

Agenda | SCA & ARCA Global Conference 2020

19-21 October 2020 | online, Berlin time (CEST)

Monday, 19 October 2020

15:00 SCA & ARCA Global: setting the stage

Chair: Holm Graessner (Tübingen, Germany)

15:00 Welcome

Thomas Klockgether (Bonn, Germany), Matthis Synofzik (Tübingen, Germany)

15:15 Keynote lecture

Towards trial-equivalent natural history studies in SCAs and ARCAs: standards and criteria for trial-readiness

Massimo Pandolfo (Brussels, Belgium)

15:50 Session 1: Clinical, patient-reported and digital biomarkers

Chair: Thomas Klockgether (Bonn, Germany)

15:50 Biomarkers and prospects for interventional trials at the preataxic stage

Luis Velázquez-Pérez (Holguín, Cuba)

16:10 Patient-reported outcome measures for ataxia (PROM-Ataxia): ready for trials?

Jeremy Schmahmann (Boston, USA)

16:30 Speech as a biomarker in ataxia: What can it tell us and how should we use it?

Adam Vogel (Melbourne, Australia)

16:40 Wearable Sensors for multicentre clinical ataxia trials: assessment in clinics/lab

Fay Horak (Chicago, USA)

16:50 Wearable Sensors for multicentre clinical ataxia trials: assessment in real life

Winfried Ilg (Tübingen, Germany)

17:00 Break

Virtual coffee rooms

Meet the speakers of Session 1

17:30 Session 2: Fluid and imaging biomarkers

Chair: Gülin Öz (Minneapolis, USA)

17:30 Neurofilaments as translational fluid biomarkers for SCA: cross-species evidence from SCA3 cohorts and mice

Matthis Synofzik (Tübingen, Germany)

17:50 Ataxin-3 proteins in biofluids
Leonard Petrucelli (Jacksonville, USA)

18:10 MRI in SCA3
Marcondes França Jr. (Campinas, Brazil)

18:30 Magnetic resonance biomarkers in ARCAs: lessons learned
Pierre-Gilles Henry (Minnesota, USA)

18:50 Flash talks I: Clinical outcomes & natural history

Chair: Vikram Shakkottai (Ann Arbor, USA)

Natural history and phenotypic spectrum of multisystemic RFC1-disease: a first large cohort study, and a novel multicenter longitudinal natural history study
Andreas Träschütz (Tübingen, Germany)

Conversion of individuals at risk for spinocerebellar ataxia types 1, 2, 3, and 6 to manifest ataxia in the longitudinal RISCA study
Heike Jacobi (Bonn, Germany)

A new tool for prompt and comprehensive visualization and comparison of cohort characteristics in spinocerebellar ataxias
Mischa Uebachs (Bonn, Germany)

Linear progression of SARA scores in SCA 1, 2, 3 and 6 patients across countries
Emilien Petit (Paris, France)

Natural history of polymerase gamma related ataxia
Friedemann Bender (Tübingen, Germany)

Validation of a German version of the Cerebellar Cognitive Affective/Schmahmann Syndrome Scale: study protocol and preliminary results
Andreas Thieme (Essen, Germany)

Flash talks II: Molecular biomarkers and treatment approaches

Chair: Julie Greenfield (Ataxia UK)

Allele-specific AAV-based silencing of mutant ataxin-3 alleviates neuropathology and motor deficits in spinocerebellar ataxia type 3
Luis Pereira de Almeida (Coimbra, Portugal)

A pharmacological treatment acting on calcium homeostasis improves motor ability and delays Purkinje cell loss in the ARSACS mouse model
Francesca Maltecca (Milan, Italy)

Association of serum neurofilament light (sNfL) and disease severity in patients with spinocerebellar ataxia type 3
Hong Jiang (Changsha, Hunan, China)

PolyQ-expanded ataxin-3: a potential target engagement marker for SCA3 in peripheral blood
Jeannette Hübener-Schmid (Tübingen, Germany)

Combined transgene and intron-derived miRNA therapy for the treatment of SCA1
Ellie Carrell (Philadelphia, PA USA)

CRISPR/Cas9-based gene knockout in Spinocerebellar Ataxia type 1 (SCA1)
Mariangela Pappadà (Ferrara, Italy)

19:20 Break

Virtual coffee rooms

Meet the speakers of Session 2 and Flash Talks I & II

19:50 SCA Working Group Results

Chair: Tetsuo Ashizawa (Houston, TX, USA)

19:50 Clinical outcome assessments
Thomas Klockgether (Bonn, Germany)

20:20 MR biomarkers
Gülin Öz (Minneapolis, USA)

20:50 Biosampling standards
Puneet Opal (Chicago, USA)

21:20 SCA Global policy
Holm Graessner (Tübingen, Germany)

21:50 SCA Global – Young Investigator Initiative

Chair: Heike Jacobi (Heidelberg, Germany) & Jennifer Faber (Bonn, Germany)

- Introduction
- Ataxia scales training
- Further steps

22:30 End meeting day 1

Tuesday, 20 October 2020

15:00 Session 3: Trial-readiness

Chair: Laura Jardim (Porto Alegre, Brazil)

15:00 From trial-ready cohorts to first clinical trials in global multicenter studies of rare diseases: learning from DIAN-TU
David Clifford (St. Louis, USA)

15:20 Natural history and treatment trial design for ultra-rare diseases
Ralf-Dieter Hilgers (Aachen, Germany)

15:40 Making outcome measures ready for regulatory acceptance: FDA qualification standards for outcome measures, biomarker and data aggregation in ultra-rare diseases
Jane Larkindale (C-Path Institute, Tucson, USA)

16:00 Neurodegenerative Disorders: regulatory aspects
Karl Broich (BfArM, Bonn, Germany)

16:20 Session 4: Industry panel / presentations on planned trials

Chair: Holm Graessner (Tübingen, Germany)

16:20 Targeting DNA damage response genes for modulation of somatic instability
Irina Antonijevic (Triplet Therapeutics)

16:30 Assessment of the novel AAV-based miQURE™ gene therapy in SCA3 animal models
Lodewijk Toonen (UniQure)

16:40 Suppression of mutant polyQ protein expression throughout the CNS in HD, SCA1 and SCA3 mice using a single CAG repeat-targeting antisense oligonucleotide.
Nicole Datson (Vico Therapeutics)

16:50 Challenges and opportunities in the deployment of volumetric MRI in SCA clinical trials
Robin Wolz (IXICO)

17:00 Phase 2 Study of CAD-1883 for Spinocerebellar Ataxia (Synchrony-1)
Christopher Kenney (Cadent Therapeutics)

17:10 Results from the Long-Term Open Label Extension Phase Analyses of BHV4157-201: A Phase IIb/III, Randomized, Double-blind, Placebo-controlled Trial of the Safety and Efficacy of Troriluzole in Adult Subjects with Spinocerebellar Ataxia
Melissa Wolf Beiner (Biohaven)

17:20 Break

Virtual coffee rooms

Meet the speakers of Session 3 and 4

17:50 Flash talks III: Digital and imaging outcome markers and disease modifiers

Chair: Julie Greenfield (Ataxia UK)

Validation of hSARA and first data from home-application
Marcus Grobe-Einsler (Bonn, Germany)

Quantifying Ataxia-Related Motor Impairments in Ataxic Rodents and Humans Using Markerless Video Tracking With Deep Neural Networks
Jana Lang (Tübingen, Germany)

Clinical scales and vestibulo-ocular reflex show changes in time since pre-clinical stages in Machado-Joseph disease/spinocerebellar ataxia type 3 (BIGPRO Study)
Camila Maria de Oliveira (Porto Alegre, Brazil)

Gait Laboratory in a Box: Objective gait assessment via smartphone analysis
Andrea H.Németh (Oxford, UK)

Cross-sectional and longitudinal assessment of the structural alterations in pre-ataxic and ataxic Spinocerebellar Ataxia Type 3 (SCA3)
Jennifer Faber (Bonn, Germany)

Whole-exome sequencing identifies novel Machado-Joseph disease-modifying genes and pathways
Mafalda Raposo (Porto, Portugal)

Flash talks IV: Molecular mechanisms and treatment approaches

Chair: Brent Fogel (Los Angeles, USA)

Mechanism of conserved ancestral haplotype in SCA10
Tetsuo Ashizawa (Houston, TX, USA)

A DNzyme that cleaves CAG repeat RNA in polyglutamine diseases
Nan Zhang (Houston, TX, USA)

Atxin-1 is signalled to DNA damage by ATM kinase
Celeste Suart (Hamilton, Ontario, Canada)

Atxn2-CAG100-KnockIn mouse spinal cord shows progressive TDP43 pathology associated with cholesterol biosynthesis suppression
Georg Auburger (Frankfurt/M, Germany)

JNK inhibitor ameliorates the SCA1 phenotype by inhibiting Bergmann glial inflammation
Chandrakath R Edamakanti (Chicago, IL, USA)

Dentatorubral-pallidoluysian atrophy (DRPLA): development of an ASO therapy and understanding the impact of ATN1 CAG expansion
Joanna A.Korecka (Boston, MA, USA)

18:20 Session 5: Existing ARCA cohorts

Chair: José Luiz Pedroso (Sao Paolo, Brazil)

18:20 ARCAs in Europe: PREPARE
Matthis Synofzik (Tübingen, Germany)

18:35 ARCAs in Europe: SPATAX
Alexandra Dürr (Paris, France)

18:50 ARCAs in Canada
Bernard Brais (Montreal, Canada)

19:05 ARCAs in USA
Brent Fogel (Los Angeles, USA)

19:20 ARCAs in Central and South America
José Luiz Pedroso (Sao Paolo, Brazil)

19:35 ARCAs in Australia
David Szmulewicz (Melbourne, Australia)

19:50 ARCAs in Asia
Osamu Onodera (Niigata, Japan)

20:05 Break

Virtual coffee rooms

Meet the speakers of Session 5 and Flash Talks III & IV

20:35 Session 6: Genes & Mechanisms

Chair: Alexandra Dürr (Paris, France)

20:35 SCAs: mechanisms, model systems and compound screening
Henry Paulson (Michigan, USA)

20:55 ARCAs: mechanisms, model systems and compound screening
Helene Puccio (Lyon, France)

21:15 Genetic platforms for collaborative ARCA and SCA gene hunting

Chair: Matthias Synofzik (Tübingen, Germany)

21:15 GENESIS
Stephan Zuchner (Miami, USA)

21:25 GPAP / RD-Connect
Sergi Beltran (Barcelona, Spain)

21:35 CAGC
Brent Fogel (Los Angeles, USA)

21:45 Round table: NGS platforms – how to collaborate within ARCA and SCA Global?
All speakers; moderator: Matthias Synofzik (Tübingen, Germany)

22:00 End meeting day 2

Wednesday, 21 October 2020

15:00 ARCA Working Group Sessions

Chair: Matthias Synofzik (Tübingen, Germany)

15:00 Defining tasks and introduction of working groups
Holm Graessner (Tübingen, Germany)

15:10 ARCA Working Group Discussions:

- #1 Clinical outcomes & registry
Chair: Rebecca Schüle, Tübingen, Germany & Thomas Klockgether, Bonn, Germany
- #2 next-generation genomics & platforms
Chair: Matthias Synofzik (Tübingen, Germany)
- #3 molecular biomarkers & biosampling
Chair: David Mengel (Tübingen, Germany), Puneet Opal (Chicago, USA), Filippo Santorelli (Rome, Italy)
- #4 MRI biomarkers
Chair: Ian Harding (Melbourne, Australia), Sirio Coccozza (Naples, Italy), Pierre-Gilles Henry (Minnesota, USA)
- #5 digital-motor biomarkers
Chair: Adam Vogel (Melbourne, Australia) & Andreas Träschütz (Tübingen, Germany)
- #6 model systems & preclinical trials
Chair: Bernard Brais (Montreal, Canada), Francesca Maltecca (Milan, Italy), Helene Puccio (Lyon, France)

- #7 policies & patient organization engagement
Chair: Holm Graessner (Tübingen, Germany) & Julie Greenfield (Ataxia UK)

16:30 Break

Virtual coffee rooms

17:00 ARCA Working Group Results

Chair: Sue Hagen (NAF)

17:00 #1 Clinical outcomes & registry

Rebecca Schüle (Tübingen, Germany), Thomas Klockgether (Bonn, Germany)

17:10 #2 next-generation genomics & platforms

Matthis Synofzik (Tübingen, Germany)

17:20 #3 molecular biomarkers & biosampling

David Mengel (Tübingen, Germany), Puneet Opal (Chicago, USA), Filippo Santorelli (Rome, Italy)

17:30 #4 MRI biomarkers

Ian Harding (Melbourne, Australia), Sirio Coccozza (Naples, Italy), Pierre-Gilles Henry (Minnesota, USA)

17:40 #5 digital-motor biomarkers

Adam Vogel (Melbourne, Australia), Andreas Traschütz (Tübingen, Germany)

17:50 #6 model systems & preclinical trials

Bernard Brais (Montreal, Canada), Francesca Maltecca (Milan, Italy), Helene Puccio (Lyon, France)

18:00 #7 policies & patient organization engagement

Holm Graessner (Tübingen, Germany), Julie Greenfield (Ataxia UK)

18:10 ARCA Wrap-up

Matthis Synofzik (Tübingen, Germany)

18:30 End of SCA & ARCA Global Conference 2020