DEVELOPMENT OF A CHILD WITH PATAU SYNDROME IN A SPECIALIZED CARE CENTER IN DEAFBLINDNESS

DESENVOLVIMENTO DE UMA CRIANÇA COM SÍNDROME DE PATAU EM UM CENTRO DE ATENDIMENTO ESPECIALIZADO EM SURDOCEGUEIRA

DESARROLLO DE UN NIÑO CON SÍNDROME DE PATAU EN UN CENTRO DE ATENCIÓN ESPECIALIZADA EN SURDOCEGUEIRA

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***A*BSTRACT**: This is a follow-up field study on the evolution of cognitive and psychomotor development of a child with total congenital deafblindness and Patau Syndrome, attending at a Specialized Care Center - Deafblindness in the interior of the State of Paraná. The child was identified as AC to guarantee the identity. The objective is to describe the child's performance in 2019 based on the methodology of *Jan Van Dijk* used for babies with deafblindness in pre-linguistic phase. Although the literature presents us with limited possibilities for these children, it was found that the professional must believe in early and continued stimulation to benefit the child's quality of life and allow hope for families. The results of Jan Van Dijk's methodology suggest that they are effective in individual care with the child, who favor alternative communication resources.

KEYWORDS: Deafblindness. Special Education. Patau Syndrome.

RESUMO: *Trata-se de um estudo de campo sobre o acompanhamento na evolução do desenvolvimento cognitivo e psicomotor de uma criança com surdocegueira congênita total e Síndrome de Patau, atendida em um Centro de Atendimento Especializado – Surdocegueira no interior do Estado do Paraná. A criança foi identificada como AC para garantir a sua identidade. Objetiva-se descrever o desempenho da criança no ano de 2019 tendo por base a metodologia de Jan Van Dijk utilizada para bebês com surdocegueira em fase pré-linguística. Embora a literatura nos apresente reduzidas possibilidades para essas crianças, verificou-se que o profissional deve acreditar na estimulação precoce e continuada para beneficiar a qualidade de vida da criança e permitir esperança às famílias. Os resultados da metodologia*

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de Jan Van Dijk sugerem ser eficazes no atendimento individual com a criança, que privilegiam recursos alternativos de comunicação.

PALAVRAS-CHAVE: Surdocegueira. Educação especial. Síndrome de Patau.

***RESUMEN**: Este es un estudio de campo sobre el monitoreo de la evolución del desarrollo cognitivo y psicomotor de un niño con sordoceguera congénita total y síndrome de Patau, visto en un Centro de Atención Especializada - Sordoceguera en el interior del Estado de Paraná. El niño fue identificado como CA para garantizar su identidad. El objetivo es describir el desempeño del niño en 2019 basado en la metodología Jan Van Dijk utilizada para bebés con sordoceguera en la fase prelingüística. Aunque la literatura nos presenta posibilidades limitadas para estos niños, se descubrió que el profesional debe creer en la estimulación temprana y continua para beneficiar la calidad de vida del niño y permitir la esperanza de las familias. Los resultados de la metodología de Jan Van Dijk sugieren que son efectivos en el cuidado individual de los niños, que favorecen los recursos de comunicación alternativos.*

***PALABRAS CLAVE**: Sordoceguera. Educación Especial. Síndrome de Patau.*

Introduction

The Deafblindness area was recognized by the Ministry of Education, in the beginning of the year 2000, and in 2002, by the Special Education Secretariat of the Ministry of Education, replaced by the Secretariat of Continuing Education, Literacy, Diversity and Inclusion (SECADI, Portuguese initials), which provided advances and possibilities for serving people in these conditions (BRASIL, 2002).

> The recognition of the deafblind as a unique individual is quite recent. In addition, although much was known about the disabilities in isolation, the studies did not show results, which only started to emerge when the deafblind person started to be recognized as a unique being (FARIA-ALMEIDA, 2004). For this reason, we adopted the word "deafblind" spelled without the hyphen because we understand, following Lagati's ideas (1993), that the deafblind presents other difficulties besides those presented by deafness and blindness, therefore, he should be considered a unique individual. In addition, deafblindness presents different degrees of hearing and vision loss, which makes it impossible to use the senses of distance; creates special communication needs; it causes extreme difficulty in achieving educational, social and leisure goals, to access information and understand the world around (ALMEIDA, 2007, p. 35).[4]

[4] O reconhecimento do surdocego como indivíduo único é bastante recente. Além disso, embora muito se soubesse sobre as deficiências isoladamente, os estudos não apresentavam resultados, os quais só começaram a surgir quando a pessoa surdocega passou a ser reconhecida como um ser único (FARIA-ALMEIDA, 2004). Por isso, adotamos a palavra "surdocega" grafada sem o hífen por entendermos, seguindo as ideias de Lagati (1993), que o surdocego apresenta outras dificuldades além daquelas apresentadas pela surdez e pela cegueira, devendo, portanto, ser considerado um indivíduo único. Além disso, a surdocegueira apresenta diferentes graus na perda da audição e visão, o que impossibilita o uso dos sentidos de distância; cria necessidades especiais de comunicação;

In the educational area, the teacher is considered a Mediating Instructor and in his practice he must consider what results can be achieved when the individual characteristics are contemplated, as Nascimento and Costa (2010, p. 243) mentioned, that

> [...] In the case of the deafblind, their linguistic and sensory condition triggers the need to obtain technological and human support from the education system that addresses their needs [...] the technological aspect refers to the systems that enable access to information such as the braille display also known as the braille line (translates the content on the computer screen to the braille system), among other technological resources available on the market and often inaccessible to the student.[5]

The Mediating Instructor[6] is also considered as an Assistive Technology resource (CAT, 2007)[7], whose definition can be described as a

> [...] knowledge area, with an interdisciplinary characteristic, which encompasses products, resources, methodologies, strategies, practices and services that aim to promote the functionality, related to the activity and participation, of people with disabilities, inabilities or reduced mobility, aiming at their autonomy, independence, quality of life and social inclusion (BRASIL, 2007, s/p).[8]

Assistive Technology resources and equipment are also used for students with congenital multiple sensory disabilities (which occur at the birth of the child or acquire before the acquisition of a language) with learning difficulties. The Mediating Instructor will promote the teaching of alternative communications and provide mediation between the person with deafblindness and/or the person with multiple sensory disabilities and their environment (MAIA, 2008), allowing the possibility of communicating effectively with people around them and receive undistorted information from those same people.

causa extrema dificuldade na conquista de metas educacionais, sociais, de lazer, para acessar informações e compreender o mundo que o cerca (ALMEIDA, 2007, p. 35).

[5] [...] No caso do surdocego, sua condição linguística e sensorial desencadeia a necessidade de obter do sistema de ensino apoio tecnológico e humano que contemple suas necessidades [...] o aspecto tecnológico refere-se aos sistemas que viabilizam o acesso à informação como o display braile também conhecido como linha braile (traduz o conteúdo que está na tela do computador para o sistema braile), entre outros recursos tecnológicos disponíveis no mercado e muitas vezes inacessíveis para o estudante.

[6] As an explanation, the teacher will be called the Mediating Instructor throughout this text.

[7] Assistive Technology is an interdisciplinary area of knowledge that encompasses products, resources, methodologies, strategies, practices and services that aim to promote functionality, related to the activity and participation of people with disabilities, inabilities or reduced mobility, aiming at their autonomy, independence, quality of life and social inclusion (CAT, 2007).

[8] [...] área conhecimento, de característica interdisciplinar, que engloba produtos, recursos, metodologias, estratégias, práticas e serviços que objetivam promover a funcionalidade, relacionada à atividade e participação, de pessoas com deficiência, incapacidades ou mobilidade reduzida, visando sua autonomia, independência, qualidade de vida e inclusão social (BRASIL, 2007, s/p).

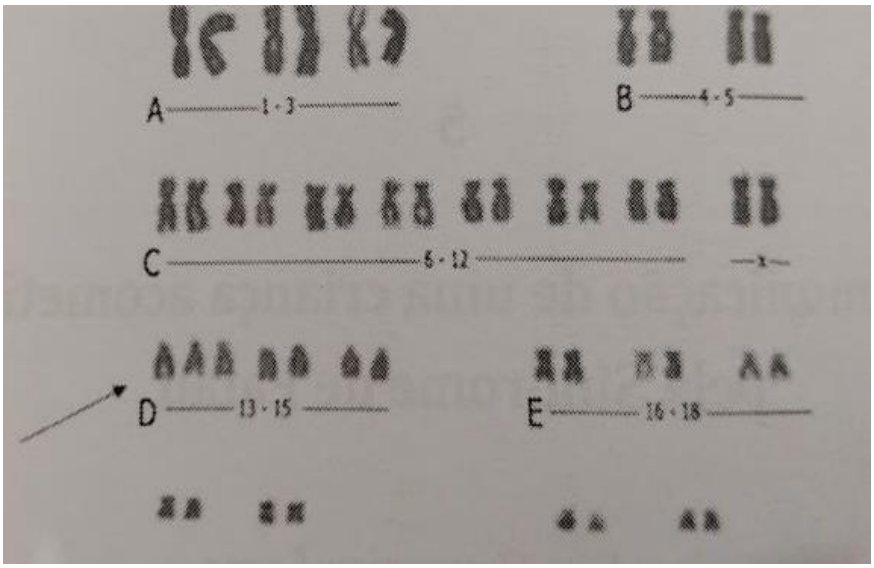
Considering the characteristic of partial or total impairment of the sensory systems of vision and hearing, the person with deafblindness presents difficulties in the academic and personal process, caused by distance, lack of mobility and accessibility. In this scenario, the teacher who acts as Mediating Instructor will establish a link between him, the child and knowledge, as the deafblind child has specific needs for his interaction with the environment of his coexistence, receiving and expressing communication, in addition to difficulties in locomotion. These facts, which can be considered a challenge for the family and mediating professionals, since the child will demonstrate the best form of communication for himself.

Not infrequently, deafblindness is associated with other disabilities and syndromes (MONACO, 2004), it is the case of the following report of a child who presents, in addition to deafblindness, Patau Syndrome associated with the set that defines the diagnosis and the clinical frame of this condition.

The first occurrence of Patau's Syndrome was recorded in the mid-1960s, by the human geneticist Klaus Patau and can be classified as a "genetic accident" in which there is one more chromosome, which can generate malformations in the nervous system (trisomy of chromosome 13), that it is possible to detect both in prenatal and neonatal and there is an estimate of 2.5% of fetuses able to exceed nine months of gestation (ROSA *et al.*, 2013; FLEITAS, 2014).

The human karyotype considered normal is formed by a set of chromosomes present in the cells of an organism that refers to the morphological characteristics of the chromosomes in number, types and shapes. In Patau Syndrome (figure 1), there are the presence of three chromosomes in group 13, which can cause external and internal malformations (SIERRA-SANTOS, 2001; ZEN *et al.*, 2008; IGNOROSA-NAVA; GONZÁLEZ-JUÁREZ, 2014), causing involvement of multiple organs and systems.

Figure 1 - Chromosome 13 trisomy - Patau syndrome



Source: Leite (*apud* LEME; LINO; WELLICHAN, 2019, p. 106)

The cure for Patau Syndrome is still unknown, even after symptom treatments and surgical interventions, but there are reports of life until the age of ten (PAZARBASI *et al.*, 2008). For the family, the diagnosis is a time to overcome challenges that may be decisive for the quality of life of a child affected by this syndrome.

In the case of AC, it was possible to monitor her development since birth in 2016 and since then, she continues to be treated and monitored by professionals from the Specialized Care Center in Deafblindness (CAE-Deafblindness), in the interior of the State of Paraná, Brazil.

The child case study in this article was seen at the CAE-Deafblindness (Specialized Care Center for Deafblindness) and has pre-linguistic (congenital) deafblindness, that is, she was born with visual and auditory impairment. Given the facts, it became necessary to think, to research with professionals who are specialists in the field of deafblindness and to discuss a method of communication proper for those who have not yet acquired any type of language. Thus, there are several methods for the communication of people with deafblindness, however, most of them need the child to be literate, which is not the case of Ac with four years and four months in May 2020.

Reys (2004) and Viñas (2004) emphasize that at birth or becoming deafblind, the first intervention and urgent measure with the child is to seek a functional communication system, so that communication is awakened or established as soon as possible. Thus, it is necessary to adapt a means of communication that meets the sensory characteristics of the deafblind child to be able to understand what is happening around them, proposing conditions to become an active and functional subject in the environment to which they belong.

In CAE-Deafblindness, the professional Mediating Instructor inserted for AC, the first sign of "mom" considering what was most familiar to her, such as her mother's touch, smell and voice (AC was also being stimulated by hearing, for using the Cochlear Implant), making it easier to understand the signal. In this way, the child, with the Tactile Libras system, will perceive the articulation, movement, location and orientation in space and time, with different information and will be able to build concepts and establish communication with her peers. In this process, it is necessary the knowledge of this system by the mediating teacher to maintain an effective communication with the deafblind person.

For the deafblind child to maintain dialogue and active participation, the family's commitment to stay at home is necessary, the guidance of the professional Mediating Instructor of CAE-Deafblindness, since Tactile Libras, for this child (AC), is indispensable for the development of her communication.

The following text was prepared with data from the official follow-up report of the AC Mediating Instructor, in 2019, with the permission of the CAE-Deafblindness and the child's parents. All ethical procedures were followed and identities were preserved. Records of consultations and reports from the family and the professional accompanying AC were consulted.

The authors' intention is to demonstrate, through the description of the report, the work developed with AC, diagnosed with the Patau Syndrome, identifying the challenges and possibilities that the child may experience when referred early to specialized care. In addition to reaffirming the need to search for the quality of life of the child and the family involved.

Case study

"AC was born in January of the year 2016, with 39 weeks, a girl, the result of a peaceful, planned and healthy pregnancy"[9] (LEME; LINO; WELLICHAN, 2019, p. 107). She had a series of complications at birth, which took her to the Intensive Care Unit (ICU), with a 25-day hospital stay, being subjected to several tests and with guidance for specific referrals in search of a diagnosis.

The diagnosis of Patau Syndrome was completed in 2018 and as a consequence, AC has no vision, hearing, brain malformation and polycystic kidneys. She underwent bowel surgery in the same year, had meningitis and for both cases, there was hospitalization.

[9] "AC nasceu no mês de janeiro do ano de 2016, com 39 semanas, uma menina, resultado de uma gestação tranquila, planejada e saudável"

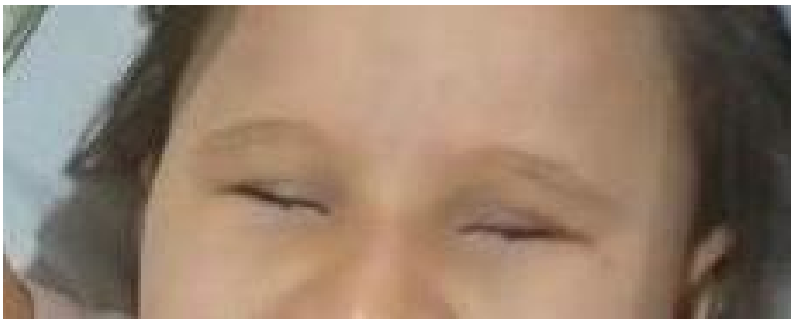
Even with the diagnosis of the syndrome and the little existing information on her development, "[...] the family's goal became the quality of life and the autonomy of the daughter"[10] (LEME; LINO; WELLICHAN, 2019, p. 109).

The meetings were sent to the CAE-Deafblindness of the city, twice a week, on Tuesdays and Thursdays, lasting one hour. The mother was present in all of them, and in some moments the aunt too, with whom the instructor, a mediation teacher, maintained a good relationship, which helped in the process of continuing the work developed there.

All activities were previously planned according to the needs and progress of AC, and considered specific care so that they were varied and stimulating for the student. They were always performed together with the mother so that she could continue working at home, since the visual and auditory stimulations must be performed daily. And especially in the case of Tactile Librasil, it is essential to work on the word at all times when it is used, therefore the importance of the family in this process.

The eye is formed from the 4th week of gestation, with the appearance and development of the optical grooves and optical vesicles, closed after birth by the myelination of the optic nerve fibers exposed to light (MOORE; PERSAUD, 2000) and the student AC presents in the visual area, microphthalmia (ICD-Q11.2) in both eyes, formed but small eye structures, eyeball with reduced AXL diameter, bilateral cataract with new vessels on the lens surface, which allows only the perception of light.

Figure 2 – Face of the eyes, which shows a decrease in the eyelid cleft



Source: Authors' collection

In the hearing area, AC has bilateral sensorineural deficiency (ICD H90.3), with a Cochlear Implant performed in 2018 in the left ear, which is still in the monitoring and adaptation phase in 2019, however, already showing possible progress.

[10] "[...] o objetivo da família passou a ser a qualidade de vida e a autonomia da filha"

Based on reports prepared by the Mediating Instructor teacher from previous years and also, by reports from the mother and aunt, it was possible to notice that AC after the implant adaptation period, had been able to carry out activities that were not successful before, such as stopping move around and pay attention to the sounds offered, turn her head to the side where the sound came from and turn again when the sound changed places. When dropping an object that made a noise when falling, AC turned to the exact side where the object had fallen, in an attempt to catch it, taking the little hands to the place where it was.

AC fits into deafblindness plus due to presenting Patau's Syndrome, which can cause severe mental retardation, in addition to severe malformations in the central nervous system and internal organs, in addition to very specific and frequent physical characteristics.

> People with deafblindness may have other associated disabilities and this condition was defined as deafblindness plus by Monaco (2004), that is, people who are born with deafblindness or were acquired and who have associations with intellectual or physical-motor disabilities and/or autism. The basic needs of people with deafblindness plus remain the same as those of a person with deafblindness without other associations. But when they present these associations, it is evident the importance of professionals who mediate communication with this population to know different ways to interact and promote access to communication, using alternative and/or expanded means and this requires even more specific training from this professional (MOLINA ; WATANABE; MAIA, 2018, authors' highlights).[11]

In cases of babies diagnosed with congenital deafblindness, as in the case of AC, the methodology adopted follows the *Van Jan Dijk* approach (1968) which established six stages for the development of language in children with deafblindness that represent a dynamic that incorporates social, sequential stimuli and cumulative (LEME; LINO; WELLICHAN; 2019), as described in the table (Chart 1) below:

Chart 1 – Stages for language development in children with deafblindness

PHASE 1 – NUTRITION	The first phase of the co-active approach. The objective of this phase is to make the child allow and accept a mediator, cooperating with the pedagogical activities proposed by him;
PHASE 2 – RESSONANCE	It consists of the interaction of people who act, communicate and move in the same harmony;

[11] As pessoas com surdocegueira podem ter outras deficiências associadas e essa condição foi definida como surdocegueira *plus* por Mônaco (2004), ou seja, pessoas que nascem com surdocegueira ou foi adquirida e que apresentam associações com deficiência intelectual ou físico-motora e/ou autismo. As necessidades básicas das pessoas com surdocegueira *plus* continuam sendo as mesmas de uma pessoa com surdocegueira sem outras associações. Mas quando apresentam essas associações, fica evidenciada a importância dos profissionais que mediam a comunicação com essa população conhecerem diferentes formas para interagir e promover o acesso à comunicação, utilizando meios alternativos e/ou ampliados e isso exige desse profissional uma formação ainda mais específica (MOLINA; WATANABE; MAIA, 2018, grifo nosso)

PHASE 3 – CO-ACTIVE MOVEMENT	Or hand on hand - characterized by the communicative expansion between the mediator and the child, using a wider space;
PHASE 4 – NON-REPRESENTATIVE REFERENCE	A reference object is used that initially will have no meaning for the child, however, it needs to be introduced before any activity is performed;
PHASE 5 – IMITATION	It represents the continuation of the coercive movement, however it is richer, as the child begins to recreate the assimilated symbolic elements in order to achieve the satisfaction of her needs;
PHASE 6 – NATURAL GESTURES	The hope is that the child will be able to initiate gestures on her own. Although these gestures are very concrete, it is the first attempt to talk about something that is missing.

Source: Devised by the authors based on Leme, Lino and Wellichan (2019)

By this approach, alternative communicative systems can assist in the development of the diagnosed and the family is an essential element to continue the work started in the specialized center. It is through the body movements of the other and through touch that the unhurt senses are stimulated.

At CAE-Deafblindness, the instructor-mediating teacher stimulated AC through *Van Dijk*'s coactive approach to stimulation with body movement, anticipation, routine and visual and auditory stimulation. Regarding the development of language for the acquisition of sound signal processing during the proposed activities and linguistic information.

In 2016 AC was submitted to Brainstem Evoked Response Audiometry (BERA) also known as *Brainstem Auditory Evoked Potential* (BAEP), which aims to examine the integrity of the auditory pathways, from the inner ear to the cerebral cortex. Presented smaller cochlea and auditory nerve and malformation in the corpus callosum (Agenesis).

Figure 3 - Total agenesis of the corpus callosum of AC

Souce: Authors' collection

Agenesis of the corpus callosum (AGCC) is a common cerebral malformation characterized by the absence (agenesis) of the corpus callosum that should have developed between the twelfth and twentieth week of gestation, and may be the result of genetic or environmental factors. The corpus callosum is responsible for the connection and communication between the two cerebral hemispheres, and causes epilepsy and delayed psychomotor development in the individual. (UTSUNOMIYA *et al.,* 1997).

The absence of the corpus callosum can be asymptomatic during the life of the human being or with slight imperceptible changes in clinical care and neurological exams. It can present in isolation or associated with other conditions: structural brain abnormalities (hydrocephalus or others), neurological dysfunction (epilepsy, macro or microcephaly, visual or auditory changes), extracerebral malformations, chromosomal abnormalities, and affects one child every thousand births. The prevalence of babies with agenesis in North America is 2.3% and in Latin countries it is unknown (UTSUNOMIYA *et al.,* 1997).

The delay in AC motor development is a relevant factor for her rehabilitation and functional capacity, being monitored since 2016 by a professional specialist in promoting interventions and capacitation for communication in people with deafblindness or multiple and sensory disabilities (MAIA, 2008)

In 2017, AC underwent 3D Resonance and Reconstruction and was referred to Speech Therapy and in 2018, underwent Cochlear Implant surgery in a specialized hospital in a city in the interior of São Paulo.

After the surgery to place a Cochlear Implant, AC underwent an adaptation process, being even reluctant to some sounds, at some moments in the beginning of the adaptation, however, it ended up adapting very well to her new reality.

Continuing the consultations initiated and reported in a previous article (LEME; LINO; WELLICHAN; 2019), the AC attendances focused on auditory stimulation, due to the greater possibility that the implant provided in the child. In addition, due to the frequency of activities carried out at CAE-Deafblindness, AC's acceptance of the activities was greater, even in the face of this exchange by the Mediating Instructor, who was recognized for her sensory touch, exploring her face and hands, accepting her contact.

AC presents perception of light, as previously described and thus, she is able to turn her head and eyes towards the illuminated objects presented, to fix her gaze and make brain connection with the lights she sees. That is how she perceive the light. She can quickly raise her hands to the illuminated object or extend them in front of her face as if she wants to touch the light or block it, according to its intensity. However, she only perceives such luminosity when there is a great contrast between light and darkness or with illuminated objects in a darkened room, because objects illuminated in a room with natural light, AC does not seem to notice.

When AC takes an object in her hands (figure 4), which she can do easily, she has the strength and dexterity to do so, she explores with a richness of details, runs her hands through the entire structure of each object, on the top and bottom sides at the same time, and explores inside parts, whenever she finds an opening to place her hands, she even realizes whether it is possible to place both hands inside the object or just one. Her dexterity with her hands allows the student to explore objects without knocking them over most of the time, holding firmly and safely.

Figure 4 – AC in activities with little balls

Source: Authors' collection

As for the different textures presented, not only through objects and toys, but also through fabrics, paint, modeling clay, water, creams and grains, AC quickly perceives the change from one texture to another, showing no reluctance for texture, despite having a preference for the softer ones, and seems to enjoy knowing, recognizing and exploring objects.

In a few months, she noticed the clue used by the Mediating Instructor for her recognition, which is a watch and a bracelet with two pendants on her left wrist, going to this clue whenever the professional receives her and greets her in the room, no longer being necessary to take her hands to the track, because with the simple touch of hands, AC already meets it with ease, greeting her with the touch of hands.

Upon hearing the well-known voice of the Mediating Instructor, AC searches for the clue she knows easily and then explores the clue as she usually explores objects, running her hands all over the place, above, below, waving the pendants on the bracelet and only after all this exploration of the track, she let go of the teacher's hands, as if waiting for the next activity.

In addition to the perception in the hands, the student also presents perception in the feet, using them, whenever possible, to explore the objects that surround her, especially when placed in the ball pool, where alone, she explores the nets around the cradle with hands and feet, picks up and holds the balls and "sinks" in the pool, turning the whole body between them.

AC recognizes the voice of her mother and other important people in her life, with whom she lives for a longer time. Sometimes she tries to remove the device from the implant. It is not possible to be sure if what bothers her is any sound or frequency modulated sound interference.

There are some sounds that can cause interference in the device, such as fans, cell phones and other noises, that is, in this case, it would not be exactly the sound that bothers you, but the interference caused in the implant device that can cause unpleasant sounds in her ears, which can bring noise to her hearing, or the simple physical discomfort of the device in her head. But when AC wants the device to be removed for some time, she takes her hands exactly where it is, repeatedly, trying to remove it, until she is able to reach her goal or someone else removes it for her.

With ease in the perception of sounds, AC when hearing something, usually turns in the direction of the sound. She likes all kinds of music, noises and sounds of toys, which arouses her curiosity whenever touched. She produces some sounds/babbles of speech, expressing her will at an adequate volume for a normal dialogue, not too loud or too low, which demonstrates that the hearing received is effectively adequate for her daily life.

In the study by Cader-Nascimento and Costa (2003, p. 145) the authors present the importance of alternative resources in the search for communication with deafblind people, and described that it is possible to

> [...] verify that the learning of alternative communication resources is possible with repercussions in other areas of human development. This leads to the assertion that if the objective of educational work is communication, then the exposure of deafblind people to all possible and available communication resources is the most viable and promising means to achieve the goal. If the goal of education is learning oral language, Tadoma proves to be an efficient method to achieve this goal. However, if the goal of education is your schooling, then in this case, your exposure to varied and distinct communication resources may not be the best way. This fact requires basic research to be carried out in search of new discoveries.[12]

The medical team at the hospital where the Cochlear Implant was performed and which follows the progress of AC, suggested that a Speech Therapist introduce the Tactile Libras method together with her speech-language work during speech therapy sessions.

The basic information necessary to carry out such work with the child was initially transmitted to the mother, however, due to lack of time, it was not possible for the teacher of the CAE-Deafblindness Mediating Instructor to speak directly with the Speech Therapist responsible for CA care, which should occur later, by the professional (Mediating Instructor) who will continue this work in 2020, as this methodology must be worked with a multidisciplinary approach for the collaboration of knowledge and exchange of information relevant to the development of AC.

Therefore, the communication attempt was initiated by Tactile Libras[13] with the word "mama", AC still did not correspond to the methodology, seeming not to have understood this idea yet. However, it is important to insist on this type of communication, because at first it is the method that best suits the child's learning needs and conditions.

[12] [...] constatar que a aprendizagem de recursos alternativos de comunicação é possível com repercussão nas outras áreas do desenvolvimento humano. Isto leva a afirmar que se o objetivo do trabalho educacional for a comunicação, então a exposição dos surdocegos a todos os recursos possíveis e disponíveis de comunicação é o meio mais viável e promissor para atingir a meta. Se o objetivo da educação for a aprendizagem da linguagem oral, o Tadoma se mostra um método eficiente para alcançar esta meta. No entanto, se o objetivo da educação for sua escolarização, então neste caso, sua exposição a recursos variados e distintos de comunicação pode não ser o melhor caminho. Este fato demanda a realização de pesquisas básicas na busca de novas descobertas.

[13] Tactile sign language is the adaptation of Libras, used by the deaf, to people with deafblindness, by its use by touch. These are signs performed on one or both hands. The position, orientation and configuration of the hands for the formation of the signals are the same and changes the signaling space and the form of reception.

Figure 5 – Tactile materials used by AC

Source: Authors' collection

Tactile Libras is one of the forms of communication used by deafblind people, using touch. Just like Tadoma[14], which also uses touch, but it captures the sense of vibration of the voice when it touches the mouth or neck of the Guide-Interpreter. Another resource used for deafblind communication is the manual alphabet used to compose messages in Braille System or the Moon System or Alphabet[15], that uses reliefs and the pictographic system itself that uses reliefs and the pictographic system itself that uses symbols and figures that uses symbols and figures (MAIA, 2008; GALVÃO, 2013; ALMEIDA, 2017; WATANABE, 2017; ANDRADE, 2018).

The aforementioned resources are being presented to AC and studied during the consultations to verify, in practice, which achieve greater results, in view of the child's particularities.

For 2020, the continuity of AC in the care at CAE-Deafblindness was recommended for visual and auditory stimulation, two to three times a week, for one or two times at a time, according to the availability of the school and family, in addition to maintaining other therapies such as Physiotherapy, Speech Therapy, Equine Therapy and Occupational Therapy sessions, in addition to the necessary medical follow-ups.

[14] Tadoma consists of a method of receptive language through touch, that is, the deafblind person places his hands and thumb on the face and mouth of the person who is speaking, which makes it possible to interpret the emission of sounds through mouth movements and vocal cords.

[15] The Moon Alphabet was invented by Dr. William Moon in 1847 in England. It consists of a relief reading based on the standard alphabet, with 14 characters used at various angles, with clear outlines. It is considered easier than Braille, and more used by the elderly and people who have lost their sight throughout their lives.

Final considerations

Although one of the symptoms of Patau Syndrome is Intellectual Disability, AC is a smart child, who easily learns and understands the proposed activities, within what is possible to understand so far, taking into account deafblindness and the syndrome.

AC expresses her opinion of what she wants and likes in her own way, making herself easily understood when she wants to end an activity, lie down or leave, for that, means were tested and monitored to see which way would bring understandable return.

AC represents a case of overcoming the initial diagnoses and studies regarding Patau Syndrome, which demonstrates how important the stimulation works are and can bring quality of life for children in these conditions. Even in the face of deafblindness aggravated by the diagnosis of a rare syndrome, treatments, strategies and resources need to be used and explored, as they can bring satisfactory results and quality of life for both AC and her family.

For parents, the feelings caused by the initial diagnosis in the face of a disability or syndrome cannot be ignored. In a mixture of feelings it is necessary to experience each one of them, in order to understand later how much each step (of stimulus or search for professionals and understanding about cognitive and motor development) will be important for your child's quality of life and for the whole family.

With a fundamental role to continue the work carried out at CAE-Deafblindness, the family needs to be guided on the best forms and strategies found in the literature or by the experience of professionals during the consultations. In addition, it provides us with important information regarding the child's development, since there is an absence of AC speech and an effective communication system, necessary situations to be worked on by a multiprofessional team, therefore, it is necessary to work together and provide guidance.

Perspectives even in the face of cases such as AC are necessary for everyone involved: for Science and health professionals, because they instigate new studies, procedures and discoveries, which allows treatments to be offered or tested; for the family, it is the opportunity to love, know, acknowledge, commit, get involved and “raise” their child regardless of the time that has been foreseen for her (in medical literature), depending on your diagnosis; for the community in general, which accompanies cases such as AC, with possibilities for reflection in various segments of society.

The work developed by the Mediating Instructor is of singular importance in the AC development process and raises important points regarding the formation and performance of this professional in the face of complex situations such as that of AC. Education and

capacitation associated with the desire to understand what was not said are essential in the process of stimulating communication.

The development of AC reaffirms the need for new studies and the search for new methodologies and improvements in the area of deafblindness and in rare syndromes such as that of Patau, which is still little discussed in national and international literature.

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REFERENCES

ALMEIDA, W. G. A guia-interpretação no processo de inclusão do indivíduo com surdocegueira. **Educ. rev.**, Curitiba, n. 65, p. 167-181, set. 2017. Available: https://www.scielo.br/pdf/er/n65/0104-4060-er-65-00167.pdf. Access: 25 May 2020.

ALMEIDA, C. A. F. A aquisição da linguagem por uma surdocega pré-linguistica numa perspectiva sociocognitivista. **Papia: Revista Brasileira de Estudos do Contato Linguístico**, v. 17, 2007. Available: http://revistas.fflch.usp.br/papia/article/view/2031/1853. Access: 26 May 2020.

ANDRADE, A. F. Surdocegueira, cartografia e decolonialidade. **Psicol. cienc. prof.**, Brasília, v. 38, n. 3, p. 595-610, set.2018. Available: https://www.scielo.br/pdf/pcp/v38n3/1982-3703-pcp-38-3-0595.pdf. Access: 21 May 2020.

BRASIL. **Educação Infantil, saberes e práticas da inclusão**: dificuldade de comunicação e sinalização – surdocegueira/múltipla Deficiência sensorial. Brasília, DF: MEC/SEESP. Caderno 5, 2002.

CAT. Comitê de Ajudas Técnicas. **Ata da Reunião VII**, de dezembro de 2007 do Comitê de Ajudas Técnicas. Secretaria Especial dos Direitos Humanos da Presidência da República (CORDE/SEDH/PR), 2007. Available: http:// http://www.mj.gov.br/corde/comite.asp. Access: 02 June 2020.

CADER-NASCIMENTO, F. A. A. A.; COSTA, M. da P. R. da. **Surdocegueira e a peculiaridade do professor na função de guia-intérprete**. Das margens ao centro: perspectivas para as políticas e práticas educacionais no contexto da educação especial inclusiva. Araraquara, SP: Junqueira & Marin, 2010.

FLEITAS, L. Síndrome de Patau o trisomía 13: reporte de caso. **Rev. Nac.**, Itauguá, v. 6, n. 2, p. 55-60, 2014. Available: http://scielo.iics.una.py/pdf/hn/v6n2/v6n2a06.pdf. Access: 21 May 2020.

GALVAO, N. C. S. S.; MIRANDA, T. G. Atendimento educacional especializado para alunos com surdocegueira: um estudo de caso no espaço da escola regular. **Rev. bras. educ.**

espec., Marília, v. 19, n. 1, p. 43-60, mar. 2013. Available: https://www.scielo.br/pdf/rbee/v19n1/04.pdf. Access: 20 May 2020.

IGNOROSA-NAVA, C. A.; GONZALEZ-JUAREZ, L. Cuidados paliativos para una muerte digna: Estudio de un caso. **Enferm. univ.**, México, v. 11, n. 3, p. 110-116, set. 2014. Available: http://www.scielo.org.mx/pdf/eu/v11n3/v11n3a6.pdf. Access: 2 June 2020.

LEME, C.G.; LINO, C.C.T.S.; WELLICHAN, D.S.P. A comunidade de uma criança acometida pela Síndrome de Patau. *In*: PAPIM, A. A. P.; ARAUJO, M. A. (Orgs.) **O processo de construção da educação para a diversidade:** um panorama científico. Porto Alegre: Editora Fi, 2019. Cap.5. p. 105-123.

MAIA, S. R *et al*. **Estratégias de ensino para favorecer a aprendizagem de pessoas com Surdocegueira e Deficiência Múltipla Sensorial**: um guia para instrutores mediadores. São Paulo: Grupo Brasil/CIDA, 2008.

MOLINA, K. S. M.; WATANABE, D. R.; MAIA, S. R. Surdocegueira: termos, conceitos e formas de comunicação. *In:* MANZINI, E. J. *et al.,* (org.). **Política d e para educação especial**. Marília: ABPEE, 2018.

MÔNACO, C. A mediação para a pessoa com surdocegueira, **apostila do curso de Formação de Mediadores**, projeto Girassol, Grupo Brasil e Embaixada do Canadá. George Brown College, 2004.

PAZARBASI, A. *et al.* Prenatal Diagnosis of Translocation 13;13 Patau Syndrome: Clinical Features of two cases. **Balkan Journal of Medical Genetics**, v. 11, n. 1, p. 69-73, nov. 2008. Available: https://www.researchgate.net/publication/236210795_Prenatal_Diagnosis_of_Translocation_1313_Patau_Syndrome_Clinical_Features_of_Two_Cases. Access: 03 May 2020.

REYES, D. A. La sordoceguera: uma discapacidad singular. *In*: REYES D. A. **La sordoceguera***:* um análisis multidisciplinar. Madrid: ONCE, 2004. p. 135-159.

ROSA, R. F. M.; et al. Achados gestacionais, perinatais e familiares de pacientes com síndrome de Patau. **Rev. Paul Pediatr**, v. 31, n. 4, p. 459-465, 2013. Available: http://www.scielo.br/pdf/rpp/v31n4/pt_0103-0582-rpp-31-04-00459.pdf. Access: 03 May 2020.

SIERRA SANTOS, L. *et al*. Un síndrome de Patau con una supervivencia que supera los pronósticos. **Medifam**, v. 11, n. 8, p. 70-74, set.2001. Available: http://scielo.isciii.es/scielo.php?script=sci_arttext&pid=S1131-57682001000800009&lang=pt. Access: 02 May 2020.

UTSUNOMIYA, H. *et al*. Dysgenesis of the corpus callosum and associated telencephalic anomalies: MRI. **Neuroradiology**, n. 39, p. 302–10, 1997. Available: https://link.springer.com/article/10.1007/s002340050414. Access: 02 May 2020.

VINÃS P. G. La educación de las personas sordociegas. Diferencias y proceso de mediación. *In*: REYES D. A. **La sordoceguera***:* um análisis multidisciplinar. Madrid: ONCE, 2004.

WATANABE, D.R. **O estado da arte da produção cientifica na área da surdocegueira no Brasil**: 1999 a 2015. 2017. 262 f. Dissertação (Mestrado em Educação) - Universidade do Estado de Paulo, USP, SP. Available: https://www.teses.usp.br/teses/disponiveis/48/48134/tde-13062017-112304/publico/DALVA_ROSA_WATANABE_rev.pdf. Access: 02 May 2020.

ZEN, P. R. G. *et al.* Apresentações clínicas não usuais de pacientes portadores de síndrome de Patau e Edwards: um desafio diagnóstico**? Rev. paul. pediatr.**, São Paulo, v. 26, n. 3, p. 295-299, set. 2008. Available: https://www.scielo.br/pdf/rpp/v26n3/15.pdf. Access: 02 May 2020.

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