



4th

Scientific International Congress on
Spinal Muscular Atrophy

GHENT

14th — 16th March 2024



PROGRAMME

ORGANISATION

medicongress

conference & association management

Noorwegenstraat 49, 9940 Evergem (Belgium)
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SMA2024@medicongress.com



SMA Europe takes great pride in inviting you to the 4th International Scientific Congress on Spinal Muscular Atrophy (SMA), from 14th to 16th March 2024 in Ghent, Belgium. Our Congress is the largest scientific congress dedicated to SMA and brings together scientists and young researchers as well as clinicians and other health-care professionals from all over the world.

SMA Europe is a non-profit umbrella organisation of SMA patient organisations from across Europe. We work together to create a better world for all those living with SMA. One of our core activities is to foster patient-relevant research in the field of SMA, to communicate the value generated from research, and, consequently, to ensure future support for research within our community.

The goal of our scientific congress is to bring together an international and multidisciplinary group of scientists and health-care professionals. We provide a venue to present and exchange breakthrough ideas relating to SMA, especially also in light of the patient relevance of their findings, and to cement existing and stimulate new collaborations.

Equally, our congress is a platform for talented young researchers. Together with experienced scientists, health-care professionals, and patient experts, they will discuss, debate and dissect significant new developments and advancements related to SMA. Moreover, they will share their visions for the future of SMA research by providing an opportunity to exchange scientific evidence and clinical experiences. Therefore, we trust that the conference will lead to ever-improved treatment and care for patients living with SMA.

The International Scientific Congress on Spinal Muscular Atrophy is a unique occasion to meet face-to-face with colleagues who are as passionate about advancing the field of SMA as you are.

All together. One Goal.

SMA Europe very much looks forward to welcoming you in Ghent!



Nicole Gusset
SMA Europe CEO and President

Scientific Congress Organisation

SCIENTIFIC COMMITTEE



Chair: Thomas Gillingwater

Professor of Anatomy at the University of Edinburgh and Editor in Chief of the Journal of Anatomy



Vice-Chair: Stefania Corti

Neurologist, Associate Professor of Neurology and Principal Investigator of Neural Stem Cell Lab at the University of Milan in Italy



Local host: Liesbeth De Waele

Paediatric Neurologist and Assistant Professor at the Faculty of Medicine, UZ Leuven NMRC for Children, Belgium



Nicolas Deconinck

Head of paediatric neurology department HUDERF, Head of the research unit of UZ Ghent NMRC



Melissa Bowerman

Senior Lecturer in Neuromuscular & Skeletal Disorders, School of Medicine, Keele University, UK



Claudio Bruno

Head, Center of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini, Genova, and Contract Professor in Paediatrics at the School of Medicine, University of Genova, Italy



Peter Claus

Scientific Director, CEO and Co-Founder of SMATHERIA, a non-profit biomedical research institute dedicated to spinal muscular atrophy (SMA) and other neuromuscular diseases preclinical and translational research, Hannover, Germany



Richard Finkel

Paediatric Neurologist and Director of the Center for Experimental Neurotherapeutics at St. Jude Children's Research Hospital in Memphis, Tennessee, USA



Ewout Groen

Senior Researcher, SMA Center of Expertise, UMC Utrecht Brain Center, Department of Neurology and Neurosurgery, University Medical Center Utrecht, The Netherlands

Scientific Congress Organisation

SCIENTIFIC COMMITTEE



Ulrika Kreicberg

Ulrika Kreicbergs, Professor of Palliative Care for Children and Young People at the Great Ormond Street Institute of Child Health, University College London, UK



Cécile Martinat

Head of INSERM/UEVE UMR 861 in I-STEM, French institute dedicated to the use of human pluripotent stem cells to study and treat monogenic diseases, Paris, France



Christian Simon

Group Leader, Carl Ludwig Institute for Physiology, Leipzig University, Germany



Charlotte Sumner

Professor of neurology at Johns Hopkins School of Medicine, USA



Ludo van der Pol

Neurologist and Professor of Neurology, head of the Netherlands SMA center at the University Medical Center Utrecht (UMCU) in The Netherlands

SCIENTIFIC CONTENT

The conference programme consists of seven sessions, workshops and poster sessions which will showcase the latest developments in the field, provide a forum for discussions and networking and highlight the challenges and future trends in SMA-related scientific disciplines and applied technologies.

For each session, researchers selected via abstract submission will have the opportunity to give a 15-minute presentation in the plenary programme.

WEDNESDAY 13th MARCH 2024 • Pre-Congress Day

Global SMA Advocacy Event

BAEKELAND ROOMS 1-3

09:00 - 12:30

Morning activities

10:30 - 11:00

COFFEE BREAK

11:00 - 12:30

Morning activities

12:30 - 13:30

LUNCH

13:30 - 14:30

Vernissage of SMA Awareness Campaign 2023

14:30 - 16:00

Afternoon activities

16:00 - 16:30

COFFEE BREAK

16.30 - 18.30

Afternoon activities

18.30 - 21.00

DINNER

ECR Event

GUISLAIN 1

13:30 - 14:30

Vernissage of SMA Awareness Campaign 2023

14:30 - 16:00

Afternoon activities

16:00 - 16:30

COFFEE BREAK

16:00 - 18:00

PRE-REGISTRATION

ICC

16.30 - 18.00

Afternoon activities

18.00 - 21.00

PRE-CONGRESS WELCOME RECEPTION BY INVITATION ONLY

THURSDAY 14th MARCH 2024

08:30 - 09:30	REGISTRATION & WELCOME COFFEE IN THE EXHIBITION AREA	
09:30 - 09:45	Welcome SMA Europe Nicole Gusset - SMA Europe CEO and President	AUDITORIUM
09:45 - 10:00	Welcome to Belgium Hilde Crevits, Deputy Minister-President of the Government of Flanders, Flemish Minister for Welfare, Public Health and Family Karel Everaert, Head of Clinics Functional Urology, UZ Ghent	
10:00 - 10:45	Is SMA cured? The future of SMA research Panel discussion with Yasemin Erbas, Cécile Martinat and Giovanni Baranello Moderator: Ankita Batla	
10:45 - 10:55	Opening of scientific & clinical programme Thomas Gillingwater, Chair of Scientific Committee	
11:00 - 11:30	COFFEE BREAK AT THE EXHIBITION AREA	
11:30 - 13:00	Session 1 - Rehabilitation in SMA - Motor neuron and spinal cord pathology AUDITORIUM Chair: Farnaz Nickpour, University of Liverpool, UK	
	01 Facilitating neuromuscular junction recovery after treatment in mouse models of spinal muscular atrophy Inga Partlova, University of Edinburgh, UK	
	02 Respiratory muscle training in patients with spinal muscular atrophy - Results of a randomized controlled trial Kim Kant-Smits, Wilhelmina Children's Hospital, University Medical Center Utrecht, The Netherlands	
	03 Bone health in adult spinal muscular atrophy (SMA) patients: Is it a forgotten risk? Channa Hewamadduma, University of Sheffield, UK	
	04 Scoliosis progression in spinal muscular atrophy type II: A comparative study between treated and untreated patients Giorgia Coratti, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy	
13:00 - 14:15	LUNCH BREAK AT THE EXHIBITION AREA	
13.10 - 14.10	Industry Sponsored Session	T. VAN RYSSELBERGHE

THURSDAY 14th MARCH 2024

14:15 - 14:30	Flash poster presentations, Session 1	AUDITORIUM
	P141 Pain in spinal muscular atrophy patients Kursat Bora Carman, Eskisehir Osmangazi University, Turkey	
	P72 Criteria for identification and accurate quantification of spinal motor neurons in healthy and disease mouse models Leonie Sowoidnich, Carl-Ludwig-Institute of Physiology, Germany	
	P13 Longitudinal changes in compound muscle action potential (CMAP) and their association with motor function in children with infantile-onset SMA in ENDEAR/SHINE Michelle Farrar, UNSW Sydney, Sydney, Australia	
	P50 Dopaminergic system role in a C. elegans model of SMA Giada Onorato, Institute of Biosciences and BioResources (IBBR), CNR, Italy	
	P121 A Combined Examination of Novel Rapid Bedside Plasma-SMN Analysis and Muscle Ultrasound May Help to Early Screen & Monitor Children with Spinal Muscular Atrophy in Clinical Setting Dian K. Nurputra, Universitas Gadjah Mada, Indonesia	
	P12 FUS protein expression and distribution in the myopathology of 5q-associated spinal muscular atrophy type 3 Heike Kölbl, Universitätsmedizin Essen Germany	
14:30 - 16:00	Session 2 - Optimising gene-directed therapeutics Chair: Rafael Yáñez Muñoz, Royal Holloway, University of London, UK	AUDITORIUM
	O5 RNA perturbation in Cerebrospinal Fluid of Spinal Muscular Atrophy Patients treated by Nusinersen Stella Gagliardi, IRCCS Mondino Foundation, Pavia, Italy	
	O6 Direct evidence of deletion and gene conversion as origin of SMA alleles revealed by long-read sequencing Maria Zwartkruis, University Medical Center Utrecht, The Netherlands	
	O7 Characterising the effect of AAV9 mediated gene therapy on the epigenetic and transcriptional stability of SMA mice Piera Smeriglio, Institut de Myologie, Paris, France	
	O8 Enhancing In Vitro SMN Protein Expression and Cell Viability through Xeno-Nucleic Acid-Based ASOs in Spinal Muscular Atrophy Cihan Tastan, Uskudar University, Istanbul, Turkey	
16.00 - 16.30	COFFEE BREAK IN THE EXHIBITION AREA	
16.00 - 17.30	Poster session 1	MINNEPLEIN
17:30 - 18:30	Parallel workshops Towards a Research Agenda for SMA Tom Gillingwater and Nicole Gusset	T. VAN RYSSELBERGHE
	Stem cell models Cécile Martinat	H. VAN EYCK
18:45	WELCOME RECEPTION IN THE EXHIBITION AREA	

FRIDAY 15th MARCH 2024

08:15 - 09:30	REGISTRATION & WELCOME COFFEE IN THE EXHIBITION AREA	
08:20 - 09:20	Industry Sponsored Session	T. VAN RYSSELBERGHE
09:30 - 11:00	Session 3 - SMN beyond the motor neuron	AUDITORIUM
	Chair: Melissa Bowerman, university of Keele, UK	
	O09	Selective loss of excitation and p53 activation cause cerebellar circuit pathology in spinal muscular atrophy Sandra Wittig, Carl Ludwig Institute for Physiology, Leipzig, Germany
	O10	Microvascular dysregulation: The missing link in SMA pathogenesis? Hazel Allardyce, University of Aberdeen, UK
	O11	Translational defects in multiple tissues from the Smn2B/- Mouse model of SMA Gaurav Sharma, Institute of Biophysics, CNR, Trento, Italy
	O12	Identification of autonomic cardiovascular dysfunction in adult SMA patients: Towards the understanding of multisystem involvement in SMA Silvia Bonanno, Fond. IRCCS Ist. Neurologico C. Besta, Milan, Italy
11.00 - 11:30	COFFEE BREAK IN THE EXHIBITION AREA	
11.30 - 13:00	Session 4 - Developmental aspects of SMA	AUDITORIUM
	Chair: Giovanni Baranello, University College London, UK	
	O13	Time-course characterization of cortical projection neuron's alterations and development in delta7 SMA mice Roberta Schellino, Dept. of Neuroscience and Neuroscience Institute Cavalieri Ottolenghi, University of Turin, Italy
	O14	Transcriptional Mechanisms Differentiating Resilient and Vulnerable Spinal and Cranial Motor Neurons from human pluripotent stem cells in SMA Morgan Gazzola, I-Stem, Corbeil-Essones, France
	O15	Isogenic patient-derived organoids reveal early neurodevelopmental defects in spinal muscular atrophy initiation Natalia Rodriguez-Muela, German Center for Neurodegenerative Diseases e.V. (DZNE), Dresden, Germany
	O16	Is SMA a developmental disease? Federica Genovese, University of Edinburgh, UK
13:00 - 14:15	LUNCH BREAK IN THE EXHIBITION AREA	

FRIDAY 15th MARCH 2024

13:10 - 14:10	Industry Sponsored Session	T. VAN RYSSELBERHE
14.15 - 14.30	Flash poster session, Session 2	AUDITORIUM
	P118 Oro facial strength in symptomatic type 1 SMA patients treated with nusinersen: Results from a prospective study involving 4 centres Milano - Rome - Brussels - Ghent Charlotte Colot, Hôpital Universitaire des enfants reine Fabiola, Belgium	
	P17 Exon7 Targeted CRISPR-Prime Editing Approaches for SMN2 Gene Editing in Spinal Muscular Atrophy Cihan Tastan, Uskudar University, Turkiye	
	P135 Towards the identification of biomarkers of disease progression and response to treatment in spinal muscular atrophy Claudia Malacarne, IRCCS Fondazione Istituto Neurologico Carlo Besta, Italy	
	P64 Unravelling the role of GABA signalling and metabolism (dys)regulation in Spinal Muscular Atrophy: results from SMAΔ7 mice cortex Giovanna Menduti, Neuroscience Institute Cavalieri Ottolenghi, University of Turin, Italy	
	P100 Electrophysiological assessment of motor unit patterns of the median nerve in adolescents and adults with spinal muscular atrophy (SMA) Leandra Ros, University Medical Center Utrecht, The Netherlands	
	P47 Network biology-based analysis of SMA: Identification of disease relevant protein targets and altered signaling in severe and mild SMA mice Ines Tapken, Smatheria gGmbH - Non-Profit Biomedical Research Institute, Germany	
14.30 - 16.00	Session 5 - New avenues in SMA research Chair: Séverine Boilée, Université de la Sorbonne, Paris, France	AUDITORIUM
	O17 Molecular and morphological characterization of primary patient-derived fibroblasts to study gene-targeting therapies for spinal muscular atrophy (SMA) Ilaria Signoria, UMC Utrecht, The Netherlands	
	O18 The Spinal Muscular Atrophy gene product regulates actin dynamics Tobias Schüning, SMATHERIA gGmbH, Hannover, Germany	
	O19 Autophagy in spinal muscular atrophy: Detrimental or a therapeutic avenue? Saman Rashid, University of Hertfordshire, London, UK	
	O20 RAINBOWFISH: Primary efficacy and safety data in risdiplam-treated infants with presymptomatic SMA Michelle Farrar, UNSW Sydney, Randwick, Australia	
16.00 - 16.30	COFFEE BREAK IN THE EXHIBITION AREA	

FRIDAY 15th MARCH 2024

16.00 - 17.30	Poster Session 2	MINNEPLEIN
17:30 - 18:30	Parallel workshops Insights from other diseases Richard Finkel & Stefania Corti	H. VAN EYCK
	Newborn screening Eduardo Tizzano	T. VAN RYSSELBERGHE
19:30	GALA DINNER AT D'OUDE VISMIJN	

SATURDAY 16th MARCH 2024

09:30 - 11:00

Session 6 - Muscles in SMA

AUDITORIUM

Chair: Frédéric Relaix, UPEC - Université de Paris Est-Créteil, France

- O21** Effect of apitegromab on motor function and patient-reported outcomes at 36 months in patients aged 2-21 years with spinal muscular atrophy
Scott Bayer, Scholar Rock, Cambridge, United States
- O22** RNA-sequencing of Type II SMA paravertebral muscle after treatment reveals two distinct molecular subtypes
Fiorella Grandi, Centre de Recherche en Myologie, Paris, France
- O23** CIC-1 inhibitor compound improves neuromuscular transmission and enhances skeletal muscle function in pre-clinical animal models of neuromuscular dysfunction
Martin Skov, NMD Pharma A/S, Aarhus N, Denmark
- O24** Taldefgrobep Alfa: Preclinical and Clinical Data Supporting the Phase 3 RESILIENT Study in Spinal Muscular Atrophy
Cliff Bechtold, Biohaven, New Haven, United States

11.00 - 11:30

COFFEE BREAK AT THE EXHIBITION AREA

11.30 - 13:00

Session 7 - Monitoring disease outcome

AUDITORIUM

Chair: Michelle Farrar, UNSW Sydney, Randwick, Australia

- O25** Aconitase as a marker of early pathological state in SMA: Data from spinal cord and fibroblasts
Serena Stanga, Università degli studi di Torino, Italy
- O26** Cold-induced weakness in adolescents and adults with spinal muscular atrophy (SMA)
Leandra Ros, University Medical Center Utrecht, The Netherlands
- O27** H-reflex as a sensitive biomarker of sensory-motor circuit dysfunction in SMA mice and patients
Christian Simon, University Leipzig, Germany
- O28** Mass-Spectrometry-based proteomics on cerebrospinal fluid identified novel potential biomarkers for Spinal Muscular Atrophy
Chiara Panicucci, IRCCS Istituto Giannina Gaslini, Genova, Italy

13:00 - 14:00

GRAB A LUNCH BOX AT THE EXHIBITION AREA

14:00 - 15:00

Award Ceremony and closing talk

AUDITORIUM

Chair: Tom Gillingwater

Award ceremony: Best oral presentation, best three posters

Nicole Gusset and Tom Gillingwater

Wrap-up and highlights of the meeting - Where do we go next?

Nicole Gusset

CLOSE

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