

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



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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

