# The 2024 International workshop on Alport Syndrome

**The Royal Hall, Xanthis Xenierou, Nicosia 1015**

## Nicosia, Cyprus

**14-16 March 2024**

**Generously sponsored by:**

Event Sponsor

REGENERON

Patient Perspective sponsor

ENYO PHARMA

Education sponsors

calliditas THERAPEUTICS

Otsuka | Visterra Otsuka Group of Companies

Also sponsored by

Kidney Research UK

Nefrontida Nephrology Center

C. Georgiou Lab Supplies

Biotronics

Supported by

TRAVERE THERAPEUTICS

STONEYGATE TRUST

ERKNet The European Rare Kidney Disease Reference Network

Alport Syndrome Alliance
A global network advancing treatments and knowledge

# Detailed agenda and list of speakers

---

**Emergency contact or logistics:**

**Jayne Perrin**
**+44 7595 942850**

**Workshop organisers:**

alport uk
a brighter future for people living with alport syndrome

biobank.cy Center of Excellence in Biobanking and Biomedical Research

REPUBLIC OF CYPRUS

University of Cyprus Medical School

The biobank.cy Center of Excellence in Biobanking and Biomedical Research is supported through the *CY*-Biobank project, which receives funding from the European Union's Horizon 2020 research and innovation program, under Grant Agreement number 857122, the Government of the Republic of Cyprus, and the University of Cyprus.

Alport UK is a founder member of the Alport Syndrome Alliance and currently drives the activities, such as organising the international workshops, until the alliance is fully operational and sustainable.

# Welcome

## Welcome to this workshop - the seventh in a series of immersive conversations - for the Alport community

Alport syndrome (AS) was named after Cecil A. Alport, who published his observations in the British Medical Journal in 1927 connecting kidney and hearing issues and later described in his book 'On Nephritis' in 1929. The eye problems were described later. Sam Clarke's inspiring short video #fightingfailure (https://youtu.be/1A1adCj-cvo) captures the rollercoaster journey of living with an isolating rare inherited condition - Alport Syndrome - that impacts many members of a family.

Groups of international researchers working on Alport in the 1960s, 70s, 80s and 90s (1990s work funded by the European Commission) demonstrated the power of collaboration with their work to progress the understanding of Alport syndrome. National patient organisations emerged, set up scientific advisory groups and raised vital funds for research in USA (Alport Syndrome Foundation), in France (AIRG France), and Italy (ASAL Onlus). Initially there were not enough research projects applying for funding. The inspiration, valuable publications and vital outputs from the various research and patient collaborations inspired the current workshop series. This in person workshop is the seventh workshop in a series of informal, immersive workshops. The previous workshops were 'in-person': Oxford, UK (2014), Göttingen, Germany (2015), Glasgow, UK (2017, Siena, Italy (2019), on-line (2021) and 'in person' Calgary, Canada (2023). Highlights from The 2019 International workshop on Alport Syndrome, Siena University, October 2019: https://youtu.be/QH8mDTmKaVU. The Genetic Variants Group, also met for a workshop in London (February 2020) just as the pandemic was taking hold across the world. Many contributors to the Genetic Variant Group are now members of expert panels within the ClinGen gene and variant curation programme, which provides an opportunity to improve variant classification for Alport syndrome and to consider disease naming within a broader context of genetic conditions.

**Despite the pandemic, more people engaged**

The fifth workshop was supposed to be in Beijing, China but was necessarily postponed because of COVID-19. However, the pandemic was an opportunity for the community to connect online instead and engage a wider more diverse community. With the inspiration and leadership from the Lennon and Miner labs and a host of patient volunteers a further series of 32 Alport online workshops featured the latest research from labs all over the world. These online workshops were moderated by young adults living with Alport Syndrome. What was so inspiring was that many of the Alport volunteers in each country personally worked on the frontline during the pandemic and kept people safe.

**Volunteering and results of the contributions**

Everyone who participates in these workshops is a volunteer and contributes their own time, on top of student studies, 'day' jobs and busy lives. The people living with Alport syndrome are incredibly grateful to everyone who contributes this time, energy and inspiration. We welcome new volunteers to the community to contribute on all aspects of the work and enable connections with an even wider international community. We should all feel very proud of the outcomes from this vibrant collaborative community – the numbers speak for themselves:

- **First workshop in 2014 in Oxford, UK**: 75 participants from 17 countries with 34 abstracts, but no pharmaceutical or commercial companies involved.
- **Fourth workshop in 2019 in Siena, Italy**: 146 participants, including 36 patients and representatives from 12 pharmaceutical companies plus 46 posters. A survey of committee members and teams celebrated: 131 publications, 16 book chapters and 38 grants – over 90% were collaborative efforts.
- **Fifth workshop online in 2021:** Over 220 registrations from 26 countries, of which a third are patients and featuring 40 posters and over 60 people contributing to the workshop this week either as presenters or moderators.
- **Seventh workshop in Cyprus in 2024:** Over 140 registrations from 19 countries, including 39 patients, with over 60 abstracts. Many pharmaceutical representatives will participate as their companies are advancing treatments, doing pre-clinical studies, or are planning/delivering clinical trials.

**What are the important ingredients in this community that make it work?**

- the strong leaders – each experts in their own right – that collaborate and find ways to work together internationally, sharing ideas and resources, avoiding unnecessary ego and hierarchy (you will note we do not use titles), in our drive to advance treatments and knowledge that creates a brighter future for people living with Alport syndrome
- the mix of diverse perspectives, volunteering and offering to contribute at many different levels, means we look at the challenges and opportunities from different angles. The perspectives come from people living with Alport syndrome, clinicians, pathologists or research scientists (basic science, genetics, kidneys, hearing, eyes), pharmaceutical or commercial company representatives, epidemiologists and more. These perspectives are also from early career people through to those with more experience.
- the personal choice, drive and motivation of individual leaders and teams to engage, volunteer or scope projects, get funding and deliver research to build knowledge and advance treatments.
- the sense of 'extended family' or 'virtual collaborative' structure, which bonds us together, means we watch out for each other, our families, our lives, our careers.
- even funding these workshops is a collaborative effort; the workshop this year is a result of UK fundraising and international sponsorship. We are very grateful this year to:
    - the organisers: who contribute huge amounts of time, energy and their own funding: **Alport UK** and **University of Cyprus' Biobank**. The biobank.cy Center of Excellence in Biobanking and Biomedical Research is supported through the CY-Biobank project, which receives funding from the European Union's Horizon 2020 research and innovation program, under Grant Agreement number 857122, the Government of the Republic of Cyprus, and the University of Cyprus.

- the very generous sponsors of The 2024 International workshop on Alport Syndrome: **Regeneron**, **Enyo Pharma**, **Calliditas**, **Otsuka**/**Visterra**, **Kidney Research UK**, **C. Geourgiou Lab Supplies**, **Biotronics Ltd, Nefrontida** and
- the very generous supporters: **Stoneygate Trust** and **Travere Therapeutics**.

**How to get the most out of the workshop**
Those who participated in our five 'in-person' workshops will know that the 'immersive' element of the workshop was because we 'locked the doors', everyone sat in a circle and contributed to the workshop discussions and asked questions. Asking questions is the key to developing knowledge and advancing treatments.

**Topics to cover in the next workshop**
There will be some topics missing from this workshop. We are not trying to cover everything. Let us know what topics are missing and we will design more specific online workshops on these topics next year to give the additional topics the focus they need. We will carry on the discussions after the workshop too, aiming to produce publications, more guidelines, further define what the Alport Syndrome Alliance needs to do to continue the process for this global network to advance treatments and knowledge.

We look forward to the conversations. Thank you for participating. Enjoy this week as we shine a light on Alport research.

Susie Gear, Constantinos Deltas, Hannah Russell

On behalf of the Workshop Organising Committee
**The 2024 International workshop on Alport Syndrome**
Administrator: Jayne Perrin

**Organising Committee for The 2024 International workshop on Alport Syndrome:**
Dr Marina Aksenova, Veltischev Research and Clinical Institute for Pediatrics of the Pirogov Russian National Research Medical University, Moscow, Russia
Professor Colin Baigent, MRC Scientist, Hon Consultant in Public Health, Oxford, UK
Assistant Professor Moumita Barua, Toronto General Hospital, Toronto, Canada
Dr Agnė Čerkauskaitė, Division of diagnosis and treatment of Rare Kidney and Metabolic Diseases, Nephrology Center, Vilnius University Hospital Santaros Klinikos, Vilnius, Lithuania
Professor Constantinos Deltas, School of Medicine, University of Cyprus and Director of biobank.cy Centre of Excellence in Biobanking and Biomedical Research, Nicosia, Cyprus
Professor Jie Ding, Department of Pediatrics, Peking University First Hospital, Beijing, China
Professor Frances Flinter, Emeritus Professor of Clinical Genetics, Guy's and St Thomas' NHS Foundation Trust, London, UK
Professor Daniel Gale, Department of Renal Medicine, University College London, UK
Professor Danica Galešić Ljubanović, Department of Pathology University of Zagreb School of Medicine, Head of Department of Nephropathology and Electron Microscopy, Dubrava University Hospital, Zagreb, Croatia
Professor Oliver Gross, Department of Nephrology and Rheumatology, Georg-August-Universität Göttingen, Germany
Professor Julia Höfele, Institute of Human Genetics, Munich, Germany
Professor Hirofumi Kai, Graduate School of Pharmaceutical Sciences, Kumamoto University, Japan
Professor Clifford Kashtan, Department of Pediatrics, University of Minnesota, USA
Professor Bertrand Knebelmann, Necker Hospital, Paris, France

Associate Professor Ron Korstanje, The Jackson Laboratory, Bar Harbor, USA
Professor Rachel Lennon, Academic Paediatric Nephrologist University of Manchester, UK
Dr Laura Massella, Division of Nephrology, , Bambino Gesù Children's Hospital - IRCCS, Rome, Italy
Professor Julian Midgley, Department of Paediatrics, Alberta Children's Hospital, Canada
Professor Jeffrey Miner, Washington University in St. Louis, USA
Professor Laura Perin, Saban Research Institute, University of Southern California, USA
Professor Judith Savige, University of Melbourne, Australia
Professor Alessandra Renieri, Professor of Medical Genetics, Director of Medical Genetics Unit, Azienda Ospedaliera Universitaria Senese viale Bracci 2, Siena, Italy
Professor Michelle Rheault, Department of Pediatrics, University of Minnesota, USA
Professor Roser Torra, Fundació Puigvert, Barcelona, Spain
Professor Neil Turner, Professor of Nephrology, University of Edinburgh and Consultant Nephrologist, Royal Infirmary of Edinburgh, UK

Patient representatives:
Dave Blatt, Alport Foundation of Australia
Maria José Cacharron, Spain
Christof Finkler and Ute Rosenthal, Alport Selbsthilfe, Germany
Susie Gear and Hannah Russell, alport uk
Mario Kolonić, Croatia
Julia Schifter, Alport Foundation Israel
Renee de Wildt, NierpatiëntenVerenigingNederland (NVN), The Netherlands
Andre Weinstock, USA
Jessie Zhang and Mr Cai, Chinese patient group, China

**Local Organising Committee**
Professor Constantinos Deltas
Dr Nicos Mitsides
Dr Gregory Papagregoriou
Dr Apostolos Malatras
Dr Christoforos Odiatis
Dr Stavroula Louka
Dr Eirini Moutsouri
Pavlos Ioannou, PhD Cand.
Ms Marina Constantinou
Ms Emily Groutidou Petridou
Ms Ioanna Christodoulou

# The 2024 Cecil Alport Awards

**Recognising achievement in researching Alport Syndrome**

1. **The best poster on Alport diagnosis**
2. **The best poster on Alport management**

# Workshop objectives and design principles for the workshop

## Workshop objectives

- **Understand the latest findings and new ideas – hearing, kidneys, basic science investigations into treatments, eyes, clinical trials and patient registries**
- **Collaborate with international colleagues in sessions on specific topics, e.g. basic science, genetics and gene editing, clinical trials**
- **Present your own research or case studies**
- **Hear patient perspectives**
- **Network with experts in related fields or related disease areas**
- **Be part of the future of Alport syndrome research and treatment.**

---

## Principles for the design of the workshop

The workshop is run as a 'lock-in' format so participants are expected to be present in the room, throughout the workshop, and for the evening activities that the Cypriot team have kindly planned for the evenings of Thursday and Friday.

It is an 'international workshop', so speaking slots are limited to allow as much time for **dialogue and discussion**. We very much welcome participation by those who are not presenting either in plenary or posters; these participants have a key role to encourage questions and deeper debate.

The Workshop Organising Committee reviewed abstracts and allocated the topics to be presented in different ways:

| *Duration* | *Type of contribution* | *Role and format* |
|---|---|---|
| 30 mins | **Keynote presentations** | Invited speakers - 25-minute talk, 5 mins Q&A |
| 15 mins | **Patient presentations** | Agreed by patients following patient meeting contributions - 10-minutes talk, 5 mins Q&A |
| 15 mins | **Priority abstracts presented** | Highest scoring abstracts invited to present - 10 mins talk, 5 mins Q&A |
| 10 mins | **Next priority of abstracts presented** | Next set of abstracts invited to present - 6 mins talk, 4 mins Q&A |
| 5 mins | **Quick-fire' presentation of abstracts** | Rest of the abstract authors invited to present 1-3 slides with key messages. 3 mins talk, 2 mins Q&A, plus invited to bring a poster |
| | **Chairs for each plenary section (in pairs)** | Introduce the topic - summarise what we do know, key concepts and key questions to answer<br><br>Summarise the topic - pull the threads together, raise additional questions or capture ideas to follow up<br><br>Facilitate the write up – lead the write up of this topic for the post-workshop publication, inviting input from as many who would like to contribute including one presenter to write a piece for their section |

---

## Social Media Policy

---

**Purpose:**

- We are keen to have a social media 'buzz' around the meeting to help get other patients and researchers interested. See the opportunity to connect in with World Kidney Day.
- Maintain confidentiality of research data while also sharing information of Alport syndrome advances with the broader scientific community and public.

1. Participants are encouraged to share their experience and photos on social media during the meeting using the hashtag **#AlportWorkshop**
2. On World Kidney Day on **Thursday 14 March**: 1) Wear your boldest, most colorful socks. 2) Photograph your socks! Either solo or in a pair, snap a pic of your socks to share. 3) Post to Twitter and/or Instagram with hashtags **#sockittokidneydisease** and **#WKD**.
3. Do not share photos or information about unpublished work without permission from the presenter
4. Do not share photos of patients without their consent
5. Do not share photos of slides or posters without the permission of the presenter

# Detailed agenda



## Wednesday 13 March

| | | |
|---|---|---|
| 7.00pm | Light dinner after patient day – *for those participating in the patient day* | **Artnaldas**<br>Address: 2 Arnaldas str. 1060, Nicosia |

## Thursday 14 March

| | | | |
|---|---|---|---|
| 8.30am | | | Registration till 9.00 |
| **9.00** | Chairs: | **Constantinos Deltas and Susie Gear** | **Plenary 1: Check in** |
| | | Every participant | **Check in individually**<br>a. Your name, your country and your role/interest in Alport syndrome<br>b. 2-4 words: Last 10 years: greatest achievement in Alport syndrome?<br>c. 2-4 words: Next 10 years: What one outcome you'd like?<br>**Please keep your answers very short** |
| | | Constantinos Deltas | **Welcome. Switching to Alport research: our journey in Cyprus** |
| | | Susie Gear | **Principles of the workshop, health and safety Agenda** |
| | | Susie Gear<br>*Video* | **Celebrating 10 years of community, research and collaboration** |
| **10.30** | | Break | |

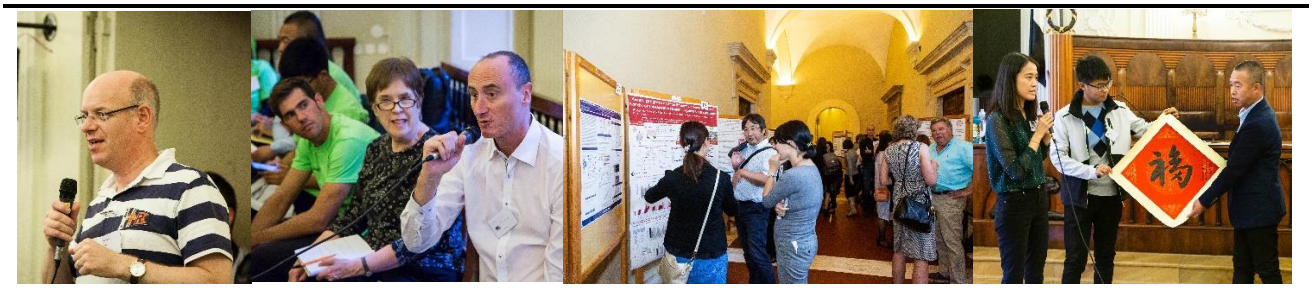
| | | | |
|---|---|---|---|
| **10.50** | | Fabrice Huré - *video* | **The mountain in his blood: living with Alport syndrome -** Mon mantra: *"Se dialyser la nuit pour revivre le jour!"* |
| | | Cyprus welcome | **Living with a rare disease in Cyprus**<br><br>**Emily Petrides**, Vice-President of the European Kidney Patients' Federation (EKPF), President of the Pancyprian Organization of People with Kidney Disorders, Board Member of the Cyprus Transplantation Council: Living with Polycystic kidney disease<br>**Constantinos Yiorkadjis**, Mayor of Nicosia<br>**Prof. Tasos Christofides**, University of Cyprus Rector<br>**Prof. Georgios Hadjigeorgiou**, University of Cyprus Dean of Medical School<br>**Dr Petros Agathangelou**, President of the Cyprus Medical Association<br>**Dr Nicos Mitsides**, President of the Cyprus Renal Association |
| **11.30** | Chairs: | **Moumita Barua, Judy Savige and Danny Gale** | **Plenary 2: Diagnosis: Naming and genotype/phenotype** |
| | | | Introduction |
| | | Mary-Beth Roberts | **Type IV collagen variant curation project** |
| | | Constantinos Deltas | **D7: Collagen IV nephropathies: The landscape of genetics research and molecular diagnosis in Cyprus** |
| | | Beata Lipska-Ziętkiewicz | **X inactivation, mosaicism, de novo and the significance of COL4A5 in girls** |
| | | Guillaume Dorval (Recorded) | **Deep intronic variants – what can we learn?** When panel testing is negative, and there is a phenotype then moving to urine to do RNA testing. |
| **1.00** | | | Lunch (Buffet) and networking |

| | | | |
|---|---|---|---|
| **2.00** | Chairs: | **Moumita Barua, and Danny Gale** | **Quickfire poster session 1**<br>Introduce rules: 'Quick fire' – 3+2 mins each |
| | | Eirini Moutsouri | **M20: Preliminary analysis of whole exome sequencing from 106 patients with hematuria and negative for COL4A3/A4/A5 pathogenic variants** |
| | | Konstantinos Tzoumkas | **D5: Identifying Genetic Contributors to Haematuria in the UK Biobank** |
| | | Judy Savige | **D10: Genetic kidney disease in familial and sporadic IgA nephropathy** |
| | | Christiana Polydorou | **D3: A COL4A4 novel founder variant causes Alport spectrum nephropathies in Cypriot families** |
| | | | Summary |
| **2.30** | Chairs: | **Danny Gale and Constantinos Deltas** | **Plenary 2: Diagnosis continued** |
| | | | Introduction |
| | | Moumita Barua | **Genetic testing in focal segmental glomerulosclerosis: in whom and when?** |
| | | Roser Torra (Recorded) | **Significance of being a 'carrier' of COL4A4 and A3 -** hottest issue in inherited kidney disease. Is it a risk factor? What should we call it? How should we manage it? Affects so many in the population. |
| | | *Discussion* | **Variants of uncertain significance –** agree guidelines for further tests |
| | | Judy Savige | **D11: AD Alport syndrome is much more common than other genetic kidney diseases that result in haematuria and proteinuria** |
| | | | Summary |
| **3.10** | | Break | |

| | | | |
|---|---|---|---|
| **3.30** | Chairs: | **Constantinos Deltas, Beata Lipska-Ziętkiewicz, Christoforos Odiatis, Stavroula Louka** | **Quick fire poster session 2**<br><br>Introduce rules: 'Quick fire' - 3+2 mins each |
| | | Ivana Trutin<br>*Presented by Tamara Nikuševa Martić* | **D1: Is it Alport syndrome or not?** |
| | | Guy Neild | **D2: Autosomal dominant kidney disease phenocopying hypertensive nephropathy in Turkish Cypriot Families** |
| | | Yaochun Zhang<br>*Presented by Kar Hui Ng* | **D6: Pathogenic Alport variants are more common in Chinese than Malays in the Singapore population** |
| | | Dipti Rao | **D13: Disease severity in patients with heterozygous COL4A3 or COL4A4 pathogenic variants and exploration of risk factors** |
| | | Lori Morton | **D24: Integrative Strategies for Chronic Kidney Disease Drug Discovery** |
| | | Matija Horaček | **D9: Comparison of patients with thin basement membrane nephropathy associated with focal segmental glomerulosclerosis to patients who have only thin basement membrane nephropathy** |
| | | Nikola Zagorec<br>*Presented by Danica Galešić Ljubanović* | **D8: Thin glomerular basement membranes in patients with idiopathic focal segmental glomerulosclerosis – meaning and the impact on prognosis?** |
| | | | Summary |

| | | | |
|---|---|---|---|
| **4.10** | Chairs: | **Billy Hudson and Rachel Lennon** | **Plenary 3: Building and maintaining basement membranes** |
| | | | Introduction |
| | | André Weinstock<br>(Live online) | **Alport Syndrome Foundation updates from the USA: Capitalism, Commercialism, and Clinical Trials** |
| | | Douglas Gould | **Establishing foundations for mechanism-based and mechanism-agnostic interventions for variants in COL4A1/A2 genes** |
| | | Brian Stramer | **Basement membrane assembly, stability and turnover in *Drosophila*** |
| | | David Sherwood | **Basement membrane linkage in *C. elegans*** |
| | | | Summary |

| | | |
|---|---|---|
| **6.10** | Buffet dinner and poster viewing | Dinner in Royal Hall – *please pick up food so you can eat whilst viewing the posters* |
| **8.15pm** | Walk to Freedom Square which is lit up for World Kidney Day till 9pm<br>*Square is designed by Zaha Hadid*<br>  | **Workshop group photo** - by photographer Dinos - in the Square in socks!<br><br>**Join the #SockItToKidneyDisease campaign to raise awareness about kidney disease.**<br>Participate by:<br>1. On March 14th, wear your boldest, most colorful socks.<br>2. Take a photo by yourself or with a friend, showcasing your funky socks.<br>3. Post a photo on social media with **#SockItToKidneyDisease**<br><br>*Thanks to Rachel Lennon for sample photo from a previous year.* |



## Friday 15 March

**8.30** Chairs: **Rachel Lennon and Neil Turner**

### Plenary 3 *continued*: Building and maintaining basement membranes

| | |
|---|---|
| | Introduction |
| Billy Hudson | **D16: Alport syndrome: Solving the mystery in pursuit of therapy** |
| Jeffrey Miner | **D4: Quantitative Assessment of the GBM's Collagen IV Networks in Paraffin Sections from Patients with FSGS Lesions and Alport Gene Variants** |
| Danica Galešić Ljubanović | **Update on the role of pathology in diagnosis** |
| Rebecca Preston - *Presented by Rachel Lennon* | **D17: Kidney matrix proteins have altered dynamics in Alport syndrome** |
| Ko Tsutsui | **D18: Localisation of basement membrane proteins in the developing mouse kidney, eye and cochlea** |
| Vanessa De Gregorio (Recorded) | **D19: Dysregulated glomerular proteins in 1-day old Col4a3 knockout mice** |
| | Summary |

**10.00** Break

**10.30** Chairs: **Jeff Miner and Laura Perin**

### Plenary 4: Glomerular disease and basic science

| | |
|---|---|
| | Introduction |
| Tobias Huber | **New avenues to target Alport syndrome and acquired pediatric kidney diseases** |
| Hirofumi Kai | **A new molecule and how it works** |
| | Summary |

**11.30** Chairs: **Danica Galešić Ljubanović and Jeff Miner**

### Quick fire poster session 3

Introduce rules: 'Quick fire' 3+2 mins each

| | |
|---|---|
| | Introduction |
| Shota Kaseda | **D14: Efficacy of Keap1-Nrf2 protein-protein interaction inhibitor for progressive phenotype in mouse model of Alport syndrome** |
| Pavlos Ioannou | **M13: Evidence that chaperone 4-PBA treatment alleviates the renal phenotype in Alport syndrome mouse models** |

| | |
|---|---|
| Lorna Milne | **D15: Deciphering the role of the glycocalyx in Alport Syndrome and pregnancy** |
| Laura Perin | **D20: Spatial transcriptomic profiling of glomeruli in Alport syndrome** |
| Kishor Devalaraja-Narashimha | **D23: A Comprehensive Transcriptomic Analysis of the Col4a3 Knockout Mouse Model of Alport Syndrome** |
| Sargis Sedrakyan presented by Laura Perin | **D21: The role of glomerular endothelial lipid metabolism in Alport syndrome** |
| Emine Bilge Caparali | **D22: Sex differences in glomerular protein expression in mice with autosomal recessive Alport syndrome** |
| Christoforos Odiatis | **M14: New Insights into mouse podocyte biology in glomerular health and Alport disease** |
| | Summary |

| | | |
|---|---|---|
| **12.30** | Buffet lunch and poster viewing | Lunch in the Royal Hall - please pick up food so you can eat whilst viewing the posters |

| | | | |
|---|---|---|---|
| **1.30** | Chairs: | **Oliver Gross, Julian Midgley and Matt Hall** | **Plenary 5: Clinical** |
| | | | Introduction |
| | | *Giulia Nutile Presenting on behalf of Italian Patient* Margherita Venturini | **ASAL listening desk** **M23: The importance of establishing a network for Alport patients: "Rete_Alport" project for accessible assistance throughout Italy** |
| | | | |
| | | Danny Gale | **So I have found a change in an Alport gene. What do I tell the patient?** |
| | | Stavroula F. Louka | **M15: MiRNA profiling of urinary extracellular vesicles (uEVs) in TBMN patients for the identification of novel disease biomarkers** |
| | | Dan Jagger | **Hearing –** update on things related to hearing. How to progress hearing research? |
| | | Irina Marcovich | **M18: Characterization of murine and human models to study Alport Syndrome pathogenic mechanisms in the inner ear** |
| | | Laith Al-Rabadi (Recorded presentation) | **M25: Assessing Alport syndrome and Thin Basement Membrane Nephropathy using Optical Coherence Tomography (OCT)** |
| | | | Summary |
| **3.00** | | Break | |
| | | | *Transition to* **Management: Pre-clinical and clinical studies, gene and drug therapies, clinical trials and registries** |
| **3.20** | Chairs: | **Neil Turner, Laura Massella and Judy Savige** | **Plenary 6: Management and treatments** |
| | | | Introduction |
| | | Hannah Russell and Heidi Zealey | **Patient journey: introduce ErkNET work on journeys** - feedback (send out as pre-read?) |
| | | Kar Hui Ng | **Alport syndrome in low resource countries** |
| | | Beata Lipska-Ziętkiewicz | **Update on ErkNET international treatment guidelines** – report from ErkNET Clinical guidelines project and where next? |

| | | | |
|---|---|---|---|
| | | | |
| | | Tom Oates | **Who to consider to test and how follow up/manage those with positive tests?** (ASN talk) |
| | | | |
| **4.50** | Chairs: | **Danica Galešić Ljubanović and Jeff Miner** | **Quick fire poster session 4**<br><br>Introduce rules: 'Quick fire' 5 mins each |
| | | | Introduction |
| | | Marina Aksenova | **M21: Aortic root dilation in boys with X-linked Alport Syndrome: Prevalence and risk factors** |
| | | Shaun Wright | **M12: Investigating the relationship between kidney disease and central nervous system homeostasis in a mouse model of Alport syndrome** |
| | | Laura Lucchetti | **M26: Cognitive profile assessment, adaptive functioning and emotional-behavioural aspects in a cohort of X-Linked and autosomal Alport syndrome patients** |
| | | Emmanuel Oduware (Recorded) | **M24: Proposal for the standardisation of national clinical guidelines for the diagnosis and treatments of suspected Alport syndrome and hereditary kidney diseases in resources challenge settings: Nigeria as a case study** |
| | | Mary Nabity | **M17: Detection of Podocyturia in Dogs with X-Linked Alport Syndrome** |
| | | Xuhui Zhong | **China perspective** |
| | | Mónica Furlano | **M27: Comparative analysis of proteinuria extent in autosomal dominant and X-linked Alport syndrome** |
| | | Valentine Gillion | **M2: Impact of thiazides on proteinuria reduction in young patients with Alport syndrome** |
| | | Jan Boeckhaus | **M3: High prevalence of hypercholesterolemia in patients with Alport syndrome at early stages of kidney disease correlates with albuminuria** |
| | | Yanqin Zhang | **M11: Course of kidney disease in Alport syndrome during and after pregnancy** |

| | | |
|---|---|---|
| | Marina Aksenova | **M22: Pregnancy and fetal outcome in women with Alport syndrome** |
| | Margriet Gosselink | **M28: Reassuring pregnancy outcomes in women with mild COL4A3-5 related disease (Alport Syndrome): genetic type of disease can aid personalized counseling** |
| | Fang Wang | **M4: How to treat prominent proteinuria mismatching with disease course in the patients with Alport syndrome** |
| | Carlotta Caprara | **M19: ADPKD and Collagen genes (COL4A3, COL4A4, COL4A5)** |
| | | Summary |
| **6.50** | Close | |

| | | |
|---|---|---|
| **7.00** | Tour round Nicosia<br>*Meet outside Royal Hall* | Partners and family welcome to join us. Free for participants, but we charge a small fee to cover tour and food for any extra people. Please pay in cash to Jayne Perrin. |
| **8.00** | Drinks, Gala dinner and awards at Venue | **Odofragma - Traditional Tavern**<br>Address: Andrea Kariou, Agios Dometios<br>*10 mins drive from Nicosia – Please meet bus at 8pm outside Royal Hall* |
| **10pm** onwards | | After dinner, buses available to return to Nicosia at 10.00 and 11.00pm. |

alportWARRIORS

www.alportuk.org

**The Workshop Organising Committee would like to thank the many**

alportWARRIORS

**on Facebook for their openness to constructively help each other, suggest ideas for research and being the inspiration for the series of International workshops on Alport Syndrome**

alport uk

a brighter future for those living with alport syndrome

## Saturday 16 March

**8.00** Chairs: **Rachel Lennon, Hirofumi Kai and Fang Wang**

### Plenary 7: Treatments

| | |
|---|---|
| | Introduction |
| Neil Turner | **Innovative new treatments and mechanisms and how to test them**. How to identify more rapid progressors. Different phases of how kidneys impacted. |
| Hans-Joachim Anders | Update on combined therapy trials in mice - **a triple therapy for Alport mice that increased their lifespan**. |
| | Discussion |
| Yupeng Chen | **Gene therapy:** engineering MiniCol4α5 for gene therapy in mice with X-Linked Alport syndrome |
| Hiroyuki Nakai | **M8: Therapeutic Efficacy of AAV Gene Therapy in a Novel Alport Syndrome Mouse Model** |
| Pinyuan Tian | **M16: Repairing Glomerular Basement Membrane Defects in Alport Syndrome** |
| Vassili Valayannopoulos | **M9: CHORD: A Phase 1/2 Open-Label, Multi-Center Trial to Evaluate Intracochlear Administration of DB-OTO Gene Therapy in Pediatric Patients with Profound Sensorineural Hearing Loss Due to Biallelic Otoferlin Mutations** |
| Mary Nabity | **M7: (AAV)2-CRISPR/Cas9 Gene Therapy in Dogs with X-Linked Alport Syndrome** |
| | Summary |

**10.00** Break

| | | | |
|---|---|---|---|
| **10.30** | Chairs: | **Julian Midgley and Neil Turner** | **Plenary 8: Registries and clinical trials** |
| | | Patient: Ryan Wicks | Personal story about participating in a trial |
| | | Danny Gale | **How RaDaR can catalyse development of treatments for rare kidney diseases**<br>**Shaping a natural history study** - Where do we start? What do clinical trials need? |
| | | Jan Boeckhaus | **M1: SGLT2-Inhibition in patients with Alport syndrome – an international, retrospective and in parts prospective observational large cohort study** |
| | | Oliver Gross | **SGLT2 treatments**. Update on Alport specific trial. E.g. present protocol and initial findings of interventional trial using SGLT2 inhibitors to treat older children with proteinuria. |
| | | Louise Oni (Recorded) | **Opportunities for clinical trials in children and the UK kidney ecosystem** |

| | | | |
|---|---|---|---|
| **11.45** | Chairs: | **Julian Midgely and Neil Turner** | **Quick fire poster session 5**<br>Introduce rules: 'Quick fire' 6+4 mins each |
| | | | Introduction |
| | | Anna Polczyk-Boron | **Finerenone: FIONA trial** |
| | | Richard Phillipson | **M6: Design of a Phase 2a study on the safety and efficacy of the NOX1/4 inhibitor setanaxib in patients with Alport syndrome** |
| | | Pietro Scalfaro | **M10: ENYO Pharma is currently developing VONAFEXOR, an antifibrotic and anti-inflammatory drug, in the Phase 2 "ALPESTRIA-1" clinical study for Alport syndrome.** |
| | | Sumit Aggarwal<br>*Presented by Danny Gale* | **Eloxx Trial update** |
| | | Fang Wang | **Update on Metformin trial in children** |
| | | Discussion | How can companies share placebo data to help our small community of patients? What placebo data is needed? |
| | | | Summary |

| | | | |
|---|---|---|---|
| **12.45** | Chairs: | **Constantinos Deltas, Moumita Barua, Jeff Miner, Rachel Lennon, Danny Gale, Tom Oates and Matt Hall** | **Plenary 9: Next steps, write up and future plans**<br>Introduction - Which publication? Who does want to get involved with a publication?<br>Summary of highlights, key questions to answer in research going forward, priorities, next steps, write-up, and future plans |
| 1.15 | | All | Check out and feedback forms |
| **1.30** | | Close and Lunch | |
| 2.30 | | Members of the Committee | **Workshop Organising Committee meeting** |
| **7.30** | | Light dinner | **Da Paolo**<br>Konstantinou Palaiologou 52, Nicosia 1015 |



**The Workshop Organising Committee would like to thank**

# Kidney Research UK,

# Stoneygate Trust and

# Travere Therapeutics

**for supporting**

**The 2024 International workshop on Alport Syndrome**