Case Report

Relato de Caso

Trajectory and outcomes of speech-language therapy in the Prader-Willi Syndrome (PWS): case report

Percurso e resultados de terapia fonoaudiológica na Síndrome de Prader-Willi (SPW): relato de caso

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ABSTRACT

The aim of this study was to describe the trajectory and the outcomes of speech-language therapy in Prader-Willi syndrome through a longitudinal study of the case of an 8-year-old boy, along four years of speech-language therapy follow-up. The therapy sessions were filmed and documental analysis of information from the child's records regarding anamnesis, evaluation and speech-language therapy reports and multidisciplinary evaluations were carried out. The child presented typical characteristics of Prader-Willi syndrome, such as obesity, hyperfagia, anxiety, behavioral problems and self aggression episodes. Speech-language pathology evaluation showed orofacial hypotony, sialorrhea, hypernasal voice, cognitive deficits, oral comprehension difficulties, communication using gestures and unintelligible isolated words. Initially, speech-language therapy had the aim to promote the language development emphasizing social interaction through recreational activities. With the evolution of the case, the main focus became the development of conversation and narrative abilities. It were observed improvements in attention, symbolic play, social contact and behavior. Moreover, there was an increase in vocabulary, and evolution in oral comprehension and the development of narrative abilities. Hence, speech-language pathology intervention in the case described was effective in different linguistic levels, regarding phonological, syntactic, lexical and pragmatic abilities.

Descritores

Síndrome de Prader-Willi/genética
Transtornos de linguagem
Obesidade/etiologia
Ansiedade/etiologia
Hiperfagia/etiologia
Transtorno do comportamento/etiologia
Estudos de intervenção
Relatos de casos

RESUMO

O objetivo deste estudo foi descrever o percurso e os resultados da terapia fonoaudiológica na síndrome de Prader-Willi, por meio do estudo longitudinal do caso de uma criança de 8 anos de idade, do gênero masculino, ao longo de quatro anos de terapia fonoaudiológica em uma clínica-escola. Foram realizadas filmagens de sessões de terapia e análise documental de informações dos prontuários referentes à anamnese, avaliação e relatórios terapêuticos fonoaudiológicos e avaliações multidisciplinares. A criança apresentou características típicas da síndrome de Prader-Willi como obesidade, hiperfagia, ansiedade, problemas de comportamento e auto-agressões. Em avaliação fonoaudiológica foram observados hipotonia orofacial, sialorréia, voz hipernasal, alterações cognitivas, dificuldades de compreensão oral, comunicação por meio de gestos e produção de palavras isoladas ininteligíveis. Inicialmente, a terapia fonoaudiológica teve o objetivo principal de promover o desenvolvimento da linguagem com ênfase na interação social por meio de atividades lúdicas. Com a evolução do caso o direcionamento principal passou a ser o desenvolvimento de habilidades conversacionais e narrativas. Foram observadas evoluções quanto à manutenção da atenção, brincadeira simbólica, contato social e comportamento. Além disso, houve aumento do vocabulário, evolução quanto à compreensão oral e desenvolvimento de habilidades narrativas. Dessa maneira, a intervenção fonoaudiológica em caso de síndrome de Prader-Willi foi eficaz em diferentes níveis, no que se refere às habilidades fonológicas, sintáticas, lexicais e pragmáticas da linguagem.

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INTRODUCTION

The scientific literature describes clinical heterogeneity and variable possibilities of intervention, for people with Prade-Willi syndrome (PWS). This study highlights Speech-Language Pathology as the specific area which must be considered in search of better outcomes, depending on the clinical phenotype of each case.

PWS was described in 1956, as a rare behavioral neurologic disorder, of genetic origin[1,2] which reaches from 1:10.000 to 1:15.000 births[3]. The etiolology refers to the absence of paternal genes, an abnormality of the long arm of chromosome 15 paternal (15q11q13) or, still more rarely, translocations, maternal disomy of chromosome 15, or abnormalities of the chromosomal *imprinting*[3].The diagnosis is based on clinical criteria and it may be definitely established by the genetic analysis.

PWS is characterized by neonatal hypotonia in 87.95 of the cases; 88.4% facial characteristics, 98.9% late neurological, psychological and motor development, 84.4% hyperphagia, 66.7% excessive gain of weight, 86.7% behavioral problems, 75.65 sleep apnea, 87.8% small and fat hands, 80% speech-language deviation, 88.9% dense and viscous saliva, 83.3% skin lesions caused by self-aggression, among other characteristics[4]. Psychiatric disorders have also been described[1,2,5,6] as obsessive-compulsive disorder, compulsive eating habit[6], psychotic symptoms[1] depression and aggressiveness, when the required food is denied[6].

Some authors have referred to the presence of repetitive behaviors, similar to those presented in cases of the autistic spectrum disorders[7], symptoms of attention deficit and hyperactivity, difficulties concerning daily life abilities and socialization[1]. It was also observed intellectual deficiency, ranging from moderate to severe[1,8] , but most of the subjects presented moderate level[8]

Concerning the aspects of speech-language pathology, several alterations were mentioned[1,9-11] and considerable variability on the severity of speech and language disturbance[1,6,10]. It was observed oral and facial hypotonia[1,6], sialorrhea[6], voice[1] and speech articulation impairment[1], restrict morphological and syntactical abilities and lexical and pragmatic difficulties[10].

Nonetheless, the previous intervention in the area of Speech-Language Pathology is taken as determinant factor for better prognosis of subjects with PWS[1]. Researches emphasize the impairment on the speech-language development and speech fluency[11], reducing the studies about the Speech-Language Pathology intervention, specially directed to lexical and pragmatic aspects of the language. The aim of this work was to describe the trajectory and the outcomes of the Speech-Language Pathology concerning Prader-Willi syndrome, through a longitudinal study of one case.

CLINICAL CASE

The present study was authorized by Free and Informed Consent Term elaborated for the specific purpose of this research, and was approved by the Ethics Committee of Research of Universidade Estadual Paulista (UNESP), under process 1417/2009.

This work is based on a clinical case study, referring to one 8-year-old male child, who was diagnosed as Prader-Willi syndrome carrier. Since data collection and data analysis were realized, a longitudinal study during and a four-year follow-up including Speech-Language Pathology intervention were carried out. Hence, filming sessions of language therapy were done, with a six-month-interval, totaling eight sessions, besides documental analysis from the register form referring to anamnese, evaluation and Speech-Language Pathology therapeutic reports, exams and multidisciplinary evaluation.

Data from Language-Speech Pathology anamnesis

When the child was 4 years old, he was referred to the clinic-school, and was initially referred to the psychology sector, for presenting excessive anxiety, biting his hands, hyperphagia and perturbation. After the psychological evaluation, he was referred to the Speech-Language Pathology sector, for demonstrating delayed speech development.

During the Speech-Language Pathology anamnesis, the mother informed that she had undergone all neonatal examinations and the child was born within the estimated period, from natural delivery, with 3.2 kg and 45 cm, without any complications. The mother informed that the baby "almost did not speak", presented a very disturbed behavior and did not know how to use objects during play time. He was able to sit without any support when was 1-year-old and started walking at 2. Concerning language development, the mother reported that the child emitted the first sounds at 4 months of age, and the first words at 1 year and a half. At 3 years of age, he was able to understand simple and complex commands and expressed himself, by saying isolated words and indicative gestures.

The child was also followed-up by an obesity support group, which included a dietitian, a speech-language pathologist, a cardiologist, a neurologist and an endocrinologist, due to feeding disorders.

Evaluation data

Clinical neurological and genetic evaluation

When the child was 5 years old, he underwent a clinical neurological and genetic evaluation and the following problems were observed: perturbation, food obsession, convulsive crises and absence, even taking anticonvulsive medicine. Concerning physical aspects, it was observed obesity and small injured hands due to self aggression. At that time, the child underwent an encephalogram, received higher doses of anti depressive and anti convulsive medication and was diagnosed with intellectual disability. Moreover, he was clinically diagnosed with Prade-Willi syndrome.

Psychiatric evaluation

The psychiatric evaluation was realized in group, with a responsible psychiatrist, a psychologist and three speech-language pathologists, authors of this work. The child pre-

sented neurological, psychological and motor delay, anxiety symptoms, hyperphagia, obesity, skin lesions due to self aggression. Besides that, it was applied the Vineland Adaptive Behavior Scale[12], which evaluated the Development Quotient of the subject. The child scored 26 for communication, 24 for social skills and 43 for daily life activities, scoring 35 for Total Development Quotient, which is characterized as moderate mental retardation. During the psychiatric evaluation, clinical examinations were carried out and signs and symptoms were observed, which confirmed the diagnoses of intellectual disability and Prader-Willi syndrome.

Otorhinolaryngological evaluation

During the therapeutic process, the child went through two otorhinolaryngological evaluations at the clinical school; it was realized a clinical otological inspection and alterations were not observed.

Hearing evaluation

Hearing evaluations were done when the child was 4 years old and then when he was 8 years of age, which showed hearing threshold according to normal pattern.

Clinical speech-language pathology evaluation

The speech-language pathology evaluation comprised the analysis of aspects of oral motricity, voice, hearing and speech. These aspects were evaluated as follows: analysis of spontaneous and directed language sample, realized by the observation of behavior and employment of vocabulary test ABFW[13], which investigated the lexical performance of the child. It was also evaluated the ludic activity through behavioral observation, in order to verify the type of action and manipulation of objects and interaction with the speech-language pathologist and the mother. Besides that, in order to complement the evaluation, were taken into consideration the data found by the employment of the Vineland Adaptive Behavior Scale, described previously, applied in group with a psychiatrist, a psychologist and a speech-language pathologist. The evaluation of the oral and facial motricity and the vocal aspects was realized by the myofunctional evaluation and passive vocal, due to the lack of comprehension and collaboration of the patient, realized through anthroscopy or visual and hearing evaluation, followed by complementary evaluation with oral inspection and cervical auscultation.

It was observed a reduced period of hearing and visual attention, becoming necessary the introduction of different sonorous and visual stimuli in short periods of time in order to keep the child involved in ludic activities. The physiological, lexical and pragmatic aspects were evaluated from the analysis of the spontaneous and directed language sample, realized through the behavioral observation. Additionally, the ludic activity was evaluated through behavioral observation, to verify the type of action and manipulation of objects and interaction during individual sessions, with the speech-language pathologist, the mother and in group sessions with other children.

The child presented interaction difficulties, cried without any apparent reason, used indicative and representative gestures, followed by isolated words like: “no” and “come here” to communicate and non systematic answers when asked. During the group sessions with other children, the child demonstrated interaction difficulties, indifference and crying.

The child demonstrated intention of communicating, through facial expression, indicative gestures and some vocalizations, but generally to express protest in relation to the activities proposed by the speech-language pathologist and intention to be isolated. During the ludic activities, he refused to play and to use objects functionally, considering that his actions and the manner of manipulation of objects, were not compatible to his age. In relation to oral emission, he demonstrated difficulties on the oral and articulatory praxis, did not name objects or pictures during tests of names and ludic activities. The oral reception was below the average for his age, being able to understand just simple verbal commands of daily vocabulary.

To complement the evaluation, it was applied the vocabulary test, ABFW[13], and it was detected a lexical repertoire below his chronological age. The patient did not answer most of the questions, not even by gestures, and made few substitutions, which were characterized as non understandable and co-hyponym. For the evaluation of oral and facial motricity and vocal aspects, it was observed oral and facial hipotony, tongue protusion, sialorrhea and hypernasal voice.

In such case, the child presented alterations referring to speech and language, syntactic, semantic and pragmatic aspects of the language and according to the speech-language pathology diagnosis, he presented language disorder.

Methods and outcomes of speech-language pathology intervention

The speech-language pathology treatment referring to the studied case lasted four years, through individual 50 minute-sessions realized twice a week. Priority was given to the interaction between the pathologist and the patient, in order to promote the cognitive development, socialization and the communication of the child, based on the pragmatic perspective. The main objective was to promote the structure of the language and consequently to increase the occurrence of intentional communicative behaviors; favoring the comprehension and the functional use of the language; besides offering orientations to the mother and to the school, in order to contribute for the development of the child.

In the beginning of the therapeutic process, the child refused to take part on the activities, presented low communicative attention, attention alterations, aggressiveness, social isolation, crying sometimes without any apparent reason and at other moments, expressing protest in relation to the activities proposed by the pathologist, restrict interactions and non connected and non understandable words. The therapy was conducted through ludic activity, trying to highlight the communicative abilities of the child and to favor the dialogical activity, including activities and games, to propitiate the interaction and help the child to understand the commands and rules of the game.

At 6 years of age, the sessions began to include activities which aimed to develop narrative, jointly to the work compri-

sing hearing attention and social interaction. After three years of speech-language pathology therapy, the child improved his conduct and interaction with the pathologist, and showed better comprehension towards simple and complex commands. The child also started dialogic activities, but did not respect the game rules and presented infantile and perseverant behavior. He showed improvement towards syntactic structure, emitting complex phrases several times, considering that sometimes it was not possible to understand, just comprehend within the context. The child was more involved in some proposed activities, presenting preferences to play with dolls, to play as a hair stylist, talk on the phone and shopping at the supermarket.

During this period, at 7 years old, some difficulties for oral expression were observed, due to the praxical alteration. But he presented complex syntactic structure and retold parts of stories. During the sessions, the child showed some irritability and aggressiveness when was not understood by the pathologist. During the same period, he started to begin some turns and to keep the theme of the conversation during a specific period of time.

During the last year of speech-language pathology therapy, the child was 8 years old, weighed 67.6 kg and was 1.46 m tall. He attended common school and resource room, did not recognize graphemes, did not take part on the pedagogical activities of the school, according to the report of the responsible teacher. He was very anxious, and demonstrated moments of self-aggression.

It was possible to verify the intention of communicating, initiative towards dialogic activity, with some difficulty in maintaining the theme, use of gestures and oral communication, with non understandable speech, due to voice apraxia and hypernasal voice. It was observed agitated behavior, impaired logical reasoning and impairment on the hearing and visual perception processes. The child understood the simple instructions, but presented difficulty to understand complex commands, characterizing the impairment towards the ability of receiving and expressing oral language.

In order to check the evolution of the child's vocabulary, the vocabulary test ABFW[13] was carried out every year, and it was possible to observe performance improvement. However, on the last evaluation, it was possible to observe many substitution processes, characterized by unintelligible segments, phonetically expressive words and co-hyponyms.

After four years of speech-language pathology intervention, it was proposed to continue the therapeutic process emphasizing the development of conversational and narrative abilities, along with attention and social interaction, considering that the child obtained a level of development which allows the employment of such aspects which are the main difficulties that the child presented after the period under speech-language pathology therapy.

DISCUSSION

The present study allowed the verification of several characteristics described in the literature, concerning the child in study, referring to Prader-Willi syndrome, such as: hyperphagia, excessive weight gain, behavioral problems, intellectual deficiency[8] psychiatric disorder [1,2,5,6] compulsive eating habit[6].

Data obtained from the clinical speech-language pathology evaluation have also corroborated for the findings from other researches, as the child presented oral and facial hypotony[1,6], sialorrhea[6], voice impairment[1], and speech articulation[1,14] and restrict morphological and syntactic abilities[10], which turned his speech non comprehensive[14]. It was possible to observe some lexical[10,14] and pragmatic[10] difficulties for maintaining the conversation topics[6]and self aggressiveness[5].

As for the speech-language pathology therapy, it is suggested that, decurrent from the clinical heterogeneity described by the literature[1] and the data found in this study, the speech-language pathology prognosis of subjects with PWS is very variable. Referring to this case studied, it was observed better performance on the communicative abilities as: increased vocabulary, initiatives and change of turn on the dialogue, maintenance of the theme and improvement on the conduct towards the pathologist.

These findings, highlight the evolution in relation to lexical and pragmatic abilities, and advance into oral language of the child, in agreement with declarations of other authors about the importance of a study focusing such aspects[1,9]. The child has also demonstrated advance in speech comprehension, but kept on presenting alterations towards this aspect, as well as for the finding in researches which reported the existence of articulation disorders, even after intervention[9].

Thus, it is fundamental to include the speech-language pathologist when previous intervention are realized by a multidisciplinary team, as many authors agree[1,6,9,11]. However, it is necessary to consider, that besides the speech-language pathology therapeutic process, other variances are involved in the communication process of the child in study, as intellectual deficiency, anxiety, convulsive episodes and absence, besides the familiar and scholastic dynamics in which he is inserted.

Studies which contribute for the characterization of the clinical phenotype of the PWS are significant, as it was highlighted in literature[15], the genetic exam is not always available at daily medical practice, so the clinical criteria become very useful. Besides, due to the scarcity of this disorder, more complex and systematic studies on behavioral interventions are still very scarce[1]. In Brazil, it is observed the scarcity of studies focusing aspects of evaluation on mental health and behavioral intervention towards PWS[5]. Hence, more longitudinal researches are necessary, concerning the clinical manifestation and speech-language pathology therapy towards this syndrome.

FINAL COMMENTS

Based on this study, it was possible to present the characteristics of a child with Prader-Wili syndrome, in relation to his physical, cognitive and behavioral development and specially the development of speech and language.

Most studies about PWS, focus the description of the genetic and phenotypic alterations, present on the studied individuals and treatment concerning the alimentary disorders of the condition, so the findings described here may help, according to the

technical knowledge of the speech-language pathologists about the manifestations, therapeutic processes and speech-language pathology prognosis referring to PWS.

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