ASSISTIVE TECHNOLOGY FOR COMMUNICATIONAL ACCESSIBILITY IN MACHADO-JOSEPH DISEASE

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ABSTRACT

Machado Joseph Disease (MJD), or type 3 spinocerebellar ataxia, is a neurodegenerative and autosomal dominant disease, which manifests with progressive ataxia, gait imbalance, diplopia and phonation disorders that result in accessibility barriers. This study aims to propose interventions to improve communicational accessibility in a 53-year-old patient with MJD. Data collection was performed after informed consent and the patient was followed-up for over one year, including assessment of quality of life and semi-structured interviews conducted with the patient, family members, and caregiver. The main difficulties were diplopia, motor coordination problems, and slowed speech. Based on the results, interventions were proposed based on the patient's needs and scientific literature to enhance her communicational accessibility. Among these interventions, highlights include the use of Google Assistant, increasing zoom and font size on the smartphone, and replacing typed texts with audio recordings, which improved the patient's communication, considering the disease stage, with an impact on quality of life. It is concluded that MJD is a disabling condition underrepresented in accessibility research. However, the importance of continuous research to adjust strategies to the progression of the disease and new communication needs over time is emphasized.

Keywords: Machado-Joseph Disease. Spinocerebellar Ataxias. Communication. Self-Help Devices. Quality of Life

1 INTRODUCTION

Machado-Joseph Disease (MJD), the spinocerebellar ataxia type 3 (SCA3), is an autosomal dominant neurodegenerative disease that is part of the group of polyglutamine (polyQ) diseases, which includes Huntington's disease, spinobulbar muscular atrophy, dentatorubral-pallidoluysian atrophy, and other spinocerebellar ataxias (SCA) in addition to SCA3 [1-4]. MJD affects approximately 1 person per 50,000-100,000 inhabitants, being the most common hereditary ataxia in the world and is progressive and irreversible, with no curative treatment available to date 3. In Brazil, a cohort study of patients with spinocerebellar ataxia in the state of Rio de Janeiro demonstrated that of the 128 confirmed cases of SCA by molecular tests, MJD accounted for 83.6% of the cases. Furthermore, it was observed that among the 107 patients with MJD, about 57.9% were women, approximately 72.9% were white, and the mean age of symptom onset was in the 4th decade of life (40.4 years) [5].

Studies on the pathogenesis and imaging of the disease have identified that some areas of the central nervous system (CNS) undergo a degeneration process leading to atrophy of the cerebellum, brainstem, basal ganglia, thalamus, some cranial nerves, spinal cord, and even the relatively affected cerebral cortex [2,6]. This process is believed to be caused by the accumulation of mutant ataxin-3, ubiquitin, and other proteins within the nuclei of neurons in the affected areas [2]. These proteins are abnormally generated due to a mutation in exon 10 of the ATXN3 gene, characterized by unstable cytosine-adenine-guanine (CAG) trinucleotide repeats. The number of repeats in the mutant allele can vary from 56 to 87 in patients with MJD. This difference can result in clinical heterogeneity among MJD patients and even within the same family [2,3].

Despite clinical variability, the main clinical feature of MJD is progressive ataxia with progressive gait imbalance accompanied by vestibular and speech difficulties. Additionally, over time, various visual and oculomotor problems such as nystagmus, saccadic intrusions, slow saccades, disconjugate eye movements, ophthalmoplegia with lid retraction, diplopia, eye bulging, and proptosis arise. Patients may also develop facial and temporal muscle atrophy, dystonia, spasticity, and amyotrophy. Dementia is not typically found, even in the advanced stages of the disease, with most patients exhibiting preserved cognitive function, despite evidence of subcortical dysfunction

and mild cognitive impairment in MJD. Survival after disease onset ranges from 20 to 25 years [4].

Complications associated with the disease, such as dysarthria, lack of motor control, and ophthalmoplegia, contribute to the accessibility difficulties faced by MJD patients, in accordance with the Brazilian Inclusion Law for People with Disabilities (Statute of the Person with Disabilities), which categorizes a person with a disability as one who has long-term physical, mental, intellectual, or sensory impairments. These impairments, when combined with one or more barriers, may hinder their full and effective participation in society, creating inequality compared to the living conditions of other people. This same law also defines the concept of assistive technology, which refers to products, equipment, devices, resources, methodologies, strategies, practices, and services aimed at promoting functionality related to the activity and participation of a person with disabilities or reduced mobility, thereby aiming at their autonomy, independence, quality of life, and social inclusion [7].

In this context, it can be observed that, due to the progression of MJD, patients will suffer from multiple combined disabilities, mainly of a physical and sensory nature that will impair their communication. Although this difficulty is widely recognized, studies investigating alternatives to improve accessible communication for these patients are scarce. In this sense, this study aims to understand the communicational accessibility needs of a 53-year-old female patient with MJD through the perspective of the patient herself, her family members, and her caregiver, and to explore communicational accessibility interventions, through assistive technologies, that meet the needs of this patient, aiming to optimize her quality of life.

2 MATERIALS AND METHODS

This study was evaluated by the Centro Universitário de Volta Redonda Research Ethics Committee and approved with the number 5.692.023.The Informed Consent Form and authorization for the recording of images and voice of the research participants (the patient, caregiver, and family members - the patient's mother and spouse) were requested, along with authorization for the research at the FOA Dr. André Sarmento Bianco Polyclinic, located in Volta Redonda, Rio de Janeiro, and data collection from the medical records. Also, for the clinical follow-up of the patient, at least two home visits were planned to map the patient's environment and the available communication accessibility conditions, such as internet access and available electronic equipment.

The patient also responded to the Brazilian version of the Medical Outcomes Study 36-Item Short-Form Health Survey (SF-36) quality of life questionnaire at the beginning of the project and after one year of follow-up to assess the patient's quality of life perception at the start and end of the study. The SF-36 is a generic quality of life assessment instrument consisting of a multidimensional questionnaire formed by 11 questions and 36 items encompassing 8 dimensions or domains: 10 items on functional capacity, 4 items on physical aspects, 2 items on pain, 5 items on general health status, 4 items on vitality, 2 items on social aspects, 3 items on emotional aspects, 5 items on mental health, and 1 item comparing the patient's current health perception to that of one year ago. The analysis of the SF-36 questionnaire results is based on the score in each domain, ranging from 0 to 100, with the worst score being zero and the best being 100. The scoring follows the SF-36 questionnaire standardization and Raw Scale calculation.

Semi-structured interviews with open-ended questions about the communication difficulties faced by the MJD patient and her perception of the proposed communication interventions at the beginning and end of the one-year project were also conducted. The patient's caregiver, who has been in service for two years, and her family members (mother and spouse) were also interviewed about their perception of the patient. The interviews were recorded in audio and video and transcribed into text.

Based on the responses obtained from the SF-36 questionnaire and the interviews, intervention proposals were developed to enhance the patient's communication accessibility. These proposals include resources and activities, mediated or not by technology, that promote independence and autonomy for those who have difficulty receiving or transmitting messages and who require specific adaptations for effective communication.

3 RESULTS

The patient, a 53-year-old female, was seen at the genetics outpatient clinic of the University Center of Volta Redonda in 2016 with a family history of MJD, as shown in the genogram (Figure 1). During this consultation, the patient reported progressive worsening imbalance, difficulty walking in a straight line, and a feeling of fatigue and "heavy legs" when she arrived home. On physical examination, instability in the standing position, gait alteration with eyes open and closed, difficulty walking in a straight line, reliance on the right leg, and difficulty lifting the left leg were observed. Based on these findings, a molecular test for the expansion of the ATXN3/SCA3 gene was requested for the patient, revealing the presence of 26 and 70 CAG repeats in each allele (normal up to 45 CAG repeats), confirming the diagnosis of MJD.

There was progression of the clinical picture in the following year with reports of dysphagia to liquids and solids, diplopia, lachrymation, difficulty holding objects, worsening postural instability and gait, requiring her to retire from her professional activities as a kindergarten educator. Over the course of five years, the patient developed the need to use a walker to move around, episodes of dysphagia with choking becoming frequent, sporadic episodes of diplopia and restless legs syndrome, that were treated with pramipexole dihydrochloride twice a day. The patient followed up with physiotherapy, speech therapy, and hydrotherapy sessions.

On physical examination, weakness of upper and lower limbs was also observed, along with mild edema and signs of venous insufficiency and edema in the lower limbs. On ophthalmological examination, long-distance diplopia with noticeable progression was reported, dysmetric saccadic movements, bilateral horizontal nystagmus, and reduced lower visual field of the right eye. The musculoskeletal examination showed a greater degree of postural instability when compared to the last consultation, ataxic gait with staggering and widened base, normal muscle tone, slight dysmetric and slowed movements, and normal diadochokinesia.

The patient was monitored from October 2022 to May 2024. Table 1 and Table 2 contain responses from semi-structured interviews regarding perceived communication difficulties and how they are apparently resolved by the patient, as viewed by the patient herself, her spouse, caregiver, and mother at the beginning of the current study.

After analyzing the interviews, assistive tools were studied to address the patient's communication needs. Consideration was given to the difficulties presented by the patient, the stage of disease progression, access to a smartphone (reported by the patient as the preferred device for communication), and financial feasibility. A 5W2H intervention plan was proposed (Table 3), focusing mainly on speech slowing and dysphagia, diplopia, and motor coordination difficulties.

Based on the discussion with the patient after the presentation of the tools, the following were implemented in August 2023: 1. increased screen zoom; 2. replacement of typed texts with audio conversations; 3. Google Assistant; and 4. Voice Access. All chosen assistive technologies were available for free and were installed/activated by the project authors on the patient's smartphone. The patient did not express interest or need in using the "Symbo Talk," "HeadMouse," and "Talkback" apps at the stage of the disease by that time. After six months of using the tools, SF-36 data collection and semi-structured interviews were conducted again, including a question about the perception of using the tools in the interview. Table 4 shows the data collected in the SF-36 questionnaire before and after implementing the tools.

In the semi-structured interviews with the patient and her family members, no changes were observed after the use of the tools in the following areas: difficulties related to communication, how the patient resolves difficulties caused by DMJ, and the difficulties that the patient and her family believe she will face in the future. Table 5 shows the responses regarding changes in the patient's daily communication after the proposed interventions.

4 DISCUSSION

Few studies have demonstrated ways to overcome communication problems generated or the use of assistive technologies in MJD. In a literature review conducted by the authors, only two studies were found that addressed ways to overcome communication accessibility barriers [8]. In a study conducted by Amery et al., four

prototypes were developed (an alphabet board, a core word board, a comprehensive communication book with words for core vocabulary and symbols for fringe vocabulary, and a comprehensive communication book with symbols for core and fringe vocabulary), organized at different levels of taxonomy, to assist in the communication of Australian Aboriginals with MJD [9]. In another work by Paulson and Shakkotai, it was only indicated that, in patients with dysarthria, alternative communication methods such as boards and digital devices could be used [10].

In the present study, we attempted to work with some accessibility applications, changes in the patient's habits, such as better utilizing voice assistive technology resources, and modifications to her smartphone that could help her overcome communication difficulties. However, due to the current state of the disease she presents, the applications "SymboTalk," "HeadMouse," and "TalkBack" were not considered useful in her daily life. These may be alternatives as the clinical condition worsens as expected in the natural progression of the disease.

Despite MJD causing various speech alterations, the change from typing messages to audio messages proved beneficial for the patient. Due to motor and ocular symptoms, the patient types slowly and sometimes makes typing mistakes. Thus, communicating via audio, despite slowed speech, facilitated her communication on social networks, according to her reports.

The "Google Assistant," which has various functions, was mainly recommended for converting speech to written messages when needed. However, the patient's slowed speech impaired the application's comprehension of the dialogue, failing to meet the objective satisfactorily at times. Although the "Voice Access" app also has a speech transcription function, the patient opted for "Google Assistant" after use, as "Voice Access" required the tool to remain active all the time, interfering with daily communication if the smartphone was near the patient, worsening the device's usability.

Enlarging the characters on the screen and the cell phone's zoom allowed for easier reading of texts and images, partially circumventing the patient's visual problems such as diplopia.

Comparing the average results of each SF-36 questionnaire domain, the domains that maintained the average were "Physical functioning" and "Physical Role". Meanwhile, there was a worsening over this period in the domains "Bodily Pain",

"Vitality" and "Social Role" The domains that showed an improvement in scores were "General Health", "Emotional Role" and "Mental Health."

In attempting to explain the reasons for the increase in these three domains, no studies were found in the literature that evaluated MJD patients in this context. In a study that used the SF-36 questionnaire in patients with Amyotrophic Lateral Sclerosis (ALS), a neurodegenerative disease that relatively shares the loss of bodily control over time, health-related quality of life showed positive correlations in the domains of "Emotional Role", "General Health", and "Social Role" [11]. According to the study authors, these aspects can significantly interfere with the perception of health-related quality of life. The improvement in the "Emotional Role" and "General Health" domains was found in the patient in the present study. One hypothesis for this positive perception may have been influenced by the intensive follow-up conducted by the researchers over the two years of care and follow-up, including home visits. Another observed issue is that the patient became more communicative on social networks when she switched from typing texts to audio messages, sending messages more frequently. This same perception was found in the spouse's interview (Table 5). This can also be reinforced by the finding of improvement in the "Mental Health" domain of the SF-36 Questionnaire.

As recommendations, the authors propose that for all patients diagnosed with MJD, the communication difficulties perceived by them should be evaluated during the initial consultations, especially: 1. Visual reading difficulties caused by diplopia; 2. Motor coordination difficulties that impair writing and typing; and 3. Oral communication difficulties due to slowed speech. Available tools should be discussed and implemented based on the discussion with the patient and their perception of the necessity. Adaptation and implementation of new tools should also be anticipated as the disease gradually progresses. In the case of the patient from this study, the choice of tools was able to partially meet some of the identified demands. It should be noted, however, that most of the chosen tools were developed as smartphone applications, so these tools would be mainly useful for other patients familiar with smartphone use. This choice was based on the patient's report that pointed out the smartphone as the main device for her communication, being already available and familiar to the patient.

It was found, however, that discussing new tools for the next stages of the disease can be challenging, as the patient needs to agree on the need related to the progression and limitation imposed by the disease.

Another issue is that new research and development of assistive technologies are needed to cover the different stages of disease progression, including tools not tested in the present study. Additionally, the research has limitations, as only one patient participated in the study and there are few publications in the literature for comparison of the findings.

6 CONCLUSION

MJD is a progressively debilitating neurodegenerative disease with no curative treatment and a poor long-term evolution, which can manifest in various ways depending on the disease stage. Patients gradually develop motor and coordination deficits, visual impairments, and speech difficulties that impact their quality of life and become increasingly demanding, requiring continuous adaptation for accessibility as the disease progresses.

This study proposes main interventions focused on assistive communication technologies, particularly smartphone applications that can aid oral communication, such as using audio messages for social media conversations; screen enlargement, text zooming, and Google Assistant for voice-to-text typing in the early stages of the disease. Voice Access did not show good adherence with the studied patient.

Other proposed applications were not tested by patients' request: Symbo Talk, Talkback, and Head Mouse, which could be alternatives for patients with a more advanced and severe clinical condition. However, the study group will continue to support the patient and offer assistive tools as the disease progresses.

Thus, this study aimed to propose interventions to assist with the current needs of the studied patient, which are considerable. Despite being simple, these measures have brought a positive impact to her life. Furthermore, these interventions may benefit other MJD patients living in similar contexts and at the same stage of the disease.

DECLARATION OF INTEREST STATEMENT

The authors declare that there are no conflicts of interest regarding the publication of this research. No financial or personal relationships with other people or

organizations have influenced the outcomes and interpretation of this study. All authors have approved the final manuscript and agreed with its submission to this journal.

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TABLES

Table 1 – Patient, spouse, mother, and caregiver responses regarding patient's communication difficulties by type and how they are resolved.

(concntinues)

Informant	Difficulties encountered	Verbal communication	Visual communication/reading	Written communication	Non-verbal/gestural communication
Patient	**Patient's complaints**	Has difficulty speaking quickly, expressing herself.	Has little difficulty. Diplopia is not currently interfering.	The handwriting has changed and she is writing more slowly.	Reports difficulty rating 7 on a scale of 0-10.
	How the patient solves the problem	Has not done anything to resolve.	Has not done anything to resolve.	She writes more slowly.	Has not done anything to resolve.
Spouse	**How the patient perceives**	Reports that the patient is speaking a bit slower.	Some difficulty.	She is writing more slowly.	Reports that the patient no longer had the habit of gesturing before the onset of MJD.
	How others perceive that the patient solves the problem	For a while, used spiral straws recommended by the speech therapist.	Does not notice.	Reports that the patient has been engaging in drawing and coloring as a habit.	Has not done anything to resolve.
Mother	**How the patient perceives it**	Reports that the patient is getting a bit nervous when speaking.	Does not notice.	Does not notice..	Does not notice.
	How the patient resolves the problem	Reports that the daughter went to the speech therapist to improve verbal communication.	Does not notice.	Does not notice.	Has not done anything to resolve.

Table 1 – Patient, spouse, mother, and caregiver responses regarding patient's communication difficulties by type and how they are resolved.

(ends)

Informant	Difficulties encountered	Verbal communication	Visual communication/reading	Written communication	Non-verbal/gestural communication
Caregiver	**How the patient perceives it**	Reports that the daughter went to the speech therapist to improve verbal communication.	Does not notice.	Little difficulty.	Difficulty moving fingers. Performs movements more slowly.
	How others perceive the patient resolves the problem	Does not notice. Has not been going to the speech therapist.	Does not notice.	Reports that the patient has been engaging in the habit of drawing and coloring.	Does not notice.

Source: The authors.

Table 2 – Responses regarding the main communication difficulties the patient currently has and the main difficulty that the patient herself, spouse, mother, and caregiver will present in the future.

Informant	What are the main communication difficulties the patient has today?	What do you believe will be the main communication difficulty the patient will encounter as the disease progresses?
Patient	Reports being unable to speak loudly and has some difficulty singing.	The lack of balance, such as not being able to bathe alone.
Spouse	Reports that the patient gets more nervous when starting to speak, cannot be interrupted while speaking, and speaks more slowly.	Not being able to communicate.
Mother	Reports that the patient wants to speak a lot and gets agitated because she cannot communicate the way she would like.	Not being able to communicate.
Caregiver	Reports that the patient is communicating more slowly.	Not being able to communicate.

Source: The authors.

Table 3 – 5W2H with the Intervention Proposal for the Patient

(continues)

WHAT	WHY	WHEN	WHERE	HOW
Need for action	Justification / benefits	Priority	Whith area	Assistive Technology Proposal
Presentation of SymboTalk to the patient.	Speech slowing and dysphagia.	July 2023	During home visit at the patient's house. App installation.	Installation on the phone and training for the use of Symbo Talk, a free application with predefined communication boards that allows messages to be spoken aloud through symbols.
Presentation of Talkback	Double vision and reading difficulty.	July 2023	During home visit at the patient's house.	Installation of the 'Talkback' app on the patient's phone and guidance on how to use it. Talkback is Google's screen reader included in Android devices. It allows control of the device without using the eyes.
Presentation of zoom magnification, with enlarged characters on the screen.	Diplopia and reading difficulty	July 2023	During home visit at the patient's house.	Screen zoom and font size increase on the cellphone. Customizing screen zoom and font size settings on your mobile device can be useful to facilitate reading and consequently enhance interaction with the device.
Presentation of “Voice Access”	Motor difficulty when typing	July 2023	During home visit at the patient's house.	Installation of Voice Access app and guidance on how to use it. “Voice Access”: Through voice commands, the app helps manipulate touchscreen interfaces.
Presentation of “HeadMouse”	Motor difficulty when typing	July 2023	Instalação no notebook da paciente durante visita domiciliar.	The "Head Mouse" will help the patient as her condition progresses. This program enables independent computer use for those without arm movements. After installation and positioning a webcam, the "Head Mouse" uses head movements to control the cursor and clicks by blinking or opening the mouth.

Table 3 – 5W2H with the Intervention Proposal for the Patient

(ends)

WHAT	WHY	WHEN	WHERE	HOW
Presentation of Google Assistant	Motor difficulty when typing	July 2023	During home visit at the patient's house.	"Google Assistant": it is a virtual personal assistant developed by Google that can perform everyday tasks such as making phone calls, sending messages, searching on Google, and also engaging in conversations with the user.
Replacement of written texts with audio in cell phone conversations.	Motor difficulty when typing. Screen reading difficulty	July 2023	During home visit at the patient's house.	Recommend that the patient advise people to avoid written forms of communication and prefer oral communications, such as sending voice messages on chat apps.

Source: The authors.

Table 4 - Values of the SF-36 questionnaire domains collected from the patient, caregiver, mother, and spouse pre and post implementation of accessibility tools.

	Patient pre	Patient post	Caregiver pre	Caregiver post	Mother pre	Mother post	Spouse pre	Spouse post	Mean pre	Mean post
Physical funcitioning	25	10	0	5	0	0	5	15	7,5	7,5
Physical Role	0	0	0	0	0	0	0	0	0	0
Bodily Pain	30	51	84	61	31	15	51	31	49	44,75
General health	17	62	25	42	35	15	37	52	28,5	42,75
Vitality	70	80	55	45	75	60	35	45	58,75	57,5
Social Role	100	50	75	100	50	62,5	37,5	25	65,6	59,38
Emotional Role	100	100	100	100	33,3	100	0	33,3	58,3	83,33
Mental health	84	80	52	56	36	56	64	68	59	65

Source: The authors.

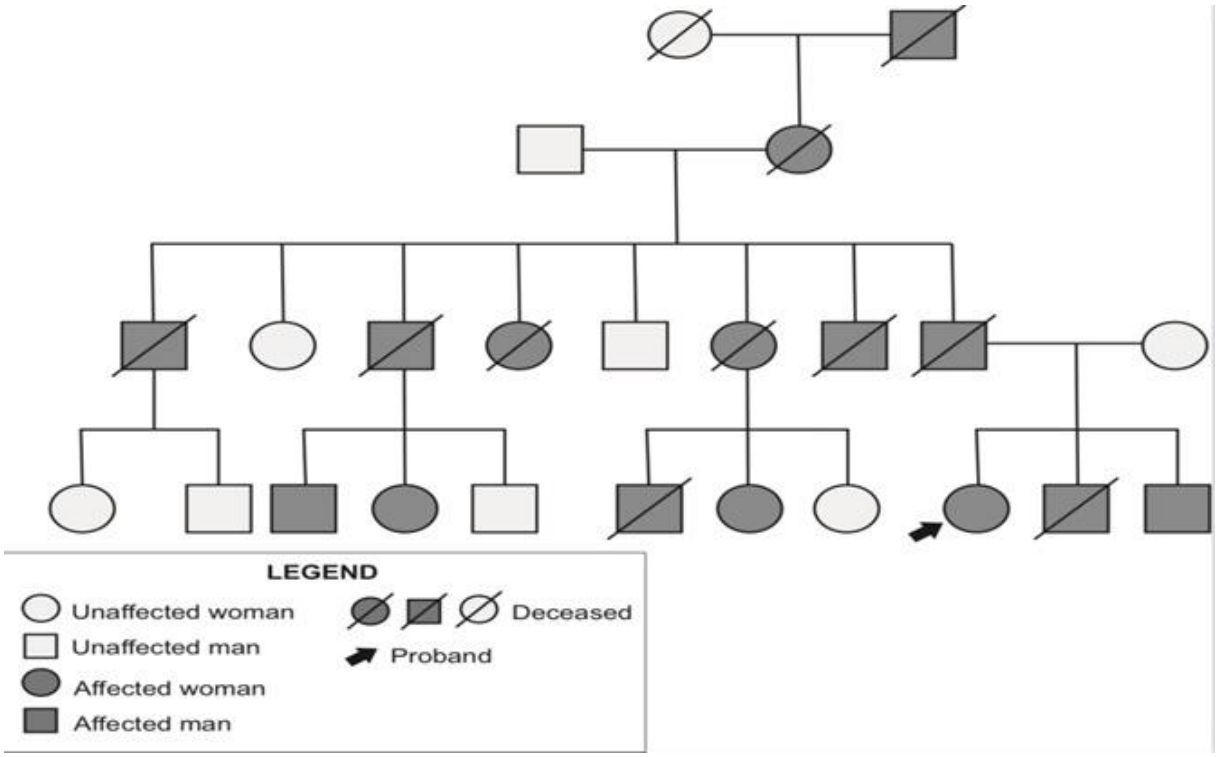
Table 5 - Responses on how the interventions performed changed communication capacity.

Describe how the interventions performed have changed your communication capacity.	**Response**
Patient	"The larger screen has been very helpful, especially for reading, with almost no difficulty." "Audio messaging has made communication easier, as typing often results in more typos." "My slow speech sometimes makes it difficult to use Google Assistant, but it has helped with conducting searches on my phone."
Spouse	"Didn't notice the screen change and its benefits." “Audio messaging has greatly helped in communicating with people via cell phone." “Didn't notice Google Assistant and its benefits.
Mother	“Didn't notice the screen change and its benefits." “Didn't notice audio messaging, as they don't use the cell phone." “Didn't notice Google Assistant and its benefits."
Caregiver	“Didn't notice the screen change and its benefits." “Audio messaging helped improve communication on the cell phone." “Didn't notice Google Assistant and its benefits."

Source: The authors.

FIGURES

Figure 1 – Patient's Genogram.



Source: The authors.

FIGURE CAPTIONS:

Figure 1 : Patient’s genogram showing individuals affected by Machado Joseph Diease.

BIOGRAPHICAL NOTES

- Lara Valentim Vale is a medical student at Centro Universitário de Volta Redonda (UNIFOA), currently in the 9th semester. She served as Administrative Director of the Slow Medicine Academic League, and as a member of the Internal Medicine Academic League and the Pediatrics Academic League. She was a member of the PET-Health Project and was a teaching assistant for Human Anatomy in the 3rd period at Unifoa. Currently, she is a member of the Otolaryngology Academic League and an intern at São João Batista Hospital in the Surgical Emergency sector and at Santa Casa de Barra Mansa in the Intensive Care Unit and Medical Emergency Center.
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