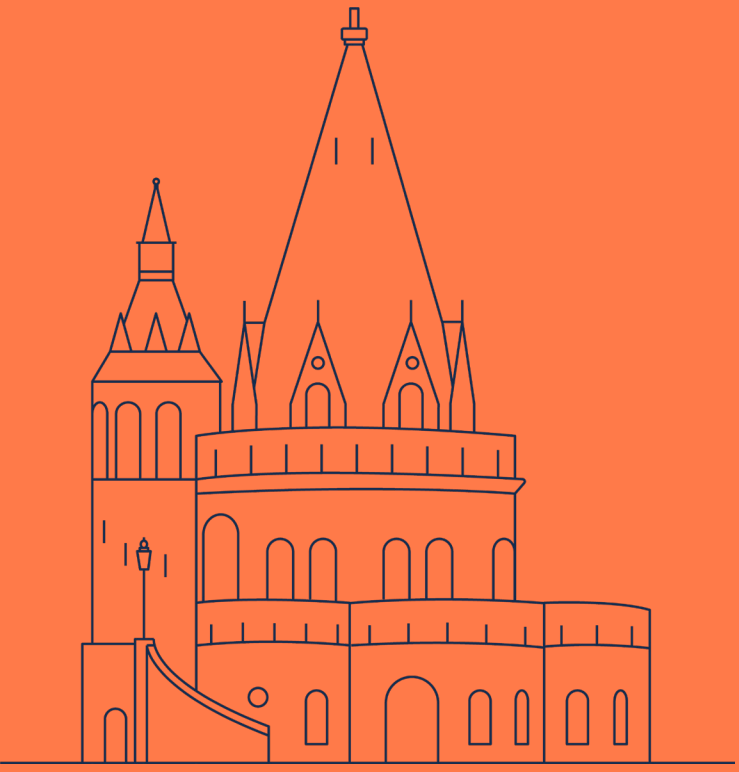
WORKSHOPS



WS1 - Organised by argenx & Karen Chen

Assessing fatigability in Spinal Muscular Atrophy: Exploring the contribution of neuromuscular junction dysfunction

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Although 3 survival motor neuron (SMN)-targeting disease-modifying therapies (DMTs) have been approved for the treatment of spinal muscular atrophy (SMA), a significant unmet medical need persists. A substantial proportion of patients are treated with ≥2 of the approved DMTs, yet many do not achieve optimal responses to these treatments. This highlights the limitations of current treatment options for patients with SMA and emphasizes the need for additional treatment options that better address disease symptoms.

Fatigability, a key symptom that significantly impacts the quality of life of patients with SMA, is emerging as a target of interest for therapeutic intervention; however, despite advances in SMN-targeting therapies, the effect on fatigability remains unclear.

Nonclinical studies have demonstrated that neuromuscular junction (NMJ) abnormalities are among the earliest and most pronounced pathological features in murine models of SMA. Some of these NMJ defects persist despite treatment with SMN-upregulating therapies. Emerging evidence indicates that modulation of the agrin–LRP4–MuSK–DOK7 signaling axis may offer a promising therapeutic strategy to ameliorate NMJ dysfunction in SMA. In addition, clinical studies have shown that NMJ functional abnormalities contribute to SMA disease pathophysiology. NMJ transmission deficits, evidenced by decremental compound muscle action potential amplitudes, persist in individuals despite treatment with DMTs. Off-label use of agents such as salbutamol, amifampridine (3,4-DAP), and pyridostigmine further supports the therapeutic potential of targeting NMJ integrity in SMA.

This workshop aims to present the role of the NMJ and to explore assessment strategies for evaluating fatigability in ambulatory and nonambulatory patients with SMA. In addition, the workshop will discuss sensitive outcome measures, including digital sensor technologies and the development of new scales to capture the intensity and duration of activity. These measures may enhance the detection of subtle changes in fatigability not captured with other assessments and support reported shifts in perceived fatigability, which may be used to measure potential treatment effects in investigational trials.

Targeting NMJ dysfunction may serve as a complementary approach to SMN-enhancing therapies, offering the potential to alleviate residual symptoms such as fatigability and muscle weakness that persist despite treatment with DMTs, thus highlighting the need for multimodal therapeutic strategies.

Acknowledgments

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WS 2 - Organised by Novartis

Beyond boundaries: Elevating SMA care for the future

This impactful and dynamic workshop is designed to explore innovative strategies and collaborative approaches to optimize care for individuals with Spinal Muscular Atrophy (SMA) Types 2 and 3. Through a moderated panel and interactive live polling session, participants will engage in meaningful dialogue, share experiences, and develop strategies to address the unmet needs in SMA care.

Workshop objectives:

- Identify and address care gaps: Explore the current gaps and challenges in SMA care, particularly for Types 2 and 3, and discuss innovative strategies to enhance patient management and support systems.
- Foster multidisciplinary collaboration: Encourage collaboration among healthcare professionals, patients, and caregivers to develop a holistic approach to SMA care that integrates medical, rehabilitative, and psychosocial support.
- Empower and advocate for change: Empower participants to advocate for improved SMA care standards by sharing insights and experiences and inspire healthcare policies and practices for better patient access and outcomes.

WS 3 - Organised by Kathryn Swoboda and Renske Wadman

Old signals, new insights: Electrophysiology as a biomarker in SMA

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In this workshop we dive into the motor unit pool, exploring the utility of electrophysiological measures as predictive biomarkers of (future) motor function in infants, children and adults with SMA.

Electrophysiologic studies (nerve conduction studies and electromyography) are potential important biomarkers to assess (pre)clinical disease activity in the motor unit, including axons, motor neurons and neuromuscular junction function. At this moment their use is limited in research and clinical care in SMA, but with the introduction of different therapeutic strategies at different disease stages it is even more important to find biomarkers that reflect disease activity and stage.

We will discuss the different opportunities and limitations of electrophysiologic studies and address the different techniques (standard nerve conduction studies, CMAP scan/ MUNIX, repetitive nerve stimulation, electromyography) and proposed protocols (which nerves, what timing, in which patient). All techniques will be explained by means of clinical cases supported with data from the literature on SMA. In addition, we will discuss the opportunities for its use in future research and clinical setting.
At the end of the workshop, the audience will know what the different techniques withhold, how to use then (and when not to) and what the future perspectives of electrophysiology in SMA are.

WS 4 - Organised by José Longatto

MAPS - Mobility, Activity, Play, and Sports
J. Longatto

Great Ormond Street Hospital

The advances in drug and gene treatments for Spinal Muscular Atrophy (SMA) have significantly increased survival, improved physical function, and enhanced condition stability. The introduction of newborn screening has further reduced the impact of severe disability in many infants and children. Despite medical progress, physiotherapy remains a key part of long-term management for many with SMA. For those who can afford it—or where health or insurance schemes allow—infants, children, and young people may access regular professional input, mobility aids, equipment, and orthotics.

Many parents and children engage in stretching, exercise, and splinting routines, though many cannot. Physiotherapy should not stop with the therapist: it must become part of daily life—not just a routine, but an integrated, natural element. Framing physiotherapy as “work” or “routine” can cause guilt, resistance, stress, and fatigue for both parents and children.

Our workshop is designed for therapists but is equally relevant to parents, teachers, and carers. It introduces a new approach to home physiotherapy: the MAPS System—Mobility, Activity, Play, and Sports. This model is family-friendly, reducing the burden of adding another routine to busy lives. It promotes movement through enjoyable activities that fit into family and school life.

To help families use MAPS, we follow four guiding principles:

- Motivation: Embedding mobility and activity into things children already enjoy—football, dancing, or play—makes participation fun and purposeful. Motivation turns exercise from a chore into a natural part of life.
- Achievability: By adapting physiotherapy into manageable tasks, MAPS ensures success for every child. Adaptive strategies let children participate at their level, building confidence without pressure.
- Participation: Physiotherapy isn’t just for the child. MAPS involves siblings, peers, and classmates so the child with SMA isn’t isolated. Framing activities as shared games makes therapy inclusive and social.
- Support: Children thrive with consistent encouragement from professionals, parents, carers, and extended family. MAPS works across school and care settings, reducing parent burden. Importantly, it does not require costly equipment.

MAPS is achievable, functional, and fun. It has proven effective with parents, children, and therapists in the UK, Portugal, Latvia, and Lithuania.

Through MAPS—Mobility, Activity, Play, and Sports—physiotherapy at home is reframed: not as a chore, but as a sustainable, enjoyable part of everyday life.

WS 5 - Organised by Esther Veldhoen and Lisa Edel

Respiratory care in Spinal Muscular Atrophy

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Respiratory muscle weakness in SMA causes reduced cough strength with recurrent respiratory tract infections and respiratory failure. Despite introduction of disease modifying therapies, respiratory symptoms are still present in many patients. For this reason, it is increasingly important to optimize supportive care to postpone or avoid respiratory failure. Assessment of respiratory function with early detection of deterioration of respiratory muscle strength is important to guide treatment.

The learning objectives of this workshop are:

- Physiology of respiratory symptoms in SMA
- Overview and (dis)advantages of different assessment tools of respiratory function used in clinical and research setting, such as lung function, tests of respiratory muscle strength, imaging studies, clinical scores.
- Evidence and use of different supportive respiratory treatments, such as airway clearance techniques and respiratory muscle training.

This will involve theoretical background, followed by practical hands-on sessions on respiratory muscle training and airway clearance, and an interactive discussion on clinical assessment tools to perform follow up on respiratory muscle weakness.
This workshop is primarily intended for clinicians (neurologists, physiotherapists, nurses, rehabilitation specialists etc). We specifically would like to invite patients to attend, as their expertise and input is useful for clinicians.

WS 6 - Organised by Simona Bertoli

Nutritional management in SMA in the new treatment era: From fundamental science to clinical practice

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Intended Audience: A multidisciplinary audience of clinicians, researchers, and patient advocates involved in SMA care.

The introduction of innovative therapies is profoundly changing the clinical phenotype of SMA, making accurate nutritional management a cornerstone for optimizing outcomes. Traditional tools like BMI are misleading in this population due to profound alterations in body composition (increased fat mass, reduced fat-free mass). Our multicenter research in Italy and the UK has addressed this by creating and validating a comprehensive assessment protocol with SMA-specific tools, including a standardized anthropometric manual, predictive equations for Resting Energy Expenditure (REE) and Fat Mass (FM%), and the first disease-specific growth charts. This interactive workshop aims to translate these research findings into daily clinical practice.

Objectives:
Present the rationale for a standardized nutritional assessment in SMA.
Train attendees on an anthropometric protocol adapted for neuromuscular disabilities.
Introduce new SMA-specific tools (predictive equations, growth charts) for accurate monitoring.
Discuss the interpretation of body composition data and its correlation with motor function.
Facilitate an interactive discussion on implementing these protocols in diverse clinical settings.

The New Clinical Challenges (10 mins): In this opening session, Prof. Simona Bertoli will discuss how new therapies reshape SMA nutritional profiles, highlighting the limitations of traditional metrics and introducing the concept of “quality of weight”, emphasizing a standardized, disease-specific assessment.
Metabolic Dysregulation in SMA Models (15 mins): This session will provide the crucial pre-clinical context for the clinical findings. Prof. Melissa Bowerman will provide the preclinical context from animal models on altered energy metabolism, brown fat activation, and the potential of targeted dietary interventions.
Impact of New Therapies in the Clinic (15 mins): Prof. Anette Hjartåker will present real-world data on how treatments affect nutritional status and dietary needs in SMA types II & III, reinforcing the need for careful and tailored nutritional support.
Practical Demonstration (25 mins): Drs. Ramona De Amicis, Silvia Gallosti, and Melis Sevim will lead a demonstration of key anthropometric measurements, explaining how to overcome difficulties like joint contractures and how to use the online calculators.

- Bulbar Dysfunction & Nutrition (10 mins): Prof. Giovanni Baranello will link neurological progression to nutritional deficits, focusing on how bulbar muscle weakness (dysphagia) is a primary driver of malnutrition.

- Round Table Discussion (15 mins): All speakers will moderate an interactive Q&A on practical implementation challenges and fostering a collaborative network for standardized nutritional care.

WS 7 - Organised by Biogen and Eduardo Tizzano

Every motor neuron matters – Biomarkers and treatment choice in SMA

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With three approved therapies, and additional treatment modalities on the horizon, SMA patients and caregivers are benefiting from increased treatment choice. However, whilst each therapy is supported by a strong evidence package, what these data mean for setting and monitoring individual treatment expectations and responses, is less clear.

- What is my capacity for improvement?
- How do I know if my treatment response is optimal?

Biomarkers such as neurofilaments, electrophysiology measures, and muscle imaging provide real-time information on the disease evolution and response to treatments at the motor unit level, offering novel perspectives on these questions.
We invite PAGs, clinicians and basic researchers with an interest in patient empowerment to join this workshop. Together we will explore how introducing a common "language" to describe the health of an individual's motor units, may support informed shared treatment decisions and better follow-up.

WS 8 - Organised by FundAME

Contractures in SMA: From scientific insights to therapeutic targets

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Contractures are a common and disabling musculoskeletal complication in SMA, reducing functional capacity, causing pain, and significantly impacting the quality of life of individuals living with the disease. Although disease-modifying therapies (DMTs) have transformed the course of SMA, contracture management remains a largely unmet clinical need and continues to rely on pre-DMT standards of care. Consequently, the management of contractures and muscle imbalance—unlike muscle function—has changed little with the advent of DMTs and still follows pre-DMT care practices. Despite the changes in the disease landscape, long term outcomes are often unsatisfactory for those with chronic SMA frequently persisting or worsening over time, highlighting the urgent need for novel therapeutic approaches. A central question—and the driving theme of this workshop—is whether contractures in SMA are solely secondary to muscle weakness, imbalance, and immobility, or whether additional SMA-specific mechanisms contribute. For instance, could SMN deficiency directly alter muscle or connective tissue properties, predisposing patients to contractures in ways distinct from other neuromuscular disorders? Preclinical evidence suggests that SMN deficiency induces intrinsic muscle abnormalities (non-neuropathic myopathy). Addressing these questions is essential for the development of novel, muscle- or tendon-targeted strategies that complement existing therapies and alleviate the burden of contractures. FundAME, SMA Europe and SMA Foundation recognize the urgent need to prioritize the understanding and management of contractures within the SMA research agenda. This workshop aims to promote a rigorous, multidisciplinary dialogue among researchers, clinicians, and patients.

Objective:

- To integrate current knowledge in basic research on muscle involvement in SMA, strategies used for its clinical management and identified patients' unmet needs, as well as therapeutic options.
- To identify new areas of research needed to improve the approach and treatment of contractures and, ultimately, the development of new approaches and specific drugs.

Acknowledgment

This workshop is offered by Roche.

WS 9 - Organised by Scholar Rock

Addressing unmet needs in SMA care through multidisciplinary collaboration and education

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Despite transformative progress, the SMA journey is not complete. Spinal muscular atrophy (SMA) has entered a new era, with survival motor neuron (SMN)–targeted treatments transforming outcomes and underscoring the importance of multidisciplinary care. Yet despite these advances, individuals with SMA and their caregivers continue to face persistent unmet needs related to muscle weakness. This ongoing weakness leads to deterioration in motor function and can negatively impact daily life and independence over time. As new therapeutic approaches emerge on the horizon, healthcare professionals (HCPs) and patient advocacy groups (PAGs) increasingly recognize critical gaps in medical education and the resources needed to support informed, collaborative decision-making in SMA care.

This interactive workshop will bring together HCPs and PAGs from the SMA community to discuss these challenges. The key topics will include: (1) the evolving unmet needs in the current and future SMA landscape; (2) the role of multidisciplinary care in optimizing SMA care; and (3) the opportunities to enhance medical education and resource development for HCPs and PAGs to support and advance SMA care.

Through case discussions, audience participation, and cross-stakeholder dialogue, the workshop will empower participants will co-create actionable insights to shape future educational priorities and strengthen support for individuals living with SMA.
This workshop is organized and sponsored by Scholar Rock, Inc. by way of financial contribution and provision of educational materials.

WS 10 - Organised by Chiara Mastella

The role of early parental empowerment in management of emergencies and urgencies at home in pharmacologically treated SMA type 1

C. Mastella, M. Foà, M. Negri, E. Pagliaccia, M. Rauso, M. Antonella Costantino

Sapre- UONPIA Fondazione IRCCS Ca Granda Ospedale Maggiore Policlinico

Spinal Muscular Atrophy Type1 (SMA1), a severe neuromuscular disorder, has historically presented significant challenges, with respiratory complications being the leading cause of morbidity and hospitalizations, particularly in the context of managing home-based emergencies. Prior to the advent of disease-modifying therapies (Nusinersen, Onasemnogene Abeparvovec and Risdiplam), care was predominantly palliative. While these new treatments have profoundly improved clinical outcomes and prognosis, the complexity of home care for SMA1 remains, and the risk of respiratory crises persists. For this reason, respiratory physiotherapy protocols and the use of tools and devices that promote chest expansion, peripheral pulmonary ventilation, and airway clearance techniques, remain fundamental in daily care.

In this workshop we would like to share our long-standing multidisciplinary experience aimed at training and empowering families, as well as all the healthcare workers who support families, on managing respiratory emergencies at home. Education and psychological support for caregivers are fundamental parts of comprehensive disease management.

Our program begins at diagnosis and evolves alongside the patient's therapeutic journey. We identified six recurrent emergency scenarios: acute airway obstruction from secretions, respiratory decline during infections, ventilator malfunction, aspiration risk from vomiting, stress-induced respiratory distress and summer dehydration. All caregivers are required to complete a Pediatric Basic Life Support course.

Initially, our focus was on palliative care, teaching families to use basic resources like chest physiotherapy, suctioning, and Ambu bags to maintain a very basic quality of life. With pharmacological advancements, the emphasis has shifted to new needs and better quality of life.

Empowered families show increased competence, confidence, and proactive care in their child's care. Our results suggest that comprehensive, evolving training, and psychological support are integral to modern SMA1 management, leading to improved outcomes, reduced hospitalizations, and enhanced family well-being.

The advent of disease-modifying therapies has profoundly transformed the prognosis of SMA1, shifting the focus from predominantly palliative care to the long-term management of a chronic condition. Our multidisciplinary training program, which integrates psychological support with ongoing practical education, has demonstrated its effectiveness in reducing hospitalizations and boosting families' competence and confidence. As the clinical landscape continues to evolve, empowering caregivers and healthcare staff is crucial for optimizing long-term outcomes and fostering a proactive approach to care.

WS 11 - Organised by Giovanni Baranello

SMA and neurodevelopmental disorders: A call for harmonized strategies

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The natural history of Spinal Muscular Atrophy (SMA) has significantly changed with the advent of SMN-enhancing therapies, leading to improved motor outcomes and prolonged survival, particularly in infants diagnosed through newborn screening. However, emerging evidence reveals that children with severe SMA—especially those with lower SMN2 copy numbers—may experience a range of neurodevelopmental disorders, including speech and language delays, autism spectrum disorder, global developmental delay, and intellectual disability. These findings challenge the traditional view of SMA as a purely motor neuron disease and underscore the need to understand its broader impact on brain development and overall quality of life.

Despite early therapeutic intervention, the long-term neurodevelopmental trajectories of these children remain uncertain. This workshop aims to address this critical gap by fostering a multidisciplinary dialogue among clinicians, researchers, patient advocates, and other stakeholders.

Key objectives include:

- Exploring the neurobiological basis of how insufficient SMN protein levels in the developing brain may contribute to neurodevelopmental impairments in children with severe SMA.
- Identifying early markers and risk factors to recognize children most vulnerable to neurodevelopmental disorders, enabling timely and targeted support.
- Discussing strategies for early intervention, family support, and developmental stimulation to mitigate the impact of these disorders and improve quality of life.
- Engaging the patient community and stakeholders to build a collaborative framework that supports families, informs healthcare systems, and promotes harmonized approaches to care and research.

Intended audience: clinicians, researchers, patients' representatives and all relevant stakeholders that are committed to improve SMA care. By bringing together diverse perspectives, this workshop seeks to catalyse a unified response to the emerging neurodevelopmental phenotype in SMA, ensuring that affected children receive comprehensive, anticipatory care that addresses not only motor but also the overall development.

WS 12 - Organised by Ewout Groen

Bench to bedside and back - Fundamental discoveries in connection with patient journeys

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This interactive workshop explores the vital connections between patient experiences, and clinical and fundamental research through compelling case studies. Patient journeys, told by the patients themselves, will be supported by clinical and scientific insights from our research team.

The recent discoveries of effective treatments for patients with SMA are based on many years of fundamental research. However, SMA models differ in many ways from humans and often do not reflect the disease course of patients. For decades, researchers suspected limited correlation between phenotypic pathology and symptoms in severe animal models and patients. As we explore symptoms beyond motor neuron dysfunction, striking resemblances between severe animal models and patients are emerging. Depletion of SMN protein leads to structural and functional changes in kidney and pancreatic cells in severe animal models. Correspondingly, clinical changes in renal function and glucose metabolism are now recognized as significant features in adults with SMA. In addition, the discovery of structural NMJ changes in mouse models was followed by recognition of corresponding fatigability and functional changes of the NMJ in adults with SMA, leading to new treatment possibilities that improve NMJ function and reduce fatigability.

Adults with SMA represent the largest patient population and face unique challenges in managing systemic complications that can significantly impact their daily functioning and quality of life. During this interactive session, patients will share their lived experiences with altered glucose metabolism, renal function changes, and NMJ dysfunction. These personal narratives will be contextualized with insights into the fundamental scientific discoveries underlying renal function, glucose metabolism, and NMJ dysfunction research, and how these discoveries have influenced modern clinical care.

Personal stories from adults living with SMA will lead this workshop. By bringing together our varying backgrounds we aim to illustrate the importance of recognizing the full spectrum of SMA-related symptoms and the critical role that patient experiences play in advancing both our understanding of disease mechanisms and the development of practical solutions for managing everyday challenges and maintaining participation in society.

Objectives:

- Understand the bidirectional relationship between animal models and patient experiences in SMA research
- Recognize non-motor symptoms as integral components of SMA pathophysiology
- Appreciate how patient narratives can inform and validate preclinical research findings
- Explore emerging therapeutic approaches for systemic SMA complications.