

Man in the Barrel Syndrome, a rare symmetrical proximal amyotrophic diplegia

Síndrome do homem no barril, um relato de caso de uma rara diplegia amiotrófica proximal

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ABSTRACT

Introduction: Brachial amyotrophic diplegia (BAD), or flail arm syndrome, historically regarded as a variant of progressive muscular atrophy, is now recognized as a distinct regional lower motor neuron phenotype within the motor neuron disease spectrum, characterized by progressive weakness and atrophy with proximal upper-limb predominance. When bilateral brachial involvement is proximal, it may manifest a clinical phenotype resembling man-in-the-barrel syndrome.

Case report: A 62-year-old man presented with progressive bilateral upper-limb weakness and amyotrophy, with proximal predominance, evolving for 23 months. Neurological examination showed a brachial-predominant pattern, proximal lower-limb weakness, no sensory abnormalities, and no clinical signs of upper motor neuron, bulbar, or respiratory involvement. Electroneuromyography demonstrated a chronic, purely motor, preganglionic neurogenic pattern. Extensive laboratory, neuroimaging, respiratory, swallowing, and genetic investigations revealed no alternative etiologies.

Discussion: The symmetrical, purely motor, lower motor neuron pattern, with proximal brachial predominance, supported the clinical phenotype of man-in-the-barrel syndrome associated with BAD. Although proximal lower-limb involvement was observed, the clinical picture remained dominated by upper-limb impairment, situating the case along the continuum between a regional brachial variant and lower motor neuron-predominant motor neuron disease.

Conclusion: This case contributes to the recognition of proximal brachial phenotypes within the motor neuron disease spectrum and reinforces the importance of systematic diagnostic workup to exclude treatable mimics and refine prognostic counseling.

Keywords: Amyotrophic Lateral Sclerosis; Motor Neuron Disease; Muscular Atrophy; Upper Extremity; Differential Diagnosis; Case Reports.

RESUMO

Introdução: A diplegia braquial amiotrófica (DBA), ou síndrome dos braços pendentes, historicamente considerada uma variante da atrofia muscular progressiva, é atualmente reconhecida como um fenótipo regional distinto de neurônio motor inferior dentro do espectro das doenças do neurônio motor, caracterizado por fraqueza e atrofia progressivas com predomínio proximal nos membros superiores. Quando o acometimento braquial bilateral é proximal, pode manifestar fenótipo clínico semelhante à síndrome do homem no barril.

Relato de caso: Homem de 62 anos apresentou fraqueza e amiotrofia progressivas e bilaterais dos membros superiores, com predomínio proximal, em evolução há 23 meses. O exame neurológico evidenciou acometimento braquial predominante, fraqueza proximal em membros inferiores, ausência de alterações sensitivas e ausência de sinais de neurônio motor superior, bulbares ou respiratórios. A eletroneuromiografia demonstrou padrão neurogênico crônico, puramente motor, pré-ganglionar. A investigação laboratorial, de neuroimagem, respiratória, da deglutição e genética não revelou etiologias alternativas.

Discussão: O padrão simétrico, puramente motor, de neurônio motor inferior, com predomínio braquial proximal, sustentou o fenótipo clínico de síndrome do homem no barril associado à DBA. Embora houvesse comprometimento proximal de membros inferiores, o quadro permaneceu clinicamente dominado pelos membros superiores, situando o caso no continuum entre variante braquial regional e doença do neurônio motor de predomínio em neurônio motor inferior.

Conclusão: Este caso contribui para o reconhecimento de fenótipos braquiais proximais no espectro das doenças do neurônio motor e reforça a importância da investigação sistemática para excluir mimetizadores tratáveis e refinar o aconselhamento prognóstico.

Palavras-Chave: Esclerose Lateral Amiotrófica; Doenças do Neurônio Motor; Atrofia Muscular; Extremidade Superior; Diagnóstico Diferencial; Relatos de Casos.

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INTRODUCTION

Amyotrophic lateral sclerosis (ALS) is a neurodegenerative disease characterized by degeneration of both upper and lower motor neurons, with a median survival of 3 to 4 years¹. Three classic clinical presentations have been described, each with distinct prognostic correlations: bulbar-onset disease, which has a more rapidly progressive course; limb-onset disease; and progressive muscular atrophy (PMA), a pure lower motor neuron form classically characterized by asymmetric, predominantly distal weakness with comparatively generalized progression^{1,2}. Rarer regional phenotypes, in which weakness remains predominantly confined to a single motor territory for one to two years before spreading to other regions, have also been described¹. Brachial amyotrophic diplegia (BAD) and leg amyotrophic diplegia (LAD) predominantly affect the upper and lower limbs, respectively, whereas isolated bulbar ALS keeps symptoms restricted to the bulbar region for a prolonged period and may show both upper and lower motor neuron involvement¹. Although BAD was historically regarded as a regional variant of PMA, it is now more appropriately considered a distinct regional lower motor neuron phenotype within the motor neuron disease spectrum, differing from classical PMA in its predominantly proximal and symmetrical upper-limb involvement, its prolonged regional confinement, and its generally more favorable prognosis².

In 1999, Katz et al. formalized BAD as a distinct nosological entity, proposing clinical diagnostic criteria for a phenotype previously recognized in the literature³. The classic BAD presentation consists of progressive, symmetrical weakness and atrophy predominantly affecting the upper limbs, typically proximal, without signs of upper motor neuron or bulbar involvement; involvement of the lower limbs may occur later in the disease course but remains comparatively milder and slower to progress^{3,4}.

This report describes a case of BAD in which the disease began in, and remained predominantly confined to, the upper limbs over a 23-month course, with proximal lower-limb involvement progressing comparatively more slowly, together with an extensive diagnostic workup negative for alternative diagnoses, contributing to the recognition of this entity in the Brazilian context.

CASE REPORT

A 62-year-old retired man with no relevant comorbidities presented with progressive muscle weakness and atrophy predominantly affecting the proximal segments of both upper limbs, beginning on September 12, 2024. At onset, he denied gait impairment or falls, and dyspnea, dysphonia, and dysarthria were neither reported

nor observed. Proximal lower-limb weakness was detected during the disease course; however, the clinical phenotype remained dominated by bilateral proximal upper-limb involvement. The process was associated with marked weight loss.

On neurological examination, muscle tone was reduced in all four limbs. Muscle strength showed a brachial-predominant pattern with marked proximal involvement. In the upper limbs, the biceps brachii was severely affected bilaterally (Medical Research Council grade 2), with less severe triceps weakness (grade 3) and relative preservation of distal muscles, including wrist extensors, finger flexors, and intrinsic hand muscles (grade 4). Proximal lower-limb weakness was also observed, mainly affecting the iliopsoas (grade 2) and quadriceps (grade 3), whereas distal lower-limb strength was relatively preserved. Muscle atrophy was most conspicuous in the upper limbs, with a proximal distribution. Fasciculations were noted in all four limbs. Deep tendon reflexes were abolished (0/4+) in all four limbs. Plantar responses were flexor and the Hoffmann sign was absent. Superficial and deep sensation were preserved, and no bulbar signs were present. Overall, the examination showed a proximal, brachial-predominant lower motor neuron syndrome, with associated proximal lower-limb involvement and without sensory abnormalities or pyramidal signs.

Laboratory investigation was unremarkable, including screening for immune-mediated, infectious, paraneoplastic, vitamin-deficiency-related, and other conditions that could mimic motor neuron disease; anti-GM1 antibodies were non-reactive. Cerebrospinal fluid analysis was normal. Electroneuromyography demonstrated active denervation, characterized by fibrillation potentials and positive sharp waves, together with chronic reinnervation, characterized by high-amplitude, long-duration, polyphasic motor unit potentials. Fasciculation potentials were also observed. Sensory and motor nerve conduction studies showed normal conduction velocities, without conduction block, supporting a chronic, preganglionic, purely motor neurogenic pattern consistent with anterior horn cell involvement. Motor evoked potentials of the upper and lower limbs did not support upper motor neuron involvement, and brain MRI with tractography showed no evidence of corticospinal tract involvement. Genetic testing revealed no pathogenic variants associated with motor neuron disease. Swallowing assessment and pulmonary function testing were normal.

Over approximately 23 months of clinical evolution, no bulbar, respiratory, or upper motor neuron involvement developed. Despite associated proximal lower-limb weakness, the overall clinical picture remained dominated by a proximal, upper-limb-predominant lower motor neuron syndrome, clinically reminiscent of the man-in-the-barrel phenotype at presentation, as shown in the photographs (Figures 1A-1B).

1A



1B



Figures 1A and 1B - Clinical photographs showing bilateral upper-limb amyotrophy with relative preservation of the lower limbs, consistent with an upper-limb-predominant motor phenotype and clinically resembling the man-in-the-barrel phenotype. Figure 1A - Frontal view; Figure 1B - Lateral view.

DISCUSSION

The patient presented bilateral, symmetrical weakness and atrophy with upper-limb predominance, without signs of upper motor neuron involvement, over a period of 23 months. The electroneuromyography findings, showing a purely motor, preganglionic neurogenic pattern, together with a negative workup for treatable causes, fulfill the diagnostic criteria for BAD³.

Katz et al. described proximal involvement as the predominant pattern in BAD, a finding also observed in the nine Brazilian cases reported by Orsini et al.^{3,4}. In the present case, although mild proximal lower-limb involvement and mild distal upper-limb weakness were also observed, the most clinically defining impairment remained bilateral, proximal, and brachial-predominant, supporting the clinical diagnosis of man-in-the-barrel syndrome associated with proximal brachial amyotrophic diplegia.

The differential diagnosis of BAD requires active exclusion of other motor neuron conditions and mimics. Multifocal motor neuropathy (MMN) was particularly relevant because of the upper-limb involvement; however, the symmetrical pattern, absence of conduction block on electroneuromyography, and negative anti-GM1 antibody testing argued against this diagnosis⁵. Adult-onset spinal

muscular atrophy (SMA type 4) was excluded by SMN1/SMN2 genetic analysis³. Spinal and bulbar muscular atrophy (SBMA) was considered less likely because of the absence of bulbar involvement, tongue fasciculations, or respiratory impairment, in addition to negative genetic testing⁶.

The Gold Coast criteria were considered because the patient presented with progressive motor impairment and lower motor neuron findings⁷. Although these criteria are useful for diagnostic classification within the ALS spectrum, the patient's initial presentation and longitudinal course were clinically dominated by marked proximal brachial involvement, without upper motor neuron signs, bulbar involvement, respiratory impairment, or sensory abnormalities. Therefore, the case was interpreted phenotypically as a brachial-predominant lower motor neuron disease presentation within the BAD/flail-arm spectrum, rather than as a classic ALS phenotype. This distinction is clinically relevant because brachial amyotrophic diplegia/flail-arm presentations may follow a slower and more regionally predominant course than classic ALS^{1,3}.

Other differentials included Hirayama disease and cervical spondylotic amyotrophy. Hirayama disease was unlikely because of the patient's age, bilateral proximal

brachial predominance, lower-limb involvement during follow-up, and absence of the typical juvenile-onset distal hand and forearm pattern⁸. Cervical spondylotic amyotrophy was considered because it may present with upper-limb weakness and amyotrophy with minimal sensory involvement; however, the symmetrical bilateral distribution, absence of radicular sensory symptoms or pyramidal signs, and broader lower motor neuron pattern favored a motor neuron disease phenotype rather than a compressive cervical etiology⁹.

The clinical course also distinguishes this presentation from classical PMA. Whereas PMA typically manifests as asymmetric, predominantly distal weakness with comparatively generalized progression, our patient showed symmetrical, predominantly proximal upper-limb weakness that remained the defining feature throughout a 23-month course, with only milder and later lower-limb involvement, a pattern consistent with BAD and associated with a more favorable prognosis^{2,10}.

Recognition of BAD has direct clinical implications, as its more favorable course, characterized by slower progression and prolonged preservation of bulbar and respiratory function, distinguishing it from classic ALS and guides prognostic counseling for patients and their families^{1,3}. This distinction may also prevent premature classification as typical ALS in patients with a regionally restricted phenotype, particularly in the absence of upper motor neuron signs or spread to other motor territories. From a therapeutic standpoint, management of BAD remains poorly defined and is usually extrapolated from classic ALS; although riluzole has demonstrated benefit in ALS, its role in slowly progressive regional variants such as pure BAD is not well established¹¹.

CONCLUSION

BAD remains a rare and frequently underrecognized motor neuron disease variant. This report contributes to its recognition in the Brazilian context and illustrates a proximal, upper-limb-predominant phenotype clinically resembling man-in-the-barrel syndrome, in which involvement of other motor territories may develop but progresses more slowly and remains comparatively milder than the brachial impairment. A systematic diagnostic workup and familiarity with current clinical criteria are essential to distinguish BAD from more aggressive forms of motor neuron disease and from potentially treatable mimics.

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