



European
Aniridia
Conference

31.05. - 02.06.
STOCKHOLM 2024



7th

**EUROPEAN ANIRIDIA
CONFERENCE**

scientific program



Aniridi Sverige



LINKÖPING
UNIVERSITY



ANIRIDIA EUROPE

Table of content

Welcome message	4
Program	6
Day one - 31 May	6
Day two - 01 June	8
Day three - 02 June	12
Speakers and abstract	14
Opening ceremony, welcome and introduction	14
Opening keynote lecture	17
Glaucoma and aniridia	18
New research areas in aniridia	20
Artificial iris, cataract, posterior segment	23
Keratopathy in aniridia	24
Stem cells and aniridia	28
Genetics and aniridia	31
Management of anirida and challenges for children	34
New therapies	38
Popular summary for patients and associations	42
Closing ceremony and acknowledgements	42
Practical A-Z	46
Exhibitors	48

Welcome to the 7th European Aniridia Conference!

It is with great pleasure and profound gratitude that I extend my warmest welcome to each of you gathered in Stockholm for the 7th European Aniridia Conference. As President of Aniridia Sweden, it brings me immense joy to see our long-held dream materialize into reality with the fruition of this remarkable event.

In less than two decades since the foundation of Aniridia Sweden, the efforts made in understanding of the complexities of aniridia as well as the development of new therapies, have been nothing short of extraordinary. The 7th European Aniridia Conference stands as a testament to our strong commitment to advancing knowledge and fostering collaboration within both the scientific community as well as the European aniridia community.

Over the next few days, we are poised to embark on an inspiring journey filled with inspiring scientific presentations, compelling narratives, and invaluable exchanges of knowledge and ideas. I am confident that this conference will catalyse progress, ignite fresh perspectives, and contribute to innovative solutions that will improve the life quality of individuals living with aniridia.



I would like to express my sincere gratitude to everyone that has over the last couple of years helped us make this event a reality. Without their strong commitment, generosity, and tremendous engagement the 7th European Aniridia Conference would not have been possible to arrange in Sweden.

May the days ahead be filled with the excitement of discovery, the joy of fellowship, and the promise of new collaborations. Let's make this conference a wonderful experience, driving progress for those affected by aniridia.

Welcome!



Ivana Kildsgaard
- President of Aniridia Sweden

31 May

Friday

DAY ONE

- 10.00 ● Registration opens
- 12.00 - 13.00 | Lunch
- 13.00 - 13.30 ● Opening ceremony, welcome and introduction
Scientific committee
Prof. **Neil Lagali**, Linköping university, Sweden;
Berit Byström, MD, Docent, University Hospital of Umeå, Sweden;
Branka Samolov, MD, PhD, S:t Erik eye hospital, Sweden;
Aniridia organizations
Barbara Poli, President of Aniridia Europe;
Ivana Kildsgaard, President of Aniridia Sweden;
Welcome speech
Beatrice Peebo, MD, Assist. Prof., Clinic Manager, S:t Erik eye hospital, Sweden
- 13.30 - 14.00 ● Opening keynote lecture
“Dry eye disease – myths and facts”,
Prof. **Tor Paaske Utheim**, MD, Oslo university hospital, Norway
- 14.00 - 15.00 ● Session: Glaucoma and aniridia
Chairs: Erlend Christoffer Sommer Landsend & Dominique Bremond-Gignac
“Glaucoma in congenital aniridia. Clinical Considerations and Management”,
Amelie Botling Taube, MD, PhD, S:t Erik eye hospital, Sweden;
“Surgical management of aniridic glaucoma”,
Prof. **Peter A. Netland**, MD, University of Virginia School of Medicine, USA
- 15.00 - 15.30 | Coffee break

- 15.30 - 17.00 ● Session: New research areas in aniridia
Chairs: Peter Netland & Tor Paaske Utheim
“Aniridia and the brain”,
James Lauderdale, Assoc. Prof., University of Georgia, USA;
“Health-related quality of life in congenital aniridia”,
Erlend Christoffer Sommer Landsend, MD, PhD, Oslo university hospital, Norway;
“Aniridia stories: Investigating congenital aniridia through narrative medicine”,
Chiara Ancona, MD, University of Brescia, Italy;
“Subjective symptoms and social consequences for patients with aniridia: Results of the pan-European COST survey study”,
Renata Schoffer, MD, Region Östergötland, Sweden
- 17.00 - 17.30 ● Panel discussion: Aniridia as a syndrome or spectrum?
Hinders and barriers for cross-European clinical management guidelines
Moderator: James Lauderdale
Participants: Peter Netland, Dominique Bremond-Gignac, Erlend Christoffer Sommer Landsend & Sophie Valleix
- 18.45 ● Meet-up and check-in at **Stockholm City Hall** inner yard –
Address: Hantverkargatan 1
- 19.00 ● Reception and buffet dinner at **Stockholm City Hall**, hosted by the City of Stockholm and the Region of Stockholm

01 June

Saturday

DAY TWO

08.30 - 09.30

Session: Artificial iris, cataract, posterior segment
Chair: Bogumil Wowra

“Artificial iris in aniridia: A dilemma”,
Vito Romano, MD, Assoc. Prof., University of Brescia, Italy;

“Posterior segment anomalies in PAX6-related congenital aniridia”,
Prof. **Dominique Bremond-Gignac**, MD, University hospital Necker enfants malades, APHP, France

09.30 - 10.30

Session: Keratopathy in aniridia
Chairs: Christina Grupcheva & Miriam Barbany Rodríguez

“Keratopathy in aniridia, clinical features and treatment options”,
Branka Samolov, MD, PhD, S:t Erik eye hospital, Sweden
& **Per Montan**, MD, Assist. Prof., S:t Erik eye hospital, Sweden;

“Lessons learned from a Swedish register study of aniridia”,
Berit Byström, MD, PhD, Umeå University, Sweden
& **Branka Samolov**, MD, Docent, S:t Erik eye hospital, Sweden;

“Outcomes of Corneal Transplantation to Treat Aniridia-Associated Keratopathy: Long Term Outcomes from UK Transplant Registry”,
Prof. **Francisco Figueiredo**, MD, Newcastle university, United Kingdom

10.30 - 11.00

Coffee break

11.00 - 12.00

Session: Stem cells and aniridia
Chairs: Stefano Ferrari & Francisco Figueiredo

“Molecular insights into aniridia-associated keratopathy”,
Berit Byström, MD, Docent, Umeå University, Sweden;

“Multi-omics analysis of aniridia stem cell-derived cornea cells”,
Dulce Lima Cunha, PhD, Radboud university, the Netherlands;

“Pluripotent stem cells as tools to study and treat limbal stem cell deficiency”,
Tanja Ilmarinen, Adj. Prof., Tampere university, Finland

12.00 - 13.30

Lunch and poster rapid fire presentations

13.30 - 14.30

Session: Genetics and aniridia
Chairs: Dominique Bremond-Gignac & James Lauderdale

“Genetic diagnosis, clinical implications and patient management”,
Prof. **Mariya Moosajee**, MD, UCL institute of ophthalmology & the Francis Crick institute, United Kingdom;

“NGS, WGS, optical mapping, and Hi-C tools to solve and interpret pathogenic mechanisms of congenital iris malformations: French experience from a cohort of more than 300 families”,
Prof. **Sophie Valleix**, MD, University hospital Necker enfants malades, France;

- 14.30 - 15.00 | Panel discussion: Challenges with genetic diagnosis, genotype-phenotype, and gene therapy; patient Q & A session
Moderator: Peter Netland
Participants: Neil Lagali, Mariya Moosajee, Sophie Valleix & James Lauderdale
- 15.00 - 15.30 | Coffee break
- 15.30 - 17.00 | Session: Management of anirida and challenges for children
Chairs: Laura Mairing & Bogumil Wowra

 - “Management of infants and children with aniridia at a German national referral center”,
Prof. **Barbara Käsmann-Kellner**, MD, Saarland university, Germany;
 - “Corneal aspects of congenital aniridia”,
Fabian Fries, MD, Saarland university medical center, Germany;
 - “Management of aniridia – European guidelines”,
Prof. **Dominique Bremond-Gignac**, MD, University hospital Necker enfants malades, APHP, France;
 - “Cohort data from 319 subjects with congenital aniridia at the Schwiete Center”,
Prof. **Nóra Szentmáry**, MD, Saarland university medical center, Germany

- 17.00 - 17.30 | Panel discussion: Differences and similarities in management of aniridia in children
Moderator: Bogumil Wowra
Participants: Dominique Bremond-Gignac, Erlend Christoffer Sommer Landsend, Laura Meuring & Barbara Käsmann-Kellner
- 19.00 | Dinner at the hotel restaurant

02 June

Sunday

08.30 - 10.30

Session: New therapies
Chairs: Neil Lagali & Stefano Ferrari

“Gene therapy approaches for aniridia”,
Prof. **Elizabeth M. Simpson**, University of British Columbia,
Canada;

“Dynamic and customized iris embedded inside a scleral
lens for aniridia patients”,
Andrés Vásquez Quintero, Assoc. Prof., Azalea Vision, Belgium;

“Innovations in sutureless transplantation of amniotic
membrane for the outpatient management of ocular
surface diseases”,
Andrew Hopkinson, Assoc. Prof., University of Nottingham,
United Kingdom;

“Updates on eyedrop therapy for mouse models of aniridia-
associated keratopathy”,
Karina Hadrian, PhD, University Hospital of Cologne, Germany;

“129S1/Svlmj Pax6 small-eye mice: A novel model for
investigating innovative Aniridia Associated Keratopathy (AAK)
therapies”,
Dina Javidjam, PhD candidate, Linköping university, Sweden

10.30 - 11.00

Panel discussion: Spinning up the wheel for new therapies,
challenges and potentials

Moderators: James Lauderdale & Stefano Ferrari
Participants: Elizabeth M. Simpson, Andrés Vásquez Quintero,
Andrew Hopkinson, Karina Hadrian & Dina Javidjam

DAY THREE

11.00 - 11.30

Coffee break

11.30 - 12.00

Q & A session for patients and experts

12.00 - 12.15

Popular summary for patients and associations
Prof. **Neil Lagali**, Linköping university, Sweden

12.15 - 12.30

Closing ceremony
Scientific committee
Branka Samolov, MD, PhD, S:t Erik eye hospital, Sweden;
Berit Byström, MD, Docent, Umeå university, Sweden;
Prof. **Neil Lagali**, Linköping university, Sweden;
Aniridia organizations
Neven Milivojevic, Vice President of Aniridia Sweden;
Ivana Kildsgaard, President of Aniridia Sweden.
Barbara Poli, President of Aniridia Europe.

12.30

Farewell lunch at the hotel restaurant

14.00

End of the conference

Speakers

Friday, 31 May

13.00 – 13.30 | Opening ceremony, welcome and introduction



**Neil
Lagali**

City and country
Linköping, Sweden

Academic/Professional title

Professor

Affiliation

Linköping University

Short biography

Neil Lagali is professor of experimental ophthalmology at Linköping University, Sweden. He leads a team of researchers focused on cornea research including rare diseases and both basic and clinical aniridia research. He has led large European research efforts to develop new knowledge and treatments for aniridia, including the COST Action ANIRIDIA-NET and EJP-RD project AAK-INSIGHT. He also serves on the scientific committee of Aniridia Europe.

Title of presentation

Introduction and Welcome to European Aniridia Conference 2024

Abstract of presentation

On behalf of the Scientific Organizing Committee of the 7th European Aniridia Conference, and with the generous support of our funders and sponsors, we are pleased to provide an exciting and up-to-date programme featuring the latest expertise, knowledge and innovations concerning aniridia and related ocular and systemic conditions. We are thankful to the expert speakers, both locally and internationally, who will provide stimulating and inspiring insights to promote further networking, collaboration, exchange and research to improve the lives of people with aniridia and as a consequence, other groups of people with related disorders. Knowledge, science, and clinical practice come together in this unique format where patients and patient organizations are also active participants. Looking forward to engaging discussions in Stockholm!



**Berit
Byström**

City and country
Umeå, Sweden

Academic/Professional title

MD, PhD, Docent

Affiliation

**University Hospital of Umeå, and
Umeå University**

Short biography

Docent, Dr Berit Byström is a senior consultant and head of the Corneal section in the University Hospital of Umeå, Sweden. She is a corneal surgeon, and she is also medically responsible for the Eye Bank in the same hospital. Her research is connected to the Dep of Clinical Sciences, Ophthalmology, Umeå University and covers the cornea in different aspects spanning over genetics, molecular perspectives as well as population studies with the use of quality registers. Dr Byström is active in the Swedish Cornea Transplant Register being a member in the steering group.

Title of presentation

Introduction and Welcome to European Aniridia Conference 2024



**Branka
Samolov**

City and country
Solna, Sweden

Academic/Professional title

MD, PhD

Affiliation

St Erik Eye Hospital

Short biography

Cataract and cornea surgeon. Senior consultant at the Anterior Segment Clinic and medical director for the Cornea department, St Erik Eye Hospital. PhD at Karolinska Institutet on Experimental studies of Corneal Neovascularisation. Chair of the steering group for Swedish Cornea Register, a national registry for cornea transplants.

Title of presentation

Introduction and Welcome to European Aniridia Conference 2024



Barbara Poli

City and country
Venice, Italy

Academic/Professional title
President of Aniridia Europe

Affiliation
Aniridia Europe

Short biography

Librarian. Born in 1965 in Venice, Italy. She works at the Library of the Fondazione Querini Stampalia and teaches bibliographic cataloguing at the University Ca' Foscari in Venice. Her son is affected by aniridia and she has been involved as a patient representative for this rare eye disease since 2003, when she was among the founders of the patient association "Aniridia Italy" and of the federation "Aniridia Europe". On behalf of the latter, she has participated to the organisation of conferences and to research projects in the field of eye rare diseases.

Title of presentation

Introduction and Welcome to European Aniridia Conference 2024



Ivana Kildsgaard

City and country
Stockholm, Sweden

Academic/Professional title
President of Aniridia Sweden

Affiliation
Aniridia Sweden

Short biography

Ivana is dedicated to supporting aniridia research, recognizing its important role in improving the quality of life for individuals affected by this rare condition. Since 2014, she has served as president of the Swedish Aniridia Association, while in 2016 she got involved in Aniridia Europe as Member of the Board. As a patient representative, Ivana actively participates in several European research projects, like ANIRIDIA-NET Cost Action #CA181169, Aniridia – Novel therapeutic tools to treat or prevent progressive cornea opacification – AAK-INSIGHT, and RESTORE Vision, a four-year project financed by the EU under the HORIZON EUROPE program. She brings a personal perspective to her advocacy, being the parent of a 15-year-old with sporadic aniridia.

Title of presentation

Introduction and Welcome to European Aniridia Conference 2024



Beatrice Peebo

City and country
Stockholm, Sweden

Academic/Professional title
Assistant Professor/Clinic Manager, MD, PhD

Affiliation
St Erik Eye Hospital

Short biography

Current position as Clinic Manager for the Surgery Department at St Erik Eye Hospital and adjunct assistant Professor at Linköping University in collaboration with Professor Neil Lagali.

Title of presentation

Welcome to European Aniridia Conference 2024

13.30 – 14.00 | Opening keynote lecture



Tor Paaske Utheim

City and country
Oslo, Norway

Academic/Professional title
Professor, MD, PhD

Affiliation
Oslo University Hospital

Short biography

Eye doctor Tor Paaske Utheim holds ten professorships split between seven universities in Norway and the United Kingdom. He is Director of Research and/or Innovation at eye departments of five hospitals in Norway. He has been rated among the top ten scientists and doctors in Norway based on publications points three years in a row. In 2012, Utheim was awarded a Fulbright Scholarship to Harvard Medical School, followed by an appointment as Adjunct Clinical Associate since 2013. He has received more than 20 academic, research, and innovation awards and honors, including the Oslo University Hospital Early Career Award for outstanding research (NOK 150 000); the Director's Award for Excellent Research at Oslo University Hospital, Ullevål; the European Society of Ophthalmology Award for the most promising Norwegian Young Clinician/Researcher; and the Medinnova's Innovation Prize of NOK 250 000. Utheim is the co-founder of the Norwegian Dry Eye Clinic and the Institute of Eye Health.

Title of presentation

Dry eye disease – myths and facts

Abstract of presentation

Dry eye disease is one of the most prevalent diseases in the world. The prevalence is increasing due to our modern lifestyle, such as extensive screen use, contact lenses, and air-condition. It was previously considered primarily a disease of the lacrimal glands, unrelated to inflammation. A recent fundamental change in our understanding of the disease demands new methods for diagnostics and treatment. In the present talk, the latest understanding of dry eye disease will be presented, and myths and facts discussed.

14.00 – 15.00 | Glaucoma and aniridia



Amelie Botling Taube

City and country
Stockholm, Sweden

Academic/Professional title
Consultant Ophthalmologist, MD, PhD
Affiliation
**St Erik Eye Hospital and Karolinska
Institutet, Stockholm**

Short biography

Amelie Taube is a Consultant Ophthalmologist and Head of the Glaucoma Service at S:t Erik Eye Hospital, Stockholm, Sweden. She did her residency and surgical training at Uppsala University Hospital and obtained her PhD in 2015 on proteomic and epidemiological studies in glaucoma at Uppsala University. Currently, her research focus is exploratory glaucoma treatment. With over 25 years of experience in cataract and glaucoma surgery in adults and children, she has a special interest in pediatric anterior segment and pediatric glaucoma. She has specialised in pediatric glaucoma and is Head of one of two centers for pediatric glaucoma surgery in Sweden. She is also on the board of the Swedish Glaucoma Society.

Title of presentation

Glaucoma in congenital aniridia. Clinical Considerations and Management

Abstract of presentation

Congenital aniridia consists of complex malformations of the eye. Glaucoma is a common feature in aniridia and is a contributing factor to visual deterioration. In this presentation, I will discuss the assessment of glaucoma in individuals with aniridia, and special considerations regarding their treatment and options in glaucoma surgery. Also, risks and potential complications to surgery will be described.



Peter A. Netland

City and country
**Charlottesville, Virginia,
USA**

Academic/Professional title
Vernah Scott Moyston Professor and Chair, MD, PhD

Affiliation
**Academic/Professional title
Vernah Scott Moyston Professor and
Chair, MD, PhD**

Short biography

Dr Netland is the Verrnah Scott Moyston Professor and Chair of Ophthalmology at UVA. He has recognized expertise in glaucoma management and has been a productive investigator. He has received numerous awards, including the Life Achievement Honor Award from AAO. He has served on numerous committees, and has served as past president of medical and ophthalmology societies, currently serving as President-Elect for the Chandler Grant Glaucoma Society. He serves on numerous committees and non-profit boards. Dr Netland was elected to the AOS in 2009. He was a founder and serves as Chair of the Board for Aniridia North America (ANA).

Title of presentation

Surgical Management of Aniridic Glaucoma

Abstract of presentation

Purpose: to provide updated information about the surgical treatments for glaucoma in aniridia. Methods: Literature and clinical comparative studies regarding surgical management of aniridic glaucoma will be presented. Results: Most children and adults with glaucoma and aniridia present with open-angle glaucoma, with a small proportion presenting with angle-closure associated with prior surgery. Evidence is favorable for use of glaucoma drainage implants in older children and adults with aniridic glaucoma. Retrospective comparisons of minimally-invasive glaucoma surgery (MIGS) in aniridic glaucoma will be presented. Conclusions: Surgical management of aniridic glaucoma is effective for controlling intraocular pressure (IOP) in aniridic glaucoma.



James Lauderdale

City and country
**Athens, Georgia,
 United States of America**

Academic/Professional title
**Director of Neuroscience, Associate
 Professor of Cellular Biology**

Affiliation
University of Georgia

Short biography

I am a developmental neuroscientist with over 30 years of experience studying the vertebrate eye and central nervous system. Much of my research is directed towards understanding the genes causal for human congenital eye disorders, such as aniridia. In addition to my research, I am active in organizations that represent, serve, and support persons affected by aniridia and their families. I am an invited expert for the Scientific Committee of Aniridia Europe and a founding board member and Scientific Chair of Aniridia North America.

Title of presentation

Aniridia and the Brain

Abstract of presentation

The PAX6 gene is essential for normal development and maintenance of the human eye and brain. Heterozygous loss-of-function mutations in PAX6 are causal for the human eye condition known as aniridia. In addition to changes in the eye, individuals with aniridia may also experience a decreased ability to smell, altered hearing, and changes in the functioning of other sensory processing systems. This talk will review what we currently know about non-eye changes associated with mutations in the PAX6 gene



Erlend Christoffer Sommer Landsend

City and country
Oslo, Norway

Academic/Professional title
MD, PhD

Affiliation
Oslo University Hospital

Short biography

I am an ophthalmologist (eye doctor) working mostly with children (pediatric ophthalmologist). My special interest is congenital eye diseases, including aniridia. In 2020, I defended my PhD thesis concerning the ocular surface and ocular fundus in aniridia. This included investigations of dry eye disease and inflammation, and changes in the retina.

Title of presentation

Health-Related Quality of Life in Congenital Aniridia

Abstract of presentation

Purpose: The aims of the presented study were to investigate health-related quality of life (HRQoL) in adults with aniridia and assess the relationships between HRQoL, psychological status, ocular health, and obesity. Methods: Twenty-nine adults with congenital aniridia participated. HRQoL was measured with standardized questionnaires. Symptoms of anxiety, depression, and obesity were assessed. We also analysed sociodemographic characteristics, and ocular and medical health variables. Results and Conclusion: Adults with congenital aniridia scored worse on certain measures of HRQoL than the general population. Poorer HRQoL was associated with increased symptoms of anxiety, depression, and obesity, and with presence of ocular pain.



Chiara Ancona

City and country
Brescia, Italy

Academic/Professional title
MD, research fellow

Affiliation
University of Brescia

Short biography

Dr Chiara Ancona is an ophthalmologist and Fellow of the European Board of Ophthalmology. She actively participates in research in corneal diseases and transplants. During her research fellow at the University of Brescia in 2023 she developed a keen interest, ended with a master, in narrative medicine. She then applied medicine narrative to the study of congenital aniridia.

Title of presentation

Aniridia Stories: investigating congenital aniridia through narrative medicine

Abstract of presentation

Congenital aniridia has a profound effect on vision, yet there has been limited exploration of the emotional and social dimensions of congenital aniridic patients. In this study, we adopt a narrative medicine approach to shed light on the illness burden experienced by these patients and their informal caregivers, offering valuable insights for clinical practice. We gathered narratives from patients of varying nationalities and ages. Our findings underscore the significance of emotional aspects in clinical interactions, highlighting their role in understanding management challenges and real-life experiences. Psychological support emerges as a critical component, spanning from clinical diagnosis to the entire care journey for both patients and their caregivers.



Renata Schoffer

City and country
Norrköping, Sweden

Academic/Professional title
MD

Affiliation
**Ögonkliniken (Eye clinic),
Region Östergötland**

Short biography

My name is Renata Schoffer and I am a resident doctor in ophthalmology. I started my residency in 2016 in Czech republic with focus on pediatric ophthalmology, especially ROP. My residency in Sweden started in 2019 and my main focus is still pediatric ophthalmology and plastic surgery.

Title of presentation

Subjective symptoms and social consequences for patients with aniridia: results of the pan-European COST survey study

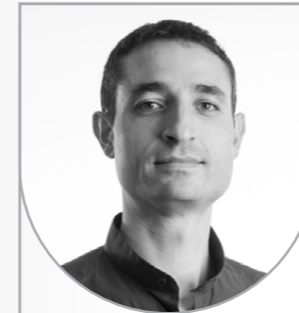
Abstract of presentation

In this study we report results from the European COST ANIRIDIA-NET patient survey study. The subjective symptoms and daily limitations for patients with aniridia related to vision were the focus of a 21-question survey, translated to 12 languages and completed via online platform. Vision, ocular symptoms, social challenges, difficulties in daily situations and activities requiring assistance were among the topics. The survey was completed by 295 aniridia patients ranging in age from few weeks to 79 years, and represented patients living in 15 countries in Europe. Results from this unique survey study will be presented and discussed.

Speakers

Saturday, 1 June

08.30 – 09.30 | Artificial iris, cataract, posterior segment



Vito Romano

City and country
Brescia, Italy

Academic/Professional title
Professor of Ophthalmology

Affiliation
University of Brescia

Short biography

Vito Romano is passionate about translational research and clinical trial design with a special interest in cornea, cataract and refractive surgery, ophthalmic imaging and ocular surface diseases. He is Professor of Ophthalmology at University of Brescia (Italy) with honorary position at the University of Liverpool.

Title of presentation

Artificial iris in aniridia: a dilemma

Abstract of presentation

The talk assesses the use of artificial iris implants in congenital aniridia, focusing on surgical techniques, clinical outcomes, complications, and study biases. It highlights the variability in outcomes and the difficulty in determining which prosthesis yields the best results. Complications, including postoperative glaucoma, are prevalent, and the mechanisms behind these issues remain unclear. Well-designed nonrandomized studies are needed. The subjective symptom improvement among patients is noted, although more quantitative measures are required. The potential benefits of improved vision must be weighed against the risks of complications, such as glaucoma and corneal issues.



Dominique Bremond-Gignac

City and country
Paris, France

Academic/Professional title
Professor
Affiliation
**University Hospital Necker
Enfants malades**

Short biography

Dominique Bremond-Gignac, MD, PhD, FEBO, Professor of Ophthalmology, Head of Ophthalmology Department with pediatric and ocular surface subspecialties at University Hospital Necker-Enfants malades and Paris Cité University in Paris. Head of Paris Orthoptic Department, associate researcher at INSERM UMRS 1138, Team 17 Research Unit, in innovation therapy. Activity is distributed in clinical practice, teaching and research. Involved with patients with aniridia disease and President of Scientific Committee of Aniridia Europe and Gêneris

Title of presentation

Posterior segment anomalies in PAX6 related congenital aniridia

Abstract of presentation

Congenital aniridia PAX6 related is characterized by a hypoplastic or absent iris and foveal hypoplasia. It has been recently shown that, in patients harboring PAX6 mutations, foveal hypoplasia is more frequently encountered than the complete absence of iris. The first study aims to characterize retinal disorders in patients with congenital aniridia PAX6 related and to characterize foveal vasculature by optical coherence tomography angiography. The second study aims to characterize optic disk hypoplasia in congenital aniridia using ultra-wide field imaging and non-mydratic retinal photography. Finally, we also investigated the relationship between optic disk hypoplasia and foveal hypoplasia.

09.30 – 10.30 | Keratopathy in aniridia



Branka Samolov

City and country
Solna, Sweden

Academic/Professional title
MD, PhD
Affiliation
St Erik Eye Hospital

Short biography

Cataract and cornea surgeon. Senior consultant at the Anterior Segment Clinic and medical director for the Cornea department, St Erik Eye Hospital. PhD at Karolinska Institutet on Experimental studies of Corneal Neovascularisation. Chair of the steering group for Swedish Cornea Register, a national registry for cornea transplants.

Title of presentation

Keratopathy in aniridia, clinical features and treatment options

Abstract of presentation

Keratopathy in aniridia is characterized by stem cell deficiency causing destabilization of epithelium and jeopardizing corneal avascularity, both prerequisite for visual ability. Together with altered immune activity and disturbed tear function this leads to impaired corneal sensibility and wound healing, pannus and vessel ingrowth, stromal thickening, reduced corneal transparency and light refracting properties. The stem cell deficiency plays the central role in aniridia-related keratopathy implicating current treatment options and limitations. The lecture covers present evidence and personal experience on surgical methods addressing corneal stem cell deficiency and critical loss of its transparency.



Per Montan

City and country
Solna, Sweden

Academic/Professional title
Assistant Professor, Senior Consultant
Affiliation
St Erik Eye Hospital

Short biography

Cataract surgeon since 1986, medical cornea since 1990 and corneal surgeon since 2000. PhD thesis on vernal keratoconjunctivitis in 2000. Author of 55 original articles.

Title of presentation

Keratopathy in aniridia, clinical features and treatment options

Abstract of presentation

Keratopathy in aniridia is characterized by stem cell deficiency causing destabilization of epithelium and jeopardizing corneal avascularity, both prerequisites for visual ability. Together with altered immune activity and disturbed tear function this leads to impaired corneal sensibility and wound healing, pannus and vessel ingrowth, reduced corneal transparency and light refracting properties. The stem cell deficiency plays the central role in aniridia-related keratopathy warranting treatment which all have limitations. The lecture covers present published evidence and personal experience of relevant surgical methods.



Berit Byström

City and country
Umeå, Sweden

Academic/Professional title
MD, PhD, Docent

Affiliation
University Hospital of Umeå, and Umeå University

Short biography

Docent, Dr Berit Byström is a senior consultant and head of the Corneal section in the University Hospital of Umeå, Sweden. She is a corneal surgeon, and she is also medically responsible for the Eye Bank in the same hospital. Her research is connected to the Dep of Clinical Sciences, Ophthalmology, Umeå University and covers the cornea in different aspects spanning over genetics, molecular perspectives as well as population studies with the use of quality registers. Dr Byström is active in the Swedish Cornea Transplant Register being a member in the steering group.

Title of presentation

Lessons learned from a Swedish register study of aniridia

Abstract of presentation

We have examined the frequency, choice of corneal transplantation techniques and outcome in aniridia-related keratopathy (ARK), in Sweden and Denmark between 2001–2016 in cases registered in the Swedish Cornea Transplant Registry. During this time, 36 eyes underwent corneal transplantation due to ARK. Penetrating keratoplasty (PK) was performed in 58.3%, a combination of PK and limbal stem cell transplantation in 13.9%, keratolimbal allograft in another 13.9%, Boston keratoprosthesis in 8.3%, and anterior lamellar keratoplasty was chosen in 5.6% of the cases. Two years follow-up data were available for 26 cases, of which most cases (61.5%) had a graft providing useful vision.



Branka Samolov

City and country
Solna, Sweden

Academic/Professional title
MD, PhD

Affiliation
St Erik Eye Hospital

Short biography

Cataract and cornea surgeon. Senior consultant at the Anterior Segment Clinic and medical director for the Cornea department, St Erik Eye Hospital. PhD at Karolinska Institutet on Experimental studies of Corneal Neovascularisation. Chair of the steering group for Swedish Cornea Register, a national registry for cornea transplants.

Title of presentation

Lessons learned from a Swedish register study of aniridia

Abstract of presentation

We have examined the frequency, choice of corneal transplantation techniques and outcome in aniridia-related keratopathy (ARK), in Sweden and Denmark between 2001–2016 in cases registered in the Swedish Cornea Transplant Registry. During this time, 36 eyes underwent corneal transplantation due to ARK. Penetrating keratoplasty (PK) was performed in 58.3%, a combination of PK and limbal stem cell transplantation in 13.9%, keratolimbal allograft in another 13.9%, Boston keratoprosthesis in 8.3%, and anterior lamellar keratoplasty was chosen in 5.6% of the cases. Two years follow-up data were available for 26 cases, of which most cases (61.5%) had a graft providing useful vision.



Francisco Figueiredo

City and country
Newcastle upon Tyne, United Kingdom

Academic/Professional title
Professor

Affiliation
Bioscience Institute, medical School, Newcastle University & Newcastle Hospital NHS Foundation Trust

Short biography

Prof. Figueiredo, MD, PhD, FRCOphth is a Consultant Ophthalmologist in the Department of Ophthalmology, Royal Victoria Infirmary (Newcastle upon Tyne, UK) and Professor of Ophthalmology at Newcastle University. He has 30 years expertise in the field of clinical and basic science research in ophthalmology, in particular in the management of ocular surface diseases. Prof Figueiredo co-founded the Bowman Club (UK Cornea Society) in 1998 and is the current secretary. He was President of MCLOSA (British Ocular Surface Society; 2012-14). His research has focused on limbal stem cells, corneal graft rejection, corneal graft outcomes, dry eye, ocular surface allergy and pterygium.

Title of presentation

Outcomes of Corneal Transplantation to Treat Aniridia-Associated Keratopathy: Long Term Outcomes from UK Transplant Registry

Abstract of presentation

The outcome of keratoplasty for Aniridia Associated Keratopathy (AAK) are generally poor. This study aims to evaluate the characteristics and outcomes of patients undergoing keratoplasty to treat AAK in the UK A retrospective registry-study, including all patients who underwent keratoplasty to treat AAK between April 2000-March 2021. Data were collected from UK Transplant Registry (NHS Blood and Transplant) at time of transplant, one-year, and two-years post-transplant. Sixty-five AAK patients received keratoplasty, nine grafts failed at one-year which equated to 75% (95% Confidence Interval (CI): 59%-85%) one-year graft survival (Kaplan-Meier). At two-years, graft survival fell to 62% (95% CI: 45%-76%, n=36). BCVA was progressively worse post transplantation. Visual gain post keratoplasty was modest with 92% of patients had 6/60 vision or worst at 2 years. The decision to perform keratoplasty in AAK patients is difficult and must be individually considered.

11.00 – 12.00 | Stem cells and aniridia



Berit Byström

City and country
Umeå, Sweden

Academic/Professional title
MD, PhD, Docent

Affiliation
University Hospital of Umeå, and Umeå University

Short biography

Docent, Dr Berit Byström is a senior consultant and head of the Corneal section in the University Hospital of Umeå, Sweden. She is a corneal surgeon, and she is also medically responsible for the Eye Bank in the same hospital. Her research is connected to the Dep of Clinical Sciences, Ophthalmology, Umeå University and covers the cornea in different aspects spanning over genetics, molecular perspectives as well as population studies with the use of quality registers. Dr Byström is active in the Swedish Cornea Transplant Register being a member in the steering group.

Title of presentation

Molecular insights into aniridia-associated keratopathy

Abstract of presentation

Aniridia-related keratopathy (ARK) comprises limbal stem cell deficiency with impaired epithelial cell adhesion, epithelial erosions, corneal conjunctivalization, and formation of a vascular pannus. With the aim to reveal histopathologic changes in ARK, we compared sections of naïve ARK corneas, removed at the time of the first transplantation, and ARK corneal buttons, removed after a failed keratolimbal allograft or failed centered/decentered transplantation, with normal human adult and fetal corneas. All ARK corneas presented analogous features, similar to fetal corneas, irrespective of earlier transplantation method, which advocates an important role for less differentiated host specific factors in the pathophysiology of ARK.



Dulce Lima Cunha

City and country
Nijmegen, Netherlands

Academic/Professional title
Postdoctoral Researcher

Affiliation
Radboud University

Short biography

Dulce Lima Cunha obtained her PhD in Genetics and Genomics in 2018 from the Innsbruck Medical University (Austria), followed by a first postdoc in Mariya Moosajee's group at UCL Institute of Ophthalmology studying inherited eye disorders, with focus on aniridia. In 2021, she moved to the Netherlands, where she is currently a Postdoc Researcher at Radboud University. Her main research interests are uncovering rare disease mechanisms using induced pluripotent stem cell (iPSC)-derived models and developing novel therapy approaches for aniridia and other rare diseases.

Title of presentation

Multi-omics analysis of aniridia stem cell-derived cornea cells

Abstract of presentation

Most aniridia patients with PAX6 mutations develop aniridia-related keratopathy (ARK), a disorder where the cornea gets progressively opaque, often leading to blindness. Using stem cells generated from aniridia patients, we examined the PAX6 role in development and function of limbal stem cells (LSC), the cells that continuously replenish the central cornea and maintain its transparency. We used RNAseq, a technique that detects gene expression changes, and are currently performing single-cell ATACseq, where we can dissect PAX6 binding sites, in aniridia LSC. These OMICS approaches will help understand the molecular changes in aniridia LSC and find novel targets and mechanisms for developing new therapies for ARK.



Tanja Ilmarinen

City and country
Tampere, Finland

Academic/Professional title
Senior researcher, adjunct professor

Affiliation
Tampere university

Short biography

I joined the Tampere university regenerative medicine team after finishing my PhD in genetics and molecular medicine from the University of Helsinki and Finnish National Public Health Institute. Currently, my research is focusing on development of stem cell-based tools for disease modeling and cell therapy treatments for ocular applications, including limbal stem cell deficiency. I have almost two decades' experience with pluripotent stem cells including ocular differentiation and characterization of graft functionality in vitro, as well as in preclinical context using different animal models. In addition, I am a co-founder of StemSight, a spin-off company developing cell therapies to address corneal blindness.

Title of presentation

Pluripotent stem cells as tools to study and treat limbal stem cell deficiency

Abstract of presentation

Pluripotent stem cells (PSCs), with their unique ability to differentiate into diverse cell types, have emerged as a transformative tool in disease modeling and therapy. We have established clinically applicable methods to differentiate ocular cells, including limbal stem cells (LSC), from PSCs. These cells have been instrumental in investigating conditions like aniridia and addressing diseases such as LSC deficiency. In this presentation, I will shortly summarize our recent work on LSC lineage commitment of aniridia patient-derived PSCs as well as in vivo transplantation studies with PSC-LSCs.



Mariya Moosajee

City and country
London, United Kingdom

Academic/Professional title
Professor
Affiliation
Moorfields Eye Hospital and UCL Institute of Ophthalmology

Short biography

Professor Mariya Moosajee is a clinician scientist; Consultant Ophthalmologist specialising in Genetic Eye Disease and Head of the Genetics Service at Moorfields Eye Hospital, Professor of Molecular Ophthalmology at UCL Institute of Ophthalmology, and Group Leader of Ocular Genomics and Therapeutics at the Francis Crick Institute in London. Her clinical focus is providing a genomic ophthalmology service for children and adults affected with genetic eye disease including aniridia. Her clinical research involves natural history studies to understand disease progression and support clinical trials. In the laboratory, she is advancing our understanding of aniridia using zebrafish disease models and human induced pluripotent stem cell derived retinal and corneal organoids.

Title of presentation

Genetic diagnosis, clinical implications and patient management

Abstract of presentation

Aniridia is a rare congenital pan-ocular condition, which is predominantly caused by mutations involving the PAX6 gene. It is inherited autosomal dominantly with high penetrance, although there is significant intra- and interfamily phenotypic variability, and one-third of cases are sporadic. In this lecture, I will highlight how we manage families presenting with aniridia, the importance of genetic investigation, counselling and implications for future patient care. There are established genotype-phenotype correlations, and as PAX6 is also expressed outside of the eye, including the pancreas and brain, systemic involvement must be considered.



Sophie Valleix

City and country
Paris, France

Academic/Professional title
Professor

Affiliation
Cochin Hospital, APHP, Cité University of Paris

Short biography

Sophie Valleix, MD, PhD, is professor of molecular genetics at Cité University of Paris in France with a biological expertise in the field of rare eye diseases. She is at the head of the molecular diagnosis unit of hereditary ocular disorders, based at Cochin Hospital in Paris, in charge of vitreo-retinal dystrophies, anterior segment dysgenesis disorders, congenital cataracts, and hereditary corneal dystrophies. She is also a molecular geneticist at SeqoIA laboratory, actively involved in the French National project (PFMG_2025) for the implementation of whole genome sequencing of hereditary eye diseases. She is a member of OPHTARA, SENSGENE and ERN-EYE.

Title of presentation

NGS, WGS, optical mapping, and Hi-C tools to solve and interpret pathogenic mechanisms of congenital iris malformations: French experience from a cohort of more than 300 families

Abstract of presentation

To optimize the genetic diagnosis of anterior segment dysgenesis disorders, we combined NGS with WGS for unsolved NGS-cases. We report the genetic spectrum of 337 French families, highlighting new disease-causing variants, and novel associations with neurodevelopmental genes. More importantly, WGS identified “extragenic” structural variants in cis-regulatory regions of PAX6 and PITX2 whose functional “in vivo” consequences on chromatin conformation were addressed by HiC studies from patient tissues. This study highlights the clinical importance of the noncoding genome in the pathogenesis of unsolved cases of aniridia and Axenfeld-Rieger disorders, improving the diagnostic yield of developmental disorders of the iris.



Mariya Moosajee

City and country
London, United Kingdom

Academic/Professional title
Professor

Affiliation
Moorfields Eye Hospital and UCL Institute of Ophthalmology

Short biography

Professor Mariya Moosajee is a clinician scientist; Consultant Ophthalmologist specialising in Genetic Eye Disease and Head of the Genetics Service at Moorfields Eye Hospital, Professor of Molecular Ophthalmology at UCL Institute of Ophthalmology, and Group Leader of Ocular Genomics and Therapeutics at the Francis Crick Institute in London. Her clinical focus is providing a genomic ophthalmology service for children and adults affected with genetic eye disease including aniridia. Her clinical research involves natural history studies to understand disease progression and support clinical trials. In the laboratory, she is advancing our understanding of aniridia using zebrafish disease models and human induced pluripotent stem cell derived retinal and corneal organoids.

Title of presentation

Syndromic aniridia and metabolomics

Abstract of presentation

PAX6-related aniridia is considered to be an isolated pan-ocular condition, but observational studies have revealed systemic features including behavioural difficulties, such as autism and attention-deficit/hyperactivity disorder (ADHD), diabetes and obesity. In this lecture, I will highlight some of the potential syndromic associations and provide an overview of our work investigating the metabolomic differences between aniridia patients and healthy control individuals, and explain how these may relate to signs of systemic involvement



Barbara Käsmann-Kellner

Academic/Professional title

Professor

City and country

Homburg/Saar, Germany

Affiliation

Dept. of Ophthalmology, University of Saarland, Germany

Short biography

Since many years I am the medical advisor to Anirie-Wagr.de Germany. My main occupation is the care of infant, junior and adult persons with Low Vision. I am retired early due to health reasons since July 1st, 2023. I continue to advise three support groups: Aniridia, Albinism, Achromatopsia. In addition, I am open to any pediatric ophthalmology questions and still lecture in the Department of Ophthalmology, University of Saarland, Germany and participate in research. Aniridia patients have always been the most challenging patients due to the progressive complications – it is a difficult task for the ophthalmologist to accompany the patients during their ordeal.

Title of presentation

Management of infants and children with aniridia at a German national referral center

Abstract of presentation

Aniridia is a rare congenital pan-ocular condition, which is predominantly caused by mutations involving the PAX6 gene. It is inherited autosomal dominantly with high penetrance, although there is significant intra- and interfamilial phenotypic variability, and one-third of cases are sporadic. In this lecture, I will highlight how we manage families presenting with aniridia, the importance of genetic investigation, counselling and implications for future patient care. There are established genotype-phenotype correlations, and as PAX6 is also expressed outside of the eye, including the pancreas and brain, systemic involvement must be considered.



Fabian Fries

Academic/Professional title

MD

City and country

Homburg, Germany

Affiliation

Department of Ophthalmology, Saarland University Medical Center, Homburg/Saar, Germany AND Dr. Rolf M. Schwiete Center for Limbal Stem Cell and Aniridia Research, Saarland University, Homburg/Saar, Germany.

Short biography

Fabian Fries is a senior consultant and ophthalmic surgeon of the Department of Ophthalmology at Saarland University in Homburg. His research interests include the development of aniridia associated keratopathy and the optimization of surgical management in this condition. He is an active clinician at the Homburg Aniridia Centre, which with over 350 aniridia patients, is one of the largest centers of excellence internationally. In addition to his recognized expertise in the clinical management of aniridia, Dr. Fries is a prolific investigator, having authored more than 80 original scientific articles, book chapters, reviews, and published abstracts. He has been working closely with the Dr. Rolf M. Schwiete Center for Limbal Stem Cell and Aniridia Research and COST members to offer patients care based on the latest research findings.

Title of presentation

Management of children with aniridia at a German national referral center

Abstract of presentation

The Homburg Aniridia Register enables more and more patients to be included and, in the long term, to make significant statements on issues with a rarity value. Recent research projects in collaboration with the Dr. Rolf M. Schwiete Center for Limbal Stem Cell and Aniridia Research and the latest results on aniridia-associated keratopathy will be presented.



Dominique Bremond-Gignac

Academic/Professional title

Professor

City and country

Paris, France

Affiliation

**University Hospital Necker
Enfants malades**

Short biography

Dominique Bremond-Gignac, MD, PhD, FEBO, Professor of Ophthalmology, Head of Ophthalmology Department with pediatric and ocular surface subspecialties at University Hospital Necker-Enfants malades and Paris Cité University in Paris. Head of Paris Orthoptic Department, associate researcher at INSERM UMRS 1138, Team 17 Research Unit, in innovation therapy. Activity is distributed in clinical practice, teaching and research. Involved with patients with aniridia disease and President of Scientific Committee of Aniridia Europe and Gêneris

Title of presentation

Management of aniridia – European guidelines

Abstract of presentation

European reference network and COST Action is dedicated to Rare Eye Diseases. Clinical Guidelines has been developed by a European group of expert ophthalmologists and from COST Project, geneticists, allied healthcare professionals and patient support groups from European countries. The aim of the guidelines is to support management and follow-up in congenital aniridia as a balanced care by establishing a standard of care for all patients affected. A systematic literature review was carried out using Pubmed and relevant databases. Meetings were organized with experts to define recommendations. Final results will be exposed.



Nóra Szentmáry

Academic/Professional title

Professor Dr

City and country

**Homburg,
Germany**

Affiliation

**Dr Rolf M. Schwiete Center for Limbal Stem
Cell and Congenital Aniridia Research,
Saarland University, Homburg, Germany**

Short biography

Professor Nóra Szentmáry studied medicine, became an ophthalmologist and received her PhD title at Semmelweis University, Budapest, Hungary. Between 2008–2010, she worked at the Friedrich Alexander University and at Saarland University, Germany, with support of a Humboldt Scholarship. In 2012, she habilitated at the Medical Faculty of Saarland University. She is working as head of the Dr Rolf M. Schwiete Center for Limbal Stem Cell and Congenital Aniridia Research at Saarland University since 2020. She was Head of the Cornea and Ocular Surface Section of the European Association for Vision and Eye Research (EVER) between 2017–2022, and EVER president in 2022–2023.

Title of presentation

Cohort Data from 319 Subjects with Congenital Aniridia at the Schwiete Center

Abstract of presentation

Anirida is a rare disease, with a global prevalence of 1 in 40,000 to 1 in 100,000. There are PAX6 gene-associated and other forms. PAX6 is a master control gene for ocular development in early embryogenesis (weeks 6 to 12). Therefore, PAX6 haploinsufficiency-associated aniridia syndrome affects almost all eye structures, and between those limbal stem cells, resulting in aniridia associated keratopathy (AAK). We summarize recent clinical data of the Homburg Aniridia Center, the effect of glaucoma treatment and lens status on AAK, central corneal microstructure, structure of corneal nerves and corneal endothelial cell properties in congenital aniridia.

Speakers

Sunday, 2 June

08.30 – 10.30 | New therapies



Elizabeth M. Simpson

Academic/Professional title

Dr

City and country

Vancouver, Canada

Affiliation

The University of British Columbia

Short biography

Elizabeth M. Simpson, BSc, MSc, PhD, is a leading scientist in mammalian genetics and genomics. The goal of her research is to improve treatment for human disorders of the brain and eye. Currently, she is focused on the development of DNA-based gene therapies in mice. She uses adeno-associated viruses and lipid nanoparticles to deliver both augmentation and genome-editing (CRISPR) therapies for treatment-resistant disorders such as Parkinson disease and aniridia (congenital blindness). Dr. Simpson is a Professor at the University of British Columbia in the Department of Medical Genetics, and an Associate Member in the Department of Ophthalmology & Visual Sciences.

Title of presentation

Gene Therapy Approaches for Aniridia

Abstract of presentation

Aniridia is a rare congenital vision-loss disease caused by heterozygous variants in the PAX6 gene. There is no long-term vision-saving therapy, but gene therapy holds promise for future treatment. Gene therapy uses nucleic acids as reagents to alter cellular behavior and thereby cure disease. Recent successes include the USA Food and Drug Administration (FDA)-approved augmentation gene therapy Luxturna for Leber Congenital Amaurosis 2. FDA-approved clinical trials for genome editing (CRISPR) gene therapies include EDIT-101 for Leber Congenital Amaurosis 10. This seminar will present the most recent work in the Simpson laboratory towards gene therapy for aniridia employing a mouse model.



Andrés Vásquez Quintero

Academic/Professional title

Chief Technology Officer/Associate Professor

City and country

Ghent, Belgium

Affiliation

Azalea Vision

Short biography

Prof. Dr. Andrés Vásquez Quintero is a highly experienced expert in stretchable electronics and wearable medical devices. He is the founder and CTO of Azalea Vision, a venture-capital-backed startup developing a smart contact lens with an active light management system. He holds a position at Ghent University as Associate Professor doing research on wearable medical devices.

Title of presentation

Dynamic and customized iris embedded inside a scleral lens for aniridia patients

Abstract of presentation

The lens is designed to benefit people with high order aberrations, presbyopia and light sensitivity (including aniridia, coloboma and ocular albinism). In this talk, Andrés will discuss the major technological milestones at Azalea Vision. Additionally, he will present the optical models and impact on visual acuity of small dynamic aperture on large pupils.



Andrew Hopkinson

Academic/Professional title

Associate Professor

City and country

Nottingham, UK

Affiliation

University of Nottingham

Short biography

Dr. Andrew Hopkinson, PhD, serves as the Chief Scientific Officer and founder of NuVision, and holds an honorary position as Associate Professor at the University of Nottingham. With 18 years in translational research in ophthalmic regenerative medicine at the University's Centre For Eye Research, Andrew's work has significantly advanced therapies for ocular surface repair and visual rehabilitation. His research primarily focused on amniotic membrane properties, leading to the innovative Tereo process and Omnigen production. Dr. Hopkinson has pioneered groundbreaking discoveries, such as a dehydrated human cornea and corneal-derived stem cell therapy, progressing from concept to pre-clinical stages. His academic career boasts over 50 scientific publications, 4 patents, and aims to develop a future portfolio of Biotherapies for clinical translation at NuVision.

Title of presentation

Innovations in sutureless transplantation of amniotic membrane for the outpatient management of ocular surface diseases.

Abstract of presentation

The use of human amniotic membrane (AM) in clinical ophthalmology has seen a significant rise, particularly with the advent of sutureless application methods. Recognised for its anti-inflammatory, anti-angiogenic, anti-fibrotic, and wound-healing properties, AM is now extensively utilised in treating both acute and chronic ocular surface diseases (OSD). Omnigen®, an innovative dehydrated AM product, addresses the limitations of traditional products, offering an accessible, effective, and user-friendly 'off the shelf' treatment option. The novel application of Omnigen with OmniLenz®, a bespoke contact lens, allows for sutureless, outpatient application, streamlining and advancing the treatment of OSD. This session explores the scientific foundation and clinical outcomes of this novel transplantation technique, underscoring its importance in enhancing OSD management.



**Karina
Hadrian**

Academic/Professional title
PhD

City and country
Cologne, Germany

Affiliation
University Hospital of Cologne

Short biography

After a Bachelor's degree and a Master's degree in Biochemistry and Molecular Medicine from Ruhr-University Bochum, Germany, I graduated from University Duisburg-Essen in 2019 with a PhD in Biology (summa cum laude) focusing on aging of the retinal pigment epithelium. Afterward, I started as a Postdoc in the lab of Prof. Cursiefen in Cologne, Germany focusing on corneal repair after inflammation.

Title of presentation

New age-dependent aniridia-like mouse model

Abstract of presentation

The Aey 80 mutant mouse was initially characterized as small-eye phenotype during development. We could now show that this mutation results in an age-dependent aniridia-like phenotype. This is characterized by reduced iris dimensions and reduced anterior chamber depth. Additionally, the mutation induces shifts in corneal thickness and a substantial ingrowth of lymphatic vessels into the cornea, accompanied by increased intraocular pressure. These alterations demonstrate the pivotal role of Pax6 in maintaining the structural integrity and functionality of the eye across different developmental stages.



**Dina
Javidjam**

Academic/Professional title
PhD Candidate

City and country
Linköping, Sweden

Affiliation
**Linköping University hospital,
Department of Biomedical and Clinical
Sciences (BKV)**

Short biography

My name is Dina Javidjam. I possess a bachelor's in molecular biology and a master's in clinical biochemistry from Mashhad University of Medical Sciences, Iran. My expertise lies in limbal stem cell culture on the amniotic membrane for transplantation, as showcased in my master's thesis centered on optimization. My passion for addressing Limbal Stem Cell Deficiency (LSCD) led me to contribute as a researcher at Charles University, Czech Republic, focusing on the long-term preservation of cultured limbal stem cells for clinical applications. Presently, I am a PhD candidate at Linköping University, Sweden. My research concentrates on Aniridia Associated Keratopathy (AAK).

Title of presentation

129S1/Svlmj Pax6 small-eye mice: A novel model for investigating innovative Aniridia Associated Keratopathy (AAK) therapies

Abstract of presentation

Aniridia-associated keratopathy (AAK) is a progressive corneal opacification associated with a deficiency of limbal stem cells. Existing treatments only temporarily relieve symptoms. To develop novel treatments, understanding the limbal niche's processes and PAX6's role is crucial. Mouse models with aniridia often have advanced AAK observable shortly after birth. In our study, we characterize a Pax6^{Sey} mouse recapitulating the human situation with delayed onset of AAK. We describe the model, features of the keratopathy and limbal niche, and evaluate new potential therapeutic options.

12.00 – 12.15 | Popular summary for patients and associations



Neil Lagali

City and country
Linköping, Sweden

Academic/Professional title

Professor

Affiliation
Linköping University

Short biography

Neil Lagali is professor of experimental ophthalmology at Linköping University, Sweden. He leads a team of researchers focused on cornea research including rare diseases and both basic and clinical aniridia research. He has led large European research efforts to develop new knowledge and treatments for aniridia, including the COST Action ANIRIDIA-NET and EJP-RD project AAK-INSIGHT. He also serves on the scientific committee of Aniridia Europe.

Title of presentation

Popular summary for patients and associations

12.15 – 12.30 | Closing ceremony and acknowledgements



Branka Samolov

City and country
Solna, Sweden

Academic/Professional title

MD, PhD

Affiliation
St Erik Eye Hospital

Short biography

Cataract and cornea surgeon. Senior consultant at the Anterior Segment Clinic and medical director for the Cornea department, St Erik Eye Hospital. PhD at Karolinska Institutet on Experimental studies of Corneal Neovascularisation. Chair of the steering group for Swedish Cornea Register, a national registry for cornea transplants.

Title of presentation

Closing ceremony and acknowledgements



Berit Byström

City and country
Umeå, Sweden

Academic/Professional title

MD, PhD, Docent

Affiliation
University Hospital of Umeå, and Umeå University

Short biography

Docent, Dr Berit Byström is a senior consultant and head of the Corneal section in the University Hospital of Umeå, Sweden. She is a corneal surgeon, and she is also medically responsible for the Eye Bank in the same hospital. Her research is connected to the Dep of Clinical Sciences, Ophthalmology, Umeå University and covers the cornea in different aspects spanning over genetics, molecular perspectives as well as population studies with the use of quality registers. Dr Byström is active in the Swedish Cornea Transplant Register being a member in the steering group.

Title of presentation

Closing ceremony and acknowledgements



Neil Lagali

City and country
Linköping, Sweden

Academic/Professional title

Professor

Affiliation
Linköping University

Short biography

Neil Lagali is professor of experimental ophthalmology at Linköping University, Sweden. He leads a team of researchers focused on cornea research including rare diseases and both basic and clinical aniridia research. He has led large European research efforts to develop new knowledge and treatments for aniridia, including the COST Action ANIRIDIA-NET and EJP-RD project AAK-INSIGHT. He also serves on the scientific committee of Aniridia Europe.

Title of presentation

Closing ceremony and acknowledgements



**Neven
Milivojevic**

Academic/Professional title
Founder and Vice President of Aniridia Sweden

City and country
Solna, Sweden

Affiliation
Aniridia Sweden

Short biography

Neven Milivojevic, a diagnosis carrier himself, founded Aniridia Sweden nearly 20 years ago, originally named The network for aniridia in Sweden. He was also the co-founder of the European federation Aniridia Europe and its first Vice President. He has an extensive experience of working with several Swedish non-governmental organisations. Professionally he has been involved in international cooperation development, and has been a political adviser at both local, regional and national level, an elected City council member, a teacher at Uppsala University and has done senior advocacy work nationally as well as internationally. As an 18-year old, Neven was awarded an international title in chess (FIDE-Master), and has since been searching for the right ways forward in life.

Title of presentation

Closing ceremony and acknowledgements



**Ivana
Kildsgaard**

Academic/Professional title
President of Aniridia Sweden

City and country
Stockholm, Sweden

Affiliation
Aniridia Sweden

Short biography

Ivana is dedicated to supporting aniridia research, recognizing its important role in improving the quality of life for individuals affected by this rare condition. Since 2014, she has served as president of the Swedish Aniridia Association, while in 2016 she got involved in Aniridia Europe as Member of the Board. As a patient representative, Ivana actively participates in several European research projects, like ANIRIDIA-NET Cost Action #CA181169, Aniridia – Novel therapeutic tools to treat or prevent progressive cornea opacification – AAK-INSIGHT, and RESTORE Vision, a four-year project financed by the EU under the HORIZON EUROPE program. She brings a personal perspective to her advocacy, being the parent of a 15-year-old with sporadic aniridia.

Title of presentation

Closing ceremony and acknowledgements



**Barbara
Poli**

Academic/Professional title
President of Aniridia Europe

City and country
Venice, Italy

Affiliation
Aniridia Europe

Short biography

Librarian. Born in 1965 in Venice, Italy. She works at the Library of the Fondazione Querini Stampalia and teaches bibliographic cataloguing at the University Ca' Foscari in Venice. Her son is affected by aniridia and she has been involved as a patient representative for this rare eye disease since 2003, when she was among the founders of the patient association "Aniridia Italy" and of the federation "Aniridia Europe". On behalf of the latter, she has participated to the organisation of conferences and to research projects in the field of eye rare diseases.

Title of presentation

Closing ceremony and acknowledgements

Practical A–Z

Conference meals

May 31st – Lunch, 12.00–13.00 in the hotel Blique by Nobis restaurant Boketto

May 31st – Reception with mingle buffet dinner,(18.45) 19.00–21.00
at Stockholm City Hall

June 1st – Lunch, 12.00–13.30 in the hotel Blique by Nobis restaurant Boketto

June 1st – Dinner, 19.00–21.00 in the hotel Blique by Nobis restaurant Boketto

June 2nd – Lunch, 12.30–14.00 in the hotel Blique by Nobis restaurant Boketto

Conference venue

At Hotel Blique by Nobis
Scientific conference session room: Blomsterkiosken

Contact organisers

For any queries, please contact us at conference2024@aniridi.se or send a message through whatsapp/phone to Neven Milivojevic, +46 70 639 00 68.

It is also possible to call the same number.

Directions

Maps and useful directions can be found on the conference web page:
<https://aniridiaconference.org/eac2024/logistics/>.

Exhibitors

At Hotel Blique by Nobis
Scientific conference session room: Blomsterkiosken

Hotel Blique by Nobis

Address: Gävlegatan 18
Phone: +46 8 557 666 00
Whatsapp: +46 8 557 666 01
E-mail: info@bliquebynobis.se

Poster presentations

At Hotel Blique by Nobis
Scientific conference session room: Blomsterkiosken

Public transport

It is easy to use the public transport in the Stockholm area. You can purchase a ticket by tapping your credit/debit card when entering the bus or train.
More information can be found at <https://sl.se/en/>.

Reception at Stockholm City Hall on May 31st

The meet-up is at 18.45 in the City Hall inner yard. There is no organised transport to the City Hall. The public blue bus no 3 leaves every 10 minutes about 250 meters from the hotel and it takes you directly to the Stockholm City Hall (bus stop: Stadshuset). Make sure you take the correct direction of the bus, which should be Södersjukhuset. The bus ride takes about 15 minutes but it might take longer in rush hour. Please allow time for transportation not to miss out on the check in. At the back of the City Hall there is a small park with some statues by Carl Eldh as well as one of the most beautiful panoramic views in Stockholm, well worth exploring. Dress code for the reception is smart casual. The hosts are the City of Stockholm and the Region of Stockholm, and they have invited us to a mingle buffet dinner.



LABORATOIRES KÔL
START-UP FOR RARE CORNEA DISEASES



22 Allée Alan Turing,
63 000 Clermont Ferrand
FRANCE

+33 (0) 463 467 818

<https://www.laboratoires-kol.com/>

contact@laboratoires-kol.com

@LaboratoiresKÔL

Omnigen®

Tereo®
processed
human amniotic
membrane



The natural
benefits of
amniotic
membrane
delivered to the
ocular surface

Omnilenz®

The benefits
of amniotic
membrane
in an outpatient
procedure



Bandage
contact lens
application of
Omnigen®
in a 4-6 minute
procedure

nu<vision®

www.nu-vision.co.uk

Support your patients' natural healing process



INNOVATIONS FOR THE EYES

We conduct research and development within all therapy areas of the eye. Our roots trace back to 1871 when the first generation of the Chibret family began manufacturing eye care medications.

From the outset, there was an understanding that all eye care medications could be simplified and improved.



Read more
about our innovations on
www.thea-nordic.com

In five generations, we have focused on innovative eye products. Family Chibret; Henri and Jean-Frédéric, Théa.

 **Théa** | let's open our eyes

Cooperation, support and sponsors

This conference would not have been possible without the kind and generous support and cooperation of our partners and donors. In addition to this our sponsors have contributed to making it possible for us to balance our finances. We are most grateful to all contributors!

Aniridi Norge

Aniridi Sverige

Aniridia Europe

AWS Aniridie-Wagr e.V.

Center for Rare Diseases, Karolinska University Hospital

City of Stockholm (Host Reception at City Hall)

Eurordis

Kronprinsessan Margaretas Arbetsnämnd för synskadade

Linköping University

Region Stockholm (Host Reception at City Hall)

Sigvard och Marianne Bernadottes forskningsstiftelse för barnögonvård

Synskadades riksförbund

Synskadades stiftelse

