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REVIEW ARTICLE

Spinal Muscular Atrophy: Crisis of neglect in care of a rare disease in Nigeria

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Abstract

Background: Spinal muscular atrophy (SMA) is a rare autosomal recessive neuromuscular disorder and a leading genetic cause of infant mortality. Progressive degeneration of spinal alpha motor neurons causes muscle weakness, paralysis, and respiratory failure. In Nigeria, limited awareness, inadequate diagnostic infrastructure, and restricted access to treatment contribute to significant unmet care needs for individuals with SMA.

Objectives: This review examines the burden of SMA in Nigeria, focusing on genetic findings, diagnostic limitations, healthcare gaps, and barriers to optimal management.

Methods: We reviewed relevant literature from PubMed, Google Scholar, and regional health databases. We analyzed genetic findings, epidemiological reports, and health policy documents to identify knowledge gaps and opportunities to improve care.

Results: Nigerians have a carrier frequency of about 1 in 60, with more survival motor neuron 1 (SMN1) copies but fewer survival motor neuron 2 (SMN2) copies than non-African populations, potentially indicating more severe disease. Limited genetic testing and newborn screening delay or miss diagnoses. High costs and limited health-system support restrict access to disease-modifying treatments, including nusinersen and gene replacement therapy. Families also face stigma, psychosocial distress, and financial hardship amid inadequate care pathways.

Conclusions: SMA remains a neglected rare disease in Nigeria. The national rare disease agenda must urgently include SMA. Priorities include expanding diagnostic capacity, implementing newborn screening, establishing a national SMA registry, raising awareness, and fostering partnerships to improve access to therapy. Bridging these gaps is crucial to achieving equitable rare disease care and improving outcomes for affected individuals.

Keywords: Genetic testing, Health policy, Nigeria, Rare diseases, Spinal muscular atrophy, SMN1/SMN2 genes

Introduction

Spinal muscular atrophy (SMA) is a rare autosomal recessive neuromuscular disorder and one of the leading genetic causes of infant mortality. It is characterized by degeneration of alpha motor neurons in the anterior horn of the spinal cord, leading to progressive proximal muscle weakness, hypotonia, and paralysis. The estimated global incidence ranges from 1 in 6,000 to 1 in 10,000 live births, with a carrier frequency between 1 in 40 and 1 in 60 individuals. [1]

Clinically, SMA presents as generalized muscle weakness and atrophy, most prominently affecting the proximal limb muscles. The condition is classified into four main subtypes (SMA types I–IV), based on age of onset and the maximum motor milestones achieved. Type I is the most severe and typically fatal in infancy without intervention, while type IV represents the adult-onset form with milder symptoms. Molecular genetic testing, particularly for homozygous deletions or mutations in the survival motor neuron 1 (SMN1) gene, has replaced muscle biopsy and electromyography (EMG) as the diagnostic gold standard. [2]

Therapeutic breakthroughs over the past decade have transformed SMA prognosis, especially when initiated early. Currently approved disease-modifying therapies include the SMN2 splicing modifiers nusinersen (Spinraza) and risdiplam (Evrysdi), as well as SMN1 gene replacement therapy via onasemnogene abeparvovec-xioi (Zolgensma). These treatments have significantly improved motor function and survival, particularly in presymptomatic infants[2]. However, such advances remain largely inaccessible in many low- and middle-income countries (LMICs), including Nigeria, where delayed diagnosis, lack of molecular infrastructure, and high treatment costs contribute to preventable disability and death.

Nigeria, Africa's most populous country with over 200 million people and more than 250 ethnic groups, offers a unique setting to study the burden and characteristics of SMA in a genetically and culturally diverse population. [3] Despite this, data on SMA epidemiology, diagnosis, and management in Nigeria and similar resource-limited contexts remain scarce. This information gap impedes the development of targeted interventions and limits integration of rare disease care into national health policy.

This narrative review synthesizes existing literature on SMA, with a global-to-local focus on its epidemiology, clinical spectrum, genetic background, diagnostic modalities,

and therapeutic strategies. It focuses on Nigeria, highlighting the challenges and opportunities for improving care delivery. The review proposes actionable recommendations, including policy reforms, expanded access to affordable genetic testing, investment in health system capacity, and locally adapted strategies to reduce the burden of SMA and advance equity in rare disease care in Nigeria.

Methods

Given the limited availability of Nigeria-specific data on spinal muscular atrophy (SMA), we used a narrative synthesis approach to explore existing evidence and identify relevant themes. We conducted a comprehensive literature search across electronic databases, including PubMed, Google Scholar, and African Journals Online (AJOL). We used a search strategy combining keywords and Boolean operators. Core terms included *"Spinal Muscular Atrophy," "SMA," "Nigeria,"* and *"Nigerian."* To broaden the search, we also used broader terms such as "rare diseases in sub-Saharan Africa" and "neuromuscular diseases in low-resource settings." We conducted the search in February 2025, with no publication-date restrictions, to include both recent and foundational literature.

We selected articles relevant to SMA in the Nigerian context, particularly those addressing prevalence, genetic characteristics, diagnosis, treatment access, health system barriers, and policy challenges. We also included grey literature, such as government reports, NGO publications, and policy briefs, to provide contextual understanding of local practices and limitations. We excluded non-English articles, abstract-only submissions, and opinion pieces without empirical support.

Figure 1 summarizes the study identification, screening, and inclusion process. From the final selection, we extracted data and organized them into three thematic areas: Diagnosis and clinical presentation, Management and access to therapy, and Health system and policy implications. This thematic organization enabled a structured synthesis of the literature and helped identify key gaps and recommendations for improving SMA care in Nigeria.

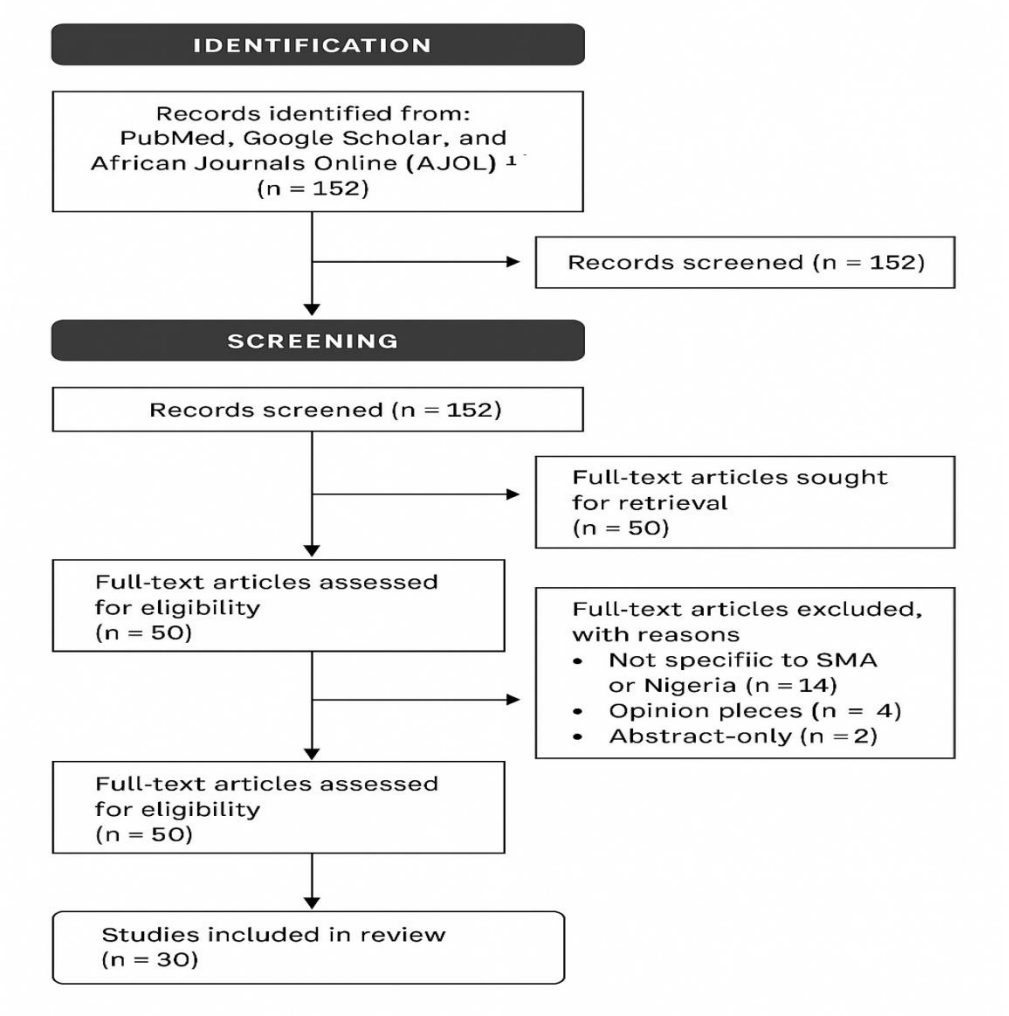

Figure 1. PRISMA-Based Flowchart of Study Selection Process. A visual summary of the article identification, screening, eligibility, and inclusion process used in the narrative review of spinal muscular atrophy (SMA) literature relevant to Nigeria.

Results and Discussion

Epidemiology and Genetic Architecture of Spinal Muscular Atrophy in Nigeria

Spinal muscular atrophy (SMA) is a severe autosomal recessive neuromuscular disorder with a global incidence estimated at approximately 1 in 10,000 to 11,000 live births. It is the second leading genetic cause of infant mortality, particularly in high-income countries such as the United States and those in Europe, where incidence rates may be higher. [4-6] Across these regions, including Australia, fewer than 9,000 individuals are estimated to live with SMA at any given time. [5]

Clinicians categorize SMA into five types (SMA 0–4) based on age of onset and severity. Type 1 is the most prevalent, accounting for about 55% of all diagnosed

cases, followed by type 2 (~30%) and type 3 (~15%). The most severe (type 0) and adult-onset (type 4) forms are rare, together comprising less than 1% of global cases. [5,7,8]

Epidemiological Evidence and Data Gaps in Nigeria

In Nigeria, epidemiological characterization of SMA remains limited, with the disorder often underdiagnosed due to lack of clinical awareness and genetic diagnostic capacity. The only available population-level estimate comes from a 2014 study that assessed SMN1 gene copy number in a cohort of 120 healthy Yoruba individuals. Two individuals had a single copy of SMN1, indicating a carrier frequency of 1 in 60 (1.67%); using Hardy-Weinberg equilibrium for autosomal recessive disorders, the authors inferred a national prevalence of approximately 1 in 14,400 live births. [9]

Despite its limitations, this study offers rare insight into the genetic epidemiology of SMA in sub-Saharan Africa, particularly in Nigeria. However, the absence of SMA-specific registries or surveillance systems, along with the scarcity of peer-reviewed Nigerian case reports, continues to hinder accurate national burden estimation.

Genetic Landscape: SMN1 and SMN2 Variability

SMA results from homozygous deletion or pathogenic variants in the SMN1 gene on chromosome 5q13, which encodes the survival motor neuron (SMN) protein, essential for the maintenance of alpha motor neurons. [7] The SMN2 gene, a near-identical paralog, acts as a disease modifier, with its copy number often correlating inversely with disease severity.

The 2014 Nigerian study found that 38% of individuals had three or four SMN1 gene copies, significantly higher than in Caucasian populations (6.3%) but lower than in Mali (53%). [9] This elevated SMN1 copy number may partially protect against disease expression by providing compensatory gene function. [6,8] However, the same study reported that 27% of Nigerians lacked any SMN2 copies, compared with 8–9% in European cohorts, suggesting a potential for more severe clinical phenotypes in individuals with SMA in Nigeria **(Table I).** [6,7,9] This unique genetic architecture, marked by both a high prevalence of functional SMN1 alleles and a comparatively low frequency of SMN2, underscores the need for context-specific diagnostic algorithms, early carrier screening, and targeted research to inform national health policy and improve outcomes for individuals living with SMA in Nigeria.

Diagnostic Challenges, Delays, and Missed Opportunities in Nigeria

The estimated lower prevalence of SMA in Nigeria (1 in 14,400 vs. global 1 in 10,000) likely reflects underdiagnosis rather than true biological rarity. Limited awareness among healthcare professionals and the public, along with inadequate diagnostic infrastructure, significantly contributes to diagnostic delays and treatment gaps in Nigeria. [10,11] Historically, many cases went unreported or undocumented because clinicians relied on clinical presentation rather than genetic confirmation. In the early 1900s, clinicians diagnosed SMA largely based on non-specific clinical features, making misclassification and underreporting common. [6] Today, while genetic testing is the global diagnostic gold standard, it remains scarce in Nigeria. Where testing is possible, high costs and long travel distances further complicate timely access. [11]

Table I: Comparative Genetic Profile of SMA-Related SMN1/SMN2 Variants in Nigerian and Non-African Populations

Population	Sample Size	% with 1 SMN1 Copy (Carriers)	% with ≥3 SMN1 Copies	% with 0 SMN2 Copies	Source
Nigerians (Yoruba)	120	1.67%	38%	27%	[9]
Caucasians	>1000 (est.)	~1.8%	6.3%	8–9%	[6,7]
Malians	N/A	N/A	53%	Not reported	[9]
Europeans (general)	Varies	~1.7–2.1%	<10%	8–9%	[6,7,9]

The diagnostic delay is notably influenced by disease severity. SMA type I, the most severe form, is often recognized and diagnosed earlier, while milder types such as SMA type III experience longer delays due to subtle symptoms requiring more extensive investigations. [12] This variability often leads patients to consult multiple healthcare providers before receiving an accurate diagnosis. [10,12] Poor health-seeking behavior, influenced by financial constraints, cultural beliefs, and limited access to quality healthcare, further worsens the diagnostic timeline. [13] Also, newborn screening for SMA, implemented in several high-income countries, has yet to be introduced in Nigeria. These delays further postpone diagnosis and therapeutic intervention, which are most effective when initiated early. [14]

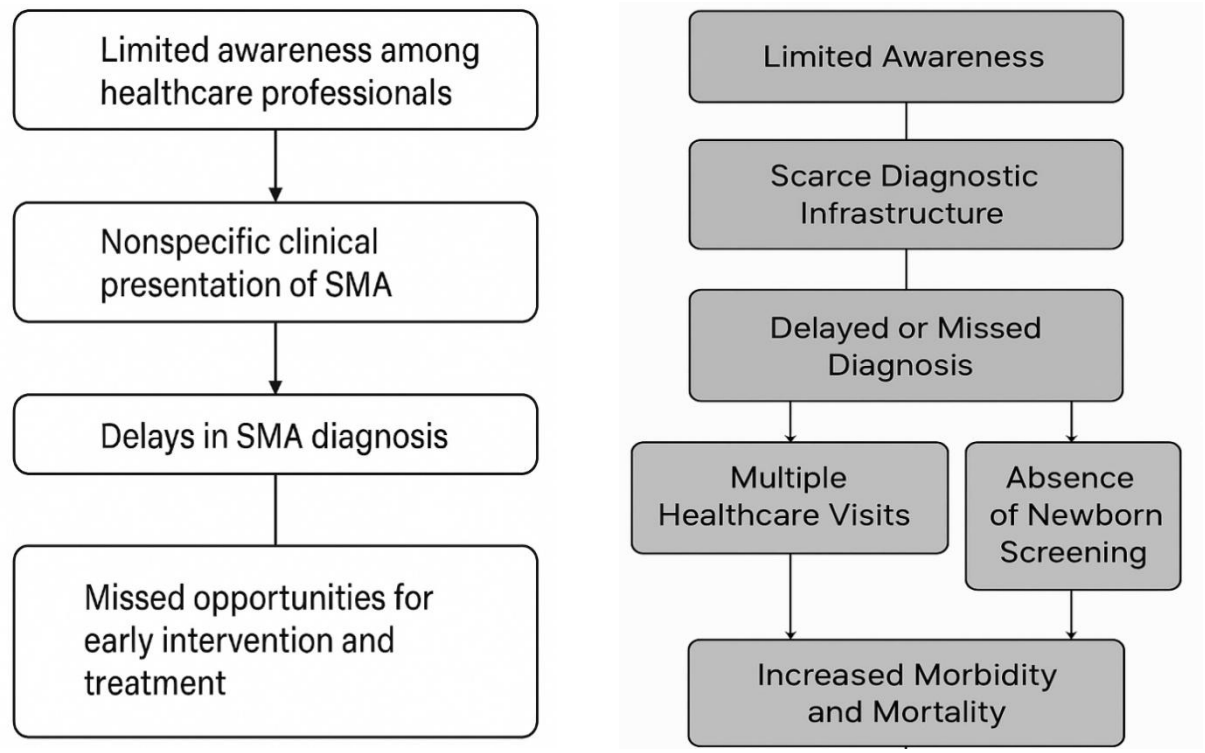

Figure 2. Integrated flowchart illustrating diagnostic challenges, delays, missed opportunities, and systemic barriers to early recognition and intervention for Spinal Muscular Atrophy (SMA) in Nigeria

As Figure 2 illustrates, these interconnected factors contribute to delayed diagnoses, missed therapeutic windows, and poor outcomes. Other constraints, cultural beliefs, and limited access to quality healthcare further delay diagnosis. [13] Ultimately, these challenges lead hospitals, policymakers, and global health organizations to grossly underestimate the true burden of SMA in Nigeria, delaying vital and potentially lifesaving interventions.

Therapeutic Disparities and Access Challenges in SMA Care in Nigeria

With the increasing recognition and diagnosis of spinal muscular atrophy (SMA) globally, significant therapeutic advances have emerged. These include disease-modifying treatments and supportive care strategies that can significantly improve survival and quality of life. However, despite these breakthroughs, stark disparities in access persist, particularly in low- and middle-income countries like Nigeria due to systemic and socioeconomic barriers. Early initiation of therapy is strongly correlated with improved outcomes in SMA, yet these access limitations lead to delayed treatment and suboptimal prognoses for many Nigerian patients.

One of the most significant barriers to treatment access in Nigeria is the

predominant reliance on out-of-pocket health expenditures and occasional donor funding. Given that SMA is a chronic and multisystemic disorder, its management requires ongoing pharmacological therapy, long-term monitoring, and supportive care. This imposes a substantial financial burden on affected families, often making timely, sustained treatment inaccessible for most. [15,16]

Furthermore, SMA care necessitates a multidisciplinary team approach involving geneticists, neurologists, pulmonologists, and physiotherapists. Unfortunately, Nigeria's healthcare system faces a critical shortage of these specialists and infrastructure. Because trained clinical geneticists and counselors are scarce, general pediatricians manage most genetic conditions, including SMA. [11] Even in tertiary institutions, care is often suboptimal, with inadequate diagnostic tools, limited rehabilitation services, and inconsistent follow-up, all of which negatively affect clinical outcomes. [17]

In addition, regulatory and pharmaceutical challenges further exacerbate inequities in access to therapy. Nigeria’s pharmaceutical regulatory landscape is characterized by weak enforcement, underfunding, and limited routine inspections. This leads to the widespread circulation of unregulated or counterfeit medications, often sold without a prescription by unqualified personnel. [18] Moreover, poor road networks, storage limitations, and fragmented drug distribution systems hinder the timely availability of approved SMA medications across the country. [19]

Altogether, these systemic healthcare deficiencies, spanning economic, infrastructural, and regulatory domains, underscore the urgent need for targeted policy reforms, subsidized treatment programs, and international partnerships. Bridging these gaps is critical to improving survival, functional outcomes, and equity in SMA care for Nigerian children.

Socioeconomic, cultural, and systemic impacts on SMA care in Nigeria

The socioeconomic burden of spinal muscular atrophy (SMA) is multifaceted and significantly affects affected families in Nigeria. Among the cost drivers, novel disease-modifying therapies pose the greatest financial challenge. Studies show that medication accounts for about 91.7% of total median SMA-related costs. [20] The average annual cost for SMA drugs varies by disease subtype and medication but can be prohibitively expensive. For example, in type 1 SMA, the most severe form, the financial

burden is often doubled because of the intensive care required. [21]

In Nigeria, where an estimated 40% of the population lives below the poverty line, this economic strain is further compounded by the predominance of out-of-pocket healthcare payments. [22] Consequently, many low-income families are unable to afford diagnosis and treatment, resulting in underdiagnosis, delayed care, and poor health outcomes. The indirect cost burden is also considerable. Studies indicate that lost productivity of the primary informal caregiver, often a parent, is the second most significant contributor to the overall socioeconomic cost of SMA. [22] These compounded financial constraints often force families to limit necessary out-of-pocket expenses, including psychological services and educational support for affected children. [22]

Nigeria lacks comprehensive health policies that adequately address rare diseases like SMA. The National Health Insurance Scheme (NHIS) does not cover rare genetic disorders, leaving treatment underfunded and reliant on international donors or philanthropic efforts. [23] Despite some advancement by agencies like the Nigeria Center for Disease Control (NCDC) in controlling infectious diseases, inherited disorders such as SMA remain largely ignored. Global assessments of rare disease policies show that Nigeria lags behind countries that have enacted orphan drug laws or dedicated funding structures. [24] The burden of care thus falls disproportionately on families, often resulting in poor treatment outcomes and further social inequities.

Cultural beliefs and societal perceptions further complicate SMA management in Nigeria. The condition's rarity and low public awareness foster stigma, misinformation, and social exclusion. A BBC article about a Nigerian mother living with SMA describes the challenges of societal misunderstanding, with disabilities sometimes perceived as divine punishment or an ancestral curse. [25] Such beliefs may discourage families from seeking medical intervention, prompting them instead to consult traditional healers or rely solely on spiritual remedies. [26] These delays reduce therapeutic windows and further burden the healthcare system.

Beyond financial and cultural challenges, inadequate psychosocial and educational support hampers SMA care in Nigeria. Numerous studies have documented the significant emotional and physical toll on caregivers. Despite medical advances, caregivers remain overwhelmed by day-to-day care demands. [27, 28] Many report high levels of stress, anxiety, depression, reduced

quality of life, and an inability to maintain employment [28]. Addressing these unmet needs requires a shift toward holistic, family-centered care that includes psychological counseling, caregiver support services, and inclusive educational planning. [29,30]

Recommendations

This article has explored the epidemiology, socio-economic burden, and current state of care for individuals living with spinal muscular atrophy (SMA) in Nigeria, while offering pragmatic strategies for improving diagnosis, treatment, and patient support. Based on these findings, the following key recommendations are proposed:

Spinal muscular atrophy (SMA) should be recognized as a national rare disease priority in Nigeria to promote policies and sustainable programs addressing limited awareness, delayed diagnosis, and restricted access to treatment. Population-specific epidemiological studies and a national SMA registry are needed to establish disease burden, characterize genetic variants, document clinical outcomes, and identify regional disparities. Develop socioeconomic interventions, including health insurance coverage, financial subsidies, and family support services, to mitigate the substantial direct and indirect costs of care.

Strengthen early diagnosis and comprehensive management by improving diagnostic infrastructure, evaluating the feasibility of newborn screening, and developing multidisciplinary care pathways. Establish national rare disease policies and SMA-specific clinical guidelines, with provisions for equitable access to genetic testing and approved disease-modifying therapies through appropriate health financing mechanisms. Public and professional awareness campaigns should engage healthcare institutions, civil society organizations, educational institutions, and policymakers to reduce stigma, encourage early presentation, and strengthen advocacy for affected individuals.

Capacity-building programs should equip clinicians, geneticists, nurses, and other relevant healthcare professionals with the knowledge and skills to recognize, diagnose, counsel, and manage SMA. In underserved communities, engage traditional birth attendants in basic awareness initiatives and appropriate referral pathways, with clear guidance on early recognition and the limits of their clinical role. Finally, national and international partnerships among research institutions, patient advocacy groups, healthcare organizations, and pharmaceutical stakeholders should be strengthened to advance research, improve access to treatment, support technology transfer, and

explore sustainable, locally appropriate approaches to SMA care.

Way Forward

The lack of comprehensive data and targeted health policies underscores the urgent need to intensify research and awareness of SMA in Nigeria. Bridging these knowledge and service gaps will not only improve care but also provide the basis for evidence-informed policymaking. Prioritizing funding for SMA-focused research is critical to unlocking innovative treatments, optimizing healthcare delivery, and establishing psychosocial support systems tailored to Nigerian realities. With deliberate investments, Nigeria can ensure SMA patients are not left behind and have the quality of life and opportunities they deserve.

Conclusion

Spinal muscular atrophy (SMA) remains grossly underdiagnosed and underreported in Nigeria, primarily due to the absence of a national registry, limited access to diagnostic infrastructure, and widespread ignorance among the public and professionals. The genetic landscape of SMA in Nigeria may differ substantially from that of European populations, with possible variations in mutation spectra and clinical phenotypes.

To bridge diagnostic and therapeutic gaps, a multi-pronged approach is essential. This includes establishing national SMA registries, raising awareness among health professionals and the public, expanding access to genetic testing and approved therapies, and aligning with global research efforts to elucidate the unique genetic and environmental determinants of SMA in Nigeria and sub-Saharan Africa. Such comprehensive action will ultimately improve diagnostic accuracy, treatment outcomes, and overall patient well-being.

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