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Relationship Between Severity of Ataxia, Quality of Life and Motor Recovery in Indian Population with Spinocerebellar Ataxia

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Abstract:

Background and Purpose: Spinocerebellar Ataxia (SCA) encompasses a group of neurodegenerative disorders characterized by progressive cerebellar ataxia. This study aims to explore the interrelationship between functional status, quality of life (QoL), and the severity of ataxia in Indian rural population with SCA patients.

Methodology: The study was observational in nature with correlational effect**.**Six patients (aged 25–70) diagnosed with SCA, confirmed by molecular or genetic testing, participated. Inclusion criteria required a minimum SARA gait score of 2 and FIM transfer score of 4, indicating the ability to walk independently without assistive devices. Exclusion criteria included concomitant brain lesions, terminal chronic diseases, unstable medical conditions, pregnancy, significant cognitive impairments, recent major orthopedic surgery, and other movement disorders. Patients were assessed for QoL, severity of ataxia and functional status using SF-36 v2, SARA, and m-FIM scales respectively. The data were collected and computed in a systematic way and then analysed by using SPSSv-26.

Results: Correlational analysis was done. The study found significant correlations between the severity of ataxia and both functional status and QoL. Higher SARA scores were associated with decreased functional independence and lower QoL scores.

Conclusion: The findings underscore the critical impact of ataxia severity on functional independence and QoL in SCA patients. Addressing ataxia severity is essential for enhancing patient outcomes. Future research should focus on interventions aimed at reducing ataxia severity to improve functional status and QoL.

Keywords: Ataxia, SARA, Quality of Life, SF-36v2, m-FIM

Introduction:

Ataxia, derived from the Greek word meaning 'not ordered,' encompasses a range of abnormal movements during voluntary actions, including incoordination, delayed movements, dysmetria (inaccuracy in targeting), dysdiadochokinesia (difficulty with rapid alternating movements), and tremor. It can manifest as a primary symptom or as part of a broader clinical context.[1] The onset of ataxia can be insidious and progressive, particularly in hereditary neurodegenerative conditions like spinocerebellar ataxias (SCAs), or it can occur acutely due to cerebellar infarctions, hemorrhages, or infections, leading to rapid and severe consequences. Cerebellar ataxia (CA), the motor dysfunction caused by cerebellar pathology, can arise from various etiologies, including neurodegenerative diseases, cerebrovascular events, multiple sclerosis, tumors, trauma, and toxins like alcohol.[2] SCAs, a prominent subset of hereditary neurodegenerative disorders, are defined by CAG trinucleotide repeat expansions in specific genes, leading to the synthesis of proteins with expanded polyglutamine (PolyQ) tracts, causing progressive cerebellar degeneration.[3]

Clinically, ataxia manifests with symptoms that depend on the type and location of cerebellar lesions. Lateralized lesions cause ipsilateral signs, while diffuse lesions result in more generalized impairments. The hallmark features of cerebellar dysfunction include gait ataxia, dyscoordination, nystagmus, dysarthria, tremors, and various motor control deficits. In SCAs, common symptoms include gait ataxia, incoordination, visual disturbances, and speech impairments, often leading to significant disability.[4,5] Current, global epidemiological studies on ataxia have estimated an overall ataxia occurrence rate of 26/100,000 in children, and for dominant hereditary cerebellar ataxia recessive hereditary cerebellar ataxia as 3.3/100,000.[6-9] Though hereditary forms, like Friedreich's ataxia and SCAs, are relatively rare, they contribute significantly to the ataxia burden. The prevalence of specific SCA subtypes, such as SCA1, SCA2, SCA3, and SCA6, varies by region, with higher incidences in certain populations, notably in Europe and Asia. Despite limited data on prevalence, the functional impact of ataxia is profound, often leading to a gradual decline in mobility, coordination, and independence.[10]

Although there is no cure for most hereditary ataxias, the focus of treatment remains on symptom management and improving quality of life. Pharmacological options like aminopyridines, acetazolamide, and coenzyme Q10 can manage specific forms of ataxia, while deep brain stimulation (DBS) and thalamic lesions may help reduce tremors.[11,12] Physiotherapy plays a critical role in enhancing balance, coordination, and mobility, fostering functional independence. A multidisciplinary approach combining medication, surgery, and rehabilitation is essential for optimizing patient outcomes.

Spinocerebellar ataxia, though progressive and degenerative, remains an underexplored topic, especially in terms of holistic factors and its severity on quality of life and motor recovery. Research on cerebellar ataxia, particularly within the Indian population, is limited. Given the profound impact of these conditions, there is an urgent need for studies that evaluate the severity of SCAs and their broader effects on the quality of life and motor recovery. This gap in research highlights the importance of further exploration into the impact of spinocerebellar ataxia, emphasizing the need for comprehensive studies to address the condition's full scope and improve patient outcomes.

Methodology:

This study adopted an observational design with a correlational focus to investigate the relationship between functional status, quality of life (QoL), and severity of ataxia in patients diagnosed with Spinocerebellar Ataxia (SCA). The research was conducted from March 2022 to June 2022 at the Indoor

and Outdoor Patient Departments of the Neurology Department at Guru Gobind Singh Medical College and Hospital, Faridkot, and the Outdoor Patient Department of Physiotherapy at the University College of Physiotherapy in Faridkot, Punjab. Ethical approval was granted by the Institutional Ethical Committee at Baba Farid University of Health Sciences, Faridkot.

A purposive sampling method was employed to select six patients from a pool of eight potential candidates, based on predetermined inclusion and exclusion criteria. Participants, aged 25 to 70 years, both male and female, had been diagnosed with cerebellar ataxia for a duration ranging from 2 months to 7 years. Inclusion criteria mandated a molecular or definitive genetic diagnosis of SCA or a strong family history of inherited cerebellar ataxia. Additionally, individuals with acquired or degenerative cerebellar ataxia who had undergone medical treatment were considered eligible. For mobility, patients were required to have a minimum SARA gait score of 2 (indicating abnormal gait and ability to walk tandem for at least 10 steps) and a minimum FIM transfer score of 4 (indicating minimal assistance for transfers between bed, chair, or wheelchair). Patients were required to stand and walk independently without the need for orthotic or assistive devices and were willing to provide informed consent.

Exclusion criteria encompassed individuals with concomitant brain lesions, terminal chronic diseases, or unstable medical conditions such as uncontrolled hypertension or unstable angina. Pregnant women, individuals with other medical conditions significantly affecting mobility, patients who had undergone major orthopedic surgery in the past six months, and those with significant cognitive impairments limiting informed consent were excluded. Patients receiving more than three hours per week of lower-body physical therapy or currently enrolled in another clinical trial were also excluded. Additional exclusion factors included the need for immediate intensive intervention for safety, receipt of botulinum toxin injections for spasticity management, and the presence of other movement disorders such as Parkinson's disease, Huntington's disease, or stroke.

After fulfilling the inclusion criteria and providing informed consent, patients were assessed using the Short form health survey (SF-36 v2), Scale for the Assessment and Rating of Ataxia scale (SARA) and m-functional independence measure scale (m-FIM) to evaluate their quality of life, cerebellar ataxia severity, and functional status, respectively. The SF-36 v2 was self-administered by the patients, with the researcher available to clarify any questions or uncertainties during the process. Both the SARA and m-FIM were administered by the researcher, as these scales require clinical evaluation. The total duration for the administration of all assessments was approximately 45 minutes. Scoring was performed immediately following the assessments, and the data were organized, computed, and analyzed using SPSS v-26.

The SF-36 v2 is a self-reported instrument that evaluates health-related quality of life across eight dimensions and two summary scales (physical and mental health), with scores ranging from 0 (worst health state) to 100 (best health state).[13] It has demonstrated excellent test-retest reliability (ICC = 0.80–0.95).[14] The SARA is a clinician-administered tool for assessing ataxia severity in eight categories, with a total score ranging from 0 (no ataxia) to 40 (severe ataxia), exhibiting high interrater (ICC = 0.98) and test-retest reliability (ICC = 0.90).[15] The FIM assesses functional independence through 18 items in two subscales—motor and cognition—scored on a 7-point ordinal scale, with total scores ranging from 18 to 126. The FIM has demonstrated strong reliability, with interrater reliability of 0.95 and test-retest reliability between 0.92 and 0.95.[16]

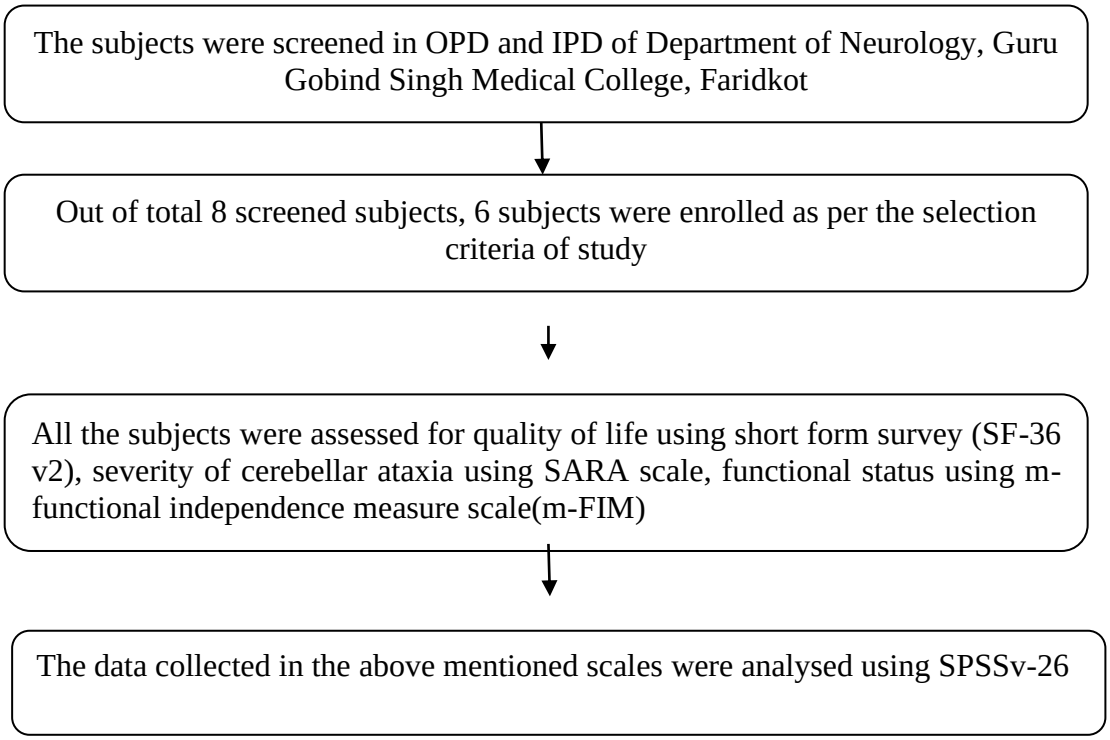

Figure 1 Flowchart of the Procedure

Figure 2 Patient performing heel shin slide using SARA scale

Results:

The study on spinocerebellar ataxia revealed the following demographic and clinical characteristics of the subjects. The mean age of the participants was 54.50 years, with a standard deviation of 11.72 years. The mean weight of the subjects was 67.33 kg, and the average height was 162.56 cm. The mean BMI was 25.45, and the mean duration of cerebellar ataxia was 38.33 months, with a standard deviation of 20.5 months, ranging from 2 months to 7 years. These findings highlight the variability in the clinical presentation of spinocerebellar ataxia among the study participants. Details are depicted in Table 1.

Table 2 presents the correlation analysis between the SARA scores and SF-36v2TM domain scores, as well as m-FIM scores. A significant negative correlation was found between physical functioning ($r = -0.85999$, $p < 0.05$) and general health ($r = -0.89039$, $p < 0.05$) with SARA scores, indicating that poorer physical functioning and general health are associated with higher severity of ataxia. Additionally, a significant negative correlation was observed between m-FIM ($r = -0.8894$, $p < 0.05$) and SARA scores, suggesting that lower functional independence is linked to greater ataxia severity. In contrast, energy (r

= -0.71684), emotional well-being (r = -0.55815), social functioning (r = -0.74933), pain (r = 0.403315), and health change (r = 0.592886) did not show statistically significant correlations (p > 0.05).

Correlation analysis between m-FIM scores and SF-36v2TM domain scores, highlighting the relationship between functional independence and various health-related quality of life parameters depicted in Table 3. A strong statistically significant positive correlation was found between physical functioning (r = 0.958, p < 0.05). Additionally, general health showed a positive correlation (r = 0.734), but the p-value was >0.05, indicating that this relationship is not statistically significant.Other SF-36v2TM domains, including energy (r = 0.597)**,** emotional well-being (r = 0.302)**,** and social functioning (r = 0.554), also showed positive correlations with m-FIM, but none were statistically significant (p > 0.05). On the other hand, pain (r = -0.697) and health change (r = -0.646) demonstrated negative correlations with m-FIM, suggesting that higher pain levels and perceived health changes may be associated with lower functional independence; however, these relationships were also not statistically significant (p > 0.05).

Table 1 Description of the demographic data and duration of Cerebellar Ataxia of the subjects with Spinocerebellar Ataxia

S No.	**Parameters**	**Mean** ± SD	**Range**
1	Age(years)	54.50 ±11.72	45
2	Weight(kg)	67.33 ±15.54	60
3	Height(cm)	162.56 ±10.5	42
4	BMI	25.45 ±3.88	15.5
5	Duration of cerebellar ataxia (months)	38.33 ±20.5	82

Table 2 Description of Correlation analysis of SARA scores with SF-36v2 scores and m-FIM scores

Parameters (SF-36v2^{TM} Domains)	**r value**	**p value**
Physical functioning	-0.85999	<0.05 (S)
Energy	-0.71684	>0.05(NS)
Emotional well being	-0.55815	>0.05(NS)
Social functioning	-0.74933	>0.05(NS)
Pain	0.403315	>0.05(NS)
Health change	0.592886	>0.05(NS)
General health	-0.89039	<0.05(S)
m-FIM	-0.8894	<0.05(S)

(S= Significant, NS= Non-Significant)

Table 3 Description of Correlation analysis of m-FIM scores with SF-36v2 scores

Parameter (SF-36v2^{TM} Domains)	**r value**	**p value**
Physical functioning	0.958	<0.05 (S)
Energy	0.597	>0.05(NS)
Emotional well being	0.302	>0.05(NS)
Social functioning	0.554	>0.05(NS)

Pain	-0.697	>0.05(NS)
Health change	-0.646	>0.05(NS)
General health	0.734	<0.05(NS)

(S= Significant, NS= Non-Significant)

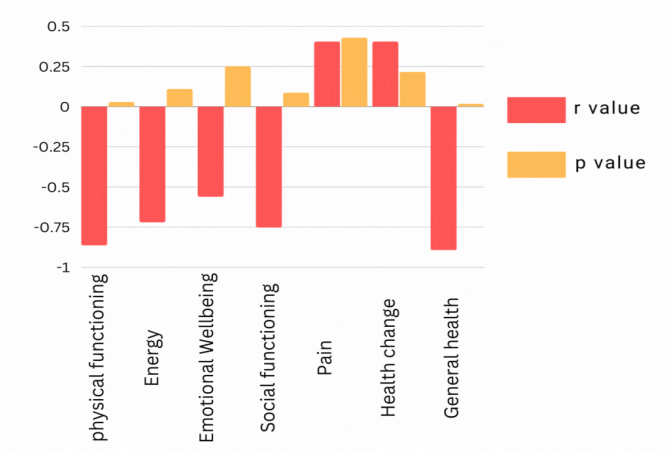

Figure 3: Graphical representation of Correlation between SARA scores with SF-36v2 domains scores

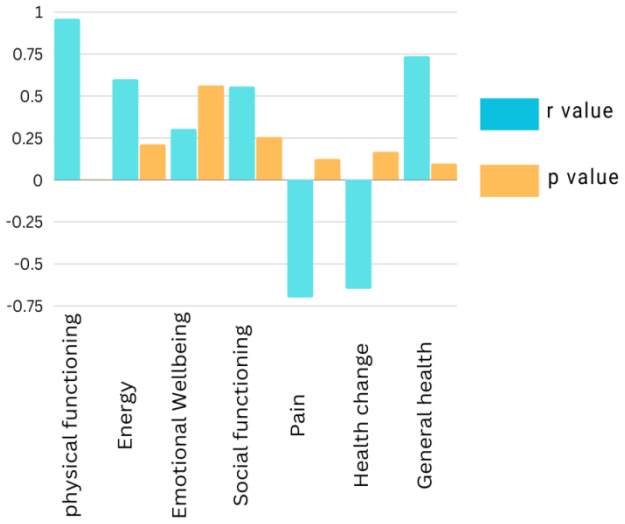

Figure 4: Graphical representation of Correlation between m-FIM scores with SF-36v2 domains scores

Discussion:

The aim of this study was to examine the relationship between the Scale for the Assessment and Rating of Ataxia (SARA), the SF-36v2 health survey, and the modified Functional Independence Measure (m-FIM) in patients diagnosed with Spinocerebellar Ataxia (SCA). The findings provide insight into the interrelationships between clinical measures of ataxia severity and quality of life outcomes.

A statistically significant negative correlation was observed between the SARA score and the Physical Functioning domain of the SF-36v2. This is consistent with the well-documented relationship between disease severity and physical performance in ataxia, where higher SARA scores (indicating greater severity) are associated with decreased physical functioning. The progressive nature of SCA, which

involves the degeneration of cerebellar structures responsible for coordination and motor control, leads to increased disability and diminished physical capacity. These findings align with Stanley's article, which reported similar negative correlations between physical functioning and disease severity in patients with neurodegenerative disorders. As SCA progresses, motor deficits result in a decline in physical independence, which directly impacts the physical health perception of individuals.[17] In a broader context, a study in Hong Kong found a significant negative correlation between ataxia severity (SARA) and the physical functioning and general health domains of SF-36v2.[18]

Additionally, a significant negative correlation was found between the SARA score and the General Health domain of the SF-36v2. This suggests that as the severity of ataxia increases, patients' perception of their overall health declines. This is likely due to the cumulative effect of motor dysfunction, which leads not only to physical limitations but also contributes to psychological stress and a diminished sense of well-being. The general health perception, being influenced by both functional and psychological factors, often correlates negatively with disease severity in chronic neurodegenerative conditions, as patients face increasing challenges in daily functioning and experience a decline in their overall quality of life.

Interestingly, the correlations between the SARA score and the Energy, Emotional Well-Being, Social Functioning, Pain, and Health Change domains of the SF-36v2 were non-significant. While it is expected that severe motor dysfunction would influence emotional well-being and social functioning, the lack of significant correlations may be attributed to a variety of factors. For instance, SCA patients may develop adaptive coping mechanisms that help preserve their emotional well-being despite motor impairment. Similarly, individuals with SCA often face social isolation due to physical limitations, but this may not be directly reflected in the quantitative scores of the SF-36v2, as social functioning can be influenced by a range of environmental and psychological factors beyond just physical disability.

The lack of a significant relationship between SARA and the Pain domain may be surprising at first glance. However, the absence of pain as a prominent symptom in SCA could explain this finding. Although ataxia impairs motor function and coordination, it may not necessarily result in chronic pain unless secondary complications such as musculoskeletal strain or injury occur. Thus, pain may not be a direct consequence of cerebellar degeneration in SCA, and other domains may be more closely tied to the physical manifestations of the disease.

A statistically significant negative correlation was found between the m-FIM score and the SARA score, indicating that as the severity of ataxia increases, functional independence, as measured by the m-FIM, decreases. This is a logical result, as more severe ataxia leads to greater disability, necessitating more assistance with activities of daily living. The m-FIM, which assesses independence in a variety of functional tasks, directly reflects the loss of autonomy as motor function deteriorates in individuals with SCA.

Furthermore, a significant positive correlation was found between the m-FIM score and the Physical Functioning domain of the SF-36v2. As functional independence increases, so does physical functioning, which is consistent with the expectation that individuals who are more physically independent perceive their physical health more positively. This reinforces the notion that motor independence is a critical component of an individual's overall health status and quality of life.

Similar to the findings with SARA, the correlations between the m-FIM score and the Energy, Emotional Well-Being, Pain, Health Change, and General Health domains of the SF-36v2 were statistically non-significant. This lack of significant correlation may suggest that functional

independence, as assessed by the m-FIM, does not always directly translate into better perceptions of emotional well-being or energy levels in SCA patients. These findings could imply that factors such as emotional resilience, psychological support, or compensatory strategies may play a significant role in how individuals with SCA cope with their condition, independent of their physical functioning. Moreover, since pain and health changes are not always predominant in SCA, they may not directly correlate with functional independence.

This study on Spinocerebellar Ataxia (SCA) has several limitations.he small sample size of six participants restricts the generalizability of the findings conducted in a single geographical area, Faridkot, the results may not represent the broader SCA population. The cross-sectional design captures data at a single point in time, making it challenging to establish causal relationships between variables. Additionally, the absence of a control group limits the ability to compare the effects of interventions with standard care. The lack of longitudinal data restricts the assessment of long-term outcomes and the sustainability of the observed effects. Lastly, the study's reliance on self-reported measures may introduce bias, as participants might overestimate or underestimate their abilities and health status. These limitations suggest the need for larger, multi-center, longitudinal studies with control groups to validate and expand upon the current findings.

In conclusion, this study illustrates the complex relationships between the clinical measures of disease severity, functional independence, and quality of life in patients with Spinocerebellar Ataxia. The significant negative correlations between SARA and both Physical Functioning and General Health domains highlight the profound impact of disease progression on physical and overall health perception. The findings regarding the m-FIM reinforce the importance of preserving functional independence in improving the quality of life for individuals with SCA. However, the non-significant correlations in other domains suggest that other psychosocial and environmental factors may influence these outcomes, underscoring the need for a holistic, multidisciplinary approach to managing Spinocerebellar Ataxia.

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