

# Schedule of Events

Registration—Lower Level

## Thursday, June 19th

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Continental Breakfast	7:30 AM - 8:30 AM	Plaza Ballroom
SMA Research Meeting	8:30 AM – 5:30 PM	Georgian Room
Cocktail Reception	5:30 PM – 7:00 PM	Arlington/Berkeley /Claredon Foyer
Meet and Greet with Families (including relay race with kids)	7:00 PM	Plaza Ballroom

## Friday, June 20th

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Continental Breakfast	7:30 AM - 8:30 AM	Plaza Ballroom
SMA Research Meeting	8:30 AM – 5:30 PM	Georgian Room
Banquet with Families	6:00 PM – 9:00 PM	Imperial/Plaza Ballroom

## Saturday, June 21st

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Continental Breakfast	7:30 AM - 8:30 AM	Plaza Ballroom
SMA Research Meeting	8:30 AM - 12:30 PM	Georgian room

Meeting Adjourns

8:30 AM Welcoming Remarks

**Human SMA Studies, Session Moderator Kathryn Swoboda**

8:40 AM Lisa Baumbach: Defining UBE-1 Mutations and Downstream Effects Underlying X-Linked Lethal SMA (XL-SMA)

9:00 AM Goran Simic: The Origin and Role of Heterotopic Motoneurons in the Pathogenesis of SMA

9:20 AM Seward Rutkove: Electrical Impedance Myography in the Assessment of Spinal Muscular Atrophy: Early Results.

9:40 AM Petra Kaufmann: The Natural History of Spinal Muscular Atrophy – Update from the PNCR Study

10:00 AM 15 Minute Break

**Clinical Trial and Newborn Screening Updates, Session Moderator Tom Crawford**

10:20 AM Eugenio Mercuri: Daily albuterol (salbutamol) in young patients with SMA type II.

10:40 AM Ching Wang: Hydroxyurea Clinical Trial in Types II/III SMA Patients

11:00 AM Kathryn Swoboda: SMA CARNI-VAL TRIAL: Randomized double-blind placebo-controlled trial of L-carnitine and valproic acid in children with SMA type II

11:20 Thomas Prior: Treatment and Prevention of Spinal Muscular Atrophy through Newborn and Population Carrier Screening

11:40 Rodney Howell: Developing a National Newborn Screening Translational Research Network

Lunch 12:00 to 1:00 PM ABC Foyer

**Regulation of SMN Expression, Splicing, and Function, Session Moderator Adrian Krainer**

1:10 PM Eric Hahnen: SMN2 gene promoter methylation: Implications for progression and therapy of SMA

1:30 PM Thomas Koed Doktor: hnRNP A1 inhibits splicing of SMN2 exon 7 by binding directly at the 3' splice site

1:50 PM Yimin Hua: Antisense Oligonucleotides Correct Human SMN2 Exon 7 Inclusion in Transgenic Mouse Tissues

2:10 PM Gordon Lutz: Oligonucleotide-Mediated SMN Expression and Functional Recovery in Mice with SMA

**Poster Session 2:30 PM to 5:30 PM**

2:30 to 4:00 PM	Group A Posters	Arlington/Berkeley/Claredon Room
4:00 to 5:30	Group B Posters	Arlington/Berkeley/Claredon Room
5:30 to 7 PM	Cocktail Hour	Arlington/Berkeley/Claredon Foyer
7 PM	Meet and Greet with Families	Plaza Ballroom

8:30 AM Morning Session Begins

**SMN Cellular Function, Session Moderator Louise Simard**

- 8:30 AM Gideon Dreyfuss: SMN deficiency causes tissue-specific perturbations in the repertoire of snRNAs and widespread defects in splicing
- 8:50 AM Klemens Hertel: Evaluation of the splicing fidelity in SMA patient cells and developing motor neurons
- 9:10 AM Virginia Mattis: SMN Read-through increases functionality compared to SMN<sup>Δ7</sup>
- 9:30 AM Eileen Workman: A SMA (type 1b) Missense Allele, SMN (A111G), is Capable of snRNP Assembly and Rescues SMA
- 9:50 AM Miroslav Dundr: De novo formation of Cajal body as a diagnostic tool for SMA
- 10:10 AM 15 Minute Break**
- 10:30 AM Heidi Fuller: Proteomic studies of SMA
- 10:50 AM Debra Shaw: Identification of novel nuclear SMN binding partners using LC-MS

**SMN Neuronal Specific Function, Session Moderator Doug Kerr**

- 11:10 AM Lingling Kong: Immaturity of neuromuscular junctions in SMA mice-implications for treatment
- 11:30 AM Umrao Monani: Reduced SMN protein impairs the maturation of neuromuscular junctions in SMA
- 11:50 to 1:00 PM Lunch -Terrace Room**
- 1:00 PM Lei Xing: Potential function of SMN in mRNA granule assembly and dynamics
- 1:20 PM Cyril Peter: Novel association of SMN with alpha subunit of COPI vesicle coat promoting complex
- 1:40 PM Brunhilde Wirth: Identification of a gender-specific protective modifier of SMA
- 2:00 PM Nicholas Parkinson: What does the SMN delta5 transgenic mouse tell us about SMN function and the relevance of profilin in SMA?
- 2:20 PM Howard Chang: A Drosophila model of human Spinal Muscular Atrophy
- 2:40 AM 15 Minute Break**

**Podium Poster Highlights and Poster Session 3:00 to 5:30 PM**

- 3:00 to 4:00 PM Podium Highlights of Selected Posters Georgian Room
- 4:00 to 5:30 PM Group C Posters Terrace Room

8:30 AM Morning Session Begins

**Drug Discovery, Moderator Arthur Burghes**

- 8:40 AM Hans Keirstead: Stem cell-derived high purity human motor neuron progenitors for research and therapy
- 9:00 AM Gregory Michaud: A Novel Therapeutic Target Protein for Spinal Muscular Atrophy
- 9:30 AM Bassem El Khodor: Phenotyping Neonatal models of neuromuscular degeneration: implications for drug candidate evaluation in the SMND7 model of SMA.
- 9:50 AM Ferrill Rose: Administration of recombinant Follistatin to a mouse model of SMA9
- 10:10 AM Matthew Butchbach: Evaluation of a novel, orally bioavailable histone deacetylase inhibitor in cell culture and mouse models of spinal muscular atrophy
- 10:30 AM 15 Minute Break**
- 10:50 AM Lili Wan: High throughput screening for drug discovery for SMA
- 11:10 AM Lee Rubin: High Content Screening for SMA Therapeutics
- 11:30 AM Sergey Paushkin: PTC Therapeutics identifies compounds that modulate SMN protein expression at the post-transcriptional level
- 11:50 AM Jonathan Cherry: Analysis of Screening Data and Validation of New Targets for the Treatment of SMA
- 12:10 AM Jill Heemskerk: Chemical Optimization of Indoprofen for the Treatment of SMA

**Meeting Adjourns**

Families of SMA would like to congratulate **Jeroen van Bergeijk** on receiving the **2nd Annual Anita McCaulay Memorial Travel Scholarship**. This travel scholarship provides funding for European students and post-doctoral fellows to attend the Annual International Spinal Muscular Atrophy Research Group Meeting. Dr. van Bergeijk is postdoctoral research assistant in the group of Dr. Peter Claus at the Institute of Neuroanatomy of the Hannover Medical School, Germany. His work concentrates on the function of SMN in neurites, in particular RhoA / Rho kinase (ROCK) signaling and G-/F-actin ratio in SMN depleted cells.