

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



**ONLY FOR PATIENTS AND FAMILIES
(WAGR patients included, All nationalities)**

Friday August 24TH 2018

Center of Reference of Rare Diseases in Ophthalmology
OPHTHARA- Necker Enfants Malades Hospital - Building Lecourbe 2
149, rue de Sèvres - 75015 Paris
Metro stations : Duroc (L13)/ Sèvres-Lecourbe (L6)

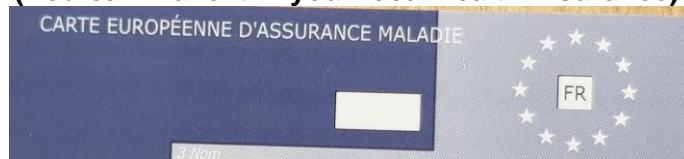
OPHTHALMOLOGICAL CONSULTATIONS FOR FOREIGN PATIENTS

With
PR BREMOND-GIGNAC, PR BORDERIE,
PR GABISON (FRANCE), PR NISCHAL (USA), DR ROBERT (FRANCE)

**8:30 APPOINTMENT FOR 12 PEOPLE
10:00 APPOINTMENT FOR 13 PEOPLE**

**PLEASE TAKE WITH YOU
The European Health Insurance Card for people from European countries**

(You can make it in your local health insurance)



4TH EUROPEAN CONFERENCE ON ANIRIDIA
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Friday August 24TH 2018

French Rares Diseases Platform

96 rue Didot - 75014 Paris

Metro station L13 : Plaisance/ Tram T3A station : Didot

ANIRIDIC PATIENTS & FAMILIES DAY (WAGR patients included, All nationalities)

With

PR ABERDAM (France) : “New therapies for aniridia”

PR SEMINA (USA) : “Aniridia and genetics counseling”

14:00/ 17:00 Room MARGUERITE

YOUTH ANIRIDIC DAY (Only for 15/ 30 years old, Youth WAGR included, All nationalities)

With

Mrs GALINA GENING (Russia) : Psycho-social impacts of aniridia

PR SEMINA (USA) : Aniridia and genetics counseling

17:00/ 19:00 Room MARGUERITE



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GENERAL ASSEMBLY OF ANIRIDIA EUROPE
Room PIVOINE 1
14:00/ 17:00
ONLY FOR
MEMBERS OF ANIRIDIA EUROPE

MEETING OF THE SCIENTIFIC COMMITTEE OF ANIRIDIA EUROPE
Room PETUNIA
17:00/ 19:00
ONLY FOR
MEMBERS OF ANIRIDIA EUROPE SCIENTIFIC COMMITTEE

DINNER
Room MARGUERITE
19:30
ONLY FOR
CLINICIANS FOR OPHTHALMOLOGICAL CONSULTATIONS ON AUGUST 24th,
GUEST SPEAKERS ON AUGUST 24th,
MEMBERS OF ANIRIDIA EUROPE SCIENTIFIC COMMITTEE and BOARD,
MEMBERS OF GENIRIS BOARD and ORGANIZATION TEAM



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



FOR HEALTH PROFESSIONALS, PATIENTS and FAMILIES

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

Saturday 25th of August

8:00/ 9:00 REGISTRATION/ PREVIEW & COFFEE BREAK

INTRODUCTION

9:00 to 9:30

9:05 Mrs G Jouanjan (France), president of Gêneris

9:10 Pr D Brémond-Gignac (France), president of Gêneris SC and Aniridia Europe SC

9:15 Mrs R Sanchez de Vega (Spain), president of Aniridia Europe

9:20 Mrs F Salama (France), member of AFM Téléthon and EURORDIS/ ERN Epag

9:25 Pr K Nischal (USA), president of WSPOS

SESSION 1 : UPDATE IN GENETICS IN ANIRIDIA

9:30 to 12:10 Moderators : Pr P Calvas (France)/ Pr E Semina (USA)

9:35 “Pax6 : what have we learned ?” - Dr P Callaerts (Belgium)

9:55 "Screening of PAX6 gene in Italian congenital aniridia patients" - Pr G Damante (Italy)

10:15 « Genetic Analysis of ‘PAX6-Negative’ Individuals with Aniridia” - Dr N Hall (UK)

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10:35 “Analysis of genotype–phenotype correlations in aniridia in Russian federation” - **Pr T Vasilyeva (Russia)**

10:55/ 11:10 COFFEE BREAK

11:10 « PAX6 unusual mutations and phenotypes » - **Pr P Calvas (France)**
 11:25 LECTURE “Genetic heterogeneity in aniridia phenotype” - **Pr E Semina (USA)**
 11:55 QUESTIONS ANSWERS

12:10/ 13:15 LUNCH

SESSION 2 : SYSTEMIC DEFICIENCY RELATED TO ANIRIDIA

13:15 to 14:40 **Moderators : Pr D Bremond-Gignac (France)**
 13:20 “The role of Pax6 and other ocular homeobox genes in circadian biology of the pineal gland” - **Dr MF Rath (Denmark)**
 13:40 “Neuropaediatrics and aniridia ; diagnostic and rehabilitation” - **Dr N Bahi-Buisson (France)**
 13:55 LECTURE “Phenotype/ Genotype correlation in aniridia” - **Pr D Bremond-Gignac (France)**
 14:25 QUESTIONS ANSWERS

SESSION 3 : SYNDROMIC ANIRIDIA

14:40 to 16:30 **Moderators : Dr M Corton (Spain)/ Pr K Nischal (USA)**
 14:45 « Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome » - **Dr L Fares Taie (France)**
 15:00 “Identification of novel and hotspot mutations in the channel domain of ITPR1 in two patients with Gillespie syndrome” - **Pr G Zanni (Italy)**
 15:15 “Improving molecular diagnosis of aniridia and WAGR syndrome using customized targeted array-based CGH” - **Dr M Corton (Spain)**

15:30/ 15:45 COFFEE BREAK

15:45 “Management of Axenfeld-Rieger with aniridia” - **Pr E Silva (Portugal)**
 16:00 LECTURE « Management of Peters Anomaly with aniridia” - **Pr K Nischal (USA)**
 16:30 QUESTIONS ANSWERS



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SESSION 4 : POSTERS SESSION

16:45 to 17:10 **Moderators : Pr D Aberdam (France)/ Pr D Bremond-Gignac (France)**

16:50 “Spectral-Domain Optical Coherence Tomography findings in 64 patients with aniridia : a retrospective study” – **Dr A Daruich (France)**

16:55 “Within-family phenotype-genotype discordance in congenital aniridia” – **Mrs H R. Pedersen (Norway)**

17:00 “Activity of Pax6+/- limbal epithelial stem cells in vivo” – **Pr JM Collinson (UK)**

17:05 “Functional splicing assays using in vitro minigenes to assess variants of uncertain significance in PAX6” - **Mrs M Tarilonte (Spain)**

SESSION 5 : ANIRIDIA, OCULAR SURFACE AND CORNEA REHABILITATION

17:10 to 18:00 **Moderators : Pr D Aberdam (France)/ Pr J Daniels (UK)/ Pr V Borderie (France)**

17:15 “In vitro modeling of aniridia-related keratopathy (ARK). Potential for drug discovery and therapy” - **Pr D Aberdam (France)**

17:30 “Biology and future therapeutic possibilities for aniridia-related keratopathy” - **Pr J Daniels (UK)**

17:45 **QUESTIONS ANSWERS**

18:00 **END OF THE DAY 1 OF THE CONFERENCE**

20:00 **GALA DINNER – CISP – “LA RESIDENCE” RESTAURANT**

ONLY FOR

GUEST SPEAKERS OF THE CONFERENCE,

MEMBERS OF ANIRIDIA EUROPE SCIENTIFIC COMMITTEE and BOARD,

MEMBERS OF GENIRIS SCIENTIFIC COMMITTEE, BOARD and ORGANIZATION TEAM



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FOR HEALTH PROFESSIONALS, PATIENTS and FAMILIES

Sunday 26th of August

8:30/ 9:00 REGISTRATION/ PREVIEW & 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)

9:10 “Aniridia and corneal ulcer therapy” - Pr E Gabison (France)

9:25 “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 “Congenital Aniridia and the Ocular Surface” - Pr Neil Lagali (Sweden)

10:10 “Histopathology Findings of Corneal Buttons in Congenital Aniridia Patients” - Pr J Alvarez de Toledo (Spain)

10:25 LECTURE “Progress Towards Gene Therapy in the Mouse Aniridic Cornea” - Pr Elizabeth M Simpson (Canada)

10:55 QUESTIONS ANSWERS



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11:10/ 11:30
COFFEE BREAK

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PRESENTATION SESSION FOR UNSELECTED POSTERS IN THE LOBBY (3 minutes by poster)

SESSION 6 : PATIENTS & FAMILY SESSION

11:30 to 12:30 Moderators : Pr D Bremond-Gignac (France) / Mrs G Jouanjan (France)

11:35 **SESSION 1** : 5 min for synthesis of the session 1 by moderators/ speakers + 10 min questions answers with patients and families

11:50 **SESSION 2** : 5 min for synthesis of the session 2 by moderators/ speakers + 10 min questions answers with patients and families

12:05 **SESSION 3** : 5 min for synthesis of the session 3 by moderators/ speakers + 10 min questions answers with patients and families

12:20 **SESSION 5** : 5 min for synthesis of the session 5 by moderators/ speakers + 10 min questions answers with patients and families

CONCLUSION

12:35 Mrs R Sanchez de Vega (Spain)

12:40 Pr D Bremond-Gignac (France)

12:45 Mrs G Jouanjan (France)

12:50/ 13:00 END OF THE CONFERENCE

13:00 to 14:00 LUNCH

