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Research Review

Early Hearing Detection and Intervention Programs: Opportunities for Genetic Services

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Congenital hearing loss is relatively frequent and has serious negative consequences if it is not diagnosed and treated during the first few months of life. Babies with hearing loss who are identified early and provided with appropriate intervention develop better language, cognitive, and social skills. As a result of improvements in screening equipment and procedures, newborn hearing screening programs have expanded rapidly in recent years, and almost 90% of all newborns are now screened for hearing loss before leaving the hospital. Because 50% or more of congenital hearing loss is due to genetic causes, providers of genetic services should play an increasingly important role in newborn hearing screening, diagnostic, and intervention services. For this to happen, parents, public health officials, and primary health care providers need to become better informed about the benefits of genetic services for children with hearing loss and their families. Providers of genetic services also need to become better informed about the current status of Early Hearing Detection and Intervention (EHDI) programs and how they can contribute to continued improvement of these programs.

KEY WORDS: newborn hearing screening; genetics of hearing loss; congenital hearing loss

INTRODUCTION

Congenital hearing loss, at a rate of approximately 3 per 1,000 births, occurs more frequently than other conditions for which newborns are routinely screened and for which early detection has proven to be beneficial [March of Dimes, 2003]. Fifteen years ago, children with congenital hearing loss were typically not identified until they were 2–years old [Toward Equality, 1988]. Such late identification results in serious negative consequences as noted by the U.S. Department of Health and Human Services:

> "It is difficult if not impossible for [many children with congenital hearing loss] to acquire the fundamental language, social, and cognitive skills that provide the foundation for later schooling and success in society. When early identification and intervention occur, hearing-impaired children make dramatic progress, are more successful in school, and become more productive members of society. [HHS, 1990, p. 460]"

Although the importance of identifying hearing loss as early as possible has been recognized for almost 60 years [Ewing and Ewing, 1944], identification during the first few months of life occurred infrequently until the development of inexpensive, accurate, and practical screening equipment in the early 1980s [Kemp and Ryan, 1993; Herrmann et al., 1995]. As such equipment became more widely available, the federal government began funding research and demonstration projects to promote and improve newborn hearing screening and diagnosis programs, or what are now widely referred to as Early Hearing Detection and Intervention (EHDI) Programs. In 1990, the U.S. Surgeon General, Dr. C. Everett Koop, M.D., issued a challenge to reduce the age at which congenital hearing loss was identified.

> "Deafness in . . . infants is a serious concern because it interferes with the development of language . . . that which sets humans apart from all other living things . . . . Early intervention actually saves money since hearing impaired children who receive early help require less costly special education services later . . . . I am optimistic. I foresee a time in this country . . . when no child reaches his or her first birthday with an undetected hearing impairment. [Northern and Downs, 1991, pp. 2–3]"

Dr. Koop's optimism was somewhat surprising, given that less than 3% of all newborns were screened for hearing loss at the time [Bess, 1993]. It was not long, however, before the percentage of newborns screened for hearing loss began to increase dramatically. As shown in Figure 1, by January 2003, approximately 87% of all newborns in the United States were screened for hearing loss before they were discharged from the hospital [NCHAM, 2003a].

The expansion of EHDI programs offers new and valuable opportunities for providers of genetic services for several reasons.

- Congenital hearing loss is the most frequently occurring birth defect.
- At least 50% of congenital hearing loss has a genetic etiology.
- Recent advances in genetic testing have led to an increase in the use of genetic services in the follow up of infants with hearing loss.
- There are significant benefits for children with hearing loss and their families if they receive genetic testing and counseling.

For providers of genetic services to become appropriately involved in EHDI programs though, they need to understand

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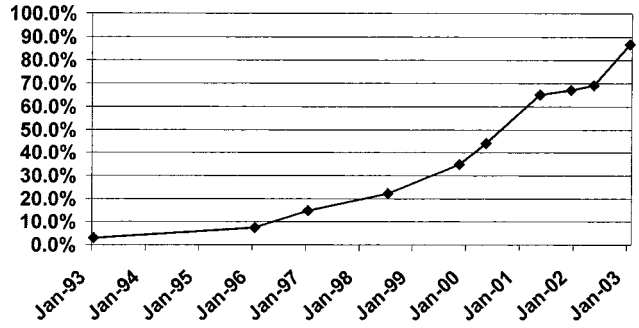


Fig. 1. Percentage of newborns screened for hearing prior to hospital discharge.

why it is so important to identify hearing loss early, the current status of EHDI programs in the U.S., and the particular ways in which genetic services can contribute to better outcomes for children with congenital hearing loss and their families.

IMPORTANCE OF IDENTIFYING CONGENITAL HEARING LOSS EARLY

The importance of early identification of congenital hearing loss stems from the relatively high frequency with which it occurs, the consequences of hearing loss if intervention is not provided, and the benefits that accrue to children and families if timely and appropriate intervention is offered.

Prevalence of Congenital Hearing Loss

How many babies in the general population are born with hearing loss? Although it seems like this should be a simple question to answer, widely discrepant figures are frequently cited. By combining the data from published reports of universal newborn hearing screening programs with data from epidemiological studies, it is possible to derive an estimate of the prevalence of permanent congenital hearing loss in which we can be confident.

Reports of screening programs. One source of information for estimating the prevalence of permanent congenital hearing loss is the published reports of newborn hearing screening programs in which babies referred from screening programs have been followed to determine how many were identified with permanent hearing loss. Data from several such studies are summarized in Table I.

Even though most programs have not been successful in determining the final hearing status of a large percentage of the babies referred from the screening program, 2–4 babies per 1,000 with permanent congenital hearing loss are being identified. If the programs that were unable to determine the hearing status of a large number of babies had been more successful with follow-up, it is likely that their prevalence rates would have been higher.

Results of epidemiological studies. How consistent are the results of these screening programs with epidemiological studies of permanent hearing loss among children? The answer to this question depends partly on how definitional issues are addressed related to severity, type of hearing loss, and age at onset.

Figure 2 shows the results of 12 studies designed to determine the number of children with bilateral permanent hearing loss in population-based cohorts ranging in size from 10,000 to over 4 million children. In each study, a large cohort of children who were representative of the general population in that country were assessed for permanent hearing loss when they were 6–12-years old. As expected, the prevalence of bilateral permanent hearing loss is substantially higher when milder hearing losses are included as shown by the dashed line in Figure 2. When the definition of hearing loss is limited to bilateral losses greater than 50 dB, prevalence is about 1.0 per thousand. However, when children are included if they have a bilateral permanent hearing loss greater than 30 dB, the prevalence increases to about 2.5 per thousand.

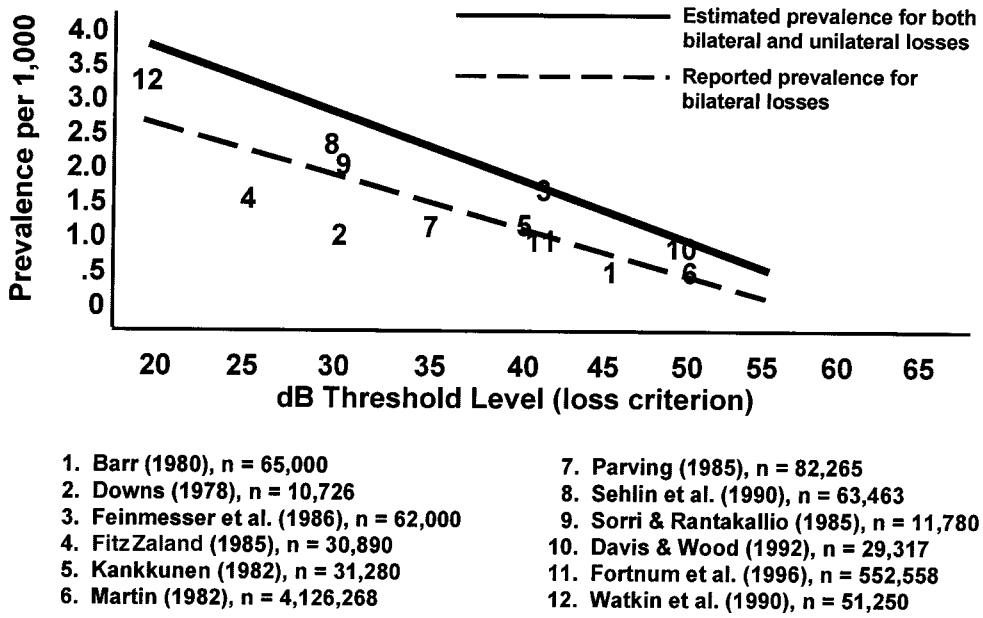
The dashed line in Figure 2 is for children with bilateral permanent hearing loss. How much would the prevalence increase if children with unilateral permanent hearing loss were included? Kinney [1953] and Brookhouser et al. [1991] show that 37–48% of children with permanent hearing loss have unilateral hearing loss. Using the results from these studies to increase the numbers shown in Figure 2 by 40% results in the estimate shown by the solid bold line in Figure 2.

But how many of the children in the studies cited in Figure 2 have late-onset loss (i.e., were born with normal hearing but later acquired a permanent hearing loss)? Although there is not yet definitive data to answer this question, a reasonable estimate can be made based on data from a large multi-center longitudinal study reported by Norton et al. [2000]. Using those data, the Joint Committee on Infant Hearing [2000] estimated that only about 2% of children with permanent hearing loss by 12 months of age had normal hearing at birth.

Coordinators of State EHDI programs that have been functioning for many years also report very few children with late onset losses [NCHAM, 2003b]. For example, Colorado, Rhode Island, and Hawaii have been screening almost all of their newborns since the early 1990s. All of these states have reasonably good tracking systems to identify children with hearing loss as they enter school. These programs also attempt to determine the status of the newborn hearing screening result for any child identified with hearing loss after the neonatal period. These states report that fewer than 5% of the children with permanent hearing loss at the time they entered school had passed the newborn hearing screening test. It is important to remember that some of these could have been congenital losses that were missed by the screening. Thus,

TABLE I. Rate Per 1,000 of Permanent Congenital Hearing Loss in Published Reports of UNHS Programs

Location of program (time)	Cohort size	Primary screening technique	%Refers lost to follow-up	Prevalence per 1,000 of hearing loss
New Jersey, Barsky-Firkser and Sun, 1997 (1/93–12/95)	15,749	ABR	41	3.30
New York, Prieve and Stevens, 2000 (1/96–12/96)	27,938	OAE & AABR	23	1.96
Colorado, Mehl and Thomson, 1998 (1/92–12/96)	41,976	AABR	52	2.56
Texas, Finitzo et al., 1998 (1/94–6/97)	54,228	OAE	31	2.15
Hawaii, Johnson et al., 1997 (1/94–6/97)	9,605	OAE	2	4.15



1. Barr (1980), n = 65,000
2. Downs (1978), n = 10,726
3. Feinmesser et al. (1986), n = 62,000
4. FitzZaland (1985), n = 30,890
5. Kankkunen (1982), n = 31,280
6. Martin (1982), n = 4,126,268
7. Parving (1985), n = 82,265
8. Sehlin et al. (1990), n = 63,463
9. Sorri & Rantakallio (1985), n = 11,780
10. Davis & Wood (1992), n = 29,317
11. Fortnum et al. (1996), n = 552,558
12. Watkin et al. (1990), n = 51,250

Fig. 2. Prevalence rates of bilateral and unilateral permanent hearing loss in population-based studies.

eliminating children with late-onset hearing loss from the data in Figure 2 would have a negligible effect on the estimates of 3–4 per 1,000 with hearing losses of 30 dB or greater.

Based on the consistency of data from newborn hearing screening programs and the epidemiological studies, a reasonable estimate of the prevalence of congenital hearing loss is 2–4 per thousand. If babies with bilateral and unilateral hearing loss of 30 dB or greater are included, the prevalence will be closer to 4 per thousand. If only bilateral hearing loss of 50 dB or greater are included, the prevalence will be significantly lower.

Consequences of Congenital Hearing Loss

Just as the prevalence of congenital hearing loss varies significantly based on the type and severity of loss, so do the consequences. Most children with severe-profound bilateral hearing loss, or what is commonly referred to as deafness, have a great deal of difficulty learning language or succeeding in school. As shown in Figure 3, data collected by Gallaudet University in their annual survey of deaf children compare reading comprehension scores of normal hearing students with those of deaf students [Schildroth and Karchmer, 1986]. These data were collected at a time that most hearing loss was not identified until children were 2½–3 years of age.

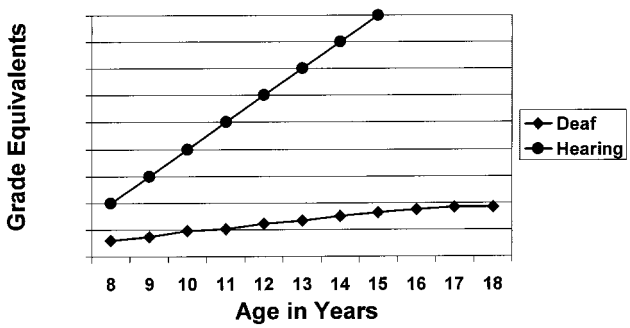


Fig. 3. Reading comprehension scores of hearing and deaf students.

As shown in Figure 3, the average deaf child was already 1½ years behind their hearing peers by the time they entered the third grade. Over time, hearing children continued to make progress with respect to reading comprehension, but deaf children made very little progress in spite of significant effort in special education programs. By the time they are 15 years old, deaf students who do not have the benefit of very early identification are 7.5 years behind their normal hearing counterparts on reading comprehension scores.

Most people are not surprised that children with severe-profound bilateral hearing loss have such difficulty learning to read. Many are surprised to learn, however, that there are serious negative effects associated with much milder forms of hearing loss. Figure 4 summarizes the results of five independently-conducted studies [Keller and Bundy, 1980; Peterson, 1981; Bess and Thorpe, 1984; Blair et al., 1985; Culbertson and Gilbert, 1986] in which children with unilateral loss were matched with hearing peers on various socio-economic and demographic variables (e.g., age, presence of other disabilities, and socio-economic status of the family).

Depending on the study, children in each group were assessed on math, language, and/or social skills. In every study, children with unilateral hearing loss lagged substantially behind the average child with normal hearing. Combining the data across all five studies, children with unilateral hearing loss scored between the 25th and 32nd centile of children with normal hearing, depending on the outcome being measured. To put this in practical terms, these data show that elementary school children with unilateral hearing loss will be 1½–2 years behind their normal hearing peers on math and language scores by the time they are in the fifth grade.

Benefits Associated With Early Identification of Hearing Loss

A frequently asked question about newborn hearing screening is whether babies with congenital hearing loss who are identified earlier do better than those who are identified later. For obvious reasons, it is not practical to randomly assign babies with hearing loss who have been identified during the first few months of life to receive intervention or not. Thus,

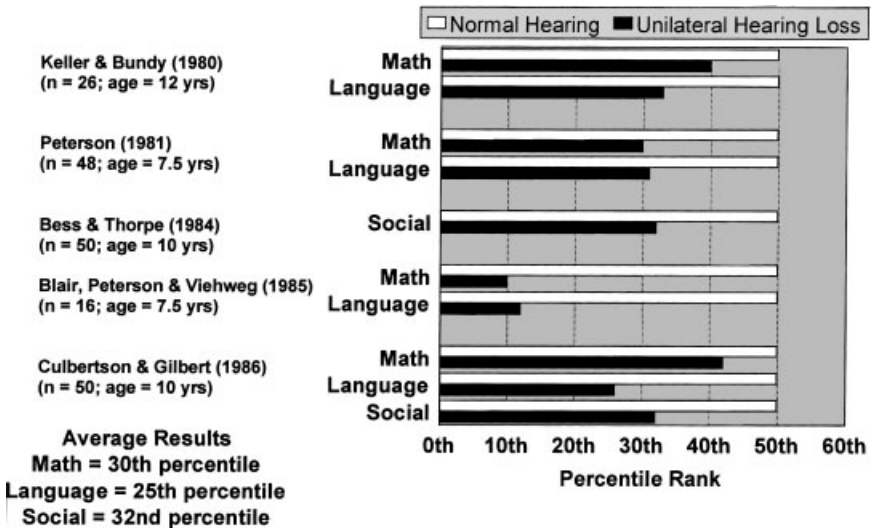


Fig. 4. Effects of unilateral hearing loss.

there are no data from prospective clinical trials that can be used to answer this question. There are, however, a number of retrospective studies in which children have been categorized into groups who were identified earlier versus later, matched on relevant variables, and assessed on developmental outcomes and success in school-related areas [Yoshinaga-Itano et al., 1998; Calderon and Naidu, 2000; Moeller, 2000].

For example, in one such study conducted at Boys Town National Research Hospital, a group of 129 children who were enrolled in an early intervention program were examined retrospectively to estimate the effect of earlier versus later intervention for children with hearing loss. Children in the early intervention program who had been enrolled prior to 6 months of age were compared to a group of 104 children enrolled in the program after 6 months of age. Means scores for the two groups on socio-economic and demographic variables were similar. As a part of the early intervention program in which these children were enrolled, language competency was assessed two times each year by people who did not know that the data would be used later to compare the effects of earlier versus later intervention. As shown in Figure 5, assessments collected at 1.8 years of age showed that the children identified before 6 months of age had a slight advantage over children identified later. The differences between the two groups became larger and larger over time. It is particularly noteworthy that by the time these children were almost 5-years old, those identified before 6 months of age were achieving within the normal range of hearing children.

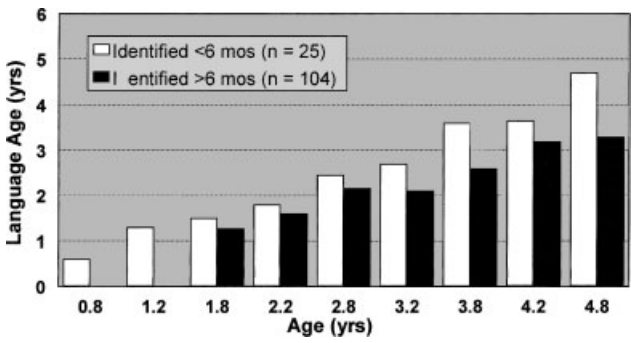


Fig. 5. Boys Town National Research Hospital Study of Earlier versus Later.

Results from the other studies [Yoshinaga-Itano et al., 1998; Calderon and Naidu, 2000] show very similar results. Even though results of randomized clinical trials are not available to answer the question of whether earlier identification leads to better outcomes for children with hearing loss, and even though each of these studies have weaknesses attributable to the lack of random assignment [see NCHAM, 2003c, for a discussion on these weaknesses], the consistency of results from multiple retrospective studies provides convincing evidence about the benefits associated with earlier intervention.

CURRENT STATUS OF EHDI PROGRAMS

With almost 90% of newborns being screened for hearing loss before leaving the hospital, newborn hearing screening is becoming the standard of care throughout the United States [NCHAM, 2003d]. This has led to a realization, however, that for children and families to experience the benefits associated with hearing screening programs, these programs must be linked more effectively to diagnosis, intervention, and family support. As reflected in the Healthy People 2010 goals [HHS, 2000] and described in more detail by the Centers for Disease Control [CDC, 2003], effective EHDI programs consist of at least the following components:

- Screening all newborns for hearing loss before 1 month of age (for most babies, this means they will be screened prior to hospital discharge).
- Obtaining definitive diagnostic data for all referred infants no later than 3 months of age.
- Enrolling babies diagnosed with hearing loss in appropriate early intervention programs before 6 months of age.
- Coordinating EHDI activities with the baby's medical home.[1]

[1]The American Academy of Pediatrics [American Academy of Pediatrics, 2003] defines a medial home as "an approach to providing health care services [...that provides] children and their families...the care that they need from a pediatrician or physician whom they know and trust. The pediatric health care professionals and parents act as partners in a medical home to identify and access all the medical and non-medical services needed to help children and their families achieve their maximum potential."

- Providing culturally competent family support to families of infants identified with hearing loss.
- Having a systematic data management and tracking system that is used for quality assurance and program evaluation activities.

The current status of EHDI programs with regard to each of these components is summarized below based on the results of a survey of State EHDI Coordinators conducted in January 2002 [White, 2003].

Screening

As shown earlier in Figure 1, there has been a dramatic increase in the past 10 years in the number of babies being screened for hearing loss prior to discharge and the number of hospitals doing universal newborn hearing screening (defined here as screening more than 90% of all births or admissions). Interestingly, no particular protocol or type of equipment has emerged as the method of choice. Approximately half of all screening programs use OAE in some way, and approximately 67% use AABR in some way (percentages sum to more than 100 because some programs use both OAE and AABR). Forty-two percent of programs do all of their screening prior to hospital discharge, while about 58% of programs use a two-stage protocol in which screening is not completed until an outpatient screening is done following discharge.

Deciding which newborn hearing screening protocol is "best" depends on the circumstances of the program and personal preferences of the people responsible. One of the most important variables is how difficult it is to get babies to come back for a second-stage or outpatient screening. Programs that have difficulty getting babies back for follow-up tend to complete the screening process prior to discharge. It should also be noted that even though there are hundreds of very successful screening programs, a significant number of hospitals are still struggling with much higher refer rates (>20%) and/or poor coverage (<85%).

Diagnosis Before 3 Months of Age

For babies who do not pass the hearing screening test, audiological diagnosis should be completed as soon as possible, but no later than 3 months of age. Unfortunately, according to State EHDI coordinators, only about half (56%) of reported diagnostic evaluations were completed by 3 months of age. Add to this the large number of children for whom follow-up data are not available (see Table I), and there is clearly a serious problem with the timely completion of diagnostic evaluations.

Such delays in diagnosis are attributable in part to the shortage of experienced pediatric audiologists. In fact, State EHDI Coordinators rated the shortage of pediatric audiologists as the most serious obstacle they face in providing effective EHDI programs. About half the states (57%) have developed written guidelines for conducting diagnostic audiological evaluations, and most (74%) have compiled a list of centers or individuals who are qualified to do diagnostic audiological evaluations for infants under 3 months of age. Unfortunately, there is not general agreement on what constitutes a qualified pediatric audiologist, and these lists are mostly comprised of self-defined pediatric audiologists.

Enrollment in Intervention Before 6 Months of Age

All states are required by federal law to provide appropriate early intervention programs for children with disabilities. However, because most state-funded early intervention programs were developed before hospital-based newborn hearing screening programs became widespread, most programs serving children with hearing loss are primarily serving those with severe to profound bilateral loss. Many of the children being identified are children with mild, moderate, and unilateral losses. Thus, early intervention services for infants and toddlers with hearing loss are in need of being re-designed and re-focused. Furthermore, State EHDI Coordinators estimate that only 53% of infants and toddlers identified with hearing loss begin an appropriate early intervention program by 6 months of age.

Infants who have hearing loss require complex, multifaceted intervention services, including medical, educational, and audiological components. The shortage of experienced and qualified pediatric audiologists often makes it difficult to fit appropriate assistive listening devices as early as desired. The lack of understanding among many primary care physicians regarding early identification of hearing loss also interferes with appropriate referrals to genetics, ophthalmology, and other specialists as recommended by JCIH [2000].

Coordination With the Infant's Medical Home

The American Academy of Pediatrics [2002] advocates that all children have access to a medical home in which care is provided that is accessible, family-centered, comprehensive, continuous, coordinated, compassionate, and culturally effective. For a newborn with hearing loss to receive such care, a primary care physician must be identified soon after the baby's birth who is familiar with that baby's circumstances, knowledgeable about the consequences and treatment of infants with hearing loss, and who is known and trusted by the family.

According to the Maternal and Child Health Bureau [2002], only about 63% of all babies in the United States are connected with such a medical home. State EHDI Coordinators estimate that the name of the physician who will care for the baby during the first three months of life is only known for about 75% of newborns discharged from the hospital. Often, these physicians are not well-informed about issues related to early identification of hearing loss. This is not surprising given the recent rapid expansion of knowledge regarding identification and treatment of hearing loss. It is unrealistic to expect all physicians to remain up to date about a condition that affects only 3 babies per 1,000. Thus, states must find ways of providing this information to physicians on an "as needed" basis. The American Academy of Pediatrics is actively working with State EHDI Coordinators to develop such informational materials, but much remains to be done.

Culturally-Competent Family Support

Having a newborn identified with a hearing loss is a difficult and challenging experience for most families. Most states (77%) provide information for parents about what to do if their child is identified with a hearing loss, but only 18% of states provide these materials in the other languages that are spoken most frequently in their state. State EHDI Coordinators estimate that only 22% of parents are confident about what to do next when their baby is diagnosed with a hearing loss, and only 31% indicate that parents in their state have a range of choices of early intervention programs which emphasize different communication alternatives (e.g., total communication, cued speech, auditory-oral). Most states (72%) indicate that they have a system in their state to assist parents of babies identified with hearing loss in making contact with other parents of babies previously identified with hearing loss.

Systematic Data Management and Tracking

Making sure that babies who are referred from screening programs receive appropriate and timely diagnostic and

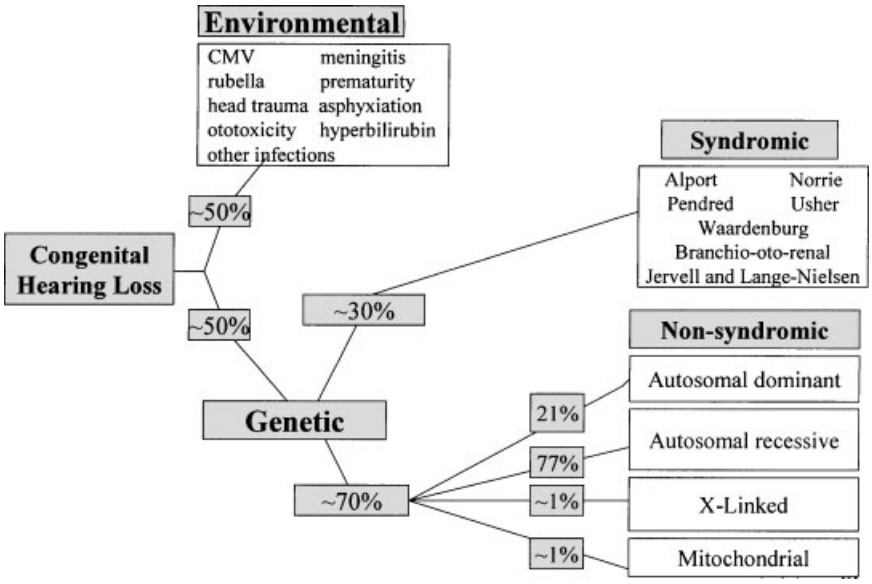


Fig. 6. Causes of hearing loss.

intervention services remains a significant challenge. As noted earlier in Table I, it is not known whether many children who were referred from the screening program ever received a diagnostic evaluation. Although many states are working to develop appropriate tracking and data management systems, this will require several years and much coordination among agencies. In the meantime, the follow-up of children remains one of the biggest challenges to the successful implementation of EHDI programs.

Systematic data collection and reporting is a prerequisite for ongoing program evaluation and quality assurance activities. Unfortunately, only a handful of states currently have adequate data collection and reporting systems in place, which means that little systematic evaluation and quality assurance work is being done.

Summary of Current EHDI Status

The current status of EHDI programs in the United States is like the proverbial glass that can be viewed as being either half full or half empty. Certainly, the likelihood for an infant or toddler with permanent hearing loss to receive timely and appropriate services is better than ever. At the same time, EHDI programs are still a long way from accomplishing the goal set by Dr. Koop in 1990 that "no child [will reach] his or her first birthday with an undetected hearing loss." To reach that goal, significant improvement is needed in the availability of pediatric audiologists, tracking and data management, program evaluation and quality assurance, availability of appropriate early intervention programs, and linkages with medical home providers.

Much has changed since the early 1990s, however. There is now a solid research and experiential basis for addressing the existing weaknesses in EHDI programs. Continuing improvement will require the commitment and resources of state health officials, hospital administrators, health care providers (particularly physicians and audiologists), and parents. The issues that need to be resolved are complex and will require stakeholders to work together over a sustained period of time.

ROLE OF GENETIC SERVICES PROVIDERS IN NEWBORN HEARING SCREENING

As shown in Figure 6, at least 50% of congenital hearing loss has a genetic origin [Gorlin et al., 1995; American College of Medical Genetics, 2002]. In fact, as more is learned about the genetics of hearing loss, we are finding that some hearing loss that was thought to be caused by environmental factors is actually linked to genetics. For example, in 1994, the Joint Committee on Infant Hearing [1995] identified the administration of aminoglycoside antibiotics as a cause of acquired hearing loss. We now know that this is only true for persons who have the mitochondrial mutation, mtDNA A1555G.

Approximately 70% of genetic hearing loss is non-syndromic, and about 30% is syndromic. Some of the most frequently-occurring and/or well-known forms of syndromic hearing loss are listed in Table II. One of the best known, although it occurs relatively infrequently, is Usher syndrome, in which babies born with hearing loss gradually lose their vision.

According to the American College of Medical Genetics [2002] and the Joint Committee on Infant Hearing [2000], families of babies with congenital hearing loss for whom the etiology of hearing loss is not well defined should be offered the option of genetic evaluation and counseling by a medical

TABLE II. Common Forms of Syndromic Hearing Loss

Syndrome	Main features (in addition to hearing loss)
Alport	Renal disease
Branchio-oto-renal	Neck cysts and renal malformation
Jervell and Lange-Nielsen	Heart problems
Neurofibromatosis Type 2	Nerve tumors near the ear
Pendred	Thyroid enlargement
Stickler	Unusual facial features, eye problems, arthritis
Usher	Progressive blindness
Waardenburg	Skin pigment changes

Adapted from Rehm et al. [2003].

geneticist. Unfortunately, most infants and toddlers with congenital hearing loss are not referred for a genetic evaluation, probably in part because many people don't understand the benefits of genetic testing for children with hearing loss. For example, in a statewide evaluation of the EHDI program done in Utah, a random sample of primary care physicians were asked to which medical specialists they would typically refer one of their patients who had "a moderate to profound bilateral hearing loss" but no definitive etiology. Only 38% indicated they would refer the child's family to a medical geneticist [Utah Department of Health, 2001].

Many people assume that the only reason to do a genetic evaluation is to estimate the probability of subsequent children in the family having a genetically-linked hearing loss. Although genetic testing enables us to estimate the chance of reoccurrence, there are many other important reasons for children with congenital hearing loss to receive a genetic evaluation and genetic counseling. For example, because so much hearing loss is syndromic, a reasonably cautious physician would need to order additional tests (e.g., electroretinograms, temporal bone imaging, electrocardiograms) to make sure that there are no associated kidney, heart, vision, or other problems. These tests are time-consuming, expensive, and often not convenient for the family. Many of these tests will be unnecessary if a genetic evaluation is done.

Genetic testing can also lead to better health care for the child. Specific examples of how this can happen are given by the American College of Medical Genetics [2002] and include:

- Vitamin A therapy may be beneficial in slowing retinal degeneration in a child with Usher syndrome.
- Early treatment of children with Jervell and Lange-Nielsen syndrome can reduce cardiac problems associated with the hearing loss.
- Biotin treatment for children diagnosed with biotinidase deficiency prevents hearing loss.
- Avoiding aminoglycoside antibiotics in children with the mitochondrial mutation mtDNA A1555G reduces the risk of developing hearing loss.

Even in cases where preventive treatments are not yet available, knowledge about syndromic hearing loss enables the medical home provider to anticipate and monitor for possible health problems associated with the child's hearing loss. For example, children with Stickler syndrome should be monitored for early retinal detachment, and children with branchio-oto-renal (BOR) syndrome should receive periodic renal examinations to identify kidney complications.

Genetic testing is also useful to dispel misinformation or concerns the parents have about what may have caused the hearing loss. Knowing that a naturally-occurring genetic mutation is responsible for the hearing loss helps many parents who are worrying about whether the hearing loss may have been caused by something they did or did not do during the pregnancy.

CONCLUSIONS

The importance of early identification and treatment of congenital hearing loss is well established based on the prevalence of the condition, the negative consequences of not intervening, and the benefits associated with early intervention programs. This information, coupled with improved equipment and procedures for newborn hearing screening has led to rapid expansion of EHDI programs. Although much remains to be done to improve EHDI programs, remarkable progress has been made during the last 15 years.

One area that has lagged behind in the development of comprehensive EHDI programs is the widespread involvement of genetic service providers. As noted earlier, a successful EHDI program must include the following components:

- Hearing *screening* of all newborns before 1 month of age.
- *Diagnosis* of referred infants before 3 months of age.
- Provision of appropriate *intervention* services to infants with hearing loss before 6 months of age.
- Coordination of services by the child's *medical home*.
- Provision of culturally competent *family support*.

Providers of genetic services have an important role to play in all of these components. Because so much congenital hearing loss has genetic origins, genetic service providers are essential to the *diagnostic* and *intervention* components. Not only can they assist in determining the etiology of the hearing loss, but the results of a genetic evaluation can indicate whether the loss is progressive and whether other health care concerns need to be addressed. Genetic services are also an important part of the comprehensive services that should be provided by the *medical home* and can contribute in important ways to providing the *family support* (both informational and emotional) needed when a child is first identified with hearing loss. Although it is not yet practical, it is very likely that evolving new technologies involving microarrays and other kinds of high through-put testing using newborn screening dried bloodspots will result in genetic analysis being used as a supplement to physiological hearing *screening* [American College of Medical Genetics, 2002]. This will be particularly important for those babies who are at risk for late-onset loss related to genetic conditions.

Thus, it is important for providers of genetic services to become informed about recent developments and the current status of EHDI programs, and to help the various stakeholders, particularly parents, primary care physicians, and public health officials, understand the valuable contribution that genetic service providers can make to effective EHDI programs.

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