2nd SCIENTIFIC & CLINICAL CONGRESS ON SPINAL MUSCULAR ATROPHY KRAKOW 2018

Thursday 25th January 2018

Key note
Where have we come, where do we go? – Arthur Burghes, PhD, Ohio State University – USA

Session 1 – SMN function importance in the context of new era

- Session chair – Basic function in splicing: RNA-mediated mechanisms of Spinal Muscular Atrophy – Livio Pellizzoni, PhD, Columbia University, USA
- Unfolding the role of SMN protein in controlling translation in vivo: implications for Spinal Muscular Atrophy – Gabriella Viero, PhD, Institute of Biophysics, CNR Unit at Trento, Italy
- Splicing analysis in a zebrafish model for Spinal Muscular Atrophy identifies transcripts important for motor neuron and Schwann cell function – Shermaine Tay, BSc, National University of Singapore, Singapore

Session 2: Neuronal specific function of SMN

- Session chair: Neuronal-specific function of SMN: Altered Axonal Actin Dynamics in Spinal Muscular Atrophy – Michael Sendtner, MD, PhD, University of Wursburg, Germany
- Converging mechanisms of p53 activation underlie selective degeneration of motor neurons in SMA – Christian Simon, PhD, University of Leipzig, Germany
- Defining Conserved Gene Networks Affected in Spinal Muscular Atrophy using Drosophila model – Taka Yokokura, PhD, Okinawa Institute of Science and Technology, Japan
- Neuronal activity regulates DROSHA via autophagy in Spinal Muscular Atrophy – Min Jeong Kye, PhD, University of Cologne, Germany
- Temporal and tissue variability of SMN protein levels in mouse models of SMA – Ewout Groen, PhD, University of Edinburgh, UK

Poster session A

Friday 26th January 2018

Industry Symposium
NOVARTIS

Session 3: SMA as a systemic disease

- Session chair – SMA as a systemic disease – Charlotte Sumner, MD, PhD, John Hopkins Medical Institute, USA
- SMN around the clock: circadian dysregulation in SMA – Melissa Bowerman, PhD, University of Keele, UK
- The development of heart defects in a mouse model of severe SMA – Simon Parson, PhD, University of Aberdeen, UK
- Abnormal fatty acid metabolism is a feature of spinal muscular atrophy – Rashmi Kothary, PhD, Ottawa Hospital Research Institute, Canada
- Identification and evaluation of new biomarkers for SMA – skeletal muscle and mitochondrial deficits – Nicole Hellbach, PhD, F. Hoffmann-La Roche Ltd

Session 4: Modifiers of phenotype

- Session chair – Protective modifiers help to unveil the cellular mechanism and to develop combinatorial therapies in spinal muscular atrophy – Brunhilde Wirth, PhD, University of Cologne, Germany
- CHP1 Reduction Ameliorates SMA Pathology by Restoring DNM1 Hyperphosphorylation and Endocytosis – Eva Janzen, MSc, University of Cologne, Germany
- Improvement of synaptic transmission at the NMJ in a mouse model of Spinal Muscular Atrophy – Rocio Tejero, PhD, University of Seville, Spain
- Comparison of independent screens on differentially vulnerable motor neurons reveals alpha-synuclein as a common modifier in motor neuron diseases – Lyndsay Murray, University of Edinburgh, UK
- RNA-Seq and Motif Analysis of Human Motor Neurons Reveals a Critical Role of SMN/SYNCRIP complex and Motif 7 in Spinal Muscular Atrophy – Stefania Corti, MD, PhD, University of Milan, Italy

Industry Symposium

AVEXIS – Gene Replacement Therapies for Monogenic Diseases, with focus on Spinal Muscular Atrophy – Dr. Samiah Al-Zaidy

Session 5: Pre-clinical combined therapies

- Session chair – Combinatorial opportunities with splice-switching ASOs in SMA – Christian Lorson, PhD, University of Missouri, USA
- Combinatorial ASO therapy using SMN-dependent and SMN-independent protection -NCALD reduction – against SMA – Laura Torres-Benito, PhD, University of Cologne, Germany
- Targeting the 5'UTR of survival motor neuron 2 (SMN2) to increase its expression in a disease model of spinal muscular atrophy – Audrey Winkelsas, BSc, National Institutes of Health, USA & University of Oxford, UK
- Dysregulated Signaling in SMA: from isolated pathway approaches to a clustered network representation – Niko Hensel, PhD, Hannover Medical School, Hannover, Germany
- Improved in vitro models of the human blood-brain barrier (BBB) using endothelial cells derived from induced pluripotent stem cells (iPSCs) for testing CNS therapeutics – Jamuna Selvakumaran, PhD, Royal Holloway, University of London, UK

Poster session B

Parallel workshops

- European collaboration – Moderated by Prof. Enrico Bertini, IRCCS Ospedale Pediatrico Bambino Gesù, Italy, and Dr. Eduardo Tizzano, Vall d'Hebron Hospital, Spain
- Big data & registries– Moderated by Prof. Hanns Lochmuller, Newcastle University, UK

Saturday 27th January 2018

Industry Symposium
BIOGEN: A multidisciplinary conversation on the evolving care for patients with SMA

Session 6: Emerging phenotypes & standards of care

- Session chair: Laurent Servais, MD, PhD, i-Motion, Institut de Myologie, France
- MRI of the cervical spinal cord and nerve roots in SMA – Marloes Stam, MD, University of Utrecht, The Netherlands
- End of Study Results from ENDEAR: Proportions of HINE-2 and CHOP INTEND Responders – Eduardo Tizzano, MD, PhD, Hospital Vall d´Hebron, Spain
- Cognitive Development, Language and use of Augmentative Alternative Communication in SMA1 Children in Italy – Grazia Zappa, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy
- Clinical challenges in the treatment of spinal muscular atrophy (SMA) with Nusinersen – Claudia Wurster, MD, University of Ulm, Germany

Session 7: Challenges of clinical trials & beyond

- Session chair: & Benefits of Earlier Treatment With Nusinersen in Infants and Children With Spinal Muscular Atrophy – Richard Finkel, MD, Nemours Children's Hospital, Florida, USA
- Clinical effects of nusinersen injections in SMA type 1 patients older than 7 months: 10 months of follow up – Karolina Aragon-Gawinska, MD, i-Motion, Institut de Myologie, France
- More than just fun and games: ACTIVE workspace volume video game quantifies upper extremity function in individuals with spinal muscular atrophy (SMA) – Linda Lowes, PT, PhD, Nationwide Children's Hospital, Columbus, USA
- FIREFISH, a multi-center, open-label trial to investigate the safety and efficacy of RG7916 in babies with Type 1 SMA: Study update and real-life experience of study implementation – Giovanni Baranello, MD, Carlo Besta Neurological Research Institute Foundation, Milan, Italy
- A long-term, open-label follow-up study of olesoxime in patients with Type 2 or non-ambulatory Type 3 spinal muscular atrophy who participated in a placebo-controlled Phase 2 trial – Francesco Muntoni, MD, PhD, University College London, UK

Conclusion

Summing-up – Professor Francesco Muntoni, University College London, UK

Closing ceremony