

From questions to impact in spinal muscular atrophy - identifying community-driven research priorities through a multi-stakeholder European initiative

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Abstract

Background Advances in medicines have transformed the outlook for people living with spinal muscular atrophy (SMA), yet many needs remain unmet. As the scientific and clinical environment evolves, aligning research priorities with the lived experience of patients is increasingly recognised as essential to ensure that investment delivers meaningful, community-relevant impact. While academic and industry agendas have yielded breakthroughs, incorporating patient and caregiver perspectives can further shape the fundamental and clinical research landscape. This project aimed to (1) identify the “Top 10” unanswered research questions in SMA from the perspectives of patients, caregivers, and healthcare professionals; (2) engage the wider research and clinical community to further refine these priorities through expert dialogue; and (3) disseminate the outcomes to raise awareness, establish a feedback loop with the community, and stimulate research and funding activity. More broadly, it aimed to illustrate how participatory approaches can inform rare disease research agendas. SMA Europe led a multinational Priority Setting Partnership using the James Lind Alliance approach. Over 900 participants from 22 countries submitted research priorities, which were consolidated and ranked through surveys and a final consensus workshop to produce a community-defined “Top 10”. In subsequent phases, a transdisciplinary expert workshop was convened to identify research areas with the highest potential impact and a dissemination campaign was launched, promoting uptake among researchers and funders.

Results The “Top 10” priorities grouped into eight research clusters, ranked by importance: 1. Regeneration of neuromuscular system; 2. Biomarkers; 3. Metabolism and diet; 4. (Personalised) Physiotherapy; 5. Understanding SMA impact on pathways, cells and tissues; 6. Bones and joints; 7. Finding solutions to manage fatigue; 8. Innovative technology. While some align with current research, others highlight underexplored areas central to quality of life. The participatory process demonstrated the role of patient organisations as conveners and co-creators in shaping research strategy.

Conclusions This initiative represents the first European-wide, patient-led initiative to define research priorities in SMA and reflects a growing shift toward inclusive governance in rare disease research, beyond Europe alone.

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By grounding biomedical innovation in community-defined priorities, it illustrates a replicable model for participatory research and transdisciplinary collaboration. SMA Europe is using the identified priorities to guide its research strategy and encourages researchers and funders to consider them in future initiatives.

Keywords Spinal muscular atrophy · Priority setting partnership · Rare diseases · Patient and public involvement · Transdisciplinary research · Participatory research · Research agenda · Community-led research

Background

The growing role of participatory research

Participatory research approaches have gained momentum across health research, particularly in the rare disease field, where conventional, top-down agenda-setting may not fully reflect the needs of the lived experiences of patients and caregivers. Patient and Public Involvement (PPI) is increasingly recognised as key to improving the relevance, equity, and real-world impact of research, especially in fields marked by knowledge gaps and limited funding [1]. Structured models such as the James Lind Alliance (JLA) Priority Setting Partnership (PSP) offer transparent, consensus-based frameworks for identifying research questions that matter most to affected communities [2, 3].

The evolving landscape of SMA

SMA is a rare autosomal recessive neuromuscular condition caused by mutations or deletions in the *SMN1* gene, leading to reduced production of the survival motor neuron (SMN) protein, essential for the survival and maintenance of motor neurons [4, 5]. The disease is characterised by a spectrum of presentations, ranging from severely impacted neonates to less severely impacted adults [6]. If untreated, SMA leads to progressive muscle weakness and atrophy, which results in loss of movement and premature death in the most severely affected individuals [7].

In the past decade three disease-modifying therapies boosting SMN levels - nusinersen, onasemnogene APOB acceptor modification, and risdiplam - have transformed clinical outcomes [8–11]. Yet, these treatments do not represent a cure and SMA remains a complex, lifelong condition with many unmet needs and open questions. These include uncertainties around long-term disease progression, impacts of therapies and combinatorial therapies, musculoskeletal complications, metabolic and nutritional impacts, bulbar and respiratory issues, and psychosocial challenges [12]. Increasingly, SMA is understood not only as a neuromuscular disorder but as a condition affecting multiple domains requiring comprehensive, individualised care [13].

Problem statement: the gap between research and real-world needs

As for many rare diseases, SMA research is shaped by structural limitations, including constrained funding for fundamental biological research and the inherent challenges of small clinical study populations [14, 15]. To date, pre-clinical and clinical research has focused primarily on SMN-dependent pathways, leading to transformative therapies that address the genetic root cause and improve neuromuscular function [8–10, 16]. However, these foundational breakthroughs do not fully address the broader spectrum of needs experienced by people living with SMA [17, 18]. Unlocking additional solutions may require looking beyond SMN biology, toward underexplored mechanisms that could inform both clinical care and future (pharmacological) therapies [19]. In this context, integrating the perspectives of individuals with SMA, their families, and frontline professionals is essential, to direct fundamental, clinical and care-related research toward real-world challenges and to ensure that limited resources are invested in areas of greatest impact for the community [20, 21].

Project rationale and aim

This project aimed to help integrate the perspectives of real-world needs of the SMA community into research priorities and represents the first formal, Europe-wide initiative to identify and disseminate community-driven research priorities in SMA. The project consisted of three interlinked phases, designed to widen stakeholder engagement across them. Phase I aimed to gather and prioritise unanswered research areas from patients, families, and healthcare professionals [3, 22]. Phase II introduced academic researchers and funders to bridge priorities with translational research, a growing approach in participatory health research [23]. Phase III aimed to disseminate the “Top 10” identified priorities, expanding the audience further to include public stakeholders and decision-makers, and ensuring that the outputs of the participatory process could inform systemic change [20].

Significance of the initiative

This initiative illustrates a shift from conducting research on people with rare diseases to conducting research with and for them, an approach increasingly recognised in the literature as essential for producing relevant and equitable outcomes [23, 24].

Anchoring research governance in lived experience, the PSP offers not only a democratically endorsed set of priorities but also a replicable model for participatory, transdisciplinary innovation in rare diseases and other conditions [20, 22]. For SMA, the PSP provides a resource of patient-defined priorities to guide decisions on which research questions to pursue and fund, translating community needs into concrete actions within research planning and governance.

This paper outlines the approach, outcomes, and broader implications of the SMA PSP, demonstrating how patient-led collaboration can complement traditional research and help shape more impact-driven strategies in rare disease science [3, 25].

Methods

The initiative was led by SMA Europe, a patient-led umbrella organisation representing 27 national SMA groups during the study, today 30. This study followed a structured, participatory approach to research agenda-setting, grounded in PPI principles and implemented in three interconnected phases (Table 1). Participation was voluntary, and digital informed consent was obtained before involvement.

Phase I: Gathering uncertainties and identification of priority topics for research

The first phase of this initiative followed version 10 of JLA PSP approach [3], with the aim of identifying the “Top 10” research uncertainties that highlight important topics for research in SMA from the perspectives of individuals with SMA, caregivers, and healthcare and social care professionals (HCPs). Respondents were invited to identify, without restriction to a particular type or field of research, questions on SMA they would like future research to answer. This phase was jointly coordinated by SMA Europe and a JLA advisor, overseen by a multi-stakeholder Steering Group and run from March 2022 to October 2023. Further methodological details are available at: Spinal Muscular Atrophy PSP Protocol | NIHR JLA [26] and Spinal Muscular Atrophy (Europe) | NIHR JLA [27].

The Steering Group included adults living with SMA, parents, clinicians, allied health professionals, and a representative from the European Neuromuscular Centre (ENMC). Members were from across Europe, including Belgium, Bulgaria, France, Germany, North Macedonia, Poland, Russia, the Netherlands, Norway, Switzerland, and the UK. Professional representation included neurologists, physiotherapists, a nutritionist, a clinical

Table 1 Overview of SMA Europe’s three-phase participatory research agenda-setting initiative

Phase	Main Activities	Stakeholders Involved	Key Outputs
Phase I • Gathering uncertainties and Identification of research priorities • JLA PSP approach	• Multilingual survey entries (1337 collected questions) • Categorising and summarising the responses, and literature screening • Prioritisation survey (582 participants) • Final consensus workshop	• People living with SMA • Caregivers • HCPs	• 52 summary questions • Shortlist of 24 research priorities • Community-defined “Top 10” research priorities
Phase II • Towards Solutions: Expert Priority-Development	• Transdisciplinary workshop (International Scientific Congress on SMA, 2024): Mapping “Top 10” priorities to research gaps and translational areas; Clustering priorities	• Phase I stakeholders • Academic researchers • Funders	• 8 thematic research clusters • Strategic research directions
Phase III • Dissemination and Community Engagement	• #WeAreOne Campaign: Co-designing outreach strategy; Social media, newsletters, direct engagement; Targeting outreach to funders and stakeholders	• Phase II stakeholders • Policy Makers • General Public	• Broad awareness of “Top 10” • Feedback loop with community • Strengthened funding momentum and implementation interest

Table 1 illustrates a) the flow of phases with their activities from initial gathering uncertainties using the James Lind Alliance (JLA) approach, to expert agenda-development, to public dissemination and community engagement, b) the enlargement of involved stakeholders over time from a core group to an inclusive-multistakeholder involvement, and c) key outputs in each phase. HCPs: Healthcare professionals

neuropsychologist, and a speech and language therapist. Meetings were held online, and a trained JLA advisor ensured adherence to the official process. A PPI specialist researcher provided analytical support throughout. The steps of the SMA PSP were as follows:

- **Step 1 – Gathering Uncertainties:** In October 2022, an online survey available in 17 languages was launched and disseminated through national SMA organisations, clinical networks, and advocacy channels, to collect answers from people living with SMA, caregivers, and HCPs representing diverse backgrounds and across the SMA continuum.
- **Step 2 – Data Processing and Question Synthesis:** All responses were screened for relevance and duplication. Using an inductive coding approach, the project lead, together with an independent reviewer, grouped similar responses, which were reviewed and refined into summary questions by a sub-group of the Steering Group.
- A targeted and structured literature review, focused on peer-reviewed publications, systematic reviews, clinical guidelines and meta-analyses, was conducted to determine questions already answered by existing evidence and exclude them. Wording was refined through feedback rounds with the Steering Group to ensure that each question reflected the intent of the community submissions.
- **Step 3 – Prioritisation Survey:** The summary questions were distributed in a second multilingual survey between June and September 2023 to individuals living with SMA, caregivers, and HCPs, who selected those questions they considered most important for future research. To ensure balance, responses were weighted equally across stakeholder groups. The 24 highest-ranking questions were shortlisted for the final consensus workshop.
- **Step 4 – Consensus Workshop:** The final step of Phase I was an in-person consensus workshop held in Haarlem in October 2023. This event brought together a balanced group of 24 participants, including adults living with SMA, caregivers, and HCPs. Using the Nominal Group Technique, a structured consensus method designed to promote equitable participation and avoid groupthink, participants iteratively reviewed, discussed, and ranked the shortlist of 24 research questions. Through small group work and collective voting rounds, the group reached agreement on the final “**Top 10” priority topics for research** for SMA, as pre-set by the JLA PSP framework.

- This workshop concluded the JLA Priority Setting Partnership and marked the transition into the next phase of the initiative.

Phase II: Towards solutions - transdisciplinary agenda-development

The second phase aimed at translating the identified priorities into actionable research pathways. This phase extended beyond the JLA framework and was grounded in principles of PPI and cross-sector collaboration.

The central activity of Phase II was a transdisciplinary expert workshop held during the **4th International Scientific Congress on SMA** in 2024. While the consensus workshop in Phase I focused on finalising the “Top 10” list, this panel-style workshop brought together a group of stakeholders, including clinicians, SMA advocates, people living with SMA, and caregivers and adding academic researchers and funders, to translate the community-defined uncertainties into more specific scientific terms, cluster them into broader thematic research areas with translational relevance and explore how the identified priority topics could inform future research strategy and investment. While the panel itself consisted of group of six invited participants, audience members were also welcome to contribute to the discussion.

Phase III: Dissemination and community engagement

Phase III focused on disseminating the outcomes of the SMA PSP and strengthening engagement across patient and research communities and beyond. This phase was designed an extension of the participatory logic of the initiative, to create transparency, encourage feedback, and mobilise cross-sectoral momentum.

A campaign titled “Top 10 Priorities. Top 10 Dreams. Together, #WeAreOne” was co-designed with community stakeholders, including researchers and pharmaceutical industries, to ensure inclusive and accessible communication. It was launched by SMA Europe and implemented across multiple channels, including social media, SMA Europe newsletters, national patient group networks, and targeted outreach to the community, researchers and funders.

Results

The results of this study are organised according to the three-phase structure of the initiative, each contributing uniquely to building an inclusive, actionable, and strategically aligned research framework grounded in lived experience (Table 1).

Phase I: Community-defined “Top 10” priority topics

927 individuals from 22 European countries participated in the Phase I open survey, of whom 487 completed it and submitted 1337 perceived research uncertainties. Responders were individuals with SMA (45%) and caregivers (36%), which represented the broad spectrum of SMA disease severity and clinical presentations, and healthcare and social care professionals (19%). Among respondents with SMA, 40% were aged 36–50 years, 26% 27–35, 13% 50–65, 9% 20–26, 7% 66 years or older, and 5% 13–19. Most caregivers (83%) were caring for a person living with SMA aged 0–19 years. The collected research uncertainties were consolidated through thematic coding, expert review, and community feedback into 52 summary questions, covering biomedical, psychosocial, quality-of-life, clinical, and care-related clusters. Those summary questions were narrowed to a shortlist of 24, through a second-round prioritisation survey, with a total of 582 participants and equal stakeholder weighting. This was followed by a multi-stakeholder in-person consensus workshop, during which they agreed on the final “Top 10” priorities.

These priorities span regenerative medicine (neuromuscular system, muscles, and bioengineering), biomarkers, metabolism and nutrition, personalised physiotherapy, multisystem impact, musculoskeletal management, fatigue and assistive technologies. While some aligned with current research efforts, others surfaced underexplored areas central to quality of life. Table 2 presents the final “Top 10” research areas, accompanied by quotes that illustrate the community perspectives. Supplementary Table 1 (see Additional File) includes the remaining research questions ranked 11 to 24.

This process exemplifies how transparency and expertise from lived experience can inform research direction. It also established the groundwork for the transdisciplinary efforts described in Phases II and III.

Phase II: Thematic research clusters

Building on the identified research priorities, Phase II aimed to translate the community-defined uncertainties into a more precise scientific language, structuring them into actionable research pathways while considering how they relate to current research gaps and funding patterns. During a transdisciplinary expert workshop hosted at the 4th International Scientific Congress on SMA in Ghent (2024), health care professionals, patient advocates, funders and academic researchers discussed how to build multi-actor research programs that respond directly to patient-defined needs while also aligning with existing scientific and policy frameworks, reviewed the priorities, examined their scientific meaning, and clustered them into eight thematic research clusters (Table 3). These clusters serve as a bridge between community-defined uncertainties and research programmes enabling targeted funding, cross-sector coordination, and translational planning. They reflect both the language of the community and the possibilities identified through scientific dialogue. The inclusion of researchers, who had not participated in Phase I, added new perspectives and helped strengthen the bridge between lived and clinical experience and scientific opportunity.

By convening this multistakeholder dialogue and initiating a continuous feedback loop, Phase II laid the groundwork for a more coherent and community-aligned research strategy for SMA in Europe. It also helped identify where targeted funding, infrastructure, or policy support may be needed to advance the most urgent priorities, marking a shift from identification of priority topics to an initial **research agenda- development**.

Phase III: From priorities to practice – mobilising for impact

The final phase focused on dissemination, advocacy, and impact mobilisation. The communications campaign *#WeAreOne* (Supplementary Table 2, Additional File) was coordinated with Phase II to: (1) raise awareness and amplify the research priorities across public and professional audiences; (2) demonstrate a feedback loop by showing participants how their input shaped the final priorities; (3) foster an informed, engaged, and active community of national SMA patient organisations who work together at the European level; (4) stimulate interest from funders, researchers, and institutions to act on the identified priorities; (5) underscored to the wider community the relevance of research, even in the current post-treatment era.

The campaign was translated by European national SMA patient organisations in their languages and disseminated it further. Communication materials were intentionally framed to support community co-ownership of the research agenda and promote continued engagement with the priorities over time.

By presenting priorities in accessible formats, leveraging national SMA organisations, and embedding PPI into outreach, this phase extended the participatory logic of the PSP. It redefined dissemination not as an endpoint, but as a driver of co-production, implementation, and accountability, and helped legitimise the participatory process and reinforced the visibility of the community’s voice beyond the SMA field.

Phase III positions the PSP as a living roadmap, whose success depends on stakeholders acting on the insights generated and on the SMA community remaining engaged in shaping future directions. Phase III is, by nature, an ongoing process. SMA Europe continues to leverage the identified priorities in its advocacy and keeps them

Table 2 Ranked community-defined SMA research priorities and community perspectives

Rank	Summarised Community (incl. HCPs) SMA Research Priorities	Examples of Individual Community-Generated Comments
1	Can nerve cells (especially motor neurons) be regenerated in people with SMA?	• Motor neuron regeneration. • Are there any medicines that can awaken the motoneurons? • Is there the prospect of a treatment to repair damaged or dead neurons and what is the potential upside? • Regeneration of nerve cells • Can standby nerve cells be activated so that they can take over functions of deceased nerve cells?
2	Are there biological markers (biological molecules in blood or other tissues) that can indicate the progression of SMA?	• Power of the biological marker to indicate the progression of SMA. • Find biological markers that tell us the stage and rate of progression of the disease.
3	Can damaged muscles be rebuilt in people with SMA? How can muscles be strengthened?	• Strengthening of the muscles affected by SMA • Possibility of action to accelerate and improve muscle growth • Drugs that can strengthen muscles additionally (with parallel reception of pathogenetic therapy) • Possibility of rebuilding lost muscle
4	Do people with SMA benefit from a specialised diet (e.g. amino acid diet) or dietary changes such as treatment with supplements?	• Dietary requirements for people living with SMA. Positive and negative effects of diet. • Focus on specialised dietary guidance • Is there a diet that can help delay muscle loss? • Research into dietary requirements, if there is best food • Dietary evidence base-keto/ AA diet • How can diet positively/negatively affect SMA.
5	What is the best physiotherapy approach for people with SMA? Does it need to be tailored to the individual (e.g. according to age and type of SMA)? Does it need to change during treatment with one of the new drugs?	• Physiotherapy approaches and monitoring for treated and non-treated patients • Standard therapy or need to be tailored? • Type I physiotherapy learnings. • Intensity of therapy input and functional outcomes • Impact of physio in rehabilitation • Physiotherapy programs and techniques (such as using vibration plates or cervical tractions)
6	What parts of the body are affected by SMA other than nerves and muscles?	• Impact of SMA on other parts of the body beside the muscles: The effect of SMA on the intestine and similar organs • What impact does SMA have on the endocrine system? • What is the impact of SMA on body parts other than muscles and nerves?
7	Does electrical stimulation of nerves and/or muscles benefit people with SMA?	• Effects of electrical stimulation on people living with SMA • Is spinal cord stimulation possible to improve arm use? • Can current/electrodes help SMA for more movement? • Can myo-stimulation techniques be used on children with SMA?
8	What is the best approach to treating and preventing problems with bones and joints (orthopedic problems) for people with SMA?	• Therapies and exercises to prevent problems with bones and joints. • Contractures in knees, tendon surgery • Hip surveillance • How to avoid hip dislocation, monitoring hips
9	What are the best ways to treat and manage fatigue in people with SMA?	• Understanding the concept of fatigue, treatment, prevention in SMA • What does fatigue look like and how can it be managed, how can fatigue be combatted • How does fatigue have on daily function in SMA? And how do people with SMA manage this? Is it effective?
10	How can new technology and assistive devices help people with SMA and their carers?	• Impact of new technology and assistive devices in SMA patients and carers. • Accessibility - facilitating access to devices • greater need for technological support and assistive devices: Often it's small things that make a world of difference, like apps that improve quality of life. • What are the assistive technologies of the future?

Table 2 presents the final “Top 10” list with the ranked research priorities identified during the SMA Priority Setting Partnership. The “Community SMA Research Priorities” are phrased as summary questions, reflecting the questions emerged from survey responses and grouped by themes. The examples included, while not exhaustive, illustrate the real-world context and experiential knowledge underpinning each question, reinforcing the role of lived experience in guiding translational rare disease research. To highlight the authenticity of the community input, the examples are reported verbatim, as submitted by respondents

Table 3 Thematic research clusters derived from the “Top 10” community-defined SMA research priorities

Thematic Cluster	Description	Community SMA Research Priorities
Regeneration of the Neuromuscular System	Restoring neuromuscular function through biological or technological means, including neuron and muscle regeneration and electrical stimulation.	1, 3, 7
Biomarkers and Disease Progression Tracking	Developing measurable indicators to track disease progression and treatment efficacy.	2
Metabolism and Diet	Understanding the metabolic impacts of SMA and optimising dietary strategies for support.	4
Personalised Physiotherapy	Adapting physiotherapy approaches to individual disease progression, treatment status, and needs.	5
Systems Biology: Multisystem Impact	Exploring how SMA affects broader body systems beyond motor neurons and muscles.	6
Musculoskeletal and Orthopaedic Management	Managing complications such as scoliosis and contractures to improve long-term mobility and quality of life.	8
Fatigue: Physical and Psychological Dimensions	Investigating physical and psychological contributors to fatigue and how to assess and mitigate them.	9
Assistive Technology and Innovation	Designing and optimising assistive devices that enhance independence and reduce caregiver burden.	10

In Table 3, each cluster was co-defined by a transdisciplinary expert group to cluster the “Top 10” priorities identified in Phase I. This framework provides a foundation for future funding strategies, cross-sector collaboration, and translational research programming

aligned with the community’s evolving needs, by maintaining strong communication between the community, stakeholders and researchers.

Discussion

This study represents a large-scale, community-driven effort to reshape research governance in the rare disease field. By applying and expanding the JLA Priority Setting Partnership (PSP) approach, SMA Europe not only produced a ranked list of research priorities grounded in lived experience but also piloted a model of participatory science for research in SMA, one that connects agenda-setting, transdisciplinary dialogue, and strategic dissemination.

An additional aim of this process is to enhance the community’s understanding of the relevance of research and how it connects to the uncertainties identified by the community itself, which are driven by the lived experiences of people living with SMA. This understanding empowers the community to advocate across Europe and beyond, raising these priorities on national research and policy agendas. For funders and researchers, the goal is to familiarise them with the community-defined priorities and provide insight into how the community perceives the field. The eight thematic research clusters offer a more scientifically grounded framing of these priorities, translating the community-defined uncertainties into actionable research areas.

Centring lived experience to reframe research

The core of the PSP contribution lies in its shift from researcher-driven priorities to a community-defined research prioritisation and offers a compelling model of transparent agenda-setting anchored in lived experience [28].

Historically, SMA research has focused predominantly on developing disease-modifying therapies targeting *SMN* genes, yielding transformative treatments such as nusinersen, risdiplam, and onasemnogene abeparvovec [8–11, 16]. Alongside these efforts, substantial clinical and care-related research has focused on improving diagnosis, monitoring, disease management, and multidisciplinary care, with these areas continuing to evolve in response to the changing therapeutic landscape [29, 30]. Altogether, these advances have reshaped the clinical landscape by slowing, stabilising, or partially reversing disease progression. At the same time, their impact has

made increasingly clear that SMA is a multisystemic condition, and a wider range of traditional and emerging patient needs, spanning biomedical, rehabilitative, and systemic areas, have become more prominent.

Through multilingual surveys and structured workshops, the PSP produced a democratically endorsed priority list of research areas which reflect how fundamental, clinical and care-related research priorities might miss critical aspects of daily life with SMA. By translating these community-defined uncertainties into a “Top 10” list of priority topics, the PSP did not merely gather opinions but created new knowledge about what matters most. By elevating priorities that extend beyond SMN protein restoration and motor function, such as fatigue, metabolic dysfunction, and assistive technology, the PSP highlights pre-clinical and clinical research needs that remain underexplored and underfunded [31–33]. These gaps underscore the urgency of a more holistic, community-informed research agenda that addresses the multisystemic nature of SMA and the real-world challenges faced by those affected [13, 24].

The process validated the epistemic authority of patients, caregivers, and frontline professionals, underscoring the value of lived experience and challenging traditional hierarchies in scientific inquiry [23, 34]. In doing so, it anchors research in the everyday realities of those affected and aligning with broader international calls for “democratising” science [25, 35, 36].

From priorities to research clusters: toward transdisciplinary innovation

To move from questions to action, the PSP convened a “Towards Solutions” workshop that mapped the ranked priorities into eight thematic clusters (Table 2) spanning across disciplines including biomedical science, rehabilitation, and bioengineering.

This framing exercise exemplifies transdisciplinary innovation as a growing imperative in rare disease research, where complex conditions often intersect multiple biological systems and fields [37]. Rather than tackling priorities in isolation, it merges them into interconnected challenges that demand integrated expertise. Even if partially addressed, these clusters have not yet delivered solution(s) that are meaningful to patients, a pattern observed across participatory research efforts across complex conditions [38].

This initiative highlights the importance of co-designed, patient-relevant translational pathways and provides a concrete opportunity to advance them [39].

Moreover, Phase III’s dissemination campaign functioned as a dynamic feedback loop, keeping the community informed, inviting reflection on the current and future needs and how best to address them, and encouraging uptake. It reframed communication not as one-way dissemination but as an ongoing participatory mechanism for aligning research with evolving community needs, an approach increasingly recognised in health research as key to closing the gap between evidence generation and real-world application [40, 41].

Strategic implications and global relevance

The SMA PSP offers insights not only for SMA, but also for the governance of rare disease research more broadly. By embedding lived experience into the architecture of priority setting and implementation, the initiative contributes to a growing international movement toward participatory research governance [42, 43].

Globally, major initiatives have advanced the integration of PPI across the research cycle, providing structured frameworks, training platforms, reimbursement models, and evaluation tools that recognise patients as co-designers and decision-makers, not just participants. In the UK, the NIHR INVOLVE (now part of the NIHR Centre for Engagement and Dissemination) and the James Lind Alliance (JLA) have laid the groundwork for embedding patient voice in research governance, with the JLA approach, applied in over 100 disease areas, widely regarded as a gold standard for stakeholder-driven priority setting [3, 44]. In Canada, the Strategy for Patient-Oriented Research (SPOR) embeds patient engagement into health research systems, while PaCER uniquely trains patients to conduct qualitative research grounded in lived experience [45, 46]. At the European level, initiatives such as EUPATI and PARADIGM have strengthened patient roles in therapeutic innovation, ethics committees, and

regulatory processes [47, 48]. The newly launched European Rare Disease Research Alliance (ERDERA) supports patient-centred research capacity across countries [49]. SMA Europe itself exemplifies sustained commitment to this approach, advancing a portfolio of participatory research activities, including co-designed clinical trial tools and treatment decision surveys, the real-world data platform OdySMA [50] and the SMAcademy [51] training initiative, and the mapping of experiences, expectations and needs of people living with SMA through the European Patient Experience Survey (EUPESMA) series [52, 53], demonstrating how patient organisations can embed lived experience throughout the research continuum [54, 55].

What distinguishes the SMA PSP is not only its multinational and multilingual execution but its ability to bridge diverse cultural and healthcare contexts within a rare disease community. Conducted across 22 countries, the initiative navigated different perceptions of disease and disability, variations in care infrastructure, and unequal access to treatments. In countries where access to therapies and early diagnosis remains limited, these remain the most urgent priorities, and ensuring access worldwide is a core commitment of the global SMA community. Despite this complexity, this initiative succeeded in looking forward and identifying a set of shared, community-defined priorities, demonstrating that it is possible to transcend national and systemic differences to articulate the common uncertainties based on experiences and unmet needs of people living with SMA. Even in conditions with approved therapies, meaningful gaps persist. Addressing these requires both broader research agendas and stronger participatory infrastructures capable of navigating and uniting such diversity.

As institutions and funders increasingly commit to co-produced, impact-driven research, the SMA PSP demonstrates that the JLA Priority Setting Partnership is a replicable model. It affirms that participatory science is not only ethically desirable but strategically necessary to advance equity, relevance, and innovation, particularly in underrepresented fields like rare diseases.

Limitations and conditions for future impact

This initiative had several limitations. Despite multilingual materials, subtle meanings may be diluted or altered during translation, language barriers and uneven geographic representation may have shaped whose voices were most heard. Also, SMA presents with a continuum of severity, and some phenotypes may have been underrepresented, resulting in research questions that reflect the priorities of certain individuals living with SMA more than others. In addition, the voices of patients and caregivers outnumbered those of healthcare professionals, potentially shaping the balance of priorities identified. While the approach for Phase I focused on identifying the “Top 10” priorities, further important questions were also identified (see Supplementary Table 1, Additional File), potentially leaving some key thematic research clusters insufficiently highlighted, especially in a highly dynamic environment as in SMA.

Moreover, this process marks a beginning, not an endpoint. The “Top 10” priorities and their thematic clustering offer a strong foundation, but new needs are likely to emerge as the natural history of SMA evolves in response to new treatments. As children and adults live longer with improved motor function, different challenges will become more salient. Ongoing, structured mechanisms to revisit and update priorities will be essential.

Finally, while this initiative has demonstrated the value and potential of participatory, transdisciplinary research design in theory and practice, translating this into mainstream research systems will require a collaborative approach among patient advocacy organisations and the research community. Realising the full promise of community-driven research will depend on continued dialogues and advocacy, from patient organisations, funders, and researchers alike, to build systems that embed lived experience not as an add-on, but as a core component of scientific governance.

Conclusions

The SMA PSP offers a compelling blueprint for reimagining SMA research from the ground up. By placing lived experience at the centre of research governance, the initiative redefined how priorities are set, knowledge is co-produced, and impact is pursued. By using a multinational and multilingual approach, SMA PSP was able to bridge diverse cultural, and healthcare contexts within a rare disease community. The resulting “Top 10” priorities for SMA reflect a community-informed, multidimensional view of SMA, encompassing biomedical, medical, rehabilitative, and technological needs that often fall outside the scope of conventional research.

Crucially, the initiative did not stop at identifying research gaps. By clustering priorities into thematic clusters, fostering transdisciplinary dialogue, and launching a participatory dissemination campaign, SMA Europe showed that patient organisations can not only define research needs, but also influence the structures through which those needs are addressed. For example, SMA Europe has been funding research projects through its “Call for Research Proposals” initiative, taking into account the community priorities identified through the SMA PSP. Likewise, the scientific programme of SMA Europe’s “International Scientific Congress on Spinal Muscular Atrophy” is shaped with consideration of the research priorities identified by the community.

The findings send a clear message: although disease-modifying treatments have transformed SMA care, the condition is far from resolved. Major unmet needs persist spanning diverse domains, including fatigue, metabolic management, systemic complications, and assistive technology. The needs of people living with SMA are not only to extend life, but to enhance its quality. These insights highlight the need both for continued research funding and for research priorities to take into account the priorities expressed by people living with SMA.

For researchers, the community-identified research priorities provide a constructive framework for collaboration: an opportunity to consider how ongoing and future work might align with the areas the community has identified as most pressing. Engaging with such participatory processes enriches both the scientific endeavour and its societal relevance.

For funders, the outcomes of this project offers a clear, community-driven rationale for prioritising investments, helping ensure that resources are directed toward research with the greatest potential to address real-world needs.

Embedding participatory governance is therefore a pathway toward more community-relevant and impacting, future-ready research, within SMA and across the wider rare disease field.

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Declarations

Ethical approval Not required - research priority setting exercises.

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