

VI EUROPEAN ANIRIDIA CONFERENCE

3,4,5 JUNE 2022

Venue: San Juan Complex PSN, San Juan de Alicante, SPAIN

DRAFT PROGRAM

FRIDAY, JUNE 3

12:00 h. Ophthalmologic study and genetic assessment for patients with aniridia and relatives at Vissum Miranza facilities.

15:00 h. - 18:00 h. Aniridia Europe Community Meeting.

20:30 h. Welcome Networking dinner (Speakers, Chairs, Committee and Board members)

SATURDAY, JUNE 4

8:30 h. Registration.

9:00 h. Welcome message. *Prof. Jorge Alió y Sanz, Miguel Hernández University; Barbara Poli, Aniridia Europe; Yolanda Asenjo García, Spanish Aniridia Association; Local Authorities*

9.30 h Introduction of the agenda and presentation of the Monographic issue on Aniridia in “Archivos”, scientific review of the Spanish Ophthalmology Society SEO and statistical report of Aniridia in Spain. *Prof. Jorge Alió y Sanz, Miguel Hernández University; Spanish Aniridia Association*

10.00 h. Molecular diagnosis of Aniridia and allied conditions.
Dr. Marta Corton, Jiménez Díaz Foundation University Hospital-IIS, Spain.

10:30 h. Role of the European Networks and Registries in the development of new therapeutic strategies for Aniridia.
Prof. J. Carlos Pastor. IOBA. Emeritus Professor University of Valladolid.

11:00 h to 14:15 h. GENETICS AND WAGR SPECTRUM (ROOM 1)

11:00 h to 12:30 h. Aniridia genetic basis: from research to clinical application. *Chairperson: Prof. Carmen Ayuso. Fundación Jiménez Díaz, Madrid, Spain*

- **11:00 h. Coming soon.**
- **11:20 h. NGS and WGS as a combined clinical diagnostic strategy for aniridia and aniridia-like disorders: a French cohort of 450 patients including a total of 185 PAX6 positive families.** *Dr. Sophie Valleix, Necker-Enfants Malades Hospital, France.*
- **11:40 h. Genetic and reproductive counselling.** *Dr. Fiona Blanco-Kelly, Jiménez Díaz Foundation University Hospital-IIS, Spain.*
- **12:00 h. Lipid Nanoparticle-Mediated Delivery of CRISPR RNP Results in Wide-Spread In Vivo Genome Editing in Mouse Cornea.** *Prof. Elizabeth M. Simpson, British Columbia University, Canada.*

12:30 to 12:45 Break. Coffee is available during the session.

12:45 to 14:15 WAGR Spectrum

Chairperson: Kelly Trout, International WAGR Syndrome Association president, USA.

- **12:45 h. Overcoming barriers and accelerating research: lessons from the WAGR syndrome patient registry.** *Kelly Trout, IWSA president, USA.*
- **13:05 h. Defining the WAGR Spectrum: Updates from the WAGR Syndrome Patient Registry.** *Dr. Jennifer Kalish, Children's Hospital of Philadelphia, USA.*
- **13:25 h. Consequences of PAX6 mutations in the brain and/or endocrine system.** *Prof. James D. Lauderdale, University of Georgia, USA.*
- **13:45 h. Nephroblastoma in patients with WAGR syndrome and clinical consequences.** *Prof. Dr. Norbert Graf, Saarland University, Germany.*

11:00 to 14:15 WHAT'S NEW IN EYE RESEARCH AND CLINICAL CARE? (ROOM 2)

Chairpersons: Pr. Jorge Alió y Sanz & Pr. Rafael Barraquer

- **11:00 h. Changes in the Crystalline Lens in Aniridia.** Pr. Rafael Barraquer, International University of Catalonia
- **11:20 h. Corneal Aspects of Congenital Aniridia.** Dr. Fabian Fries, Saarland University, Germany.
- **11:40 h. Cultivated stem cells for corneal failure due to limbal deficiency in congenital aniridia.** Dr. José María Herreras, University Institute of Applied Ophthalmobiology IOBA, Spain.
- **12:00 h. Light management and vision correction with smart contact lenses.** Prof. Andrés Vásquez Quintero, Ghent University, Belgium.

12:20 to 12:35 Break. Coffee is available during the session

- **12:35 h. Pheno-genotypic correlations in foveolar aplasia in aniridia.** Dr. Pilar Casas de Llera, Fernández- Casas clinic, Spain.
- **12:55 h. Aniridia: relentless inflammation: mechanisms and potential new treatment.** Giulio Ferrari, Vita-Salute San Raffaele University, Italy.
- **13:15 h. Cataract surgery in the aniridic eye.** Dr. Juan Álvarez de Toledo, Barraquer Clinic, Spain.
- **13:35 h. Aniridic Glaucoma: Mechanisms and Management, including MIGS.** Prof. Peter Netland, University of Virginia, USA.
- **13:55 h. Neuroophthalmological findings in aniridia.** Prof. Barbara Käsmann-Kellner, Saarland University, Germany.

14:15 Lunch

15:30 h. Aniridia-net COST action: European cooperation in aniridia research.

Chairperson: Dr. Juliana Martínez Atienza.

- **15:30 h. Overview of the latest activity in collaborative European aniridia research.** Prof. Neil Lagali. Linköping University, Sweden
- **15:45 h. Foveal Hypoplasia Grading in 95 Cases of Congenital Aniridia: Correlation to Phenotype and PAX6 Genotype.** Dr. Dominique Bremond Gignac, OPTHARA, France

- **16:00 h. Identification and validation of small compounds to treat aniridia-related keratopathy.** *Dr. Daniel Aberdam. INSERM. France.*
- **16:15 h. iPSC as a tool for understanding aniridia-associated keratopathy.** *Prof. Heli Skottman, Tampere University, Finland.*
- **16:30 h. Regenerative medicine and cellular therapy for bilateral limbal stem cell deficiency.** *Dr. Mariya Moosajee. Moorfields Eye Hospital, UK.*
- **16:45 h. Questions and answers / Panel discussion**

17:20 - 17:35 Coffee Break

17:35 h to 19:00 h. Presentation of selected posters and communications. Rapid fire session for Early Career Scientists.

SUNDAY, JUNE 5

9:00- 12.00 h. Saturday panels: summary for patients by a representative of each table.

- **9.00-9.25 h.** Genetics Basis Summary.
 - *Prof. Carmen Ayuso, Prof. Carmen Ayuso. Fundación Jiménez Díaz, Madrid, Spain*
- **9.30- 9.55 h.** WAGR Spectrum Summary.
 - *Kelly Trout, IWSA, USA.*
- **10.00-10.55 h.** Eye Research and Clinical Care Summary.
 - *Dr. Juan Álvarez de Toledo, Barraquer Clinic, Spain.*

11:00 h-11.20 h. Coffee Break

- **11.30- 11.55 h.** Saturday COST Action Summary.
 - *Prof. Neil Lagali, Linköping University, Sweden*

12:00 h. Round tables for patients.

12.00-13.00 h. Social inclusion at school:

- **12.00 h.** Early attention and stimulation. *Sonia Rodríguez. Spanish National Organisation of the Blind (ONCE)*
- **12.30 h.** European project: "School for all, Seeing the invisible". *Victor García. RedTree.*

13. 00-13.30 h. International collaboration and life story:

- **13:15 h.** Collaboration and support to Spanish speaking countries. *Spanish Aniridia Association*
- **13:15 h.** Living with Aniridia. *Yolanda Asenjo, Spanish Aniridia President, Spain*

13:45 h. Official closing act. *Prof. Jorge Alió y Sanz, Miguel Hernández University; Barbara Poli, Aniridia Europe; Ivana Kildsgaard, Aniridia Sweden; Yolanda Asenjo García, Spanish Aniridia Association; Local Authorities.*

14:15 h. Lunch



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