

## CLINICAL INQUIRIES

From the  
Family Physicians  
Inquiries Network

# What are appropriate screening tests for infants and children?

## EVIDENCE-BASED ANSWER

There is adequate evidence for screening neonates for hemoglobinopathies, congenital hypothyroidism, phenylketonuria (strength of recommendation [SOR]: **A**), and cystic fibrosis (SOR: **B**). Vision screening should be done for those younger than age 5 years (SOR: **B**). High-risk children should be tested for tuberculosis (TB) (SOR: **B**) and lead

toxicity (SOR: **B**). Few data exist to guide frequency and timing of these screening tests, so the following timing recommendations are based on consensus opinion (SOR: **C**): test for visual acuity yearly starting at age 3 years; test for TB and lead once between the ages of 9 and 12 months, and repeat for high risk or exposure.

## CLINICAL COMMENTARY

### Obtain family history; order additional screening tests if history suggests them

Why do states differ so much in the neonatal screening tests that they routinely perform? Some states screen for only a few genetic diseases, others for more than 40. Most states do neonatal hearing screening despite limited evidence of utility, while only one quarter of all states have neonatal cystic fibrosis screening programs, a condition for which there is probably better evidence for screening. While we might like to think that good science alone would dictate screening policy, the

economic circumstances of each state, variable interpretation/quality of the research reviewed, and legislative priorities (among many reasons) probably play at least as much a roll. For any test, its accuracy is only as good as the pretest probability of the disease for which it is being used. Our yield for cystic fibrosis screening will be higher in families with a history of cystic fibrosis. This is the key point—you still need to obtain a family history and order additional screening tests if the history suggests them.

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### Evidence summary

There are many opinions and recommendations about what constitutes quality health surveillance for children. However, many screening tests for children lack evidence of effectiveness and information on harms.<sup>1</sup> The scope of this question required use of evidence published in high-quality systematic reviews. The US Preventive Services Task Force (USPSTF) provides the

most rigorous evidence on which to base recommendations.<sup>2</sup> Medline was searched for any additional individual studies of interest. The USPSTF has conducted reviews for selected screening tests for children; the **TABLE** summarizes those with sufficient evidence to recommend them. We identified 1 additional evidence-based recommendation from the Centers for Disease Control and Prevention. This

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**TABLE**

**US Preventive Services Task Force evidence-supported testing for children**

TEST (SOR)	POPULATION	USPSTF COMMENTS	AAFP	AAP
<b>UNIVERSAL SCREENING</b>				
Neonatal hemoglobinopathy (A)	Newborns		Strongly recommends	Recommends once at 2 to 4 days of life, but before age 1 month
Neonatal phenylketonuria (A)	Newborns—repeat at 2 weeks if <24 hrs old at discharge		Strongly recommends	
Congenital hypothyroidism (A)	Newborns		Strongly recommends	
Vision screening for strabismus, amblyopia, and refractive error (B)	Before age 5	Type of screening tests vary with age; evidence inadequate to recommend specific test	Recommends	Start objective testing yearly at age 3
<b>HIGH-RISK SCREENING</b>				
PPD test for tuberculosis (A)	Children at high-risk for TB	Risks for TB: HIV, close contacts of persons with TB, immigrants from countries with high TB prevalence, low income populations, and residents of long-term care facilities	Strongly recommends screening high-risk children	Screen high-risk children at 12 months and or upon recognition of high-risk factors
Lead toxicity (B)*	Infants at risk at 12 months of age at risk for lead exposure	Risks for lead†	Screen infants at high risk	Screen high-risk infants at 9 to 12 months. Repeat at age 24 months for those at high-risk
<p>* This document is currently being updated; the recommendation may or may not change.                      † Risk are living in a house older than 1950 with peeling paint or remodeling, living near heavy traffic or lead industry, living with someone who has elevated lead levels or whose job/hobby involves lead exposure, using lead-based pottery, or taking remedies that contain lead.                      AAFP: American Academy of Family Practice recommendations from: <a href="http://www.aafp.org/PreBuilt/RCPS_August2005.pdf">www.aafp.org/PreBuilt/RCPS_August2005.pdf</a>.                      AAP: American Academy of Pediatrics recommendations from: <a href="http://aappolicy.aappublications.org/cgi/content/full/pediatrics;105/3/645">aappolicy.aappublications.org/cgi/content/full/pediatrics;105/3/645</a>.</p>				

report, based on a systematic review, recommends cystic fibrosis screening in neonates based on moderate benefits and low risks of harm.<sup>3</sup>

The **TABLE** summarizes the evidence supporting universal childhood screening for hemoglobinopathies, congenital hypothyroidism, phenylketonuria, and visual defects; and high-risk childhood screening for tuberculosis and lead toxicity. The **TABLE** also lists the recommendations from the American Academy of Pediatrics

(AAP) on frequency and timing of screening as guided by consensus opinion.

The USPSTF recommendations supporting screening for hemoglobinopathies, congenital hypothyroidism, and phenylketonuria are considered standard of care. The USPSTF believes that updating these 1996 recommendations would have little impact on clinical practice.

The USPSTF recommendations supporting vision screening found no direct evidence supporting screening for visual

**FAST TRACK****USPSTF's recommendation on screening for lead levels is being revised and may change**

acuity. One fair-quality controlled study (N=3490) showed a decreased prevalence of amblyopia in the screened group and evidence that treatment of amblyotic risk factors prevents amblyopia. A Cochrane review of this topic showed insufficient evidence for visual screening of older (school-aged) children; for amblyopia, no data sufficient for analysis was found.<sup>4,5</sup>

The USPSTF recommendation to screen asymptomatic high-risk children for TB is based on the effectiveness of early intervention (14 controlled trials) and the accuracy of the Mantoux test.

The USPSTF document on screening for lead levels is currently being revised and the recommendation may change. Although no controlled studies directly show that screening high-risk children for lead exposure improves clinical outcomes, several lesser-quality studies create a logical path to this conclusion.

The USPSTF finds there is insufficient evidence to recommend for or against performing the following screening tests in children: blood pressure screening; screening for overweight in children and adolescents; and iron deficiency screening in asymptomatic infants. Both Cochrane Systematic Reviews and USPSTF found insufficient evidence to support universal hearing screening, including neonatal hearing screening.<sup>6</sup> The USPSTF makes no recommendation regarding screening high-risk children for hyperlipidemia.

The USPSTF recommends that the following tests should not be performed in children because there is good evidence that the harms outweigh the benefits: thyroid cancer screening in children and bacteriuria screening in asymptomatic nonpregnant children.

**Recommendations from others**

There are numerous guidelines recommending various sets of preventive services for children, but there are few evidence-based recommendations. The AAP recommendations can be found in *Guidelines for Health Supervision III*.<sup>7</sup> The AAP also publishes policy statements and guidelines in the journal *Pediatrics*. The American Academy of Family Practice's (AAFP) recommendations on health supervision can be found at: [www.aafp.org/PreBuilt/RCPS\\_August2005.pdf](http://www.aafp.org/PreBuilt/RCPS_August2005.pdf).

A summary of the AAFP and the AAP recommendations on each of the USPSTF supported tests is in the **TABLE**. While AAFP and USPSTF recommendations concur, AAP recommendations differ in recommending hearing screening for all newborns, iron deficiency screening at 9 months of age, screening for lipid disorders in children at risk starting at 24 months, and screening urinalysis at age 5 years.

**REFERENCES**

1. Moyer VA, Butler M. Gaps in the evidence for well-child care: A challenge to our profession. *Pediatrics* 2004; 114:1511-1521.
2. US Preventive Services Task Force. Guide to Clinical Preventive Services [website]. Available at: [www.ahrq.gov/clinic/cps3dix.htm](http://www.ahrq.gov/clinic/cps3dix.htm). Accessed on June 28, 2006.
3. Grosse SD, Boyle CA, Botkin JR, et al. Newborn screening for cystic fibrosis: evaluation of benefits and risks and recommendations for state newborn screening programs. *MMWR Recomm Rep* 2004; 53(RR-13):1-36.
4. Powell C, Porooshani H, Bohorquez MC, Richardson S. Screening for amblyopia in childhood. *Cochrane Database Syst Rev* 2005; (3):CD005020.
5. Powell C, Wedner S, Richardson S. Screening for correctable visual acuity deficits in school-age children and adolescents. *Cochrane Database Syst Rev* 2005; (1):CD005023.
6. Puig T, Municio A, Meda C. Universal neonatal hearing screening versus selective screening as part of the management of childhood deafness. *Cochrane Database Syst Rev* 2005; (2):CD003731.
7. *Guidelines for Health Supervision III*. Elk Grove Village, Ill: American Academy of Pediatrics; 1997. (Updated 2002).