Systematic screening and new genetic methods help ensure early diagnostics, treatment and habilitation of infants with congenital hearing loss

Infants who cannot hear

Deafness and serious hearing impairment occur in somewhat more than one in one thousand newborns. New discoveries in the physiology of hearing and technological innovations in hearing aids and cochlear implants have provided a basis for examining newborns for hearing loss and thus also for early treatment and habilitation. Hearing screening of newborns is undertaken in maternity wards and neonatal intensive care units.

A precondition for introducing screening for a disease in a population is that appropriate treatment of this disease is available (1). Searching for diseases that cannot be treated is not very meaningful. Further requirements for screening to be introduced include that the condition in question must represent a considerable health problem, that suitable examination methods with high sensitivity and specificity are available and accepted in the population, that the screening procedure and the treatment are cost-effective and that they are provided as a permanent scheme. These principles were formulated in 1968 and remain the valid guidelines issued by the World Health Organization. This issue of the Journal of the Norwegian Medical Association includes a presentation of a study of hearing loss in neonates (2) that complies with the regulations for screenings. This was started several years before the national guidelines for hearing screening of neonates became available in 2006 (3).

Early diagnosis of deafness and hearing impairment in children in Norway is based on measurement of otoacoustic emissions as an expression of functioning outer hair cells in the cochlea (4). Furthermore, auditory brainstem response audiometry combined with measurement of stable, frequency-specific auditory responses are necessary for determining a likely hearing threshold, which is a precondition for choosing an appropriate habilitation method.

Nelson and collaborators, who have written the article, have undertaken a number of improvements of the screening method over a period of ten years (2). Infants who do not pass the screening must be given especially high priority in further examinations at hearing centres, so that habilitation can be initiated at the recommended age, from six months. To detect hearing loss that develops after birth, the authors have demonstrated the importance of paying constant attention to language development seen in relation to hearing.

The introduction of new vaccines in the national vaccination programme, especially the vaccine against rubella, has caused intrauterine infections to decrease as a cause of deafness and hearing impairment. Better control of environmental causes of hearing impairment has caused the relative importance of hereditary hearing loss to increase. Developments in medical genetics have enabled us to detect the cause of early deafness and hearing impairment more frequently. Comprehensive registries of genes and gene loci that are associated with hereditary hearing loss have been established (5). More than 140 genes have been identified. A considerable proportion of the population are carriers of such genes. A preponderance of autosomal recessive transmission means that deafness most often occurs completely unexpectedly in families, and most deaf and seriously hearing-impaired children have parents with normal hearing. The Norwegian Internet portal for medical genetic analyses (6) provides an overview of the genetic analyses that are undertaken in Norway with regard to deafness.

Digital hearing aids were brought into use approximately 20 years ago and have significantly improved the habilitation of infants and toddlers whose remaining hearing is sufficient to benefit from a hearing aid. The development of cochlear implants occurred at the same time as hearing screening became possible (7, 8). The preconditions for achieving good results in the development and use of the implant included close collaboration between institutions with competence in ear surgery, audiology, neurophysiology, microelectronics, pedagogy and linguistics. This could bring forth the knowledge that was necessary for development of methods for direct electrical stimulation of the acoustic nerve and the central auditory pathways. In deaf children, it was established in particular how important early exposure to sound via a cochlear implant is for achieving appropriate language development. Around the turn of the millennium, the use of multichannel cochlear implants with up to 25 frequency-coded electrodes increased in children in steadily younger age groups, with good results. As a result of early treatment and habilitation, most of these children can go to regular schools when provided with necessary educational support.

As of the end of 2013, altogether 576 children under 18 years had been provided with a cochlear implant in Norway (K. Rasmussen, the CI unit, Rikshospitalet, personal communication). The number of children who underwent surgery remained stable during the period 2000–13, with approximately 37 patients per year. In 2000 no children underwent surgery before the age of two years, while the average age in 2012 amounted to nine months for children whose impairment had been detected by the neonatal screening. This shows that early diagnosis in a successful screening programme over a relatively short time has resulted in considerable improvements in the habilitation of deaf and seriously hearing-impaired children. In light of figures from comparable countries, Rikshospitalet covers the estimated need for the required number of cochlear implants at the appropriate time for taking full advantage of neuronal plasticity and sensitive periods in the development of auditory and language functions in children (9). Diagnostics, examination and initiation of habilitation must take place on a continuous basis. Only a modest number of patients are involved each year, and children and their parents should be offered optimal treatment.

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