

COMMUNICATION ASPECTS IN WERDNING-HOFFMAN SYNDROME: A CASE REPORT

Aspectos da comunicação na síndrome de Werdning-Hoffman: estudo de caso clínico

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ABSTRACT

Werdnig-Hoffman Syndrome (WHS) is a hereditary neuromuscular disorder characterized by progressive muscle weakness and atrophy, which prevents motor skills development. Mental development is preserved, vivacity and intelligence stand out in contrast to poor motor activity. The subject affected by this syndrome, has physical limitations for effective interaction, which can cause communication problems. The purpose of this study is to analyze the communicative possibilities of a child with Werdnig-Hoffman Syndrome during the speech and language therapeutic process. It is a case study of four years old female child, female, attended speech and language therapy service in a multiple disabilities rehabilitation center, from August 2013 to April 2014. The clinical assessment showed that the patient communicated by facial expressions and rare vocalizations. During the therapeutic process, the interaction was dialogically sustained through responses to yes/no questions: raising the eyebrow concomitant to eyes wider open for a "yes" and eyebrow contraction for "no", with light denial head movement. Though predominantly silent and motionless, the patient expressed her subjectivity by means of a communicative conduct that although incipient indicated symbolic activity related to integration and interpretation of reality.

KEYWORDS: Communication; Case Studies; Speech, Language and Hearing Sciences

■ INTRODUCTION

Werdnig-Hoffman Syndrome (WHS) is a hereditary severe neuromuscular disease with early and rapid onset (before six months of age), characterized by progressive muscle atrophy and weakness that prevents the development of motor skills, due to severe motor and respiratory harm, associated to important symmetric hypotonia involving the muscles of the hip, upper back, neck, upper and lower members. Body extremities become flaccid, the lower extremities in external rotation and the upper ones in abduction. Another characteristic is weak crying and non-effective coughing¹⁻³.

In this clinical condition, mental development is preserved and vivacity and intelligence stand out, in contrast to these patients' poor motor activity. Life expectation is short, usually around two or three years of age, due to the progressive paralysis of the respiratory muscles⁴⁻⁶.

Regarding the treatment of WHS, there are extremely important measures that should be adopted in order to allow for a multi-professional approach, involving mainly Physical Therapy, Nursing, Speech-Language Pathology and Audiology and Occupational Therapy, in addition to constant medical follow-up, thus favoring a reduction of hospital stays and improvement in the subject's quality of life¹. Patients depend on technological and/or pharmaceutical devices that are pivotal for their survival, such as mechanical ventilation, instruments that will aid in nutrition, breathing and drug administration. Due to the illness' chronic characteristic and to the dependency on mechanical

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ventilation, patients may receive necessary care from part of the healthcare team at home^{7,8}.

Due to these characteristics and the physical limitations from the early months of life, the subject with WHS has difficulties in interacting with others that may cause communication problems and, therefore, demands early Speech-Language Pathology intervention. Although the publications regarding Speech-Language Pathology and Audiology in this field are few and many times without access to the themes, there are studies in Medicine, Physical Therapy, Occupational Therapy and Education that approach the interaction and/or communication of these children⁹.

Supplementary and/or alternative communication (SAC) in these cases is also a possibility that can aid in the process of communication. However, some of these children, even in the absence of oral speech, do not adapt to these communication-aiding procedures and prefer to use facial expressions and rudimentary sounds to express themselves.

About the silent conduct, it is worth mentioning Orlandi who states that silence is founding (and not necessarily silencing) since from it, words may emerge. Thus, even if the subject does not use oral language to express himself, his silence may be filled with meanings and "seen as a means of communication, not as a means of non-communication"^{10,11}.

Based on these considerations, the purpose of this study is to analyze the communicative possibilities of a child with Werdnig-Hoffman Syndrome throughout the Speech-Language Therapy process.

■ CASE PRESENTATION

This study was approved by the Ethics Committee of the Pontifical Catholic University of São Paulo, under number 1.076.665/2014.

History

This is a case study of a child with WHS, undergoing speech-language therapy sessions in a rehabilitation center for multiple disabilities in São Paulo, between August 2013 and April 2014.

Four year-old, female patient, only child of a young couple who had a desired and planned pregnancy. She was born of natural birth, weighing 3.780 kg and 49 cm, was breastfed and released from the hospital with her mother after three days of birth.

At four months, the mother complained to the pediatrician that the child did not support her cervical spine, which was not considered a relevant clinical sign. At seven months, the child manifested symptoms of a strong flu, with difficulties to eat and breathe. She was admitted to the hospital but her

respiratory condition did not improve, and thus she was sedated and placed on a breathing tube. During this period, the WHS diagnosis was confirmed by the neurologist. While at the hospital (three months) she underwent a tracheostomy and gastrostomy. Since then, there have been motor and muscular harm manifestations. Upon hospital discharge, she moved only her right hand, her toes and breathed through mechanical ventilation, and thus required a home care support team composed by a nursing professional (24/7), three physical therapy sessions and one Speech-Language pathology session per day and a weekly doctor's and nurse's visitations for follow-ups.

At two years of age she started to attend an early intervention group in a multiple disability facility. She stayed in this group during the year of 2012 and, in 2013 she began special schooling in the same institution. In August 2013, at 3 years and 5 months of age she began individual speech-language therapy sessions. When seeking for this type of care, her mother reported that, even though her daughter communicated and was understood by people close to her daily activities, she wished for her to develop oral speech while she could also use SAC.

Speech-Language Therapy Process

The patient attended weekly sessions, of 30 minutes each. For this study, nine months of speech-language therapy were comprised, from August 2013 to April 2014. She used an adapted wheelchair (inclined, with a support to keep her cervical spine and head erect), a respirator attached to the tracheotomy and was monitored by an oximeter to permanently verify heartbeats and O₂ saturation. There was a table attached to the wheelchair in order to make activities easier to accomplish.

Orofacial motricity assessment was made through clinical observation and included intraoral and extraoral morphological observation and analysis, assessment of orofacial mobility, muscle tone, and sensitivity and of the oral functions of chewing, swallowing, breathing and speaking. There was hypotonia as well as reduced mobility and sensitivity (cheeks, lips, tongue and chin), small mouth opening, reduced tongue mobility, tongue fasciculation, excess saliva and abundant drooling. Chewing and swallowing were absent. Sometimes, she manifested a wish to try some kind of food (in soft consistency) that was put in her mouth. With the food inside her mouth she made small tongue movements and more saliva was produced than usual. Therefore, she had a few seconds to taste the food and then it was removed with gauze.

Regarding the language assessment, conducted through clinical observation in a dialogic and playful

context, it was verified that the patient communicated through facial expressions and rare nasalized vocalizations. She paid attention to the environment, always observing what was happening around her. Her adequate verbal comprehension, reacting in a coherent manner according to context stood out. She established visual contact with the therapist, accepting physical contact and returning with a slight smile.

Throughout the therapy process, interaction was sustained dialogically through the therapist's questions that were answered affirmatively or negatively by the patient. For "yes" she raised her eyebrow along with greater opening of her eyes and a slight smile. For "no" she contracted her eyebrow and made a slight denial movement with her head. Many times, vocalizations were not understood by the therapist, but her mother and/or nursing assistant who were there during the sessions would clarify the content.

Inicialmente, nas primeiras 06 (seis) semanas de terapia fonoaudiológica, foram realizadas tentativas de utilização da CSA, com computador e *Software Board Maker*, para confecção da prancha. A paciente realizava a escolha das figuras utilizando os recursos de "sim" e "não" descritos e, com o direcionamento do olhar, localizava-as na prancha com facilidade.

In spite of the encouragement from the therapist, Family members and caregiver, the patient refused to use this device, insisting in facial expressions and very few vocalizations as means of communication, through which she maintained rudimentary dialogues, until she was fully understood. For instance, when she wanted to express something and her vocalizations were not understood by the therapist, she quickly looked to her caregiver or mother, who interpreted their meaning most of the time.

RESULTS

After two months of speech-language therapy, it became evident that the SAC resource was not effective, at that moment, from a functional communication point of view. The therapist also gradually became an interlocutor, attributing possible meanings to the patient's vocalizations (always in a low voice with nasal and imprecise articulation) associated to the others expressive resources, i.e. facial expressions, slight head movements and silent pauses, that could be interpreted in spite of her nearly inert body.

In this specific context, a moment stands out when the child came to the session with an angry/cross facial expression, accompanied by the new

nursing assistant who was still adjusting and, therefore, did not understand her most times. This fact seemed to justify her irritability.

During the following session, the same professional reported that the girl taught her to use the iPad, in order to show her favorite software. Using her looks (expressing "yes" or "no") she directed the caregiver's gestures. This effectiveness in communication worked in several situations, as shown in the dialogue contexts below, where the patient and the therapist are sitting face to face.

Situation 1:

Therapist: was your birthday last week?

Patient: (raises her eyebrow alongside a greater eye opening and a slight smile)

Therapist: Oh, how nice! Was there a party at your house?

Patient: Answers "yes" (raises her eyebrow alongside a greater eye opening and a slight smile)

Therapist: was there a party here at school too?

Patient: Answers "yes"

Therapist: Oh, I got it! There was no party at school...

Situation 2:

Patient: (sstares at the message board on the wall)

Therapist: - What are you looking at, the letters?

Patient: Answers "yes".

Therapist: - Is your letter in this name?

Patient: Answers "yes".

Therapist: - And what is the letter of your name, this one? (showing a random letter)

Patient: Answers "no" (eyebrow contraction and slight denial head movement)

Therapist: - No? Is it this one? (pointing to the correct first letter of her name)

Patient: Answers "sim".

Therapist: - Oh, good. Let's write your name, cut it and put it up on the board too?

Patient: Answers "yes".

Situation 3:

Therapist: Now I will choose a game here for us to play together.

(therapist takes the tablet and clicks on the button that shows several application icons and the Picture of a baby on the background)

Patient: (starts to blink her eyes and raise her eyebrow)

Therapist: Do you want to show me a game that you like?

Patient: Answers "no".

Therapist: Then what do you want? When the tablet lit up you started to blink...

Patient: Answers "yes".

Therapist: is it the picture of the baby?

Patient: Answers "yes".

Therapist: oh, it's the Picture! Do you like the baby? Do you want to know whose picture this is?

Patient: Answers "yes" and vocalizes [ã u]

Therapist: You want to know who is this baby, don't you?

Patient: Answers "yes".

Therapist: It's my niece!

Paciente: (smiles)

The fragments of speech-language therapy sessions mentioned above show the effectiveness of the communication in the therapist/patient partnership, which also happens in the child's family and educational contexts.

It is known that motor and speech impediments are severe, but they do not prevent communication, the symbolic frame through which this child turns herself into a subject. She is, in fact and by right, an "I" to someone else, even though her relationship and social circles are small, restricted almost fully to the family and institutional environments where she receives multi-professional educational and health care.

■ DISCUSSION

The results show that in the cases of patients suffering from severe organic conditions associated to the absence of oral speech, as in HWS, the Speech-Language Pathologist must assume that language, as an intersemiotic activity, goes beyond the domain of speech. Thus, the interaction among subjects is not determined exclusively by a common code that is shared¹².

According to Dolto¹³, acting is language. In other words, language is the desire to find others, and

thus to establish communication and be accepted by them. Thus, body is language when it produces meaning, keeping and feeding this desire¹⁴. In this direction, the child studied in this research, in the condition of desiring subject and in spite of the organic deprivations, is constituted and recognized as such by her interlocutors.

The biological and psychic singularity of this child tells of her experience in language and of how it marks the forms of communication and its subjective constitution¹⁵. The roles played by the parents and the ways they turn the child's experience into words will trace her psychism¹⁶. In this study, the parent's acceptance of this child, how different she is and the deprivations she deals with, was remarkable, and what seems fundamental is her ability to circulate and sustain her communicative relationships.

In this perspective, the process of constituting oneself includes the realization of parental expectations, and may become difficult when the real baby does not correspond to the parents' ideals due to organic conditions. The birth of a baby with an organic deficit that is congenital or acquired very early may derail the parents' eyes from the subject/child to the illness or disability, which holds a strong potential to harm the child's subjective constitution. In the case that was studied this seems to not have happened¹⁷.

■ CONCLUSION

In the presented case there was acceptance by the family and the therapist, allowing the child, in spite of the severity of her clinical condition, to circulate through language, affirming herself as a subject.

Even though predominantly silent and with an inert body, the patient expresses her subjectivity through communicative conducts that, albeit precarious, show symbolic activity in the insertion and interpretation of reality.

RESUMO

A Síndrome de Werdnig-Hoffman é uma doença neuromuscular hereditária, caracterizada pela atrofia e fraqueza muscular progressiva, que inviabiliza o desenvolvimento de habilidades motoras. O desenvolvimento mental encontra-se preservado, vivacidade e inteligência destacam-se, em contraste à precária atividade motora. O sujeito acometido por essa síndrome tem limitações físicas para a efetiva interação com o outro, o que pode acarretar problemas de comunicação. Este estudo tem como objetivo analisar as possibilidades comunicativas de uma criança com Síndrome de Werdnig-Hoffman no decorrer do processo terapêutico fonoaudiológico. É um estudo de caso de uma criança com quatro anos de idade, gênero feminino, filha única (de casal jovem), atendida em terapia fonoaudiológica em centro de reabilitação de deficiências múltiplas no período de agosto de 2013 a abril de 2014. Na avaliação fonoaudiológica observou-se que a paciente comunica-se por expressões faciais e raras vocalizações. No decorrer do processo terapêutico, a interação foi sustentada dialogicamente por meio de perguntas da fonoaudióloga respondidas afirmativa ou negativamente pela paciente. Para “sim”, elevava a sobrancelha concomitante à maior abertura dos olhos e um discreto sorriso. Para “não”, contraía a sobrancelha e realizava leve movimento de negação com a cabeça. Embora predominantemente silente e corporalmente inerte, a paciente expressa sua subjetividade por meio de condutas comunicativas que, mesmo precárias, indicam atividade simbólica na inserção e interpretação da realidade.

DESCRITORES: Comunicação; Estudos de Caso; Fonoaudiologia

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