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Review Article

Aetiology of congenital hearing loss: A cohort review of 569 subjects

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ABSTRACT

Objective: Newborn hearing screening was implemented in Flanders about fifteen years ago. The aim of this study was to determine the aetiology of hearing loss detected by the Flemish screening programme.
Methods: From 1997 to 2011, 569 neonates were referred to our tertiary referral centre after failed neonatal screening with Auditory Brainstem Responses. In case hearing loss (HL) was confirmed, further diagnostic testing was launched. A retrospective chart review was performed analysing the degree of HL, risk factor and aetiology.
Results: Metabolic disorders (0.5%), infectious diseases (35.8%), congenital malformations (6.1%) and genetic abnormalities (19.8%), whether or not syndromic, were retained. In 35% of the subjects no obvious aetiology could be determined in the current study.
Conclusion: In contrast to the literature findings, this series shows a genetic syndromic cause in 80% of the genetic bilateral HL cases. On the other hand connexin positive diagnoses were mostly underrepresented in this study, showing the need for better screening.
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Contents

1. Introduction

The first European Consensus Development Conference on Neonatal Hearing Screening in Milan, 1998 stated that intervention for bilateral permanent hearing loss is considered to be most successful if commenced in the first few months of life [1]. Therefore, identification by screening at or shortly after birth has the potential to improve quality of life. This was confirmed by the Joint Committee in Infant Hearing, recommending a screening of all babies prior to 1 month of age. Diagnostics should be completed before the age of 3 months and for those with confirmed hearing impairment intervention should be initiated before 6 months of age. In many countries newborn hearing screening programmes started throughout the nineties [2]. In general two different methods are used for hearing screening. One is based on oto-acoustic emissions (OAE), the other on AABR. In Flanders the first universal neonatal hearing screening (UNHS) covering a whole region, started in 1997, organised by Kind & Gezin, a Flemish Child Welfare organisation based on AABR, also referred to as the Algo®-test, named after the apparatus used for this purpose [3]. The referral rate of this programme after a second fail at the test was 0.33% for the first 5 years [4], for which further investigation in a reference centre for infant hearing loss is required. The causes of congenital HL are multiple and its evolution is partly determined by its aetiology. As stated before, detection of HL at a very young age is important to provide an early full diagnostic work-up. Language, social and emotional development can only fully develop if a quick detection and treatment of HL are implemented [5,6]. The results of the first five years of hearing screening in

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Flanders showed that 86% of affected children attended regular schools [7]. Data gathered in 2008 by Kind & Gezin, the Flemish Child Welfare organisation, mentioned a yearly target newborn population range of 99.1%. More than half of the cases (67%) were tested before the age of six weeks in a non-medical environment. 94% of the neonates are tested before an age of six weeks [4]. Almost every child in the Neonatal Intensive Care Unit (NICU) is tested before dismissal, however hearing screening may not be the first priority due to serious associated pathology. After referral to a reference centre a complete ENT examination and AABR threshold determination is performed. In case of bilateral affection it is completed by additional exams by ophthalmologists, paediatricians and geneticists. Imaging and biochemic analysis are also performed. The aim of this study is to investigate the cause of newborn hearing loss, detected by UNHS and explored after referral to our tertiary referral centre, with a special interest regarding bilateral hearing impairment.

2. Materials and methods

A comprehensive chart review of 569 children (327 boys, 242 girls), of which 463 were referred to our tertiary referral centre after AABR testing by Kind & Gezin and 106 were picked up by ABR screening in the NICU of our hospital, was retrospectively performed regarding the aetiology of the HL. All these children were referred between 1997 and 2011. Hearing loss was attributed to several etiological categories: HL of genetic (syndromal, non-syndromal) origin, non-syndromal malformation, cytomegalovirus (CMV), serous otitis media, HL of unknown aetiology and a residual category with other rare causes of HL. After diagnostics one other group showed normal hearing. Data acquisition and statistical analysis were performed using Microsoft Excel (Microsoft Corporation, Redmond, WA, USA). Upon referral, the parents were questioned by the ENT specialist regarding the psychomotoric development of the child, familial disorders and a number of risk factors, according to the Joint Committee on Infant Hearing (JCIH) recommendations [2]. A clinical ENT examination checked for deformities of the outer ear with particular attention for anomalies related to the first branchial arches and a micro-otoscopy was performed. All children were examined for potential phenotypic characteristics of syndromes. Audiological evaluation consisted of different investigations: click evoked auditory brainstem responses (ABR), oto-acoustic emissions (OAE) and auditory steady state responses (ASSR). Also a 1000-Hz tympanometry was performed to investigate the middle ear function. After this extensive first evaluation in which a possible hearing loss was confirmed, further diagnostics were launched. Special efforts were made in good guidance of the parents after confirmation of HL. Fig. 1 provides an overview of the diagnostic work-out depending of the clinical situation, anamnesis and extent of HL (bilateral versus unilateral). In unilateral hearing loss cases further diagnostics were sometimes conducted based on single subject specific reasons. For example in case of unilateral middle ear atresia CT scan was conducted at a later stage.

The degree of hearing loss was calculated using pure-tone thresholds at 0.5, 1, 2 and 4 kHz (PTA0.5-4) of behavioural audiograms. If none available, results from the ABR evaluation were used. The degree of HL was defined as: mild HL (30–50 dB HL), moderate (50–70 dB HL), severe (70–90 dB HL) and profound (>90 dB HL) [8]. In our centre syndromal hearing loss is considered as an HL combined with one or more of these characteristics: mental retardation, a major somatic malformation (microcephaly, microtia cardial abnormality, renal anomaly, limb deformity) or a dysmorphia (defined as the presence of at least three minor

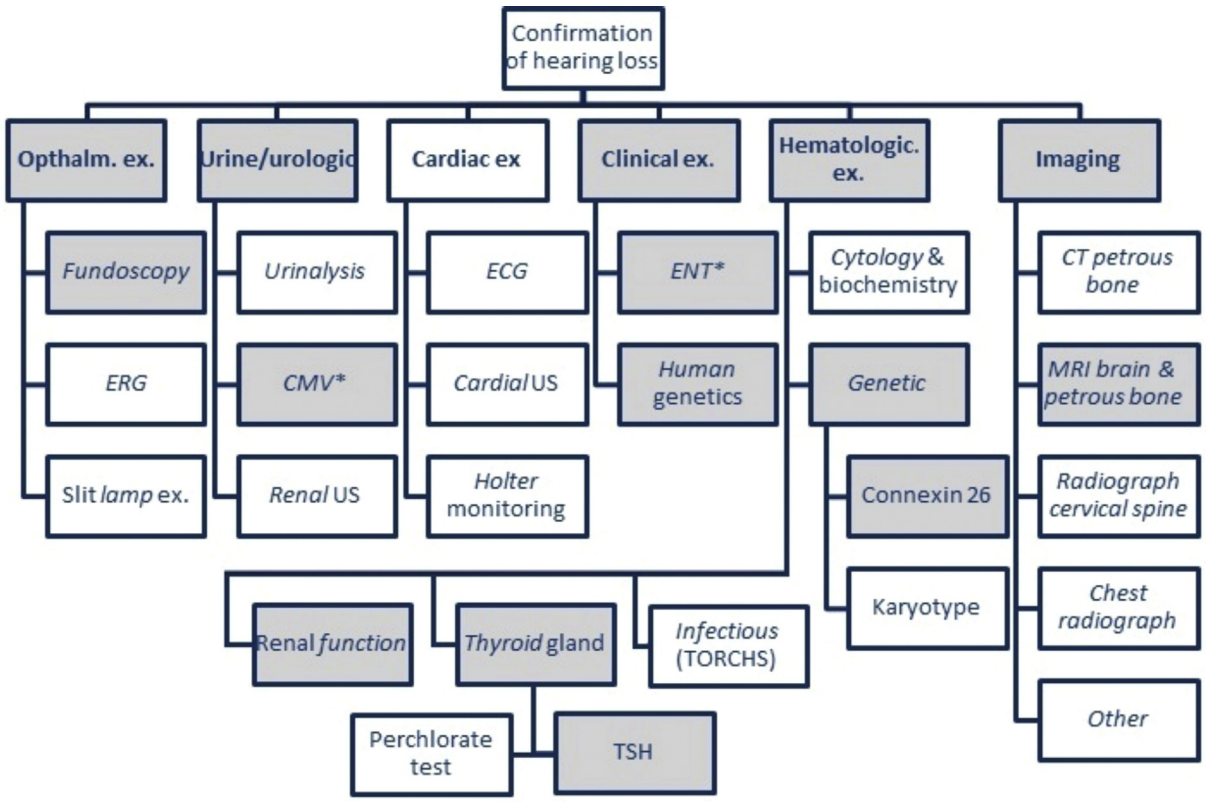

Fig. 1. Overview of tests initiated in bilateral HL. Grey shaded tests are the standard evaluation. Others are optional. * CMV urinalysis and clinical ENT examination are the only standard tests in unilateral HL. Opthalm, ophthalmological; ex., examination; ERG, electroretinogram; US, ultrasound; ECG, electrocardiogram; TSH, thyroid stimulating hormone, CT, computer tomography; cerv., cervical.

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