Aetiologic diagnosis of hearing loss in children identified through newborn hearing screening testing

Diagnosi eziologica dell’ipoacusia nei bambini identificati attraverso lo screening audiologico neonatale

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SUMMARY

With the implementation of universal newborn hearing screening (UNHS) programmes and early diagnosis and treatment of hearing problems, the need has clearly emerged to implement and carry out a systematic and coordinated protocol for the aetiological diagnosis of permanent hearing impairment (PHI). Within the framework of the Italian Ministry of Health project CCM 2013 “Preventing Communication Disorders: a Regional Program for early Identification, Intervention and Care of Hearing Impaired Children”, it has been decided to consider the problems relative to aetiological diagnosis of child PHI within UNHS programmes. The specific objective was to apply a shared diagnostic protocol that can identify the cause in at least 70% of cases of PHI. For this part of the project, four main recommendations were identified that can be useful for an efficient aetiological diagnosis in children affected by PHI and that can offer valid suggestions to optimise resources and produce positive changes for third-level audiological centres.

KEY WORDS: Aetiology • Hearing loss • Children • Newborn hearing screening • SWOT analysis

RIASSUNTO

Parallelamente alla attuazione dei programmi di screening audiologico neonatale e di diagnosi audiologica e trattamento precoci, si è resa evidente la necessità di mettere a punto e attuare un protocollo per la diagnosi eziologica della sordità, che sia sistematizzato e che si coordini, senza interferire, con il percorso diagnostico audiologico. Nell’ambito del progetto del Ministero della Salute CCM 2013 “Programma regionale di identificazione, intervento e presa in carico precoci per la prevenzione dei disturbi comunicativi nei bambini con deficit uditivo” è stata presa in considerazione la problematica relativa alla diagnosi eziologica della ipoacusia infantile nell’ambito dei programmi di screening audiologico neonatale. L’obiettivo specifico è quello di attuare il protocollo diagnostico per ottenere una definizione della causa della ipoacusia in almeno il 70% dei casi con diagnosi audiologica confermata. Nell’ambito di questa parte del progetto, sono state individuate quattro principali raccomandazioni utili nella ricerca di una diagnosi eziologica nei bambini affetti da ipoacusia, che possono costituire, per i centri audiologici di III livello, dei validi suggerimenti per ottimizzare le risorse e produrre cambiamenti positivi.

PAROLE CHIAVE: Eziologia delle ipoacusiche infantili • Screening udittivo neonatale • Analisi SWOT

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Introduction

With the implementation of UNHS programmes and early diagnosis and treatment of hearing problems 1,2, to implement and carry out a systematic and coordinated protocol for the aetiological diagnosis of permanent hearing impairment (PHI) has clearly emerged. In this respect, progresses in molecular genetics and in the treatment of pre- and perinatal infections have improved both diagnostic and therapeutic possibilities in the fields of congenital and prelingual PHI, so today an accurate and early aetiological diagnosis plays an even more important and sometimes decisive role. Consequently, once the identification of PHI has been made, it is advisable to implement a well-structured programme for aetiological diagnosis based on a multidisciplinary approach 3.

Concerning the treatment and management of children with hearing problems, the early identification of the causes of the disorder offers a number of advantages: it can avoid expensive and unnecessary tests, provide psychological benefits to relatives who are made aware of the causes of the impairment, offer important genetic infor-
mation both for the child and the family, and supply prognostic information, which can help identify risk factors, prevent complications, and allow an early diagnosis of associated problems, with the possibility to prevent the effects of the disorder. The information obtained from aetiological diagnosis can therefore be useful in the management of the child with PHI.

The importance of an aetiological diagnosis is underlined by the recent position statements of the Joint Committee on Infant Hearing of 2007 and 2013, in which it has been recommended that every newborn diagnosed with PHI should start a protocol for diagnostic assessment of the causes of the disorder. However, in many of the newborn hearing screening protocols, the possibility of carrying out an aetiological diagnosis is not taken into account.

Within the framework of the Italian Ministry of Health project CCM 2013 “Preventing Communication Disorders: a Regional Program for early Identification, Intervention and Care of Hearing Impaired Children”, it has been decided to consider the problems relative to the aetiological diagnosis of childhood PHI in UNHS programmes. As a result, one of the objectives was to obtain an early aetiological diagnosis through the implementation of uniform and timely procedures, in particular with regard to common causes (CMV, GJB2/GJB6 mutations, inner ear malformations). The specific aim was to apply a shared diagnostic protocol that can identify the cause in at least 70% of cases of PHI.

**Material and methods**

A review of the international literature was made to collect information, aiming at a strategic analysis of the activities associated with aetiological diagnosis. The achieved data were then analysed, taking into account the current protocols and programmes of aetiological diagnosis for evaluation of the positive and negative aspects of the entire process. This first evaluation allowed us to perform a preliminary SWOT analysis (Strengths, Weaknesses, Opportunities, Threats) on aetiological diagnosis. SWOT analysis was performed by reporting the following: Strengths, S; Weaknesses, W; Opportunities, O; and Threats, T in aetiological diagnosis. In SWOT analysis, Strengths and Weaknesses were referred to the inner aspects of the system under examination, while Risks and Opportunities regarded the external conditions that can affect system performance. The achieved data were examined, discussed and integrated by a team of 21 professionals involved in the prevention, diagnosis and treatment of newborn hearing impairment (audiological physicians, otolaryngologists, audiometrists, speech therapists, paediatricians, psychologists, audoprosthesis and clinical specialists for cochlear implants, families) in third-level centres in which UNHS programmes are available. In order to obtain recommendations and general guidelines from SWOT analysis, a TOWS matrix was created, which allowed combining strengths with opportunities (strategy S-O), strengths with threats (strategy S-T), weaknesses with opportunities (strategy W-O) and weaknesses with threats (strategy W-T). The detailed description of the SWOT and TOWS matrix analysis procedure can be found elsewhere in this issue.

**Results**

This article will discuss the problems of SWOT analysis carried out by the team of expert operators, and the final TOWS analysis constructed on the basis of the results obtained. The working team provided 32 answers for category S, 44 for category W, 30 for category O and 30 for category T, for a total of 136. These data were then grouped according to type and area of reference. The main key points obtained are listed in Table I.

**Strength key points analysis**

Four main topics emerged from SWOT analysis, which represent the strengths featuring the area of aetiological diagnosis: the existence of a protocol for aetiological diagnosis (46.87%), multidisciplinary collaboration (34.37%), facilitated access to audiologic assessment 4 (12.5%), communication to the families of the outcomes of diagnosis 2 (6.25%). These strengths can be analysed in further detail:

**Existence of protocols for aetiological diagnosis used in some third-level audiologic centres**

This category contains all the replies concerning the possibility of structuring a shared protocol for the achievement of an aetiological diagnosis, starting from the protocols used in some structures. The presence of a shared protocol could allow third-level centers to standardise methods and procedures for aetiological diagnosis. The access to aetiological data is extremely important because it can help to better delineate future progress in child development, by addressing the therapeutic choices in a safer and more conscious manner. In order to optimise the procedure of aetiological investigation, a point of strength is represented by the possibility to carry out the diagnostic programme in a single centre in which all involved specialists are present (audiological physician, otolaryngologist, paediatrician, ophthalmologist, child neuropsychiatrist, geneticist, radiologist, etc.). In order to favour participation, prenatal diagnostic protocols could be activated at the Departments of Neonatology to allow precocity of intervention.

**Multidisciplinary collaboration**

This category contains all the replies concerning multidisciplinary collaboration among the professionals involved
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Due to the variety of the investigation’s multidisciplinary collaboration, this represents an important resource for the entire procedure of aetiological research. In particular, paediatric (dismorphological paediatrics) and genetic counselling have proven to be an important factor for early detection of PHI associated with other syndromes.

Facilitated access to audiologic assessment
This category contains all the replies concerning facilitated access to audiologic diagnosis. Facilitated access to audiologic assessment and short waiting times are essential for early identification of impaired hearing and to be able to rapidly start the entire process of aetiological research.

Communication to the families of the outcomes of diagnosis
This category contains all the replies concerning the communication of aetiological diagnosis to families. The identification of PHI aetiology is an important requirement for many parents, and for this the family should be informed about the outcomes of the various aetiological investigations, and on the possible prognosis of their child development.

Weakness key points analysis
Six main topics emerged from SWOT analysis, which represent the Weaknesses featuring the area of aetiological diagnosis: lack of standardisation in diagnostic procedures (27.27%), limited multidisciplinary collaboration (20.45%), difficulties in communication with the family (13.63%), limited knowledge (13.63%), management difficulties (13.63%) and obstacles in data collection (11.36%) (Table IIb). These weaknesses can be analysed in further detail:

Lack of standardisation in procedures
This category contains all the replies concerning the lack of standardisation in the procedures relative to implementation of an aetiological diagnosis, associated with the lack of a universally shared protocol (at least on a national scale). The identified weaknesses are mainly due to the absence of guidelines for the treatment of the patient, both regarding times and modalities, resulting in a “personal” management of aetiological diagnosis. Difficulties reported in the management of diagnostic protocols are even greater in patients with syndromic PHI.

Scarce multidisciplinary collaboration
This category contains all the replies concerning multidisciplinary collaboration among the team members participating in the definition of aetiological diagnosis. Collaboration problems are strictly connected to the difficulties in disseminating and sharing the results of the aetiological investigations submitted to the patient by the staff members (e.g. outcomes of post-natal virologic diagnosis), and to the obstacles in coordinating the different diagnostic activities, especially when the professionals involved are not working in the same structure. Communication with the territorial Service and Paediatric Neuropsychiatry has proved to be particularly difficult.

Difficulty of communication with families
This category contains all the replies concerning the difficulties in communicating with the family of the hearing impaired child. The first difficulty emerges from the fact

Table I. Main key points extrapolated from questionnaires.

Table Ia. Strengths.

<table>
<thead>
<tr>
<th>Strength key points</th>
<th>Frequency (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Existence of a protocol for aetiological diagnosis</td>
<td>15 (46.87%)</td>
</tr>
<tr>
<td>Multidisciplinary collaboration</td>
<td>11 (34.37%)</td>
</tr>
<tr>
<td>Facilitated access to audiologic assessment</td>
<td>4 (12.5%)</td>
</tr>
<tr>
<td>Communication to families of the outcomes of diagnosis</td>
<td>2 (6.25%)</td>
</tr>
</tbody>
</table>

Table Ib. Weaknesses.

<table>
<thead>
<tr>
<th>Weakness key points</th>
<th>Frequency (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lack of standardisation in the procedures</td>
<td>12 (27.27%)</td>
</tr>
<tr>
<td>Scarce multidisciplinary collaboration</td>
<td>9 (20.45%)</td>
</tr>
<tr>
<td>Difficulty of communication with the family</td>
<td>6 (13.63%)</td>
</tr>
<tr>
<td>Limited knowledges</td>
<td>6 (13.63%)</td>
</tr>
<tr>
<td>Management Difficulties</td>
<td>6 (13.63%)</td>
</tr>
<tr>
<td>Scarce information technology for data collection</td>
<td>5 (11.36%)</td>
</tr>
</tbody>
</table>

Table Ic. Opportunities.

<table>
<thead>
<tr>
<th>Opportunity key points</th>
<th>Frequency (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Implementation of shared protocols for aetiological diagnosis</td>
<td>10 (33.3%)</td>
</tr>
<tr>
<td>Shared database for aetiological data collection</td>
<td>9 (30%)</td>
</tr>
<tr>
<td>Multidisciplinary collaboration</td>
<td>9 (30%)</td>
</tr>
<tr>
<td>Other</td>
<td>2 (6.6%)</td>
</tr>
</tbody>
</table>

Table Id. Threats.

<table>
<thead>
<tr>
<th>Threats key points</th>
<th>Frequency (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Resources</td>
<td>9 (30%)</td>
</tr>
<tr>
<td>Scarce standardisation in the approach to aetiological diagnosis</td>
<td>9 (30%)</td>
</tr>
<tr>
<td>Difficulty in communication</td>
<td>7 (23.3%)</td>
</tr>
<tr>
<td>Lack of legal support</td>
<td>3 (10%)</td>
</tr>
<tr>
<td>Other</td>
<td>2 (6.6%)</td>
</tr>
</tbody>
</table>

Table I (a b c d) The table shows the frequency of the topics that emerged in the categories Strength, Weakness, Opportunity and Threat. (n = 231)
that the family is not always given a clear and complete explanation of the entire programme, and therefore parents cannot understand the importance of defining aetiological diagnosis and of the different steps involved in the diagnostic process. Another critical point is represented by transmission of the test results from the third-level laboratory (genetic and other tests) to the child’s family. The communication difficulties increase when the patient’s place of residence is distant from the audiologic centre of reference.

**Limited knowledge**
This category contains all the replies concerning the limited knowledge of some aspects of aetiological diagnosis. The availability of limited epidemiological data is associated with the partial knowledge of a definite aetiological diagnosis. There are also limited data in relation to the sensitivity and specificity of the various tools of aetiological investigation.

**General management difficulties**
This category contains all the replies concerning the general management difficulties concerning the process of aetiological diagnosis. The most relevant point of weakness is represented by the presence of long waiting-times to obtain diagnostic tests and consequently to obtain the necessary clinical data for an aetiological diagnosis. Other critical points concern the absence of dedicated staff and the costs necessary for diagnostic tests.

**Difficulty in collecting data**
This category contains all the replies concerning the collection of aetiological diagnostic data. The lack of uniformity in data collection standards, the impossibility of telematic data sharing and the absence of shared databases make the exchange of information between the problematic for the different specialists involved in the aetiological research to exchange information, determining an incomplete collection of the elements necessary to reach a diagnosis.

**Opportunities key points analysis**
Three main topics representing the Opportunities characterising aetiological diagnosis emerged from SWOT analysis: implementation of shared protocols for etiological diagnosis (33.3%); shared database for the collection of aetiological data (30%); multidisciplinary collaboration (30%). These points of strength can be analysed in further detail:

**Implementation of shared protocols for aetiological diagnosis**
This category contains all the replies concerning the advantages of diagnostic aetiological protocols for the implementation of shared protocols. All third-level centres must be highly specialised, and possess the tools and competences for aetiological assessment in their institutes, or within affiliated structures. The study of CMV represents a major field of investigation for the general protocol of aetiological research. Gynaecologist and parents should be sensitised on this issue so as to promote early aetiological diagnosis, by activating protocols of prenatal screening for the research of CMV during pregnancy.

**Shared database for the collection of aetiological data**
This category contains all the replies concerning the advantages of a database for the collection of aetiological data that could lead to the creation of shared databases. The availability of these databases could promote the exchange of information among the different specialists involved, and could allow the collection of useful epidemiological data, and to establish guidelines for treatment.

**Multidisciplinary collaboration**
This category contains all the replies concerning the advantages of multidisciplinary collaboration. The communication network represents a strong need within the process that will allow audiologists, otolaryngologists, neonatologists and family paediatricians (to mention only some of the specialists) to perform aetiological diagnosis. All professionals of the multidisciplinary team (physicians and paramedics) need to possess some basic knowledge on the main stages of the process: aetiology of PHI, investigation methods of the main causes and prognosis of development for each aetiological factor. The possibility to access training opportunities could be useful for knowledge standardisation.

**Threats key points analysis**
From the SWOT analysis four main topics emerged, which represent the risks characterising the area of aetiological diagnosis: lack of resources (30%), insufficient standardisation in the approach to aetiological diagnosis (30%), difficulties in communication (23.3%) and lack of legal support (10%). These risks can be analysed in further detail as follows:

**Resources**
This category contains all the replies concerning the risks derived from the lack of resources, associated with the difficulties in covering the healthcare costs necessary to conduct an aetiological investigation.

**Scarce standardisation in the approach to aetiological diagnosis**
This category contains all the replies concerning the risks derived from the limited standardisation in approaching aetiological diagnosis. The lack of uniformity is most evident considering the competences of the third-level centres situated on the national territory, and it is likely to be related to the lack of shared guidelines. The training and the preparation of centres involved in aetiological diagnosis is variable, and diagnosis can be underestimated.
Furthermore, the resources for detection may be invested in a partial and unorganised way.

**Difficulties in communication**
This category contains all the replies concerning the Risks derived from the difficulties of communication in the field of aetiological research. The lack of communication between the different professional figures involved represents a risk for aetiological diagnosis and for future management of outcomes. In particular, there is a risk correlated to partial communication of the results during aetiological tests performed in third-level centres.

The difficulties in communication involve not only professionals, but also impair communication between healthcare operators and parents. Parents may sometimes show a scarce interest in the programme of aetiological diagnosis since they cannot understand its importance. The difficulties in communicating with the family are increased in all the situations in which there is social discomfort, and the lack of resources can make it even more difficult to adhere to the protocols of aetiological diagnosis. The cultural background is also important for an efficient communication. Difficulties in communication can also be enhanced by the presence of families with different cultural backgrounds. In these cases, the search for the causes of PHI may assume a more or less relevant connotation.

**Lack of legal support**
This category contains all the replies concerning the risks deriving from the lack of legal support. The lack of legal support defining requisites and competences of the third-level centres regard to aetiological diagnosis represents a risk for the standardisation of the approaches to aetiological diagnosis.

**Discussion**
The TOWS matrix, which relates Strengths-Opportunities, Strengths-Risks, Weaknesses-Opportunities, Weaknesses-Threats, was created starting from the data of SWOT analysis. An analysis of this type allowed to compare internal and external aspects to the system, to obtain specific recommendations and guidelines for the optimisation of the process of aetiological diagnosis. The recommendations can constitute an excellent cause for reflection for third-level audiologic centres, and can supply suggestions to optimise resources and produce positive changes. A total of 17 recommendations were obtained from the analysis and discussion of the data (Table II).

From the strategic study performed it was possible to identify four principal areas representing the strongest elements of strength for aetiological diagnosis. The drawbacks should be minimised and the opportunities implemented:

- implementation of shared protocols for aetiological diagnosis of infant hearing loss and definition of a shared

### Table II. TOWS matrix (see text for explanation).

<table>
<thead>
<tr>
<th></th>
<th>Strength (S)</th>
<th>Weakness (W)</th>
</tr>
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</table>
| **SO strategy** | 1. Implementing shared protocols for aetiological diagnosis of infant hearing loss  
2. Defining shared timing of the different investigations included in the protocol of aetiological assessment  
3. Creating structured and formalised multidisciplinary teams for audiologic, medical and aetiological assessment of children with impaired hearing  
4. Implementing a database or information system for data storage and exchange of information among various professionals  
5. Optimising times and modes of communication to the families on the importance of aetiological diagnosis, investigation programs, results of tests and their meanings |  |

| **ST strategy** | 1. Optimising resources for the implementation of standardised and shared protocols for aetiological diagnosis  
2. Optimising the modes of access to the structures/operators involved in aetiological investigations to streamline procedures and reduce waiting times  
3. Improving communication among healthcare operators, and between operators and families by exploiting, optimising and exporting database and information systems available in some regional realities and dedicated personnel of associations  
4. Identifying moments shared between operators and families to communicate the results of the aetiological examinations and of their meanings |  |

| **WO strategy** | 1. Establishing and standardising times and modes of access to the structures and specialists involved in aetiological diagnosis  
2. Optimising and standardising the collaboration of the multidisciplinary team  
3. Improving communication among the members of the multidisciplinary team by implementing databases or information systems, or moments of multidisciplinary meetings  
4. Promoting systems of information and training addressed both to the operators forming the multidisciplinary team and to the families on the importance and meaning of aetiological assessment |  |

| **WT strategy** | 1. Motivating institutions and associations on the importance of operator training  
2. Involving the associations in informing the families on the importance and meaning of aetiological assessment  
3. Stimulating the institutions and associations on the need for legal support, by fostering the implementation and realisation of protocols for aetiological diagnosis  
4. Sensitising institutions and associations on the importance of databases and systems for the collection and exchange of information concerning aetiological diagnosis programmes |  |
The first recommendation concerns the necessity to include the child with confirmed diagnosis of hearing impairment in national protocols for aetiological diagnosis. Adherence to audiologic protocols for neonatal screening and consequent early diagnosis of child hearing impairment are associated with the need to reach an aetiological diagnosis within the first months of life. In compliance with JCIH, every child confirmed with the diagnosis of impaired hearing should be included in a diagnostic protocol aimed at determining the cause of the problem. In around 50% of cases, referred children with Phi confirmed, every child confirmed with the diagnosis of Phi, through which they managed to identify the cause of impaired hearing in approximately 1/3 of cases.

The definition of the aetiology of deafness provides important prognostic information both for the evolution of the defect for global development of the child and for outcomes of prosthesis-rehabilitation. Furthermore, early aetiological diagnosis can favour the identification of problems associated with PHI, by facilitating the activation of targeted and prompt recovery intervention, and provide important information for parent and family from both a genetic and prognostic point of view. No shared protocols currently exist (at a national level) for aetiological diagnosis in UNHS referent children, in which the presence of impaired hearing has been confirmed. Each Centre of Reference (III level) conducts the search independently using the means available.

From the study of the literature it appears evident that the UNHS programs envisage structured and shared protocols for aetiological and medical diagnosis, alongside the protocol of audiologic diagnosis. In this respect, Declau et al. have reported a protocol for aetiological diagnosis applied to refer newborns at UNHS for which the PHI presence was confirmed. The screening allowed to identify the causes of PHI in around 50% of cases. Leenheer et al. proposed a step-by-step protocol for aetiological assessment of referred newborns at screening, with confirmed diagnosis of impaired hearing. More recently, Lemmens et al. implemented a protocol of aetiological diagnosis which they applied to a group of 505 children with confirmed diagnosis of PHI, through which they managed to identify the cause of impaired hearing in approximately 1/3 of cases.

In 2014, the American College of Medical Genetics and Genomics published guidelines for clinical assessment and aetiological diagnosis of impaired hearing, and faced the problem of prelingual hearing impairment.

The congenital infection from CMV represents a meaningful example of the importance of early aetiological diagnosis. At present, congenital infection from CMV seems to be the only significant infectious pre-natal cause of congenital or prelingual hearing impairment, owing to the fact that toxoplasmosis, epidemic parotitis and rubella have become rare thanks to the prevention performed during pregnancy and vaccination campaigns. In industrialised countries, moreover, congenital infection from CMV is estimated to be the most common congenital infection, with a prevalence at birth around 0.3-0.6%; therefore, congenital infection from CMV is currently considered the major non-genetic cause of childhood hearing impairment. Approximately 10% of newborns with congenital infection are symptomatic at birth, and among these there is a high risk of developing neurological sequelae, including neurosensorial PHI. Of the remaining 90% of asymptomatic newborns, around 6-23% have or will develop PHI. In case of congenital infection from CMV, hearing impairment can be present at birth or can occur months or even years after birth, and in more than 50% of cases it has a progressive trend. With regard to the possibility of treating newborns affected by this congenital infection, over the last few years evidence has emerged that antiviral therapy with ganciclovir (intravenous) or vanganciclovir (oral) administered in the first month of life can be effective in reducing the risk of neurological problems, and in particular in preventing the progression of impaired hearing.

It is important to perform diagnosis of congenital infection of CMV as early as possible, in order to be able to distinguish between congenital and acquired infection, and to offer the child the possibility of pharmaceutical treatment. Today, the method considered to be the most effective is testing for viral DNA in urine or saliva within the first 21 days of life.

To date, from the analysis of literature, there seem to be no neonatal screening programmes for this type of pathology. Within the framework of the general protocol for the research of the aetiology, the study of CMV thus represents an important field of investigation. Implementation of protocols for neonatal screening for CMV infection, restricted to refer newborns at neonatal audiologic screening, can be fundamental for early aetiological diagnosis. Recently, Williams et al. published a study on the feasibility of screening for congenital infection from CMV, concluding that saliva screening, aimed at UNHS referred newborns, is feasible and well accepted by families, and allows newborns with PHI from congenital CMV infection to benefit from pharmacological therapy with antiviral drugs.

The protocol of neonatal audiologic screening of the Tuscany Region (obligatory from November 2007) expects that the congenital infection from CMV is investigated early in all referred newborns with otoemission at screening, through viral DNA testing in urine by PCR. This will
make it possible to distinguish any CMV congenital infection from other infections contracted later. To our knowledge, this type of investigation is not yet routinely performed in other neonatal audioligic screening protocols. Moreover, since congenital infection from CMV seems to be a factor predisposing to foetal death in the uterus, premature birth and low-weight conditions due to gestational age (SGA: small for gestational age), our working group has looked for the presence of CMV infection in all SGA newborns and in premature infants admitted to the Operative Unit of Neonatology at the University Hospital in Pisa from November 2005 to April 2009. In the study, an association was found between congenital infection from CMV, premature birth (3.03%) and SGA newborns (3.7%)\(^1\).

In addition to neonatal investigations for diagnosing congenital infections from CMV, another external possibility is the creation of a network for collaboration among gynaecologists. These specialists are the first to get in contact with the future mothers, and therefore could immediately sensitise them on the problem, favouring early diagnosis of maternal infections from CMV.

The implementation and performance of shared protocols of aetiological assessment would allow to obtain not only an early aetiological and medical diagnosis, with all the benefits mentioned above, but also a common approach among the various structures so as to guarantee greater uniformity of intervention. More precisely, the protocol should establish the times and ways of access to the structures and the various specialists involved, defining the timing of the various investigations. The definition of these aspects could streamline the researching procedures and positively affect healthcare costs. The absence of shared protocols sometimes leads to a useless repetition of the same diagnostic tests on the same child in different centres (e.g. genetic tests), in the event that a family decides to consult various audiologic centres, with a waste of resources.

Each third-level audiologic centre should possess the competences and means to manage the entire process of etiological research. The possibility of joining only one centre of reference would support the child and its family at the same time, and reduce the dispersion of clinical data.

The second recommendation identified within the field of aetiological diagnosis regards the possibility of structuring a formalised multidisciplinary team for audiologic, medical and aetiological assessment of the hearing-impaired child.

The process leading to the definition of an aetiological diagnosis involves operators belonging to different disciplines. The protocol followed by our centre mainly involves the following specialists: audiological physician, geneticist, neonatologist, ophthalmologist, neuroradiologist, child neuropsychiatrist and paediatrician. In a later stage, and according to the specific needs of the patient, other operators can be involved. Taking on global responsibility for the patient, with integrated interventions, is based on the synergy and collaboration of all the members of the multidisciplinary team, even when they belong to units that are located in different centres. It is important that the professionals involved in the team are connected by a solid and efficient communication network that is able to guarantee a constant and bi-directional exchange of information. Towards this end, the figure of the family paediatrician plays an important role: considering the continuous relation that paediatricians have with the family of the child, their collaboration is fundamental to monitor the development of the aetiological investigation process.

An important aspect of the second recommendation concerns the training of the members of the multidisciplinary team. Participation in a protocol of aetiological diagnosis requires the acquisition of basic notions that allow all the members of the team to understand the importance of the investigations and their prognostic outcomes. An opportunity in this field is represented by the participation in specific training courses. In this regard, we report the positive experience of the Tuscany Region, which organised compulsory training courses for the family paediatrician in the years 2009-2010. The course, divided into two days, faced the main themes of child with PHI, and included a session aimed at checking the knowledge that had been learned. This initiative allowed to give greater uniformity to the audioligic knowledge of paediatricians, and had a positive impact on the procedures of management of all at-risk patients as well as of the patients with previously confirmed diagnosis. The possibility of involving institutions and associations, so that they may promote the realisation of specific training projects in the field of childhood hearing impairment, could represent a strategy designed to standardise the know-how of multidisciplinary team members. The third recommendation concerns the possibility for the members of the multidisciplinary team to access databases or systems for the data storage and the exchange of information.

Access to the data concerning aetiological investigations and outcomes by all the professionals of the team can stimulate multidisciplinary collaboration, thus facilitating the exchange of information. Update of information included in the database (tests performed, times, results of the investigations, evaluation gaps, future appointments, etc.) would allow the professionals responsible for the child to check whether the protocol of aetiological diagnosis has been followed correctly or whether further investigation is necessary. In addition, systematic data collection represents an opportunity for the realisation of future studies, both epidemiological, and on the aetiology of child hearing impairment. Within the framework of neonatal audioligic screening in the Tuscany Region, a telematic
registry (database) has been established for the collection of data concerning newborns that were referred to UNHS. The database will report the audiologic and aetiological outcomes and prosthetic rehabilitation treatment undertaken. The registry will also contain the data concerning referred newborns at screening who present elements of risk for progressive hearing impairment, or for late onset, so that they can be followed-up. The registry will need to be constantly updated by the healthcare staff operating at the different levels (neonatology, audiology, third-level centres of reference).

The possibility of referring to a shared database clashes with the risk correlated to the lack of resources, influencing both the design and diffusion of the tool, as well as the scarce availability of the personnel destined to its compilation.

In the absence of a shared database, drafting of the written reports containing the results of the assessments and tests – and also the availability of telephone numbers and e-mail addresses of reference – could favour the exchange of information among healthcare operators.

Even in this case, a major external risk is represented by the impossibility to use the resources that could be exploited to involve the staff responsible for these activities. The final recommendation concerns the need to involve the parents in the plan of definition of aetiological diagnosis. Full adhesion to any protocol of aetiological diagnosis requires good parental collaboration. The respect of all these passages is closely correlated to the possibility of understanding the importance of each step for a diagnosis and, in turn, the importance of the diagnosis for the implications it may have on a child’s development. The external risks of this recommendation regard the difficulties in achieving clear and exhaustive communication with the family. The specialists will have to make sure that the family is fully aware of the types of investigation considered by the aetiological protocol and of the modes and times necessary for it to be performed. Once a diagnosis has been made, the results will have to be communicated to the family, by explaining the implications it may have in terms of prognosis for the progress of the disturbance and in general for the development of the child. The language used with the relatives will need to be adapted to their socio-linguistic characteristics (e.g. foreign parents with poor competence in Italian language), and the specialists will need to be sure that the parents have understood the contents of the message. In order to facilitate the entire process, it might be useful to plan moments of shared participation between operators and families during which the outcomes of the aetiological examinations and their meanings will be explained.

Conclusions

With the implementation of newborn hearing screening programmes, the need to implement effective and stand-ardised protocols for aetiological diagnosis has clearly emerged, to be carried out promptly, for the identification of the cause of PHI and the presence of any comorbidities or associated disabilities, so as to rapidly offer an optimal and customised treatment to each child affected by prelingual PHI. Within the framework of this project, following a first SWOT based on a review of the international literature, a second overall SWOT analysis was elaborated, resulting from an exchange and discussion among 21 experts in paediatric audiology. The data obtained were then used for the realisation of a TOWS matrix from which four main recommendations were identified, useful for the search of aetiological diagnosis for childhood PHI:

1. implementation of shared protocols for etiological PHI diagnosis in UNHS referred newborns, for whom the presence of PHI was confirmed and a shared programme of timing for several diagnostic investigations was defined;
2. creation of structured and formalised multidisciplinary teams for audiologic, medical and aetiological assessment of the hearing-impaired child;
3. implementing databases or systems for data storage and exchange of information among various professionals;
4. optimising the times and modes of communication to the family about the importance of aetiological diagnosis, investigation programmes, outcomes of tests and their meanings.

For third-level audioligic centres, the recommendations obtained from this process represent valid suggestions for the optimisation of resources and creation of positive changes.

References

Aetiologic diagnosis of hearing loss in children identified through newborn hearing screening testing


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