

# 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

15:00 Welcome

Thomas Klockgether (Bonn, Germany), Matthias 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

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

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

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

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

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

- Introduction
- Ataxia scales training
- Further steps

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

22:30 End meeting day 1

## Tuesday, 20 October 2020

### 15:00 Session 3: Trial-readiness

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

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

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)

### Flash talks IV: Molecular mechanisms and treatment approaches

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)

Ataxin-1 is signalled to DNA damage by ATM kinase

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)

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**

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

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

18:50 ARCAs in USA  
Brent Fogel (Los Angeles, USA)

19:05 ARCAs in Canada  
Bernard Brais (Montreal, Canada)

19:20 ARCAs in Central and South America  
José Luiz Pedroso (Sao Paulo, 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**

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**

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: Matthis Synofzik (Tübingen, Germany)

**22:00 End meeting day 2**

## Wednesday, 21 October 2020

### **15:00 ARCA Working Group Sessions**

15:00 Defining tasks and introduction of working groups

Holm Graessner (Tübingen, Germany)

15:10 ARCA Working Group Discussions:

- #1 Clinical outcomes & registry
- #2 next-generation genomics & platforms
- #3 molecular biomarkers & biosampling
- #4 MRI biomarkers
- #5 digital-motor biomarkers
- #6 model systems & preclinical trials
- #7 policies & patient organization engagement

**16:30 Break**

**Virtual coffee rooms**

### **17:00 ARCA Working Group Results**

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 Cocozza (Naples, Italy), Pierre-Gilles Henry (Minnesota, USA)

17:40 #5 digital-motor biomarkers

Adam Vogel (Melbourne, Australia)

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**