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# Effectiveness of group speech therapy for patients with Machado Joseph Disease: a series of cases

## Eficácia de terapia de fala em grupo para pacientes com a doença de Machado Joseph: uma série de casos

## Eficiencia de la terapia del habla en grupo para pacientes con Enfermedad de Machado Joseph: una serie de casos

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***Abstract***

**Objective**: To verify the benefit of group speech therapy in speech intelligibility of patients with Machado Joseph's disease (MJD). **Methods**: A series of cases was carried out, with patients seen in a speech therapy clinic for neurodegenerative adults in a referral hospital in southern Brazil. Patients with the molecular diagnosis of MJD were included. Speech recordings were performed before and after the intervention. Subsequently, the speech excerpts underwent auditory-perceptual analysis by 3 trained speech therapists and calibrated to a Kappa index ≥ 0.90, blind to speech collections and acoustic analysis in the Praat software. Speech therapy was performed in a group, consisting of four weekly sessions of fifty minutes. Each session was divided between speech exercises and guidance on strategies to optimize communication. **Results**: The sample consisted of 5 patients with a mean age of 39.8 years (± 16.51) and disease duration of 10 years (± 8.15). Four (80%) participants received an initial speech therapy

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**Authors' contributions:**
BGSA, RM, AA: Data collection, Methodology, Drafting the article.
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diagnosis of mild dysarthria and one (20%) of moderate dysarthria. After the intervention, there was no improvement in the diagnosis of dysarthria, however it was found that 60% (n = 3) of the participants showed improvement in the speech motor bases: articulation, 40% (n = 2), prosody and resonance and 40% (n = 2) worsened in breathing. The acoustic analysis showed an improvement in maximum phonation time (MPT) in 3 (60%) of the 5 patients. **Conclusion**: Despite the little improvement in specific parameters of the acoustic analysis, there was an improvement in speech functionality from the auditory perceptual analysis, improving the speech intelligibility of this sample.

**Keywords:** Machado Joseph Disease; Dysarthria; Rehabilitation; Speech, Language and Hearing Sciences.

## *Resumo*

**Objetivo:** Verificar o benefício de terapia fonoaudiológica em grupo na inteligibilidade de fala de pacientes com Doença de Machado Joseph (DMJ). **Método:** Realizou-se uma série de casos, com pacientes atendidos em um ambulatório de fonoaudiologia para adultos neurodegenerativos em um hospital de referência no sul do Brasil. Foram incluídos pacientes com o diagnóstico molecular de DMJ. Realizaram-se coletas de fala pré e pós-intervenção. Posteriormente, os trechos de fala passaram por análise perceptiva-auditiva por 3 fonoaudiólogas treinadas e calibradas a um índice Kappa ≥ 0.90, cegas às coletas de fala e por análise acústica no *software* Praat. A terapia fonoaudiológica foi realizada em grupo, composta por quatro sessões semanais de cinquenta minutos. Cada sessão foi dividida entre exercícios de fala e orientação sobre estratégias para otimizar a comunicação. **Resultados:** A amostra foi composta por 5 pacientes com média de idade de 39,8 anos (±16,51) e tempo de doença de 10 anos (±8,15). Quatro (80%) participantes receberam diagnóstico fonoaudiológico inicial de disartria leve e um (20%) de disartria moderada. Após a intervenção, não houve melhora no diagnóstico de disartria, contudo verificou-se que 60% (n=3) dos participantes apresentaram melhora na articulação, 40% (n=2) na prosódia e ressonância e 40% (n=2) apresentaram piora na respiração. Na análise acústica observou-se melhora no tempo máximo de fonação (TMF) em 3 (60%) dos 5 pacientes. **Conclusão:** Verificou-se melhora na funcionalidade da fala através da análise perceptiva auditiva, porém com pouca melhora em parâmetros específicos da análise acústica.

**Palavras-chave:** Doença de Machado Joseph; Disartria; Reabilitação; Fonoaudiologia.

## *Resumen*

**Objetivo**: Verificar el beneficio de la logopedia grupal en la inteligibilidad del habla de pacientes con enfermedad de Machado Joseph (EMJ). **Metodos**: Se realizó una serie de casos, con pacientes atendidos en una clínica de logopedia para adultos neurodegenerativos en un hospital de referencia en el sur de Brasil. Se incluyeron pacientes con diagnóstico molecular de EMJ. Se realizaron grabación del habla antes y después de la intervención. Posteriormente, los extractos del habla se sometieron a un análisis auditivo-perceptivo por 3 logopedas capacitados y calibrados con un índice Kappa ≥ 0,90, ciegos a las grabación del habla y al análisis acústico en el software Praat. La logopedia se realizó en grupo, consistente en cuatro sesiones semanales de cincuenta minutos. Cada sesión se dividió entre ejercicios de habla y orientación sobre estrategias para optimizar la comunicación. **Resultados**: La muestra estuvo formada por 5 pacientes con una edad media de 39,8 años (± 16,51) y una duración de la enfermedad de 10 años (± 8,15). Cuatro (80%) participantes recibieron un diagnóstico inicial de terapia del habla de disartria leve y uno (20%) de disartria moderada. Tras la intervención, no hubo mejoría en el diagnóstico de disartria, sin embargo se encontró que el 60% (n = 3) de los participantes mostró mejoría en las bases motoras del habla: articulación, 40% (n = 2), prosodia y resonancia. y el 40% (n = 2) empeoró en la respiración. El análisis acústico mostró una mejora en el tiempo máximo de fonación (TMF) en 3 (60%) de los 5 pacientes. **Conclusion**: A pesar de la pequeña mejora en los parámetros específicos del análisis acústico, hubo una mejora en la funcionalidad del habla a partir del análisis de percepción auditiva, mejorando la inteligibilidad del habla de esta muestra.

**Palabras clave:** Enfermedad de Machado Joseph; Disartria; Rehabilitación; Patología del habla y Lenguaje.

## Introduction

Spinocerebellar ataxia type 3 (SCA3), also known as Machado-Joseph disease (MJD), is an autosomal dominant neurodegenerative disease caused by the repeat expansion of the CAG sequence of the ATXN3 gene at chromosome 14q32.11. The prevalence of MJD is higher in countries with inhabitants of Portuguese and Azorean descent[2]. In Rio Grande do Sul, Brazil, and Portugal, the minimum estimated prevalence is 3 cases per 100,000 inhabitants[3,4]. Due to a range of possible underlying CAG repeat expansion, researchers have noted significant clinical pleomorphism. As such, MJD has been traditionally subdivided into three types, according to the age of onset. Generally speaking, the earlier the onset, the more severe the symptoms[5].

Among the most frequent symptoms of MJD are ataxia, dysphagia, dysarthria, and eye problems[6]. As the disease progresses, these symptoms can translate into important daily obstacles that affect a patient's quality of life. Regarding speech, mixed dysarthria with cerebellar and hypokinetic components is the most frequent type among patients with MJD. This type of speech impairment is characterized by imprecise articulation, altered prosody, distorted consonants and vowels, prolonged intervals, slow rate, a rough or tense voice, vocal instability, and monopitch[7]. The younger the patient at the onset of the disease, the more frequent the presentation of monopitch and impaired prosody. Furthermore, articulation, diadochokinetic rates, resonance, and prosody have shown a moderate correlation with the number of CAG triplet repeats[7].

Although dysarthria is a frequent MJD symptom, few international or national studies[7-9] have described the clinical characteristics or possible approaches to speech therapy for this patient population. We found two case reports about speech therapy for dysarthria caused by MJD. The Lee Silverman[9] (LSVT) method was used in the treatment and the authors reported improvement in all speech subsystems, fewer signs of dysphagia, and enhanced, voice-related quality of life.

Another study[10] described the process of assessing and managing speech therapy for a patient with MJD. The patient presented impaired breathing, articulation, phonation, and resonance. The speech therapy was initially based on weekly sessions during which the patient's condition was monitored and orofacial myofunctional exercises were planned for practice at home. The authors reported moments of improvement interspersed with moments of stability or worsening clinical status, due to the progression of the disease.

Intensive home therapy was found to be effective for patients with autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)[11]. Seven participants underwent a 4-week intervention specifically tailored for cerebellar ataxia. The researchers observed improvement in speech intelligibility and naturalness after treatment.

A recent review[8] concluded that, although there is insufficient data regarding the effectiveness of speech therapy for patients with ataxia, it is essential for this population. Considering the limited amount of evidence concerning the effectiveness of speech therapy for patients with dysarthria caused by MJD, this study aimed to address this issue and test a therapeutic approach.

## Method

This is a case series that relied on convenience sampling. Participants visited a speech therapy clinic for neurodegenerative adults at a renowned hospital in southern Brazil. Based on having a molecular diagnosis of MJD, and a finding of impaired spontaneous speech intelligibility during the first visit to the outpatient clinic, patients were invited to participate in the study. Other inclusion criteria were normal cognitive thresholds and the availability to participate in all phases of therapy. Individuals who missed one session or the final assessment were excluded. The initial sample consisted of eight patients. Three were excluded for not completing all the therapy sessions. This study was approved by the hospital ethics committee under approval number 2018-0323. All participants signed an informed consent form.

For the speech assessment, the same speech tasks were recorded before and after the course speech therapy. These samples were recorded in an outpatient service room. An Acer(®) notebook, model Aspire One, and a KARSECT HT-9(®) microphone were coupled with an Andrea Pureaudio USB(®) adapter. Individual files were saved and archived using Audacity(®) software, labeled with the name of each patient and the date of collection. Participants were asked to sustain the vowel /a/

with a single breath. They were asked the question "What route did you take to get here?" to elicit spontaneous speech for the recording.

Subsequently, three blinded speech-language pathologists with at least three years of experience in the field, trained and calibrated with a ≥ 0.90 kappa test score, performed an auditory-perceptual evaluation of the speech samples. These examiners had undergone training during which they listened to impaired speech samples and classified them. Afterward, the inter-examiner agreement was tested against a gold standard voice specialist for calibration. Following calibration, all the examiners participated in other speech analysis projects before taking part in our analysis. The speech of the participants was classified according to each speech subsystem and degree of impairment: normal, mild, moderate, or severe. In addition, an acoustic analysis of voice was performed using Praat(®) software. The parameters of shimmer, jitter and maximum phonation time (MPT) were analyzed.

The speech therapy was held in a group format, for a total of four weeks. Participants attended weekly fifty-minute sessions. Each session was divided into speech exercises and guidance on strategies to enhance communication and improve speech intelligibility (Chart 1). For this reason, therapy planning focused on separate and combined breathing, articulation, and phonation control tasks. The planning that was used in this study was based on references addressing voice and speech therapy for patients with neurological diseases.

**Chart 1.** Therapy sessions

| Week | Speech Subsystem | Exercises |
|---|---|---|
| Week 1 | Respiration | - Sustaining phonemes and syllables - /s/, /z/, /sa/ and /za/ - using diaphragmatic breathing. Five repetitions each. |
| | Articulation | - Alternating over-articulated, sequenced vowels - /a/, /i:/, /u:/. Ten repetitions.<br>- Reading monosyllabic and disyllabic words aloud, with feedback from the patients in the group. |
| | | - Completing full tongue circles around closed lips. Five repetitions in either direction. |
| Week 2 | | Revision of all exercises. |
| | Articulation | - Over-articulating the trisyllabic sequence /pataka/. Five sets of ten repetitions. |
| | Articulation/ Respiration | - Reading sentences aloud, with feedback from the patients in the group.<br>- Practicing spontaneous speech with guidance and feedback from the speech-language pathologist. |
| Week 3 | | Revision of all exercises. |
| | Articulation/ Respiration | - Reading aloud written excerpts of the patients' choosing, as well as reading samples from the speech-language pathologist, with feedback from patients in the group.<br>- Practicing spontaneous speech with guidance and feedback from the speech-language pathologist. |
| Week 4 | | Revision of all exercises. |
| | Articulation/ Respiration | - Reading aloud written excerpts of the patients' choosing, with feedback from the patients in the group.<br>- Practicing spontaneous speech with guidance and feedback from the speech-language pathologist. |

Original Table.

## Results

The final sample consisted of five participants, three women, and two men. Table 1 shows the participants' sociodemographic data.

In Table 2, we present the results of the acoustic analysis, before and after therapy. Regarding the MPT, before therapy, all participants presented scores below the average scores of normality for their sex. After the group speech therapy, 60% (3) of the patients improved their MPTs, but still achieved scores below the normal range described in the literature[13]. Regarding shimmer, this parameter remained impaired before and after speech therapy. As for jitter, 40% (2) of the patients showed improvement.

**Table 1.** Sociodemographic data

| Patient | 1 | 2 | 3 | 4 | 5 |
|---|---|---|---|---|---|
| Sex | F | M | M | F | F |
| Age | 23 | 24 | 39 | 56 | 57 |
| Disease duration (years) | 2 | 12 | 5 | 8 | 23 |
| Walking aid | No | No | Wheelchair | Wheelchair | Cane |
| Age at disease onset (years) | 21 | 12 | 34 | 48 | 34 |
| Formal education (years) | 12 | 9 | 11 | 7 | 5 |
| MMSE | 29 | 29 | 30 | 20 | 19 |

F: female; M: male; MMSE: Mini-Mental State Examination.

**Table 2.** Results of the acoustic analysis parameters, before and after therapy

| Parameter | Patient 1 | Patient 2 | Patient 3 | Patient 4 | Patient 5 |
|---|---|---|---|---|---|
| **MPT (seconds)** | | | | | |
| Before therapy | 5.85 | 0.07 | 8.06 | 5.58 | 5.25 |
| After therapy | 9.22 | 8.73 | 10.29 | 3.79 | 3.34 |
| Average values of normality[a] | 14 | 20 | 20 | 14 | 14 |
| **Jitter (Local %)** | | | | | |
| Before therapy | 1.19 | 0.36 | 1.99 | 0.39 | 0.27 |
| After therapy | 0.66 | 2.94 | 1.16 | 0.67 | 1.79 |
| Average value of normality[12] | | | ≤ 1..04% | | |
| **Shimmer (Local %)** | | | | | |
| Before therapy | 7.63 | 9.80 | 14.14 | 10.34 | 10.40 |
| After therapy | 7.48 | 17.10 | 16.37 | 10.75 | 17.11 |
| Average value of normality[13] | | | ≤ 3.81% | | |

MPT: maximum phonation time

Table 3 shows the results of the auditory-perceptual analysis. Sixty percent (60%, n=3) of the participants showed improved articulation, and 40% (n=2) demonstrated improved prosody and resonance. However, 40% (n=2) presented worse breathing.

**Table 3.** Results of the Auditory-Perceptual Evaluation

| | Patient 1 | Patient 2 | Patient 3 | Patient 4 | Patient 5 |
|---|---|---|---|---|---|
| **Articulation** | | | | | |
| Before therapy | Mild impairment | Mild impairment | Mild impairment | Moderate impairment | Moderate impairment |
| After therapy | Normal | Mild impairment | Normal | Normal | Moderate impairment |
| **Phonation** | | | | | |
| Before therapy | Mild impairment | Mild impairment | Mild impairment | Mild impairment | Moderate impairment |
| After therapy | Mild impairment | Mild impairment | Mild impairment | Mild impairment | Moderate impairment |
| **Prosody** | | | | | |
| Before therapy | Normal | Mild impairment | Normal | Normal | Mild impairment |
| After therapy | Normal | Normal | Normal | Normal | Normal |
| **Respiration** | | | | | |
| Before therapy | Normal | Normal | Mild impairment | Mild impairment | Moderate impairment |
| After therapy | Mild impairment | Mild impairment | Mild impairment | Mild impairment | Mild impairment |
| **Resonance** | | | | | |
| Before therapy | Normal | Normal | Mild impairment | Normal | Moderate impairment |
| After therapy | Normal | Normal | Normal | Normal | Mild impairment |
| **Dysarthria** | | | | | |
| Before therapy | Mild | Mild | Mild | Mild | Moderate |
| After therapy | Mild | Mild | Mild | Mild | Moderate |

Original Table.

## Discussion

This case series aimed to verify the effect of group speech therapy on patients with an SLP diagnosis of MJD-related dysarthria. Based on the results in the previous section, we suggest there was a positive effect on the speech intelligibility of these patients, even with a short term of therapy (4 weeks). Specifically, improvement was observed in 3 of the 5 speech subsystems, namely articulation, prosody, and resonance. In the acoustic analysis, we found that patient MPTs did improve, although they did not reflect average scores of normality[12,13]. MJD is a degenerative disorder, so it is understood that symptoms will not go into remission. As such, enhanced speech intelligibility can allow patients to better enjoy communication and quality of life. Our data corroborate studies[7-10] that also reported improved speech intelligibility after speech therapy.

The choice of the group therapy format addressed the high demand at the outpatient clinic, located in a hospital belonging to the public health network in Brazil. This format provided a space for individuals with the same disease and speech difficulties to participate in an exchange. Although it is a frequent practice in speech therapy, especially within the context of the Brazilian Unified Health System (SUS), there are few published studies on its effectiveness. The group therapeutic approach in Brazilian speech therapy emerged in the 1980s, due to the large number of individuals who needed care in Basic Health Units, and the smaller number of professionals available for individual healthcare[14.]

We found literature reviews[14-16] that investigated the benefits of group speech therapy. The main benefits of this approach that the researchers reported were the opportunity for subjects to construct knowledge together, and the exchange of experiences which can help to improve speech-language symptoms and modify patient perception. Furthermore, the space to discuss and share with their peers increases patient motivation. Over time, through the therapeutic bond, they begin to adhere more easily to therapeutic plans. The group format introduces new possibilities for relationships in which the agents convey a great amount of information, and share experiences and knowledge.

Most of the studies were performed with adults and elderly people, in the areas of language, voice, and hearing[14-16]. We found a report of an objective

assessment of group care for patients with dysarthria in which the participants showed improvement in the speech subsystems of prosody, respiration, phonation and articulation. Furthermore, during the subjective assessment, the subjects reported improvement in all the speech subsystems they received therapy for [17].

Our study limitations include the small sample size, the short term of speech therapy, failure to use a quality of life questionnaire, and the possible heterogeneity among the patients' MJD types. There was a loss of approximately 25% of patients, due to physical mobility and transportation issues. The centralization of specialized health services in capitals makes it a challenge for patients with rare diseases to have access to treatment. The number of sessions was based on research[11] and followed the operational protocol of the hospital clinic. We believe that further sessions could have resulted in better acoustic analysis scores. Although no speech-related quality of life questionnaire was used, patients expressed a high degree of satisfaction and a desire to continue in therapeutic care.

## Conclusion

After undergoing group speech therapy, the MJD patients with dysarthria presented better speech intelligibility, particularly regarding the speech subsystems of articulation, prosody, and resonance. However, the results of the acoustic analysis only showed improved maximum phonation times.

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