Cochlear Implants in Children Diagnosed with CHARGE Syndrome

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Abstract

Introduction The CHARGE association (coloboma of the eyes; heart disease; atresia of the choanae; retarded growth and development; genital hypoplasia/genitourinary anomalies; ear anomalies and/or hearing loss) was first described in 1979 by Hall, and among its main features is hearing loss. This study presents a case aiming to establish relationships between performance on Infant Toddler Meaningful Auditory Integration Scale (IT-MAIS) and Meaningful Use of Speech Scales (MUSS) tests and the analysis of hearing and language categories of a patient diagnosed with CHARGE syndrome, before and after cochlear implant (CI) surgery.

Case Report A 7-year-old girl was diagnosed with CHARGE. She had severe sensorineural hearing loss and was a prelingual unilateral CI user. We analyzed data from the patient's medical records regarding therapies and video recordings.

Results The patient showed positive results in all evaluations after CI. IT-MAIS rose from 5 to 90% following the use of CI. MUSS also rose, from 75 to 72.5%, after use of CI. Classification of Auditory Skills changed from category 1 before use of CI to category 6 after use of CI. Classification of Language Skills changed from category 1 before use of CI to category 3 after use of CI. The CI is an aid but there are many factors in the therapeutic process, and great heterogeneity in individuals diagnosed with CHARGE should be investigated.

Conclusion The development of listening and language skills after CI use was demonstrated by IT-MAIS and MUSS tests, and categorization of speech and hearing in this child with a diagnosis of CHARGE syndrome shows that CI can be an effective technological resource to provide information on hearing as one source for language construction.

Keywords

- ► charge syndrome
- ► cochlear implants
- ► hearing
- ► language

Introduction and Literature Review

The CHARGE association, or CHARGE syndrome, was first described in 1979; in 1981 the acronym CHARGE (coloboma of the eyes; heart disease; atresia of the choanae; retarded growth and development; genital hypoplasia/genitourinary anomalies; ear anomalies and/or hearing loss) was proposed to portray this set of findings.[1]

The auditory sensory deficit may jeopardize learning, especially due to injury in the acquisition and development of oral language, which varies depending on the type and degree of hearing loss. Even a mild loss can interfere with oral language development of children and their academic

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success. There is no doubt, however, that children diagnosed with severe and profound hearing loss are more susceptible to significant lags in language acquisition and in the educational process.[2,3]

Cochlear implants (CIs) are electronic devices of high biomedical technology, developed to perform the function of cochlear hair cells that are damaged or missing. CIs provide direct electrical stimulation of the remaining nerve fibers in individuals with severe and profound hearing loss, allowing the transmission of electrical signal to the auditory nerve to be decoded by the cerebral cortex. The CI provides the feeling of listening and the quality required for the perception of speech sounds.[4]

Considering the use of CIs in patients diagnosed with CHARGE syndrome, it is important to pay attention to variation of temporal bone anomalies and anatomical variations of facial nerve pathway, factors that may hamper or prevent surgery.[5]

CI centers are gaining more experience and application criteria for surgery are in expansion, but children with additional disabilities continue to be topic of discussion. Many centers perform implant surgery in children with additional disabilities, but this population is very diverse and presents unique challenges. The literature suggests that 30 to 40% of children with sensorineural hearing loss have an additional disability.[6]

Therefore, it is vitally important to monitor the progress and assess the evolution in these cases, as well as to disseminate the scientific results, to aid in decision making of challenging cases with multiple disabilities.

To verify the hearing abilities in very young children, we used the Infant Toddler Meaningful Auditory Integration Scale (IT-MAIS) questionnaire, adapted by Castiquini and Bevilacqua.[7] This is a substantial auditory integration scale for young children, which also searches children's spontaneous auditory behaviors in daily life situations, using examples in three different areas of the auditory skills development. These areas include changes in vocalization associated with device usage, being alert to environmental sounds, and assigning meaning to sounds. Using information provided by parents, the examiner scores each question by the frequency of behavior occurrence that ranges from 0 ("Never shows this behavior") to 4 ("Always shows this behavior").

For evaluation of development of language skills, we used an adaptation of the Meaningful Use of Speech Scales (MUSS), which is an oral language assessment questionnaire with closed questions whose main objective is to evaluate the spoken language use by the child.[8]

The experience of using CIs in children with CHARGE syndrome is still incipient and the international literature reports few clinical cases, as in the studies of Bauer et al,[9] Lanson et al,[10] Southwell et al,[11] and Meinzen-Derr et al.[12] This study presents a case aiming to establish relationships between the performance in IT-MAIS and MUSS tests and the analysis of hearing and language categories of this patient with a diagnosis of CHARGE syndrome, before and after CI surgery, in an attempt to contribute to these results about the expectations for other similar cases.

Case Report

This project was approved by the ethics committee and research at the Catholic University of Brasilia, under protocol 241/2010. Upon approval, data collection was conducted during March and April 2011. The subject of this study was a 7-year-old girl diagnosed with severe sensorineural hearing loss, which occurred before language acquisition—in other words, she was a prelingual unilateral CI user and diagnosed with CHARGE syndrome. The main way of communicating with the survey participant was through spoken language. The survey participant and her guardian consented to take part in the survey.

At the time the research was performed the participant attends regular school and is in her second year (old third grade of elementary school) and is monitored twice a week. The patient has speech therapy sessions twice a week, lasting 1 hour, with family participation.

Data from medical records of patient, comprising the record of the therapies and video recordings of therapy sessions, were analyzed. The participant and the family answered questions on the IT-MAIS and the MUSS.[7,8]

Upon collection, the data gathered before and after cochlear implantation were compared. The hearing and language development were classified according to categories of hearing and language.[13,14] Data collected were predominantly descriptive because it is a cross-sectional study comparing the data obtained prior to cochlear implantation with the data obtained after a certain time of CI use.

Results

In analyzing the records, the patient has most features of CHARGE syndrome (►**Table 1**). The patient medical records included the date the CI was activated, sensorial privation time (from birth), and time of auditory development as described in ►**Table 2**. The results of applying the assessment before surgery (2007) and the most current data (2011) are presented in ►**Tables 3** and **4**.

Regarding the evolution of language categories (►**Table 5**), in 2007, without CI, the patient's performance was rated category 1, and in 2011, using CI, the performance was rated category 3 (►**Table 5**). In the categories of hearing (►**Table 6**),

Table 1 CHARGE syndrome main features

Coloboma	Cardiac alteration	Atresia of the choanae	Retarded growth and development	Genital hypoplasia	Hear alteration
Yes	Yes	Yes	Yes	No	Yes

Table 2 Survey records

Current age	Age at CI activation	Sensorial privation time	Time of auditory brain development
7 y, 9 mo	4 y, 4 mo	4 y, 4 mo	3 y, 5 mo

Abbreviation: CI, cochlear implant.

in 2007, without the CI, the patient was in category 1, and in 2011, with the use of CIs, the patient was in category 6.

With a hearing age (time of use/CI activation) of 3 years and 5 months, the patient is currently in the highest category of hearing, category 6, recognition of open-set word. This child is able to hear words out of context and extract sufficient phoneme information, and she can recognize words solely through hearing, which differs from results found in 2007 without the use of CI, when the patient was in category 1, detection. This child detects presence of the speech signal.

Discussion

The child in this study presented positive results in all evaluations after cochlear implantation, as expected from the literature with nonsyndromic cases. The child made significant gains in the IT-MAIS, which is a scale designed to access the significance of hearing loss for the child in the use of sounds in a daily life situation; her scores rose from 5 to 90% following the use of CIs.

Bauer et al studied six patients diagnosed with CHARGE syndrome who used CIs; the IT-MAIS was used in only two patients, one of whom progressed within 6 months from 7.5 to 77.5% and the other patient from 7.5 to 15% after 6 months of implantation.[9]

Lanson et al, in study with 11 children diagnosed with CHARGE syndrome, concluded that the patients had varying results and some limited degrees of hearing benefit from CIs.[10] These authors also emphasized the importance of counseling parents regarding realistic expectations from CIs.

Regarding the MUSS, which is a structured interview with parents and covers information about the frequency with which the child shows significant oral language behaviors in his or her daily routine, the results post–cochlear implantation were lower compared with the results of IT-MAIS, rising from 7.5 to 62.5%, demonstrating that the patient had greater oral communicative abilities upon activation of the CI.

The patient reached category 6 of hearing after cochlear implantation, which is equivalent to a step more advanced listening. Comprehension is the more refined hearing ability,[4] because it requires that the individual understands the meaning of the message. To understand, the subject must possess the domain of listening skills mentioned earlier, such as detection, discrimination, and recognition. The patient must be able to hear words out of context and extract sufficient phoneme information and to recognize the word solely through hearing.

Bauer et al, in a study of children diagnosed with CHARGE syndrome, found a patient who after a year of CI advanced to the recognition ability and another patient who after 4 years of CI use progressed to the comprehension skill in an open hearing, equivalent to the category 6.[9] In the present survey, the patient, after 3 years and 5 months of CI use, progressed to the hearing comprehension in open set.

As seen in ►**Table 6**, the patient progressed in oral language, although the gains were small, leaving category 1 ("This child does not speak and can present undifferentiated vocalization") and moving into category 3 ("This child builds sentences"). The child in this study has deficits including several factors cited as features of CHARGE syndrome, which may justify the slow progress in the language category.

The development of listening and language skills depends on several factors, such as[15]:

- degree and time of hearing loss
- age at detection and intervention
- the child's characteristics: cognitive style, ability to build language, psychological aspects (memory and attention), and emotional development
- family characteristics: attitudes and skills of parents and siblings, suitable environment, acoustic environment at home and school context, which favors the development of listening skills
- therapist and/or teacher properly performing work

In hearing impaired patients, additional disabilities may impact the development of language as related to the use of CIs,[16] and children with multiple disabilities are more prone to additional use of Total Communication,[17] which is an educational philosophy for the deaf that has an oral and manual code (such as spontaneous gestures, manual alphabet, Portuguese flagged, and others), compared with children with only one additional disability.

The patient receives speech therapy by the Aurioral method, which prioritizes hearing as described by Bevilacqua and

Table 3 Ranking of the IT-MAIS after 3 y and 5 mo of cochlear implant use

2007, without cochlear implant	2011, using cochlear implant
5%	90%

Abbreviation: IT-MAIS, Infant Toddler Meaningful Auditory Integration Scale.
Source: Castiquini and Bevilacqua.[7]

Table 4 MUSS score

2007, without cochlear implant	2011, using cochlear implant
7%	62.5%

Abbreviation: MUSS, Meaningful Use of Speech Scales.
Source: Nascimento and Bevilacqua.[8]

Table 5 Classification of language skills

Categories	2007	2011	Language development
1	Without cochlear implant		This child does not speak and can present undifferentiated vocalization.
2			This child speaks only isolated words.
3			This child constructs simple sentences of 2 or 3 words (after cochlear implants).
4			This child constructs sentences with 4 or 5 words and uses connecting elements (pronouns, articles, prepositions).
5		With the cochlear implant	This child constructs sentences of more than 5 words, using connecting elements, conjugating verbs, plurals, etc. She is fluent in oral language.

Source: Proposed by Bevilacqua et al.[14]

Table 6 Classification of auditory skills

Categories	2007	2011	Auditory ability
0			This child does not detect speech in situations of normal conversation (speech detection threshold of $>$ 65 dB).
1	Without cochlear implant		Detection: This child detects the speech signal.
2			This child differentiates between words by suprasegmental features (duration, tone, etc.); example: *hand* versus *shoe*, *house* versus *boy*.
3			Starting word identification: This child differentiates between closed-set words based on phonetic information. This pattern can be demonstrated with words that are identical in length but contain multiple spectral differences; example: *refrigerator* versus *bike*, *cat* versus *home*.
4			Identification of words through vowel recognition: This child differentiates between closed-set words that differ primarily in the vowel sound; example: *foot*, *powder*, *shovel*; *hand*, *my*, *me*.
5			Identification of words through consonant recognition: This child differentiates between closed-set words that have the same vowel sound but contain different consonants; example: *hand*, *bread*, *so*, *dog*, *floor*.
6		With cochlear implant	Recognition of open-set word: This child is able to hear words out of context and extract sufficient phoneme information, and recognize the word solely through hearing.

Source: Geers.[13]

Formigoni.[15] As the patient of this study shows characteristics other than the hearing disability, the Total Communication method could assist her in developing listening skills and language.

The CI is an aid, but there are many factors in the therapeutic process, and great heterogeneity in individuals diagnosed with CHARGE syndrome should be investigated, not only to enable the development of appropriate auditory behavior but to ensure that this influence more positively impacts the development of language.

Conclusion

The development of listening and language skills were demonstrated by gains in IT-MAIS and MUSS and categorization of speech and hearing in this child with a diagnosis of CHARGE syndrome; this study shows that CIs can be an effective technological resource to provide information on hearing as one source for language construction.

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