JOURNAL OF THE ROYAL SOCIETY OF NEW ZEALAND
2025, VOL. 55, NO. 3, 547–573
https://doi.org/10.1080/03036758.2024.2399340

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REVIEW ARTICLE

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A treatment-focused approach to medical investigations for hearing loss in infants

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ABSTRACT

This integrative review synthesises two related but distinct fields: **Early Hearing Detection and Intervention (EHDI)** and **Medical Investigation of Childhood Hearing Loss**. Both fields involve medical evaluations for permanent hearing loss, yet they differ in their perspectives, timing, and target populations. By integrating these bodies of literature, this review introduces a novel, treatment-focused approach to the medical investigation of permanent prelingual hearing loss. Given that the first two years of life represent a critical period for neural plasticity and brain development, this approach holds significant potential for improving patient outcomes. Additionally, it offers opportunities to enhance clinical efficiency and advance equity in the management of childhood hearing loss.

ARTICLE HISTORY
Received 30 November 2023
Accepted 27 August 2024

HANDLING EDITOR
Grant Searchfield

KEYWORDS
Early hearing detection and intervention (EHDI); equity; medical investigations; aetiology; prelingual hearing; natural sleep MRI; congenital cytomegalovirus; genetic

Introduction

Hearing loss in infancy has a profound effect on many aspects of human development. It is a health condition that is insidious, and if left untreated has irreversible effects (Ching et al. 2006). It has been almost 30 years since the formal recommendation that hearing should be tested in newborn infants in a universal manner (Joint Committee on Infant Hearing 1995). We now observe widespread adoption of newborn hearing screening programmes internationally (Busse et al. 2021). Early Hearing Detection and Intervention (EHDI) has been in place for over 10 years in New Zealand where there is a united approach to infant screening and diagnosis of hearing loss (Ministry of Health 2016b). However, the medical workup for these infants is highly variable between centres.

Prelingual hearing loss refers to hearing loss that is present in early infancy (Chari and Chan 2017). The term encompasses both congenital hearing loss and hearing loss acquired soon after birth (Chari and Chan 2017), both of which require high-quality care during the first two years of life to minimise long-term disability (Yoshinaga-

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Itano 2014). These types of hearing loss are detected by newborn hearing screening programmes. In contrast postlingual hearing loss, hearing loss which begins after the first two years of life, has much less impact on life course (Yoshinaga-Itano et al. 1998; Morton et al. 2022), due to the neural foundations for language developed in this time (Moore and Linthicum 2007). Hearing loss which starts after the newborn period but before two years of age has varying impacts on life course, and EHDI prorgammes attempt to detect this through targeted surveillance based on risk factors (Ministry of Health 2016b).

The NZ health system includes 20 centres (previously District Health Boards). Despite recent integration into a national health system, these centres continue to operate in a predominantly independent manner. Most of the EHDI programme in NZ sits within this government-funded public health system. The exception to this is the two cochlear implant centres which receive additional support from charitable organisations and through fundraising efforts. Observed in NZ is a culture of limiting expense where possible, which is in line with the Choosing Wisely campaign (Te Tāhū Hauora Health Quality & Safety Commission 2023). Although medical workup for hearing loss in a newborn is not specified in the paediatric otolaryngology subsection of Choosing Wisely (McDonough et al. 2021), there is a minimal investment in seeking the aetiology of hearing loss for newborn infants who refer to screening in most districts. Typically a 'wait and see' approach is adopted. There are observable geographical differences in the investment of resources into infant hearing loss across NZ; potential influencing factors for this include differing levels of rurality, the proportion of the Indigenous population and ethnic minorities, size, staffing stability, and staff experience.

The critical treatment phase of an EHDI is the first two years of life, as this is the critical window of neuroplasticity in the developing brain (Yoshinaga-Itano et al. 1998). For families who choose spoken language, hearing aids and/or cochlear implants are worn throughout life. Regardless of whether a family chooses spoken or sign language, however, it is essential to begin building the neural pathways for language within the first two years of life to minimise long-term disability (Yoshinaga-Itano et al. 1998; Moore and Linthicum 2007). The quality and breadth of the intervention during the treatment phase have cascading impacts, eventually affecting social, cultural, educational and vocational aspects of life (Yoshinaga-Itano 2014).

The development of language is a necessary precursor to executive function measured at pre-school, and well-developed executive function is associated with adulthood success (Moffitt et al. 2011). The treatment phase of EHDI not only impacts the life course but also impacts intergenerational transfers of advantage (Morton et al. 2022). As the consequences of treatment for hearing loss during the first two years span a lifetime, this review proposes that NZ take a treatment-focused approach to medical investigations, which acknowledges that early life is the opportune time to invest healthcare resources into the condition of prelingual hearing loss.

Further support for a thorough medical work up at the start of the treatment phase comes from evidence that early-life brain development is influenced by multiple factors, which interact in a cumulative manner (Wallander et al. 2021). If two or more influencing factors are negative, a measurable impact is observed in preschool-age assessment of executive function (Wallander et al. 2021). Hearing loss is a challenge for a developing brain, as are social factors such as maternal mental health, household financial

stress (Wallander et al. 2021), or residential mobility (Jelleyman and Spencer 2008). A late diagnosis of a multi-organ, or syndromic condition, adds complexity and subsequent strain to the situation. The NZ health system does not have a direct influence on social factors; however, it does have direct control over the medical work up at the start of the treatment phase. Through thorough medical work up, and appropriate treatment, the strain on early-life brain development is reduced, and children with other challenges such as those living in hardship, are one step closer to well-developed executive function, and on the trajectory towards adulthood success.

Similar to many areas of healthcare, the current EHDI programme shows inequities for NZ Māori. For example, the benchmark of achieving the diagnostic assessment before three months of age was achieved for 69% of children with ethnicity identified as Other (including NZ European), as compared to 44% of Māori infants (Ministry of Health 2016a, 2023). To not further exacerbate ethnic inequities, it is necessary to acknowledge the importance of world view, culture, and language in treatment models. If these factors are ignored when designing systems, barriers to access are created. The importance of worldview and system design has been recently acknowledged, with legal implications (The Waitangi Tribunal 2023).

To date, two published qualitative studies provide insight into the views of Māori regarding EHDI programmes (Crisp 2010; Smiler 2014) as well as one unpublished study (Dargaville 2002). These studies are concordant with research on other health topics and explain that the Māori world view has heightened the importance of relationships (Ware et al. 2018) and social collectivism (Hickey and Wilson 2017). A major learning from this literature is that engagement with Māori infants can be improved when connections are made and maintained with Māori communities (hapu and iwi) who are well connected with families (whānau).

Although the cultural world view is important, other factors such as the long-lasting negative effects of colonisation such as unfair land acquisition, the criminalising of traditional healers (Barrett et al. 2022), and the numerous forms of racism (Adcock et al. 2023) cannot be ignored. There is an intersectionality of numerous forms of disadvantage for many Māori (Simon-Kumar et al. 2022). Acknowledging the added barriers for Māori through social disadvantage, the practical delivery of medical investigations for hearing loss in newborns needs to be considered carefully. Creating systems that keep hospital-based appointments to a minimum and involve community-based indigenous-led service providers wherever possible is paramount to facilitating engagement, and not exacerbating inequities for Māori.

Evaluation of outcomes is an important part of the feedback loop of health programmes (McGill et al. 2021), particularly in resource-limited settings, and for programmes that span numerous departments and sectors. Measurement of outcomes facilitates awareness of equity, adding further importance for countries where equity of outcomes is legally mandated (New Zealand History 1840). In NZ, the standard of practice guidelines includes the '1-3-6' timeliness goals outlined in the Universal Newborn Hearing Screening and Early Intervention Programme (UNSEIP) National Policy and Quality Standards (Ministry of Health 2016b). These guidelines recommend screening infants before one month of age, providing the diagnostic assessment before three months of age, and providing intervention before six months of age (Ministry of Health 2016b). What is lacking, however, is a measurement of outcomes, which is

noted to be a similar gap for many American programmes (Yoshinaga-Itano 2014). Language development is the primary aim of EHDI programmes, however, considerable effort would be needed to measure language outcomes in a culturally and linguistically appropriate manner for the multiethnic population of NZ (Reese et al. 2015). Given the interplay of language with executive function, broader developmental measures would also be sensible (Stika et al. 2015). Later childhood milestones may be easier to measure and would also be informative, such as rates of high school completion or absence of involvement in the justice system.

This integrative review (Cronin and George 2023) combines two fields of literature. The first is the traditional medical specialist-led diagnostic work up for infants presenting with hearing loss, for which some guidelines do exist (Brown and Smith 2013; British Association of Audiovestibular Physicians 2015b, 2015a, 2016; Liming et al. 2016; British Association of Audiovestibular Physicians 2018a, 2018b; Sung et al. 2019; Sung 2022). The second field of literature is Comprehensive EHDI Programmes, which advocates for medical investigations to occur within the EHDI programme, an emerging idea in the literature (Shearer et al. 2019; Thorpe and Smith 2020). The first field of literature is based on a traditional model that offers the benefit of the medical specialist having a high level of knowledge. Alternatively, the second body of work focuses on the integration of medical investigations into an EHDI programme, wherein opportunities to act earlier and through an integrated team approach exist. Comprehensive EHDI programmes also allow an opportunity to address different population sizes, including all newborns (universal approach), the 2% who refer on the hearing screen (hearing-targeted approach) or specifically the 0.2% who are identified with permanent hearing loss (The Joint Committee on Infant Hearing 2019).

Integrating these two relatively siloed fields of literature has the potential to enhance the medical management of infants with hearing loss. The public health system in NZ faces several challenges, including recovery from the COVID-19 pandemic. Medical specialists are typically hospital-based, which can limit accessibility and result in long wait times. Additionally, the commonly used 'wait and see' approach in NZ may be influenced by the timing of interactions between medical specialists with the diagnosed newborn. A model that involves the wider EHDI workforce allows early time points to be considered, with the potential for improvements in accessibility, geographical consistency and equity.

The treatment-focused approach to medical investigations for prelingual hearing loss encourages consideration of task shifting. Task shifting involves handing on tasks from one workforce to another (Expert Panel on Effective Ways of Investing in Health 2019). It is often used to shift tasks from professionals who have long training times and are consequently expensive in healthcare to those with shorter training requirements who may be more abundant (Pokorny et al. 2020). Task shifting promotes working as an integrated, patient-focused team with the division of labour between professional groups based on current evidence rather than traditional roles (Expert Panel on Effective Ways of Investing in Health 2019). In addition to creating efficiency and supporting adaptability, other potential benefits include combining medical specialist knowledge with indigenous-led delivery (Reidy et al. 2023), resulting in improved access and cultural safety.

This literature review aims to reduce the variation and geographical inequities that exist in the medical work-up for infants with hearing loss. A secondary but important

aim is to identify approaches that avoid exacerbation of the ethnic inequities that exist in NZ, with the aim of improving equity for children with prelingual hearing loss.

Method

Search terms

The search terms used are shown in Table 1. Boolean terms and search domains were adjusted as needed between the two databases: Scopus (Title for groups 1, 2 and 4, and Title/Abstract/Keywords for group 3) and Web of Science (Title for all four groups).

The search was limited to between January 2014 and May 2024. It was expected that sentinel papers older than 10 years would be cited often in the literature gathered by the search, and could therefore be pearled. The imposed 10-year-limit resulted in a unique collation of the literature and revealed that there is not yet consistent use of terminology for medical investigations focused on optimising care over the first two years of life. For example, some important studies that focused on medical investigations in the newborn period still used the term 'childhood' or 'paediatric', meaning these needed to be included in the search terms, and the resulting literature gathered was large, and included studies that included older paediatric ages. The search criteria were not limited to English papers only. The differing societal structure of China lead to unique insights into newborn genetic screening. The refinement of studies is detailed in Figure 1 and resulted in 239 studies being included.

Critical appraisal

Integrative reviews collate studies from empirical and theoretic primary sources and a wide variety of methodologies (Hopia et al. 2016). It is therefore seen as too complex to use many formal critical appraisal tools for the many different study designs (Whittemore and Knafl 2005). For integrative reviews which include studies with wide-ranging methodology, Whittemore and Knafl (2005) recommend using a formal appraisal tool only for studies that are outliers. Outliers were those studies that show a result that is contradictory to most studies. None of the papers included in this review showed contradictory or controversial results.

Table 1. Search terms used to gather literature.

1	neonat* OR infant OR child* OR newborn OR 'new born' OR congenital OR paediatric OR pediatric
	AND
2	(hearing W/5 (loss OR impairment OR deficit)) OR 'Hard of Hearing' OR deaf* OR (hearing W/5 screening) OR 'early hearing detection and intervention' OR ehdi
	AND
3	'aetiolog*' OR 'etiolog*' OR (caus* W/5 hearing) OR 'medical work up' OR 'clinical management' OR 'investigation*' OR gene* OR genom* OR 'cCMV' cytomegalovirus OR 'medical work-up' OR ophthalm* OR ecg OR echocardiogram OR (otologic W/5 treatment) OR 'guideline*'
	AND NOT
4	'Otoacoustic emission*' OR ('late-onset' W/3 hearing) OR implant* OR (absence W/3 'hearing loss') OR eczema OR adult* OR acquired OR model OR consanguin* OR sudden OR 'delayed referral' OR 'Valganciclovir' OR 'case report' OR model* OR literacy OR diabetes OR meningit* OR postmeningit* OR attitudes OR women OR perceptions OR 'brain stem' OR questionnaire* OR brainstem OR 'Cat' OR 'Autism' OR 'Mouse' OR 'Mice' OR 'Dog' OR 'Cats' OR 'Dogs' OR 'Ex vivo' OR school OR literacy OR vocabulary OR cancer OR cisplatin OR 'equalization tube' OR patient OR family

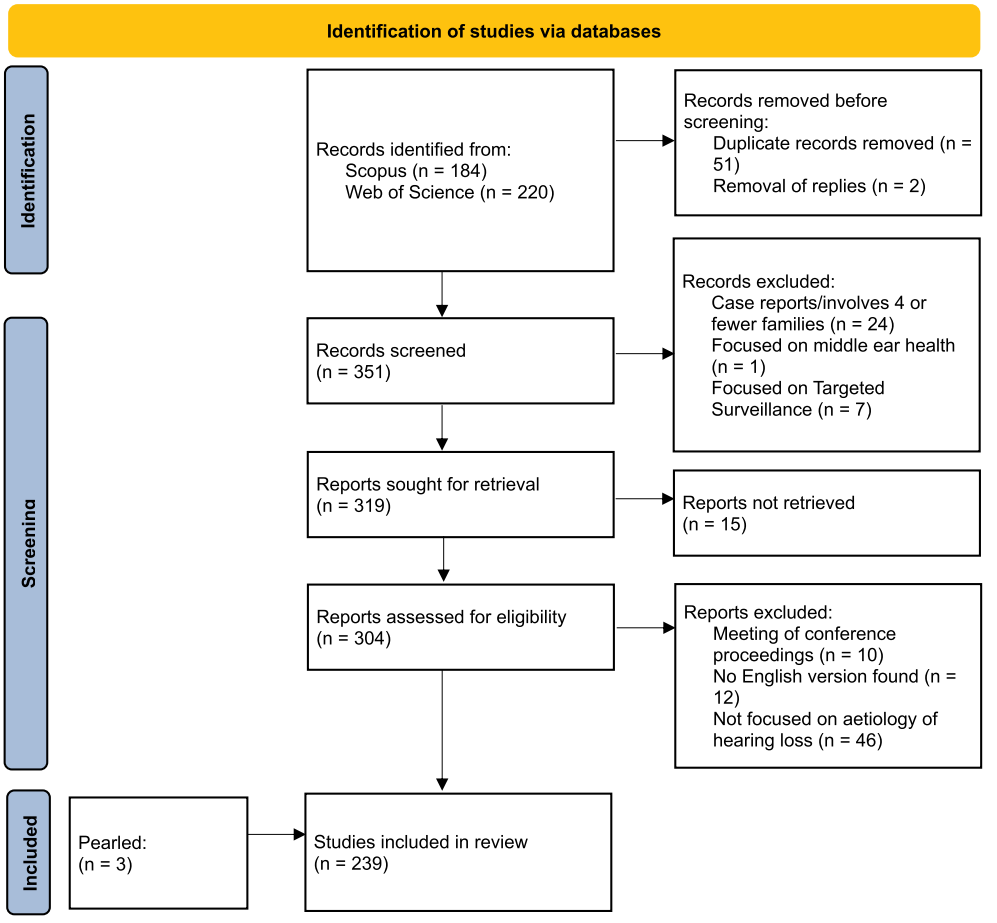

Figure 1. Flow chart of articles included.

Through integration of the gathered literature, the results detail medical investigations that alter case management over the first two years of life. Their potential for integration into an EHDI programme is described, as are any necessary considerations to improve or avoid exacerbation of inequities.

Results

Congenital cytomegalovirus

Early medical investigation is essential to identify the most common cause of non-genetic sensorineural hearing loss, cCMV, which accounts for 15–20% of permanent childhood hearing loss (Morton 2006; Grosse et al. 2008). Screening results are most accurate when completed in the first three weeks of life, as the virus is rampant in the general population and may be present in mother's milk. A positive titre from saliva or urine could be the result of secondary exposure if testing is delayed. Knowledge of cCMV status impacts the treatment phase, as 50% of infants with prelingual sensorineural hearing loss who are

positive for cCMV will experience worsening hearing thresholds (Korver et al. 2017; Dedhia et al. 2018; Shearer et al. 2019; Satterfield-Nash et al. 2020; Thorpe and Smith 2020; Ahmed Hameed Hasan et al. 2021; Jin et al. 2022; Johansson et al. 2022; Lee et al. 2023; Silva et al. 2023). The heightened risk of hearing loss progression associated with cCMV alters case management over the first two years, as it includes more frequent audiological monitoring, and more social support for families who are struggling. An additional justification for being aggressive about establishing cCMV status in a newborn is that cCMV-positive infants often have other neurological sequelae that require child development team involvement in early life (Akrich et al. 2023).

There is evidence from both traditional medical specialist-led approaches to medical work-up (Sung et al. 2019; Gustafson and Corbin 2021) as well as from comprehensive EHDI programmes (Wentland et al. 2018; Shearer et al. 2019) for a much-needed shift in the approach to cCMV testing away from an individual focus (determined by the medical specialist, post-hearing loss diagnosis) to a population approach with testing performed before meeting the medical specialist. The population-wide focus would allow cCMV status to be known for all cases of prelingual hearing loss identified through EHDI and the test would be performed at a younger age, improving the test's specificity for cCMV, as distinct from the acquired CMV infection which has little to no impact on the treatment phase.

Testing for cCMV can be performed as a universal (Ronchi et al. 2017) or a hearing-targeted approach (Harrison 2015; Demmler-Harrison 2016; Dedhia et al. 2018; Vancor et al. 2019; Thorpe and Smith 2020). When the EHDI 1-3-6 goals are met, cCMV testing within the first 21 days of life is a viable option for the hearing screening workforce (Thorpe and Smith 2020; Fourgeaud et al. 2022). A hearing-targeted approach involves doing a cCMV test for all infants who refer from the hearing screen, which is 2% of live births (Ministry of Health 2016b). The lower test numbers and borrowing the workforce and systems from the existing EHDI programme contribute to the feasibility of this hearing-targeted approach to cCMV testing (Shearer et al. 2019; Webb et al. 2022).

A targeted approach to obtaining an accurate indication of cCMV for the 0.2% with confirmed prelingual hearing loss would yield a hit rate of 0.04% of live births that are both cCMV positive and have prelingual hearing loss (Grosse et al. 2008). The cost of this testing regime may be justified firstly by the significant impact that cCMV status has on the treatment phase (increased risk of progression and other neurological sequelae) and secondly due to the cCMV testing regime being within the control of the health system. While many social factors can influence an infant's early-life brain development and are beyond the control of health systems, the decision to test for biological factors such as cCMV and subsequently tailor treatment approaches is clearly within the purview of healthcare systems. It is estimated that cCMV prevalence is higher in Māori infants (Jeong 2023), and this is evident from ethnic differences measured in pre-school (O'Brien et al. 2009), and the over-representation of Māori in populations experiencing deprivation (Fowler et al. 2022). Acknowledging this, and the cumulative interactions observed between factors that negatively influence brain development (Wallander et al. 2021), testing for cCMV would be expected to positively contribute towards equity of outcomes for the EHDI programme.

Although there are a number of different forms of early-life cCMV testing occurring, there is an overall international trend to adopt early-life cCMV testing (Gantt 2023), as

well as progressing from a targeted to a universal approach to testing. Testing for cCMV can occur after 21 days, however, as the likelihood of having acquired cCMV increases a two-step test method may be required. The British protocol recommends polymerase chain reaction (PCR) testing of urine or saliva up to 1 year of age for those diagnosed with sensorineural hearing loss (British Association of Audiovestibular Physicians 2016, 2015b, 2015a, 2018b). If negative, the result can be trusted, however, if positive the newborn Dried Blood Spot (DBS) needs to be checked to distinguish acquired from congenital cCMV (British Association of Audiovestibular Physicians 2016). If the DBS is positive the result can be trusted, as the test has good specificity (Elise et al. 2021). If the DBS is negative, however, uncertainty remains, as many current DBS laboratory techniques can give false negative results (Choi et al. 2009). Although not ideal, the population for whom there is uncertainty is small, and this two-step method can be completed with little or no change in systems in most NZ hospital laboratories.

Currently, there is no protocol or common practice related to cCMV testing in NZs. Therefore, an alternative first step could be considered: task shifting from the medical specialist to the audiologist. A urine or saliva sample could be collected at the time of the hearing loss diagnosis, allowing lab testing and results to be available to the medical specialist before they see the infant. Another method, proposed by Lee et al., showed clinically useful results from combining cCMV culture which has high specificity (98.7%) with PCR testing which has high sensitivity (100%) (Lee et al. 2023). Appendix presents a graphical representation of some of the different options available for integrating cCMV testing with the EHDI programme.

Genetics

Genetic testing for infants with hearing loss can be completed for many purposes. The purpose of genetic testing and its impact on the treatment phase need to be considered. When the focus of genetic testing is to identify the cause of the hearing loss and test order prioritises diagnostic yield, it may not be treatment focused. For example, when an infant presents with bilateral symmetrical sensorineural hearing loss (SNHL) and a family history of similar hearing loss (factors associated with higher diagnostic yield) genetic testing is unlikely to alter case management. This is particularly true if the older generations show a pattern of hearing stability and an absence of a syndrome (common with GJB2 mutations). Genetic testing in this scenario is not treatment-focused. In contrast, when an infant has no family history of hearing loss and no obvious aetiology for their diagnosed hearing loss, genetic testing to check for rare syndromes becomes important as it will alter case management, despite these genetic mutations being rare and therefore having low diagnostic yield. The identification of rare syndromes that are not clinically obvious in the newborn period leads to earlier intervention and support from child development professionals. Rare syndromes that can occur with prelingual hearing loss that are not clinically obvious in the newborn period are termed non-syndromic mimics (Sloan-Heggen et al. 2016). Improvements in genetic testing speed and efficiency make testing for numerous rare syndromes possible.

Genetic testing has rapidly progressed in recent years, from single/direct gene tests to rapid testing methods termed massively parallel sequencing (MPS) or next-generation sequencing (NGS). NGS includes testing a prescribed panel of genes (or micro-array)

or whole exome or whole genome sequencing (Al-Ani 2023). A Deafness panel or a whole exome test analyses 0.014% and 2% of the genome, respectively, making these tests more efficient than whole genome (100%) testing methods (Parker and Bitner-Glindzicz 2015). Different combinations of genetic test methods are used in different labs, influenced by the technology available, and the need for efficiency. It is noteworthy that copy number variants make up a significant portion of genetic hearing loss (Shearer et al. 2014; Sloan-Heggen et al. 2016); some test techniques, such as karyotyping, are not able to identify these.

Many of the studies included in this review focus on a small number (1-4) of the most prevalent genes accounting for hearing loss (Fang et al. 2015; Häkli et al. 2015; Vona et al. 2015; Xiong et al. 2016; Plevova et al. 2017; Yu et al. 2018; Torre-Gonzalez et al. 2022; Hoang et al. 2023). They show that gene testing can be achieved on a large scale for children with hearing loss, however, current approaches lack the ability to check for rare non-syndromic mimics, thus reducing the test's benefit for the treatment phase. Mutations in the GJB2 gene are the most common form of genetic hearing loss in most populations, however, identification of GJB2 mutations as the cause of a diagnosed hearing loss has minimal impact on the treatment phase. Confirmation of pathogenic GJB2 mutations does lower the likelihood of a more significant hearing loss aetiology and, as with all genetically inherited conditions, knowledge of the inheritance pattern may influence reproductive choices for some families.

The orientation to genetic testing described in other studies differs in that it acknowledges the benefit of identifying non-syndromic mimics early (Sloan-Heggen et al. 2016; Boudewyns et al. 2023). This includes Jervell and Lange-Nielsen, Ushers syndrome, and Turners syndrome. Knowledge of these syndromes, and hence the other organs or systems involved, allows early medical care for these conditions. An important consideration is the morbidity/mortality associated with the syndrome, for example, knowledge of Jervell and Lange-Nielsen, which involves long QT syndrome and is associated with sudden infant death, can trigger medical intervention that can be life-supporting (Pabba and Chakraborty 2023).

Genetic testing does not need to be performed within the first few weeks of the infant's life and therefore can be left until the EHDI programme narrows the population to the 0.2% of infants with permanent hearing loss. There is one well-defined exception to this, however. A universal approach to testing for the genetic mutation in the 12S rRNA gene, which underlies sensitivity to ototoxicity from aminoglycoside treatment, would be beneficial (Howell 2019; Shearer et al. 2019; Thorpe and Smith 2020; Nunez-Batalla et al. 2021). Through screening, the use of aminoglycosides for infants who have the mutation can be avoided, if possible, thus preventing hearing loss from occurring. A second reason for universal testing is to detect genetic mutations that are associated with hearing loss in infants who pass the physiological test of hearing. These genetic conditions may cause late onset or mild hearing loss. Further research is needed to quantify the benefits of monitoring infants who pass the physiological newborn hearing screen but are identified as having a genetic mutation for hearing loss (Howell 2019; Shearer et al. 2019; Thorpe and Smith 2020; Gustafson and Corbin 2021; Nunez-Batalla et al. 2021). Universal approaches to genetic testing are long-standing in some countries (Lu et al. 2018; Dai et al. 2019), however, the adoption of a universal approach is currently unlikely in NZ due to the government-funded health system and the '*Choosing Wisely*' culture. What

is more plausible for NZ is to adopt genetic testing post-diagnosis of hearing loss, involving 0.2% of infants.

Massively parallel sequencing, a form of NGS, involves assessing a prearranged 'panel' of genes, which allows faster laboratory testing techniques. There would be one panel only for the condition of prelingual hearing loss, taking away the need for complex decision-making by the medical specialist (Sloan-Heggen et al. 2016). With the simplicity associated with a prelingual hearing loss panel, pre-test counselling would involve routine considerations of risks. This approach would benefit from task shifting to improve accessibility and cultural safety.

Some studies mention the use of broad genetic testing approaches, but link the use of these to the presence of syndromic features (Sung et al. 2019). This method fails to recognise the importance of syndromes which commonly present as non-syndromic mimics in early infancy, a key time for treatment-focused medical investigations (Sloan-Heggen et al. 2016). Other studies choose a stepwise approach, testing for common single gene mutations, due to not having access to a broad genetic test option (Liming et al. 2016; Liddle et al. 2022). An alternative strategy when there is a lack of access to broad genetic testing is to arrange a medical assessment half-way through the treatment phase. Within the population of infants diagnosed with prelingual hearing loss identified through an EHDI programme, we expect a portion to be non-syndromic mimics. If a service does not have access to a broad genetic work-up then a second medical check-up will allow identification of some non-syndromic mimics, for example, those showing early signs of developmental delay or craniofacial abnormalities (Akrich et al. 2023). These early and subtle signs may be more likely diagnosed by a medical specialist than a primary care provider (general practitioner or Plunket nurse). If task shifting from the medical specialist was to occur, it may be more effective to involve and upskill professionals who have a rapport with the family, e.g. the hearing habilitation audiologist, or those familiar with early signs of developmental delays, e.g. a neurodevelopmental therapist. Whether or not an EHDI programme has a broad approach to genetic testing or uses a second medical check-up approach, it is important that there is uniform and agreed upon access across the whole programme. Without this, there are likely to be geographical and cultural inequities.

Difficulties arise in the field of genetics regarding variations of unknown significance (VUS), and what to do with this information. The number of genes identified as causes of hearing loss is increasing, and many have not yet been identified (Van Camp 2021), which leads to some literature recommending repeat lab assessments in future years. Choosing to do a second test for genetic mutations later in life has implications for patient flow through a hospital system, the clinical workload of clinicians, and consideration of the practical utility of knowledge regarding genetic mutations as individuals move out of the treatment phase, and families move out of their reproduction phase and hence the risk of the gene mutation occurring for future infants becomes irrelevant. These implications are not addressed by this review but warrant further research and discussion.

A common feature of the gathered literature is that there is a need for a multidisciplinary team which includes a genetic counsellor (Sloan-Heggen et al. 2016; Korver et al. 2017; Lu et al. 2018; Dieleman et al. 2019; Judge et al. 2019; Sung et al. 2019; Jiang et al. 2020; Niu et al. 2020; Satterfield-Nash et al. 2020; Thorpe and Smith 2020;

Gustafson and Corbin 2021; Nunez-Batalla et al. 2021; Torre-Gonzalez et al. 2022; Boudewyns et al. 2023; Silva et al. 2023). There is Comprehensive EHDI literature that reminds us that, when approaches for a whole EHDI programme are being considered, the impacts on the workforce also need to be considered (Shearer et al. 2019). This is noted in the latest position statement on infant hearing (The Joint Committee on Infant Hearing 2019).

There are societal benefits for multi-ethnic populations of moving from a step-wise approach to a broad approach to genetic testing and concurrently altering the focus of genetic testing from aiming to determine the cause of the hearing loss to instead aiming to identify conditions that influence the treatment phase. The step-wise approach starts with a genetic mutation which has the highest prevalence in the population and, therefore highest diagnostic yield. This will likely be skewed by the majority ethnicity and is likely to place minority ethnicities, which include indigenous peoples in most colonised nations, at a disadvantage. The prevalence of different frequencies of mutations varies across ethnic groups (Chan et al. 2011; Torre-Gonzalez et al. 2022; Ma et al. 2023), and therefore a single gene approach will be more beneficial to some ethnicities than others. It is noteworthy that, of the literature included in this review, the studies that focus on a few genes and a population-wide approach are based in the predominantly monoculture societies of Taiwan and China (Lu et al. 2018; Jin et al. 2022). Torres-Gonzalez et al. explain that GJB2 is the most common mutation for European/Americans and those of European descent (Torre-Gonzalez et al. 2022). Single gene testing which favours the majority ethnicity is not optimal for NZ's multi-ethnic population.

As well as genetic testing being important for the identification of comorbidities including significant medical conditions and having the potential to identify infants who pass screening but are at risk for later hearing loss, genetic testing can also be used to help elucidate the hearing loss diagnosis. Some rare conditions are difficult to diagnose in the presence of middle ear effusion (MEE) which is common in young children. This includes auditory neuropathy spectrum disorder (ANSD), which has a number of genetic causes (Kim et al. 2022; Dedhia and Park 2023). The measurement of otoacoustic emissions is helpful for the diagnosis of ANSD, but MEE often prevents accurate measurement of the emissions. Another example is the diagnosis of permanent hearing loss when there are good bone conduction thresholds and poor air conduction thresholds without microtia (The Joint Committee on Infant Hearing 2019). If this rare condition co-occurs with MEE, the hearing loss appears no different from a temporary hearing loss caused by MEE, and the infant may be discharged (Te Whatu Ora – Health New Zealand 2023). This can leave infants with permanent hearing loss without vital early intervention (typically a bone-conduction hearing aid). ANSD and permanent conductive hearing losses are examples of conditions with genetic causes (e.g. mutations in OTOF and POU3F4). To help in a diagnostic, treatment-oriented manner, genetic testing for POU3F4 would need to occur for all infants with conductive hearing loss at the time of the diagnostic ABR (a subset of 2%). As MEE is more common in Māori and Pacific children (Paterson et al. 2006; Digby et al. 2014), and populations experiencing material hardship (Zhang et al. 2014), a hearing-targeted approach for these mutations would improve equity of the EHDI programme.

Imaging

Magnetic Resonance Imaging (MRI) is considered the imaging modality of choice for prelingual hearing loss (Funamura 2017; Chin et al. 2020). MRI and computerised tomography (CT) can be complementary (Robson et al. 2023), however, MRI is superior at diagnosing some conditions (Lee et al. 2018), does not expose the infant to radiation, and identifies the small subset of infants who would benefit from CT (O'Brien et al. 2021).

MRI features prominently in literature focused on investigations directed by a medical specialist (De Schrijver et al. 2019; Judge et al. 2019; Sung et al. 2019; Niu et al. 2020; O'Brien et al. 2021; Qian et al. 2021; Faistauer et al. 2022; Liddle et al. 2022; Olivier et al. 2022; Boudewyns et al. 2023). MRI does not feature in studies whose focus is the incorporation of medical investigations into an EHDI programme (Shearer et al. 2019; Thorpe and Smith 2020), however, this review suggests this is warranted. The literature focused on the diagnosis of aetiology recommends MRI for both unilateral (British Association of Audiovestibular Physicians 2016; Lee et al. 2018; Vos et al. 2022) and bilateral presentations of SNHL (British Association of Audiovestibular Physicians 2015b, 2015a). Considering our goal of optimising the treatment phase, this review determines that MRI is essential for a subset of hearing losses. Firstly, when the family would like to consider a cochlear implant, for bilateral severe to profound hearing loss, to check for cochlear nerve deficiency (CND) (Liming et al. 2016; Dedhia et al. 2018; Johansson et al. 2022). With a diagnosis of bilateral CND, spoken language may not be an option and families without prior knowledge of sign language need to be supported to adopt early sign language. Secondly, a diagnosis of bilateral ANSD may also be caused by bilateral CND. Thirdly, unilateral ANSD, which may be associated with central nervous system abnormalities, warrants early assessment by a paediatrician (Roche et al. 2010).

Some studies offered MRI based on hearing loss severity being moderate or worse (Liddle et al. 2022). This is to identify congenital abnormalities with the cochlear or vestibular peripheral organs. The condition that has the most impact on the treatment phase is enlarged vestibular aqueduct (EVA), as this condition has a heightened risk of hearing loss progression and may change the way parents approach care for their infant (e.g. reducing the risk of a head knock) (Noordman et al. 2015). As the presentation of hearing loss which could be caused by an EVA is wide ranging, the numbers in this category are higher than the hearing loss presentations associated with CND. Due to this, and the often-strained radiological resource, progression to a national approach to MRI to check for EVA could occur following improvements in referral pathways.

If attempted early (before three months of age) MRI can be performed without sedation, via natural sleep (Sung et al. 2019; Johansson et al. 2023). Avoiding sedation significantly reduces the health resources required for MRI, and there is evidence to support this being safer for the infant's long-term cognitive health (Kilic et al. 2021). Literature included in this review does not clearly identify task separation and instead highlights the importance of the multidisciplinary team working together (De Schrijver et al. 2019; Shearer et al. 2019; Niu et al. 2020; Thorpe and Smith 2020; Gustafson and Corbin 2021; Faistauer et al. 2022), which supports the consideration of audiologist initiation of MRI based on the diagnosed hearing loss.

The current EHDI timeliness goals in NZ are '1-3-6' (months) for screening-diagnosis-treatment. Within a successful programme, the actual age that each infant is seen for

diagnostic testing could vary, ranging between 3 weeks and 3 months of age. Acknowledging that the literature supports a 3-month cut-off age for attempting natural sleep MRI, a programme performing to 1-3-6 goals would allow a natural sleep option for MRI for some, but not all. Based on the trends observed in the measured timeliness goals (Ministry of Health 2016a), it can be hypothesised that in NZ those seen later are those who face barriers to engagement. This group is skewed towards Māori and those experiencing deprivation. This situation is not equitable. A shift to a 1-2-3 strategy, with the aim of completing all diagnoses by 2 months of age, has been recommended (The Joint Committee on Infant Hearing 2019) and would allow a larger proportion of infants to have the option of a natural sleep MRI, potentially improving equity of access to medical investigations.

Ophthalmology evaluation

The motivation for undertaking ophthalmology evaluation is firstly due to the higher prevalence of ocular pathology in children with hearing loss (Gürtler et al. 2017; Wentland et al. 2018) and secondly, due to the compounding impact a deficit in two major primary senses would have on language, socio-emotional, and cognitive development (Chari and Chan 2017; Gruber et al. 2019).

Many studies recommend that ophthalmology evaluation should be completed for infants with hearing loss, but do not suggest an age (Brown and Smith 2013; Carey and Palumbos 2016; Chen and Oghalai 2016; Liu et al. 2019) and others state that it should occur immediately following diagnosis and be repeated at multiple ages throughout childhood (British Association of Audiovestibular Physicians 2015b, 2015a, 2016). Although ocular assessment in the newborn period may be limited, some studies do specify referral for ophthalmology evaluation at age 3–4 months (Kilic et al. 2021; Acke et al. 2022); other studies specify later, but still within the treatment phase, e.g. 1 year of age, (British Association of Audiovestibular Physicians 2015b, 2015a, 2016).

While numerous studies have investigated the prevalence of ocular pathology in children with hearing loss across a wide range of ages (Prosser et al. 2015; Köylü et al. 2016; Altiaylik Ozer et al. 2018; Gruber et al. 2019), further research and focused literature reviews are warranted to elucidate the optimal timing for ophthalmologic evaluations in infants with prelingual hearing loss. Usher's syndrome is the most common eye disorder in children with hearing loss and the route to diagnosis will change with progress towards a genetic panel; however, ophthalmic evaluations may also be beneficial to check for other conditions (Gruber et al. 2017; Hawley et al. 2017).

Electrocardiogram

An established correlation exists between prelingual SNHL and congenital heart conditions, a subset of which have genetic aetiologies (Fenrich et al. 2022; Yang et al. 2024). While genetic panels offer an improved means to identify individuals with Jervell and Lange-Nielsen (JNL) syndrome, there are documented cases of prolonged QT intervals without the corresponding genetic mutation for JNL (Ergül et al. 2021; Fenrich et al. 2022). There are also other electrocardiographic abnormalities for which there are unknown implications (Mehta et al. 2016; Suma et al. 2022). Further research is warranted

to determine which cases of permanent newborn hearing loss require an electrocardiogram (ECG), particularly when there is a genetic method for identifying JLN syndrome.

Vestibular assessments

There is an association between hearing and vestibular dysfunction, which is unsurprising given the close anatomical relationship between the peripheral organs and lateral neural pathways involved in the hearing and balance systems (Wang et al. 2021). Wang et al. (2021) argue that as both hearing loss and vestibular dysfunction impact quality of life, their co-occurrence warrants investigation. These authors reference evidence of improvement in motor development delay after intervention from a 2004 study by Rine et al. (Rine et al. 2004). Martens et al. demonstrated that performing vestibular assessments at six months of age for all cases of newborn SNHL can be achieved (Martens et al. 2022), however, a targeted approach is likely more feasible in countries without any current paediatric vestibular testing protocols or workforce. A targeted approach would involve testing groups that have known risk, for example, those with severe or profound hearing loss, hearing loss caused by meningitis, cCMV or cochleovestibular anomalies (Martens et al. 2022), CND (Tsukada and Usami 2021), unilateral hearing loss (Birdane et al. 2016), or those showing signs of motor delay (Janky and Yoshinaga-Itano 2022). Notably, these are all common types of prelingual hearing loss and testing these high-risk groups could be labour-intense.

Using numerous test methods Wang et al. showed that rates of vestibular dysfunction were similar between syndromic and non-syndromic cases of permanent hearing loss. However, failing the Cervical Vestibular Evoked Myogenic Potential (cVEMP) assessment was higher in syndromic cases. They argue that vestibular testing using cVEMP will fail to detect vestibular dysfunction in non-syndromic cases of permanent hearing loss. Using cVEMP only and testing at six months of age Martens et al. found that 13.9% of infants with permanent prelingual hearing loss had vestibular pathology (Martens et al. 2019; Martens et al. 2022). They argue that the intervention for these infants improves motor development and overall quality of life. As there is limited testing of vestibular function in NZ, the workforce needs to be considered to ensure that testing does not increase geographical inequities in service provision. Currently, there is scarce evidence for how and when testing should occur, and the value of testing vestibular function in infants with hearing loss, however, this is likely to be clarified in the near future with highly regarded authors taking an interest in this topic (Janky and Yoshinaga-Itano 2022).

Discussion

Guidelines for investigations following the diagnosis of hearing loss in children are not lacking (Brown and Smith 2013; British Association of Audiovestibular Physicians 2015b, 2015a, 2016; Liming et al. 2016; British Association of Audiovestibular Physicians 2018a, 2018b; Sung et al. 2019; Sung 2022). There are, however, few guidelines on this topic focused on newborn infants, (Liming et al. 2016), and none which are orientated around optimising intervention over the first two years of life (treatment focused).

Through integration of both comprehensive EHDI literature and medical-led investigations of aetiology literature, this review results in the identification of medical

interventions that are treatment focused. The results also identify the potential for improvements in efficiency and equity.

The authors' observation of variation in practice throughout NZ is in line with published literature that adherence to guidelines in this area is generally low (Wilson et al. 2005; Kenna 2021). Some variation in practice is associated with the type of medical specialist reviewing the patient (Wilson et al. 2005; Kenna 2021). Variation is also seen, however, between individuals within the same medical specialty and the same workplace (Wentland et al. 2018).

The solution proposed by both Wilson et al. and Kenna is that medical specialists should do more medical investigations following the diagnosis of hearing loss and should also follow recommended guidelines (Wilson et al. 2005; Kenna 2021). In contrast, this review has highlighted that a treatment-focused approach that involves a multidisciplinary team and task shifting may be a better way forward for achieving equitable access to medical investigations for children with prelingual hearing loss identified through EHDI programmes. Support for this comes from evidence that traditionally published guidelines have not highlighted the importance of equity, and consequently, strict adherence to the medical specialist-led model could exacerbate inequities in service provision (Boyles et al. 2023).

Traditionally, following a diagnosis of hearing loss, a child is seen by a medical specialist who will then consider ordering medical investigations (Rutherford et al. 2011; Wentland et al. 2018). Although medical specialists are highly educated in this area, their ability to make a positive contribution to patient outcomes is reduced by the timing of the flow of patients to them from the EHDI programme. This review suggests that a hearing-targeted or audiologist-initiated approach to medical investigations should be considered as an approach that could improve patient outcomes. This approach allows patients to be protected from negative effects associated with the long wait times that can occur with referral to another service. This is critically important for some investigations (cCMV and MRI). Educating audiologists on which subsets of infants with prelingual hearing loss would benefit from the various medical investigations available closes the gap between the medical specialist knowledge and the workforce making the hearing loss diagnosis and working closely with the family over the first three years of the infant's life.

The collected literature has been in support of early-life ophthalmology evaluation. This contrasts with current NZ guidelines supported by ophthalmologists, as prelingual hearing loss is not listed as a reason for targeted ophthalmology referral (The Paediatric Society of New Zealand and Starship Foundation 2022). The guidelines do include premature infants, those who show global development delay, and those with a family history of vision loss in childhood. It is noteworthy that in NZ, universal ophthalmology screening approaches exist. These include a check for the red reflex after birth and at six weeks of age (Newborn Clinical Network 2022) and a vision screen (primarily for amblyopia) as part of the B4 School Check at 4–5 years of age (Hamm et al. 2019). The latter screen is outside the age focus of this review and shows similar problems with poorer access for Māori and Pacific peoples and for those from communities with higher deprivation (Findlay et al. 2020). The next step forward for clarity regarding optimal ophthalmology evaluation for infants with hearing loss requires the support of NZ-based ophthalmologists. Given co-occurring vision loss is rare but important (Lin et al. 2011), consideration of targeted audiologist-led screening options warrant consideration.

Audiologist-initiated MRI is not a new concept, as this is already used with adult populations internationally (Abbas et al. 2018; Eakin et al. 2022). Currently, in NZ, it is the audiologist who makes the diagnosis of hearing loss, explains the diagnosis to the family, and refers the infant to the cochlear implant programme. MRI could be initiated by the audiologist before the infant is seen by the medical specialist. It is hypothesised that this would decrease the national average age of MRI completion, as well as improve utilisation and success rate of the natural sleep method. Reducing the need for sedation or anaesthesia would be a cost-saving for the health sector.

Using genetic testing to help distinguish temporary from permanent conductive hearing loss is described, however, this challenge in the diagnosis may be unique to NZ. Many American EHDI programmes do not routinely include bone conduction testing (Findlen and Schuller 2020), and audiological assessments may be repeated until MEE has cleared or is treated. This technique as the limitation of contributing towards delays in diagnosis (The Joint Committee on Infant Hearing 2019; Warner-Czyz et al. 2024) however has the strength of allowing for diagnosis of permanent conductive hearing loss due to ossicular fixation or third window effects (Dasgupta et al. 2023; Kempfle and Remenschneider 2024).

Appropriate investigation for complex cases, such as non-syndromic mimics, is highly important for that individual and their family, however, these cases are rare. This can lead to medical support for infants with prelingual hearing loss being de-prioritised in times of resource shortage. Anecdotally, a reduction in this support by medical specialist departments has been seen in the post-COVID time period in the NZ health sector. When there is a low incidence of a condition, these investigations can be seen as low priority and disregarded by medical staff in rural centres, where there are more pressing basic health issues for the population and specialised testing is less available and more expensive to access. Similar explanations are likely to underly the lack of data on investigations of aetiology of hearing loss for low and middle-income countries, including the expense, the reliance on highly trained medical specialists, and the time-consuming and often inconclusive nature of the results of investigations (Jiang et al. 2020).

Agreeing on a base minimal approach (essential medical investigations) and providing access to these across the region that the EHDI programme covers would likely improve geographical equity. Task shifting can help mitigate resource strain as a reason for medical investigations not being completed. Task shifting could also facilitate indigenous-led delivery improving accessibility and equity for indigenous populations.

Limitations

This study excluded literature focused on middle ear health. Although not the focus of this review, the authors acknowledge that poor middle ear health, and its ensuing fluctuating hearing loss can impact auditory neural development in the prelingual period (Altamimi et al. 2023). Poor middle ear health is more prevalent for Māori and Pacific populations (Nash et al. 2023), and as there are no ethnicity-directed provisions within the current NZ EHDI, we encourage novel approaches (Dickinson et al. 2018) and further research in this area.

This review adopts a medical view of prelingual hearing loss as a sensory loss that bears potential for intervention that can enhance or restore functional use of hearing for communication and social, educational and vocational development of an individual.

The focus of the review is on early and accurate diagnosis so that appropriate treatments can be offered to families, including both oral and sign language support. The authors recognise and respect that a non-medical model of deafness which embraces Deaf culture and chooses to use sign (rather than oral) language, is a choice that families of children with hearing differences have the right to make (Lillo-Martin et al. 2023). For these families, early detection remains important to facilitate support for sign language or other forms of communication within the neuroplastic period.

Conclusion

This integrative review describes and promotes change in the way medical investigations for prelingual hearing loss are typically conducted in NZ. Rather than being prioritised for their diagnostic yield and implemented by a medical specialist at one point in time and in a centralised location, priority is given to medical investigations which significantly impact treatment, supporting effective intervention over the first two years of life. The proposed approach is goal-orientated (supporting well-developed language centres in the brain) and equity-focused. This approach considers the influence the child's age has on the utility of a medical investigation and the effects of clinician location and waiting time on equity. We describe how the umbrella of EHDI's multiple non-medical workforces can be utilised and systems and protocols can be adjusted to benefit both patient and family, as well as reducing the cost to the healthcare system. The changes proposed in this article are relevant for other countries pursuing equity of outcomes from their EHDI programme. To facilitate further discussion, research and potential changes in clinical practice the following set of questions are proposed for determining which medical investigations could be brought into an EHDI programme:

(1) Equity – Will the change improve equity?
(2) Treatment-focused – Do the results of the investigation influence intervention over the first two years of life?
(3) Time critical – Are there benefits from it being done when the child is very young?
(4) Efficiency – Does bringing the investigation into the programme improve the overall cost to the health sector?

With the shift from independent districts to a combined NZ public health system, this review provides guidance for intervention framing, essential medical investigations, and proposals for improving the equity and efficiency of NZ's EDHI programme.

Disclosure statement

No potential conflict of interest was reported by the authors.

Funding

This work was supported by Lottery Health Research [Grant Number 9107-3727163]; Lindsell Richards Education Fund [Grant Number 9107-3728325]; Braemar Charitable Trust [Grant Number 9107-3727386].

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Appendix

Interaction between EHDI benchmarks and medical investigations

EHDI timeline and goals: S = Screen, D = Diagnose (dark grey), T = Treatment begins (treatment phase is lighter grey), Med. Review = Review by a medical specialist.

	The first two years of life for infants with permanent hearing loss											
	Age (1-month increments)						Age (3-month increments)					
	1	2	3	4	5	6	9	12	15	18	21	24
	S		D			T						
For cCMV testing to occur with the gold standard test techniques the test needs to occur under 3 weeks of age. The newborn hearing screening workforce are a potential workforce, and this could be universal or hearing-targeted.	cCMV											
For cCMV testing to occur for the fewest infants it would need to occur after the diagnosis of SNHL, a subset of 0.2% of live births. Audiologists could collect urine or saliva, and ask for consent to test the DBS if needed.			cCMV									
If the EHDI timing was adjusted to 1-2-3 benchmarks , it would allow for improved accuracy of cCMV testing as well as cCMV testing for a very narrow portion of the population, those with SNHL (i.e., a subset of 0.2% of live births).		cCMV										
If the EHDI timing was adjusted to 1-2-3 benchmarks, it would allow the MRI to take place prior to 3 months of age (the commonly cited cut-off age for attempting natural-sleep MRI).		MRI										
A medical specialist review at 12 months of age allows improved detection of craniofacial abnormalities, general development delays and social influences on development. Genetic testing for non-syndromic mimics early but post diagnosis, would complement this medical review.								Med. Review				
	S	D	T									

EHDI timeline and goals: S = Screen, D = Diagnose (dark grey), T = Treatment begins (treatment phase is lighter grey), Med. Review = Review by a medical specialist.