



International Congress for Ataxia Research

ATAXIA
Ataxia UK

FARA
Friedreich
Ataxia
Research
Alliance
fara.org

NAF
National Ataxia
Foundation
naf-foundation.org

NOVEMBER 1-4, 2022 DALLAS, TEXAS

Conference Daily Program

Tuesday 1st November

- 10am-2pm** **Registration, Exhibitor Check-in and Poster Set-up**
Registration & Exhibitor Set-up in Crystal Ballroom Foyer
Poster Set-up in Crystal Ballroom
- 10:00am** **Young Investigator Meet & Greet**
ICAR Trainee Organizing Committee
Crystal Ballroom Foyer
- 11:00am** **Mentorship Panel**
Panelists: Prof. Massimo Pandolfo (McGill University), Prof. Gülin Öz (University of Minnesota), Prof. Alexandra Durr (Salpêtrière University Hospital), Prof. Stefan Pulst (University of Utah), Prof. Helene Puccio (University of Lyon)
Chairs: Dr. Gabriela Bolzan (Universidade Federal do Rio Grande do Sul), Dr. Elisabetta Indelicato (Medical University of Innsbruck)
Lalique Ballroom
- 2:00pm** **Welcome to ICAR**
Mr. Andrew Rosen (National Ataxia Foundation)
ICAR Co-chairs: Dr. Stefan M. Pulst (University of Utah), Prof. Massimo Pandolfo (McGill University)
Crystal Ballroom
- 2:15pm** **(Keynote) The Future of Ataxia Clinical Trials: Strategies for Successful Academic Industry Partnerships**
Speaker: Dr. Nina Schor (NINDS/NIH)
Chair: Dr. Stefan M. Pulst (University of Utah)
Crystal Ballroom
- 3:00pm** **(Panel) Sharing Experience and Perspective: Living with Ataxia**

Chair: Ms. Jennifer Farmer (Friedreich's Ataxia Research Alliance)
Crystal Ballroom

3:40pm **Break & Exhibitor Exploration**
Crystal Ballroom Foyer

4:00pm **Parallel Breakout Sessions & Workshops**

Breakout: Disease Mechanisms

Chairs: Dr. Vikram Shakkottai (UT Southwestern), Ms. Celeste Stuart (McMaster University)

Crystal Ballroom

4:00pm *Uncovering the Regional and Cell-Type Specific Contributions Underlying Selective Cerebellar Degeneration in Spinocerebellar Ataxia Type 2*, Ms. Ashley Robbins (The Children's Hospital of Philadelphia)

4:15pm *Axonal swellings in a mouse model of ARSACS*, Dr. Amy Smith-Dijak (McGill University)

4:30pm *Examining the basis for age-dependent neuronal dysfunction in spinocerebellar ataxia type 6 (SCA6)*, Ms. Haoran Huang (UT Southwestern)

4:45pm *Reactive Bergmann glia play a central role in spinocerebellar ataxia inflammation via the JNK pathway*, Dr. Puneet Opal (Northwestern University)

5:00pm **Selected 5-min Flash Talks:**

Cotranslational degradation of mutant sarsin explains lack of genotype-phenotype correlation and defines molecular diagnosis in ARSACS patients, Dr. Daniele De Ritis (San Raffaele Scientific Institute)

Premature transcription termination induced by expanded GAAs leads to frataxin deficit in Friedreich's ataxia,

Workshop: Imaging

Chairs: Dr. Gülin Öz (University of Minnesota), Dr. Ian Harding (Monash University)

Lalique Ballroom

Dr. Marek Napierala (UT Southwestern)

Targeting ATAXIN-2 modulates p53-dependent apoptosis, Dr. Mandi Gandelman (University of Utah)

The bittersweet interrelation between O-GlcNAc transferase and the Machado-Joseph disease protein ataxin-3, Ms. Priscila Pereira Sena (University of Tuebingen)

5:30pm Break / Room Transition

5:40pm Clinical Grand Rounds I

Chairs: Dr. Stefan M. Pulst (University of Utah), Prof. Massimo Pandolfo (McGill University)

Speakers: Dr. Arnulf Koeppen (Albany VA Medical Centre), Dr. Deborah Hall (Rush University Medical School), Dr. Ricardo Currò (University College London) Crystal Ballroom

6:30-7:30pm Welcome Reception
Crystal Ballroom Foyer

Wednesday 2nd November

7:45am Breakfast
Garden Court

8:20am Day 2 Program Announcements
Ms. Jennifer Farmer (Friedreich's Ataxia Research Alliance)
Crystal Ballroom

8:30am Plenary: Disease Mechanisms
Chairs: Dr. Stefan Pulst (University of Utah), Prof. Jérôme Honnorat (Université de Lyon)
Crystal Ballroom

8:30am (Invited Talk) Autoimmune cerebellar ataxia: What is new in 2022? Prof. Jérôme Honnorat (Université de Lyon)

9:00am *Specific cerebellar spike signatures determine the presentation of cerebellar movement disorders*, Dr. Meike Van Der Heijden (Baylor College of Medicine)

9:15am *Altered calcium signaling in Bergmann glia contributes to decreased firing rate of Purkinje cells and motor deficits in a*

mouse model of Spinocerebellar ataxia type 1 (SCA1), Dr. Marija Cvetanovic (University of Minnesota)

9:30am *CGG repeat-elicited neurodegeneration in Fragile X Tremor-Ataxia Syndrome*, Dr. Peter Todd (University of Michigan)

9:45am *Sense and antisense RAN proteins in the CAG•CTG polyglutamine spinocerebellar ataxias*, Dr. Monica Banez Coronel, (University of Florida)

10:00am **Break & Exhibitor Exploration**
Crystal Ballroom Foyer

10:30am **Parallel Breakout Sessions and Workshops**

Breakout: Disease Mechanisms II

Chairs: Prof. Esther Becker (University of Oxford), Dr. Layne Rodden (Friedreich's Ataxia Research Alliance)

Crystal Ballroom

10:30am *Disruption of the CoQ10 biosynthetic Complex Q causes mitochondrial dysfunction and Ca²⁺ imbalance in Purkinje neurons in COQ8A-ataxia*, Dr. H el ene Puccio (IGBMC)

10:45am *Hyperexcitability and hypertrophy in the inferior olivary nucleus of the spinocerebellar ataxia type 1 brainstem*, Mr. Logan Morrison (UT Southwestern)

11:00am *Exploring the missing heritability in SPG7 heterozygous carriers with Whole Genome Sequencing*, Dr. Marie Coutelier (Sorbonne Universit e)

11:15am *A shared mechanism for SCA35 and gluten ataxia*, Dr Chih-Chun Lin (Columbia University)

11:30am **Selected 5-min Flash Talks:**

Breakout: Biomarkers

Chairs: Dr. Elisabetta Indelicato (Medical University of Innsbruck), Dr. Louise Corben (Murdoch Children's Research Institute)

Lalique Ballroom

The glucocorticoid receptor as a biomarker and neuronal therapeutic target of a disease-improving bile acid in SCA3/MJD, Dr. Jorge Diogo Da Silva (University of Minho)

Data-derived wearable digital biomarkers predict Frataxin gene expression levels and longitudinal disease progression in Friedreich's Ataxia, Prof. Richard Festenstein (Imperial College London)

Multi-omics analysis reveals very long chain ceramides as potential biomarkers and therapeutic target in Friedreich's ataxia, Dr. Clementina Mesaros (University of Pennsylvania)

Sensory and corticospinal signs before ataxia onset in SCA1 and SCA3: the READISCA study, Dr. Sophie Tezenas du Montcel (Sorbonne Universit e)

Selected 5-min Flash Talks:

Molecular Mechanisms of SCA48, Dr. Matt Scaglione (Duke University)

Genotype-phenotype correlation in RFC1 repeat expansion disease, Dr. Andrea Cortese (University College of London)

Reduction of BACE1 expression attenuates motor deficits and neuropathology in spinocerebellar ataxia type 1 mice, Dr. Jaehong Suh (Massachusetts General Hospital / Harvard Medical School)

Abnormalities and sex-difference in muscle histomorphology in Autosomal recessive spastic ataxia of Charlevoix-Saguenay, Prof. Elise Duchesne (Université du Québec à Chicoutimi)

Cerebrospinal fluid proteomic analysis in Friedreich ataxia, Prof. Massimo Pandolfo (McGill University)

Gait biomarker allow to capture robust longitudinal change in Spinocerebellar ataxia type 3 (SCA3) within one year, Dr Winfried Ilg (Hertie Institute for Clinical Brain Research)

Quantitative motor assessment of upper limb ataxia with Q-motor: a cross-sectional validation study including novel ataxia tasks, Dr. Andreas Träschütz (Hertie Institute for Clinical Brain Research)

A natural history study to track brain and spinal cord changes in individuals with Friedreich's ataxia: The TRACK-FA protocol, Dr. Helena Bujalka (Monash University)

12:00pm

Lunch and Poster Session

Lunch in Garden Court, Posters in Crystal Ballroom

12-1pm Even # posters present

1-2pm Odd # posters present

2:00pm

Plenary: Cerebellar Non-motor Circuits and Functions

Chairs: Dr. Peter Tsai (UT Southwestern), Dr. Ian Harding (Monash University)
Crystal Ballroom

2:00pm **(Invited Talk) Cerebellum and non-motor behaviors: cerebellar contributions to autism**
Dr. Peter Tsai (UT Southwestern)

2:30pm *Cerebello-cerebral functional connectivity in SCA3 prior to ataxia onset: Resting state fMRI findings from READISCA*, Dr. Sheeba Anteraper (Carle Foundation Hospital)

2:45pm *The Cerebellar Cognitive Affective Syndrome Scale in Spinocerebellar Ataxias: A CRC-SCA/READISCA Study*, Dr. Louisa P. Selvadurai (Massachusetts General Hospital)

3:00pm *Impulsivity Trait in Cerebellar Ataxia and Parkinson Disease*, Dr. Sheng-Han Kuo (Columbia University Irving Medical Center)

3:15pm *Cognitive-affective manifestations in ataxic and pre-ataxic phases of Spinocerebellar Ataxia type 3/Machado-Joseph Disease*, Dr. Gabriela Bolzan (Universidade Federal do Rio Grande do Sul)

3:30pm **Break & Exhibitor Exploration**
Crystal Ballroom Foyer

4:00pm **Parallel Breakout Sessions & Workshops**

| | Breakout: Emerging Therapies (Preclinical) | Breakout: Imaging | Workshop: Cerebellum Neuroanatomy & Physiology |
|---------------|--|---|---|
| | <u>Chairs:</u> Dr. Beverly Davidson (The Children's Hospital of Philadelphia), Dr. Ronald Buijsen (Leiden University Medical Center) | <u>Chairs:</u> Prof. Marcondes França (Universidade Estadual de Campinas), Prof. Gilles Naeije (Université libre de Bruxelles) | <u>Chairs:</u> Dr. Roy Sillitoe (Baylor College of Medicine), Dr. Henry Paulson (University of Michigan) |
| | Crystal Ballroom | Lalique Ballroom | Waterford A/B |
| 4:00pm | <i>CRISPR gene-editing rescues molecular and motor phenotypes in Fmr1 CGG knock-in mice</i> , Dr. Carolyn Yrigollen (Children's Hospital of Philadelphia) | <i>Longitudinal multimodal MRI in SCA3: Evaluation of imaging biomarker candidates from the early pre-ataxic to the late ataxic stage</i> , Dr. Jennifer Faber (DZNE) | <i>Intra-Cerebellar Regional Molecular Differences Confer Vulnerability in Spinocerebellar Ataxia Type 1 Pathology</i> , Ms. Katie Hamel (University of Minnesota) |
| 4:15pm | <i>Progress towards a viral gene therapy for Christianson syndrome</i> , Dr. Collin Anderson (University of Utah) | <i>Pons and middle cerebellar peduncle diameters are diagnostic of Multiple System Atrophy of the cerebellar type (MSA-C)</i> , Dr. Christopher Stephen (Massachusetts General Hospital / Harvard Medical School) | <i>Characterization of cerebellar astrocyte reactivity and metabolism in a new mouse model of Friedreich's ataxia</i> , Dr. Frida Loria (Hospital Universitario Fundación Alcorcón) |
| 4:30pm | <i>Intranasal delivery of Extracellular Vesicles carrying silencing sequences alleviates Spinocerebellar Ataxia Type 3 (SCA3)</i> , Mr. David Rufino-Ramos (University of Coimbra) | <i>Dorsal Root Ganglia Imaging is a Potential Biomarker in Friedreich's Ataxia</i> , Dr. Rafaella Tacla (University of Campinas) | <i>Impaired reinforcement learning in patients with cerebellar ataxia</i> , Dr. Christian Johannes Amlang (Columbia University) |

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|--------|--|--|---|
| 4:45pm | <i>Inhibiting Hsp90 as a therapeutic approach for ARSACS</i> , Dr. Suran Nethisinghe (University College of London)) | <i>Tract-specific spinal cord diffusion tensor imaging in Friedreich's ataxia</i> , Prof. Marcondes C. França Jr (University of Campinas) | <i>Patterned Purkinje Cell Loss During Normal Aging in Mice</i> , Ms. Sarah Donofrio (Baylor College of Medicine) |
| 5:00pm | <p>Selected 5-min Flash Talks:</p> <p><i>Development of an ATXN3-targeted siRNA therapy for Spinocerebellar ataxia type 3</i>, Dr. Maria do Carmo Costa (University of Michigan)</p> <p><i>Metformin decreases RAN proteins and improves behavioral phenotypes in SCA8 BAC mice</i>, Dr. Setsuki Tsukagoshi (University of Florida)</p> <p><i>Dentatorubral-pallidoluysian atrophy (DRPLA): ASO therapeutic development and understanding the impact of ATN1 CAG expansion</i>, Dr. Joanna Korecka (Brigham and Women's Hospital / Harvard Medical School)</p> <p><i>A CAG expansion-selective chemical screen identifies compounds that selectively reduce CAG-expansion transcript levels across spinocerebellar ataxias</i>, Dr. Hannah Shorrock (University at Albany-SUNY)</p> | <p>Selected 5-min Flash Talks:</p> <p><i>In vivo evaluation of dentato-thalamo-cortical tract integrity in Friedreich ataxia using diffusion MRI</i>, Dr. Mario Tranfa (University "Federico II")</p> <p><i>Long-term Cardiac magnetic resonance imaging study in Friedreich's Ataxia</i>, Dr. Thiago Rezende (University of Campinas)</p> <p><i>Neuroinflammation in the cerebellum and brainstem in Friedreich ataxia: an [18F]-FEMPA PET study</i>, Dr. Ian Harding (Monash University)</p> <p><i>Natural history of magnetic resonance imaging in the pre-ataxic stage of Machado-Joseph Disease: BIGPRO study</i>, Prof. Laura Bannach Jardim (Hospital de Clinicas de Porto Alegre)</p> | <p>Open Discussion</p> |
| 5:30pm | Break / Room Transition | | |
| 5:40pm | Parallel Breakout Sessions & Workshops | | |

Clinical Grand Rounds 2

Chairs: Dr. Stefan Pulst (University of Utah), Prof. Massimo Pandolfo (McGill University)

Speakers: Prof. Marios Hadjivassiliou, (Royal Hallamshire Hospital), Dr. Penina Ponger (Tel-Aviv Sourasky Medical Center), Dr. Dana Sugar (Rush University)

Crystal Ballroom

Workshop: Science Dissemination to Lay Audiences

Chairs: Ms. Celeste Suart (McMaster University), Dr. Mohamed Elsaey (The Technische Universität Braunschweig)

Lalique Ballroom

6:30pm **End of Program**

7:00pm **Trainee Trivia & Pub Night (Registration Required)**
Ida Claire

Thursday 3rd November

7:45am **Breakfast**
Garden Court

8:20am **Day 3 Program Announcements**
Ms. Sue Millman (Ataxia UK)
Crystal Ballroom

8:30am **Plenary Session: Emerging and Existing therapeutics**
Chairs: Dr. Beverly Davidson (The Children's Hospital of Philadelphia), Prof. Massimo Pandolfo (McGill University)
Crystal Ballroom

8:30am **(Invited Talk) *Emerging and existing therapies in ataxias***, Dr. Beverly Davidson (The Children's Hospital of Philadelphia)

9:00am *Cerebellar tDCS in Friedreich Ataxia: A randomized, double-blind, sham-controlled, crossover trial*, Dr. Gilles Naeije (Universite libre de Bruxelles)

9:15am *GeneTACTM small molecules increase frataxin in a mouse model of Friedreich ataxia, restore FXN and improve mitochondrial function in patient-derived cells, and achieve sustained biodistribution in CNS and heart in rats and non-human primates*, Dr. Nancy Levin (Design Therapeutics)

9:30am *CLR01, a molecular tweezer, attenuates motor dysfunction and pathology in SCA3 in vivo models*, Prof. Patrícia Maciel (University of Minho)

9:45am *Restoring calcium homeostasis in Purkinje cells arrests neurodegeneration and neuroinflammation in the ARSACS mouse model*, Dr. Andrea Del Bondio (San Raffaele Scientific Institute)

10:00am **Break & Exhibitor Exploration**
Crystal Ballroom Foyer

10:30am **Parallel Breakout Sessions & Workshops**

| | Breakout: Cell and Animal Models | Breakout: Clinical Outcome Assessments and Natural History Studies | Workshop: Inherited Ataxias: from clinical presentation to genetic diagnosis |
|----------------|--|--|---|
| | <p><u>Chairs:</u> Dr. Roy Sillitoe (Baylor College of Medicine), Dr. Rosella Abeti (University College of London)</p> | <p><u>Chairs:</u> Dr. Jeremy Schmahmann (Massachusetts General Hospital), Dr. Michelle Tosin (Rush University Medical Center)</p> | <p><u>Chairs:</u> Dr. Elisabetta Indelicato (Medical University of Innsbruck), Dr. Gabriela Bolzan (Universidade Federal do Rio Grande do Sul), Prof. Sylvia Boesch (University of Innsbruck), Prof. Andrea Nemeth (University of Oxford)</p> |
| | Crystal Ballroom | Lalique Ballroom | Waterford A/B |
| 10:30am | <i>Generation of mechanosensory neurons from human pluripotent stem cells</i> , Ms. Amy Hulme (University of Wollongong) | <i>The FA App: A Smartphone/Tablet Platform for Global Virtual Research in Friedreich Ataxia</i> , Dr. Ian Harding (Monash University) | |
| 10:45am | <i>Patient-specific iPSCs reveal vascular dysfunction in Friedreich's Ataxia</i> , Dr. Jarmon Lees (St. Vincent's Institute of Medical Research) | <i>What matters to patients – a framework and resource for development of meaningful outcomes in ataxias</i> , Prof. Matthis Synofzik (University of Tübingen) | |
| 11:00am | <i>Spinocerebellar ataxia type 1 characteristics in patient-derived fibroblast and induced pluripotent stem cell-derived neuronal cultures</i> , Dr. Ronald Buijsen (Leiden) | <i>The FA-HI & FACR-HI: Development and Validation of Two Novel Friedreich's Ataxia Outcome Measures</i> , Mr. Spencer Rosero (University of Rochester) | |

University Medical Center)

11:15am

A preclinical behavioral assay and analytical platform to model genetic background, age, and zygosity in CHIP-dependent ataxia phenotypes (SCA48/SCAR16), Prof. Jonathan Schisler (University of North Carolina at Chapel Hill)

Longitudinal observation of clinical scales and oculomotor neurophysiology since the pre-ataxic stage of Machado-Joseph disease: BIGPRO study, Prof. Laura Bannach Jardim (Hospital de Clinicas de Porto Alegre)

11:30am

Selected 5-min Flash Talks:

Activation of the type I interferon response associated with frataxin knockdown in iPSC-derived cardiomyocytes, Dr. M.Grazia Cotticelli (Children's Hospital of Philadelphia)

Neurodegenerative synergy between RAN translation and CGG repeat RNA toxicity in rodent models of FXTAS, Ms. Samantha Grudzien (University of Michigan-Ann Arbor)

An induced pluripotent stem cell-based model to investigate proprioceptive neuronal pathology in Friedreich ataxia, Dr. Chiara Dionisi (Université Libre de Bruxelles)

Novel genetic modifiers of SCA3/MJD: an EMS screening in a C. elegans model of the disease, Prof. Patrícia Maciel (University of Minho)

Selected 5-min Flash Talks:

Development and validation of the dysarthria impact scale, Prof. Adam Vogel (The University of Melbourne)

The S-Factor, a new measure of disease severity in spinocerebellar ataxia: Findings and implications, Dr. Louisa Selvadurai (Massachusetts General Hospital)

FA-CHILD – A 3-year, 6 Month-Interval Natural History Study in Children with Friedreich's ataxia, Dr. Christian Rummey (Clinical data science GmbH)

Gait, electromyography and synergies analysis of Freidreich's ataxia, Dr. Gessica Vasco (Bambino Gesù Children's Hospital)

12:00pm **Lunch and Poster Session 2**
Lunch in Garden Court, Posters in Crystal Ballroom

12-1pm Even # posters present

1-2pm Odd # posters present

2:00pm **Plenary Session: Interactive Debate**
Chair: Dr. Deborah Hall (Rush University Medical Center)
Crystal Ballroom

- 1) *Are gene-based therapies going to be the answer for ataxias?* Dr. Peter Todd (University of Michigan), Dr. Deborah Hall (Rush University)
- 2) *Should statistically significant change (for example in a biomarker) or clinically meaningful change be the outcome measure in ataxia clinical trials?* Dr. Alexandra Durr (University Hospital Salpêtrière), Dr. David Lynch (University of Pennsylvania & Children's Hospital of Philadelphia)
- 3) *Are ataxia animal models appropriate for pre-clinical testing of trinucleotide repeat-targeted therapeutics, like gene therapy?* Dr. Laura Ranum (University of Florida), Dr. Vikram Shakkottai (UT Southwestern)

3:30pm **Break & Exhibitor Exploration**
Crystal Ballroom Foyer

4:00pm **Parallel Breakout Sessions & Workshops**

Breakout: Emerging Therapies (Clinical)

Chairs: Dr. Alexandra Durr (University Hospital Salpêtrière), Dr. Gabriela Bolzan (Universidade Federal do Rio Grande do Sul)

Crystal Ballroom

4:00pm *Safety and Pharmacokinetics of Single- and 13-Day Multiple-Dose Administration of CTI-1601, a Frataxin Replacement Therapy for Friedreich's Ataxia*, Dr. Nancy Ruiz (Larimar Therapeutics, Inc.)

4:15pm *Home Aerobic Training versus Balance Training for Cerebellar Ataxia: A Randomized Controlled Trial*, Dr. Scott Barbuto (Columbia University Irving Medical Center)

4:30pm *SpeechATAX: A rater blinded randomized controlled trial of intensive*

Workshop: SARA Scale

Chairs: Dr. Jennifer Faber (DZNE), Dr. Heike Jacobi (University of Heidelberg)

Waterford A/B

home-based biofeedback therapy for dysarthria progressive ataxia, Prof. Adam Vogel (The University of Melbourne)

4:45pm Autophagy as a treatment pathway in Spinocerebellar Ataxia: SLS-005 (Trehalose injection, 90.5 mg/mL for intravenous infusion), Dr. David Biondi (Seelos Therapeutics Inc)

5:00pm **Selected 5-min Flash Talks:**

Engage-Ataxia: Preliminary results after 1 year from a physical activity coaching intervention in individuals diagnosed with ataxia, Dr. Chelsea Macpherson (Columbia University)

A Phase 1/2 Study of the Safety and Efficacy of LX2006 Gene Therapy in Participants with Cardiomyopathy Associated with Friedreich's Ataxia, Dr. Jay Barth (LEXEO Therapeutics Inc.)

A randomised placebo-controlled crossover trial of micronised resveratrol as a treatment for Friedreich ataxia, Prof. Martin Delatycki (Murdoch Children's Research Institute)

Cerebellar transcranial direct current stimulation in spinocerebellar ataxia type 3: a randomized, double-blind, sham-controlled trial, Dr. Roderick Maas (Radboud University Medical Center)

5:30pm **End of Program**

6:30pm **Busses leave hotel for Offsite Dinner: Boot Scootin' Night Out**
Circle R Ranch

Friday 4th November

8:00am **Breakfast & Poster Take Down (must be removed by 10:30am)**
Garden Court

9:00am **Plenary Session: Late-Breaking Research**

Chair: Prof. Andrea Nemeth (University of Oxford)
Crystal Ballroom

9:00am *Variiegated silencing in Friedreich ataxia*, Prof. Sanjay Bidichandani (University of Oklahoma Health Sciences Center)

9:20am *Identification of β -III-spectrin actin binding modulators for treatment of SCA5*, Dr Adam Avery (Oakland University)

9:40am *Direct utility of natural history data in analysis of clinical trials: Propensity match-based analysis of Omaveloxolone in Friedreich ataxia using the FA-COMS dataset*, Dr. David Lynch (University of Pennsylvania & Children's Hospital of Philadelphia)

10:00am *An intronic GAA repeat expansion in FGF14 causes autosomal dominant adult-onset ataxia (SCA50, ATX-FGF14)*, Dr. David Szmulewicz (University of Melbourne)

10:20am *Expansion of a Deep Intronic FGF14 GAA Short Tandem Repeat in Late-Onset Cerebellar Ataxia*, Dr. Bernard Brais (McGill University)

10:30am **Conference Summary and Awards**
Dr. Stefan M. Pulst (University of Utah)
Crystal Ballroom

10:45am **Final Remarks**
Mr. Andrew Rosen (National Ataxia Foundation)
Crystal Ballroom

10:50am **Break / Room Transition**

11:00am **Trainee Flash Talks for Patients, Caregivers and Friends (open to all ICAR attendees)**
Chairs: Dr. Layne Rodden (Friedreich's Ataxia Research Alliance), Dr. Ronald Buijsen (Leiden University Medical Center)
Waterford A/B

1:00pm **Ataxia Global Initiative conference starts**
Lalique Ballroom

Appendix:
Descriptions of ICAR Workshops

ICAR 2022 Imaging Workshop

Chaired by:
Dr. Ian Harding, Monash University
Prof. Gulin Oz, University of Minnesota

Overview:

Everything you wanted to know about neuroimaging, but were too afraid to ask! This workshop will provide an introductory overview of common human MRI acquisition approaches, analysis tools, and outcome measures that are used in ataxia research and clinical trial contexts. The workshop will be targeted to academic and industry scientists with limited knowledge or experience with neuroimaging. (Note: we will not address clinical radiological practise, ie. radiological diagnosis, or specific data analysis approaches).

There will be plenty of time for interactions with the panel, so please bring lots of questions!

Learning Objectives:

- Entry-level knowledge base to facilitate research/trial design decisions, academic literacy, and communications with neuroimaging/radiology collaborators
- Overview of common human brain (and spinal cord) MRI data types, tools, and outcome measures used in ataxia research & trials
- Understanding potential biomarker and disease characterisation use cases

Speakers and Topics:

1. Introduction and General Considerations (Speaker: Dr. Ian Harding)
2. Volumetric MRI (Speaker: Dr. Thiago Rezende)
3. Diffusion MRI (Speaker: Dr. Christophe Lenglet)
4. Quantitative Susceptibility Mapping (Speaker: Dr. Ian Harding)
5. Magnetic Resonance Spectroscopy (Speaker: Dr. Gulin Oz)
6. Functional MRI (Speaker: Dr. Sheeba Anteraper)
7. Open Discussion

ICAR 2022 Cerebellar neuroanatomy and physiology Workshop

Chaired by:

Dr. Roy Sillitoe, Baylor College of Medicine

Dr. Henry Paulson, University of Michigan

Overview:

Cerebellar structure and function are both intimately linked to the onset, progression, and severity of ataxia. However, it is unclear how circuit mis-assembly, neurodegeneration, and altered function (in the absence of a clear pathology) converge on the same cerebellar microcircuit and ultimately cause the same behavioral abnormalities. This workshop will consider the cellular architecture and connectivity of the normal and ataxic cerebellum to gain a better understanding of how cerebellar abnormalities drive disease-related behavioral symptoms. Motor and non-motor impacts will be considered. Discussions will focus on both human and animal models, with the latter providing an opportunity to highlight novel experimental approaches such as virus-mediated circuit mapping, optogenetics, DREADDs, deep brain stimulation, and molecular connectomics. The workshop will be of interest to trainees and faculty from both research and clinical settings.

Speakers (10 min talks + 5 min Q&A):

Ms. Katie Hamel - Intra-Cerebellar Regional Molecular Differences Confer Vulnerability in Spinocerebellar Ataxia Type 1 Pathology (poster sub)

Dr. Christian Johannes Amlang - Impaired reinforcement learning in patients with cerebellar ataxia

Ms Sarah Donofrio – Patterned Purkinje Cell Loss During Normal Aging in Mice

Dr Frida Loria - Characterization of cerebellar astrocyte reactivity and metabolism in a new mouse model of Friedreich's ataxia

Open discussion (30 minutes)

Science Dissemination to a Lay Audience

Chaired by:

**Celeste Suart, McMaster University
Dr. Mohamed Elsaey, Suez Canal University**

Overview:

Want to share your research findings and scientific knowledge with the public, but you are not sure where to start? This workshop will provide an introduction to key concepts and strategies for communicating with lay audiences about scientific and medical information. We will review common errors and difficulties with lay audience communication, followed by best practices for lay writing. Participants will have the opportunity to apply what they have learned through workshop activities. Prior to the workshop, we ask that all attendees identify an abstract from a previously published paper and bring it with them to the session (digitally or hard copy).

Learning Objectives:

By the end of this session, participants will be able to:

- Identify common challenges scientists encounter when writing for lay audiences
- Describe the Inverted Triangle Model of lay article writing
- List best practices for clear communication with non-specialist audiences
- Apply skills learned in the workshop to transform a scientific abstract into a lay abstract

Topics (60mins total):

1. Introduction and Overview of Session
2. Common Errors in Lay Audience Communication
3. The Inverted Triangle Model of Writing
4. Best Practices for Lay Audience Communication
5. Writing a Lay Abstract
6. Open Discussion

ICAR 2022 Scales Training

Chaired by:

**Dr. Jennifer Faber, DZNE Bonn
Dr. Heike Jacobi, University of Heidelberg**

on behalf of the Ataxia Global Young Investigator Initiative

Overview:

One of the central aims of the Ataxia Global Young Investigator Initiative is the education and training to assure high quality of clinical assessments by trained examiners. The workshop will cover the most important scales for the assessment of ataxic patients, in clinical practice but also for research purposes. We will introduce the scales and show patient videos to practice assessment and highlight some common pitfalls.

This workshop is directed to residents/trainees working in the field of ataxias.

ICAR 2022 Clinical Workshop
Inherited Ataxias: from clinical presentation to genetic diagnosis

Chaired by:

Prof. Sylvia Boesch; University of Innsbruck
Prof. Andrea Nemeth, University of Oxford
Dr. Elisabetta Indelicato, PhD; University of Innsbruck
Dr. Gabriela Bolzan, PhD; University of Porto Alegre

Overview:

Every young attendee at ICAR knows really a lot about one or maybe some forms of ataxia. What about a complete, clinical practise oriented overview: what I do have to search and how in the first evaluation of a patient with ataxia? Which genetic testing am I supposed to choose? This workshop will provide a clinical flow chart to guide the evaluation of a patient with ataxia, starting from clues to rule out secondary and sporadic forms, key clinical aspects in the evaluation of a genetic ataxia to indications and limitations of the different genetic diagnostic tools (e.g. analysis of copy number variants, gene panels, whole-exome and whole-genome sequencing).

The workshop addresses contents relevant for the clinical neurological practise, updated on the light of the latest literature and is targeted to residents/trainees.

Learning Objectives:

- Provide a structured approach in the first evaluation of a patient with ataxia in the clinical practise.
- Provide a clinically oriented knowledge on different type of genetic testing available with indications and limitations in the setting of genetic ataxias.

Speakers and Topics (90mins total):

1. Introduction (Speaker: Dr. Elisabetta Indelicato and Dr. Gabriela Bolzan) *ca. 5-10 min*
2. Clinical Part (Speakers: Prof. Sylvia Boesch, Dr. Elisabetta Indelicato, Dr. Gabriela Bolzan) *40 min*
 - Ruling out acquired and sporadic forms; pitfalls and overlapping clinical pictures
 - Genetic Ataxias: pragmatic clinicogenetic classification, case presentations, differential diagnosis
 - Focus on dominant Ataxias.
 - Focus on recessive Ataxias.
3. Genetic part (Speaker: Prof. Andrea Nemeth) *30 min*
 - Overview about available genetic tools in ataxia diagnostics (PCR, Sanger Sequencing and NGS techniques). Possibilities and limitations of each tool as well as current application in the clinical practice.
4. Questions & Discussion *10-15 min*