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Screening for Congenital Hypothyroidism: US Preventive Services Task Force Reaffirmation Recommendation Statement

US Preventive Services Task Force

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- The US Preventive Services Task Force (USPSTF) makes recommendations about preventive care services for patients without recognized signs or symptoms of the target condition.
- Recommendations are based on a systematic review of the evidence of the benefits and harms and an assessment of the net benefit of the service.
- The USPSTF recognizes that clinical or policy decisions involve more considerations than this body of evidence alone. Clinicians and policy-makers should understand the evidence but individualize decision-making to the specific patient or situation.

SUMMARY OF RECOMMENDATION & EVIDENCE

The US 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 US, most state-based screening programs utilize serum thyroxine (T₄) 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 of screening for congenital hypothyroidism in newborns is substantial.

CLINICAL CONSIDERATIONS

Patient Population Under Consideration

This recommendation applies to all infants born in the US. 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 US: Primary TSH with backup T₄; and primary T₄ 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-T₄ strategies, and methods designed to identify both primary and central hypothyroidism. Future research 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

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* Members of the Task Force at the time this recommendation was finalized. For a list of current Task Force members, go to www.ahrq.gov/clinic/uspstfab.htm.

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