Impact and benefits of early hearing screening

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8 GENERAL DISCUSSION AND CONCLUSION

In the DECIBEL-study several aspects concerning permanent childhood hearing impairment and newborn hearing screening are studied.

In this thesis the following objectives have been addressed:

1. Determine the aetiology of permanent childhood hearing impairment in children in the Netherlands.
2. Determine the sensitivity of the newborn hearing screening program and the characteristics of the children unidentified by NHS.
3. Study the effect of newborn hearing screening on developmental outcome in 3-5 year old children with permanent childhood hearing impairment compared to children in distraction hearing screening.

In this chapter the findings on these subjects are summarized and discussed. Furthermore suggestions for future research and policy on these topics are presented. It closes with remarks on the relevance of the study and final conclusions.

OBJECTIVE 1:
Determine the aetiology of permanent childhood hearing impairment in children in the Netherlands.

WHAT WAS KNOWN?

The cause of permanent childhood hearing impairment is generally assumed to be hereditary in 50%, non-genetic in 25% and unknown in 25%.[6-8] However, because of methodological and national (policy, immunization programs) reasons, the reported proportions attributed to the different aetiological causes differ greatly. The leading non-genetic cause of permanent childhood hearing impairment is reported to be congenital cytomegalovirus infection[10] and isolated hearing loss the most frequent long-term consequence.[57;58] Although most infants with a congenital cytomegalovirus infection do not show any signs or symptoms at birth (85%), about 15% will develop permanent sequelae including hearing loss.[11-13] It has been suggested that newborn hearing screening may fail to detect children with progressive or delayed-onset hearing loss linked to congenital cytomegalovirus infection.[59]

WHAT IS NEW?

Neither the systematic review nor the results of diagnostic investigations in children participating in the DECIBEL-study support the general assumptions on the proportions figure (CHAPTER 2).[6-8] The following distribution is proposed: 40% hereditary origin, 30% acquired origin and one-fourth of unknown cause. A category of miscellaneous causes is added which makes up the total of 100%. The proportions in this aforementioned calculation were found to vary with the degree of hearing loss. The results of the systematic review and the DECIBEL-study supported the importance of diagnostic investigations to identify the aetiology of hearing loss. Even when only one specific congenital infection (cytomegalovirus) and limited genetic diagnostic investigations (without clinical consultation) were performed, the cause of permanent childhood hearing impairment was identified in a substantial proportion of children.

Congenital cytomegalovirus was detected in 23% of the children with profound hearing loss and in 8% of all young children with hearing loss participating in the DECIBEL-study (CHAPTER 3). The general and language developmental outcome of children with permanent childhood hearing impairment and congenital cytomegalovirus infection was found to be significantly lower than the development of children with permanent childhood hearing impairment without congenital cytomegalovirus infection.

We found that doctors involved in mother and child care in the Netherlands do not posses optimal knowledge on cytomegalovirus infection (CHAPTER 4). Not only did doctors miscalculate the

chance of encountering a child with congenital cytomegalovirus infection in medical practice, but they were also not well informed about the risk factors for transmission and symptoms. Including congenital cytomegalovirus infection in the differential diagnosis in symptomatic newborns is crucially important; otherwise children may be left undiagnosed, with possibilities for treatment and follow-up not explored.

WHAT NEEDS TO BE DONE IN POLICY?

Parents deserve thorough information about possibilities for aetiologic diagnostic investigations. This is important not only for reasons of family-planning but also with regard to the prognosis of development of their child with hearing loss. It is essential for professionals caring for these children (pediatricians, otolaryngologists, general practitioners and audiologists) to be aware of local or national multi-disciplinary initiatives for specialty consultation. Improved, standardized and systematic investigations of the aetiology of permanent childhood hearing impairment will finally decrease the number of unknown causes.

As to date, prevention is the only universal opportunity to decrease the proportion of children with congenital cytomegalovirus infection, awareness and knowledge of professionals involved in mother and child care in the Netherlands needs to be improved. We consider that publications on this topic, not only in scientific but also in general media, will contribute to this.

WHAT NEEDS TO BE DONE IN RESEARCH?

Although we did not find that hereditary causes account for 50%, it is likely that the proportion attributed to genetic origin will increase when the possibilities for diagnostic investigations develop further and are more widely used in the next few years. Parents and sibs of children participating in the DECIBEL-study contribute for that reason in further research by the otogenetic laboratory of Radboud University Nijmegen Medical Centre.

It would be of interest to study the developmental outcome of children with permanent childhood hearing impairment caused by factors other than congenital cytomegalovirus infection. In that way children who need specific habilitation programs can be identified and their developmental prognosis may be more accurately determined. Since the presence of congenital cytomegalovirus infection is related to the developmental outcome in children with hearing loss, it is important that possibilities for screening, follow-up and treatment of these children are explored.

OBJECTIVE 2:
Determine the sensitivity of the newborn hearing screening program and the characteristics of the children unidentified by NHS.

WHAT WAS KNOWN?

Newborn hearing screening has been introduced to identify hearing loss early in life. However, sometime during childhood children may not only acquire hearing loss, but some may also present with delayed onset and progressive hearing loss. Earlier studies have provided figures on the prevalence of hearing loss in live newborns with reports varying between 0.94[3] to 1.18[2] per 1000, between 1.33 (CI95% 1.22-1.45)[4] and 1.44 per 1000(CI95% 1.41-1.48) at the age of 5-10 years and 1.63 per 1000 (CI95% 1.59-1.67) at the age of 8-13 years.[5] This increase in prevalence during childhood suggests that, with a newborn hearing screening program, it is not unlikely that approximately 40% of children with permanent hearing loss of congenital cause are unidentified and need to be diagnosed later than the neonatal period. One earlier (smaller) study determined that the proportion of true cases identified by NHS was 92%.[94]

WHAT IS NEW?

As described in CHAPTER 5 we found that the prevalence of permanent childhood hearing impairment in the 2003-05 cohort in the Netherlands following newborn hearing screening was 0.97 per 1000 live newborns. This prevalence of hearing loss at birth is in accordance with international estimates. However, the number of children known with permanent childhood hearing impairment at the age of 5, whom we could identify in our country, was much lower than expected. The national immunization program for children, the screening for irregular erythrocyte antibodies and Human Immunodeficiency Virus in pregnant women and also the low prevalence of congenital cytomegalovirus infection in our country could have contributed to this lower than expected prevalence. We found that the sensitivity of the current newborn hearing screening program was high. The proportion of children unidentified by newborn hearing screening but with permanent childhood hearing impairment at the age of 3-5 years was 0.19 per 1000 newborns screened (documented acquired hearing loss excluded). Of these children the majority presented for evaluation of hearing before the age of 3 (75%) and had a moderate degree of hearing loss (>50%). The latter suggests that the hearing level of these children could have been just under the detection threshold used in newborn hearing screening. Since the group unidentified by newborn hearing screening was not very large we were not able to define a main cause of hearing loss in the children with an initially negative screen at newborn hearing screening.

Compared to the previously used DHS, all program characteristics increased importantly: It is reassuring for professionals that the proportion of children unidentified by NHS in not that large. For parents it is comforting that NHS leads to earlier diagnosis and has a much larger positive predictive value compared to DHS.

WHAT NEEDS TO BE DONE IN POLICY?

Newborn hearing screening is a valuable and expensive resource and regular evaluation is necessary to optimize its performance. If one would strive to identify all children with true permanent childhood hearing impairment by newborn hearing screening, the detection level for hearing loss should be decreased or the implementation of additional hearing screening programs should be explored. To identify (among all live newborns screened) the currently unidentified proportion of children per year (38/195.000), an enormous (financial and practical) effort would be necessary.

The results of our study suggest that not all children identified with a positive screen for hearing loss were known to the Audiology Center at the age of 3-5 years. This may be caused by late mortality (especially in neonatal intensive care graduates), emigration or a lower degree of hearing loss during diagnostic evaluation. An adequate and secured track and chase system following newborn hearing screening could improve our insight into the yield of newborn hearing screening. A well-managed database, to which the Audiology Centers provide input on definite results concerning the hearing of an individual child, would facilitate regular evaluation. Cooperation in such a project by all partners involved in child hearing is essential to monitor and optimize the quality of newborn hearing screening in the near future.

When discussing options for future research to optimize the yield of newborn hearing screening even further, the abovementioned remarks on the relatively high sensitivity of the current hearing screening program should be kept in mind. It is not likely that all suggestions proposed below, will be cost-effective in the light of the results presented in this thesis.

WHAT NEEDS TO BE DONE IN RESEARCH?

It has been suggested that another moment (and probably type) of hearing screening later in childhood, in addition to newborn hearing screening, would be essential to identify all children with hearing loss. Future research is necessary to determine the feasibility and the cost-effectiveness of this 'second' hearing screening. Choices should be made concerning the age at screening, the population to be screened (all children or high risk) and the method used (physiological or behavioral-observational). The current hearing screening at school-age is therefore a program to evaluate. Although we found that up to the age of 5 the absolute increase in number of children with permanent childhood hearing impairment is not large, evaluation of

this school-age screening program will ensure us if we did not overlook a relevant number of children.

It has also been suggested that the yield of newborn hearing screening could be increased by the introduction of screening for the presence or absence of causative agents for hearing loss. One could think of screening for connexine 26 (the major cause of hereditary hearing loss) or screening for congenital cytomegalovirus. Children with a positive screening result for congenital cytomegalovirus could be closely followed up audiologically to detect hearing loss as early as possible and even treatment can be discussed. Currently a randomized clinical trial is planned by the Leiden University Medical Center. The results of this trial should provide us with information on the cost-effectiveness of audiological (and developmental) follow-up of congenital cytomegalovirus positive children and with information on the feasibility of anti-viral treatment in this population.

OBJECTIVE 3:
Study the effect of newborn hearing screening on developmental outcome in 3-5 year old children with permanent childhood hearing impairment compared to children in distraction hearing screening.

WHAT WAS KNOWN?

Auditory input is considered to be essential for adequate development and social functioning. Therefore it seems important to have information on the child's hearing abilities early in life in order to create opportunities for early amplification and habilitation. Earlier studies on this topic were descriptive in nature and based on convenience samples. In these studies early hearing screening and subsequent timely intervention was found to have positive consequences for speech and language skills.[21;31;37;87-91] When compared to children with permanent childhood hearing impairment with no screening or only targeted screening of high risk infants, children following newborn hearing screening were found to have a broad range of developmental advantages in childhood. [21;31;34;89;108-110] Children with hearing loss were found to be at risk in areas such as behaviour, emotion and quality of life.[111] Improved developmental outcome, however, is to be expected only when early identification of permanent childhood hearing impairment is followed by early amplification and intervention.[21] The Joint Committee on Infant hearing recommends that intervention following a refer at hearing screening should start no later than at the age of 6 months.[15]

WHAT IS NEW?

To overcome financial, practical and ethical considerations precluding the initiation of a randomized trial to date[21;37], we used the regional differences in the assignment of type of hearing screening created by national policy, as an instrumental variable. Since the type of hearing screening offered was independent of the prognosis of hearing of the individual child, this study, with its 'naturally' randomized design, is expected to be as credible as a randomized trial.[38-41]

We found that early hearing screening by newborn hearing screening results in better language and general development in children with permanent childhood hearing impairment at the age of 3-5 years. We also found a significantly better social development and quality of life when compared with children in distraction hearing screening (CHAPTER 7). The advantage for children in newborn hearing screening was even larger, when children with a known abnormal development caused by a congenital infection with cytomegalovirus, the presence of which could be known prior to hearing screening, were excluded from the analysis. Taking the aetiology of

hearing loss into account appeared to be essential when judging the developmental outcome of children with hearing loss.

Early hearing screening was not followed by early intervention in all the children in our study (CHAPTER 6 AND 7). The results of our study suggest that, as well as implementation difficulties, professionals at Audiology Centres differed in their advice to parents concerning the need for amplification and/or parents varied in the priority they gave to amplification, due to their disbelief in the presence of PCHI or the importance of habilitation for PCHI. The degree of hearing loss was an important predictor of age at amplification, with the children with moderate degrees of hearing loss being amplified the latest. The delay between identification and amplification might have resulted in a reduction of the developmental differences found between children with permanent childhood hearing impairment in the newborn hearing screening and the distraction hearing screening.

Since the newborn hearing screening program, as well as the hearing screening for neonatal intensive care graduates have now been running for some years, it is more likely that amplification following a positive screen for hearing loss is achieved earlier nowadays in the Netherlands.

In summary, in the DECIBEL-study we were the first to confirm in a pseudo-randomized nationwide study, that newborn hearing screening leads to a broad range of advantages in outcome in childhood. It is important to realize that the development of children with permanent childhood hearing impairment who were offered newborn hearing screening is still not comparable to that of a normally developing child with normal hearing.

WHAT NEEDS TO BE DONE IN POLICY?

For continuous evaluation of the performance of hearing impaired children following newborn hearing screening it would be useful when the earlier mentioned track and chase system also requested information on developmental outcome. The development of all children with permanent childhood hearing impairment was found to be not regularly assessed by health care professionals. When it was evaluated, it was unclear if this was initiated because of suspected underperformance of the individual child or simply because it was the policy of the Audiology Center where the child was known. We advise regular assessment and the use of the results of ´gold standard´ methods to evaluate development to ensure the input for the track and chase system (non verbal intelligence test, Reynell test for expressive and receptive language) until validation of parental questionnaires is completed.

Professionals and parents should be aware of the importance of timely intervention. Although habilitation can be regarded as a personalized path, international recommendations on the age of diagnosis and start of amplification cannot be 'freely' interpreted. We hope that the results

of the DECIBEL-study, in which all Audiology Centers willingly cooperated, will contribute to the understanding of the importance of the recommended time-frame periods following a positive screening for hearing loss at newborn hearing screening.

WHAT NEEDS TO BE DONE IN RESEARCH?

Newborn hearing screening and the age at start of intervention are important factors in the prognosis of developmental outcome in children with permanent childhood hearing impairment. For a better understanding it is essential to identify the contribution of other variables (e.g. aetiology, communication, habilitation and education). Information on these variables has been collected in children participating in the DECIBEL-study and is available for further analysis.

Children admitted to a neonatal intensive care unit were, with regard to their developmental prognosis, not included in the analysis on developmental outcome. More research is necessary to determine the effect of early hearing screening on developmental outcome in this high-risk population.

RELEVANCE

A nationwide screening program cannot exist without regular evaluation and (if necessary) adjustments. The results as presented in this thesis give an overview of the yield and the effectiveness of the current newborn hearing screening program and insight in the characteristics of children with hearing loss.

This study covers several aspects concerning the care for children with permanent childhood hearing impairment. First, the results are important for national and international policymakers in developing further screening- and preventive strategies. Secondly, the results give professionals insight into the prevalence of childhood hearing loss and the characteristics of these children. Thirdly, the results are expected to contribute to the improvement of timing of diagnostics and intervention programs. Finally, we hope that in the future the results will assist parents of children with permanent childhood hearing impairment in achieving optimal development of their child in the hearing world.

FINAL CONCLUSION

This is the first time developmental outcome following hearing screening in children with permanent childhood hearing impairment was studied in a unique, pseudo-randomized design. We found that early detection of hearing loss by newborn hearing screening (and subsequent early intervention) is beneficial for the development of children with permanent childhood hearing impairment, although their development at 3-5 years is not yet comparable to that of children with normal hearing. We showed that the yield of the current hearing screening program is large, but that there is ample room for improvement in the timing following early detection of hearing loss. We would like to emphasize that the aetiology of permanent childhood hearing impairment should be investigated and the cause taken into account when evaluating the development of the child.