Disease Severity and Its Associations With Non-Ataxic Signs, Cognition, Mobility, and Balance In Spinocerebellar Ataxia: A Cross-Sectional Study

Gravidade Da Doença e Suas Associações Com Sinais Não Atáxicos, Cognição, Mobilidade e Equilíbrio Na Ataxia Espinocerebelar: Um Estudo Transversal

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ABSTRACT

Background: Spinocerebellar ataxias (SCA) are progressive neurodegenerative disorders. Severity ratings are commonly used to describe disease progression, yet how ataxia severity relates to extracerebellar (non-ataxic) signs, cognitive screening, mobility, and balance remains incompletely understood.
Objective: To examine associations between ataxia severity and non-ataxic signs, cognition, mobility, and balance in SCA and to explore the influence of functional stage.
Methods: In this cross-sectional study, 42 individuals with SCA were assessed with the Scale for the Assessment and Rating of Ataxia (SARA), Inventory of Non-Ataxia Signs (INAS), Berg Balance Scale (BBS), Timed Up and Go (TUG), and Mini-Mental State Examination (MMSE). Participants were classified by Klockgether stage (stage 1 vs stage 2). Associations with SARA were tested using Spearman's rho; partial correlations controlled for Klockgether stage.
Results: Stage 2 showed higher SARA scores and worse function than stage 1, with lower BBS scores and longer TUG times (all p<0.001). INAS was higher in stage 2 but not significant (p=0.056), and MMSE did not differ (p=0.780). SARA correlated with INAS (rho=0.385; p=0.012), BBS (rho=−0.852; p<0.001), and TUG (rho=0.773; p<0.001), but not with MMSE (rho=0.167; p=0.289). After adjustment, associations remained for BBS (rho=−0.752; p<0.001) and TUG (rho=0.642; p<0.001), but not for INAS (rho=0.266; p=0.089).
Conclusion: Greater ataxia severity was strongly associated with poorer balance and mobility, supporting combined use of SARA with BBS and TUG to characterize functioning and guide physiotherapy planning. Lack of association with MMSE should not be interpreted as absence of cognitive impairment; domain-specific cognitive testing and longitudinal studies are warranted.

Keywords: Spinocerebellar Ataxias; Disease Progression; Cognition; Mobility Limitation; Postural Balance; Activities of Daily Living.

RESUMO

Introdução: As ataxias espinocerebelares (SCA) são doenças neurodegenerativas progressivas. As classificações de gravidade são comumente usadas para descrever a progressão da doença, mas ainda não se compreende completamente como a gravidade da ataxia se relaciona com sinais extracerebelares (não atáxicos), triagem cognitiva, mobilidade e equilíbrio.
Objetivo: Examinar as associações entre a gravidade da ataxia e sinais não atáxicos, cognição, mobilidade e equilíbrio na SCA e explorar a influência do estágio funcional.
Métodos: Neste estudo transversal, 42 indivíduos com SCA foram avaliados com a Escala de Avaliação e Classificação da Ataxia (SARA), o Inventário de Sinais Não Ataxicos (INAS), a Escala de Equilíbrio de Berg (BBS), o Teste Timed Up and Go (TUG) e o Mini-Exame do Estado Mental (MMSE). Os participantes foram classificados pelo estágio de Klockgether (estágio 1 vs estágio 2). As associações com a SARA foram testadas usando o rho de Spearman; correlações parciais controladas para o estágio de Klockgether.
Resultados: O estágio 2 apresentou pontuações SARA mais altas e pior função do que o estágio 1, com pontuações BBS mais baixas e tempos TUG mais longos (todos p<0,001). O INAS foi mais elevado no estágio 2, mas não significativo (p=0,056), e o MMSE não diferiu (p=0,780). A SARA correlacionou-se com o INAS (rho=0,385; p=0,012), o BBS (rho=−0,852; p<0,001) e o TUG (rho=0,773; p<0,001), mas não com o MMSE (rho=0,167; p=0,289). Após o ajuste, as associações permaneceram para o BBS (rho=−0,752; p<0,001) e o TUG (rho=0,642; p<0,001), mas não para o INAS (rho=0,266; p=0,089).
Conclusão: Uma maior gravidade da ataxia esteve fortemente associada a um pior equilíbrio e mobilidade, apoiando a utilização combinada do SARA com o BBS e o TUG para caracterizar o funcionamento e orientar o planejamento da fisioterapia. A falta de associação com o MMSE não deve ser interpretada como ausência de déficit cognitivo; são necessários testes cognitivos específicos do domínio e estudos longitudinais.

Palavras-chave: Ataxias espinocerebelares; Progressão da doença; Cognição; Limitação da mobilidade; Equilíbrio postural; Atividades da vida diária.

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INTRODUCTION

Spinocerebellar ataxia (SCA) is a heterogeneous group of hereditary diseases characterized by various genetic mutations that lead to the progressive degeneration of the cerebellum and its connections[1]. This degeneration compromises motor coordination, balance, and, consequently, functionality[2]. As the disease progresses, individuals with SCA experience increasing difficulties in activities of daily living, an increased risk of falls, and a gradual loss of functional independence[3,4]. SCA 3, also known as Machado-Joseph disease, is the most prevalent subtype globally, accounting for approximately 15% to 45% of diagnosed cases[5]. Although all types of SCA share progressive cerebellar degeneration, each subtype has distinct clinical manifestations that vary according to the regions of the nervous system affected[1,6]. In addition to the characteristic ataxic signs, individuals with SCA may also present extracerebellar manifestations (non-ataxic signs), such as areflexia, hyperreflexia, urinary dysfunction, and spasticity[7].

Beyond motor dysfunction, studies indicate that cerebellar disorders are associated with cognitive deficits, including executive dysfunction, visuospatial difficulties, memory deficits, and reduced verbal fluency[8]. Because cognition can influence everyday task performance and safety, cognitive impairment may add to functional limitations even among individuals with similar levels of motor severity. However, despite evidence of both motor and cognitive dysfunction in SCA, there is a lack of research investigating how these domains interact to shape overall functioning. It remains unclear whether cognitive performance is associated with ataxia severity and if cognition relates to functional outcomes such as mobility and balance.

Clinical tools such as the Scale for the Assessment and Rating of Ataxia (SARA), the Berg Balance Scale (BBS), and the Timed Up and Go (TUG) test are commonly used in SCA[9]. In clinical and research settings, measures of mobility and balance are frequently used as indicators of functional performance and fall-related risk. Nevertheless, the relationships between ataxia severity, cognition, and functional performance (mobility and balance) remain insufficiently characterized in people with SCA. Given the progressive and complex nature of SCA, a more integrated approach to assessment is essential. Characterizing the associations between ataxia severity, cognitive screening, and functional performance (mobility and balance) may update rehabilitation strategies aimed at preserving independence and quality of life.

Therefore, this study aims to examine how ataxia severity (SARA) relates to functional performance (mobility and balance) and cognitive screening in individuals with SCA. Specifically, we will test correlations between SARA and MMSE, BBS, and TUG, and compare outcomes across disease stages. Non-ataxic signs (INAS) will be explored as a complementary clinical feature to better characterize the sample. These analyses may help identify key variables for therapeutic planning.

MATERIALS AND METHODS

This is a cross-sectional observational study, carried out and described using the STROBE checklist[10]. The study was conducted between November 2024 and October 2025 and followed the ethical guidelines established by CNS resolution 466/12 and was approved by the local ethical committee (CAAE 70797823.1.0000.5235). In this study, for the cross-sectional analysis, we used the initial data from a randomized clinical trial registered on the clinicaltrials.org website (Registration Number NCT06267222). AI-assisted tools were used for English-language editing and stylistic refinement of the manuscript. No AI tools were used for data analysis, statistical computation, or generation of results. The authors reviewed and approved all content and take full responsibility for the final version.

Individuals with SCA of any type who were on the registers of public hospitals, and private doctors and physiotherapists registers were invited to take part in the study. The eligibility criteria were investigated using an anamnesis form which also contained questions relating to sociodemographic data (gender, age, age at symptom onset, disease duration). The inclusion criteria were as follows: aged between 20 and 70 years old, regardless of gender or ethnicity; diagnosed with spinocerebellar ataxia of any type by a neurologist; with mild to moderate severity of ataxia according to the Klockgether scale; able to walk for 2 meters even with the use of a walker, cane or crutch; with a score ≥21[11] on the Mini-Mental State Examination (MMSE[12,13]); and with no other concomitant neurological alterations. Volunteers were excluded if they were illiterate; were undergoing any other physiotherapeutic or experimental drug intervention during the study; or had musculoskeletal, neurological, or cardiorespiratory disorders that prevented them from performing the tests. Those individuals eligible read and, if they agreed, signed the informed consent form. The participants were then assessed using the instruments described below.

The SARA scale was developed to assess the severity and progression of cerebellar signs in ataxias[14]. It was validated for Portuguese[15]. The SARA is an eight-item functional-based instrument that assesses gait, posture, sitting, speech disorders, limb coordination and postural disorders. The SARA score is the sum of eight item scores: gait (score 0-8), posture (0-6), sitting (0-4), speech (0-6), finger chasing (0-4), nose-to-finger test (0-4), rapid alternating hand movements (0-4) and heel-to-toe sliding (0-4). The maximum total SARA score is 40 points and indicates more severe ataxia[16].

The presence of non-ataxic signs (signs unrelated to ataxia) was assessed using (INAS). The inventory comprises 30 items, grouped into 16 extracerebellar (non-ataxic) signs. Therefore, the inventory score can range from 0 (absence of non-ataxic signs) to 16 (severe extracerebellar involvement)[17]. The signs include areflexia, hyperreflexia, plantar extensor response, spasticity, paresis, amyotrophy, fasciculation, myoclonus, stiffness (rigidity), chorea, dystonia, resting tremor, sensory symptoms, urinary dysfunction, cognitive dysfunction, and brainstem oculomotor signs[17].

The (BBS) assesses dynamic and static balance through 14 tasks. Each task is graded on a 5-point ordinal scale ranging from 0 to 4 for a maximum score of 56. A score of 0 is given when the individual is unable to perform the task, and a score of 4 is given when they can complete the given task independently. Other factors that affect the points given are the time taken to complete the task, the length of time a position can be maintained, and the amount of supervision or assistance required[18].

The (TUG) is a simple performance test commonly used to assess functional mobility. It assesses mobility, balance, walking ability and the risk of falling in older adults. It is a simple, easy-to-apply, quick test with excellent intra- and inter-examiner reliability, good convergent validity, and sensitivity to detect small changes[21]. In this test, the individuals are asked to get up from a sitting position, walk 3 meters, turn around 180°, walk the 3 meters again, and sit down again. The TUG can be used to predict the risk of falls. A time of 35 seconds or more predicts falls; completing the test in less than 15 seconds predicts a reduced risk of falls.

The Mini-Mental State Examination (MMSE) is a test that allows the assessment of cognitive function and screening for dementia. It has been widely studied and used in clinical settings to detect cognitive decline, to follow up dementia and to monitor response to treatment[12,13]. The MMSE was developed with the aim of differentiating between psychiatric patients with organic and functional conditions. In Brazil, the MMSE was translated into Portuguese[11]. At the time, it was noted that the total score depended on the educational level of the individual being assessed[11,13].

For study stages, see figure 1.

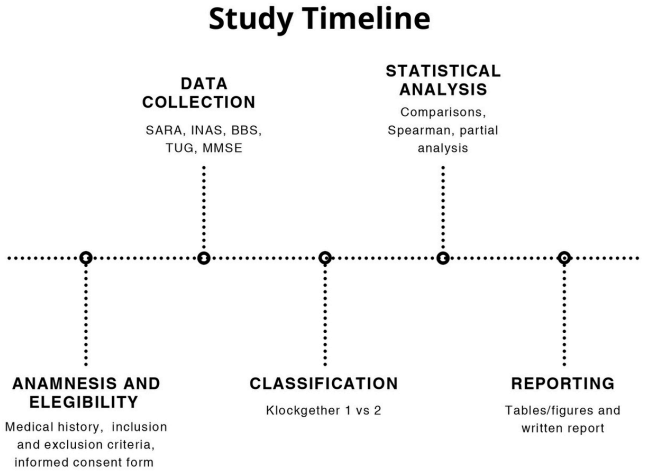

Figure 1. Study Timeline.

Statistical Analysis

Most of the data showed a non-normal distribution (Shapiro-Wilk test). For this reason, a non-parametric approach was used. The Mann-Whitney test was used to compare groups.

The association between the MMSE, INAS, BBS and TUG variables and the SARA score was estimated using Spearman's correlation coefficient (rho). In addition, a partial correlation analysis was carried out, controlling for the Klockgether stages[23], to consider the progression of the disease (stage 0 = no walking difficulties; stage 1 = onset of the disease, as defined by the onset of walking difficulties; stage 2 = loss of independent walking; stage 3 = confinement to a wheelchair).

RESULTS

Forty-two individuals with SCA were assessed and classified according to Klockgether et al. (1998) into stage 1 (n=21) and stage 2 (n=21). Most participants had SCA3 (stage 1: n=18; stage 2: n=21), while individuals with SCA2 (n=1), SCA5 (n=1), and SCA7 (n=1) were all classified as stage 1. Sample characteristics are present in table 1.

Table 1. Sample Characteristics

	Result
Sex, n (%)	
Female	27 (64.3%)
Male	15 (35.7%)
Age (years)	46.0 [41.0 – 53.3]
BMI (kg/m²)	25.7 [23.4 – 27.6]
Symptom duration (years)	7.0 [5.0 – 11.0]

Age, BMI and Symptom duration as Median [IQR].

Group comparisons are presented in Table 2. Mann–Whitney tests showed no between-group differences for demographic variables (age, body mass, height, and BMI) or symptom onset time (all p≥0.170), and no differences were observed for MMSE scores (p=0.780). In contrast, stage 2 participants showed greater disease severity and worse functional performance, with higher SARA scores and longer TUG times, and lower BBS scores (all p<0.001). INAS values were higher in stage 2, but this difference did not reach statistical significance (p=0.056).

Table 2. Comparison of clinical-functional outcomes between Klockgether stages

Variable	Stage 1 median [IQR]	Stage 2 median [IQR]	Mann–Whitney U	p
SARA	10.0 [7.0–11.5]	15.5 [12.0–19.5]	63.0	<0.001
INAS	2.0 [1.0–3.0]	4.0 [1.0–5.0]	145.0	0.056
MMSE	26.0 [25.0–27.0]	26.0 [25.0–29.0]	209.0	0.780
BBS	48.0 [46.0–54.0]	39.0 [28.0–45.0]	387.0	<0.001
TUG (s)	12.7 [9.5–15.8]	24.0 [15.9–63.0]	70.0	<0.001

Note. Values are median [interquartile range]. Between-group comparisons were performed using the two-sided Mann–Whitney U test. Values are median [IQR]. Between-group comparisons were performed using the two-sided Mann–Whitney U test. Abbreviations: SARA, Scale for the Assessment and Rating of Ataxia; INAS, Inventory of Non-Ataxia Signs; BBS, Berg Balance Scale; TUG, Timed Up and Go; MMSE, Mini-Mental State Examination.

Associations between ataxia severity (SARA) and clinical-functional outcomes are shown in Table 3 and Figure 2. SARA was significantly associated with INAS (small positive correlation), BBS (strong negative correlation), and TUG (strong positive correlation), while no significant association was observed with MMSE (trivial correlation).

Table 3. Association of SARA scores with other clinical-functional outcomes

Outcome	Spearman ρ (95% CI)**	p	Partial ρ* (95% CI)**	p*
MMSE	0.167 (−0.138, 0.442)	0.289	0.178 (−0.107, 0.463)	0.261
INAS	0.385 (0.082, 0.650)	0.012	0.266 (−0.015, 0.546)	0.089
BBS	−0.852 (−0.925, −0.689)	<0.001	−0.752 (−0.858, −0.584)	<0.001
TUG	0.773 (0.539, 0.896)	<0.001	0.642 (0.353, 0.820)	<0.001

* Partial correlation adjusted for Klockgether stage (1 vs 2).
** 95% CI estimated by bootstrap (1,000 iterations).
Abbreviations: SARA, Scale for the Assessment and Rating of Ataxia; INAS, Inventory of Non-Ataxia Signs; BBS, Berg Balance Scale; TUG, Timed Up and Go; MMSE, Mini-Mental State Examination.

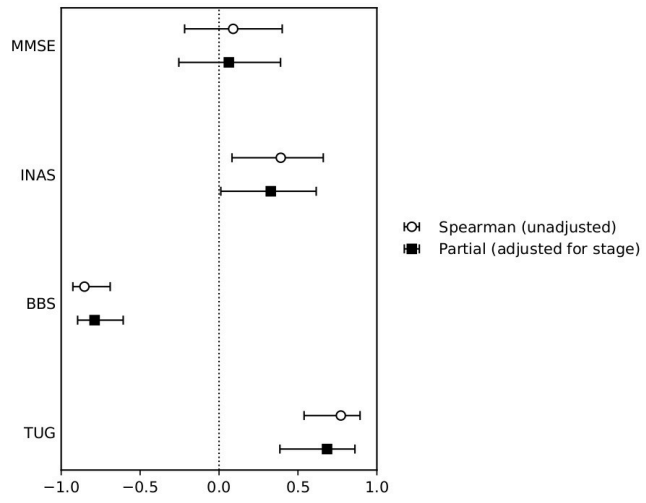

Figure 2. Spearman and partial correlations (adjusted for Klockgether stage) between SARA and clinical-functional outcomes.

Scatter plots illustrating these relationships are shown in Figure 3 (A–C), stratified by Klockgether stage. Partial correlation analyses adjusted for Klockgether stage (Table 2, Figures 1 and 2) showed an overall attenuation of correlation coefficients, indicating that disease stage partly accounts for the observed associations. Even after adjustment, significant associations remained between SARA and both BBS (strong negative correlation) and TUG (moderate-to-strong positive correlation), whereas the association with INAS was no longer statistically significant.

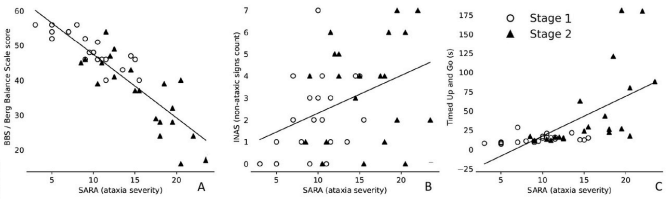

Figure 3. Scatter plots showing the associations between ataxia severity (SARA) and clinical-functional outcomes, stratified by Klockgether stage (1 vs 2). (A) Balance performance (Berg Balance Scale, BBS). (B) Extracerebellar burden (Inventory of Non-Ataxia Signs, INAS). (C) Functional mobility (Timed Up and Go, TUG). Each point represents one participant; solid lines indicate fitted linear trends.

DISCUSSION

This study shows that ataxia disease severity, quantified by SARA, is a central marker of functional decline in SCA, with strong and stage-independent associations with balance (BBS) and mobility (TUG). In contrast, no association was observed between ataxia severity and global cognitive screening (MMSE), suggesting that routine screening may not capture the cognitive domains most frequently affected in SCA.

Cognition and disease severity

Regarding disease severity, MMSE scores were not associated with SARA in our sample. The absence of a significant correlation between MMSE scores and SARA severity should not be interpreted as absence of cognitive impairment. Rather, it likely reflects limitations of global screening instruments, which may be insufficiently sensitive to executive, visuospatial, and fluency domains commonly reported in SCA. Previous studies have described cognitive deficits in SCA populations, particularly in SCA3. A study[24] reported impairments in visual and verbal memory, visuospatial/constructive abilities, and verbal fluency in individuals with SCA3. Another study[25] further emphasizes that cognitive characterization may help guide therapeutic strategies and improve quality of life. Mechanistically, altered cerebello-cerebral functional connectivity has been suggested as a potential contributor to cognitive symptoms in SCA. Taken together, these findings support the use of more comprehensive,

domain-specific neuropsychological batteries in future studies and clinical practice, particularly targeting executive and visuospatial functions.

Disease severity (SARA) and functional decline (BBS/TUG)

Individuals in the more advanced functional stage (stage 2) had higher SARA scores and clearly worse performance on balance (lower BBS) and mobility (longer TUG times), indicating a marked functional impact of disease progression. Importantly, the associations between SARA and both BBS and TUG were strong in unadjusted analyses, and they remained significant after controlling for Klockgether stage. This suggests that SARA captures meaningful functional impairment beyond stage-based classification alone and supports its clinical use as a severity anchor when interpreting performance-based functional measures. These results align with prior work showing progressive loss of walking ability and balance with increasing ataxia severity[27,1,4,28]. The relationship between increasing SARA and poorer mobility performance is also consistent with evidence that gait and functional mobility deteriorate as ataxia progresses[29,16].

Impact of non-ataxic signs (INAS)

In unadjusted analyses, higher SARA scores were associated with higher INAS scores, suggesting that extracerebellar burden tends to increase with greater ataxia severity. However, this relationship weakened and was no longer statistically significant after controlling for functional stage. In addition, although INAS values were higher in stage 2, the between-stage difference did not reach statistical significance. Together, these findings suggest that non-ataxic signs may co-occur with increasing severity but that their association with SARA is partly explained by functional staging. Clinically, extracerebellar features such as areflexia, urinary dysfunction, and spasticity may still contribute to the overall burden and complexity of management, even when their independent statistical contribution to SARA is attenuated[7]. Overall, our findings suggest that disease severity remains the primary clinical anchor for interpreting functional impairment, while extracerebellar signs may add complexity without fully explaining severity–function relationships.

Interpretation of the partial correlation analysis

Adjustment for Klockgether stage led to an overall attenuation of correlation coefficients, indicating that functional stage accounts for part of the associations observed between SARA and other outcomes. Nevertheless, the persistence of significant associations for BBS and TUG after adjustment supports the robustness of the severity–function link, reinforcing the use of balance and mobility tests as clinically meaningful “functional thermometers” to complement SARA during evaluation and follow-up.

Limitations and implications for rehabilitation

This study has limitations. The sample size was modest and predominantly composed of SCA3, which may limit generalizability to other genotypes. The use of the MMSE as the sole cognitive measure likely reduced sensitivity to executive and visuospatial deficits. Additionally, the cross-sectional design precludes causal inference and limits conclusions about trajectories over time. Despite these limitations, the findings have practical implications for rehabilitation: incorporating SARA alongside BBS and TUG may improve functional profiling, help stratify fall risk, and guide intervention planning (dose, progression, and safety). A multidimensional assessment that also documents extracerebellar signs and includes more sensitive cognitive evaluation may further support individualized rehabilitation strategies.

On the other hand, the study has several strengths, most notably its comprehensive multidimensional clinical assessment using a battery of validated instruments (SARA, INAS, BBS, TUG, and MMSE), which provides a holistic understanding of the motor and cognitive impairments in SCA patients. A significant contribution of this research is its focus on non-ataxic signs (via the INAS scale), highlighting their additional clinical characterization in the disease's functional impact. Furthermore, the methodological rigor is evidenced by the stratification of participants into Klockgether functional stages and the application of partial correlations to control for disease progression, ensuring more robust findings. These results offer direct clinical relevance, providing evidence-based insights that can guide targeted rehabilitation strategies and improve balance and mobility management in this population.

CONCLUSION

Greater ataxia severity (SARA) was strongly associated with poorer functional performance, particularly balance (BBS) and mobility (TUG). These findings support the clinical use of BBS and TUG as practical functional indicators to stratify fall risk and guide rehabilitation planning, including exercise dosing and safety considerations. Importantly, the associations between SARA and both BBS and TUG remained significant after adjustment for functional stage (Klockgether), suggesting that severity captures meaningful functional impairment beyond stage-based classification. Although no association was observed with global cognition on the MMSE, this should not be interpreted as absence of cognitive impairment; brief global screening may be insufficiently sensitive to the executive and visuospatial domains often affected in SCA. Future studies should include larger,

subtype-informed samples and longitudinal designs to clarify trajectories and to better characterize cognitive–motor relationships using domain-specific neuropsychological batteries.

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