

4TH EUROPEAN CONFERENCE ON ANIRIDIA
“Aniridia, iris and cornea developmental anomalies”



Saturday August 25th / Sunday August 26th 2018

CISP Ravel - 6 avenue Maurice Ravel - 75012 PARIS - FRANCE

Metro stations : Porte de Vincennes (L1)/ Bel Air (L6)

Tram T3A stations : Alexandra David-Néel/ Montempoivre

FOR HEALTH PROFESSIONALS

Saturday 25th of August

8:00/ 9:00 COFFEE BREAK

INTRODUCTION

9:00 to 9:30

Mrs G Jouanjan (France), president of Gêneris

Pr D Brémond-Gignac (France), president of Gêneris SC and Aniridia Europe SC

Mrs R Sanchez de Vega (Spain), president of Aniridia Europe

Mrs F Salama (France), member of AFM Téléthon and EURORDIS Board

Pr K Nischal (USA), president of WSPOS

SESSION 1 : UPDATE IN GENETICS IN ANIRIDIA

9:30 to 12:05 Moderators : Pr P Calvas (France)/ Pr E Semina (USA)

9:30 “Pax6 : what have we learned ?” - Dr P Callaerts (Belgium)

9:50 "Screening of PAX6 gene in Italian congenital aniridia patients" - Pr G Damante (Italy)

10:10 « Genetic Analysis of ‘PAX6-Negative’ Individuals with Aniridia” - Dr N Hall (UK)

10:30 “Analysis of genotype–phenotype correlations in aniridia in Russian federation” - Pr T Vasilyeva (Russia)

10:50/ 11:05 COFFEE BREAK



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11:05 « PAX6 unusual mutations and phenotypes » - Pr P Calvas (France)

11:20 LECTURE « Non pax6 mutation with aniridia » - Pr E Semina (USA)

11:50 QUESTIONS ANSWERS

12:05/ 13:15 LUNCH

SESSION 2 : SYSTEMIC DEFICIENCY RELATED TO ANIRIDIA

13:15 to 15:15 Moderators : Pr D Bremond-Gignac (France)/ Pr B Kaesmann-Kellner (Germany)

13:15 “Aniridia visual impairment : how to improve low vision ? “ Pr B Kaesmann-Kellner (Germany)

13:30 “The role of Pax6 and other ocular homeobox genes in circadian biology of the pineal gland” - Dr MF Rath (Denmark)

13:50 “Neuropaediatrics and aniridia ; diagnostic and rehabilitation” - Dr N Bahi-Buisson (France)

14:05 « Diabetes and obesity associated to aniridia » - (France)

14:20 LECTURE “Phenotype/ Genotype correlation in aniridia” - Pr D Bremond-Gignac (France)

14:50 QUESTIONS ANSWERS

15:05/ 15:25 COFFEE BREAK

SESSION 3 : SYNDROMIC ANIRIDIA

15:25 to 17:10 Moderators : Dr M Corton (Spain)/ Pr K Nischal (USA)

15:25 « Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome » - Dr L Fares Taie (France)

15:40 “Identification of novel and hotspot mutations in the channel domain of ITPR1 in two patients with Gillespie syndrome” - Pr G Zanni (Roma, Italy)

15:55 “Improving molecular diagnosis of aniridia and WAGR syndrome using customized targeted array-based CGH” - Dr M Corton (Spain)

16:10 “Management of Axenfeld-Rieger with aniridia” - Pr E Silva (Portugal)

16:25 LECTURE « Management of Peters Anomaly with aniridia” - Pr K Nischal (USA)

16:55 QUESTIONS ANSWERS

SESSION 4 : POSTERS SESSION

17:10 to 17:25 Moderators : Pr D Aberdam (France)/ Pr D Bremond-Gignac (France)

Flash presentation of the best posters selected by the scientific committee of the conference



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SESSION 5 : ANIRIDIA, OCULAR SURFACE AND CORNEA REHABILITATION

17:25 to 18:00 Moderators : Pr D Aberdam (France)/ Pr J Daniels (UK)/ Pr V Borderie (France)
17:30 “In vitro modeling of aniridia-related keratopathy (ARK). Potential for drug discovery and therapy” - **Pr D Aberdam (France)**
17:45 “Biology and future therapeutic possibilities for aniridia-related keratopathy” - **Pr Julie Daniels (UK)**

18:00 END OF THE DAY 1 OF THE CONFERENCE

Sunday 26th of August

8:30/ 9:00 COFFEE BREAK

INTRODUCTION

9:00 to 09:05 Mrs G Jouanjan (France) - Pr D Brémond-Gignac (France)

SESSION 5 : ANIRIDIA, OCULAR SURFACE AND CORNEA REHABILITATION

9:05 to 11:00 Moderators : Pr D Aberdam (France)/ Pr J Daniels (UK)/ Pr V Borderie (France)

9:05 “Aniridia and corneal ulcer therapy” - **Pr E Gabison (France)**
9:20 “Aniridia and Boston type I keratoprosthesis : anatomical survival and visual prognosis, post-surgical visual outcome and complications” - **Dr MF de la Paz (Spain)**
9:40 “Limbal stem cells niche” - **Pr V Borderie (France)**
9:55 « *Cell therapy for regeneration of the corneal epithelium in aniridic patients. Aniridia: Recent Developments in Scientific and Clinical Research* » - **Pr A Shortt (UK)**
10:10 “Congenital Aniridia and the Ocular Surface” - **Pr Neil Lagali (Sweden)**
10:25 “Histopathology Findings of Corneal Buttons in Congenital Aniridia Patients” - **Pr J Alvarez de Toledo (Spain)**
10:40 LECTURE "Progress Towards Gene Therapy in the Mouse Aniridic Cornea" - **Pr Elizabeth M Simpson (Canada)**

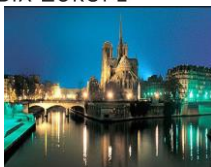
11:10 QUESTIONS ANSWERS



4TH EUROPEAN CONFERENCE ON ANIRIDIA “Aniridia, iris and cornea developmental anomalies”



ANIRIDIA EUROPE



11:25/ 11:35 COFFEE BREAK

SESSION 6 : PATIENTS & FAMILY SESSION

11:35 to 12:50 Moderators : Pr D Bremond-Gignac (France) / Mrs G Jouanjan (France)

11:35 SESSION 1 : 5 min for synthesis of the session 4 by moderators/ speakers + 15 min questions answers with patients and families

11:55 SESSION 2 : 5 min for synthesis of the session 4 by moderators/ speakers + 15 min questions answers with patients and families

12:15 SESSION 3 : 5 min for synthesis of the session 4 by moderators/ speakers + 15 min questions answers with patients and families

12:35 SESSION 5 : 5 min for synthesis of the session 4 by moderators/ speakers + 15 min questions answers with patients and families

CONCLUSION

12:55 to 13:00

Mrs R Sanchez de Vega (Spain)

Pr D Bremond-Gignac (France)

Mrs G Jouanjan (France)

13:00 END OF THE CONFERENCE

13:00 to 14:00 LUNCH

