF recommends the service. There is high certainty that the net benefit is moderate or there is moderate certainty that the net benefit is moderate to substantial.	Offer/provide this service.
C	The USPSTF recommends against routinely providing the service. There may be considerations that support providing the service in an individual patient. There is moderate or high certainty that the net benefit is small.	Offer/provide this service only if there are other considerations in support of the offering/providing the service in an individual patient.
D	The USPSTF recommends against the service. There is moderate or high certainty that the service has no net benefit or that the harms outweigh the benefits.	Discourage the use of this service.
I Statement	The USPSTF concludes that the current evidence is insufficient to assess the balance of benefits and harms of the service. Evidence is lacking, of poor quality or conflicting, and the balance of benefits and harms cannot be determined.	Read “Clinical Considerations” section of USPSTF Recommendation Statement. If offered, patients should understand the uncertainty about the balance of benefits and harms.

TABLE 2

USPSTF Levels of Certainty Regarding Net Benefit

Definition: The U.S. Preventive Services Task Force defines certainty as **"**likelihood that the USPSTF assessment of the net benefit of a preventive service is correct**".** The net benefit is defined as benefit minus harm of the preventive service as implemented in a general, primary care population. The USPSTF assigns a certainty level based on the nature of the overall evidence available to assess the net benefit of a preventive service.

Level of Certainty	Description
High	The available evidence usually includes consistent results from well-designed, well-conducted studies in representative primary care populations. These studies assess the effects of the preventive service on health outcomes. This conclusion is therefore unlikely to be strongly affected by the results of future studies.
Moderate	The available evidence is sufficient to determine the effects of the preventive service on health outcomes, but confidence in the estimate is constrained by factors such as: - the number, size, or quality of individual studies; - inconsistency of findings across individual studies; - limited generalizability of findings to routine primary care practice; or - lack of coherence in the chain of evidence. As more information becomes available, the magnitude or direction of the observed effect could change, and this change may be large enough to alter the conclusion.
Low	The available evidence is insufficient to assess effects on health outcomes. Evidence is insufficient because of: - the limited number or size of studies; - important flaws in study design or methods; - inconsistency of findings across individual studies - gaps in the chain of evidence; - findings not generalizable to routine primary care practice; or - a lack of information on important health outcomes. More information may allow an estimation of effects on health outcomes.

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