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Case reports

Cochlear implant in a child diagnosed with Dandy-Walker Syndrome Variant: a study case

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ABSTRACT

Dandy-Walker Syndrome Variant presents itself as a milder form of Dandy-Walker Syndrome, with less pronounced vermis hypoplasia, and hearing impairment is among its characteristics. This study aimed to report the case of a male patient aged 4.5 clinically diagnosed with Dandy-Walker Syndrome variant, a cochlear implant user, who was referred to rehabilitation services and followed up by a multidisciplinary team. The patient underwent therapy assisted by an audiologist/speech therapist and a physiotherapist between June 2016 and December 2016, totaling 20 sessions, with emphasis on the Aurioral approach. His evolution regarding hearing and motor abilities was evaluated through standardized instruments that helped to catalogue the patient's evolution and responses in an empirical way. The development of his auditory and motor skills, evaluated through standardized tests used as parameters of therapeutic evolution, demonstrated that rehabilitation, performed by a multi-professional team, can be satisfactorily applied in the management of cases where deafness does not appear as the only associated factor. It is suggested that a cochlear implant, despite the difficulties peculiar to the syndrome in question, can be an effective resource to acquire oral language and reach more complex stages related to hearing and language skills.

Keywords: Cochlear Implant; Dandy-Walker Syndrome; Psychomotor Performance; Rehabilitation; Language

INTRODUCTION

Dandy-Walker Syndrome is characterized by cystic dilatation of the fourth ventricle, by aplasia, either total or partial hypotrophy of cerebellar vermis, and it is a non-familial syndrome in which cerebral malformations with corpus callosum agenesis, heteropsias, lissenphaly, and stenosis of the aqueduct of Sylvius may occur. The Dandy-Walker variant presents itself as a milder form, with less pronounced hypoplasia of the vermis and hydrocephalus is less common [1,2].

Auditory deficit is among its main characteristics. Bilateral sensory-neural deafness may be part of the clinical picture of the syndrome, and a cochlear implant can be an option to reduce the effects of deafness on oral language acquisition[3]. Treatment involves a multidisciplinary team and the prognosis is variable according to the phenotype[4].

The objective of this study was to evaluate the auditory, linguistic and motor skills of an implanted child included in a speech therapy program with aurioral approach by a multidisciplinary team.

CASE PRESENTATION

The research was approved by the Ethics Committee of the State University of Amazonas, normative act 1.679.634, the Free and Informed Consent Form was signed by the people in charge, according to Resolution CNS 466/2012. The casuistry of this research consisted of a child, KL, 4.5 years of chronological age, male, diagnosed with profound bilateral sensorineural hearing loss at 1 year and 8 months of age, due to the Dandy-Walker syndrome variant.

Data from the patient's record show audiological exams performed before the implant: Transient Otoacoustic Emissions and Evoked Otoacoustic Emissions, Distortion Product with absent responses in all frequency bands in both ears. In the electrophysiological hearing assessment, Brainstem Auditory Evoked Potentials were not detected bilaterally. The parameters used to perform the examination by air conduction were click stimuli, with alternating polarity and frequency rate of 27.1 clicks per second. As for the wave morphology at 100 dB HL, the V wave was identified with bilaterally increased absolute latency time, OE / OD with minimum response level at 95 dB HL. By bone conduction a click stimulus was used, with alternating polarity and frequency of 27.1 clicks per second, without contralateral masking, OE / OD with no response at 60 dB HL.

Tympanometry: bilateral type A curve. Absence of contralateral and ipsilateral stapedial reflexes. Behavioral hearing test with no response to the instruments and no observed VOR (Vestibulo-Ocular Reflex). From this point on, there was a selection, indication and adaptation process of PSAP (Personal Sound Amplification Device/Product) in the institution that referred the patient to this service. The patient received, at the time, a pair of appliances, model NAIDA I UP, brand Phonak, adapted in bilateral shell mold. Free Field audiometry indicated auditory thresholds with PSAP in: 250 Hz-80dB HL, 500Hz-100 dB HL, 1000Hz-100dB HL, 2000HZ-100dB HL, 4000Hz-100dB HL, that is, the child was not able to detect speech even with the use of personal sound amplification devices, and undergoing therapy based on the aurioral approach.

CI surgery was performed at 2.5 years of age, with full insertion and adequate functioning of all electrodes, verified in impedance telemetry, in addition to adequate neural response. The inserted device was the Harmony model by Advanced Bionics. At 2.6 months of age the implant electrodes were activated, with the speech processor programmed with the HiRes 120 speech coding strategy. According to the family, the child makes effective use of the CI, however, has not used the PSAP in the contralateral ear, despite the indication/recommendation, since the implant activation. After CI, the free-field audiometry presented 35 dB on average and logoaudiometry 25 dB.

The diagnosis is Dandy-Walker Syndrome Variant. Magnetic resonance imaging of the encephalon indicates cerebellar vermis hypoplasia and mild dilatation of the cisterna magna (Figures 1 and 2).

Figure 1. Magnetic resonance imaging, axial section evidencing hypoplasia of the cerebellar vermis

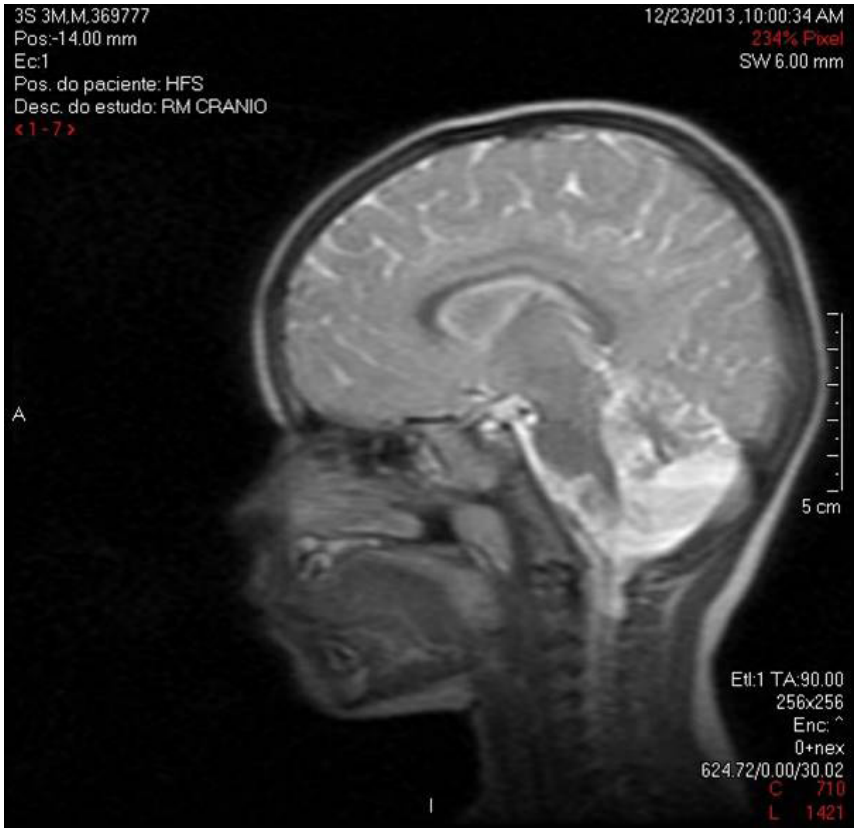

Figure 2. Magnetic resonance imaging, sagittal section evidencing cerebellar reduction and slight dilatation of the cisterna magna

A karyotype study was performed, which revealed there was no consanguinity between the parents and no history of gestational complications. The patient presented delayed psychomotor development, significant psychomotor agitation and changes in balance. The child was included in a therapeutic process with an audiologist/speech therapist and a physiotherapist, in joint care, always using the CI device, between July 2016 and December 2016, totaling 20 sessions, with emphasis on the Aurioral approach. He presented a hearing age of 22 months at the beginning of the service. The therapeutic goals and objectives have prioritized certain aspects of rehabilitation to develop auditory skills such as: detection, discrimination, recognition and comprehension of oral language, associated to improvement of motor coordination and balance adequacy. At the beginning of each session, the proper functioning of the CI device was checked through the Ling sounds[5], in which six phonemes representing the audibility of speech sounds were evaluated.

The data of the patient's chart containing the records of the therapies performed were analyzed. With this in mind, the evolution of the patient was evaluated through specific protocols, according to the child's age, which helped establish a comparison in a given period of therapeutic care, as well as catalogue and evaluate the patient's evolution and responses empirically.

For the evaluation of hearing and language skills, parents responded to the Significant Hearing Integration Scale protocol for small children - IT-MAIS, translated and validated for Portuguese[6], besides the classification of hearing and language development according to hearing and language categories [7,8]. For the evaluation of motor skills, during care, the Developmental Coordination Disorder Questionnaire was used. DCDQ-Brazil, whose aim was to detect motor development disorders, through which it is possible to identify if motor difficulties interfere significantly in everyday activities.[9]

The so-called IT-MAIS (Significant Hearing Integration Scale for young children) is a protocol used to verify the auditory abilities of young children and aims to evaluate the perception of speech in children under 4 with profound hearing impairment. It is organized in 10 simple questions related to spontaneous auditory behaviors in everyday situations, it aims to evaluate three areas of development, such as changes in vocalization associated with the use of the device, alert of environmental sounds and attribution of meaning to sound. Each question on the scale corresponds to 5 points, ranging from 0 (zero) to 4 (four), 0 = never, 1 = rarely, 2 = occasionally, 3 = often and 4 = always. These questions add up to 40 points that correspond to 100%, indicating optimal auditory development in relation to attention and recognition of speech sounds.

Thus, the application was performed through an informal interview with parents, who were asked to report, as faithfully as possible, the child's auditory behavior, and provide examples of day-to-day development. At the end of the application, scores were assigned according to the categories proposed in the instrument, and the hearing ability was classified according to the hearing categories [8,10].

Category 0 – no speech detection. This child does not detect speech in normal conversation situations (speech detection threshold > 65 dB).

Category 1 - detection. This child detects the presence of the speech signal.

Category 2 - pattern of perception. This child differentiates words by supra segmental characteristics (duration, tone, etc.). e.g.: mão (hand) X sapato (shoe), casa (house) X menino (boy).

Category 3 - initiating word identification. This child differentiates between words in a closed set based on phonetic information. This pattern can be demonstrated with words that are identical in duration but contain multiple spectral differences, e.g. geladeira (refrigerator) X bicicleta (bicycle), gato (cat) X casa (house).

Category 4 - word identification through vowel recognition. This child differentiates between words, in a closed set, which differ primarily in the sound of the vowel. e.g.: pé (foot), pó (powder), pá (shovel); mão (hand), meu (mine/my), mim (me).

Category 5 - identification of words by means of consonant recognition. This child differentiates between words, in a closed set, which have the same vowel sound, but contain consonant differences. e.g.: mão (hand), pão (bread), tão (so/too), cão (dog), chão (ground).

Category 6 - word recognition in open set. This child is able to hear words out of context and extract enough phonemic information and recognize the word exclusively through listening.

For classification of hearing and language categories, behaviors, auditory responses and oral communication were also evaluated in interactive situations through activities in the therapeutic environment[11]. It was possible, therefore, to record clinical observations and classify the hearing and oral language aspects, as originally proposed by the therapeutic

goals. In the end, scores were assigned according to language categories.

DCDQ-Brazil

To evaluate the skills and motor development, the Development Coordination Disorder Questionnaire (DCDQ), developed in Canada and adapted to Brazilian Portuguese for Brazilian children, was used[9]. The instrument has a specific questionnaire for the detection of motor development disorders. It consists of fifteen items that evaluate the performance of the child in different everyday life situations, whose questions are divided into three groups: motor control during movements, fine / written motor and general coordination. The items were scored on a five-point scale (1 = nothing like your child) and 5 (extremely similar to your child), with a maximum score equal to 75. The higher the score, the better the child's motor performance.

After completing the questionnaire, the scores of each item were added to obtain the final score. The maximum score, adding the points of the three areas, is 75. Three cut-points were developed for identification of DCD (Developmental Coordination Disorder). A score 0-46 indicates that the child has DCD or may have DCD. A score 47-75 indicates that the child probably does not have DCD. The authors reiterate that the final result should be compared to the observation of the child both in a therapeutic environment and in an informal environment. Thus, the instrument serves to screen and identify children who are at risk for DCD, Developmental Coordination Disorder, in which it is possible to identify if motor difficulties significantly interfere with activities of daily life[9].

It should be observed that all the evaluation instruments described were applied in two different moments. The first evaluation was performed in July 2016, when the child was referred by another service to assistance, and the second was performed after 6 months of therapy, in December 2016.

RESULTS

Evaluation of the development of auditory and language skills

Table 1. Results from the first evaluation on after 6 months of therapy

Assessment	1st Evaluation made in July 2016	2nd Evaluation made in Dec. 2016
IT-MAIS	32.5% Report of increase in vocalizations after the use of CI, attention to the call of the patient's own name in a quiet environment. Attention to the environment sounds.	52.5% Patient responds to his name spontaneously in different environments, can discriminate environmental sounds in relation to speech sounds, and discriminates different voices .
Hearing Category	2	4
Language Category	1	3

Legend: CI =Cochlear Implant

With regard to hearing and language skills, the percentage started to set at 52.5% in IT-MAIS. The improvement of the performance of the hearing and language categories can be evidenced by the better results in the auditory comprehension, arranged in Tables 2 and 3.

After analyzing Tables 2 and 3, it can be seen that the patient presented improvement in oral language, moving from category 1 (the child does not speak and can present undifferentiated vocalizations) to category 3 (this child builds sentences). Despite difficulties in aspects related to Dandy-Walker syndrome, these results guarantee quality of life. This information comes from parental reports when the questionnaires are applied.

Table 2. Evolution of language skills[6]

Categories	July 2016	Dec.2016	Language Development
1	Starting Age: 4.5 Hearing Age: 22 months		This child does not speak and can present undifferentiated vocalizations.
2			He speaks only isolated words.
3		Age: 4.11 Hearing Age: 28 months	He builds simple sentences composed of 2 or 3 words (After Cochlear Implant).
4			He builds sentences of 4 or 5 words, and starts using connective elements (pronouns, articles, prepositions).
5			This child builds sentences of more than 5 words, using connective elements, conjugating verbs, using plurals, etc. He is fluent in oral language.

Table 3. Evolution of Auditory Skills[7]

Categorias	Jul/2016	Dez/2016	Development of Auditory Skills
0			This child does not detect speech in normal conversation situations (speech detection threshold> 65 dB)..
1			Detection: This child detects the presence of the speech signal.
2	Starting Age: 4.5 Hearing Age: 22 months		This child differentiates words by supra-segmental traits (duration, tone, etc.), e.g. mão (hand) X sapato (shoe); casa (house) X menino (boy).
3			Beginning of the identification of words This child differentiates between words in a closed set based on phonetic information. This pattern can be demonstrated with words that are identical in duration but contain multiple spectral differences, e.g. geladeira (refrigerator) X bicicleta (bicycle), gato (cat) X casa (house).
4		Age: 4.11 Hearing Age: 28 months	Identification of words through vowel recognition. This child differentiates between words in a closed set that differ primarily in terms of the vowel sound, e.g. pé (foot), pó (powder), pá (shovel); mão (hand), meu (mine, me).
5			Identification of words through recognition of the consonant. This child differentiates between words in a closed set that has the same sound as the vowel, but contains different consonants, e.g. mão (hand), pão (bread), tão (so), cão (dog), chão (ground).
6			Recognition of words in an open set. This child is able to hear words out of context and extract enough phonemic information, and recognize the word exclusively through hearing.

Assessment of the development of motor skills

It is important to note that the evaluation instruments were applied in two moments, an initial evaluation and another after six months of care to make it possible to determine a parameter of evolution of the child's responses, as shown in Figure 3.

Evaluation DCQD/Brazil				
Items evaluated at DCDQ / Brazil sessions	1st Evaluation July 2016		2nd Evaluation Dec. 2016	
	Instrument Score	Obtained Score	Instrument Score	Obtained Score
I. Motor control during movement.	30	17	30	18
II. Fine motor skills and writing skills.	20	17	20	17
III. General motor coordination.	25	11	25	15
TOTAL SCORE	75	45	75	50
* Obtained Values	Score <46		Score = 50	
	The instrument suggests development coordination disorder or the patient is suspected to have it.		Probably does not indicate the disorder.	

* Instrument reference values: Results from 15 to 46 points are considered indicative of developmental coordination disorder and suggest individuals are considered at risk of having the disorder. Scoring from 47 to 75 show individuals may not have developmental coordination disorder.

Figure 3. Evaluation of Developmental Coordination Disorder

Thus, in the initial evaluation, observing the indicative values of the instrument to verify the significant disorder in motor coordination, the child presented scores lower than 46 for his age group, indicative of difficulties in coordination, reaching, in the second evaluation, a score equal to 50, indicating a satisfactory evolution, during which, it was possible to notice the improvement of the motor control during movements, fine motor and general coordination, despite the balance limitations imposed by the syndrome.

In this process, 15 aspects distributed in the DCDQ-Brazil sessions were evaluated, including motor control during movement, fine motor skills and writing and global motor coordination.

DISCUSSION

In the case presented herein, the patient's age of diagnosis is considered late (1 year and 8 months old), due to his long-term hearing loss as well as his not performing well when using PSAPs, which may negatively interfere with the development of hearing and language, seeing that a relevant part of the neuronal plasticity period of the central pathways was compromised.

Some studies confirm that the performance of the implanted child in the evaluated areas is directly related to the factors, such as: frequent use of the device, the age the patient was when the surgery was performed, time of sensory deprivation, hearing disability etiology, family involvement in the therapeutic process, in addition to the existence of impairments associated with hearing loss, among others [5,7,10-12].

However, despite these criteria and the association of this Syndrome with deafness, the participant, after being included in the therapeutic program, 6 months after the first IT-MAIS test, presented scores with a 20% increase, with significant gains in the hearing category , indicating that the CI contributed to a better auditory perception of this child, who started to speak more words with emission of sentences of up to two or more elements, at the end of this period, indicating a good evolution in terms of auditory and language skills. Evolving from Hearing Category 2 to 4.

It is observed that in the first application of the instrument (Table 1), there was an increase in vocalizations after the activation of the CI, a fact that can be attributed to the auditory feedback provided by the implant.

Besides, the application of questionnaires to parents, as well as the relationship between professionals and the family, enabled the extraction of important information related to auditory and linguistic development, as well as reports on the frequency

of these abilities appearing in the child's daily life, as some studies corroborate[10,12,13].

Studies described in the literature regarding the benefit of the implant in relation to hearing and language skills in cases involving the syndrome are quite scarce, only one publication suggests that the presence of Dandy-Walker syndrome cannot be considered a contraindication for surgery and the use of the cochlear implant[3]. However, other studies have described that, in children with hearing impairment, language development may be close to that of normal children if impairment is mild. On the other hand, children with more serious disorders may present a development that is below the expected. The use of CI in these cases would improve the perception of speech, although this improvement is often significantly lower than that found in children who do not have these additional disorders, which reinforces the present findings in this study[14-16].

It is important to mention that there are studies that point out that the development of hearing and language skills may be slower in children with syndromes other than deafness, especially in relation to expressive language, an aspect that can be verified in the findings of this study, in which the child presented, with regard to language skills, slower results when compared to hearing abilities[14,16].

Regarding the development of motor skills, the results showed that when comparing initial evaluation with reevaluation there was a significant improvement in trunk balance, plantar support, coordination and overall movement. Research has shown that both the cerebellum and the prefrontal cortex are two important areas for motor functions and attention aspects of the subject, which are more disturbed in patients with the syndrome[17,18]. Thus, it is important to report that the child presented, at first, psychomotor difficulties during the performance of activities involving motor coordination and balance, in addition to hearing impairment. Because of this, multidisciplinary management was regarded relevant considering the presented symptoms, a language rehabilitation program, associated with the motor activities in a global way, offered the patient an adequate development in a holistic way. Moreover, the instrument was used to early identify whether or not there was a developmental coordination disorder, in order to allow adequate intervention and support to reduce the negative consequences associated with this condition.

Specific cases, such as this one, lead us to have discussions in the implant centers regarding the indication of CI in children with hearing loss disorders. Those who opt for implantation aim to minimize auditory sensory deprivation, improve interaction with the environment, understand language and, consequently, have quality of life. Although the results described in these studies by researchers fall short of those presented by their peers without associated syndromes, sensory input is described as a facilitating factor for interaction and access to auditory and language skills [3,19-21].

Cochlear implant centers are gaining more experience and the application criteria for surgery are being expanded, but children with additional disabilities continue to be a topic of discussion. Many centers perform surgery on children with additional disabilities, but this population is quite diverse and presents unique challenges. Literature suggests that 30-40% of children with sensorineural hearing loss have an additional deficiency[15,18].

Other studies suggest that multidisciplinarity is able to add to the intervention context several benefits. Thus, it results in a rehabilitation program with language therapy and psychomotor exercises in patients with changes in motor and speech development, which emphasizes the importance of early diagnosis and a multidisciplinary team [4,17,19].

Due to the motor limitations of the child, the assistance of a professional physiotherapist contributed to expand his functions, as well as the modification of neurological and motor difficulties, including cognitive and communication aspects.

CONCLUSION

The development of auditory skills and motor development, demonstrated in the IT-MAIS and DCDQ-Brazil test results, respectively, used as parameters of the therapeutic evolution of this child with diagnosis of the Dandy-Walker Variant, showed that therapeutic rehabilitation, performed with both support and a multiprofessional team, can be applied satisfactorily in the management of cases where deafness does not appear as the only associated factor.

In addition, it suggests that a cochlear implant can be an efficient resource for acquiring oral language and reaching complex stages related to hearing abilities, despite the peculiar difficulties caused by the syndrome, it promotes access to sounds, minimizes auditory sensory deprivation, favors interaction with the environment and undeniable quality of life for the child and his family.

When analyzing the specific case of this patient, one can consider the scope of positive aspects in relation to the benefits obtained with the device.

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