

# Language in Treacher Collins Syndrome: a dialogical analysis

## A linguagem na Síndrome de Treacher Collins: uma análise dialógica

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#### **ABSTRACT**

The Treacher Collins Syndrome or Mandibulofacial dysostosis is due to genetic mutations and characterized by craniofacial malformations. Children with this syndrome may present cognitive, linguistic and psychomotor difficulties. There are few publications that discuss the complexity of its therapeutic aspects, especially those focused on language clinical evolution. The present study aims to analyze a speech - language clinical work on oral language of a boy who has this syndrome, considering the dialogical nature of language. This is a longitudinal and prospective case study, carried out in a university clinic located in the south of Brazil, during four years, from 2012 to 2016. Data were collected from weekly recordings of the patient interacting with his therapists, and also from his record files. The results indicate that this child presented oral language appropriation evolution. Despite his vocal production and phonemes articulation's difficulties, due to his craniofacial alterations that characterize this syndrome, the dialogical activities established between the child, his therapists and his family, caused gradual changes in his language use. Initially, he used gestures, facial mimics, pointing, which were understood only by people who were part of his daily life. Nowadays, he still uses gestures, but he also began to use oral language to participate in interactive practices, which indicates his autonomy to interact with other people.

**Keywords:** Mandibulofacial dysostosis; Clinical evolution; Language; Child development; Family relations

#### **RESUMO**

A Síndrome de Treacher Collins ou Disostose Mandibulofacial é decorrente de mutações genéticas e caracterizada por malformações craniofaciais. Crianças com essa síndrome podem apresentar dificuldades cognitivas, linguísticas e psicomotoras. São raras as publicações que discorrem sobre a complexidade de seus aspectos terapêuticos, especialmente, voltados à evolução clínica vinculada à linguagem. O presente estudo objetiva analisar o processo terapêutico voltado à oralidade de um menino com essa síndrome, considerando a natureza dialógica da linguagem. Trata-se de um estudo de caso longitudinal e prospectivo, realizado em uma clínica-escola de uma Universidade, situada no sul do Brasil, durante quatro anos, desde 2012 até 2016. Os dados foram coletados a partir de gravações semanais do paciente em interação com os seus terapeutas, sendo, também, considerados os registros arquivados em seu prontuário. Os resultados indicam que a criança apresentou evolução no que se refere à apropriação da linguagem oral. Apesar das dificuldades na produção vocal e na articulação de fonemas, decorrentes de alterações craniofaciais próprias da síndrome em questão, as atividades dialógicas estabelecidas entre o menino, seus terapeutas e sua família, propiciaram mudanças gradativas no seu posicionamento em relação ao outro e à linguagem. Inicialmente, ele fazia uso de gestos, mímicas faciais, apontamentos, os quais eram compreendidos apenas pelas pessoas que faziam parte do seu cotidiano. Atualmente, além dos recursos gestuais, ele passou a usar a oralidade para participar de práticas interativas, indicando mais autonomia para interagir com seus interlocutores.

**Palavras-chave:** Disostose mandibulofacial; Evolução clínica; Linguagem; Desenvolvimento infantil; Relações familiares

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#### **INTRODUCTION**

Treacher Collins Syndrome (TCS) is a dysfunction due to genetic mutations and characterized by craniofacial malformations. In 1900, it was diagnosed by the British ophthalmologist Treacher Collins, being named after him. However, in 1944, from the description performed by Franceschetti Klein, it was also adopted the term mandibulofacial dysostosis to refer to this syndrome<sup>(1)</sup>. Its incidence is around 1/25,000 and 1/70,000 of live births<sup>(2)</sup>.

The occurrence of genetic malformations, characteristic from the TCS, cause conditions that interfere negatively in the quality of life of people affected by this disorder. The most common signs and symptoms are: antimongoloid (downward slanting) palpebral fissures, lower-eyelid coloboma, sparse eyelashes, malar hypoplasia and micrognathia<sup>(3)</sup>. Ear deformities are also observed in the outer ear canal and middle ear<sup>(4)</sup>. Dental deformities are severe, causing facial nerve anteversion, open bite and contraction of the orbicular and mentonian muscles when the patient tries to close his/her mouth<sup>(5)</sup>.

A small lower jaw, associated with a posteriorly displaced tongue may cause breathing conditions, in addition to mobility disorders in the organs related to feeding and speech-sound production<sup>(5)</sup>. Therefore, although the observed malformations at birth do not develop, they frequently bring about conditions that harm the global development of the subjects with TCS, impairing their psychomotor, cognitive and linguistic performance<sup>(6)</sup>.

Due to the unique complexity of the craniofacial deformities caused by the TCS, and its outcomes, patients' treatment, affected by mandibulofacial dysostosis, must be multidisciplinar, comprising diverse knowledge fields, such as orthodontics, otorhinolaryngology, ophthalmology, speech-language therapy, neurology, psychology, among others, depending on the dynamics of each case<sup>(7,8)</sup>. However, despite the interest of several specialities, scientific publications on the clinical aspects of that syndrome are scarce, mainly the ones addressing therapeutic follow-up<sup>(5)</sup>.

From a speech-language pathological view, the scarce literature addressing the theme points to significant breathing, swallowing problems, and a condition of high and narrow palate, occasionally a cleft palate, are issues that the area must assess and follow-up<sup>(9)</sup>. As for hearing, in 30% to 50% of the cases, the affected subjects have severe bilateral conductive hearing loss due to the malformation of the three small bones in the middle ear, or stenosis of the outer ear canal<sup>(4)</sup>.

Concerning the orality in patients with TCS, micrognathia and posteriorly displaced tongue are elicited as factors which impair sound articulation and oronasal ressonance, causing speech unintelligibility<sup>(10)</sup>. The oral deformities cause discursive problems, interferring negatively in the social participation of these patients, pointing to the need of a language-focusing follow-up. In the speech-language clinic based on a dialogical perspective<sup>(11)</sup>, subjects' organic issues are considered in light of the social relations that each one is inserted, in a unique way.

In this perspective, the speech-language clinic considers the social environment as constituting the subjects' cognitive dynamics, to the extent that language organizes the roles that they take on in several contexts that they participate, discursively guiding their speech<sup>(12)</sup>. The therapeutic follow-up, grounded in the dialogism includes, in its approach, interactive verbal and non-verbal aspects, broadening their possibilities of meaning and linguistic, discursive reorganization of patients with TCS.

Thus, considering the dialogical nature of the language, which does not exclude the organism from their social meanings<sup>(11)</sup>, the present study aims to analyze the therapeutic process, oriented to the oral language of a boy with TCS.

#### **FEATURING THE CLINICAL CASE**

The current study was approved by the Ethics Board on Research, register 8910/11, and complied with the criteria of Resolution 196/96, Brazilian Council of Health. Research participant's legal guardians signed the Free Informed Consent. It is a prospective, longitudinal case study, elaborated from a clinical speech-language intervention of a child with Treacher Collins Syndrome, whose diagnosis was performed by his pediatrician. Fictitious names were given to the child and his family in order to keep their confidentiality: Marcos, the patient; Sofia, his mother; Dante, his father; and Mário, his brother.

Marcos has been treated at a teaching clinic from the graduation in Speech-Language Therapy of a University in Southern Brazil, weekly therapies, 40 minutes each. For data analysis, four years of speech-language treatment were considered, since the first contact of the child's parents with professionals and interns in Speech-Language Therapy in the middle of 2012, when Marcos was twenty-two months of age, until 2016, when the patient had already turned six years old.

For data collection, different aspects described in the patient's medical record were considered, such as the initial interview with his parents, speech-language assessment, daily records, semiannual reports, interdisciplinary contacts, feedback to the patient and his family. In addition, the interactive situations between Marcos and his therapists were videotaped in an SM-T116BU, 4.4 android version. It should be pointed that, each year, a different intern treated Marcos, once it was a teaching clinic.

Marcos is a boy who was born in a city in Southern Brazil in the second semester of 2010. At the beginning of his therapeutic process, his mother, Sofia, stated having been hard to deal with TCS. She commented that went through embarrassing situations when his son needed to leave home, due to his facial image.

She informed that, according to the doctors, his son did not have any life expectations due to the severe degree of the syndrome. Despite the traumatic experience, according to his mother, "Marcos is a wonderful child, worth any efforts... he's smart and understands very well what's going on around him." Sofia reported that, as soon as he was born, Marcos needed to be intubated urgently.

After birth, the child had three respiratory arrests and two cardiac arrests because, as a consequence of the Treacher Collins Syndrome, he had upper airways obstructions. Two hours and thirty minutes after his birth, his guardians' permission was requested for an emergency tracheotomy, which he uses until nowadays. His parents could see Marcos only 24 hours after his birth. He was admitted to the Intensive Care Unit (ICU) and stayed in hospital for 45 days. In the ICU, he was inserted a nasogastric tube for feeding, as he had difficulty in swallowing.

After leaving the hospital, Marcos needed to undergo a jaw surgery and a gastrostomy. At six months of age, he underwent another surgery for eyelid correction. Since then, he has already undergone several reconstructive and corrective surgeries. However, he still makes use of the tracheostomy tube to increase his respiratory possibilities, as well as the feeding tube.

Regarding the family dynamics, Marcos lives with his parents and Mario, his brother, who is two years older than him. His father is a state school teacher, and his mother, despite having quit working out in order to dedicate full time to his family, currently teaches Chemistry. Dante also takes care of Marcos, and the couple shares the housework. At home, Marcos plays with his brother and parents, actively participating in the family activities.

Marcos used to attend kindergarten, in a non-systematic way, once he often had to be hospitalized and undergo several surgeries. His parents understood the importance for him to interact with other children, and wanted him to stay at school. However, in 2014, there was a complaint, as he needed a nurse beside him to be frequently aspirated. Due to that, during the data collection, Marcos was a long time away from school and, therefore, without interacting with other children and teachers in the school setting.

Concerning Marcos' hearing, Sofia, in her first interview in 2012, explained that her son had intact middle ear, but he did not have ear drums and canals. In 2013, he had moderate hearing loss with bilateral mixed component, and made use of the BAHA hearing aids, presenting functional gain of 40dB.

In the Oral Motricity, in 2012, Marcos had difficulty in breathing and swallowing, did not have lip occlusion, and reduction of the upper airways flow. His cheeks and eyes were asymmetric, retrognathic jaw, and did not have movements to capture, prepare and chew food. Currently, Marcos has been followed up by two gastroenterologists and a nutritionist, and can swallow liquids, but in the past, he could not swallow even his own saliva.

In 2012, Marcos's voice was too low. In 2013, to intensify his vocal production, there was an attempt to use a defenestrated tracheostomy tube. However, it was a very unsuccessful attempt, as Marcos did not have physiological structure to use such a device, therefore, it had to be removed.

In relation to language, the main focus of this study, Sofia reported, at the beginning of the clinical process, Marcos could make himself understood among his family, using gestures, facial mimics, and lip and tongue movements. However, most of the time, listeners who were not part of his daily life, could not understand what Marcos meant by using such resources. At that initial moment, his mother stated that she wished Marcos could talk, disregarding his malformations. After 20 months, Marcos started to add sound to his speech, leading Sofia to point, in February of 2014, that "today he mimes and speaks with the gestures, as well."

As for his speech-language therapy, specifically centered on his language, it should be pointed out that, in his clinical process, his TCS-related organic impairments, as well as his relationship with therapists and family, in several enunciative situations, were taken into consideration. The therapeutic objectives were focused on Marcos's interactive possibilities. His gestures, facial mimics, pointing, vocal productions were meaningful throughout the therapeutic process, according to subsequent discussion.

#### **DISCUSSION**

The strategies used in the clinical treatment focused on activities considering the living practice of the language, thus, during the speech-therapy sessions, dialogues about the child's daily life were carried on, his tasks, his family relations, his leisure activities, his requests. Moreover, diversified playful activities - soccer match, plays related to the super heroes, drawings, painting, making clay artifacts, objects with clay—were built with the participants of the clinical practice, that is, between the child and his therapists.

In this sense, understanding that language is marked by the flow of dialogical activities taking place within actual enunciative situations, during the clinical session, Marcos was encouraged to use several semiotic resources, complying with his initiatives to draw, play, cry, finding the space to elaborate and organize the difficulties along his interaction with the other. Until mid-2015, he basically produced vowel sounds [a, e, E), at low intensity.

In spite of trying to use the augmentative alternative communication boards, Marcos did not want to use that strategy to communicate. At the end of 2015, he started to produce, in dialogical situations, sounds that followed the melody of songs that were sung to and by him. In 2016, the fact which called the attention was that despite his organic impairments, related to retrognathic jaw, tongue placed forward, reduction of the upper airways flow, difficulty in lip occlusion, Marcos could establish oral dialogues with his therapist.

In order to elicit the oral productions elaborated between the child and his therapist, four episodes of the speech-language therapeutic sessions, held in the first semester of 2016, were selected. These episodes, shown in tables, were transcribed with the pauses, intonations and stretches present in patient's and therapist's oral language, placed in turns organized in ascending numbers. However, despite Marcos's speech displays exchanges and distortions, the transcription was not performed literally, as such enunciations were interpreted by the therapist from the enunciative context, where interaction was produced. The use of the letter T stands for the therapist's participation, and the letter M indicates Marcos's speech. The conversational markers followed the indicators adopted in the Standard Urban Spoken Language Project/São Paulo State, Brazil (Projeto NURC/SP)<sup>(13)</sup>.

In Table 1, it is perceived that Marcos takes over as the subject in the enunciative situation. Initially, considering that the therapist could not understand his orality, he turns to graphic resources to indicate what he meant. In turns 03 and 05, he makes use of gestures to indicate an affirmative and the size of the animal in the photo, the theme of the established dialogue.

Later, in turn 07, Marcos starts to make use of the orality. He takes the initiative by questioning the therapist and report that, initially, he could not understand how the term used to designate the black color could be used to name a dog. That is, he seems to show his outrage while considering that colors do not suit to be proper nouns. When she perceived his outrage, the therapist explained the reason why she named her dog Black. Thus, only after her explanation, Marcos resigned himself, stated by the vowel stretching produced in turn 13.

In this dialogical excerpt, it is possible to observe Marcos making use of different resources in order to participate in the enunciative situation. He scribbles in a piece of paper, makes use of gestures and orality to carry on a dialogue with the therapist about a photo, interested in interacting with the other. Therefore, from a linguistic-discursive point of view, it is possible to state that this child keeps a semantic coherence to take part in a dialogue that puts him in a discursive production.

In Table 2, Marcos uses orality in a contextualized way to answer questions elaborated by the therapist, in turns 02 and 04, to agree with her, and also to disagree with her in turn 16. In addition, he makes effective use of facial mimics and gestures to participate in the conversation. From a dialogical standpoint, it can be stated that, although that child had started using orality from the end of 2015, he was already inserted in the discursive flow much before that.

In Table 2, evidenced above, he shows to be involved in a certain social organization which privileges the image of super heroes. He also unveils that he recognizes the physical setting

surrounding him to the extent that he can tell the therapist where she should write a super-hero name by using the adverb *here* coherently with the produced enunciation in turn 06.

In Table 3, it is observed that the child establishes verbal interaction, using the orality to make affirmatives or denials, in turn 13, to state his opinion. In turn 30, in addition to answering in a contextualized way, the question elaborated by the therapist, saying that he is tired, he builds a sentence with subject, verb and object, reporting that he wanted to go home, showing that he masters the Portuguese syntax. Therefore, besides answering sentences elaborated by the other, he can elaborate their own.

Table 1. The data in Table 1 were produced on 04/11/2016, while the therapist (T) shows Marcos a Picture of her dogs

Turn	Symbol	Transcription	Indicator/NURC
01	M	((M tries to ask his therapist something, she does not understand. He takes paper and starts to scribble, as if signing something)) ${}^{\prime}$	(( )) transcriber's description
02	Т	do you want to know my dogs' names?	? question
03	M	((nods))	(( )) transcriber's description
04	Т	Elvis	
05	M	((mimes it is big, pointing to the bigger dog in the picture))	(( )) transcriber's description
06	Т	Preta (Black)	
07	M	no:o the name	: vowel stretching
08	Т	Preta (Black)	
09	M	O NOME (THE NAME)	capital letter - stressed intonation
10	Т	but my dog's name is: Black	: vowel stretching
11	M	é? (Is it?)	? question
12	Т	because she is really black, we named her like that	
13	M	a:	: vowel stretching

M.'s age.: five years and six months

**Table 2.** The therapist (T) and Marcos are talking, on 05/30/2016, about super heroes, and she introduces some super-heroes stickers to build a panel with Marcos

Turn	Symbol	Transcription	Indicator/NURC
01	Т	I don't know who that isis that the widowva	pause
02	M	É (Yeah)	
03	Т	now, we can't remove the sticker	
04	M	não dá (we can't)	
05	Т	and where am i going to write the super heroes' names?	? question
06	M	aqui Ó: (here)((points to where the therapist should write))	capital letter – stressed intonation; : vowel stretching; (( )) transcriber's description
07	Т	that one is the	pause
08	M	Capitão América (Captain America)	
09	Т	Captain America	
10	M	that one is ((didn't remember the super hero's name))	pause;
			(()) transcriber's description
11	Т	and that one here is	pause
12	M	Thor	
13	Т	Ó: ((points to a super-hero))	capital letter - stressed intonation;
			: vowel stretching
			(( )) transcriber's description
14	M	é esse é o thor né (isthat's thorright)	pause
15	Т	i'm going to write here black widow	pause
16	M	não é (it's not)	pause
17	Т	who is it, then?	? question
18	M	((facial expression indicating that he doesn't know))	(( )) transcriber's description
19	Т	we have to do some research	pause

M.'s age: five years and seven months

Table 3. The therapist (T) and Marcos are putting a puzzle of super heroes together and painting it on 06/06/2016

Turn	Symbol	Transcription	Indicator/NURC
01	Т	which paint are you going to use?	? question
02	M	hum	pause
03	Т	let me take a bit of the excess	
04	M	pronto? (ready?)	? question
05	Т	readyare you going to paint it all in red?	? question
06	M	É (Yeah)	
07	Т	let me take the excess	pause
08	M	não precisa (not necessary)((while he paints, he tries to sing))	pause
		pronto (ready) ((difficult to understand his speech))	(( )) transcriber's description
09	Т	(black?)	? question
10	M	Não (No)	
11	Т	tell me the name of the color you want	
12	M	[aa]	[ ] phonetic transcription
13	Т	white?	? question
14	M	É (Yeah)	
15	Т	which super hero do you like best?	? question
16	M	Hulk, homem aranha preto (Hulk, black spider man)	
17	Т	Do you lend your brother your toys?	? question
18	M	não gosto (don't like)	
19	Т	doesn't he like to play?	? question
20	M	((unintelligible speech))	(( )) transcriber's description
21	Т	he likes to play with the black spider man do you want to keep on painting?	pause
			? question
22	M	((stops the activity and thinks))	(( )) transcriber's description
23	Т	i can draw this super heroi can draw you know?	? question
24	M	É (Yeah)	
25	Т	you help me	
26	M	TCHARAN ((puts the brush in the paint and walks around the room))	capital letters - stressed intonation
			(( )) transcriber's description
27	Т	Marcos i'm asking if you want me to draw the black spider man?	? question
28	M	sim:(yeah)	: vowel stretching
29	Т	are you tired?	? question
30	M	tô quero ir pra casa (yeahi want to go home)	pause

M.'s age: five years and seven months

Also in Table 3, Marcos evidences, in turns 08, 12 and 20, that he participates in the dialogue, producing sounds to make up unintelligible speech. In turn 11, for example, the therapist asks about the color he wanted to use for a proposed activity. He answers by making use of vowel sounds [aa], which are interpreted by the therapist as meaning the white color (branca, in Portuguese), evidencing Marcos's difficulty in producing the consonant group [br], as well as the velar consonant [k].

In this case, the therapist should investigate if the difficulty can be confirmed, investing in placing the speech sounds, according to the child's possibility. Anyway, it is worth pointing that, in turn 16, Marcos says *homem aranha preto* (black spider man), that is, his speech follows the phonological pattern of the Portuguese language, in addition, the consonant group [pr] is produced, showing that the unvoiced sound [p] is already part of this child's orality.

In turn 19, the therapist asks a question related to Marcos's routine, involving the patient's older brother's preferences. It is a dialogical excerpt on a routinely discussed subject in the speech-language therapeutic follow-up, which includes the child's family. However, in this specific fragment, Marcos answered the therapist in such a way that he could not be understood. In these

situations, the therapist should pay attention to the possibility for Marcos to produce unintelligible sounds intentionally when he is not interested in interacting. If that is the case, the therapist should interpret the attitude so that Marcos can think about it and, if possible, reorganize it. Marcos has the right of not being willing to interact in all speech-language sessions, and do what is proposed by his therapist. That understanding allows him to be respected as a unique subject, taking over as an active, responsive participant in the verbal interaction flow. Thus, Marcos needs to consider that he can and should tell his therapist when he is not willing to carry out certain activities. Thus, the listener will effectively have a place in the discursive chain, enabling him to be recognized as a unique subject, participant in unrepeatable enunciative situations.

In Table 4, as well as in the former ones, Marcos evidences that he can participate in the enunciation, providing an answer after the other to justify his actions, according to his therapist's request. In turn 02, he answers the therapist's question coherently, with a negative. In turn 07, he justifies the negative, explaining that he had not gone to school because he had a sore throat. In this turn (07), it is also possible to observe Marcos's syntactic and semantic organization, to the extent that he uses the verb

Table 4. The Therapist (T) and Marcos are, on 06/13/2016, talking about Marcos's daily activities

Turn	Symbol	Transcription	Indicator/NURC
01	Т	but then did you go to school?	pause
02	M	Não (No)	
03	Т	why?	? question
04	M	porque sim (because i didn't)	
05	Т	because i didn't? is that an answer? ((laughing))	? question; (( )) transcriber's description
06	Т	why didn't Marcos go to school?	? question
07	M	porque tô com dor de garganta:(because i've a sore throat)	: vowel stretching
08	Т	are you taking medicine?	? question
09	M	é (yeah)((points to his throat)))	(( )) transcriber's description
10	Т	a: medicine for sore throat:	: vowel stretching;
			pause

M.'s age: five years and eight months

to be, first person singular, in the present tense, in a coherent way in the dialogue. In this dialogue, Marcos shows, again, his progress in the semantic and syntactic aspects concerning the oral language, by producing, according to these discursive possibilities, complete enunciations, coherent with the enunciative situation.

In the four dialogues presented here, Marcos shows that he is inserted in the discursive chain, and therefore, he can participate in enunciative processes, mixing the use of gestures, mimics, pointing and orality itself. Initially, at the beginning of the speech-language therapeutic treatment, according to data from his records, Marcos could not even get into the therapy room by himself. He needed the frequent presence of his parents. In the beginning, Sofia and Dante interpreted Marcos's actions to his therapist. Then, he took over, as a subject able to enunciate and be enunciated, as he recognized himself inserted in a symbolic chain, in the clinical speech-language therapeutic process.

The enunciation, evolving from the interaction of two or more subjects, is delimited by its immediate situation, and concomitantly, reflects a broader social structure<sup>(14)</sup>. Marcos's family, according to Sofia's reports, since his birth, has been trying to set apart from a social view grounded in the exclusion and disbelief from what is different. Despite all the TCS-related difficulties, it is a family who has invested in the potentialities of their child, working to improve his quality of life. In this sense, the family role stands out along the process of Marcos's language appropriation.

In the dialogical perspective, the other is relevant in the constitution of each subject, as the enunciations are addressed to someone, and a response is expected from the listeners, active participants in the discursive practices immersed in living contents.

In Marcos's case, according to his history, it is possible to follow his participation in all household tasks, being interpreted in his actions and respected his singularities. His parents, despite their son's organic disabilities, have seen Marcos as an individual able to understand and participate in the dialogical flow of the community that he lives in. In the child's records, it is possible to follow his parents' reports on the interactions that they establish with Marcos every day. He has participated in all family meals since his first year of age, even not being able to have oral feeding. He sits at the table with his parents and brother and joins the talks, during lunch and dinner. In those moments, his family members discuss their routine and Marcos participates in the discussions by means of different semiotic

expressions, mimics, orality, which are interpreted by his parents and brother.

Besides integrating family routine, he plays with his older brother and participates in leisure activities with his parents, visits his grandparents, uncles and cousins, being compelled by his parents to express himself by means of gestures or orality. In the situations that he is not understood by his listeners, his mother takes on the role of mediator and interprets the interactions that Marcos is involved in.

In a dialogical perspective, understanding is a process which implies former meaning of all involved in a dialogue. Therefore, the dialogical activity depends on the responsive understanding of each participant that interferes in the enunciation and, consequently, in the other's enunciation<sup>(15)</sup>. By his family, Marcos has been treated as someone who has conditions to understand the events that he experiences. Therefore, his parents attribute meaning to his actions, thus, he can participate in sociodiscursive situations, showing to understand them in a responsive way.

Similarly, the therapeutic treatment tried to potentialize Marcos's interactive possibilities, giving him space so that he could use all his resources – body language, facial mimics, vowel intonations, orality - to reassure himself as a listener. The therapists, grounded in a dialogical perspective of the language<sup>(11)</sup>, tried to interact with Marcos, taking him as a unique subject, constituted in the interaction of the social voices that surround him. In this sense, understanding that enunciation cannot be taken, in a simplistic way, by the psychophysiological circumstances of the subjects that participate in it<sup>(11)</sup>, the speech-language therapeutic follow-up set apart from explanations based on physical and physiological aspects and focused on Marcos's semiotic productions. Thus, beyond the physiological mechanism involved in Marcos's sound productions, his follow-up highlighted the fact that dialogical activity is connected with the social environment where it is produced.

Marcos's clinical process took in consideration, besides the organic disabilities due to the TCS, the relationship that he can establish with his therapists and family, in diversified enunciative situations. It is possible to follow, in this child's story and in the four dialogical events shown, that this boy is able to participate in dialogical situations. He understands the other's orality and makes himself orally understood, expressing his opinions, agreeing or disagreeing with what is proposed to him, elaborating, in a meaningful way, questions and producing complete sentences.

#### **FINAL CONSIDERATIONS**

The current study aimed to analyze the therapeutic process, oriented to the oral language, from a boy with TCS in a dialogical perspective. In this sense, it was evidenced that this child could participate in enunciative situations, focusing on Marcos's early discursive possibilities, elicited by means of several semiotic resources, such as gestures, facial mimics, body language.

Despite the craniofacial malformations that Marcos has, due to the TCS, this boy gradually began to make use of the orality, to take over his role of listener and participate in the dialogical flow. He has been in the process of oral language appropriation, and it should be pointed out the outcomes that a clinical speech language therapy produced,, by mediating the relation between this child and orality grounded in the dialogical perspective. Similarly, it should be highlighted his family's fundamental participation so that Marcos could place himself as a language subject.

Concluding this manuscript, it is possible to state that the dialogical approach enabled the organization of a therapeutic follow-up, which was able to join actions that, beyond the organic aspects involved in speech production, focused on a child with TCS and his unique history, expanding his interactional possibilities, and providing him with more autonomy to interact with his listeners. Finally, it deems to point out that the formerly mentioned speech-language therapeutic intervention has contributed to the process of oral language appropriation of this child. However, further studies should be carried out on this theme, widening the understanding of Speech-Language Pathology on language appropriation in patients with TCS.

### **REFERENCES**

- Medina JMP, Escobar MCA, Alvarez AYT, Hernández JFP, Carbot DM. Síndrome de Treacher-Collins. Presentación de un caso. Rev Méd Electrón. 2014;36(2):211-6.
- Alfonso LS, Centelles IA. Síndrome de Treacher Collins en una familia cubana. Presentación de caso. Rev Habanera Cienc Méd. 2016;15(3):408-17.
- 3. Leyva CJ, Restrepo GM. Síndrome de Treacher Collins: revisión de tema e presentación de caso. Revista Universitas Médica. 2014;55(1):64-70.

- Polanski JF, Plawiak, AC, Ribas A. Hearing rehabilitation in Treacher Collins Syndrome with bone anchored hearing aid. 2015;3(44):483-87. http://dx.doi.org/10.1016/j.rppede.2015.08.016.
- Rodrigues BGS, Silva JLO, Guimarães PG, Formiga CKMR, Viana FP. Evolution of a child with Treacher Collins syndrome undergoing physiotherapeutic treatment. Fisioter Mov. 2015;28(3):525-33. http:// dx.doi.org/10.1590/0103-5150.028.003.AO11.
- Boku A, Hanamoto H, Kudo C, Morimoto Y, Sugimura M, Tooyama M, et al. Airway management for Treacher Collins syndrome with limited mouth opening. Open Journal of Anesthesiology. 2013;3(2):90-2. http://dx.doi.org/10.4236/ojanes.2013.32022.
- Toledo IC, Azevedo RA, Alves-Rocha A, Silva-Feles DA, Silva-Neta I, Pires-Borges K. Síndrome de Treacher Collins – relato de caso. Rev. Odont. 2016;16(9):945-52.
- Nogueira BML, Silva TN, Nogueira BCL, Silva WB, Menezes SAF, Menezes TOA. Genetic autosomal dominant disorders: a knowledge review. Int J Odontostomatol. 2015;9(1):153-8. http://dx.doi.org/10.4067/ S0718-381X2015000100023.
- Cassab TV, Tonello C, Dutka JCR, Yoshida MM, Alonso N, Antoneli MZ. Alterações de fala na síndrome de Treacher Collins. Revista Brasileira de Cirurgia Craniomaxilofacial. 2012;15(2):69-73.
- Massi G, França DR, Santos RS, Ribas A, Fonseca VD, Guarinello AC, et al. Speech language pathology findings in a Treacher Collins syndrome patient. Int Tinnitus J. 2016;20(1):31-5. http://dx.doi.org/10.5935/0946-5448.20160006. PMid:27488991.
- Bakhtin MM. Marxismo e filosofia da linguagem: problemas fundamentais do método sociológico na ciência da linguagem. 14. ed. São Paulo: Hucitec; 2010.
- Coudry MIH. Caminhos da neurolinguística discursiva: o velho e o novo. In: Coudry MIH, Freire FMP, Andrade MLF, Silva MA, editores. Neurolinguística discursiva: teorização e prática clínica. Campinas: Mercado de Letras; 2011. p. 279-399.
- 13. Galembeck PT, Carvalho KA. Os marcadores conversacionais na fala culta de São Paulo (Projeto NURC/SP). Intercâmbio [Internet]; 2010 [citado em 2017 Out 14]; 6:2746. Disponível em: https://revistas.pucsp.br/index.php/intercambio/article/view/4100/2746
- Mariani BZP, Guarinello AC, Massi G, Tonocchi R, Berberian AP. Speech language therapy practice in a bilingual dialogical clinic: case report. CoDAS. 2016;28(5):653-60. http://dx.doi.org/10.1590/2317-1782/20162015287. PMid:27849242.
- Ucedo DM, Santos KP, Santana APO. Language in frontotemporal dementia: an analysis in light of Enunciative-Discursive Neurolinguistics. CoDAS 2017;29(4):e20160154. http://dx.doi.org/10.1590/2317-1782/20172016154.