Conference Endnote Address:
A Quick Look Back and a Charge Going Forward

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Abstract

“If, when a child becomes two years old, they suspect that he is handicapped by deafness or partial deafness, they may apply to their Local Education Authority, which must arrange for a medical examination.” (Ewing & Ewing, 1947)

There is a long and storied history of newborn hearing screening and intervention in our professions. This history reflects the efforts of numerous pioneers all of whom have contributed to remarkable improvements in the lives of children with hearing loss and their families over the decades. Today, we have the knowledge, technology, and systems in the developed world that will pave the way for children with hearing loss to maximize their potential and live satisfying and productive lives. However, there is still much work to do. There are hearing healthcare disparities within countries and across the globe – disparities that might be reduced by technological linkages between the developed and undeveloped worlds. Furthermore, we are beginning to see new opportunities for genetic screening for hearing loss and a possibility of identifying those children who have unapparent hearing loss at birth. The future promises the possibility of our preventing, not just treating, some types of childhood hearing loss. This presentation highlights some of the many opportunities ready for us to embrace today and some awaiting us tomorrow – opportunities that we can hardly imagine today.

A Look Back

One thing we can always count on is that time will change our perspective on things – the way we view things from the past, the way we view things in the present, and the way we will view things in the future. In 1913, the Children’s Bureau (now the Maternal Child Health Bureau) published the first in a series of monographs on mother and infant care in response to high infant mortality rates throughout the United States (U.S.). As stated on the opening pages of one monograph, “It (the monograph) endeavors to present the accepted views of the best authorities at the present time” (West, 1914; p. 7). Our views of the recommendations from that series of monographs have definitely changed over time. For example, one recommendation to new mothers was “The beneficial effect of sunlight is best obtained when the rays reach the skin directly. Clothing or ordinary window glass keeps out the ultra-violet rays – the rays that prevent and cure rickets. Sun baths may be begun when the baby is about 3 or 4 weeks old” (West, 1914; p. 42). Of course, we view a baby’s exposure to direct sunlight much differently now. Today, the American Academy of Pediatrics recommends that babies “avoid direct sunlight” (Hagan, Shaw, Duncan, 2008; p. 5). Numerous examples of such scientific opinions that were widely accepted at the time but are rather alarming today, exist in these early publications.

Just as standards of care for infants have changed over time, our views about the appropriate care of children with hearing loss have changed as well. In 1947, Ewing and Ewing, preeminent leaders and advocates of early intervention and education for deaf children in England, wrote a comprehensive account of their views and experiences entitled “Opportunity and the Deaf Child”. In this text, the Ewings stated “If, when a child becomes two years old, they (parents) suspect that he is handicapped by deafness or partial deafness...they must arrange for a medical examination” (pp. 8-9). Almost 10 years later, they modified their recommendations in “New Opportunities for Deaf Children”, stating “…the writers are convinced that all babies should be given screening tests of hearing, by the ninth to twelfth month”
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In the 1990s, audiologists transitioned from behav-
orial screening methods to physiologic methods – audi-
tory brainstem responses and otoacoustic emissions.
Enthusiasm with those new approaches to screening
led to an eagerness to implement universal screening
programs throughout the U.S. Although few would
argue that early identification of hearing loss was not a
noble objective, debates ensued in this country about the
best way to proceed with implementation (e.g., Bess &
Paradise, 1994). The disagreements generated by these
deliberations stimulated numerous studies that resulted
in a body of research that demonstrated the feasibil-
ity of mass newborn hearing screening and, ultimately,
the benefits of same. Today, we have newborn hearing
screening tools with good reliability, sensitivity, and
specificity. Although some countries are still struggling
to get screening programs in place, feasibility has been
demonstrated in numerous countries including Aus-
tralia, Canada, the U.S, and some European countries
where between 80% and 97% of newborns are screened
(Lehnhardt, 2009; NIDCD, 2011). As a result of these ef-
forts, permanent hearing loss in infants is now identified
on average by two to four months of age, as opposed to
two to three years of age just a couple of decades ago in
the U.S. (White, Forsman, Eichwald, & Munoz, 2010).

And, while we have successfully implemented new-
born screening programs, we are still challenged by
initiating timely intervention services. Reports from the
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that only 57% of those babies who do not pass their
newborn screenings are documented as having received
follow-up care. Although no strong evidence exists iden-
tifying the factors that contribute to loss-to-follow-up,
numerous reasons have been proffered including family
proximity to resources (ASHA, 2008). Today, we are
just beginning to see the vast possibilities of widespread
telepractice for reducing the impact of distance from
early intervention resources for families. Through tele-
practice, we can now provide remote newborn hearing
screening, audiologic assessments, hearing aid support
services, and cochlear implant programming. In addition,
we can provide remote consultations with parents and
professionals all around the globe, and we can provide
eyeal intervention services directly in a child’s home. We
are only seeing the tip of the iceberg in terms of what
we can accomplish with telepractice (Eikelboom, Atlas,
Mbao, & Gallop, 2002). Forty-two percent of states in
the U.S. report having implemented some telepractice
efforts for providing intervention to infants and children
with hearing loss (NCHAM, 2010). Of those 42%, 79%
are in the planning or pilot stages – clearly, we have a
long way to go but the promise of effective intervention
through remote practice is upon us. We can look forward
to reducing the hearing health care disparities currently
in existence around the world by engaging these new
technologies.

A Charge Forward

Just like the Children’s Bureau publications in the
1920s, throughout this conference our speakers have
attempted to present the most current, state-of-the-art
views on issues related to childhood hearing loss. No
doubt that in years to come, we will get a chuckle from
some of the views presented over the last few days – per-
haps some of our current ideas will not be successful or
long lasting. But, we are still challenged to face the same
questions faced by those who came before us. Where
do we go from here? What are our future directions for
improving the lives of those with hearing loss and their
families? Are there new and improved ways to identify
hearing loss early?

One such challenge is how we will face the inevitab-
ility of genetic screening for hearing loss. At least two
thirds of permanent hearing loss present at birth has a
genetic cause and 70% of that is non-syndromic. But,
even in the case of syndromic hearing loss, symptoms
may be non-apparent at birth. Hearing loss is genetically
quite heterogeneous and involves mutations in many
genes (Van Camp, Willems, & Smith, 1997). Therefore,
selecting a single gene or a group of genes for testing will
not allow for the prediction of hearing loss risk. For each
of the six most frequent causative genes that have been
identified to date for autosomal recessive non-syndromic
hearing loss (GJB2, SLC26A4, MYO15A, OTOF, CDH23, TMC1), at least 20 mutations have been reported (Phillips et al., 2013). GJB2 is a small gene that contains a single coding exon but over 300 different mutations have been reported (Human Gene Mutation Database, n.d.). It is suspected that over 500 genes, with some containing hundreds of mutations, are likely to contribute to hearing loss. Furthermore, interactions among several genes could be required for some individuals to exhibit hearing loss. Despite the current availability of some limited testing for specific genes that are believed to cause hearing loss, the reality of genetic complexity still makes it difficult to predict which newborns will develop hearing loss.

To counter this limitation in our current ability to identify genetic hearing loss, it is likely that technology will yield the possibility of entire genome sequencing within 10 years. Although that is an exciting prospect, it is accompanied by many challenges. Moving from genetic screening for babies known to have hearing loss to population-based screening will require considerable public input, advances in technology, and improved knowledge of genetic causes of hearing loss. The benefits of such screening and the potential limitations will need careful consideration – much like our transition from high-risk screening to universal screening for hearing loss in the 1990s. However, the stakes today are likely even higher. Incidental findings will be a concern (Phillips et al., 2013). That is, if the whole genome is sequenced, what are our ethical or moral obligations to report all mutations that could give rise to future genetic diseases rather than just the mutations associated with hearing loss? It is probable that one-disease/condition screening programs will no longer be sustainable given the implementation costs. Advocates for early identification of hearing loss in infancy will likely be part of a larger genetic screening initiative for multiple childhood conditions and disorders in the future.

In addition to genetic testing of infants, there are efforts underway that can potentially prevent hearing loss in children that are not yet fully understood or commonplace in our provision of care. One such example is exploration of the audiologic impact of aminoglycoside exposure in neonates with MTRNR1 mutations. These mutations lead to hearing loss in patients treated with aminoglycoside antibiotics, which is distinct from the ototoxicity associated with therapeutic levels of these drugs. However, the precise role of MTRNR1 mutations in pediatric aminoglycoside ototoxicity has not yet been fully explored. The American College of Medical Genetics recommends testing for these mutations in patients who have been identified with hearing loss (ACMG, 2002). In the future, we may be able to genotype mothers while pregnant to determine which infants are at increased risk of aminoglycoside ototoxicity and utilize alternative treatments.

Another challenge to identification of early hearing loss is congenital cytomegalovirus (cCMV), the leading cause of non-genetic sensorineural hearing loss in children. Ninety to ninety-five percent of infants born with cCMV will have no apparent clinical abnormalities at birth and will not be identified via routine examination. Therefore, infants with cCMV are likely to pass a newborn hearing screening only to have 10-15% develop hearing loss later in childhood. Clearly, if we could identify those babies with cCMV, they could receive targeted audiologic monitoring resulting in quick intervention as indicated. Although definitive testing for cCMV is available, such methods are not automated and, therefore, are not suited for mass newborn screening. Studies have been ongoing to determine fast and effective methods for mass cCMV screening of newborns. Evaluation of polymerase chain reaction (PCR) screening methods that use dried blood spots and methods that assay infant saliva have been the focus of research attention in the last decade (e.g., Boppana et al., 2011).

Another approach to prevent hearing loss, as opposed to simply identifying/treating, is through the use of protective agents when utilizing ototoxic chemotherapies. A good example of such chemotherapies is cisplatin, one of the most ototoxic drugs known, for the treatment of pediatric cancers, resulting in hearing loss in between 20% to 90% of children receiving the drug. Young children are at the greatest risk for acquiring hearing loss from cisplatin, which is typically permanent, bilateral, and high frequency. Sodium thiosulfate has provided protection against platinum-induced hearing loss in animals and human patients and several other thiol-based agents have also shown otoprotection in animal models (Dickey, Muldoon, Kraemer, & Neuwelt, 2004; Doolittle et al., 2001).

The discussion herein suggests a fundamental change in the future of newborn hearing screening, and identification and management of childhood hearing loss more generally. Whereas the past and current purpose of screening has been to identify children who have hearing loss, the future goal is likely to include additional components of preventing hearing loss and identifying children who are likely to have hearing loss but are not yet manifesting symptoms. Performing anticipatory testing does not identify patients with a dis-
ease per se but, rather, those with an increased risk for an adverse reaction to medication, or those with special dosing requirements, thus allowing for the avoidance of certain medications that could result in hearing loss. Perhaps we will return to a type of “high-risk” screening after all. And, rather than requiring children and families to travel to large healthcare institutions for services, we may be reaching out to them in their own remote communities via telepractice. The elimination of hearing health care disparities around the globe is almost within our reach!

References


