

Stories about life and treatment onset of a patient with Treacher Collins syndrome: case study

Narrativas sobre a vida e o início do tratamento de uma paciente com Síndrome de Treacher Collins: um estudo de caso

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ABSTRACT | The Treacher Collins syndrome (TCS) is a rare anomaly of craniofacial development with variable clinical manifestation. The main goal of this study was to analyze the experience of a 26-year-old woman with TCS who initiated her rehabilitation in adulthood, aiming to deepen the possible impacts on her development. After approval by the Institutional Ethics Review Board (CAAE 00981418.3.0000.5441), the case study was conducted by documental analysis of the multiprofessional patient records and a semi-structured interview recorded in audio, transcribed in full and qualitatively analyzed by the Content Analysis technique of Bardin. Two theme categories were evidenced that indicated the life experience of the participant divided in two periods: (1) "Development up to adulthood", which included the subcategories: "What's wrong with me?"; "Relationship with the mother"; "I suffered bullying: challenge of school education" (2) "Being someone with Treacher Collins syndrome", and subcategories: "There are others like me: knowing the diagnosis"; "Treatment onset: challenges and expectations"; "Psychology and the interdisciplinary team"; "But I am happy today: plans for the future". Since TCS is a facial malformation, it may involve living with the stigma of not presenting a face within the imposed social standards. The present case presented emotional suffering, worsened because she did not ask for help. Conversely, the diagnosis of the syndrome allowed the process of identification with other affected individuals, the onset of rehabilitation and experience of positive impacts on her quality of life. Psychology participated as a space for hearing and possibility of reflecting about the rehabilitation process.

KEYWORDS: Treacher Collins syndrome. Rehabilitation. Family. Interdisciplinary team.

RESUMO | Síndrome de Treacher Collins (STC) é uma anomalia do desenvolvimento craniofacial rara de manifestação clínica variável. Este estudo teve por objetivo principal analisar a experiência de uma mulher de 26 anos com STC que iniciou a reabilitação na idade adulta, buscando aprofundar possíveis impactos para o seu desenvolvimento. Com aprovação do Comitê de Ética em Pesquisa (CAAE 00981418.3.0000.5441), realizou-se um estudo de caso clínico por meio da análise documental do prontuário multiprofissional e uma entrevista semiestruturada gravada em áudio, transcrita integralmente e analisada qualitativamente pela técnica de Análise de Conteúdo de Bardin. Evidenciaram-se duas categorias temáticas que sinalizam a experiência de vida da participante dividida em dois momentos: (1) "Desenvolvimento até a fase adulta" que abarcou as subcategorias: "O que eu tenho?"; "Relacionamento com a mãe"; "Eu sofri *bullying*: desafio da escolarização" (2) "Ser alguém com Síndrome de Treacher Collins", e subcategorias: "Existem outros como eu: conhecendo o diagnóstico"; "Início do tratamento: desafios e expectativas"; "A psicologia e a equipe interdisciplinar"; "Mas hoje eu sou feliz: planos para o futuro". STC, sendo uma malformação facial, pode implicar em conviver com o estigma de não apresentar o rosto dentro dos padrões sociais impostos. No caso analisado, apontou-se sofrimento emocional, sendo este intensificado pelo não pedido de ajuda. Por outro lado, o diagnóstico da síndrome possibilitou o processo de identificação com outros sujeitos acometidos, o início da reabilitação e a vivência de impactos positivos em sua qualidade de vida. A psicologia figurou como espaço de escuta e possibilidade de reflexão sobre o processo de reabilitação.

PALAVRAS-CHAVE: Síndrome de Treacher Collins. Reabilitação. Família. Equipe interdisciplinar.

Introduction

Craniofacial development is complex and susceptible to many abnormalities. Approximately one-third of congenital anomalies affect both the head and face, with more than 700 variations of them described so far. Facial dysostoses are a set of rare anomalies of the craniofacial skeleton which are heterogeneous regarding etiology and clinical manifestations. They appear as an abnormal sequence of development in the first and second pharyngeal arches and their derivatives, affecting the maxilla, mandible, and hyoid support structures. They can be subdivided into mandibulofacial dysostosis and acrofacial dysostosis (Terrazas, Dixon, Trainor & Dixon, 2017). The most described mandibulofacial dysostosis is Treacher Collins syndrome (CTS).

When describing how CTS was named throughout time, Andrade et al. (2005) highlight that the diagnosis is called like that because Treacher Collins described its essential components in 1900. The authors indicate that the syndrome incidence varies between 1:40,000 to 1:70,000 live births, regardless of gender or race, with autosomal dominant transmission and variable expressiveness with a great diversity of clinical manifestations.

Variability in the clinical expression of the syndrome makes it difficult to create intervention guides. Nevertheless, the rehabilitation steps aim to preserve the person affected since early childhood by going through multidisciplinary assessment (ophthalmology, otorhinolaryngology, speech-language pathology and audiology, plastic surgery) and surgical interventions in a rehabilitation process that lasts until adulthood (Alfonso & Centeles, 2016). The authors also highlight the importance of psychological support for both the family and the subject being treated due to malformations and the several surgical steps required.

CTS can be characterized by micrognathia (small jaw); malar hypoplasia; antimongoloid, oblique palpebral fissures; lower eyelid coloboma (often with absence of cilia); cleft palate; auricular hypoplasia or dysplasia and middle ear atresia, which can lead to conductive hearing loss (Pollo Medina, Álvarez, Torres, Placeres, & Morales, 2014). The large variation in CTS phenotypic expression can make diagnosis difficult, so it is important the differential diagnosis of other syndromes, such as Nager and Miller syndromes,

which have some similar impairments. Likewise, genetic counseling is complex and necessary as the syndrome has an autosomal dominant inheritance, which means nearly 40% of cases have a family history and 60% appear to be due to new mutations (Passos-Bueno & Splendore, 2001).

Lodovichi et al. (2018) investigated the quality of life perception of individuals with CTS, both with and without ear deformities, and found no difference between them as all subjects scored high on quality of life. The authors mention possible aspects that may have generated this result, such as adherence to an appropriate, individualized treatment protocol in a specialized center; opportunity for longitudinal psychological monitoring for developing coping tools for insults and bullying by peers; and understanding the treatment protocol for minimizing anxiety in people with ear deformities as they become aware of the appropriate time for reconstruction procedures. The authors thus emphasize the importance of specialized treatment and an interdisciplinary team to favor the development of individuals with CTS with a higher quality of life, minimizing the aesthetic and functional impacts caused by the syndrome.

De Martini (2011) conceptualizes rehabilitation as an educational, social, and therapeutic-related work that can shift based on peculiar needs and situations. People undergoing rehabilitation seek to restore or create new resources to resume life activities in the best possible way. The author thus emphasizes that professionals need to understand that a strictly technical discussion – which does not consider socioeconomic status, family issues, and individual choices – will not be enough. Thus, he highlights interdisciplinary work is essential for an adequate, effective rehabilitation process.

The psychologist's practice in any health institution must aim to deal with suffering, which is a situation triggered by illness or hospitalization (Mutarelli, 2015). The author adds that hospitals are essentially healing institutions, and psychologists working in such places should advocate for health promotion to minimize or avoid additional emotional suffering. He also defines these professionals as the bearers of the foreign gaze, directing the team to the patients' singularity, seeing them as unique and bearer of self-knowledge. Psychologists must weave with their teams the integration between patients' life history, illness process, and treatment.

It is important to stress the scarcity of literature on the syndrome that describes the role of psychology in the rehabilitation process or the psychological aspects involved in living with CTS. Given how rare this syndrome is and the particularities of its clinical manifestations, there are only case reports in the published literature directed to very specific aspects of rehabilitation, such as surgical, otorhinolaryngological, and physical therapy interventions.

Thus, this study aims to analyze the experience of a patient with CTS who started rehabilitation in adulthood and sought to deepen the possible impacts on her development, as well as to describe the role of psychology as a party in the interdisciplinary rehabilitation team.

Methods

This study was performed after approval by the Research Ethics Committee at the University of São Paulo's Hospital for Rehabilitation of Craniofacial Anomalies (HRAC/USP), Approval No. 3.051.962, Certificate of Presentation for Ethical Consideration (CAAE) No. 00981418.3.0000.5441. During the study period, HRAC/USP had 125 patients diagnosed with CTS registered, of which only one patient met the following inclusion criteria: 18 years of age or older; regardless of gender; clinical diagnosis of CTS; no diagnostic comorbidity; treatment onset after 18 years of age. The 26-year-old female patient was admitted to the hospital on April 18th, 2018 without having undergone any specific treatment for CTS. She was invited to participate in this study when undergoing individual care in the institution's psychology sector, when she learned about the study, its objectives and methods, as well as signed the Informed Consent Form.

She was advised that participating in this study would expose her to personal information, which could be risky as it could bring back memories of potentially painful experiences and situations. Therefore, the researching psychologist could interrupt the study whenever needed and offer psychological support to minimize any impacts. The participant was aware that she could stop participating in the study without any impact on her rehabilitation at the institution.

To achieve the proposed objectives, a clinical case study was performed through documentary analysis

of the multidisciplinary medical record to define the clinical (diagnosis, therapeutic stages) and psychosocial aspects (socioeconomic profile). Extra time was scheduled in the psychology sector so the researcher could learn about the participant's individual experience. The sessions were offered in a private room. A semi-structured interview was conducted by the researching psychologist, starting from three initial questions: what is like to live with CTS regarding personal, family, and social aspects; talk about your treatment; how do you perceive the role of psychology in your treatment. This interview was audio recorded with the participant's permission and analyzed by using Laurence Bardin's content analysis technique.

Thus, following its assumptions, the analysis was performed in three stages. Initially, the material was organized through the full transcription of the recorded audio and an initial floating reading in search of indicators. The material was then explored through new readings so that the data could be coded in record units. This process was built aimed at the content that responded to the proposed objectives. Finally, data listed were treated and interpreted, thus building, through grouping by similarity and differentiation, the thematic categories and subcategories relevant to the theme. Caregnato and Mutti (2006) name these steps as pre-analysis, material exploration, and treatment of results and interpretation. Interpretation and inference, according to Câmara (2013), is the moment when the researcher makes raw results significant and valid. For Bardin (2009), the inference in the content analysis is an induction tool to investigate the causes starting from the effects, and interpretation will only make sense when comparing the data obtained with the theoretical foundation. This process was performed in the discussion.

Case description

The study participant is single, female, and 26 years old. She reported orthodontic braces as the only treatment she underwent since 2016. In an initial assessment at the institution, she met with the craniofacial team, which included craniofacial surgery, neurosurgery, speech-language pathology and audiology, genetics, psychology, orthodontics, and social work professionals. The consultations were

individual, and afterward the team met to discuss the case and define conduct.

The genetics sector confirmed the diagnostic hypothesis of CTS, identifying the following clinical (or phenotypic) characteristics: absence of cleft lip and palate; downward oblique palpebral fissures; bilateral lower eyelid coloboma; hypoplasia/agenesis of the zygomatic arch; mandibular hypoplasia; discrete, prominent, slightly altered ears. The main complaint was dyspnea.

The social assessment scored lower average socioeconomic classification and highlighted the following characteristics: completed high school, working as a stock handler, living with three siblings and a nephew, reported good family relationships and motivation for treatment.

After the initial assessment, the surgically assisted rapid maxillary expansion procedure (SARME) was indicated as the surgical procedure to expand the maxillary segments. The procedure was performed on July 11th, 2018. She is currently scheduled to undergo maxillary, mandibular, and chin orthognathic surgery.

Results

Analysis of the interview allowed us to identify that the participant's life experience can be didactically divided into two moments. The first shows the thematic category "Development Until Adulthood," which includes the following subcategories: "What do I Have?;" "Relationship With Her Mother;" "I Was Bullied: The Challenge of Going to School." The second moment highlighted the thematic category "Being Someone with Treacher Collins Syndrome," which in turn covered the subcategories: "There are Others Like Me: Understanding the Diagnosis;" "Treatment Onset: Challenges and Expectations;" "Psychology and the Interdisciplinary Team;" "Today I Am Happy: Plans for the Future." The significant contents of each category and subcategory will be described, and the participant's speeches will be in the spotlight to exemplify the reported experience. The first moment of the participant's life was described in the following thematic category:

Development Until Adulthood

This category covered the striking aspects of the participant's life experiences until adulthood as a person who had always had facial malformation but did not know that was an indicator of CTS. These are the thematic subcategories:

What do I Have?

The participant's life experiences were marked by growing up feeling different, having marks on her face, and not understanding what she had. She experienced countless situations in which being different made people offend her, including experiences in the family environment as well as in other social situations even in childhood. She would feel different yet fully capable. These experiences marked her development.

"I would look in the mirror and not know what I was, like ' Does it have a name? Do other people have it? I knew nothing at all... So it was hard for me to figure out. "

"... And the children and other people would make jokes, asking if I had suffered a car accident. That would always happen to me but I was a child, and children don't get it. You can cuss them, call them names, and they will still hug you. "

Her way of facing situations of discrimination in her life experience became evident. She would try not to react to the remarkably painful situations but ended up experiencing intense psychological suffering. Nonetheless, she had faith and would hope things would be better.

"... all my life I have let go of what people have done to me. I wouldn't talk back, I just accepted ... I never let myself down for being isolated. When I was a teenager I would spend some nights crying."

Relationship With Her Mother

The participant's report made clear the aspects of her relationship with her mother, the strong bond between them, and the impacts of the way her mother dealt with her malformation and her non-treatment at the appropriate age. She reported realizing that her mother wanted to be able to take care of her health, but socioeconomic, family, and even emotional conditions limited her possibilities.

There was a specific moment when she told her mother she wanted to seek treatment and understood her mother's difficulties in that situation. Their strong bond is her what helps her fight back all difficulties she experiences.

"I remember she said: Do you want to work on your case? And I said: I do, I really do! I couldn't take it at that moment. My eyes teared up, I started crying, and so did she. She suffered more than me with the problem. She said that at the right time God would act, and then I didn't have the courage anymore. So for basically two days she was, like, feeling down, swallowing her tears. And I understood her, it was very difficult for her. My mother had 17 children, she worked a lot in her life, and when she turned a certain age she was no longer healthy. So the one who needed to be taken care of wasn't me, it was her. And that's what I did. I took care of her until her last day."

When reflecting on her life story, the participant wonders why she never talked to her mother about possible treatments and regrets it.

"the most amazing thing is that I couldn't communicate that to my mom, and I don't know why. Today I regret it because I didn't sit down with her and told her I knew she never wanted to get started with my treatment and all, but now I have the strength to go ahead and take that burden off her."

Later, the strong bond they have is evident again as the participant does not hold the mother responsible for her non-treatment. She understands the circumstances that may have influenced the mother's passive attitude towards the birth of a daughter with malformation and shows how much she felt loved by her mother.

"... and thanking me for not asking her. I never judge her. I think that as a daughter I saw the suffering in her eyes and if it were for her I would have been treated many years ago. But she had no strength, so I respected that part, and the mother's heart says everything, right? So she knew that things would happen at the right time."

The participant realizes that the way her mother treated her, giving her hope and stimulating her faith, was her support to face difficult situations daily.

I Was Bullied: The Challenge of Going to School

Going to school was marked by numerous bullying situations for the participant. During her adolescence this experience became worse, marking the way she acted in the classroom, as well as how she dressed and took care of her hair. This experience caused her great psychological suffering. She experienced that without complaining at school or to her family, so she did not receive any appropriate help, and her faith was her greatest source of support.

"I think the worst phase was adolescence at school. I was bullied a lot; I had no friends... Everyone was very prejudiced... I took all the bullying and prejudice in silence. Sometimes I would cry at night, tears would flow from my eyes, and I would ask God for strength so I could continue studying, living, being happy, looking at myself. I struggled a lot, I had many nicknames, it was horrible! I don't know how I managed to finish high school..."

Even in the face of psychological and sometimes even physical violence, the participant's goal was to finish school so she could work later. The fear of being offended shaped her behavior at school, so she looked for resources to keep motivated, such as reading.

"'I will finish and start working right away.' That desire to finish school and start working, but at the right time, was something I can't even explain. I even ask myself: how did I make it? Despite everything I made it. I think it was strength and determination, because I really, really wanted to learn."

Her school experience was marked by her seeking to protect herself from the violence she was submitted to, so whenever possible she avoided exposing herself by not participating in important moments in school life and hiding school events from her family. However, she mentions the intense psychological suffering of not experiencing these important stages of school life.

"I remember those graduation photos I didn't take because I'd be exposed at school, so I didn't go. Then I regretted it. It was silly, I should have taken the photo, whether I would get criticized or not... I knew the students were going to make a joke right there if I took the picture, or if I received my diploma after finishing high school. So I didn't go either. But that hurt me a lot, I should have gone... And I didn't tell my mother either, I wouldn't speak. There were things she didn't even know about."

The second moment of the participant's life, after she learned about her diagnosis, included the following thematic category:

Being Someone with Treacher Collins Syndrome

Discovering that her facial features were indicators of the syndrome diagnosis was a remarkable moment that started a process of identification with other people with CTS, favoring her understanding of her difficulties and envisioning a treatment process. From the participant's worldview, understanding her diagnosis was like "her time had arrived, because God does not forsake anyone". Consequently, the following thematic subcategories analyzed:

There are Others Like Me: Understanding the Diagnosis

The participant grew up feeling different, with marked facial features and functional difficulties in breathing. Thus, understanding that these aspects were indicators of a syndrome caused her ambivalent feelings, such as shock and identification. It allowed her to meet other people who experience difficulties like her own, or even more intense since CTS has very variable clinical manifestations. It was a milestone in her personal experience as she could understand her suffering was somehow shared by other people with experiences very similar to hers, or even more intense, favoring her process of emotional maturation process and facing difficulties.

"When I found out, it seemed like everything opened up. Everything became clearer for me [...]. It was a natural shock because I was not the only one who has it, several people have it, so I thought, wow, people with much more serious problems, going through serious surgeries, dealing with other people and everything. Mine is the syndrome, but a little mild, and people at school already thought it was different and were prejudiced. Oh, my God!"

Treatment Onset: Challenges and Expectations

Learning about the syndrome also made it possible to learn about the possible treatment for her difficulties, making the participant very motivated and aware of the long, challenging process ahead of her. Going through rehabilitation in a reference center offered her an identification process with other people with CTS and the development of a bond of trust with the team that treats her.

The importance of getting treatment here is that I met children and adults in the same situation, each with different difficulties and overcoming in different ways... It was wonderful because I was welcomed, and the humanization and the affection we have here are immense. "

The participant has already undergone the first surgical stage of treatment. It is a moment surrounded by expectations and intense challenges, such as understanding what each procedure can bring about, the process of hospitalization and recovery in the postoperative period.

"I would feel butterflies in my stomach, all the anticipation. I would get anxious, I wanted it to happen soon, to find out whether it would work out ok. It was all I wanted, I wouldn't even think about anything else, it was this huge joy. Pain is part of the process, but after you go through the pain, even when I was still in pain, I was happy. The palatal disjunction was performed so I felt the device activation, and every time they activated it, it hurt a lot. But even in all that pain, I was still happy, because it would soon be over. It was so fast, I couldn't even imagine the pain I went through, because the happiness was greater, it was bigger than anything.

Recovering from the surgical process involves postoperative care that changes the person's daily routine, such as liquid food, rest. Additionally, the aesthetic changes caused by the recovery process impact on the individual's feeling of well-being as they cause unwanted transitory impacts as reported by the participant.

"...I was hospitalized for 15 days. It swelled! My cheeks were very swollen, my chin was gone. And I stayed at home for a long time. I would wear a mask to go to work. I was ashamed because my teeth had gone so far out that I put a finger through them... So I avoided going out because people would look at me, wouldn't understand, and ask many questions, so I'd rather stay indoors and avoid those questions."

The participant states that that first intervention met her expectations and has already impacted her self-esteem, feeling of well-being. Even with the negative impacts of the recovery process, she is motivated for the next surgical steps as she aims for greater aesthetic and functional comfort in her breathing.

"I also have this great expectation despite the intense pain. But I don't even think about that part. I think I've already gone through a worse phase. It's the details. I know people who don't dare to face orthognathic surgery. After your first surgery, you end up doing everything because the disjunction is no different from the one I'm going to do. You break a bone here, lose some weight, then get ashamed [during recovery from surgery]. It isn't cool, but then over time you bring it to a perfect end, and this is what happened."

Psychology and the Interdisciplinary Team

When the participant was approached about her experience with psychology professionals during her treatment, she mentions she has already undergone psychotherapy in her hometown and highlights the benefits of this self-care process.

"... It's been nearly two years since I started seeing a psychologist. I started, then I quit, then started back again, and it was very good. It is a very important part of the treatment because sometimes we manage to say things to a psychologist that we wouldn't have the courage to talk about with another person. "

The participant states that psychological care during the follow-up sessions at the rehabilitation center is important for her treatment. She adds that if he had had access to this space of self-knowledge before, she could have had different experiences.

"... When you make an appointment with the psychologist you can vent everything you need, and I think that is good. It is great to be able to help the patient. I'm speaking for myself. I didn't tell you, but I didn't have with my mom the conversation I'm having with you, and I regret that I didn't. Maybe if I had a follow-up session with a psychologist before I would have managed to talk to her, but I wasn't prepared."

She adds that in psychology care she could understand what the postoperative process would feel like emotionally wise:

"I think [having psychological care] is paramount. Patients must have that so that they can be aware of what they will go through, whatever they will face, so they can learn how to deal with it. I wasn't aware that my teeth would get further apart, and that I would be self-conscious about it. But I started to open my mind so I could prepare myself beforehand, and thank God it worked, I got my mind ready... there is a process, and it takes time to get to the point He wants you to get..."

The participant understands the difference between the professional roles of the team and psychologist, and she also values the work of all the professionals involved in her treatment.

"... we have other questions for the professionals, and they have their language, which is totally different, about what will happen in the surgery and such, how this will or won't be like, they are careful like that. They don't want to scare the patient. I understand their side [surgical team], I don't think it's easy, but psychology awakens this clarity in the patients, it makes us feel very safe. And then I became aware it would turn out great later. So it encourages the patient to make the decision and face the treatment."

Today I Am Happy: Plans for the Future.

The participant expresses gratitude for her personal experience and shows that her faith made her stronger in the face of such intense experiences. She has plans to start studying and to be able to experience moments that were marked by emotional suffering during her basic schooling process.

"...it's a lot of expectations. I believe I feel safer, more comfortable. I think when I get to the classroom I will feel at ease, discuss an assignment, talk, laugh. It'll be very good. I think everything changes, not only physically, but my mind as well. I feel safer, I feel like talking and asking questions. I feel I'm getting stronger and I have already felt a little change. I believe there will be much more. "

The awareness of her syndrome and her personal history marked by prejudices allowed the participant to reflect on affective relationships with people with the same syndrome, as well as about having children.

"... I have this syndrome; I see it as a normal thing. I don't see any problem if I had a relationship with a person with CTS, sure! Now I don't think about the option of having a child with CTS. It's not that I don't accept it, it's the difficulty that he or she may face in the future. I don't want anyone going through what I went through. So people may think it is prejudice, but it's not. Only those who have it know the obstacles they have to face. Living in the hospital, having to overcome many barriers in society and at work, thinking they don't have what it takes when they actually do. "

The team's geneticist's guidance allowed the participant to reframe her dream of becoming a mother through adoption, for example.

"The geneticist said: it's like throwing a coin, it's heads or tails. I said: look, there are so many ways nowadays to have a child. You can adopt a child who is suffering at an orphanage. That was never difficult for me. I think helping others is much better than giving up on whatever your plan is, which is something that may bring you immense difficulty. It's not that I have prejudice, not really!"

Discussion

The participant's first moment of life, in which the category "Development Until Adulthood" stood out, approached her personal experiences marked by her lack of knowledge about her syndrome, as well as prejudice and psychological violence (bullying). It also indicates how the coping process of her mother, who is her reference figure, is entangled with the confrontation of her condition.

Alves (2016) discusses how the concept of beauty and aesthetics impose itself in existence since physical appearance has an important meaning in people's psychosocial well-being, associating beauty (social construct) with kindness. Thus, the disfigured face deviates from the recommended beauty standards, and the person with facial deformity starts to live with the stigma. This mark, which stands out and moves away, makes it impossible for the affected person's other attributes to be noticed. The stigma of living with a facial malformation in a society with unattainable beauty standards has impacted her development.

The author adds the face as the expression of personal identity, how one is recognized, always revealing subjectivity, fundamental in verbal and non-verbal communication, and that physical attraction also permeates the "beautiful" face. Thus, deeply understanding what the face means for the individual implies being aware that the person can't see him or herself without the help of a mirror. They are seen through other people's eyes and their reactions. The mouth and the smile make up this face and make possible an immense range of meanings and representations. It is through them that one understands and takes part in the world, as well as understands sensations and feelings. Any impairments in these elements can impact

the individual's social participation as the values associated with beauty or its absence will be closely connected to the acceptance of that individual in the environments in which he or she will participate (Alves, 2016).

Therefore, the questions experienced by the participant throughout her development until she is aware of her syndrome, highlighted in the subcategory "What do I have?", are understood. She experienced all the stigma of having a face that is different and distances herself from the standard of beauty socially required. Also, those whom she interacted with built their impressions of the participant, assuming possible disabilities and mocking her.

Psychology is concerned with the effects of stress, which understood as a reaction of the organism to a threat to its stability, involving physical, psychological, mental, and hormonal components (Lipp & Malagris, 2011 as mentioned in Seidl et al., 2018) in the physical and mental health of the person who experiences it. Analysis of the experience described by the participant found the presence of a continuous stressor (something that generated the stress reaction) in her development, which is prejudice and the emotional impact of that process, such as crying and emotional fragility. However, the desire and motivation to overcome these situations are also often perceived, such as when she does not give up on attending school.

Still, according to Seidl et al. (2018), another important concept in this context is coping, understood as cognitive and behavioral efforts aimed to handle the demands of internal or external demands considered as a personal resource overload. Thus, efforts aim to manage (reduce, minimize, or tolerate) these demands of the person-environment relationship, which is a dynamic and reciprocal relationship. The participant experienced these processes without asking for help.

The interview showed the role that the relationship with the mother meant for the non-treatment at the appropriate age and also for the process of coping with the difficulties experienced by the participant, thus highlighting the subcategory "Relationship With Her Mother."

The participant's mother faced vulnerable socioeconomic conditions. Also, the birth of a child with impairments or disabilities can cause a crisis in the family, damaging their identity, structure, and functioning. Sometimes the family finds itself unprepared to face such a reality that will require breaking expectations, new emotional demands, and living with the child with different needs. The family seeks to adapt and reorganize in the face of that reality, but this adjustment may require time. Thus, some families manage this transformation while others have greater difficulty, showing hopelessness and perceiving its structure threatened, with reduced capacity to react to this crisis (Barbosa, Balieiro & Pettengell, 2012).

According to De Martini (2001), disability since birth can impact the developmental impairment when the situation is interpreted with fear and ignorance, resulting in environments of overprotection or depreciation, impacting the child's desire to explore his body and world.

The child's first years of life are a crucial period for the development of resources for their entire life, which later we understand as his motor, physiological, affective, or psychic aspects. These are directly involved in the relationships that the person has with the physical space, with their own body, and that of others (his body image), with their interests, abilities, and so on (De Martini, 2011, p. 2266). The result of this development process, whether a facilitator or not, will impact rehabilitation. For dependent, passive adults, the process of rehabilitation will mean allowing for the first time the development of resources to undertake whatever they need to do. That may mean a profound change in the person's representations of themselves and their relationships (De Martini, 2011). This process was identified in the participant of this study.

The several aspects that may have influenced the participant's mother's coping with the daughter's malformation condition impacted her lack of knowledge that such condition indicated the diagnosis of a syndrome, as well as the lack of treatment at the appropriate age. However, the participant understands these possible aspects and does not directly blame the mother. Nevertheless, she experiences moments of ambivalent feelings for not having asked for help and never having proposed a conversation about the issue with her mother.

When considering family and disabilities, Regen (2011) indicates that the role of a stable family is to enable a safe training ground in which children can learn to be more human, to love, to build their personality, to develop their self-image, and to relate to the broader and unchanging society in which they were born. That indicates the family's major importance as the first social cell of which they are a part and the main responsible for individual and social formation. With its specific characteristics, the strong relationship the participant had with her mother made it possible to learn important values such as respect, kindness with other people, and religious faith, which are her foundation for facing personal difficulties.

When observing the participant's development before her syndrome diagnosis, the last subcategory listed is the experience of the schooling process marked by intense bullying. Throughout her school life, being different brought her psychological violence and numerous teasing and offenses, which are processes that produced intense psychological distress for her. Bullying as a social phenomenon is understood by Moreira (2012) as intentional and repeated acts of physical or psychological violence, practiced by an individual or a group of individuals, aiming to intimidate or assault another individual, which may occur in a school or business environment and produces harmful effects on the victim.

Being subject to bullying for many years can lead to a certain psychological disruption, but it is crucial to understand that it is present at the base of a society in a structural, socioeconomic, and cultural manner, and especially in human morality. Managing this situation, therefore, depends on the family, school and society triad, each playing their educational role (Moreira, 2012).

The participant describes that the most difficult period of her school experience was during adolescence, which was both when she had no friends and the aesthetic issue has a more serious impact. Abreu and Zacharias (2016) refer that this phase of human development pervades questions, changes, concerns about the body and uncertainties. It is a difficult period as self-image and self-esteem are impacted by the aesthetic standards imposed by society and reinforced by the media, as already previously reflected. They add that women are, in fact, the main target of this issue and may suffer emotional impact when they fail to meet the imposed standard,

surrounded by feelings of frustration, fear, anguish, and insecurity, which can lead to depression.

Experiencing adolescence as a natural moment of questioning and physical and psychological changes, linked to facial malformation, intense bullying, and a less active posture, without asking the family or school for help, was a source of intense emotional suffering for the participant. Her faith produced hope for some change and made her stronger so she could endure the transition, complete basic schooling, and work in the future, as well as find help in resources such as literature.

Still according to Abreu and Zacharias (2016), the family is key to managing problems during adolescence, especially when there is bullying at school, because the family is the environment that shapes and shows the world, as well as teaches people to move forward. The individuals in this dynamic, as well as the school, must be partners to understand the seriousness of the problem and encourage the creation of a space for awareness (Abreu & Zacharias, 2016).

Gómes-Ortiz, Del Rey, Bolamos, and Ruiz (2014) sought to study the possible relationships between parenting styles and the parties involved in bullying (aggressors, victims, victim-aggressors) and found some research that proposed possible relationships, specifically regarding communication between parents and children and problems of victimization and violent school conduct, that indicates a greater chance of involvement with this phenomenon in adolescents who perceive negative communication with parents. The authors, however, make it clear that these analyses are incipient and may depend on the social, cultural, and school reality in which the bullying phenomenon is occurring. Therefore, it is possible to indicate a probable relationship between the participant's role as a passive victim and her ineffective communication with her mother. She sometimes wished to propose important conversations to her mother but was constantly concerned with her well-being since her mother seemed to suffer when the daughter's malformation was the subject. The participant then remained silent and did not receive the help she needed.

The analysis identified a second moment that surfaced the thematic category "Being a Person With CTS,"

which can be didactically divided into subcategories: "There are Others Like Me: Understanding the Diagnosis;" "Treatment Onset: Challenges and Expectations;" "Psychology and the Interdisciplinary Team;" and "Today I Am Happy: Plans for the Future."

Learning about her diagnosis of CTS was a moment of emotional ambivalence for the participant as she first experienced shock, but then could understand she was not the only one and met other individuals with the same syndrome, which has varying degrees of impairment. When discussing the impact of a diagnosis, Oliveira-Cardoso, Garcia, Santos, and Santos (2018) explain that, at that moment, individuals need to redefine their routine and review their plans. Uncertainties and insecurities are raised, and treatment is announced, allowing the beginning of a crisis that is surrounded by emotional fragility. Considering that, individuals need to feel welcomed and supported in this delicate period of their life. Upon learning about her diagnosis, the participant could meet others with the same syndrome and identify with them, perceiving herself as belonging to a group that experiences similar difficulties, either at a functional or social level.

The subcategory "Treatment Onset" describes this experience as a moment marked by expectations and challenges. The participant had already performed her first surgical intervention and described the impacts experienced during the hospitalization period, such as swelling and pain. During the postoperative moment at home, she had to adapt to temporary aesthetic and functional changes, such as a gap between her teeth and breathy voice, until she could identify the positive impacts of this intervention, such as feeling better about herself and self-care.

Carvalho, Martins, and Barbosa (2012) highlight that individuals with dentofacial deformity tend to have high levels of anxiety, reduced satisfaction with facial and dental image, reduced self-esteem, and feeling of insecurity, as described by the participant at several moments during the interview. Therefore, they add there must be good preparation of this individual to manage the emotional impacts throughout an orthodontic surgical intervention as these are invasive interventions in the most exposed area of the body (face).

Currently, the participant looks forward to her next orthognathic surgery but is aware of the processes involved in the surgical procedure. Orthognathic surgery allows for improvement in dental occlusion, facial aesthetics, and masticatory and respiratory functions. The main motivation for patients is the aesthetic impact, which is usually accompanied by expectations of associated psychological benefits (Carvalho et al, 2012).

Mutarelli (2015) points out that being in the hospital environment impacts a break in the routine of the individual who is experiencing curative care, based on the medical model. Thus, in this context, it is up to the psychologist to seek to offer conditions so that hospitalization can be healthily perceived from a psychic point of view, and the experience of illness can be integrated into the individual's history, acting as health promotion.

By exploring the participant's perception of the role of psychology throughout her treatment, the thematic subcategory "Psychology and the Interdisciplinary Team" became evident. The participant values the care provided by the whole team responsible for her treatment and can specify the roles of each professional. The hospital environment in the curative model favors that the relationship established between the health team and the patient/family is extremely impersonal. That means each professional with their expertise performs their care procedures without necessarily knowing the patient's uniqueness. It is then up to the psychologist in this environment, who is trained to offer an individualized view, to take the place of bringing to the other team professionals the patient's idiosyncrasies, understanding the patient's emotional functioning and helping the team and patient to seek coping strategies for the possible issues that are impacting the treatment (Vieira and Waischunng, 2018).

The participant observes that the consultations with the institution's psychology team were essential for preparing her for the surgical procedure, providing opportunities in which she could critically reflect on the processes that would involve the surgery,

postoperative moment, and adaptation to the changes resulting from the procedure. Additionally, the hospital psychologist's role is highlighted, which is to work with the health care team to promote support and safety to patients and their families through collaboration in decision-making regarding the conduct to be taken (Borges & Soares, 2018).

Patients sometimes receive information from doctors and surgeons that make them calm or that cause questioning, so they need a space to listen, freedom to externalize their feelings about the situation they face. Therefore, according to Saldanha, Rosa, and Cruz (2013), it is up to the psychologist, as a professional who works with the multidisciplinary team, to help patients and their family to deal with the emotional content that emerges through fantasies, fears, and doubts after these conversations and guidance sessions with health care teams.

As a last subcategory, "Today I Am Happy: Plans for the Future," the participant's motivation regarding her future was clear, indicating plans to obtain higher education, and especially the impact with the awareness of having a syndrome and her decision about having children. When she understood the genetic risk of having a child with the same diagnosis, she reinterpreted her maternal dream through the possibility of adoption. Genetic counseling (GC) has several objectives, ranging from defining a diagnosis to understanding the prognosis and risk of occurrence or recurrence of the investigated genetic issue. Thus, GC has the crucial purpose of enabling conscious and balanced decision-making about the health of oneself and family members (Hannum, Miranda, Brito, Costa-Neto, and Cruz, 2015).

The health care area has adopted the term "resilience" to refer to those who have a positive adaptation to a threat. The participant's life story is marked by doubts, fears, prejudice, emotional suffering, and intense desire to move forward, as well as faith in waiting for the "good to come". From that, it can be suggested that she is a resilient person, understanding that being resilient does not mean the absence or immunity to risks, but a dynamic process of adaptation in the face of adverse experiences (Seild et al, 2018).

Conclusions

This study analyzed the experience of a person with CTS and could present the specifics of her life history, impacted by her not knowing her diagnosis, resulting in the late start of her rehabilitation process. That clarified the impacts on her development, such as intense emotional suffering during basic schooling, less active posture in facing her issue, and deficient communication with his mother, even though the affective bond between them had been positive. Treatment onset can also be noted as an important opportunity to reframe her life experience.

However, it is necessary to highlight the limitation of this study when analyzing a single case. Given the possibilities of clinical manifestations and characteristics of the syndrome rehabilitation, it is important to ensure that each person affected by CTS can experience the functional, psychological, and social impacts in a unique way from their subjectivity and life history, as well as participate in different experiences from those reported here.

However, it is worth noting that the notes highlighted here signal important issues to which rehabilitation teams should pay attention. These include the importance of the family when facing the diagnosis throughout the subject's development, the presence of situations of violence in the school environment and possible less active postures by the victim, and the importance of psychological support in the treatment process, especially in surgical moments, in line with what the literature indicates as the role of the psychologist in a health care team. There is a need for further research with a qualitative view that could expand the knowledge on the experience of being a person with CTS and the rehabilitation process in several aspects: does knowing the diagnosis from birth and starting rehabilitation at the ideal age lead to less emotionally distressing experiences? What is the role of family communication in coping with bullying situations? These are relevant questions that arose after this study and deserve investigation.

Contributions of the authors

Guedes EG participated in the conception, design, search and analysis of research data, interpretation of results, writing, and forwarding of the scientific article. Ribas MC and Abramides DVM participated in the conception, design, interpretation of results, and writing of the scientific article.

Conflicts of interest

No financial, legal or political conflicts involving third parties (including but not limited to government, companies and private foundations) have been declared for any aspects of the work submitted (including but not limited to grants and funding, participation in advisory council, study design, preparation manuscript, statistical analysis).

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