Newborn Hearing Screening Programs and Their Impact on Early Usher Diagnosis

Presented at

First International Symposium on Usher Syndrome and Related Disorders

Omaha, Nebraska

by

Karl R. White

National Center for Hearing Assessment and Management

www.infanthearing.org

Universal Newborn Hearing Screening is not a new idea…

"[There is] an urgent need to study further and more critically methods of testing hearing in young children . . . during this first year the existence of deafness needs to be ascertained . . . training needs to be begun at the earliest age that the diagnosis of deafness can be established."

Ewing IR, Ewing AWG. 1944. The ascertainment of deafness in infancy and early childhood. ***The Journal of Laryngology and Otology*** 59:309-333.

What I Hope You Will Remember From This Presentation

1. A rising tide lifts all ships
2. Education is important, but expensive
3. Science and advocacy can and should be partners
4. Lessons learned from newborn hearing screening


Montreal School
For the Deaf
Group 4 1977-78

Spring is my favorite season. The sun shines bright. The flowers begin to grow. I like spring.

What enabled us to move from ….

There → to → Here?

Earlier Identification of Hearing Loss

Availability of Better Assistive Listening Devices

High Quality Early Intervention Programs that focus on teaching LANGUAGE

Advocacy and Public Policy Initiatives

Why is Early Identification of Hearing Loss so Important?

- Hearing loss occurs more frequently than any other condition for which population-based screening is done

Frequency of Congenital Hearing Loss?

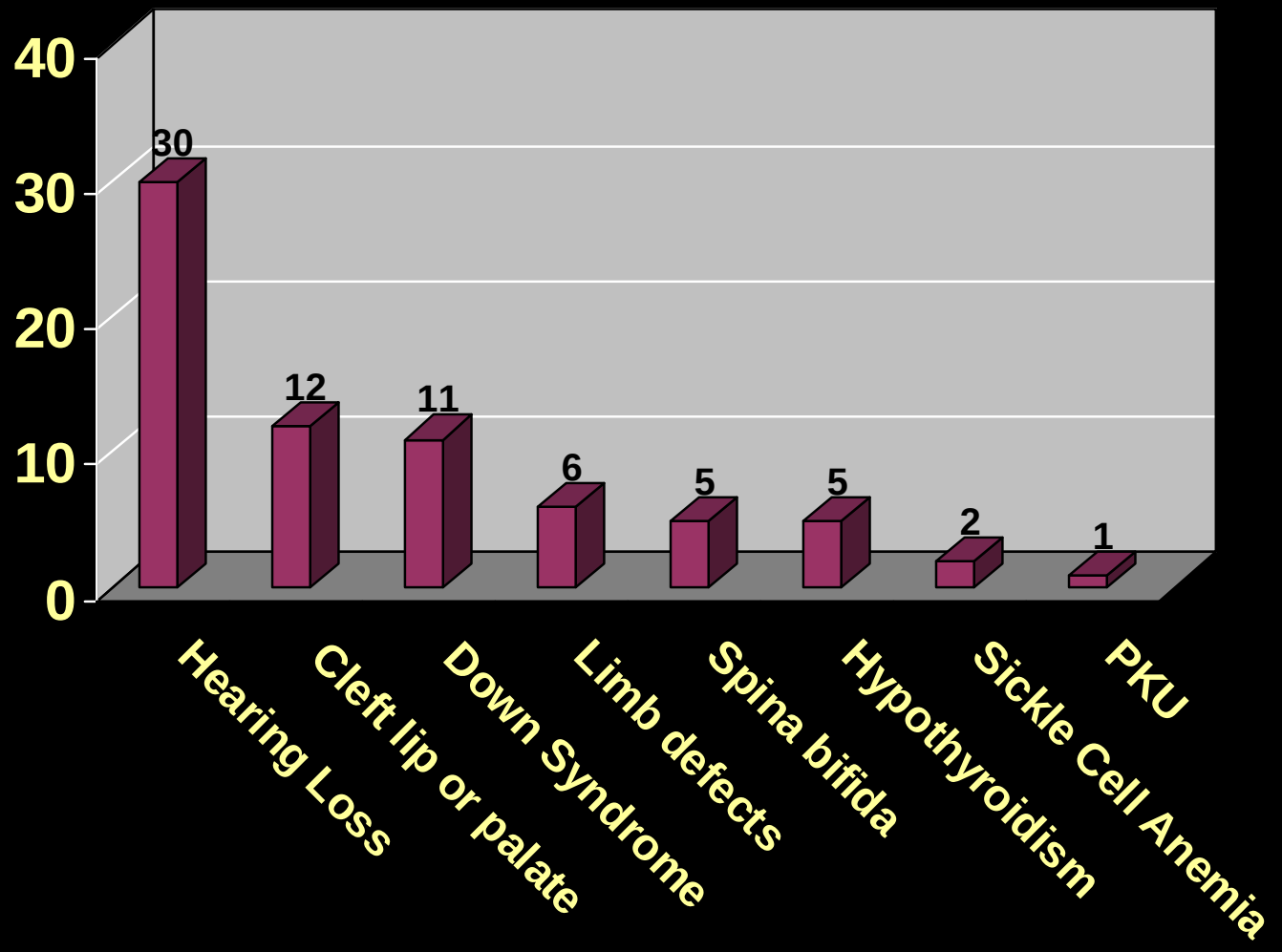
- **1 per 1,000**
- **2 per 1,000**
- **3 per 1,000**
- **6 per 1000**

Rate Per 1000 of Permanent Childhood Hearing Loss in EHDI Programs

Site	Sample Size	Prevalence Per 1000
Rhode Island (3/93 - 6/94)	16,395	1.71
Colorado (1/92 - 12/96)	41,976	2.56
New York (1/96 - 12/96)	27,938	1.65
Utah (7/93 - 12/94)	4,012	2.99
Hawaii (1/96 - 12/96)	9,605	4.15
Massachussets (1/04 – 12/04)	78,515	2.87

Adapted from White KR (2003). The current status of EHDI programs in the United States. *Mental Retardation and Developmental Disabilities Research Reviews, 9*(2), 79-88.

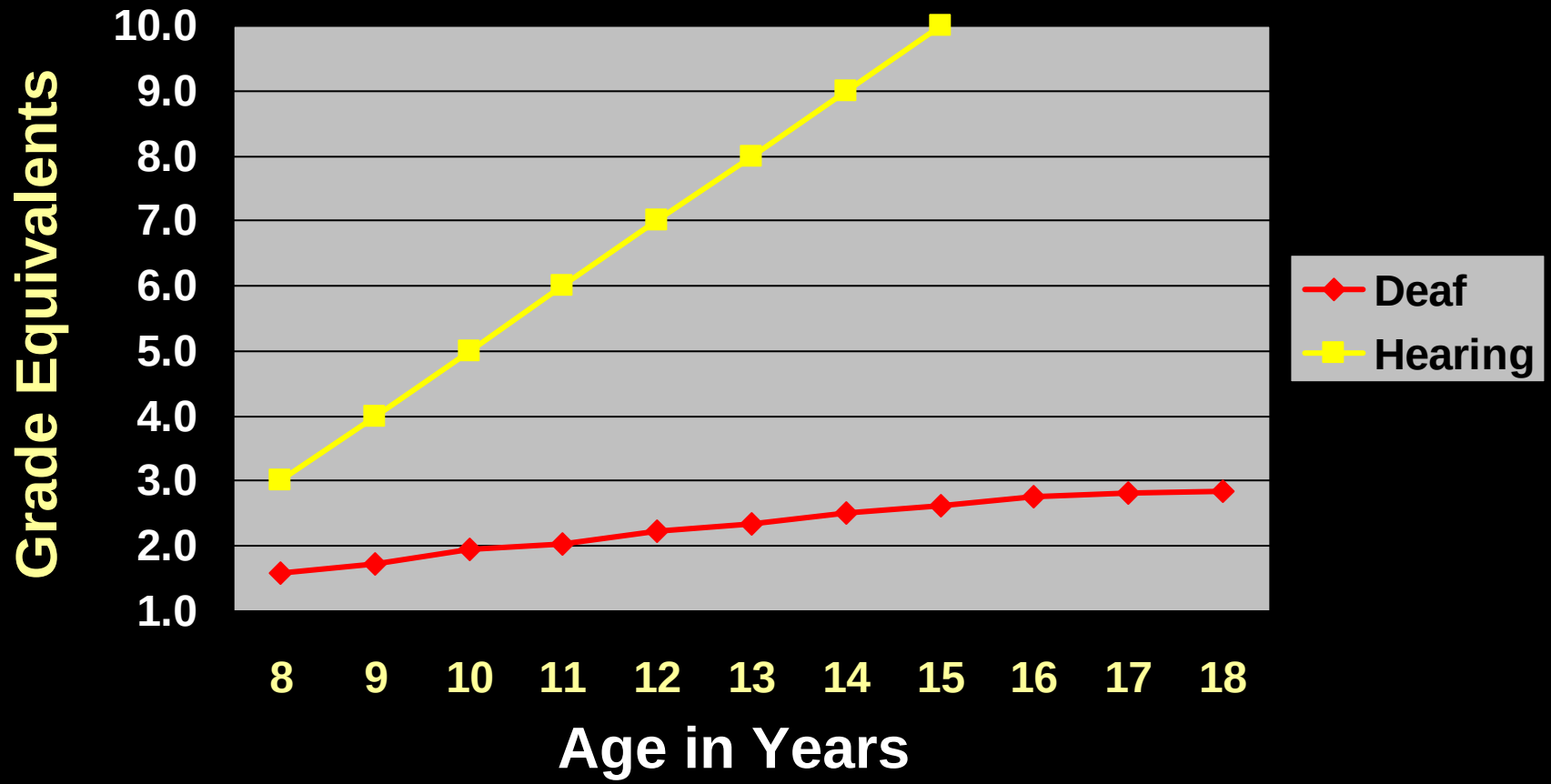
Incidence per 10,000 of Congenital Conditions



Why is Early Identification of Hearing Loss so Important?

- Hearing occurs more frequently than any other birth defect.
- Undetected hearing loss has serious negative consequences.

Reading Comprehension Scores of Hearing and Deaf Students



Schildroth, A. N., & Karchmer, M. A. (1986). Deaf children in America, San Diego: College Hill Press.

Effects of Unilateral Hearing Loss

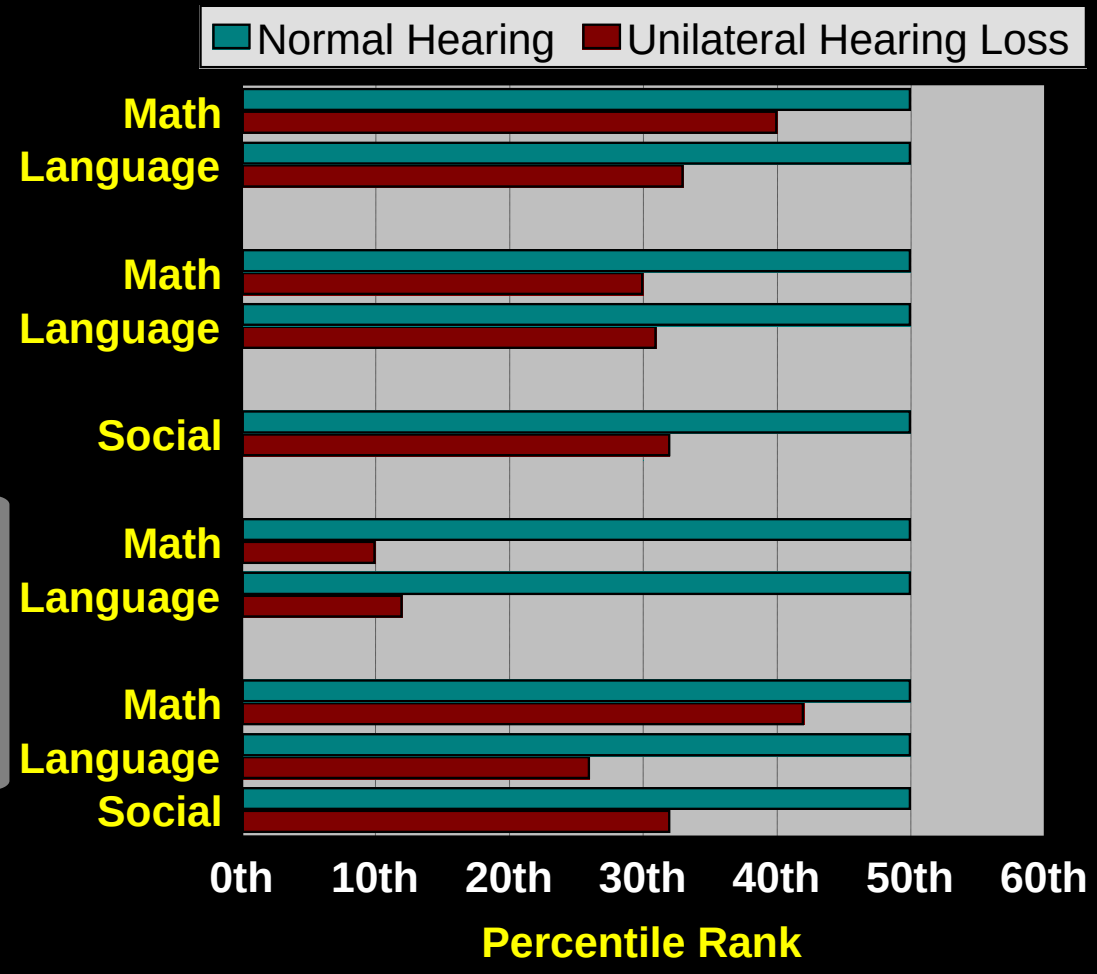
Keller & Bundy (1980)
(n = 26; age = 12 yrs)

Peterson (1981)
(n = 48; age = 7.5 yrs)

Bess & Thorpe (1984)
(n = 50; age = 10 yrs)

By 3rd grade, the average child is ~24 months behind his or her peers in math, language and social skills

Average Results
Math = 30th percentile
Language = 25th percentile
Social = 32nd percentile



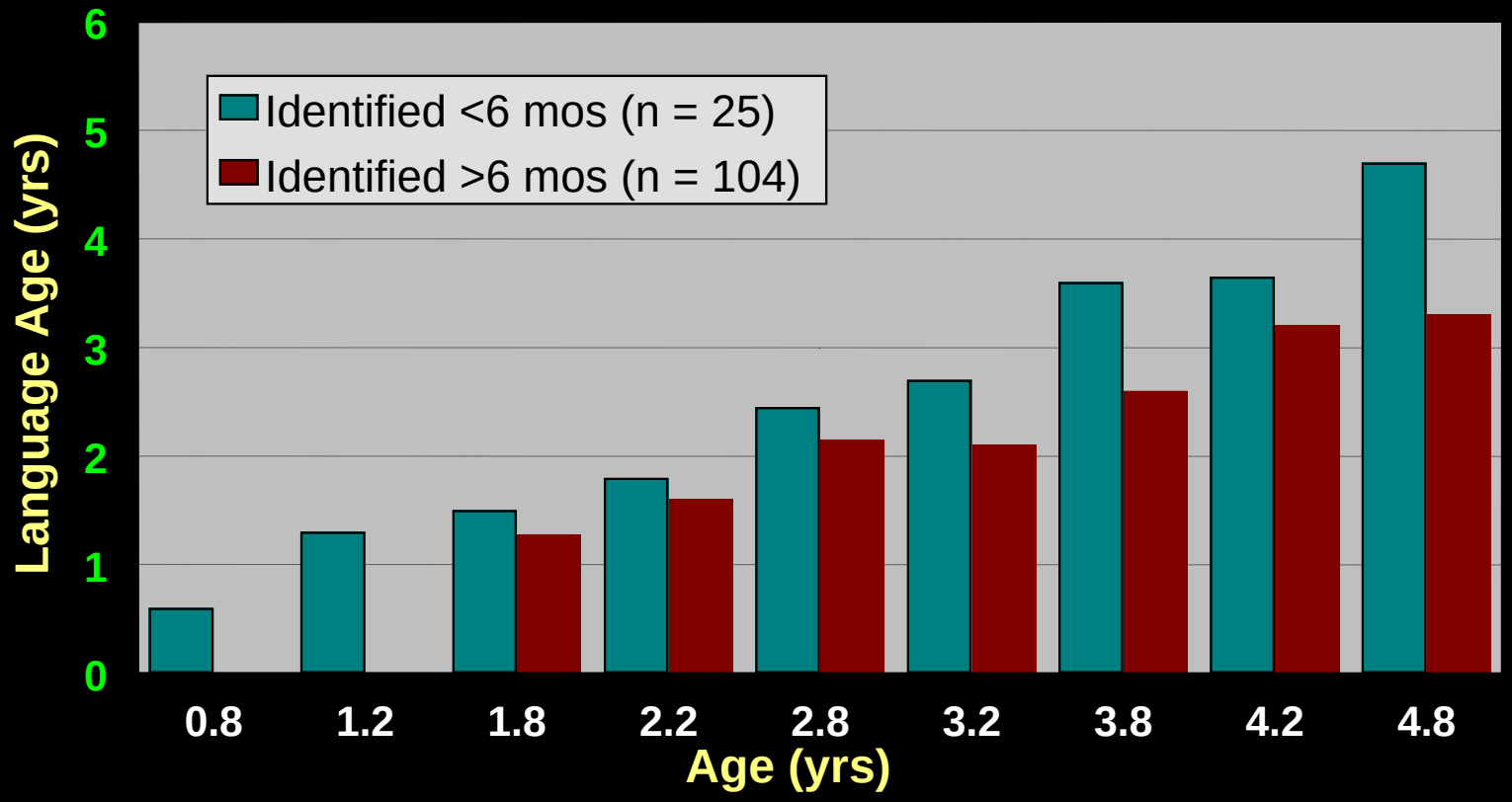
Why is Early Identification of Hearing Loss so Important?

- Hearing loss occurs more frequently than any other birth defect.
- Undetected hearing loss has serious negative consequences.
- There are dramatic benefits associated with early identification of hearing loss.

Boys Town National Research Hospital Study of Earlier vs. Later

129 deaf and hard-of-hearing children assessed 2x each year.

Assessments done by trained diagnostician as normal part of early intervention program.

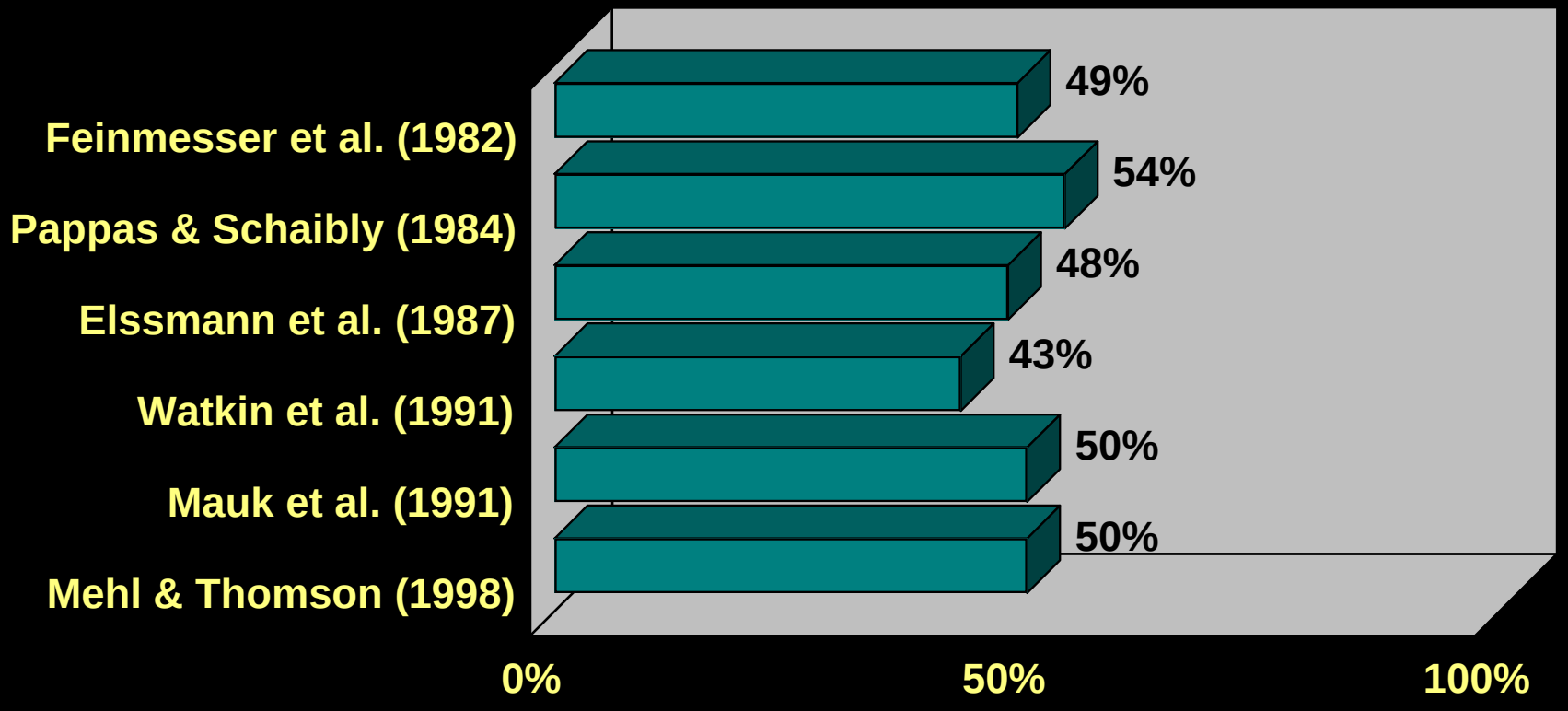


Moeller, M.P. (1997).Personal communication moeller@boystown.org

Newborn Hearing Screening Prior to 1990

- **Conventional Auditory Brainstem Response**
  - **Accurate, but too expensive**
- **High Risk indicators**
  - **Only about 50% of children with congenital hearing loss exhibit one or more of these high risk indicators**

What Percentage of Hearing Impaired Children were High Risk as Infants?



Accuracy of High Risk Based UNHS Programs
Mahoney and Eichwald (1987)

Program operational from 1978-1995.

JCIH indicators incorporated into legally required birth certificate.

Computerized mailing and follow-up, and free diagnostic assessments at regional offices and/or mobile van.

Program now discontinued because:

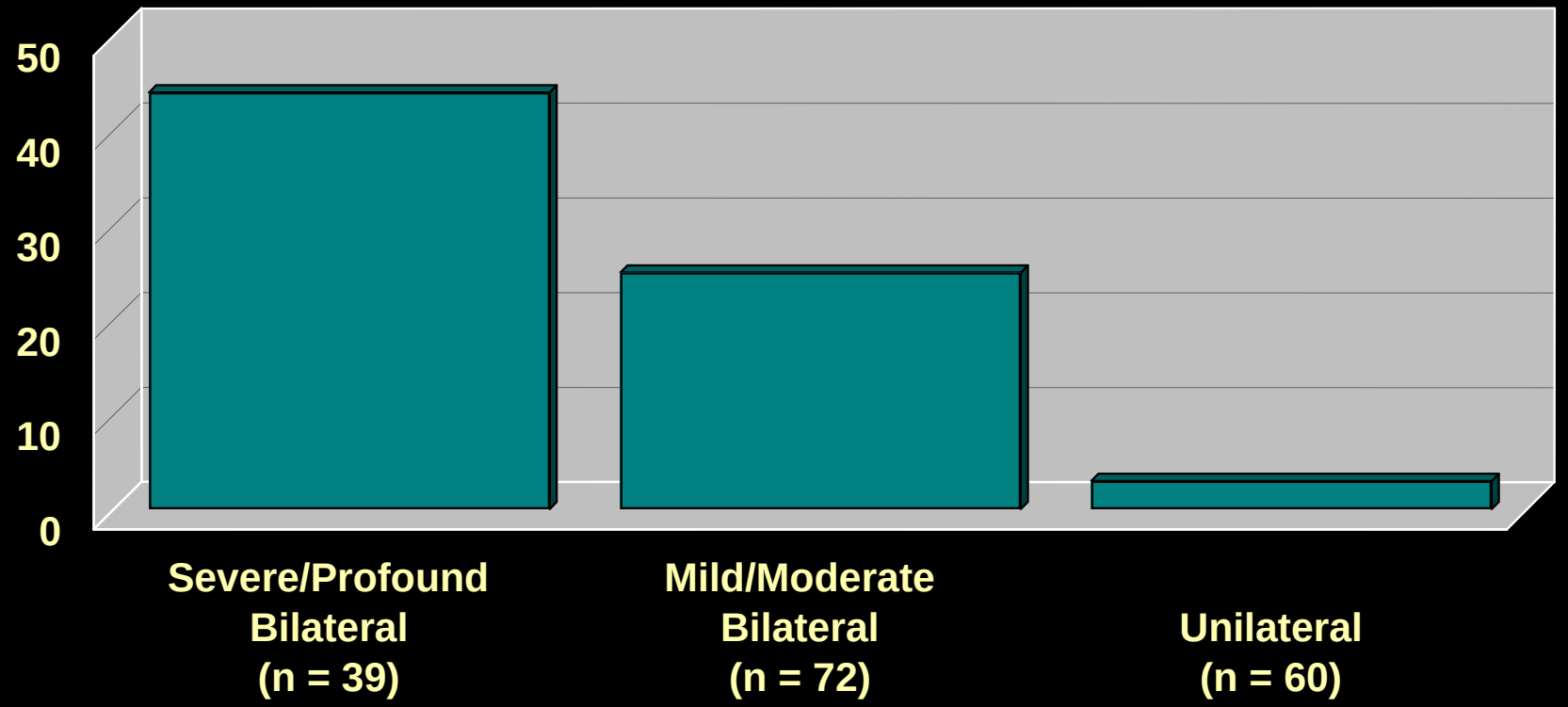
- parents only made appointments for about 1/2 the children who had a risk indicator.
- only about 1/2 of the children with an appointment showed up.
- difficulty obtaining accurate information from hospitals for some risk indicators.

Mahoney, T.M., & Eichwald, J.G. (1987). The ups and "downs" of high-risk hearing screening: The Utah statewide program. Seminars in Hearing 8(2), 155-163.

Newborn Hearing Screening Prior to 1990

- Auditory Brainstem Response
  - Accurate, but too expensive
- High Risk indicators
  - Only about 50% of children with congenital hearing loss exhibit high risk indicators
  - Only about ½ of those with high risk indicators make an appointment for further testing and only about ½ of those are ever tested
- Behaviorally-based hearing screening
  - Expensive
  - Inaccurate

Percentage of Children with Permanent Hearing Loss Identified by the Infant Distraction Test Performed at 8 Months of Age



Watkin, P. M., Baldwin, M., & Laoide, S. (1990). Parental suspicion and identification of hearing impairment. Archives of Disease in Childhood, 65, 846-850.

From 1988-1993vthe first large-scale clinical trial of universal newborn hearing screening was conducted

-- the Rhode Island Hearing Assessment Project ---

SEMINARS IN HEARING—VOLUME 14, NUMBER 1 *February 1993*

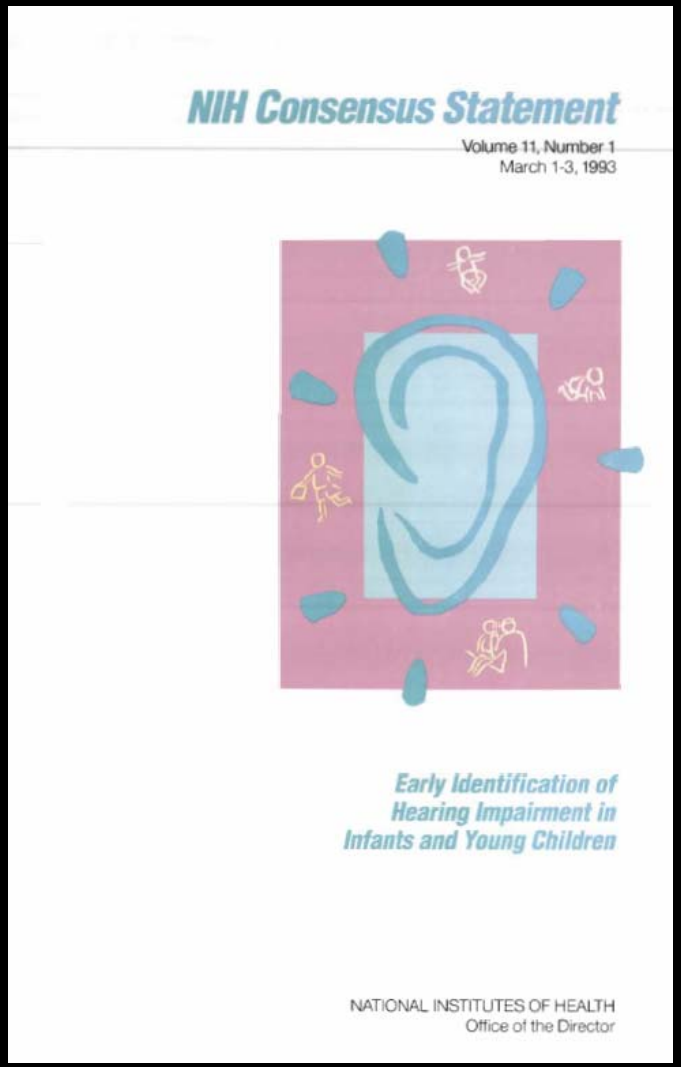
UNIVERSAL NEWBORN HEARING SCREENING USING TRANSIENT EVOKED OTOACOUSTIC EMISSIONS: RESULTS OF THE RHODE ISLAND HEARING ASSESSMENT PROJECT

Karl R. White, Ph.D., Betty R. Vohr, M.D., and Thomas R. Behrens, Ph.D.

The earlier that hearing loss can be identified and intervention begun, the better the prognosis for the child in areas ranging from language development to academic success, social interactions, and successful participation in society.[1] Indeed, early identification of significant hearing loss is so important that the U.S. Department of Health and Human Services (HHS) recently set a goal to reduce to 12 months the average age at which signifi-

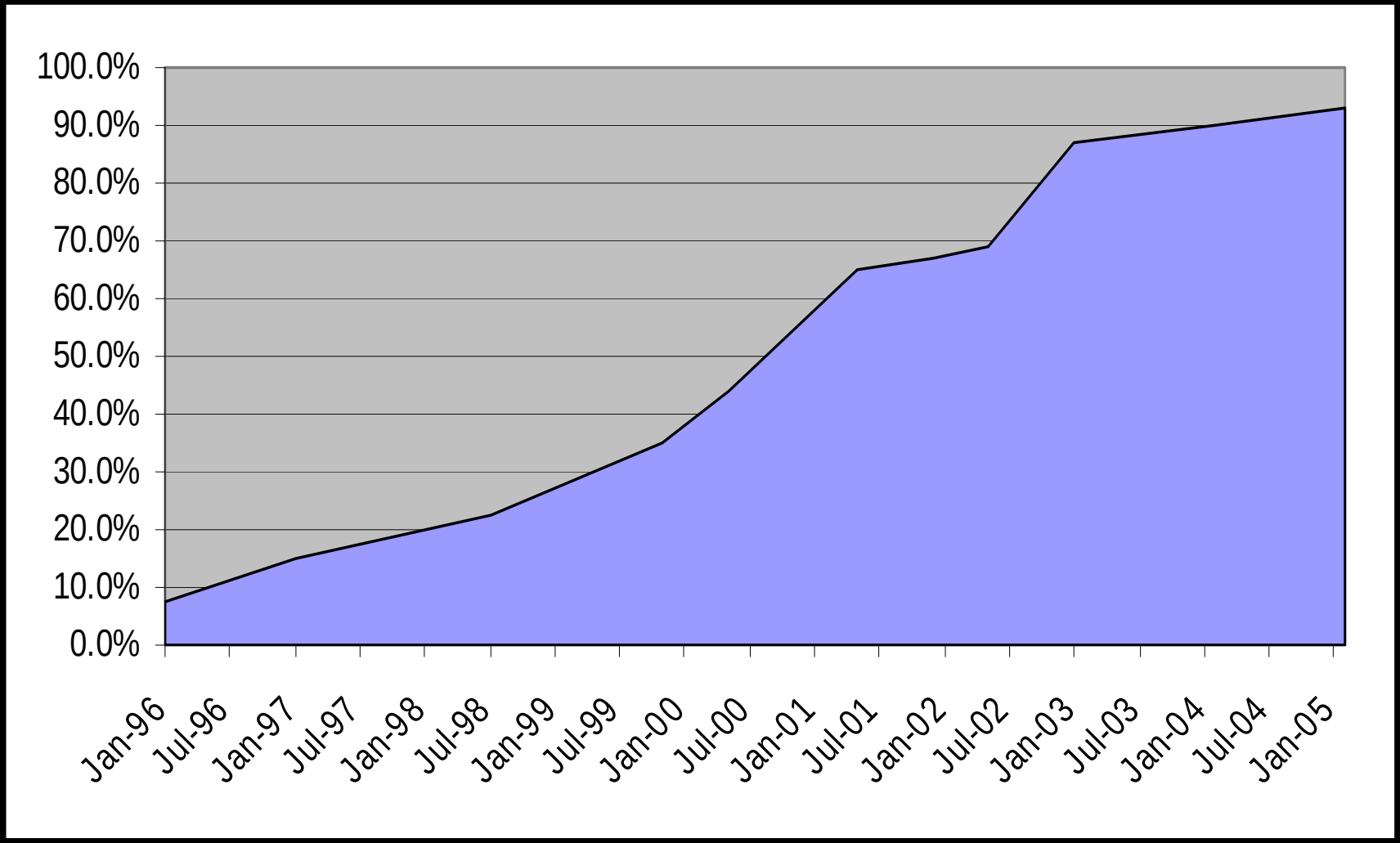
of using auditory brainstem response (ABR) to identify hearing loss among infants and toddlers.[4,5] Such research certainly contributed substantially to the American Speech-Hearing Language Association's (ASHA) recommendation of ABR as the preferred method for screening the hearing of newborns.[6] However, the expense of doing ABR testing of newborns was very likely what led to ASHA's recommendation that it be done only with

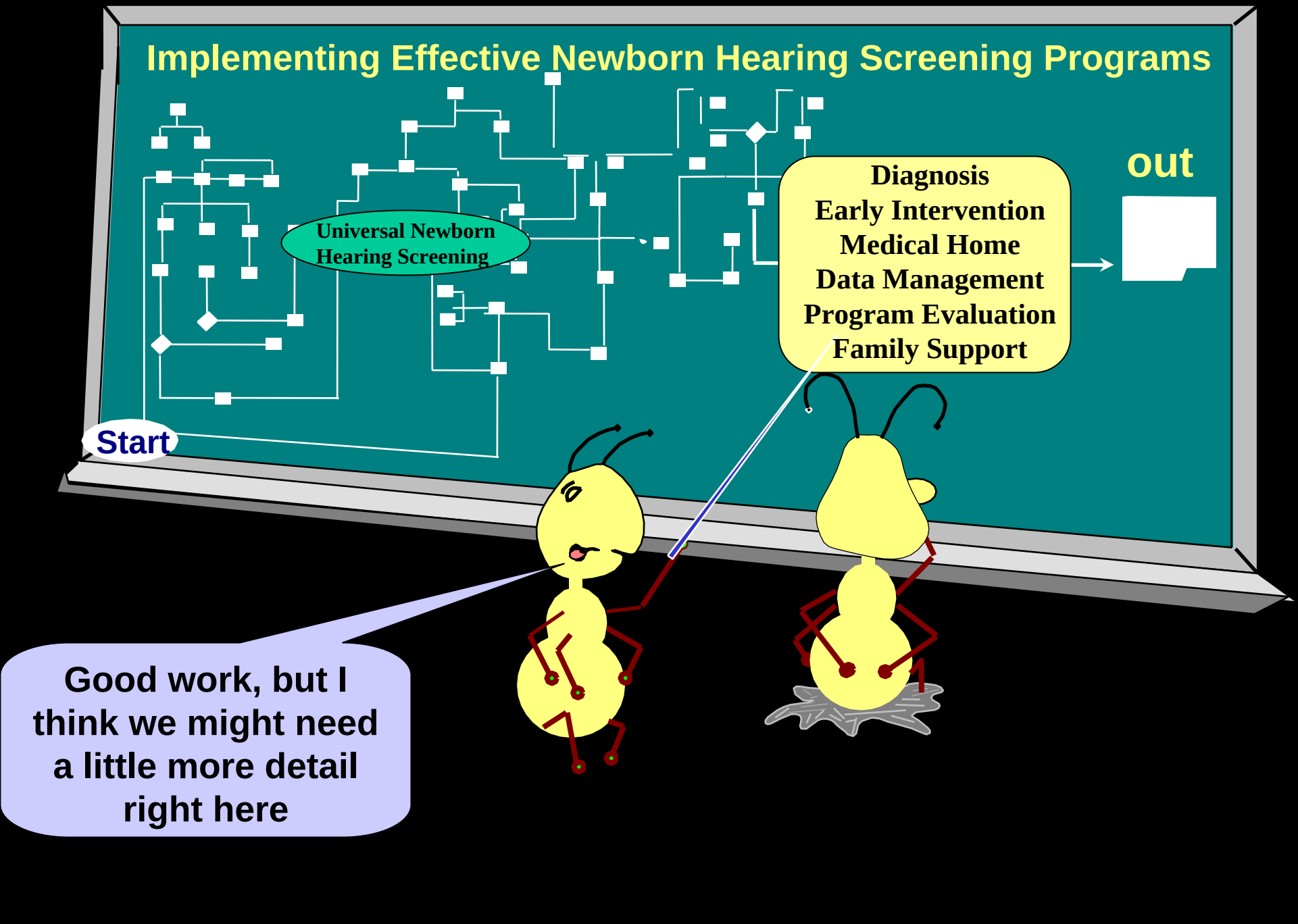
In March, 1993 an NIH Consensus Panel concluded that:



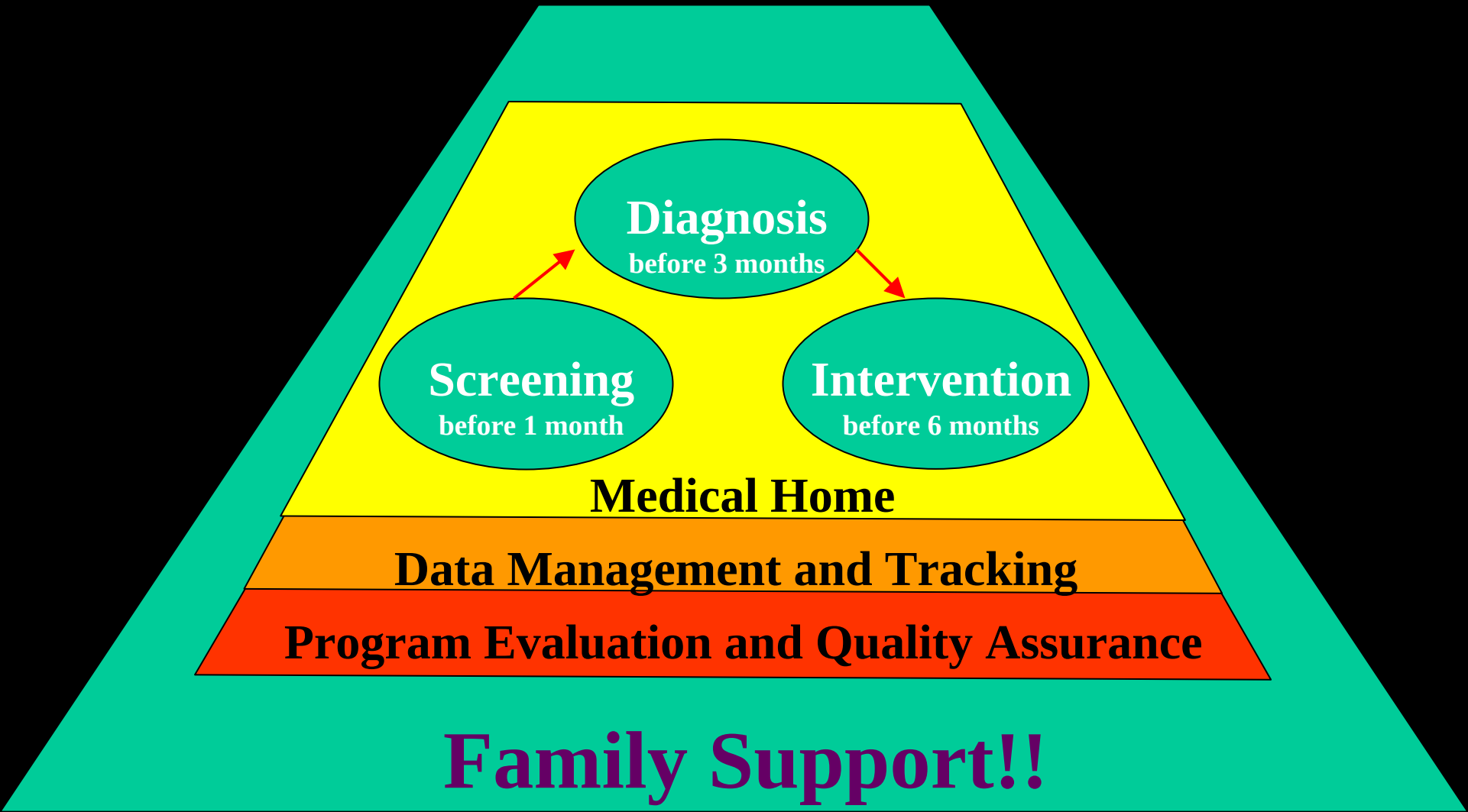
- The average age of diagnosis of hearing loss remains constant at about 2 ½ years of age.
- All infants should be screened for hearing loss…this will be accomplished most efficiently by screening prior to discharge from the well-baby nursery.
- Identification of hearing loss must be seen as imperative for all infants

Percentage of Newborns Screened for Hearing in the United States




Implementing Effective Newborn Hearing Screening Programs
Universal Newborn Hearing Screening
Diagnosis
Early Intervention
Medical Home
Data Management
Program Evaluation
Family Support
out
Start
Good work, but I think we might need a little more detail right here

The Impact of Early Hearing Detection and Intervention (EHDI) Programs on Early Usher Diagnosis



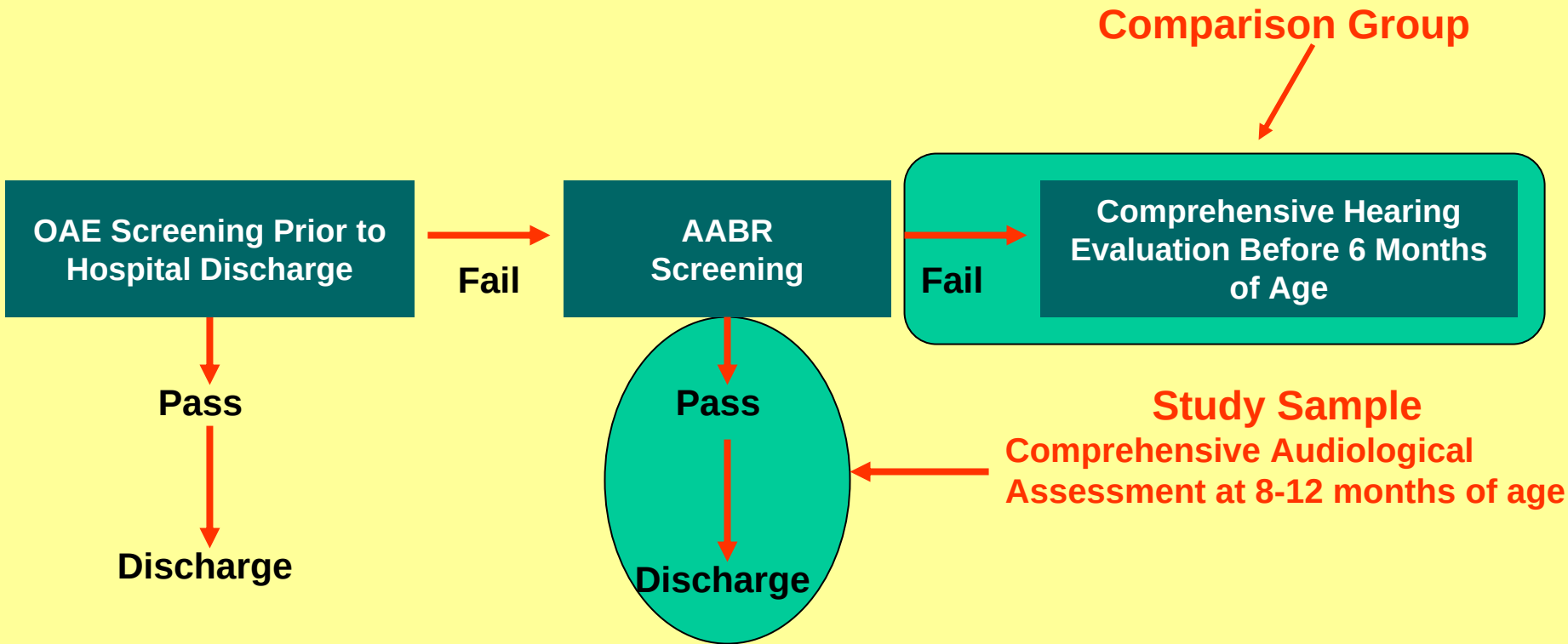
The Impact of Early Hearing Detection and Intervention (EHDI) Programs on Early Usher Diagnosis

1. A Rising Tide Lifts All Ships
2. Education and Public Awareness
3. Advocacy and Policy Initiatives.

Status of EHDI Programs in the US:
Universal Newborn Hearing Screening

- With ~95% of infants screened, newborn hearing screening has become the “standard of care”
- There are hundreds of excellent programs - - - regardless of the type of equipment or protocol used
- Some programs are still struggling with high refer rates and poor follow-up

Does a 2-stage (OAE/AABR) newborn hearing screening protocol miss babies with mild hearing loss?



How Many Additional Babies with Permanent Hearing Loss were Identified?

	Comparison Group (Fail OAE/ Fail AABR)	Study Group (Fail OAE/ Pass AABR)	Total
Number of Babies	**158**	**21**	**179**
Prevalence per 1,000	**1.82**	**.55***	**2.37**

***Adjusted for proportion of OAE fails that enrolled**

Represents 23% of all babies with PHL in birth cohort

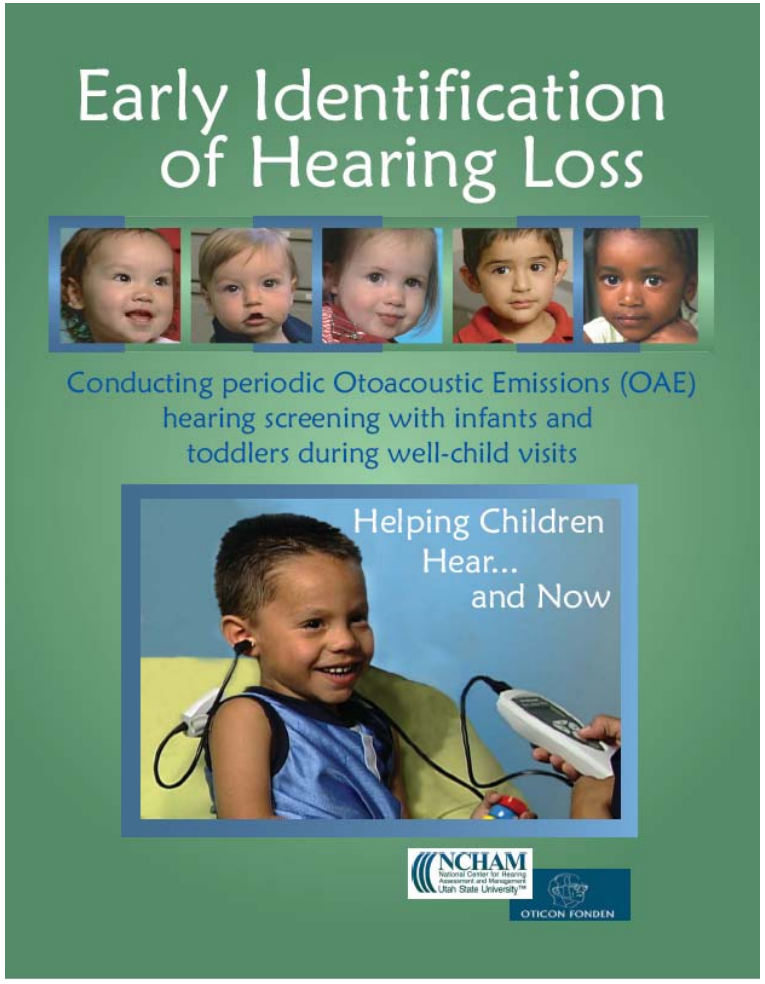
Johnson J, White KR, Widen JE, Gravel JS, James-Trychel M, Kennalley T, Maxon AB, Spivak L, Sullivan-Mahoney M, Vohr BR, Weirather Y, & Holstrum J (2005). A multi-center evaluation of how many infants with permanent hearing loss pass a two-stage OAE/A-ABR newborn hearing screening protocol. *Pediatrics*, *116*(3), 663-672.

The Hearing Head Start Project

- Feasibility study from 2001-2004
- 69 programs in 3 states with 3,000+ children screened
- Identified 2 per 1,000 with permanent hearing loss and 20 per 1,000 with unidentified transient losses
- Programs now being replicated in 12 additional states

Eiserman WD, Shisler L, Foust T, Buhrman J, Winston RL, White KR (In Press). Screening for hearing loss in early childhood programs. *Early Childhood Research Quarterly*.

Hearing Screening During Well Child Visits to Health Care Providers



- Pilot studies and materials development 2005-2006
- Worked with American Academy of Pediatrics to develop recommended policy changes
- Training and implementation materials available from www.HearAndNow.org

Status of EHDI Programs in the United States

- Universal Newborn Hearing Screening
- Effective Tracking and Follow-up as a part of the Public Health System

Rate Per 1000 of Permanent Childhood Hearing Loss in EHDI Programs

Site	Sample Size	Prevalence Per 1000
Rhode Island (3/93 - 6/94)	16,395	1.71
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Rate Per 1000 of Permanent Childhood Hearing Loss in EHDI Programs

Site	Sample Size	Prevalence Per 1000	% of Refers with Diagnosis
Rhode Island (3/93 - 6/94)	16,395	1.71	
Colorado (1/92 - 12/96)	41,976	2.56	48%
New York (1/96 - 12/96)	27,938	1.65	
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Rhode Island (3/93 - 6/94)	16,395	1.71	42%
Colorado (1/92 - 12/96)	41,976	2.56	48%
New York (1/96 - 12/96)	27,938	1.65	67%
Utah (7/93 - 12/94)	4,012	2.99	73%
Hawaii (1/96 - 12/96)	9,605	4.15	98%
Massachussets (1/04 – 12/04)	78,515	2.87	89%

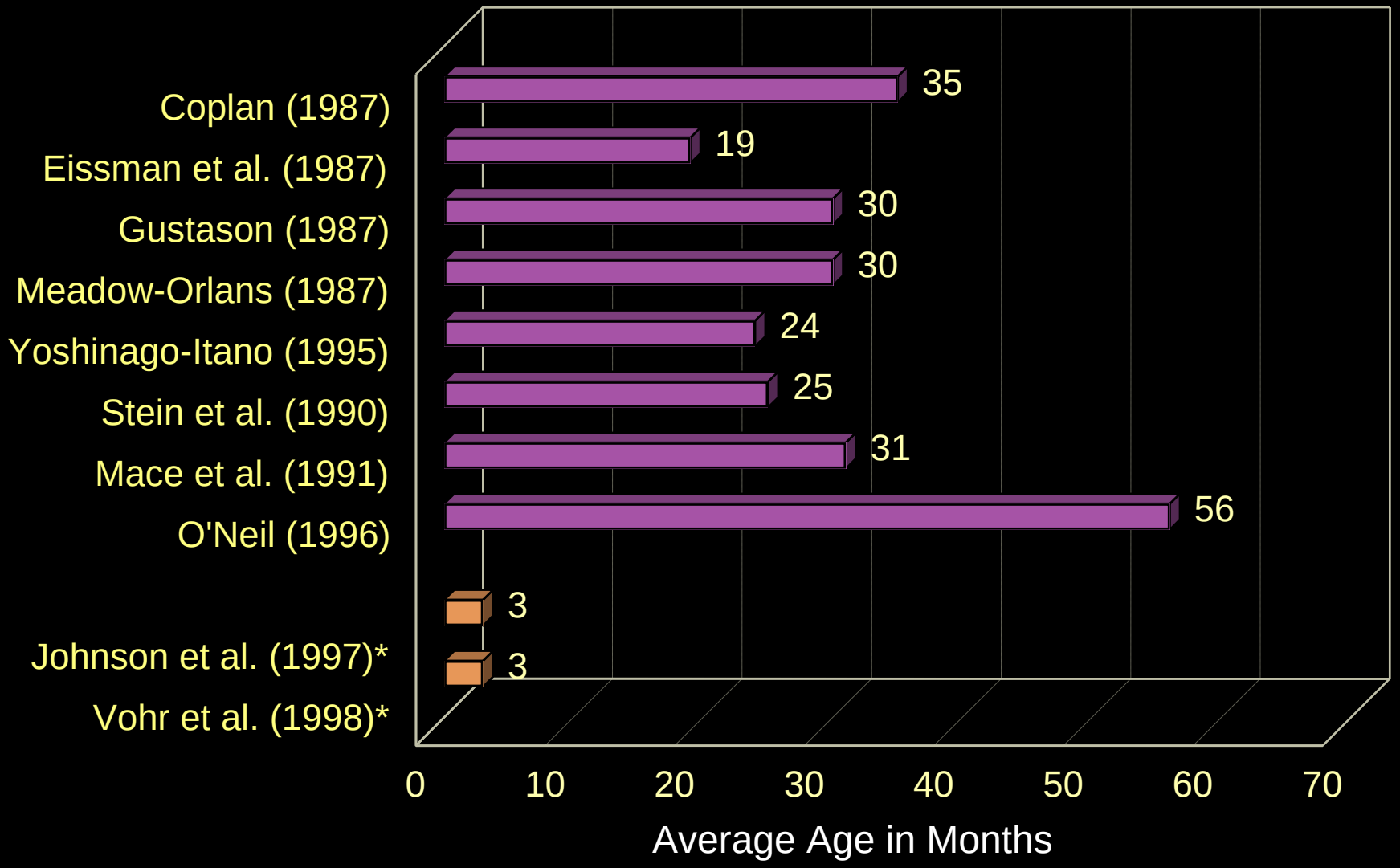
Tracking and Data Management

- 89% of states have created a statewide tracking system
  - information submitted for 80% of the births in 2003
  - 72% have individual identifying data --- up from 32% in 2001
- 57% track babies until at least 3 years of age
- Linkages with other Public Health Information systems are expanding (eg, Vital Statistics, heelstick, EI, Immunizations)

Status of EHDI Programs in the United States

- Universal Newborn Hearing Screening
- Effective Tracking and Follow-up as a part of the Public Health System
- Appropriate and Timely Diagnosis of the Hearing Loss

Confirmation of Permanent Hearing Loss



Status of EHDI Programs in the US: Audiological Diagnosis

- Equipment and techniques for diagnosis of hearing loss in infants continues to improve
- Severe shortages in experienced pediatric audiologists delays confirmation of hearing loss
- State coordinators estimate only 56.1% “receive diagnostic evaluations by 3 months of age

Status of EHDI Programs in the United States

- Universal Newborn Hearing Screening
- Effective Tracking and Follow-up as a part of the Public Health System
- Appropriate and Timely Diagnosis of the Hearing Loss
- Prompt Enrollment in Appropriate Early Intervention

Status of EHDI Programs in the US:
Early Intervention

- Current system designed to serve infants with bilateral severe/profound losses---but, majority of those identified have mild, moderate, and unilateral losses
- State EHDI Coordinators estimate that only 53% of infants with hearing loss are enrolled in EI programs before 6 months of age
- Public or insurance funding is seldom available for high quality hearing aids

Most Early Intervention Programs for Children with Hearing Loss are "Missing the Mark"

- 95% of all newborns with hearing loss have parents with normal hearing.
- When parents in North Carolina were given a choice

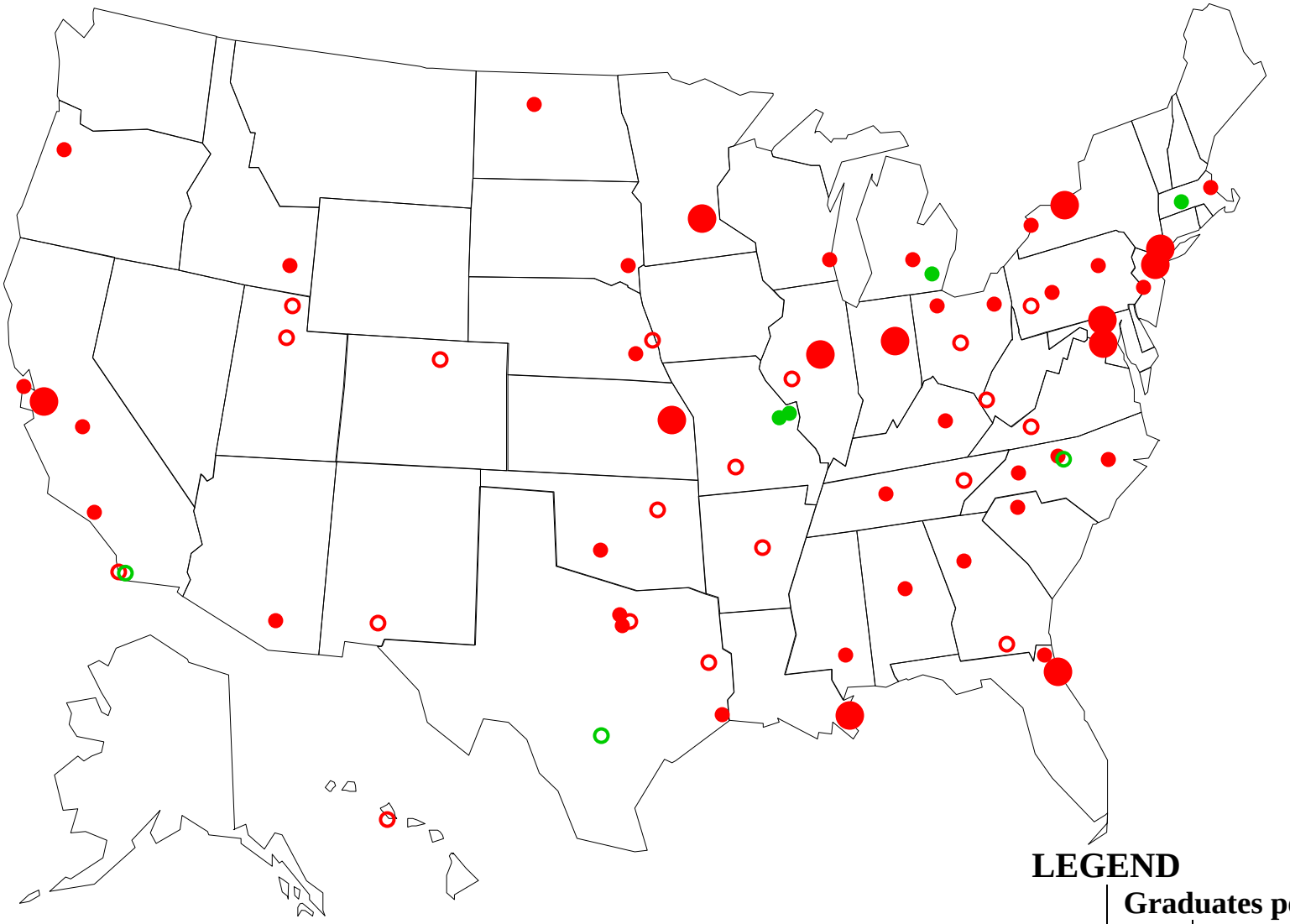
  In 1995: 60% chose sign-language options; 40% chose auditory-oral
  In 2005: 15% chose sign-language options; 85% chose auditory-oral

- The number of cochlear implant for children under age 5 has quadrupled in the last 4 years (to 2000+ implants per year)

Mitchell RE and Karchmer MA. Chasing the mythical ten percent: Parental hearing status of Deaf and Hard of Hearing students in the United States. *Sign Language Studies*. 2004: 4(2), 138-163.

Brown C. Early intervention: *Strategies for public and private sector collaboration*. Paper presented at the 2006 Convention of the Alexander Graham Bell Association for the Deaf and Hard of Hearing. 2006 Pittsburgh PA.

Primary Emphasis of Personnel Preparation Programs for Teachers of Deaf and Hard of Hearing



LEGEND

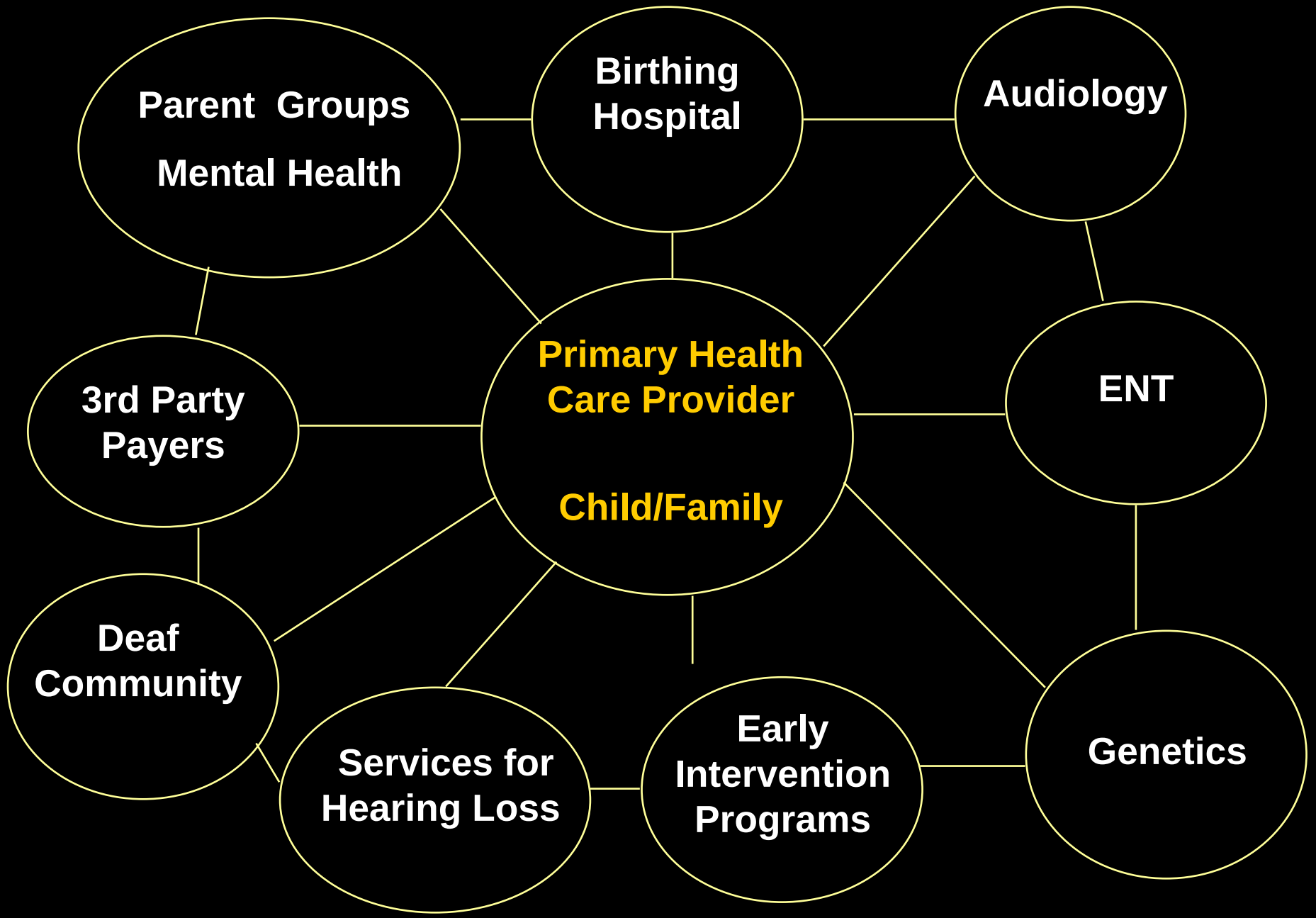
Primary Emphasis	Graduates per Year: 1-5	6-15	16+
Sign Language-based	○	●	●
Spoken Language-based	○	●	●

Note: Although many programs describe themselves as providing "comprehensive" services, most have a primary emphasis on a specific approach as indicated by the curriculum offerings, the placement of graduates, the type of practicum available, etc. Classification of programs on this map considered those factors in conjunction with annual self-report survey data from the 2004 and 2005 issues of the ***American Annals of the Deaf.***

Status of EHDI Programs in the United States

- Universal Newborn Hearing Screening
- Effective Tracking and Follow-up as a part of the Public Health System
- Appropriate and Timely Diagnosis of the Hearing Loss
- Prompt Enrollment in Appropriate Early Intervention
- A Medical Home for all Newborns

EHDI and the Medical Home



Educating Primary Health Care Providers About Early Identification of Hearing Loss

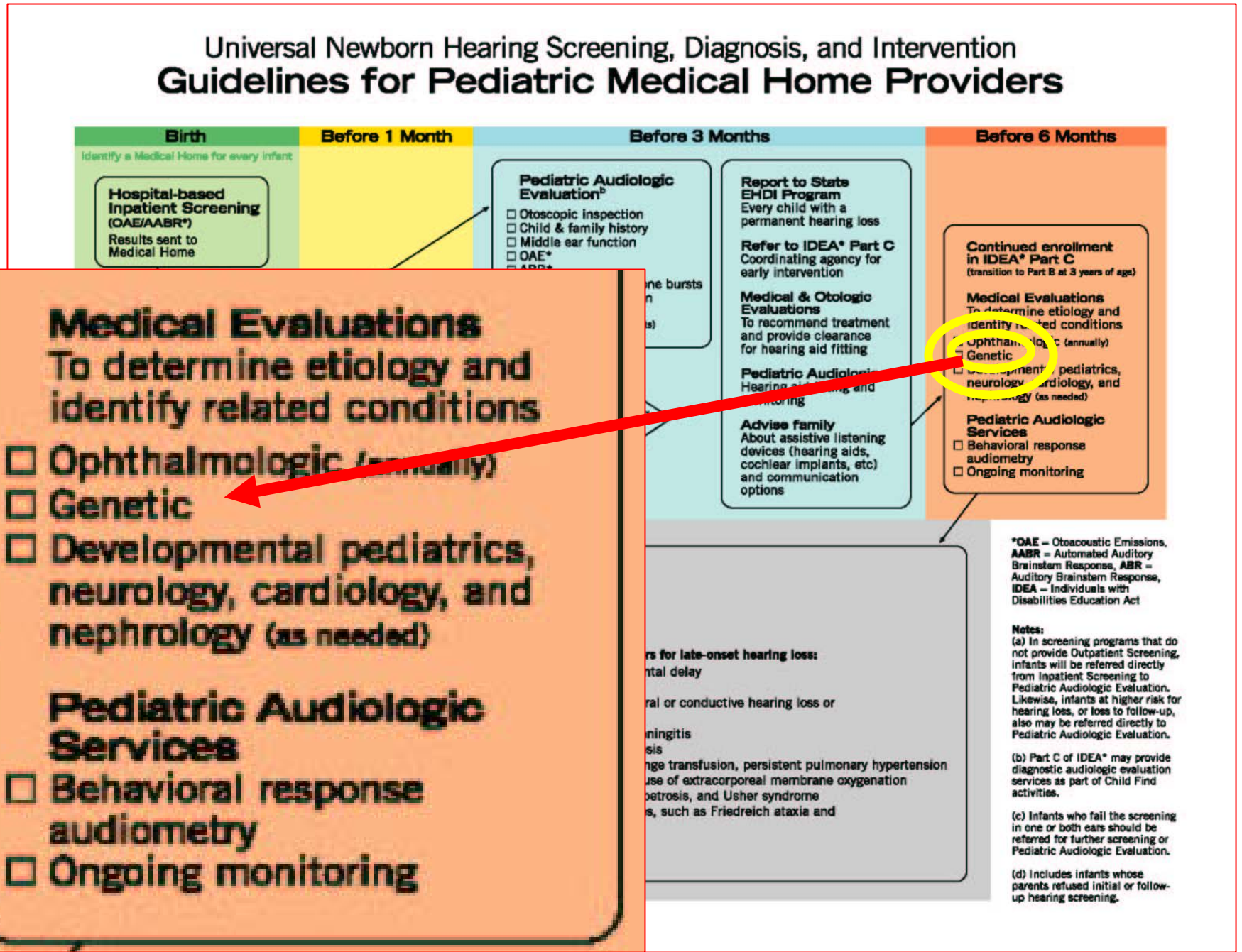
Assume a newborn for whom you are caring is diagnosed with a moderate to profound bilateral hearing loss. If no other indications are present, would you refer the baby for a(n):

	Always or Often
Ophthalmological evaluation	0.6%
Genetic evaluation	8.9%
Otolaryngological evaluation	75.6%

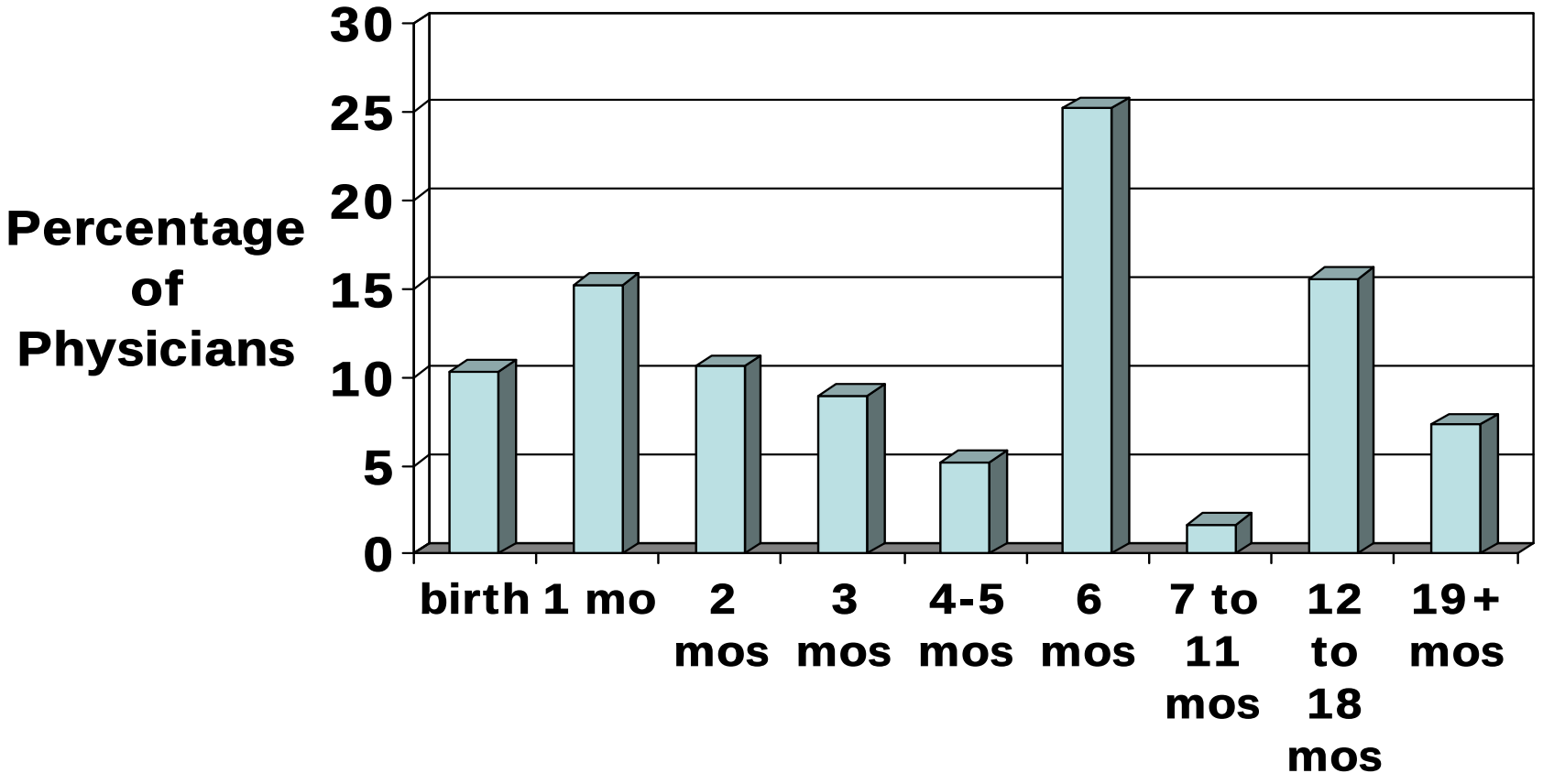
Responses of 1975 physicians in 21 states

Moeller MP, White KR, & Shisler L (in press). Primary care physicians' knowledge, attitudes and practices related to newborn hearing screening. *Pediatrics*.

American Academy of Pediatrics



When can an infant be fit with hearing aids?

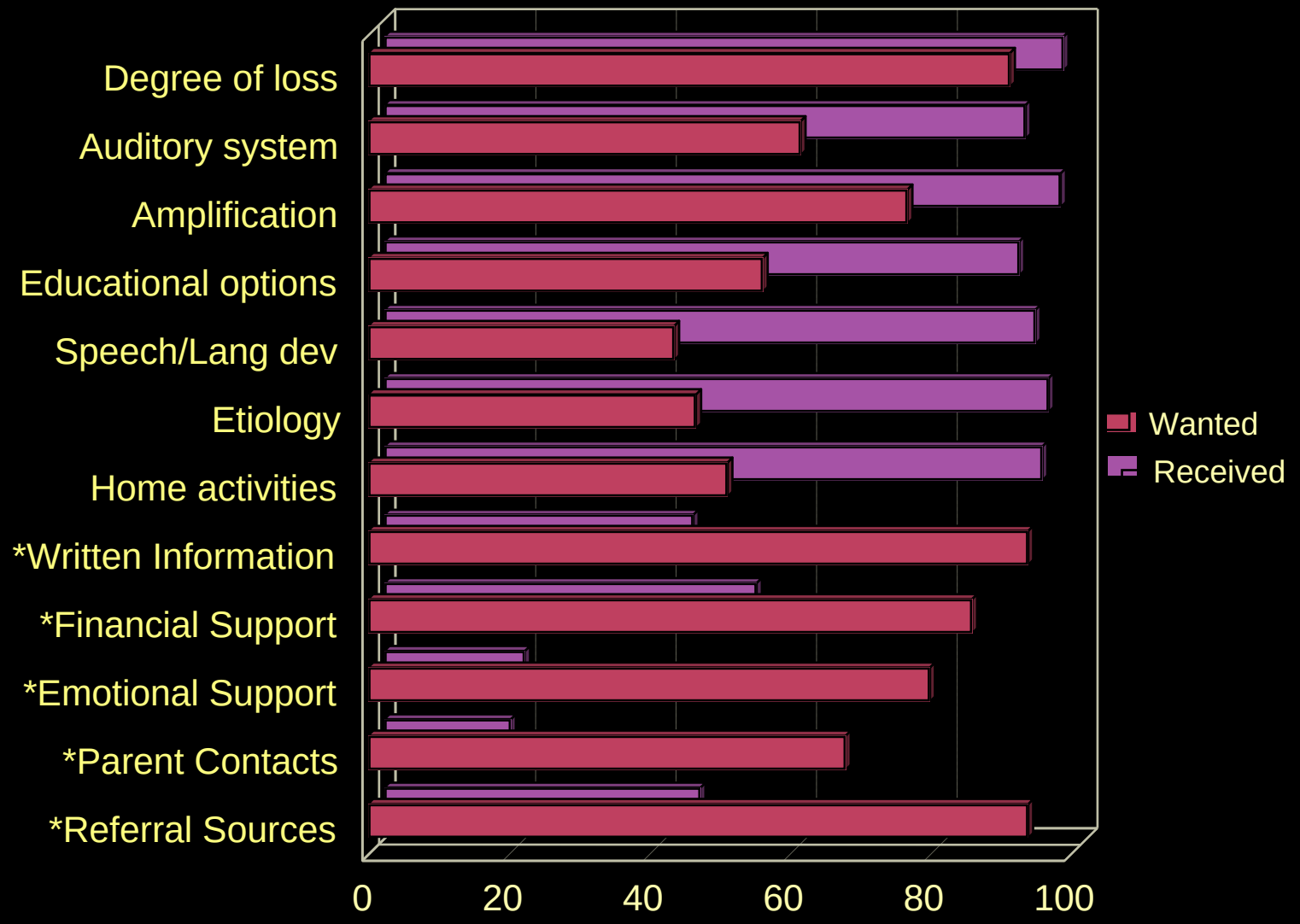


Type of Physician	Age at which hearing aids can be fit				
	<=1 mo	2-3 mos	4-6 mos	7-11 mos	12+ mos
Pediatrician (n=1145)	**36.3%**	**16.9%**	**29.0%**	**2.1%**	**15.6%**

Status of EHDI Programs in the United States

- Universal Newborn Hearing Screening
- Effective Tracking and Follow-up as a part of the Public Health System
- Appropriate and Timely Diagnosis of the Hearing Loss
- Prompt Enrollment in Appropriate Early Intervention
- A Medical Home for all Newborns
- **Culturally Competent Family Support**

Information Wanted vs. Received by Parents at Hearing Loss Confirmation

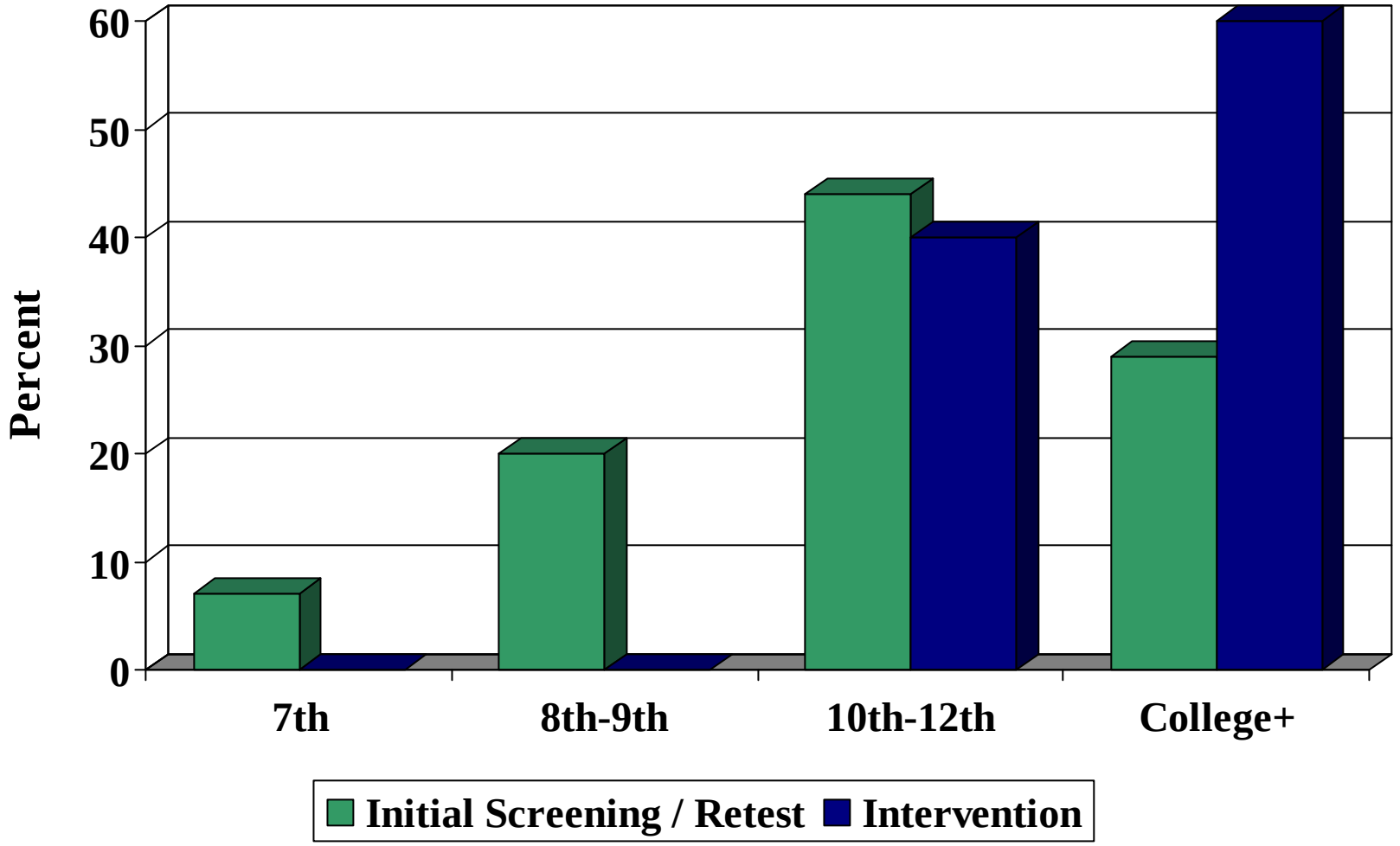


Martin, George, O'Neal, & Daly (1987); *Sweetow & Barrager (1980)

Are current EHDI materials effective?

Brochure Readability

Gold Standard Readability: ≤6th Grade



Policy and Legislative Initiatives with Local, State and Federal Partners

Take Home Messages

- **The world has changed for infants and young children with permanent hearing loss**
- **Screening is only the first (and the easiest!) step**
- **Just as scientific and technological advances have made the revolutionary changes of the last 15 years possible --- more are coming**
- **Education and advocacy are the foundation on which future progress will be built**
- **Usher Syndrome is one of many specific conditions (but an important one) that will benefit from effective comprehensive screening for permanent hearing loss**

www.infanthearing.org

http://www.infanthearing.org/

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Links & Links

NAVIGATION

Who We Are

- Our Background
- Nat. EHDI Resource Center
- Research Projects
- The Staff

EHDI Components

- Newborn Hearing Screening
- Diagnostic Audiology

NCHAM
National Center for Hearing Assessment and Management
Utah State University™

• 2006 SE Regional EHDI Conference •

Registration is open for the Southeast Regional EHDI Conference: Partnering for Progress to held on October 5-7, 2006 in Jackson, Mississippi
Click for more information

To ensure that all infants and toddlers with hearing loss are identified as early as possible and provided with timely and appropriate audiological, educational, and medical intervention, an early hearing detection and intervention (EHDI) program should comprise three basic components—newborn hearing screening, audiological diagnosis, and early intervention. Threaded throughout these components should also be some key elements—culturally-competent family support, medical home, data management, legislative mandates, and program evaluation tools. Follow the links below to find information about these basic components and key elements, and about other related EHDI resources and information.


EHDI Components

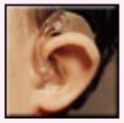

EHDI Resources


State Information


Legislative Activities


NCHAM Items

Annual EHDI Meetings


EHDI Workshops

May/June 2002 · Vol. 4 · No. 3

Genetics Evaluation Guidelines for the Etiologic Diagnosis of Congenital Hearing Loss

Genetic Evaluation of Congenital Hearing Loss Expert Panel

The advent of hearing screening in newborns in many states has led to an increase in the use of genetic testing and related genetic services in the follow-up of infants with hearing loss. A significant proportion of those with congenital hearing loss have genetic etiologies underlying their hearing loss. To ensure that those identified with congenital hearing loss receive the genetic services appropriate to their conditions, the Maternal and Child Health Bureau of the Health Resources and Services Administration funded the American College of Medical Genetics to convene an expert panel to develop guidelines for the genetic evaluation of congenital hearing loss. After a brief overview of the current knowledge of hearing loss, newborn screening, and newborn hearing screening, we provide an overview of genetic services and a guideline that describes how best to ensure that patients receive appropriate genetic services. The significant contribution of genetic factors to these conditions combined with the rapid evolution of knowledge about the genetics of these conditions overlaid with the inherently multidisciplinary nature of genetic services provides an example of a condition for which a well-integrated multidisciplinary approach to care is clearly needed. **Genet Med 2002:4(3):162–171.**

Key Words: *newborn screening, genetic testing, congenital hearing loss*

Appropriate management of all persons identified with congenital hearing loss, as defined above, requires a comprehensive genetic evaluation.

…ly common in the human population. …ring loss is estimated to occur in about 1 …ately 50% of cases are thought to be due … and the remainder to genetic causes[1,2] …ormer include acoustic trauma, ototoxic …oglycosides), and bacterial or viral infec… …ytomegalovirus (CMV). Approximately …ssociated with genetic factors are classi… …he deafness is not associated with other …ine a recognized syndrome). In the re… …re than 400 forms of syndromic deafness …e of associated clinical findings.[1,2] The …s widely among the many forms of syn… …includes both conductive and sensori… …be unilateral or bilateral, symmetrical or …ssive or stable.[4]

…y of nonsyndromic hearing impairment …t the deficits are most often sensorineu… …usually subdivided by mode of inheri… …% of NSHI is autosomal recessive, 22% is …d 1% is X-linked. The associated "DeaF… …signated DFNB (autosomal recessive), …minant), and DFN (X-linked). A variable …haps less than 1%, is due to mitochon… …he proportion may be much higher (10%–20%) in some populations[5,6] (Fig. 1). As a general rule, individuals with autosomal recessive NSHI have profound prelingual deafness, while dominant mutations lead to a more variable phenotype. More than 90% of children with congenital profound autosomal recessive NSHI are born to parents with normal hearing, while the remaining 10% or less are born to deaf parents.

Over the past 5 years, remarkable progress has been made identifying new hearing impairment loci and cloning new genes for deafness. To date, at least 77 loci for NSHI have been mapped: 40 autosomal dominant, 30 autosomal recessive, and 7 X-linked.[7] As of July 2001, 50 auditory genes have been identified and sequenced including 14 for autosomal dominant disorders, 9 for autosomal recessive, 2 for X-linked, 5 mitochondrial, and at least 31 genes for syndromic hearing loss. In some cases, different mutations at the same locus have been found to cause syndromic and nonsyndromic forms of deafness. Although significant advances have been made, it is clear that more genes and mutations await discovery. Information about these genes and their protein products is revolutionizing our knowledge of the molecular processes involved in hearing and enhancing our understanding of how the alteration of these processes can lead to hearing loss. This knowledge may lead to mutation-specific therapies that can delay or prevent certain forms of genetic deafness such as the avoidance of aminoglycoside therapy in those with specific mitochondrial mutations.

History of newborn screening programs

Newborn screening programs for heritable disorders began in the early 1960s.[8] They have evolved into the current public health newborn screening systems that include screening for metabolic diseases, hemoglobinopathies, endocrine disorders, cystic fibro-

…al Genetics, 9650 Rockville Pike, Bethesda, MD 20814-3998.
Go to www.genetest.com/acmg for a printable copy of this document.

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JOINT COMMITTEE ON INFANT HEARING YEAR 2000 POSITION STATEMENT:
Principles and Guidelines for Early Hearing Detection and Intervention Programs

The Year 2000 Position Statement and Guidelines were developed by the Joint Committee on Infant Hearing. Joint committee member organizations and their respective representatives who prepared this statement include (in alphabetical order) the American Academy of Audiology (Terese Finitzo, Ph.D., chair; and Yvonne Sininger, Ph.D.); the American Academy of Otolaryngology–Head and Neck Surgery (Patrick Brookhouser, M.D., vice-chair; and Stephen Epstein, M.D.); the American Academy of Pediatrics (Allen Erenberg, M.D.; and Nancy Roizen, M.D.); the American Speech-Language-Hearing Association (Allan O. Diefendorf, Ph.D.; Judith S. Gravel, Ph.D.; and Richard C. Folsom, Ph.D.); the Council on Education of the Deaf whose member organizations include: Alexander Graham Bell Association for the Deaf and Hard of Hearing, American Society for Deaf Children, Conference of Educational Administrators of Schools and Programs for the Deaf, Convention of American Instructors of the Deaf, National Association of the Deaf, and Association of College Educators of the Deaf and Hard of Hearing (Patrick Stone, Ed.D; Joseph J. Innes, Ph.D. and Donna M. Dickman, Ph.D.*); and the Directors of Speech and Hearing Programs in State Health and Welfare Agencies (Lorraine Michel, Ph.D.; Linda Rose, MCD; Thomas Mahoney, Ph.D.). Ex officios to the JCIH include: Evelyn Cherow, MA (American Speech-Language Hearing Association); Deborah Hayes, Ph.D., (Marion Downs National Center for Infant Hearing); and Liz Osterhus, MA and Thomas Tonniges, M.D. (American Academy of Pediatrics)

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, the Council on Education of the Deaf (see
rs of Speech and Hearing Programs in State

… families should be offered the option of genetic evaluation and counseling by a medical geneticist