Guidelines for DBS screening: Newborn Screening Expands: Recommendations for Pediatricians and Medical Homes – Implications for the System

The American Academy of Pediatrics (AAP) believes that the medical care of infants, children, and adolescents ideally should be accessible, continuous, comprehensive, family centered, coordinated, compassionate, and culturally effective. It should be delivered or directed by well-trained physicians who provide primary care and help to manage and facilitate essentially all aspects of pediatric care. The physician should be known to the child and family and should be able to develop a partnership of mutual responsibility and trust with them. These characteristics define the "medical home."

2009 clinical report in *Pediatrics* from the Committee on Practice and Ambulatory Medicine the Section on Otolaryngology–Head and Neck Surgery, which focuses on the JCIH guidelines for medical homes


In June 2008, a panel of experts met in Como, Italy at the NHS 2008 Conference to develop Guidelines for the Identification and Management of Infants and Young Children with Auditory Neuropathy.

The conference was sponsored by the Bill Daniels Center for Children’s Hearing – Children’s Hospital Colorado. Deborah Hayes, Ph.D., Co-Chair of the Bill Daniels Center for Children’s Hearing, moderated the conference.

“Auditory neuropathy” is a relatively recent clinical diagnosis used to describe individuals with auditory disorders due to dysfunction of the synapse of the inner hair cells and auditory nerve, and/or the auditory nerve itself. Unlike patients with sensory hearing loss who show clinical evidence of impaired outer hair cell function, patients with auditory neuropathy show clinical evidence of normally functioning outer hair cells. Individuals with auditory neuropathy typically demonstrate impaired speech understanding, and show normal to severely impaired speech detection and pure tone thresholds. It has been shown that auditory neuropathy affects an individual’s ability to process rapidly changing acoustic signals, known as auditory temporal processing.

The 2009 Clinical and Laboratory Standards Institute (CLSI) guidelines, Newborn Screening for Preterm, Low Birth Weight, and Sick Newborns; Approved Guideline (I/LA31-A), focuses on the best practices for NBS of preterm and other at-risk newborns in SCBUs. State NBS and international programs may be reviewing their current protocols in light of this recommendation, as it has been well received as good practice.
Provider Web Sites & Tools

ACMG ACTion sheets

American College of Medical Genetics ACTion sheets and confirmatory algorithms for DBS screening

EHDI algorithm, page 1 (flowchart)

Early Hearing Detection and Intervention (EHDI) Algorithms for infant hearing screening

EHDI algorithm, page 2 (customizable referral resource)

Medical Home Portal

Reliable information and resources to help physicians and parents care for children and youth with special health-care needs (CYSHCN)

National Center for Medical Home Implementation

Resource for health professionals, families, and anyone interested in creating a medical home for all children and youth

The Medical Home Toolkit was developed by the Center for Medical Home Improvement and the National Center for Medical Home Implementation.

Medical Home Toolkit:

Medical Home Action Plan

Tool for providing a Medical Home Action Plan

Family Centered Care Coordination Assessment

Tool to assess your family-centered care

Care Co-Management Among Patient/Family, Pediatric Medical Home and Specialists

Tool to enhance communication and sharing of care and knowledge among families and professionals

Child/Adolescent Health Assessment

Screening questionnaire to improve patient care

Pediatric Care Plan

Tool to document the medical summary, emergency information, and the child’s care plan.

NCCRCG

The National Coordinating Center for the Genetic and Newborn Screening Service Collaboratives (NCC). NCC focuses on bringing quality genetic and newborn screening (NBS) services to local communities, and building bridges between public health, primary care/Medical Home, geneticists and other specialists, and families and consumers.

NCHAM

National Center for Hearing Assessment and Management

NCHAM state map

National Center for Hearing Assessment and Management Web site provides contact information for the state programs and state legislation, guidelines, Web site and family educational materials

NCHAM State Map

National Center for Hearing Assessment and Management State Map

NNSGRC State Map

National Coordinating Center for the Genetic and Newborn Screening Service Collaboratives State Map provides information and resources in the area of newborn screening and genetics to benefit health professionals, the public health community, consumers and government officials

State EHDI/UNHS Mandates: Summary Table

MCHB/ HRSA

Maternal and Child Health Bureau – Health Resources and Services Administration: Newborn Screening

Newborn screening and genetics public health infrastructure initiatives

The Genetic Alliance Newborn Screening Clearinghouse (Baby’s First Test)

Baby’s First Test – Newborn Screening: The Facts

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Newborn Screening Facts

- A brief introduction to the most important information on newborn screening

Genetics and Family History

- [http://www.babysfirsttest.org/about-newborn-screening/genetics](http://www.babysfirsttest.org/about-newborn-screening/genetics) Information on why it is important for all babies to have newborn screening, even if there isn’t a family history of a condition, and how genetic conditions are inherited

Screening Resources

- A brief description and links to several government and nonprofit agencies working to improve health through newborn screening

Conditions Screened by State

- [http://www.babysfirsttest.org/](http://www.babysfirsttest.org/) Every state has slightly different requirements for newborn screening. This section reviews factors that influence how states choose their screening panels and how to find out more information.

AHRQ – CAHPS

Agency for Healthcare Research and Quality Consumer Assessment of Healthcare Providers and Systems (CAHPS)

The CAHPS program develops and supports the use of a comprehensive and evolving family of standardized surveys that ask consumers and patients to report on and evaluate their experiences with health care. These surveys cover topics that are important to consumers, such as the communication skills of providers and the accessibility of services.

CMHI Family/Caregiver Survey

Center for Medical Home Index – The Medical Home Index (MHI) is a validated self-assessment and classification tool designed to translate the broad indicators defining the medical home (accessible, family-centered, comprehensive, coordinated, etc.) into observable, tangible behaviors and processes of care within any office setting. It is a way of measuring and quantifying the "medical homeness" of a primary care practice.

Family-Centered Care Self-Assessment Tools from Family Voices:

- Family Tool
- Provider Tool
- User Guide

Family-centered care is a key aspect of quality in health care for children, youth, and their families.

These tools are designed to:
1. Increase outpatient health-care settings' and families' awareness about the implementation of family-centered care and,
2. Provide an organized way for health-care settings to assess current areas of strength and identify areas for growth, plan future efforts and to track progress.

Medical Home Learning Collaborative:

- Comprehensive Care Planning

Information about the essentials of comprehensive care planning for children with special health-care needs (CSHCN). Three distinct types of documents present medical information plans, emergency plans, and working (action) care plans. When combined appropriately for CSHCN (based upon need), these tools make up a comprehensive care plan.

National Athletic Trainers’ Association:

- Consensus Statement: Sickle Cell Trait and the Athlete

In a recent review of nontraumatic sports deaths in high school and college athletes (1), the top four killers, in order of occurrence, were: cardiovascular conditions, hyperthermia (heatstroke), acute rhabdomyolysis tied to sickle cell trait, and asthma. Acute exertional rhabdomyolysis (explosive muscle breakdown) from sickle cell trait is the least understood of these conditions. The
purpose of this Task Force is to raise awareness of this condition and provide measures to reduce the risk of exertional collapse related to sickle cell trait.

A Fact Sheet From the AAP and HRSA

In the United States, sickle cell disease occurs in approximately 1 in 2,500 newborns. It is more prevalent than any other condition identified by newborn blood screening.

Template to create a Newborn Screening Refusal Waiver

This new tool and resource kit provides materials for health supervision care from infancy through adolescence. It is designed to accompany and support Bright Futures: Guidelines for Health Supervision of Infants, Children, and Adolescents, Third Edition.

Postpartum depression is the most common complication of childbearing. The 10-question Edinburgh Postnatal Depression Scale (EPDS) is a valuable and efficient way of identifying patients at risk for perinatal depression. The EPDS is easy to administer and has proven to be an effective screening tool.

This handout addresses the commitment to health promotion and disease prevention in partnership with community members and health and education professionals.

Tool to help determine where your practice is in relation to community pediatrics activities.

Checklist to help you understand linkages you have developed and where gaps remain. It also contains a list of possible community resources.

This one-page handout is designed to help families by providing information on topics such as family readiness, feeding, safety, and routine baby care. This age-appropriate form also offers families information about what to expect at the next well-child visit, the 2- to 5-day (first week) visit, to help families prepare a list of questions and concerns for their health care provider.

Tool to be completed by the parent or guardian; it covers essential medical screening questions and parental concerns and questions about the newborn and home environment.

Questionnaire to be used to elicit family concerns about their infants health care

Available in English and Spanish. This tool summarizes critical information parents need in the first few days following a newborn’s discharge from the nursery. It can be adapted to meet your practice needs. Note: These tools are also useful when discussed and given to parents during the prenatal visit about what to expect when the baby is born.

NPR’s Health Blog – Pediatricians Need To Help ‘Sad Dads’

“Fathers who are depressed are far more likely to spank their 1-year-olds than dads who aren’t.”

Patient- and family-centered care is an approach to the planning, delivery, and evaluation of health care that is grounded in mutually beneficial partnerships among health-care providers, patients, and families. It redefines the relationships in health-care.
Risk Factors for Hearing Loss


APPENDIX 2: Risk Indicators Associated With Permanent Congenital, Delayed-Onset or Progressive Hearing Loss in Childhood

Family Handouts and Web Sites

AAP HealthyChildren.org

HealthyChildren.org is the only parenting Web site backed by 60,000 pediatricians committed to the attainment of optimal physical, mental, and social health and well-being for all infants, children, adolescents, and young adults.

Purpose of Newborn Hearing Screening

Babies learn from the time they are born. One of the ways they learn is through hearing. If they have problems with hearing and do not receive the right treatment and early intervention services, babies will have trouble with language development.

Listen-Up-About-Why-Newborn-Hearing-Screening-is-Important

Newborn hearing screening focuses on identifying hearing loss early. Catching problems sooner rather than later can make a big difference in a child's development.

Centers for Disease Control and Prevention

CDC has FREE brochures, posters, fact sheets, and more for parents, health-care providers, and public health professionals.

NCAA – A Fact Sheet for Students and Athletes – Sickle Cell Trait

Use this fact sheet to advise students and athletes of what they need to know about sickle cell trait.

Card File Registry

Card registry system based on one detailed by John Oldham and Michele Maunder. Oldham J, Maunder M. Who are your patients? Patient information in practice planning and performance improvement. Manag Care Q. 1999 Summer;7(3):35-44. Knowledge of patients above the individual level is important for any quality improvement effort in the office practice, and for informing decisions for the organization's strategic direction on service delivery. Pivotal in this knowledge is the development of a registry of patients for an office, and understanding of how that is linked to improvement methods.

Boys Town My Baby's Hearing

Boys Town National Research Hospital Web site in English and Spanish, My Baby's Hearing, for families with questions about newborn hearing screening and for families with infants identified with a hearing impairment. Includes downloadable pdf resources for professionals to use for family education purposes.

Hands and Voices

A parent-driven, parent/professional collaborative group for deaf and hard-of-hearing children.

Consumer resource section of the NCCRCG Regional Collaborative

The NCC provides resources to assist consumers and their families in accessing information about genetic services and resources at a national level.

Monaco family story

StephenMonaco.org Web site:

Stephen Angelo Monaco was born on October 26, 1997. Due to lack of comprehensive newborn screening, Stephen’s inherent metabolic disorder, Isovaleric Acidemia, went undetected, and he appeared to be a normal, healthy child. At age three and a half, within a 24-hour period, he suffered a life-threatening metabolic crisis that resulted in severe brain damage, leaving him as a severely disabled child with complicated medical issues.
Every state in the nation now has a newborn hearing screening program, and 2007 data from the Centers for Disease Control and Prevention showed that 97% of newborns in the United States were screened for hearing loss. However, 46.1% infants who failed their hearing screening did not receive a diagnostic evaluation and are considered "lost to follow-up" (LTF). The document *Loss to Follow-Up in Early Hearing Detection and Intervention*, developed by the ASHA Working Group on Loss to Follow-Up, includes a systematic review of the evidence related to LTF and concludes that the available evidence provides neither meaningful direction in identifying the patients/families at highest risk for LTF nor in decreasing that risk (ASHA, 2008).


A briefing paper from the Secretaries Advisory Committee on Hereditable Disorders in Newborns and Children.

Newborn screening programs are state-based with variable policies. Guidance regarding the retention, storage, and use of portions of newborn screening dried blood spots that remain after screening (residual specimens) was first published in 1996. Since then, newborn screening programs have paid increased attention to specimen storage and usage issues.

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Fathers' Depression Related to Positive and Negative Parenting Behaviors With 1-Year-Old Children

WHAT'S KNOWN ON THIS SUBJECT: Paternal depression affects fathers' interactions with their children. However, little is known regarding the association between paternal depression in fathers of young children and specific parenting behaviors commonly discussed at well-child visits.

WHAT THIS STUDY ADDS: Depressed fathers were nearly four times more likely to report spanking and less than half as likely to report consistently reading to their 1-year-old children. Notably, 77% of depressed fathers reported talking with their child’s doctor in the previous year.PEDIATRICS; originally published online March 14, 2011.

The clinical report from the Committee on Practice and Ambulatory Medicine the Section on Otolaryngology – Head and Neck Surgery Congenital or acquired hearing loss in infants and children has been linked with lifelong deficits in speech and language acquisition, poor academic performance, personal-social maladjustments, and emotional difficulties. Identification of hearing loss through neonatal hearing screening, regular surveillance of developmental milestones, auditory skills, parental concerns, and middle-ear status and objective hearing screening of all infants and children at critical developmental stages can prevent or reduce many of these adverse consequences. This report promotes a proactive, consistent, and explicit process for the early identification of children with hearing loss in the medical home. An algorithm of the recommended approach has been developed to assist in the detection and documentation of, and intervention for, hearing loss.

PEDIATRICS. 2009;124:1252–1263

Ethical Issues With Genetic Testing in Pediatrics

From the AAP Committee on Bioethics:

This statement reviews considerations for the use of genetic technology for newborn screening, carrier testing, and testing for susceptibility to late-onset conditions. Recommendations are made...
promoting informed participation by parents for newborn screening and limited use of carrier testing and testing for late-onset conditions in the pediatric population. Additional research and education in this developing area of medicine are encouraged.

**PEDIATRICS. 2001, June; Vol. 107, No. 6**

The aims of this study were to determine parent and provider knowledge and awareness of newborn screening; to gather opinions from parents, providers, and newborn screening professionals about the content and timing of newborn screening education; and to use consensus data to formulate recommendations and to develop educational materials for parents and providers.

**PEDIATRICS. 2006, May; Vol. 117, Num. 5,**

Sickle cell trait or the carrier state for sickle cell anemia occurs in high frequency among people of African-American and/or Hispanic descent, but it can also occur in people of all ethnicities. For nearly three decades, screening for sickle cell disease and related hemoglobinopathies has been part of state-mandated universal newborn screening programs in the United States. When properly conducted, testing for hemoglobinopathies also detects the carrier state for sickle cell disease and other hemoglobinopathies. These results have lifelong validity and for most people testing does not need to be repeated.

*The Medical and Research Advisory Committee (MARAC) of the Sickle Cell Disease Association of America, Inc. in collaboration Key Opinion Leaders and Federal partners developed carrier screening recommendations from proceeding from meetings convened on December 17, 2009 and February 18, 2010. The MARAC 2009 – 2010 roster is listed in Appendix B.*

**Screening U.S. College Athletes for Their Sickle Cell Disease Carrier Status**

This briefing paper outlines issues surrounding the screening of young athletes for their sickle cell disease carrier status (or sickle cell trait), a genetic condition. The Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) has developed the report to advise the Secretary of the U.S. Department of Health and Human Services about the rule of the National Collegiate Athletic Association requiring testing for sickle cell trait in all incoming Division I student athletes.

**SECRETARY’S ADVISORY COMMITTEE ON HERITABLE DISORDERS IN NEWBORNS AND CHILDREN**

Learn more about new methods for screening newborns from the leading experts in pediatrics whose insight and expertise make PREP Audio the most relevant audio journal available. Whether you’re in the car or in the gym, you’ll learn from the experts when you listen to each audio issue.

**Data Standards for Electronic Reporting from the U.S. National Library of Medicine**

*The index was originally published in November 2003 in Medical Home Crosswalk To Reimbursement. The information was*

**AAP Medical Home Coding Fact Sheet**

**Coding Resources**

**Newborn Screening Coding and Terminology Guide:**

**AAP Bookstore**

**PREP AUDIO JUNE 2007: THE NEW NEWBORN SCREENING**

**Sickle Cell Disease Association of America, Inc. Carrier Screening Recommendations**
developed by Margaret McManus, Alan Kohrt, Joel Bradley, and Linda Walsh in collaboration with the Center for Medical Home Improvement, the American Academy of Pediatrics, and the National Institute for Children’s Healthcare Quality. Funded by the Maternal and Child Health Bureau, U.S. Department of Health and Human Services through the Maternal and Child Health Policy Research Center.

Payment and Finance

Section of the National Center for Medical Home Implementation Web site

Strategies for Billing, Coding and Getting Paid Appropriately – A Guide for Family Physicians,

Each January brings changes to every physician office – there are always new and deleted billing codes (CPT and HCPCS II), new deductible amounts to be collected from patients, new and revised billing requirements from health insurance plans, and sometimes new payment amounts from third-party payers. It can be challenging for a medical office staff to be aware of all these changes and ready to adapt and implement them in your practice management procedures and the daily work flow of your office.

AAP Coding for Pediatric Preventive Care 2012

This quick reference booklet includes guidance on how to properly code for pediatric preventive medicine.

Developmental Screening Coding Fact Sheet

Developmental screening, surveillance, and assessment are often complemented by the use of special tests, which vary in length. This coding fact sheet provides guidance on how pediatricians can appropriately report limited and extended developmental screening and testing services.

Early Hearing Detection and Intervention (EHDI) Coding Fact Sheet

This Coding Fact Sheet provides a guide to coding for pediatric hearing screening. Strategies and a template letter for pediatric practices to handle payer denials and contractual issues are provided in “Denial Management and Contract Negotiation for Hearing Screening Services”

For more resources on coding, visit AAP Practice Management Online or contact the AAP Coding Hotline at 800/433-9016 extension 4022 or aapcodinghotline@aap.org

Improvement Tools

All Ideas for Closing Gaps

Tool to help team plan PDSA cycles

Improvement Planning Worksheet

Print version of tool to collect and enter patient data

NBS Data Collection Tool

Grid of all measures for NBS, including calculations

NBS Measures

Survey to assess current office procedures for NBS

NBS Practice Survey

NBS Tools

List of tasks to help staff confirm/document NBS milestones

NBS Checklist

Customizable NBS office procedure document

NBS Office Protocol

The ACT sheets and algorithms are designed primarily as educational resources for physicians to help them provide quality medical services, and adherence to them does not necessarily ensure a successful medical outcome. The ACT sheets and algorithms should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the
health-care provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient’s record the rationale for any significant deviation from these ACT sheets and algorithms.