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Case reports

Mitochondrial disease and augmentative and alternative communication: a clinical case study

Doença mitocondrial e comunicação suplementar e alternativa: estudo de caso clínico

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ABSTRACT

There are few Brazilian speech-language pathology studies about patients with mitochondrial disease diagnostic. The genetic metabolically clinical diseases are very frequent and show symptoms that demand speech-language pathology interventions (miofunction oral changes, auditory and complex communication needs). In this paper, the results indicate the possibility of augmentative and alternative communication work with this population. Objective is to describe the augmentative and alternative communication use results of the work with a child with mitochondrial disease. The therapeutic proceedings focus on the social language meaningful and contextualized activities use, with the help of diary activities photos and the Picture Communications Symbols. The results shows an increase on individual oral language function, when it maximizes discursive practice, especially when it has a communicative intention on the interactional context.

Keywords: Speech, Language and Hearing Sciences; Language; Mitochondrial Diseases; Communication Aids for People with Disabilities

RESUMO

Poucos são os estudos fonoaudiológicos brasileiros sobre pacientes com o diagnóstico de doenças da cadeia respiratória mitocondrial. Esse quadro clínico é uma das doenças genéticas do metabolismo mais frequentes e apresenta sintomas que demandam intervenções fonoaudiológicas (alterações miofuncionais orais, auditivas e dificuldades na aquisição da linguagem oral). Neste estudo, articula-se a possibilidade de trabalho com esses sujeitos na abordagem da comunicação suplementar e alternativa. Objetivou-se descrever os resultados da utilização da comunicação suplementar e alternativa no atendimento de uma criança com doença mitocondrial. Os procedimentos terapêuticos enfatizaram o uso social da linguagem por meio de atividades significativas e contextualizadas, com o apoio de fotos e de figuras do Picture Communication Symbols de atividades cotidianas. Os resultados obtidos apontaram aumento na funcionalidade da linguagem oral do sujeito, ao favorecer suas práticas discursivas, especialmente quanto à intenção comunicativa no contexto interacional.

Descritores: Fonoaudiologia; Linguagem; Doenças Mitocondriais; Auxiliares de Comunicação para Pessoas com Deficiência

INTRODUCTION

Mitochondrial respiratory chain disorders, also known as mitochondrial cytopathies, mitochondrial diseases or oxidative phosphorylation disorders, are a group of clinically heterogeneous inherited metabolic disorders caused by defects in mitochondrial ATP production[1].

Establishing a diagnosis of these disorders is challenging due to the wide range of possible clinical features, the non-standardization of diagnostic criteria, and the presence of frequent secondary oxidative phosphorylation disorders. The diagnostic criteria are: clinical assessments and genetic and laboratory tests[2].

The main clinical presentations involving central nervous system tissues are ataxia, myoclonia, psychomotor retardation, psychomotor regression, dystonia[3], muscle weakness, intolerance to physical exercise, sensorineural hearing loss, movement coordination issues, seizures, learning disabilities, optic atrophy, pigmentary retinopathy, ophthalmoplegia, cardiomyopathy, diabetes, autism, growth atrophy, peripheral neuropathy, dementia, and multiple lipomas, in addition to frequent episodes of respiratory issues[4].

Because they are considered rare diseases, mitochondrial disorders (MD) did not use to be scientifically investigated in a systematic manner. However, over the last 15 years there has been a significant increase in scientific research into the issue. Epidemiological studies have been conducted with the aim of confirming that mitochondrial diseases are, indeed, among the most prevalent genetic metabolic disorders, representing substantial financial investments in the healthcare industry[5]. The prevalence is estimated at 10 to 15 cases per 100,000 births, similar to that of well-known neurological disorders, such as amyotrophic lateral sclerosis (ALS) and muscular distrophies[1].

The prognosis for MD is poor, with high mortality rates and premature death[6]. Mortality ranges from 10% to 50%/year after diagnosis, among children, and from 5% to 20%/year after the beginning of clinical treatment among adults[7]. Cases that show good clinical progress are rare[8,9].

Brazilian clinical speech & language studies into this disease are basically associated with hearing losses[10] and orofacial motricity disorders[11]. However, in addition to oral (primarily due to hypotonia) and auditory myofunctional disorders, other clinical manifestations include learning (reading and writing) difficulties as well as spoken language disorders, which justify a therapeutic approach based on Augmentative and Alternative Communication (AAC)[12].

AAC may be employed as a supplementary resource to existing speech or as a replacement for non-functional speech, by means of facial expressions, gestures, graphic symbols, pictures and/or writing, as a means of promoting interpersonal communication so as to benefit social interaction, school performance and self-esteem[13].

The purpose of this paper is to present the results of employing AAC in the treatment of a child with mitochondrial disease.

PRESENTATION OF THE CLINICAL CASE

This study has been approved by the home institution's Research Ethics Committee as per report no. 1.227.183 CAAE: 48729315.5.0000.5482. The participant's legal custodians have signed the Free and Informed Consent Term, thus agreeing with the conduction and disclosure of the research and its results.

Patient's history

The patient is an 11-year-old boy diagnosed with mitochondrial disease who has been under the care of a specialized genetics clinic since he was one year old, and has since then been taking medications to replace the amino acids his body cannot produce: l-carnitine, thiamine, coenzyme, vitamins C and E, riboflavin and folic acid. He undergoes periodic hearing and vision evaluations, with results within normal standards.

He goes to a regular private school and during the study was attending the second and third years of elementary school. The school's main complaint was about his behavior, especially in regard to his interaction with other students and his aggressiveness, both of which were associated with the patient's communication difficulties.

His communication with his family was based mostly on oral language, with the support of some non-symbolic gestures like pointing[12], always associated with vocalizations. His relatives reported having trouble understanding him.

The speech-language evaluation found: presence of rare vocalizations or monosyllabic words (as the patient's phoneme articulation is limited, it is often difficult to understand the word) and use of gestures and body language, with low functionality due to his limited discursive autonomy and communicative

intentions, associated with repetitive speech, i.e., he repeats his own speech. All this led to irritability – he would slap his interlocutor or his own head – whenever he was not understood.

RESULTS

The material analyzed regards the speech-language therapy based on the AAC approach during the period between May 2014 and May 2015, in 45-minute sessions conducted once a week. The therapeutic procedures emphasized the social use of language through meaningful, contextualized activities, supported by photos and drawings of daily activities provided by the Picture Communication Symbols (PCS), the system employed by AAC. The activities were chosen according to the patient's interests, and sometimes he picked the symbol himself. Games, history books and toys (ball, toy cars) were explored, while at the same time presenting the pictures so they might assist the communication. The child used the pictures spontaneously, usually by pointing, but any gesture, such as looking, grabbing, or even biting, was accepted as a response and interpreted orally by the therapist.

During the therapeutic process a notebook-shaped communication board was created and used as a means of supporting oral language within different interactional contexts (speech therapy, school and home activities), represented by theme boards (pictures organized according to themes associated to the patient's most significant experiences).

The speech-language therapy included monthly guidelines given to the patient's educators, with the aim of helping them make use of AAC strategies, thus maximizing the child's interactions. These strategies basically refer to the presence of pictures in therapy, used the therapist along with speech, and at times spontaneously pointed at or picked up by the patient himself. These pictures were composed of drawings on paper representing activities usually associated with the context.

The family approach consisted of individual interviews with the parents, as well as their participation in some sessions.

The results of this intervention will be presented in accordance with the following parameters: communicative intention, discursive autonomy, language functionality and non-verbal resources.

The use of AAC led to significant changes: after one year of therapy, the patient expanded his vocalizations with the support of AAC symbols, which proved to be effective resources to supplement speech. His communicative intention improved, leading to some discursive autonomy, and, consequently, an increase in the functionality of his spoken language.

The following clinical excerpts illustrate these results:

1. Therapist (T) and Subject (S) sitting down, interacting with the help of the AAC board (Appendix 1).
S: Car (points at the car picture)
T: Do you want to play with the car?
S: aaah (agreeing)
T: Whose is that? (pointing at the video game picture)
S: Teteu (that's what he calls himself)
T: it is yours?
S: (unintelligible)
T: Who do you play with?
S: uuh (in a low, unintelligible voice)
T: Who played with you?
S: Mommy (with no support from the picture)
T: Mommy played video game?
S: Yeah
T: What did you do this week?
S: (Points at the video game picture and looks at T)
T: Video game?
S: aaah (agreeing)
T: You played video game? With whom?
S: Daddy
T: With daddy? Does daddy know how to play?
S: (makes a "no" gesture with his head)

2. (S leafing through the notebook).
S: Uh! (points to a picture representing a TV show he likes a lot).
T: What is that? BBB! It is over, isn't it?
S is facing three pictures representing games that were shown to him at a previous moment.
S: ahh
T: What do you want?
S: *Cocah* (for "*brincar*", the Portuguese word for "play") – looks at T
T: Do you want to play?
S: uuhh (agreeing)
T: What do you want to play?
S: eeh (agreeing) – looks and points at the picture representing the game "Pop-up Pirate" and looks at T.
T: Do you want to play with the Pirate?
S: yeah

The pictures prove to be an effective resource to encourage dialogue, both when introduced by T (excerpt 1) as when introduced by S (excerpt 2).

3. (S and T are at the beginning of the session)
S: Hiiii
T: Hi!
S: *tetem*? (for "*tudo bem*?", Portuguese for "all right?")
T: I am, and you?
S: hiiii, *tetem*?
S takes dialogue turns; however, when unaided by the pictures his speech tends to be repetitive.

4. (S and T are talking without the AAC board).
S: *Pexi, pexi* (a sound similar to "*peixe*", the Portuguese word for "fish")
T: *Peixinho*? ("Little fish?")
S: uuhhhh
T: What?
S: Swim
T: The little fish swims?
(Dialogic rupture)

5. (S and T are talking, using the AAC board)
S: Daddy (points at the picture representing the father)
T: Daddy is waiting for you outside.
S: Car
T: He's in the car?
S: aaah (clapping affirmatively)

It may be seen that in both contexts (excerpts 4 and 5) S takes on the role of active interlocutor. However, T's interpretations of his utterances are benefitted by the pictures, promoting a discursive symmetry, i.e., a consensual picture-interpretation correspondence. The interaction elements in the exchange structure are symmetrical when understood as a relative agreement expressed through accepting the interpretation for negotiation, which will become an object of knowledge[13].

6. (S and T are talking, using the AAC board)
T: Which one is your bicycle? (showing two pictures: a bicycle and a tricycle).
S: *Bicique* (pointing to the bicycle)
T: Bicycle!
S: uuuuh (showing agreement in his intonation)
T: I see
S: eeeh (with the same intonation)

During the therapeutic process, using AAC helped increase the patient's vocabulary: he repeats new words with the aid of the pictures, trying to articulate them adequately and generalizing their use in different contexts. In the excerpt above we may even notice a semantic subcategorization.

7. (S and T are talking, without the AAC board).
S: aaah
T: What?
S: *Sussuh* (looking out the window)
T: Sun?
S: aaah (pointing to the window)
T: No sun today. It's raining!
S: aaah, *sussuh*
T: There is no sun today! There is a lot of rain...

The intelligibility of his speech is still hindered by his orofacial myofunctional limitations, and this is when the AAC symbols are particularly useful to give effective support to the dialogue, as in the following excerpt:

8. (S and T are talking, with the AAC board on the table).
S: *Cocah*
L: Uh? (not understanding)
S: *Cocah* (repeating, and pointing at the picture representing a Coca-Cola)

As the treatment progressed, the patient's use of the AAC expanded to include his family, so that, whenever he wants to say something, he now seeks the support of his notebook in order to express his desires, needs and intentions. It bears stressing that his mother is usually his interlocutor of choice in these instances.

DISCUSSION

In this study, AAC proved effective to establish and sustain the dialogue. However, to make this possible it was necessary to constantly attribute meaning to the patient's utterances[14], which are still linguistically precarious. That is, so as to go beyond mere metalinguistic activities (naming, repeating) and towards a discursive structure aimed at expressing subjective content, even faced with the significant limitations imposed by the patient's pathology[15]. Only then may the AAC symbols evolve from being mere signs to acquiring a polysemic, variable and flexible character within the intersubjective context between interlocutors[14], stepping away from the conception

that overvalues the formal aspects of language to the detriment of its discursive function and effectiveness in interactions[15].

Thus the use of AAC in the present case became an alternative to low-functionality spoken language, allowing for the development of some discursive autonomy, albeit still precarious.

On the other hand, the significant presence of unintelligible elements in the subject's speech is compatible with the orofacial myofunctional disabilities inherent to the pathology[11]. However, such limitation did not prevent the dialogue, as the T sought to interpret these segments according to the interactional context, which proved effective, particularly when the AAC was employed[16].

CONCLUSION

The results of the present study indicate that AAC, associated with a non-formalistic conception of language (i.e., that which privileges the expression of subjectivity) has expanded the functionality of the subject's language by benefitting his discursive practices, especially regarding the communicative intention in the interactional context.

Therefore, the authors recommend that further research into ACC focus on and give emphasis to a conception of language that underpins its procedures, which is not typically the case in the existing literature on this issue.

REFERENCES

1. Rodenburg RJT. Biochemical diagnosis of mitochondrial disorders. J Inherit Metab Dis. 2011;34:283-92.
2. Rötig A. Human diseases with impaired mitochondrial protein synthesis. Biochim Biophys Acta. 2011;1807:1198-205.
3. DiMauro S, Rustin P. A critical approach to the therapy of mitochondrial respiratory chain and oxidative phosphorylation diseases. Biochim Biophys Acta. 2009;1792(12):1159-67.
4. Nasseh IE, Tengan CH, Gabbai AA. Doenças Mitocondriais. Rev Neurociências. 2001;9(2):60-9.
5. DiMauro S, Hirano M, Kaufmann P, Mann J. Mitochondrial psychiatry. In: DiMauro S, Hirano M, Schon EA, editors. Mitochondrial Medicine. Abingdon: Informa Healthcare. 2006. p. 261-77.
6. Chinnery P, Majamaa K, Turnbull D, Thorburn D. Treatment for mitochondrial disorders. Cochrane DatabaseSyst Rev. 2006a;1(1):CD004426
7. Naviaux RK, Nguyen KV. POLG mutations associated with Alpers' syndrome and mitochondrial DNA depletion. Ann Neurol. 2004;55(5):706-12.
8. Zeviani M, Carelli V. Mitochondrial disorders. Curr Opin Neurol. 2003;16(5):585-94.
9. Debray FG, Lambert M, Lortie A, Vanasse M, Mitchell GA. Long-term outcome of Leigh syndrome caused by the NARP-T8993C mtDNA mutation. Am J Med Genet A. 2007;143A(17):2046-51.
10. Bonassa-Mourato AC, Lança SM, Iório MCM, Assencio-Ferreira VJ. Comparativo da Progressividade das Deficiências Auditivas em Pacientes Portadores de Miopatias Metabólicas. Rev. CEFAC. 2002;4(1):67-70.
11. Mendes LTB, Ferreira ZCF. Fase Oral da Deglutição em Pacientes com Doenças Mitocondriais. 19º. Congresso Brasileiro e 8º. Internacional de Fonoaudiologia. [base de dados da internet]. 2011. São Paulo – SP. p.170. Disponível em: http://www.sbfa.org.br/portal/anais2011/trabalhos_select.php?tt=Busca&id_artigo=170
12. ASHA: American Speech Language Hearing Association. What is Augmentative and Alternative Communication? Disponível em: http://www.asha.org/public/speech/disorders/AAC/#what_is
13. Lier MF, Palladino RRR, Maia EAM. Simetrização e Assimetrização na Comunicação Pré-linguistica. In: Rojo RHRR, Cunha MC, Garcia ALM. Fonoaudiologia & Linguistica. São Paulo: EDUC; 1991. p.11-24.
14. Vasconcellos R. Clinica de linguagem: da entrevista à implementação da CSA, suas heterogeneidades e a inclusão Escolar. In: Chun RYS, Reily L, Moreira EC. Comunicação Alternativa: Ocupando Territórios. São Carlos: Marquezine & Manzini: ABPEE; 2015. p. 129-43.
15. Panhan H. A tecnologia no espaço clínico e terapêutico fonoaudiológico. Temas sobre Desenvolv. 2001;10(58/59):56CE-8CE.
16. Vasconcellos R. Clinica de linguagem e seus efeitos singulares no encontro entre falas de terapeuta e paciente com paralisia cerebral. In: Passerino LM, Bez MR, Pereira ACC, Peres A. Comunicar para Incluir. Porto Alegre: Ponto e Virgula Editora; 2013. p. 313-28.

APPENDIX 1. Augmentative and Alternative Communication Board