Programme

10-year anniversary Conference Alternating Hemiplegia of Childhood and *ATP1A3* diseases

and

10th Symposium on *ATP1A3* in disease 2022

Hybrid: Edinburgh and online

19th-21st October 2022

Royal College of Physicians Edinburgh,
9 Queens Street

Times shown are British Summer Time (BST)

Join in with the conversation:
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Wednesday 19th October 2022

Day 1: The Past - AHC and *ATP1A3*, the last 10 years

09:00-10:00
Registration and coffee for delegates and speakers
Main Foyer in Conference Centre

09:00-09:30
Opportunity for families to gather in person and discuss priorities for next 3 days
Meeting room 1/2

Session 1.
Alternating Hemiplegia of Childhood (AHC) and *ATP1A3*: an overview
Chair: **Professor Sanjay Sisodiya**
Lecture theatre and online

10:00
Welcome by organising committee. A reminder of why we are all gathering – focus on those with the lived experience of Alternating Hemiplegia of Childhood & *ATP1A3* diseases.
Followed by a short playing of the video 'Human Timebombs'
Katherine Behl, AHC UK and Conference organising committee

10:10
Keynote presentation
ATP1A3 disease –phenotypic description to gene discovery
Plenary talk discussing the basic science perspective of the gene discovery
Professor Kathleen Sweadner, Harvard University

10:40
The evolving clinical spectrum of AHC and related conditions
Professor Hendrik Rosewich, University Medical Center, Goettingen

11:00
What is the role of *ATP1A3*?
Professor Poul Nissen, Aarhus University

11:20
Panel discussion

11:30-11:45 *Coffee break, poster viewing and networking*
Main Foyer in Conference Centre

Session 2:
The development of animal models in the study of *ATP1A3* diseases – what can they tell us?
Chair: **Professor Arn Van den Maagdenberg**
Lecture theatre and online

11:45
State of the art historical overview on animal models of *ATP1A3*-related disorders
Dr Steve Clapcote, University of Leeds

12:15
Panel discussion

12:30
The ATP1A3 standing committee
Dr Hendrik Rosewich and **Dr Tsveta Schyns**

12:45-13:45 *Lunch, poster viewing and networking*
Main Foyer, Conference Centre

Session 3: Collaborative science – the AHC and *ATP1A3* community and what it has brought Chair: **Johanna Brown** *Lecture theatre and online*
13:45 The lived experience of CAPOS **Ms Sonal Sumaria**
14:05 *(virtually)* The diagnostic criteria of AHC and *ATP1A3* diseases **Professor Mohamed Mikati,** Duke University
14:25 What does it mean to have a 'broken' *ATP1A3* pump? **Professor Arn Van den Maagdenberg,** Leiden University Medical Centre
14:45 Panel discussion
14:55-15:15 *Coffee break, poster viewing and networking*
Session 4: Moving forwards towards new nosology and classification Chair: **Katherine Behl** *Lecture theatre and online*
15:15 Day in the life of a parent……predictably unpredictable **Johanna Brown,** AHC UK and Conference Organising Committee
15:30 Debate: What's in a name? How should AHC be named and classified for families, clinical practice, and research? **Professor Sanjay Sisodiya** **Professor Hendrik Rosewich**
16:10 Panel discussion
16:20-16:30 Learning points from the day Closure

16:30: Optional historical tour of the Royal College of Physicians of Edinburgh Libraries

Thursday 20th October 2022

Day 2: The Present. AHC & *ATP1A3* diseases – where are we now, and where are we going?

08:00-08:30
Registration and coffee for delegates and speakers
Main Foyer in Conference Centre

08:25
Opening of Day 2 – Lived experience of AHC and *ATP1A3* diseases
Lecture theatre and online

Session 1:
Sharing current research on AHC and *ATP1A3* diseases: the life-course clinical perspective
Chair: **Dr Simona Balestrini**
Lecture theatre and online

08:30
Why are natural history studies crucial for understanding the disease and potential future treatments? *Learning from other rare conditions*
Professor Andreas Brunklaus, University of Glasgow

08:50
Addressing the genotype-phenotype correlation in AHC and *ATP1A3* diseases
Dr Aikaterini Vezyroglou, Great Ormond Street Hospital, University College London

09:10 *(virtually)*
ATP1A3 mutations cause polymicrogyria
Professor Renzo Guerrini, University of Florence

09:30
Transition from childhood to adulthood
Dr Eleni Panagiotakaki, University Hospitals of Lyon

09:50
AHC – a lifelong disease. Long-term follow-up of adults with AHC
Dr Marco Perulli, Catholic University of The Sacred Heart, Rome

10:10
Panel discussion

10:20-10:35 *Coffee break, poster viewing and networking*
Main Foyer in Conference Centre

Session 2:
Key dilemmas for clinicians, researchers, and families
Chair: **Dr Aikaterini Vezyroglou**
Lecture theatre and online

10:35
How do we prevent delay in a diagnosis of AHC and *ATP1A3* diseases?
Dr Ailsa McLellan, Royal Hospital for Children & Young People, Edinburgh

10:55
Sleep issues in AHC and *ATP1A3* diseases
Dr Simona Balestrini, University College London and University of Florence

11:15 *(virtually)*
Treatment complexities in AHC and *ATP1A3* diseases: dystonia management
Professor Manju Kurian, Great Ormond Street Hospital, University College London

11:35 *(virtually)*
Treatment complexities in AHC and *ATP1A3* diseases: Flunarizine – to use or not to use?
Professor Masayuki Sasaki, Tottori University Japan

11:55
How can we create a clinical trial for AHC and *ATP1A3* diseases? *Learning from other rare diseases*
Professor Stéphane Auvin, Université de Paris

12:15
Panel discussion

12:25-13:35 *Lunch, poster viewing and networking*
Main Foyer, Conference Centre

Session 3:
Back to the lab
Chair: **Dr Steve Clapcote**
Lecture theatre and online

13:35
Rescue of Na/K-ATPase mutational effects by secondary mutation: Perspective for future pharmaceutical intervention in *ATP1A3* neurological disease
Professor Bente Vilsen, Aarhus University

13:55
Molecular mechanisms behind symptoms in *ATP1A* 3 and 1 mutations
Professor Anita Aperia, Karolinska Institutet

14:15
ATP1A3 expression: spinal cord/motor function
Professor Gareth Miles, University of St Andrews

14:35
Updates from the TREAT AHC research study: what drugs are being tried?
Dr Danilo Tiziano, Catholic University of the Sacred Heart, Milan

14:55 *(virtually)*
Possible future therapeutic target? The γ-Benzylidene Digoxin Derivative BD-15
Prof Leandro Barbosa, Universidade Federal de São João del-Rei

15:15
Panel discussion

15:15-15:30 *Coffee break, poster viewing and networking*

Session 4:
AHC and *ATP1A3* diseases: many facets, many needs
Chair: **Professor Helen Cross**
Lecture theatre and online

15:30
Introduction: The value of the Multi-Disciplinary Team (MDT)
Professor Helen Cross, Great Ormond Street Hospital, University College London

The need for an MDT to manage AHC – how should this be composed?
Discussion from clinicians involved in MDT management of AHC and ATP1A3 diseases on how their specialty can feed into the MDT at a local and national level

15:40
Cardiology, **Professor Juan Kaski**, Great Ormond Street Hospital, University College London

15:50 *(virtually)*
Gastroenterology, **Professor Mohamed Mikati,** Duke University

16:00 Speech and Language therapy, **Mr Steven Rose,** Great Ormond Street Hospital, London **16:10** Physiotherapy, **Dr Agnieszka Stępień,** University of Physical Education, Poland **16:20** Community Paediatrics/holistic palliative care, **Dr Helen Aspey,** Great North Children's Hospital, Newcastle **16:30** Pain Medicine**, Dr Suellen Walker**, Great Ormond Street Hospital, University College London **16:40** Respiratory, **Dr Don Urquhart,** Royal Hospital for Children & Young People, Edinburgh **16:50** Psychiatry, **Dr Boris Chaumette,** Reference Center for Rare Psychiatric Diseases Paris
17:00 Panel discussion: Standard of care of AHC patients and development of clinical consensus for AHC/*ATP1A3* diseases
17:20-17:30 Learning points from the day Closure

19:00: Scottish Welcome and Drinks reception in the New Library, Royal College of Physicians of Edinburgh

19:20: Formal three course dinner in the Grand Hall, Royal College of Physicians of Edinburgh

21:30-22:30: Coffee and drinks in the New library with music

22:30:00:00: Scottish Ceilidh

Friday 21st October 2022

Day 3: The Future for AHC/*ATP1A3* diseases, clinical practice, and research

08:00-08:30
Registration and coffee for delegates and speakers
Main Foyer in Conference Centre

08:30
Opening of Day 3 – Lived experience of AHC and *ATP1A3* diseases
Memorial for those with AHC or *ATP1A3* diseases who have died
Followed by recording from Filippo Franchini, parent and AISEA committee member
Lecture theatre and online

Session 1: Driving forward research and understanding in rare diseases: how can patients and families be involved?
Chair: **Katherine Behl**
Lecture theatre and online

09:00
Good Diagnosis: Improving the experience of diagnosis for people with rare conditions
Ms Natalie Frankish, Genetic Alliance UK

09:20
Patient-driven registries
Ms Isabella Brambilla, epiCARE patient rep and Dravet Syndrome registry co-ordinator

09:40 *(virtually)*
How to engage patients for faster transfer of research results to clinical practice
Claire Nolan, Head of Engagement, International Bureau of Epilepsy

10:00
Panel discussion

10:10-10:30 *Coffee break, poster viewing and networking*
Main Foyer in Conference Centre

Session 2:
Moving forwards: clinical trials
Chair: **Dr Ailsa McLellan**
Lecture theatre and online

10:30
A clinical scale for AHC/*ATP1A3* clinical trials
Dr Elisa de Grandis, University of Genoa

10:50
CBD in context in the management of rare epilepsies.
Professor Finbar O'Callaghan, Great Ormond Street Hospital, University College London

11:10
Panel discussion

Session 3:
Moving forwards: gene therapy strategies
Chair: **Professor Arn Van Den Maagdenberg**
Lecture theatre and online

11:20
Learning from other neurological diseases – progress in gene therapy
Professor Mimoun Azzouz, University of Sheffield

11:40 *(virtually)*

AAV9-mediated *ATP1A3* gene therapy: an update **Professor Cat Lutz,** Jackson Laboratory
12:00 *ATP1A3* gene editing: Using CRISPR for *ATP1A3* diseases **Mr Alexander Sousa,** Harvard University
12:20 Antisense oligonucleotide therapy: a possible target for AHC/*ATP1A3* diseases **Professor Al George,** Northwestern University
12:40 Panel discussion
12:50 Prize for best poster
12:55 Closure of conference, summary and key highlights of the conference and consensus on targets for future research **Summary by researcher, clinician, and patient organisation representative**
13:10 Lunch *Main Foyer, Conference Centre*