

Unlocking the Potential of Machine Learning and Artificial Intelligence for Early Detection and Monitoring of Spinal Muscular Atrophy Type 3

A Narrative Review in Low- and Middle-Income Countries

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ABSTRACT

Spinal Muscular Atrophy Type 3 (SMA3), or Kugelberg-Welander disease, is a later-onset and relatively milder form of SMA, but delayed recognition can still cause progressive weakness, loss of ambulation, scoliosis, fatigue, and reduced quality of life. In low- and middle-income countries (LMICs), early diagnosis and monitoring are often limited by subtle symptoms, restricted genetic testing, specialist shortages, fragmented follow-up, and weak referral systems. This paper presents a conceptual framework for using artificial intelligence (AI), machine learning (ML), and Internet of Things (IoT) enabled tools to improve SMA3 detection, monitoring, and care delivery in LMICs. The framework integrates multimodal data collection, including demographic, clinical, motor function, diagnostic, treatment, patient-reported, wearable, smartphone-based, and health system data. These inputs are processed through cloud-based data preparation, including standardization, cleaning, privacy protection, missing data handling, multimodal integration, and feature extraction. AI/ML models may support disease progression prediction, risk stratification, early detection of motor decline, treatment response prediction, phenotype clustering, and longitudinal monitoring. The resulting outputs include clinical decision support, personalized rehabilitation, early warning alerts, patient dashboards, national registries, efficient resource allocation, and policy evidence. Successful implementation requires low-cost digital tools, interoperability, ethical governance, multidisciplinary collaboration, local validation, and explainable AI. This approach may enable earlier intervention and improve outcomes for SMA3 patients in resource-limited settings.

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**Introduction**

Spinal Muscular Atrophy Type 3 (SMA3), also known as Kugelberg-Welander disease, is a milder, later-onset form of SMA characterized by slowly but progressive muscle weakness and motor function decline [1–4]. SMA3 onset presents after 18 months of age with clinical signs of walking difficulty, frequent falls, and proximal muscle weakness, especially in the lower limbs [2]. The underlying cause is a homozygous deletion or mutation in the Survival Motor Neuron 1 (SMN1) gene on chromosome 5q13, which leads to reduced levels of functional SMN protein, which is essential for motor neuron survival [5–8]. SMA3 is attributed to the presence of a higher number of SMN2 gene copies, which partially compensate for the lack of SMN1 by producing limited amounts of functional SMN protein [5,9,10]. Despite this genetic advantage, patients with SMA3 will still experience a gradual decline in muscle strength and ambulatory function over time [1–3]. While life expectancy is usually unaffected, as the disease progresses, individuals may develop scoliosis, fatigue, and finally loss of ambulation [2,11].

In many low- and middle-income countries (LMICs), SMA3 remains underdiagnosed due to its subtle early symptoms, absence of universal screening programs, and limited healthcare professional awareness. The lack of genetic diagnostic infrastructure and follow-up systems further exacerbates delays in recognition [12–16]. Hence, affected individuals may miss the opportunity for early intervention, rehabilitation planning, potentially lifelong independence, and affect quality of life [12,13,17,18].

Machine Learning (ML) and Artificial Intelligence (AI) technologies offer promising avenues for scalable, cost-effective screening, monitoring, and personalized rehabilitation strategies in SMA [19–24]. These tools may improve community-based detection through symptom modelling, wearable sensor data analysis, and remote assessment [25–27]. Their implementation may not require extensive medical infrastructure, making them highly suitable for LMIC contexts.

This study aims to develop a conceptual framework for integrating Artificial Intelligence (AI), Machine Learning (ML), and Internet of Things (IoT) technologies to improve the early

detection, continuous monitoring, and personalized management of Spinal Muscular Atrophy Type 3 (SMA3) in low- and middle-income countries (LMICs). The framework addresses persistent challenges in resource-limited settings, including delayed diagnosis, limited access to genetic testing, shortages of specialists, fragmented follow-up, and weak referral systems, by combining multimodal clinical, wearable, smartphone, and health system data into an AI-driven decision support ecosystem. The novelty of this study lies in proposing a comprehensive, end-to-end digital health architecture that integrates multimodal data acquisition, cloud-based preprocessing, explainable AI/ML analytics, IoT-enabled longitudinal monitoring, and health system integration within a single framework specifically designed for SMA3 care in LMICs. Unlike previous studies that primarily focus on isolated AI prediction models or wearable monitoring technologies, this framework simultaneously emphasizes clinical decision support, personalized rehabilitation, early warning systems, national registry development, and policy-informed resource allocation while incorporating interoperability, ethical governance, affordability, and local validation as essential implementation components.

Literature Review

A. Role of AI

AI has demonstrated a remarkable ability in recognizing abnormal motor patterns through video or gait analysis [25,28]. By training deep learning models on large datasets of children's movements, AI may identify subtle deviations in posture, balance, and walking patterns associated with SMA, especially SMA3 [22,25,28]. Gait abnormalities such as a Trendelenburg sign, toe walking, or compensatory upper body movements may be quantified and tracked over time. Such systems can be integrated with wearable sensors or even standard smartphone cameras for community-level screening [26,27]. This non-invasive and scalable approach holds great promise for remote or underserved areas lacking paediatric neurologists.

Smartphone-based tools provide an accessible platform for parents, teachers, and community health workers to detect early signs of neuromuscular disorders [29]. Applications may include checklists and interactive features to guide observation of red flags in SMA such as Gowers' sign, difficulty standing from the floor, or climbing stairs [1–4]. These tools may also prompt users to record and submit videos for AI-assisted analysis or teleconsultation in cloud-computing-based digital media content distribution by engaging non-specialists in surveillance; early detection becomes a shared responsibility at the community level [30]. This empowers families to seek early intervention and minimizes the risk of missed cases.

AI-powered decision support systems can be embedded into electronic medical records or standalone applications for use by general practitioners in LMICs. Such systems analyse patient

history, developmental milestones, physical findings, and caregiver concerns to generate a list of differential diagnoses. When SMA3 is suspected, the system may recommend referral, further evaluation, or supportive tests based on local resource availability by fine-tuning the model [31]. These tools may reduce diagnostic errors and support capacity building in primary care settings. Ideally, this system can run its function offline or with limited connectivity to make it suitable for rural deployment.

One of the key advantages of AI and digital screening tools is the ability to flag potential SMA cases before significant disability occurs [22]. Early recognition allows for timely initiation of physiotherapy, orthotic support, and non-pharmacological interventions such as electrical stimulation that may preserve motor function [32,33]. Monitoring deterioration before loss of ambulation is significant in SMA3, where progression can be slow and deceptive [10,15,34]. Preventing avoidable complications such as contractures or scoliosis can significantly enhance long-term outcomes [12]. These technologies thus may shift care from curative to preventive, which is especially impactful in LMIC healthcare systems.

B. Applications of ML in SMA3

ML models may potentially revolutionize SMA3 care by recognizing early-stage decision-making of SMA patients using clinical and demographic data. One of the possible applications is risk stratification using motor milestone data and family history, where ML algorithms can learn from annotated datasets to identify children at higher likelihood of SMA diagnosis and progression [19,20,23,24,28]. For example, delayed independent walking, difficulty climbing stairs, or a positive family history can be weighted to calculate a predictive risk score. These models may also forecast long-term ambulation outcomes based on baseline functional assessments and age at onset [20]. This enables clinicians and families to plan proactively, including educational adaptations, environmental support, and mobility aids. Such predictive analytics can also facilitate earlier referrals, even before genetic testing is confirmed [24,35].

It is feasible in LMIC settings to train ML models using non-genetic inputs, such as demographics, symptom checklists, and video-observed behaviours. These inputs can be collected by community health workers, teachers, or parents without the need for laboratory or clinical infrastructure. Through supervised learning, ML models can detect patterns correlating with SMA3 and differentiate it from other neuromuscular disorders [24]. Locally relevant datasets improve the model's contextual accuracy, ensuring it reflects population-specific variations in development and presentation. This democratizes the diagnostic process and reduces overreliance on centralized hospital systems. In turn, it facilitates earlier detection and intervention in rural or underserved regions.

To support scalability and real-world impact, a new concept called TinyML models can be embedded into low-resource mobile applications optimized for offline use. These applications require minimal processing power and can operate on standard Android smartphones where mostly available across LMICs [36]. It may guide users through structured assessments, symptom checklists, and video submissions analysed by embedded models. They may also include multilingual support, caregiver education, and referral recommendations. The user interface must be designed for usability by non medical person, such as community nurses or parents, to support broad uptake. This approach ensures ML's benefits are not limited to high-resource settings but extend equitably to all affected populations.

Methods

This study employed a conceptual framework development approach based on a comprehensive narrative review and systems design methodology. Relevant literature was identified from peer-reviewed publications indexed in major scientific databases, including Scopus, PubMed, Web of Science, and IEEE Xplore, focusing on studies published between 2019 and 2025. The literature search used combinations of keywords such as *Spinal Muscular Atrophy Type 3*, *Kugelberg-Welander disease*, *artificial intelligence*, *machine learning*, *Internet of Things*, *wearable sensors*, *digital health*, *clinical decision support*, and *low- and middle-income countries*. Additional references were identified through backward citation tracking to ensure comprehensive coverage of foundational studies.

The proposed framework was designed according to widely accepted digital health principles, emphasizing interoperability, explainable artificial intelligence (XAI), ethical governance, data security, scalability, affordability, and local validation. Rather than developing or validating a predictive algorithm, this study provides a theoretically grounded implementation framework intended to guide future empirical research and digital health deployment for SMA3 management in low- and middle-income countries.

Results and Discussion

A. Conceptual Framework for LMICs

An integrated digital platform offers a unified solution that connects data collection, intelligent analysis, and actionable outputs to support the early detection and personalized management of SMA3. Data inputs can include caregiver-recorded videos of walking or standing, wearable-derived mobility data (such as step count or gait rhythm), and structured symptom checklists assessing motor milestones and red flags [24,28]. These multimodal inputs allow for a comprehensive functional profile without the need for hospital-based evaluation. Community health workers or parents may upload data via a mobile app, enabling remote assessment even

in low-resource areas. The platform's design should support data collection over time, enabling longitudinal tracking and trend analysis. This continuous monitoring helps detect subtle motor decline that may not be evident during brief clinical encounters. Fig. 1 summarizes the integrated workflow of the digital SMA3 platform, including multimodal data collection, ML-based analysis, personalized outputs, and system-level monitoring.

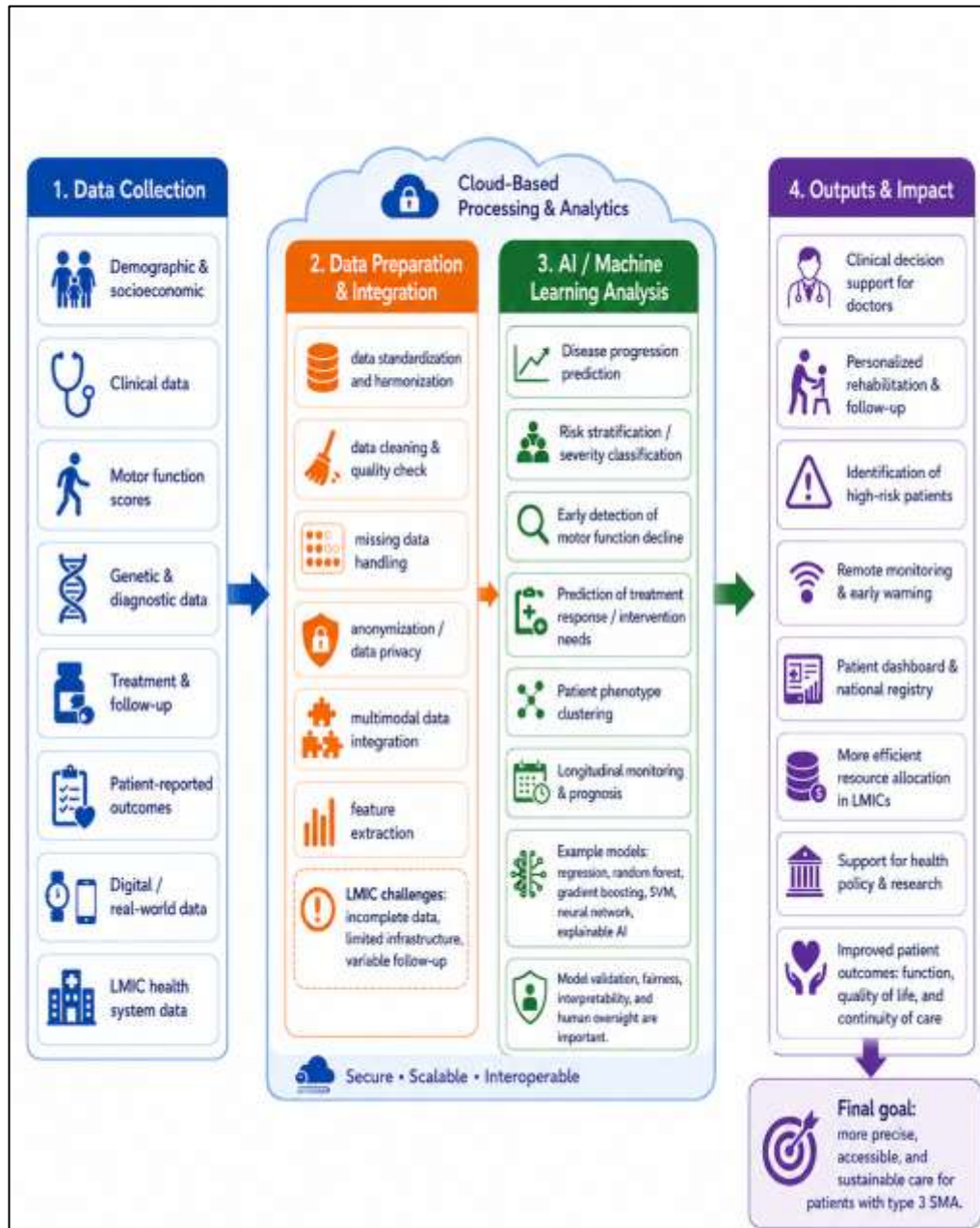


Fig. 1. Integrated workflow of the digital SMA3 platform, including multimodal data collection, ML-based analysis, personalized outputs, and system-level monitoring.

The analytics component is powered by ML algorithms trained to predict disease severity and progression risk based on the collected data [24,28]. These algorithms can recognize delays in motor development, generate risk scores, and issue automatic alerts when thresholds are exceeded. By combining historical and real-time inputs, the system enhances diagnostic confidence and supports proactive management. Alerts can be routed to caregivers, primary care providers, or referral centres to prompt timely action. The platform may also categorize patients into severity levels (e.g., mild, moderate, severe) to guide follow-up frequency. These analytics are critical in LMICs where specialists are scarce, and misdiagnosis is common.

Outputs from the platform are tailored for end-user relevance and clinical utility [24,28]. Caregivers receive easy-to-understand guidance, such as tips for mobility support, environmental modifications, or early physiotherapy exercises. Health workers are provided with referral recommendations based on severity predictions and local service availability. Physiotherapists may receive suggested exercise plans adapted to the patient's current function, encouraging personalized and efficient intervention. For cases flagged as urgent, the system can generate referral letters or connect users with teleconsultation services. This closed-loop feedback ensures that data collected is translated into meaningful, timely actions. It will foster collaboration between families, health workers, and rehabilitation specialists.

For real-world scalability, the platform can be deployed through existing maternal-child health or school health programs in LMICs [37]. These programs already serve as contact points for developmental surveillance and vaccination, making them ideal for layered screening initiatives. Health workers trained in basic motor assessment can be empowered with digital tools to enhance accuracy and reach. Integrating SMA surveillance into routine visits ensures population-level coverage without requiring new infrastructure. Furthermore, it aligns with WHO goals for early detection of childhood disabilities within primary care. Embedding digital SMA screening in such programs creates a sustainable, community-rooted solution.

To support decision-makers and clinicians, the platform can offer cloud-based dashboards accessible to primary health centres and district hospitals [19,24,28,30]. These dashboards visualize trends in motor function, alert histories, patient risk profiles, and referral patterns. Aggregated data can help identify regional hotspots, track adherence to follow-up, and optimize resource allocation. Secure, encrypted access ensures patient privacy while allowing multi-tiered visibility for different healthcare roles. Remote experts can also use the dashboard for asynchronous consultation or second opinions. Ultimately, this centralized data view strengthens health systems' ability to monitor neuromuscular disorders and intervene earlier, even with limited manpower.

B. Challenges and Enablers in LMIC Context

Challenges in deploying digital health solutions for SMA3 in LMICs include digital illiteracy among both caregivers and frontline healthcare providers. Many individuals may lack familiarity with smartphones, apps, or data input procedures, reducing the effectiveness of user-centered technology [38,39]. Training and ongoing support will be necessary to ensure meaningful engagement and minimize misuse or underutilization.

Infrastructure limitations such as inconsistent electricity, poor internet connectivity, and lack of access to compatible devices pose additional barriers [39]. These limitations are especially pronounced in rural and remote areas where the need for early detection may be greatest. Without infrastructure support, even the most advanced tools may fail to deliver impact at the ground level.

Cost and cultural acceptance of wearable or mobile technology can also hinder adoption [39]. Families may not afford or prioritize devices, especially when symptoms are mild, or diagnosis is delayed. In some communities, unfamiliarity or distrust toward technology-based health interventions can limit participation unless culturally sensitive approaches are used.

On the other hand, enablers include the growing mobile phone penetration across LMICs, with increasing numbers of households owning smartphones [40]. This trend opens new avenues for mobile-based screening, monitoring, and caregiver education. As device access improves, so does the potential for scalable digital health solutions.

There is increasing interest from NGOs and government bodies in advancing disability care and digital health, especially as part of universal health coverage agendas [41]. These stakeholders can provide financial support, policy integration, and program sustainability. Their involvement ensures that digital SMA platforms are not just pilot projects but part of long-term health system strengthening.

C. Ethical and Policy Considerations

Ensuring informed consent and child data protection is essential when collecting and analysing sensitive health information through digital platforms [24,42]. Caregivers must clearly understand how their child's data will be used, stored, and shared, especially in communities with limited digital literacy. Strong safeguarding such as anonymization, encryption, and ethical oversight must be in place to protect patient data and uphold trust [42].

Efforts must also focus on avoiding the exclusion of marginalized populations, such as those in rural, low-income, or low-literacy settings. Without inclusive design and outreach strategies, digital health innovations may widen existing health disparities [24]. Equitable access requires culturally adapted interfaces, community outreach, and offline functionality for areas with limited connectivity.

There is an urgent need for national and regional policies to guide the use of AI and ML in health diagnostics, particularly in LMICs [24,43]. Clear frameworks are required to regulate algorithm safety, transparency, accountability, and integration into public health systems. Without policy guidance, implementation may be fragmented or face resistance from regulators and health authorities.

Finally, public-private collaborations are key to funding, scaling, and sustaining digital SMA initiatives. Partnerships between governments, NGOs, academic institutions, and technology companies can align technical innovation with health priorities. These collaborations can help lower costs, ensure interoperability, and accelerate deployment across diverse settings.

D. Future Directions and Recommendations

There is an urgent need for pilot studies that test open-source digital tools for early detection and monitoring of SMA3 in real-world LMIC settings. Open-source frameworks allow for cost-effective customization, community input, and local adaptation. Such research will generate practical evidence on feasibility, accuracy, and scalability.

Machine learning models require diverse, high-quality datasets to perform reliably across populations. Establishing global and regional SMA datasets, including demographic, clinical, and motor performance data, is essential. Data sharing collaborations can reduce bias, improve generalizability, and accelerate algorithm refinement [24]. Bridging clinical and technical expertise is critical for successful development and implementation of AI/ML tools in SMA care. Neurologists can provide domain-specific insights, while AI engineers ensure model performance and scalability. Public health professionals play a key role in integration within existing health systems and ensuring community engagement.

Frameworks for regulatory approval and integration into LMIC healthcare systems to transition from research to routine use, regulatory pathways must be established for AI and IoT-based diagnostics. Clear frameworks are needed to assess safety, accuracy, data privacy, and ethical use in paediatric populations. Policies must also support integration into primary care workflows, especially in resource-limited settings.

Conclusion

Although SMA3 is generally less severe than earlier onset forms, delayed recognition can still lead to progressive functional decline and substantial long-term disability. Early identification and continuous monitoring are therefore essential to preserve mobility, independence, and quality of life. AI, ML, and IoT-based tools offer scalable and affordable solutions to support detection, risk stratification, and longitudinal follow-up. In low- and middle-income countries, these technologies may help bridge gaps in specialist access, strengthen local

clinical decision-making, and improve continuity of care. With thoughtful, ethical, and context-specific implementation, digital health innovations can meaningfully enhance outcomes for patients living with SMA3.

Conflict of Interest

The author declares that there is no conflict of interest.

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