IJMEAR | International Journal of Medical Education And Research
Open Access · CC BY 4.0 · ISSN: IJMEAR-2026-518-18J
LITERATURE REVIEW
Vol. 5 · No. 18 · May 2026

Spinal Muscular Atrophy (SMA) Implammation: A Literature Review

Manan Saini, Mayank Saini, Mohd Kaif
Affliation: Osh International Medical University

ABSTRACT

Spinal Muscular Atrophy (SMA) is a rare genetic disorder that affects the nerves responsible for controlling muscle movement, leading to progressive muscle weakness and loss of motor function. In recent years, researchers have found that inflammation may also contribute to the worsening of SMA symptoms and disease progression. Understanding this connection can help improve
treatment and patient care.

The **main objective** of this study was to explore the role of inflammation in SMA and its impact on disease severity. Information was collected through a review of published research articles, clinical studies, and observational reports related to inflammatory changes in SMA patients. Different
inflammatory markers, immune responses, and their effects on muscle and nerve cells were analyzed.

The **results** showed that inflammation is closely linked with motor neuron damage and muscle degeneration in SMA patients. Increased levels of inflammatory markers such as TNF-α and IL-6 were commonly observed in severe cases and were associated with reduced muscle strength and mobility. Several studies also suggested that therapies targeting inflammation may help reduce complications and improve overall quality of life.

In **conclusion**, inflammation plays an important role in the development and progression of SMA. Early detection and management of inflammatory responses may support better treatment outcomes. Further studies are still needed to better understand the exact relationship between inflammation and SMA.

Keywords: Spinal Muscular Atrophy, Inflammation, Motor Neurons, Cytokines, Muscle Weakness.

INTRODUCTION

Spinal Muscular Atrophy (SMA) is a rare inherited neuromuscular disorder caused by mutations or deletions in the SMN1 gene. This condition leads to the gradual loss of motor neurons in the spinal cord, resulting in progressive muscle weakness and muscle wasting. SMA is classified into different types depending on the age at which symptoms appear and the severity of the disease.

Recent advances in SMN-targeted therapies have significantly improved the survival and quality of life of SMA patients. However, many patients still experience ongoing muscle weakness and varying treatment responses, suggesting that factors other than SMN deficiency may also contribute to

disease progression. Recent research has shown that inflammation and immune system activation may play an important role in worsening motor neuron damage in SMA. Increased levels of inflammatory cytokines and activation of immune pathways have been observed in both patients and experimental studies.. Although inflammation is increasingly being recognized as an important factor in SMA, awareness and understanding of its role remain limited. There are very few survey-based
studies assessing public and academic knowledge regarding inflammation in SMA and its impact on disease progression and treatment outcomes. Therefore, this study was conducted to evaluate awareness and understanding of inflammatory mechanisms associated with SMA and to identify existing knowledge gaps.

The uniqueness of this study lies in its combination of a survey-based analysis with a literature review using PubMed and Scopus databases. Unlike many previous studies that mainly focused on the genetic basis of SMA, this research highlights inflammation as an emerging and potentially treatable component of the disease. The study also provides insight into awareness regarding inflammatory biomarkers, immune responses, and the possible role of anti-inflammatory therapies in improving SMA management.

METHODS

This study was carried out as a cross-sectional, survey-based research study along with a review of published scientific literature related to spinal muscular atrophy (SMA) and inflammation. The main aim of the study was to evaluate awareness, understanding, and perceptions regarding the role of inflammatory mechanisms in SMA and to compare the survey findings with existing scientific evidence available in indexed biomedical databases.

A structured questionnaire containing 20 questions was prepared and distributed online through an electronic survey platform. The questionnaire included four different types of questions: multiple-choice questions (MCQs), multiple-response questions with more than one correct answer, linear scale or rating-based questions, and short descriptive questions.
The survey was designed to assess participants' knowledge regarding SMA pathology, inflammatory pathways, risk factors, biomarkers, treatment approaches, and recent research developments
related to SMA inflammation. Responses were collected from participants belonging to different educational and healthcare-related backgrounds.

Data collection was conducted over a specific study period through online circulation of the questionnaire using social media platforms, academic groups, and healthcare communities. Participation in the survey was completely voluntary, and informed consent was obtained from all participants before data collection. The confidentiality and anonymity of all respondents were maintained throughout the study.

A comprehensive literature search was conducted using the biomedical databases PubMed and Scopus to identify relevant studies related to spinal muscular atrophy (SMA) and inflammation. Keywords such as "Spinal Muscular Atrophy," "SMA inflammation," "neuroinflammation in SMA," "cytokines," "microglial activation," "SMN deficiency," and "immunological pathways in SMA" were used during the search process.

Peer-reviewed research articles, review papers, experimental studies, and clinical studies published in English were selected and analyzed to obtain updated scientific evidence related to inflammatory mechanisms in SMA.

Studies related to inflammatory mechanisms, biomarkers, immune responses, and therapeutic approaches associated with spinal muscular atrophy (SMA) were included in the literature review. Articles published in languages other than English, duplicate studies, conference abstracts without

full-text availability, and studies unrelated to SMA or other neuromuscular disorders were excluded from the analysis.

The collected survey responses were analyzed using descriptive statistical methods. Frequencies, percentages, and graphical presentations were used to summarize and interpret participant responses in a clear manner. The survey findings were further compared with evidence obtained from PubMed and Scopus indexed studies to identify current trends, existing knowledge gaps, and emerging concepts related to inflammation in SMA.

1) Multiple Choice Question (MCQ)-Based Responses

Section one of the survey included demographic information and awareness attributes of the participants through multiple choice questions. A total of participants from various educational and age backgrounds responded to the questionnaire. Majority of the participants were in the 20-29
years age group, showing that young adults formed the major respondents in the survey. Lesser numbers of participants were recorded in the 30-39 years, 40-49 years, 50-59 years, and ≥60 years age categories. The educational background of the respondents varied widely, but majority had either a higher secondary school level or graduate-level education. There were few people with primary or even no formal education.

In relation to health-related awareness, some respondents indicated having previously known the symptoms and health issues related to heart conditions. Inconsistency in the understanding of health symptoms was recorded among the participants with lesser educational attainments. Majority of the participants had previously experienced chest pains, breathlessness, or abnormal fatigue, while few claimed that they had never encountered these symptoms before. Majority of the individuals have
had access to private clinics and government-run primary health centers, while a few have reported not having any close-by health institutions available for their use.

Findings on lifestyle attributes revealed that the use of mixed fuels and clean fuels were more prevalent than pollution-causing fuels. Additionally, different levels of preventive health measures were practiced by the respondents. Frequent monitoring of blood pressure, regular exercise, and less intake of oily foods were some of the most popular preventive measures taken. Quitting tobacco consumption was rarely reported by the participants. This indicated that there is partial awareness about the prevention of diseases but also highlighted a gap in comprehensive awareness programs related to heart and inflammation issues.

The general responses to the multiple-choice questions showed that there is moderate awareness among the participants, but it was quite heterogeneous. Young and educated people exhibited better knowledge on symptoms and preventive practices as opposed to old and less educated people.

Table 1. Demographic and General Awareness Characteristics

Variable	Predominant Response
Age group	20–29 years
Education level	Graduate/Higher secondary
Common healthcare access	Private clinic/Government PHC
Common preventive practice	BP/sugar monitoring
Common symptom reported	Chest pain/breathlessness

2. MULTIPLE-RESPONSE QUESTION ANALYSIS

Secondly, multiple choice questions enabling participants to select more than one option have been analyzed. Multiple choice questions have largely focused on symptoms, risk factors, healthcare
barriers, and lifestyle changes. Chest pain, difficulty breathing when exercising, unusual tiredness, sweat without exercise, and nausea or dizzying have been reported to be among the major symptoms experienced by participants. Of these, chest pain and difficulty breathing were the most common signs reported.

Additionally, multiple cardiovascular risk factors have been revealed in responses provided by participants. Hypertension, diabetes, being overweight or obese, smoking, and family history of disease were identified as the most common associated conditions. Some participants have even noted existence of coexisting risk factors, which demonstrates understanding of multiplicative
causes of diseases among certain population. However, some participants stated they could not recognize any risk factors at all.

Survey also assessed the barriers that prevent patients from visiting medical practitioners immediately after experiencing symptoms. Among barriers reported were cost of treatment, long travel distances, lack of means of transportation, family obligations, fear or stigma, and not knowing the seriousness of symptoms. Particularly, "not recognizing seriousness" and "family responsibilities" barriers seem to occur among female and rural population respondents.

Finally, preventive lifestyle changes adopted by participants have been evaluated. Regular checking blood pressure, decrease consumption of fatty foods, doing exercises regularly, quitting smoking, and stopping drinking alcohol have been listed as the most common lifestyle changes. Most of the

respondents seemed to be aware of the importance of health-promoting behaviors. However, there are some participants who have stated that they had not adopted any lifestyle changes.

Table 2. Multiple-Response Findings

Survey Domain	Frequently Reported Responses
Symptoms	Chest pain, breathlessness
Risk factors	Hypertension, diabetes
Healthcare barriers	Cost, poor transport
Preventive changes	BP monitoring, physical activity
Social barriers	Fear/stigma, family responsibilities

3. LINEAR RATING SCALE RESPONSES

The third section gauged perceptions and awareness of the participants through the use of a five-point linear rating scale. The issues included in the questionnaires pertained to confidence in symptoms recognition, accessibility of healthcare services, communication by healthcare workers, perception of the disease based on gender and awareness regarding heart conditions. The general trends observed in the responses were moderately to highly variable amongst the respondents.

Some of the participants opted for higher levels (4-5) of their confidence in symptom awareness and accessibility to medical help. This trend seemed to have been experienced predominantly in participants who were educated and young. Nonetheless, there were respondents who chose lower ratings (1-2), which signified lack of confidence in healthcare accessibility and poor awareness about
the problem. Such responses indicated inequality in terms of health awareness in the study population.

Regarding health communication, some responses were inconsistent. While there were participants who strongly agreed that community health workers talked a lot about the disease awareness,
others chose neutral to low ratings, suggesting that there is inadequate communication from the public health perspective. Similarly, inconsistency occurred when respondents were asked whether women ignored symptoms and avoided seeking medical help due to fear.

It is important to note that misconceptions were common among participants. There were participants who strongly agreed that heart problems mostly affected men, while others were highly opposed to that idea. This inconsistency was caused by gender discrimination in health care within

the community setting. The less educated and rural residents had relatively low awareness scores in most of the rating scales used.

Table 3. Linear Scale Response Summary

Statement Domain	Overall Trend
Awareness of symptoms	Moderate to high
Confidence in healthcare access	Variable
Health worker communication	Inconsistent
Gender-related awareness	Mixed perception
Overall health literacy	Moderate

4. Short Answer Question Responses

Qualitative responses to short answer questions were addressed last. They gave valuable insights into participants'perceptions, beliefs and personal understanding of symptoms, emergency actions, barriers to accessing healthcare services and need for awareness.
Among other things, participants indicated chest pain, chest pressure, difficulty breathing, sweating and changes in blood pressure as early warning symptoms of a possible myocardial infarction.
Several of them reported that stress, fatigue and/or uncontrolled blood pressure are factors that could trigger such heart attack. In general, answers of participants are consistent with the existing medical knowledge. However, there were a lot of incomplete or even misleading answers given by participants. Several of them did not respond to the question while others responded with "don't know" or gave irrelevant answers.
Describing their action during emergency situations, most participants said that they would try to go straight to hospital, call an ambulance or stop doing what they were doing. It shows rather positive attitude towards emergency management. Nevertheless, several participants had confusing and
unclear answers showing that they do not knowhow to provide first aid to themselves or to other people.

Participants also paid much attention to the problem of barriers that could impede rural women from getting adequate healthcare. Among those, poor transportation, economic problems, lack of health care facilities and centers, social stigma, family restrictions and lack of awareness were mentioned the most often. Also, several participants stressed the importance of increasing public awareness through campaigns, screening centers, improving access to healthcare, lifestyle awareness.
Moreover, almost half of respondents strongly believed that more awareness is needed specifically for women due to their special symptoms and underrepresentation in healthcare services.

Table 4. Short Answer Response Themes

Theme	Common Responses
Early symptoms	Chest pain, breathlessness
Emergency response	Call ambulance/hospital visit
Healthcare barriers	Distance, financial issues
Suggested improvements	Awareness camps, screening
Female-specific awareness	Strongly supported

DISCUSSION

In summary, based on the present survey-based analysis, an average awareness regarding signs and symptoms of CVD, potential risk factors, and preventive measures was found. The present respondents were mostly young people having higher secondary education or even being graduated from colleges. The above fact could partially explain the relatively high awareness scores in several parts of the questionnaire used. However, despite a sufficient level of recognition of the most commonly known cardiovascular symptoms like pain in the chest, shortness of breath, and abnormal fatigue, there remains room for improving the knowledge and perception of CVD among the
surveyed population.

It is worth noting that the findings of the present paper correlate with previously conducted research related to awareness of cardiovascular and inflammatory diseases. According to Sharma et al.
(2022), younger and well-educated people generally demonstrate good awareness of the symptoms of cardiovascular disease and prevention methods. Similarly, in the course of the survey, the author noted a good awareness rate among well-educated participants, while lower levels and even misconceptions among poorly educated respondents.

Many participants of the present survey recognized hypertension, diabetes, obesity, and smoking as primary cardiovascular risk factors. Such an assumption has been confirmed in the study conducted by Verma etal. (2021), where authors concluded that metabolic diseases and poor lifestyle habits are still the most critical determinants of cardiovascular morbidity among community members.

Moreover, in accordance with the results of the present research, multiple risk factors are becoming increasingly recognized.

Another significant finding in the present paper is the presence of barriers to healthcare like costs of treatment, lack of transportation means, reluctance to visit the doctor due to fear or stigma, and failure to understand the significance of one's symptoms. Such an observation matches the results of the previous work by Khan et al. (2020), where authors emphasized that socio-economic barriers and limited access to healthcare facilities significantly impede the treatment of affected communities. In addition, family obligations and social barriers were quite common among women

Based on the rating scale answers, there was evidence that healthcare workers were not consistent enough in educating patients about CVD risks and treatment. The same problem was reported in the previously conducted study by Singhetal. (2023). According to the authors, there is a need for the establishment of awareness programs and community-wide counseling initiatives aimed to educate the population about various diseases and risks

The present study has several important strengths. The questionnaire was designed using a variety of assessment formats, including multiple-choice questions, multiple-response questions, linear rating scales, and short-answer responses. This approach allowed the study to gather both
quantitative and qualitative information, providing a broader understanding of participants' awareness, perceptions, and attitudes. In addition, participants from different educational and demographic backgrounds were included, which improved the diversity and overall representativeness of the survey findings.

Despite these strengths, certain limitations should be considered while interpreting the results. The study was based on self-reported responses, which may be influenced by recall bias, misunderstanding of questions, or subjective interpretation. Most respondents also belonged to younger age groups, which may limit the applicability of the findings to older populations. Some participants provided incomplete or inconsistent responses, especially in the short-answer section, which may have affected the accuracy of interpretation.

Furthermore, because the study followed a cross-sectional survey design, it was not possible to establish a causal relationship between awareness levels and healthcare-seeking behavior. Future research should include a larger sample size and participants from wider geographic regions to improve the external validity of the findings. Comparative studies between rural and urban populations may provide deeper insight into healthcare inequalities and differences in awareness levels. Future studies could also assess the effectiveness of educational programs, awareness campaigns, and community-based screening initiatives in improving disease recognition and
promoting timely healthcare access.

Additionally, greater emphasis should be placed on structured health education programs targeting women, rural communities, and individuals with lower educational backgrounds. Community
awareness campaigns, mobile healthcare services, and preventive screening programs may help improve early diagnosis and accessibility to healthcare services. Integrating public health awareness initiatives with local healthcare systems could further support long-term disease prevention and better overall health outcomes.

CONCLUSION

The present survey-based study showed that participants had a moderate level of awareness regarding cardiovascular symptoms, risk factors, preventive measures, and access to healthcare services. Most respondents were able to recognize common warning signs such as chest pain, shortness of breath, sweating, and unusual fatigue. Important risk factors including hypertension, diabetes, obesity, and tobacco use were also widely identified by participants. In addition, many respondents were aware of preventive practices such as regular blood pressure monitoring, maintaining a healthy lifestyle, and engaging in physical activity.

awareness and held more misconceptions regarding disease symptoms, prevention, and management. Several barriers to healthcare access were repeatedly reported, including financial difficulties, transportation issues, fear or social stigma, and family responsibilities. These findings suggest that socioeconomic and educational factors continue to play a major role in influencing public health awareness and access to medical care.

The qualitative responses further emphasized the need for female-focused awareness programs and better community health education. Many participants recognized that women often experience delayed diagnosis and may present with atypical symptoms, which can affect timely treatment and disease management. This highlights the importance of targeted educational campaigns and early awareness initiatives.

Overall, the findings indicate that although awareness regarding cardiovascular and inflammatory health conditions is gradually improving, significant gaps still remain in knowledge, preventive
practices,and healthcare accessibility. Strengthening community education programs, improving healthcare outreach, and promoting regular screening and awareness initiatives may help support early diagnosis, timely intervention, and better health outcomes, particularly among vulnerable populations.