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# Effect of speech-language therapy in Machado-Joseph disease: case report

## Atuação fonoaudiológica na doença de Machado-Joseph: relato de caso

Thames dos Santos Marques<sup>1</sup> , Cristina Lemos Barbosa Furia<sup>1</sup> , Juliana Onofre de Lira<sup>1</sup> 

### ABSTRACT

Machado-Joseph disease is the most prevalent form of spinocerebellar ataxia in Brazil, and has dysphagia and dysarthria among its main clinical signs. This case report aims to ascertain the effects of intensive speech-language intervention in a patient with Machado-Joseph disease. Data collection was performed based on speech and swallowing assessment protocols and self-assessment protocols specific to swallowing-related and communication-related quality of life. Quantitative assessments of acoustic parameters were also performed. The intervention was administered through the Lee Silverman method, which is an intensive program aimed at increasing vocal intensity. The results of clinical and instrumental evaluations showed improvement in all motor parameters of speech (respiration, phonation, resonance, articulation, and prosody), besides a reduction in dysphagic signs. Regarding vocal quality, there was a decrease in hoarseness and instability, regularization of jitter and shimmer, increased vocal intensity, and improved coordination of words and phrases by expiration, as well as slight improvement of diadochokinesis. After intervention, self-assessment of swallowing-related quality of life was unchanged or improved in the domains directly related to food, but reduced in emotional domains. The patient reported satisfaction in all domains of voice-related quality of life, and scores were increased in all domains. We conclude that intensive intervention was beneficial for the participant and positively impacted their quality of life.

**Keywords:** Spinocerebellar ataxia type 3; Machado-Joseph disease; SCA3; Dysphagia; Deglutition disorders; Dysarthria; Lee Silverman voice treatment; Quality of life

### RESUMO

A doença de Machado-Joseph é a forma de ataxia espinocerebelar de maior prevalência no Brasil e tem como alguns dos principais sinais clínicos a disfagia e a disartria. Este relato de caso objetivou verificar os efeitos da intervenção intensiva fonoaudiológica em um paciente com a doença de Machado-Joseph. A coleta de dados foi realizada a partir de protocolos de avaliação de fala e deglutição e protocolos de autoavaliação de qualidade de vida, em relação à deglutição e comunicação. Também foram realizadas avaliações quantitativas de parâmetros acústicos. A intervenção foi administrada por meio do método Lee Silverman, programa intensivo que visa ao aumento da intensidade vocal. A partir das avaliações clínicas e instrumentais, os resultados demonstraram melhora em todas as bases motoras de fala, respiratória, fonatória, ressonantal, articulatória e a prosódia, além da diminuição dos sinais disfágicos. Na qualidade vocal, houve diminuição de rouquidão e instabilidade, regularização de *jitter* e *shimmer*, aumento da intensidade vocal, melhora na coordenação de palavras e frases por expiração e, ainda, melhora discreta da diadococinesia. Após intervenção, a autoavaliação de qualidade de vida relacionada à deglutição apresentou valores iguais ou maiores nos domínios diretamente ligados à alimentação, porém, os domínios emocionais diminuíram. O paciente relatou satisfação em todos os domínios da qualidade de vida em voz e foram obtidos valores maiores em todos os domínios. Concluiu-se que a intervenção intensiva beneficiou o participante e impactou positivamente sua qualidade de vida.

**Palavras-chave:** Ataxia espinocerebelar tipo 3; Doença de Machado-Joseph; SCA3; Disfagia; Desordens da deglutição; Disartria; Método Lee Silverman; Qualidade de vida

Study carried out at Curso de Fonoaudiologia, Faculdade de Ceilândia – FCE, Universidade de Brasília – UnB – Ceilândia (DF), Brasil.

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**Conflict of interests:** No.

**Authors' contribution:** TSM, CLBF and JOL conceived and designed the study; participated in data collection, analysis, and interpretation; wrote or reviewed the manuscript for important intellectual content; and gave final approval.

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## INTRODUCTION

The spinocerebellar ataxias (SCAs) are neurodegenerative diseases characterized by an increase in CAG (cytosine/adenine/guanine) trinucleotide repeats in certain genes<sup>(1)</sup>. SCA3, also known as Machado-Joseph disease (MJD), is the most prevalent form in Brazil<sup>(1)</sup>. Clinically, it is characterized by progressive ataxia, ophthalmoplegia, and varying degrees of pyramidal signs (such as hyperreflexia, spasticity, dysphagia, dysarthria), extrapyramidal signs (such as dystonia, stiffness and bradykinesia), or peripheral neuropathy with areflexia or amyotrophy<sup>(2)</sup>.

Dysarthria refers to speech disorders caused by failures in muscle control of speech-language mechanisms, resulting from insults to the central or peripheral nervous system<sup>(3)</sup>. Dysarthria changes one or more of the many functions—including phonation, respiration, articulation, resonance, and prosody—which must act in a concerted manner for adequate emission of speech<sup>(3)</sup>. In MJD, dysarthria is expected to worsen as the disease progresses. In one study<sup>(2)</sup> of 47 families affected by MJD, dysarthria was observed in 25% of individuals at 10 years of disease progression and 50% at 15 years. The key manifestations of ataxia-related dysarthria are generally characterized by vocal tremor, effort, and harshness during phonation (abnormalities in vocal stability), irregular interruptions during articulation, prolongation of phonemes and distortion of vowels and consonants, prosody featuring excessive stress, no differentiation of tonicity, and unsystematic excessive vocal loudness<sup>(3)</sup>.

Dysphagia—difficulty in passage of food from the oral cavity to the stomach—is one of the symptoms commonly present in the SCAs. It has considerable impact due to clinical complications and is directly associated with one of the leading causes of death, aspiration pneumonia<sup>(2)</sup>. The most frequent manifestations in MJD are constant choking, including on saliva, and weight loss<sup>(2)</sup>. Approximately 70% of patients develop dysphagia by the eighth year of disease progression and, after 15 years, this symptom progresses to a moderate or severe degree<sup>(2)</sup>.

Few studies have been conducted on speech-language therapy interventions in MJD. One case report<sup>(4)</sup> of a patient with mild dysarthria assessed 2 months of a weekly speech-language therapy intervention focusing on phonatory and respiratory function. The patient's condition alternated between stable and deteriorating and, despite follow-up with guidance and exercises every 2 months, dysarthria became more severe. In another intervention study including four cases of MJD diagnosed with mild dysphagia<sup>(5)</sup>, sessions consisting of orofacial myofunctional exercises, adaptation to consistencies, and direct therapy with facilitating, compensatory, and clearance maneuvers were held once weekly for 6 weeks. After the intervention, three of the four patients did not present any change in dysphagia and one experienced progression of manifestations.

A review of the literature found no reports of speech-language therapy interventions using the Lee Silverman Voice Treatment (LSVT) method in cases of MJD. However, in a patient with dysarthria due to cerebellar dysfunction secondary to thiamine deficiency<sup>(6)</sup>, the results suggested an increase in mean fundamental frequency and improvement in articulatory precision and intonation in the short and long term.

The LSVT method is an easy-to-learn, low-cognitive-demand program aimed at increasing vocal intensity through phonatory effort. Unique features include its focus on specific aspects, such

as intensive daily training, focus on voice, and self-monitoring<sup>(7)</sup>, and it has been shown to be effective in the treatment of dysarthria and dysphagia. Initially proposed for the treatment of Parkinson's disease (PD), it has also been used for other neurological conditions, such as progressive supranuclear palsy and multiple sclerosis, as well as presbyphagia<sup>(8-10)</sup>.

Although reports of LSVT interventions in MJD are not found in the literature, the method has been applied to a patient with ataxia<sup>(6)</sup>, who showed improvements in certain aspects of speech. The authors pointed out that ataxia-related dysarthria involves difficulty in adjustment of motor learning, that is, the ability to detect and correct errors, automate, and store the new necessary movement patterns. There is also difficulty in coordinating and stabilizing movement. These functions are performed by the cerebellum, in association with other areas, such as the striatum, limbic system, and prefrontal cortex. The aforementioned difficulties create a need for constant monitoring and control when attempting to execute movements. When movements are intended to create speech, these difficulties can become even worse because of the associated linguistic and cognitive demands.

Thus, according to Sapir et al.<sup>(6)</sup>, effective treatments for ataxic dysarthria must not require conscious effort for adjustment of automated pre-morbid speech patterns, should be of brief duration, and should impose no cognitive demands. LSVT meets all of these criteria and also acts to improve the vocal instability that occurs in these patients. Within this context, the objective of this case report was to ascertain the effects of intensive speech-language therapy using the LSVT method in an individual with progressive Machado-Joseph disease, from the points of view of the examiners and of the participant himself.

## PRESENTATION OF CASE

The study project was approved by the Faculdade de Ceilândia (CEP/FCE) Research Ethics Committee under protocol number CAAE 09573219.6.0000.8093 and the participant provided written informed consent.

The patient was a 33-year-old male who had graduated from advertising college and was self-employed as a freelancer in animation, graphic design, and video production. He presented to the outpatient neurology clinic of a university hospital for speech-language therapy. Six years previously, he had received a diagnosis of SCA3. His initial symptoms—difficulty walking and speech disorders, especially after intake of alcoholic beverages—had developed around 20 years of age. He had a family history of MJD; his father had also been diagnosed and died of sepsis secondary to aspiration pneumonia after 16 years of disease progression.

The initial and final evaluations were carried out by two speech-language therapists with clinical experience. An analysis of inter-rater agreement was performed. The patient was appropriately autonomous and reported independence for all activities of daily living. However, he required an assistive device (orthopedic walker) for ambulation and a caregiver when leaving the house. He was communicative and was able to respond to all commands verbally and independently. His behaviors were contextualized. He reported no weight loss since his diagnosis, claimed to be

monitored by a dietitian, and complained of frequent choking and dietary restrictions (dry foods). He reported never having undergone speech-language therapy interventions.

For clinical evaluation, the Dysarthria Protocol<sup>(3)</sup> was used, which consists of isolated and combined exploration of the five motor components of speech (respiration, phonation, resonance, articulation, and prosody). Vocal quality was classified using the GRBASI scale<sup>(11)</sup>: G, grade (overall degree of hoarseness), R, roughness (irregularity of vocal-fold vibration, hoarseness, bitonality), B, breathiness (degree of air escaping from the glottis), A, asthenia (weakness and loss of vocal power), S, strain (hyperfunctional use of phonation), and I, instability (fluctuation of vocal quality over time). The GRBASI is a 4-point scale used to assess the degree of vocal deviation from the norm: 0, normal; 1, slight or mild; 2, moderate; and 3, marked.

Speech intelligibility assessment was performed by having the patient read aloud a list of 10 monosyllabic words and 11 phrases arranged per protocol. The speech samples were transcribed phonetically by a lay listener, exactly as heard, and corrected based on their correspondence to the target words. If the transcription did not match the target word or phrase, that word or phrase was assigned a score of zero. After analysis, the percentage of correct answers was calculated. The initial diagnostic hypothesis from the speech-language pathology standpoint was dysarthria.

Speech samples were collected with a digital recorder, with the participant sitting in an upright posture and the microphone placed at a distance of 15 cm. Acoustic frequency weightings were performed by analysis of the sustained vowel /a/ in Voxmetria software (CTS Informática). The acoustic parameters selected for comparison were mean fundamental frequency (F0), jitter, shimmer, glottal to noise excitation (GNE) ratio, and the phonatory deviation diagram (PDD).

The F0 corresponds to the number of vocal fold pulses or cycles in one second. In men, values in a range of 80 to 150 Hz are expected<sup>(12)</sup>. Jitter is determined by the variation in frequency; when altered, it suggests decreased control of vocal fold vibration. It is associated with a rough vocal quality and, in the Voxmetria software suite, has a maximum normal value of 6.0%. Shimmer is determined by the variation in vocal intensity and, when altered, suggests a reduction in glottal resistance or a mass lesion. It correlates with the presence of noise during voice emission and with breathy voice quality<sup>(12)</sup>. In the Voxmetria software, values of up to 6.5% are considered normal. The GNE ratio is a parameter that measures whether the vocal signal originates from vibration of the vocal folds or from turbulence of the air current generated in the vocal tract. It is also associated with breathiness<sup>(13)</sup>. In Voxmetria, values from 0.5 to 1.0 dB are considered normal. The PDD is a feature exclusive to Voxmetria which correlates the acoustic measurements of jitter, shimmer, and their equivalents on the horizontal axis and the GNE ratio on the vertical axis, combining these parameters into a two-dimensional plot<sup>(13)</sup>.

For clinical evaluation of swallowing, the Joint Protocol for Functional Assessment of Swallowing<sup>(14)</sup> was used to characterize the events of the oral and pharyngeal phases. Positioning of the oral sensorimotor system at rest was structurally adequate; however, it was hypertonic and exhibited reduced mobility, velocity, amplitude, and precision of movements. The liquid (100 mL water), honey-thick (100 mL thickened water), and

solid (saltine cracker) consistencies were tested. Ad lib boluses of each consistency were offered. After functional assessment of swallowing, the diagnostic hypothesis from a speech-language pathology standpoint was refined to mild-to-moderate neurogenic oropharyngeal dysphagia.

Self-assessments were conducted using the Quality of life in Swallowing Disorders (SWAL-QOL) Protocol, which has been validated for MJD<sup>(15)</sup>. The protocol assesses 10 domains, which include swallowing as a burden, eating desire and duration, symptom frequency, communication, fear of eating, mental health, sleep, and fatigue, each measured on a Likert scale from 1 (“always”) to 5 (“never”). The food selection and social/role functioning domains allow answers based on the degree of agreement, also on a scale of 1 to 5, ranging from “agree completely” to “disagree completely”. The score is obtained by adding the values of each domain and dividing the sum by the number of items in each one. Scores range from 0 to 100. The higher the score, the higher the swallowing-related quality of life.

The Voice-Related Quality of Life (V-RQOL) questionnaire<sup>(16)</sup>, which consists of a list of 10 possible voice-related problems scored on a Likert scale from 1 (“it never happens and it’s not a problem”) to 5 (“it happens all the time and it really is a problem”), was also administered. The score is calculated according to a specific algorithm; the higher the score, the higher the voice-related quality of life.

Due to the similarity of the patient’s condition and a case of dysarthria resulting from cerebellar dysfunction previously described in the literature<sup>(6)</sup>, the intervention consisted of the Lee Silverman Voice Treatment (LSVT) method<sup>(7)</sup>, which aims to increase vocal intensity through an intensive program of calibration and hierarchical exercises involving spontaneous speech and structured reading. The LSVT was applied by a third speech-language therapist who had been certified in the method. As standard, 16 individual 1-hour sessions were administered, on four consecutive days per week for 4 weeks. In all sessions, a stopwatch and sound level meter placed 30 cm apart were used. The participant was instructed to practice at home on the days of the week in which no sessions were scheduled.

Reassessment took place 1 week after the last session and lasted approximately 2 hours. Chart 1 describes the clinical results of speech articulation and deglutition assessment (practitioner-centered outcomes), while Chart 2 shows the results of the self-assessment protocols (participant-centered outcomes).

Improvement was observed in all motor components of speech (respiration, phonation, resonance, articulation, and prosody), as was a reduction in signs of dysphagia. Regarding vocal quality, there was a reduction in hoarseness and instability, normalization of jitter and shimmer, increase in vocal intensity, and improvement in coordination and number of utterances (words and phrases) per exhalation, as well as a slight improvement in diadochokinesis. Self-assessment with the SWAL-QOL<sup>(15)</sup> showed higher scores in the eating desire and duration, symptom frequency, food selection, communication, role functioning, and sleep domains. Nevertheless, domains of a more emotional nature, such as fear of eating and mental health, scored lower than at baseline. The burden and fatigue domain scores remained unchanged. On reassessment with the V-RQOL<sup>(16)</sup>, higher scores were obtained in all domains, demonstrating improvement in the participant’s voice-related quality of life.

**Chart 1.** Clinical examination findings (practitioner-centered outcomes)

	BEFORE INTERVENTION	AFTER INTERVENTION
<b>RESPIRATION</b>	1- Mixed chest/clavicular type	1- Chest type
	2- Oronasal breathing mode	2- Oronasal breathing mode
<b>PHONATION</b>	1- Maximum phonation time (MPT) /a/=11s. Hard (glottal) vocal onset G2 R2 B2 A0 S1 I2 2- s/=26s / /z/=16s s/z ratio: 1.6 = suggestive of glottic hypercontraction 3- Words per exhalation: - Number counting = 2 PE - Text reading = 15 PE Rough/breathy, strained utterances  4- Low pitch 5- Weak/appropriate loudness	1- Maximum phonation time (MPT) /a/= 10s Near-simultaneous voice onset, G1 R1 B0 A0 S0 I1 2- /s/= 15s/ /z/= 15s s/z ratio: 1.0 = within normal limits  3- Words per exhalation: - Number counting = 4 PE - Text reading = 11 PE Reduction in roughness, breathiness, and strain. Improved coordination of respiration and phonation  4- High pitch 5- Appropriate loudness
<b>RESONANCE</b>	1- Laryngopharyngeal, moderate	2- Laryngopharyngeal, slight
<b>ARTICULATION</b>	1- Labial movements, /i – u/: inconsistent/irregular, with tremor; velocity: 5 seconds for 5 repetitions 2- Lingual movements, /ka – ta/: distortion of phonemes and associated facial movements 3- Speech intelligibility > 80% - Monosyllables: 80% - Full sentences: 54.5%	1- Labial movements, /i – u/: reduced inconsistency and tremor, velocity: 7 seconds for 5 repetitions 2- Lingual movements, /ka – ta/: only slight incoordination 3- Speech intelligibility > 80% - Monosyllables: 100% - Full sentences: 90.9%
<b>PROSODY</b>	1- Inadequate marking of stressed syllable; no change in intonation within and between sentences	1- Sporadic inadequacy in marking of stressed syllable; improved intonation for interrogative sentences.
<b>ORAL MOTOR PARAMETERS</b>	1- Diadochokinesis: inconsistent in frequency and intensity, tremor of structures of phonation and articulation, no identifiable identical interval between syllables, syllables distorted and poorly produced, decreased labial range of motion	1- Diadochokinesis: improved consistency in frequency and intensity, more appropriate interval between syllables, few or no distorted or poorly produced syllables, appropriate labial range of motion
<b>SOUND LEVEL MEASUREMENT</b>	Mean: 75 dB	Mean: 82 dB
<b>DEGLUTITION (LIQUID/HONEY-THICK CONSISTENCY)</b>	- Decreased bolus ejection - Oral transit time adequate - Decreased oropharyngeal coordination - Laryngeal elevation decreased - Choking and coughing present - Cervical auscultation abnormal - Bolus sensation - Desaturation (96% SpO <sub>2</sub> to 93% SpO <sub>2</sub> ) during deglutition of liquid consistency. No desaturation during deglutition of honey-thick consistency - No vocal changes	- Improved bolus ejection - Oral transit time adequate - Improved oropharyngeal coordination - Appropriate laryngeal elevation - No choking or coughing - Cervical auscultation normal - Absence of bolus sensation - Oxygen saturation stable during deglutition
<b>MASTICATION (SOLID CONSISTENCY)</b>	- Bites with incisors - Simultaneous bilateral mastication	- No vocal changes - Bites with incisors - Simultaneous bilateral mastication
<b>DEGLUTITION (SOLID CONSISTENCY)</b>	- Decreased bolus ejection - Oral transit time slowed (to position and eject bolus) - Laryngeal elevation decreased - Presence of oral residue - Decreased oropharyngeal coordination - No vocal changes - Bolus sensation, liquid required to clear bolus	- Improved bolus ejection - Oral transit time adequate - Laryngeal elevation adequate - Absence of oral residue - Improved oropharyngeal coordination - No vocal changes - Absence of bolus sensation

**Subtitle:** MPT = maximum phonation time; PE = per exhalation; dB = decibels; SpO<sub>2</sub> = peripheral oxygen saturation

**Chart 2.** Participant-centered outcomes

	<b>BASELINE</b>	<b>FINAL</b>
<b>SWAL-QOL BURDEN DOMAIN</b>	75	75
<b>SWAL-QOL EATING DESIRE AND DURATION DOMAIN</b>	30	34
<b>SWAL-QOL SYMPTOM FREQUENCY DOMAIN</b>	32	36
<b>SWAL-QOL FOOD SELECTION DOMAIN</b>	30	50
<b>SWAL-QOL COMMUNICATION DOMAIN</b>	20	40
<b>SWAL-QOL FEAR DOMAIN</b>	25	17
<b>SWAL-QOL MENTAL HEALTH DOMAIN</b>	22	10
<b>SWAL-QOL SOCIAL FUNCTIONING DOMAIN</b>	10	28
<b>SWAL-QOL SLEEP DOMAIN</b>	40	50
<b>SWAL-QOL FATIGUE DOMAIN</b>	50	50
<b>V-RQOL SOCIAL-EMOCIONAL DOMAIN</b>	0	50
<b>V-RQOL PHYSICAL FUNCTIONING DOMAIN</b>	25	50
<b>V-RQOL TOTAL SCORE</b>	15	50

Subtitle: SWAL-QOL = Quality of life in Swallowing Disorders; V-RQOL = Voice-Related Quality of Life

## DISCUSSION

The objective of this case report was to ascertain the effects of an intensive speech-language therapy intervention with the LSVT method in a patient with MJD. Clinical and instrumental assessment revealed improvement in all motor components of speech (respiration, phonation, resonance, articulation, and prosody), as was a reduction in signs of dysphagia.

Although previous reports of LSVT-based interventions in MJD were not found in the literature, in a similar case<sup>(6)</sup> of dysarthria due to cerebellar dysfunction, the authors reported short- and long-term improvements, such as increased phonatory strength, stability, and articulatory precision, better intonation, and speech intelligibility. These findings were similar to those of the present study, although long-term gains have not been assessed. However, according to the literature<sup>(7)</sup>, the gains obtained with the method persist for approximately 2 years in cases of Parkinson's disease, which allows maintenance and future follow-up.

Although ataxia is constant in MJD, it also features changes in the pyramidal, extrapyramidal, and peripheral nervous systems, resulting in multiple changes in vocal quality, such as breathiness, roughness, and strain<sup>(17)</sup>. The patient's vocal quality was initially classified as  $G_2R_2B_2A_0S_1I_2$ , indicating overall abnormality, hoarseness, breathiness, and instability with moderate deviation and slight strain at the end of utterances. After the intervention, the score had improved to  $G_1R_1B_0A_0S_0I_1$ , indicating suppression of breathiness and strain and reductions in the degree of hoarseness and instability.

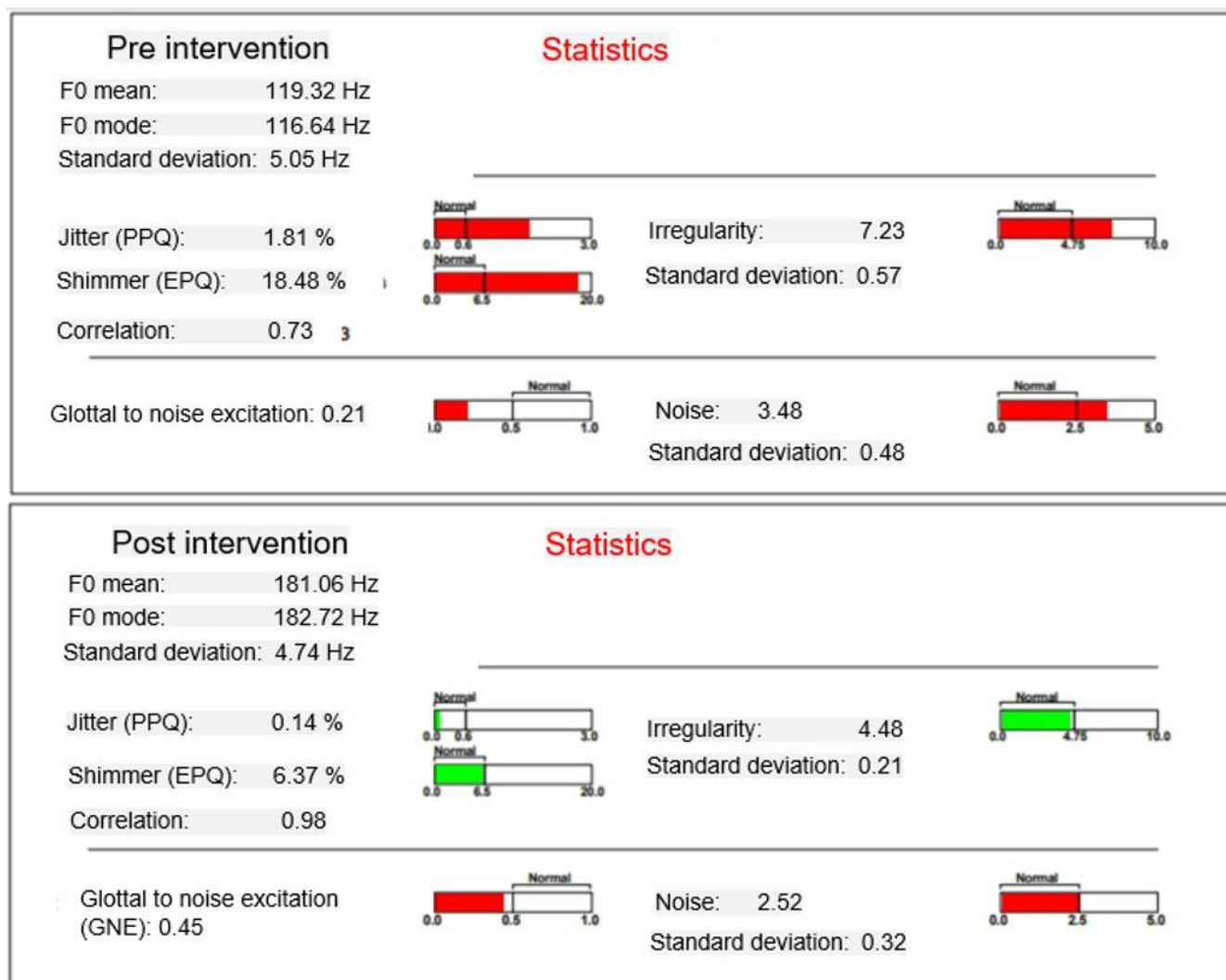
On time-course assessment, the maximum phonation time (MPT), which estimates airflow control during phonation<sup>(12)</sup>, was reduced by a few seconds after the intervention. However, the MPT is measured on the basis of sustained emission of a vowel, and difficulties in carrying out this test are common in cases of neurological disorders, because such emission requires precise central nervous system control in coordinating the myoelastic forces of the larynx and aerodynamics of the air current<sup>(12)</sup>. Despite this fact, there was a qualitative increase in the participant's perceptual analysis, as demonstrated by improvement of vocal onset to near-instantaneous, decrease in strain, and improvement in phonation stability (Chart 1). The participant's F0 increased from 119.32 Hz to 181.06 Hz

after the intervention (Figure 1), which is consistent with the results of the method in a similar case<sup>(18)</sup>. The post-intervention F0 exceeds normative values for men (up to 150 Hz)<sup>(12)</sup>, thus corresponding to a high-pitched voice. However, fluctuations in frequency are to be expected in patients with cerebellar changes, due to the lack of proprioceptive brain monitoring<sup>(17)</sup>. The change in resonance after the intervention (Chart 1) and improvement in voice projection and intensity may also have contributed to the increase in fundamental frequency.

After the LSVT intervention (Figure 1), the participant's jitter and shimmer also improved to normal range. The acoustic parameters demonstrated an increase in vocal fold vibration control, an increase in glottic resistance, and a decrease in breathiness, consistent with the perceptual interpretations of the participant's voice. Although the communicative manifestations of Machado-Joseph disease are different from those found in Parkinson's disease, our results are in agreement with previous studies of patients with Parkinson's disease treated with LSVT<sup>(7)</sup>, which demonstrated improvement in glottic closure and vocal fold vibration after the intervention.

The participant's GNE ratio (Figure 1) increased from 0.21 dB to 0.45 dB (near-normal range). This increase in GNE ratio explains the reduction in breathiness in the participant's voice<sup>(13)</sup>; however, the GNE was still abnormal because the participant's vocal quality remained hoarse, despite improvement in acoustic and perceptual values. Before the intervention, the PDD<sup>(13)</sup> was concentrated in the upper right quadrant, suggesting moderate deviation; after the intervention, variation was observed in the representation quadrant, approaching normality and with a more concentrated distribution of points (Figure 2).

In a sample of 31 individuals diagnosed with MJD, one study<sup>(17)</sup> found that articulation was characterized by consonant inaccuracy, prolongation of sounds, distorted vowels, repetition, and interruptions of articulation. Despite manifestations similar to those observed in the aforementioned study<sup>(17)</sup>, such as articulatory imprecision and phonemic distortions, our patient also presented with lip tremors and associated facial movements at baseline. After the intervention, there was a decrease in inconsistency and tremor during articulatory movements. In addition, a decrease in velocity of articulatory movements was observed—a factor that considerably impacted the improvement of his speech intelligibility, and a known positive consequence of LSVT as described elsewhere in the literature<sup>(6)</sup>.



**Figure 1.** Mean F0, jitter, shimmer, and glottal to noise excitation ratio before and after intervention

**Subtitle:** F<sub>0</sub> = fundamental frequency; PPQ = pitch perturbation quotient; EPQ = energy perturbation quotient; GNE = glottal to noise excitation ratio

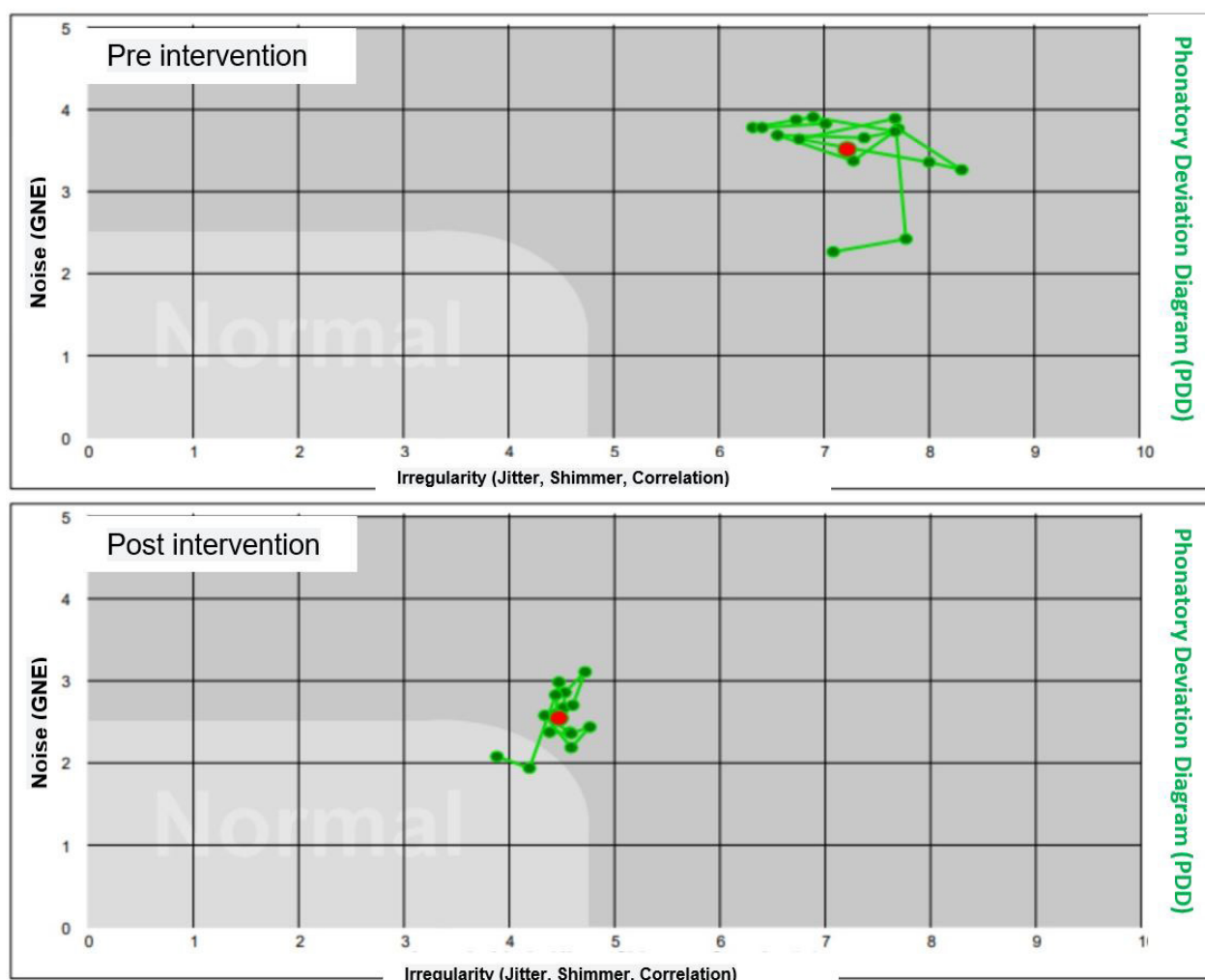
Assessment of diadochokinesis reflects an individual's neuromotor maturity and integration. In the same study of MJD mentioned above<sup>(17)</sup>, the articulatory diadochokinesis of the sample was considered slow and irregular, as in the patient described in the present case report. After the LSVT intervention, the participant's diadochokinesis showed slight improvement. However, Wolf et al.<sup>(17)</sup> noted that parameters such as younger age of disease onset and a higher CAG repeat burden have a moderate correlation with slowness on diadochokinesis assessment. Although diagnosed at 27, the participant reported noticing manifestations of the disease as early as age 20, which somewhat explains his slight improvement in this test. It should be noted that the LSVT method has been scientifically proven to yield benefit in Parkinson's disease, which has a different pathophysiology than MJD.

Nevertheless, after intervention the participant's speech was better paced, with less inconsistency in frequency and intensity, no tremor or syllable distortion, and the interval between syllables was appropriate, as was the amplitude and movement of the lips. Improved control of meter, rhythm, and verbal fluency was also observed in the participant's speech—factors that positively influenced his overall communication ability.

Wolf et al.<sup>(17)</sup> found that prosody was one of the most affected systems in their sample of individuals with MJD, due to changes in rhythm, reduction of stress, variation in velocity, inappropriate pauses, and prolonged intervals. Likewise, the participant in this case report had inadequate marking of stressed syllables and a relatively flat affect. After the LSVT intervention, he started to better adjust the tone of his words and improved his intonation, especially for interrogative sentences. However, the examiners agreed that prosody could be further explored, which would require specific intervention with more traditional training methods.

After intervention, clinical evaluation of swallowing revealed effective bolus ejection and oropharyngeal coordination and absence of coughing, choking, vocal changes, or oxygen desaturation. A pilot study<sup>(18)</sup> found that application of the Lee Silverman method in cases of Parkinson's disease improved not only phonation but also neuromuscular control of the entire upper aerodigestive tract. Therefore, we believe the benefits observed in the present case are attributable to the intervention.

The patient's view of the intervention, as obtained from self-assessment protocols, was consistent with the clinical examination and objective assessments. SWAL-QOL<sup>(15)</sup> scores



**Figure 2.** Phonation disorder diagram plot before and after intervention

**Subtitle:** GNE = glottal to noise excitation ratio

after the intervention were reduced only in domains of an emotional character, such as mental health and fear of eating, unlike in a previous study, also carried out after a speech-language therapy intervention<sup>(6)</sup>, which listed the fear of eating domain as one of the three most improved after therapy. We suggest that these results are due to the fact that the participant was young, educated about the progression of his disease, and was instructed by the examiners on the concept of dysphagia, its causes and consequences. Scores in all V-RQOL domains<sup>(16)</sup> improved after the intervention, expressing how the participant's perception of his own speech changed. This demonstrates that the participant identified himself as more communicative, socially interactive, and found it less difficult to speak loudly. Satisfaction in relation to the intervention and its outcomes indicates that a high degree of benefit was achieved, with consequent improvements in the participant's quality of life.

## CLOSING REMARKS

Studies such as this one aim to explore the behavior of MJD in response to speech-language therapy, and can contribute to the development of tools for both practitioner-centered and patient-centered assessment after the intervention. The LSVT

proved to be a viable option for intensive intervention in cases of speech and swallowing disorders in MJD. Limitations include the single-subject design, which prevents generalization of our findings and comparison of the benefits of the intervention in other individuals with MJD. Further studies are needed to attest to the effectiveness of the Lee Silverman method in cases of neurogenic dysarthria and dysphagia caused by Machado-Joseph disease, with particular focus on the long-term effects of such interventions, in order to maximize the quality of life of these patients and guide their speech-language therapy management.

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