

# 3rd European Aniridia Conference

## and Patient's Day, Duisburg, Germany

Location: Schauinsland-Reisen Travel Agency Headquarter, Stresemannstr. 80, 47051 Duisburg

Program Outline from August 26th to August 28th, 2016

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Friday, 26 <sup>th</sup> of August	Patient's Day (13:00 – 18:00)	Patiententag (13:00 – 18:00)
	<b>3<sup>rd</sup> Scientific European Aniridia Conference</b>	<b>3. Europäische Wissenschaftliche Aniridie-Tagung</b>
<b>Saturday, 27<sup>th</sup> of August</b>		
<b>09:00 – 10:40</b>	General ophthalmological manifestations of aniridia, Genetics	Allgemeine Augenbeteiligungen bei Aniridie, Genetik
<b>11:00 – 11:20</b>	Official opening of the 3 <sup>rd</sup> European Aniridia Conference	Offizielle Eröffnung und Begrüßung zur 3. Europ. Aniridietagung
<b>11:20 – 12:30</b>	Developmental and cellular biology	Zelluläre und entwicklungsbezogene Biologie
<b>12:30 – 13:30</b>	Lunch and Poster presentation	Mittagsimbiß und Postervorstellung
<b>13:30 – 15:15</b>	Experimental Ophthalmology	Experimentelle Ophthalmologie
<b>15:45 – 18:15</b>	Clinical Cornea, Neuroscience and Low Vision, Quality of Life	Klinische Hornhautthemen, Sehbehinderung, Lebensqualität
<b>19:00</b>	Closed Session: Lecturers' dinner with AWS & AE representatives	Geschlossene Veranstaltung: Abendessen Referenten, AWS, AE
<b>Sunday, 28<sup>th</sup> of August</b>		
<b>09:00 – 10:25</b>	WAGR – Syndrome & Wims Tumor	WAGR – Syndrom & Wilms Tumor
<b>10:45 – 12:00</b>	Cataract and Glaucoma in Aniridia	Katarakt und Glaukom bei Aniridie
<b>12:00 – 13:00</b>	Lunch and Poster presentation	Mittagsimbiß und Postervorstellung
<b>13:00 – 16:00</b>	Summary of important conference messages for patients (in German), announcements (eg 4 <sup>th</sup> European Aniridia Conference in Paris), acknowledgements of sponsors and supporting organisations, closing remarks and Farewell	Zusammenfassung wichtiger Tagungsergebnisse (auf Deutsch), Vor-Ankündigungen (zB 4. Europäische Aniridie-Konferenz in Paris), Danksagungen an Sponsoren und unterstützende Organisationen, Schlussworte und Verabschiedung



Supported by COST action BM1302

**3RD EUROPEAN  
ANIRIDIA CONFERENCE**

**27<sup>th</sup> – 28<sup>th</sup> of August, 2016  
Duisburg, Germany**



## SCIENTIFIC PROGRAM

**PLEASE NOTE:** Only the first speakers are named in the time schedule, please see [abstract booklet](#) for [complete](#) lists of authors  
Please observe the given lecture times (in brackets) to avoid overrunning the session times  
The posters should be discussed during lunch break. Abstracts in the booklet are ordered according to the time schedule.

### SATURDAY AUGUST 27<sup>TH</sup>, 2016

CHAIR OF SESSION

#### CLINICAL OPHTHALMOLOGY – HISTORY OF ANIRIDIA AND OPHTHALMOLOGY OF ANIRIDIA

09:00 – 09:45	Erlend S. Landsend, NO	Spectrum of ophthalmological findings in PAX6 related aniridia	(12)	Berthold Seitz Neil Lagali
	Barbara Käsmann-Kellner, GER	Clinical findings in Aniridia without PAX6 Mutation	(10)	
	Hyun Taek Lim, KOREA	Clinical Features of Korean Patients with Congenital Aniridia with PAX6 mutation and without	(10)	
	Jens Martin Rohrbach	History and future of aniridia	(12)	

#### GENETICS

09:45 – 10:40	Veronica v. Heyningen, UK	PAX6 and other genes: mutations and phenotypes	(20)	Daniel Aberdam Veronica van Heyningen
	Hanno Bolz, GER	Genetics of aniridia	(15)	
	Daniel Aberdam, FR	Cellular modelisation of aniridia for molec. characterization and gene therapy	(20)	

10:40 – 11:00	<b>COFFEE BREAK</b>			
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## OFFICIAL OPENING OF THE 3RD EUROPEAN ANIRIDIA CONFERENCE

11:00 – 11:20	Denice Toews-Hennig, President Aniridie-WAGR Support Group Germany <b>AWS</b>	(5)	
	Barbara Käsmann-Kellner, GER, German Aniridia Center, Medical Advisor <b>AWS</b>	(5)	
	Asbjørn Akerlie, President of Aniridia Europe <b>AE</b>	(5)	
	Manfred Osenger, Governing Mayor of the City of Duisburg	(5)	

## DEVELOPMENTAL AND CELLULAR BIOLOGY

11:20 – 12:30	Cheryl Gregory-Evans, CA	Latest developments in RNA therapies for aniridia	(20)	Tor Utheim Lorenz Latta
	James Lauderdale, USA	Neuroanatomical changes in adult humans and mice deficient in Pax6	(20)	
	Ursula Schlötzer-Schrehardt, D	Molecular anatomy of the limbal stem cell niche	(20)	
12:30 – 13:30	<b>LUNCH AND POSTER DISCUSSION: EDEN ET AL: LONG TERM FOLLOW UP OF PATIENTS WITH CONGENITAL ANIRIDIA</b>			

## EXPERIMENTAL OPHTHALMOLOGY AND MEMBERS OF COST

13:30 – 15:15	Tor Paaske Utheim, NO	Treatment options for limbal stem cell deficiency	(20)	Claus Cursiefen Neil Lagali
	Oygunn Utheim, NO	Impact of storage conditions on human cultivated corneal cells	(15)	
	Neil Lagali, SE	Imaging limbal stem cell niche degradation and progression of aniridia related keratopathy in several patient cohorts	(20)	
	Che Connon, UK	Role of stromal stiffness in maintaining corneal epithelial phenotype: importance to homeostasis and corneal bioengineering	(20)	
	Cusiefen & Lagali	Current and future European research on rare and complex ocular surface diseases (e.g. Aniridia)	(15)	
	Lorenz Latta, GER	Opportunities and disadvantages of aniridia research process in clinical setting	(10)	

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15:15 – 15:45	<b>COFFEE BREAK</b>
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## CLINICAL OPHTHALMOLOGY: CORNEA

15:45 – 16:50	Gerd Geerling, GER	Corneal diagnostic tools for ocular surface disease (20)	Gerd Geerling Nguyen Nhung
	Claus Cursiefen, GER	Boston-keratoprosthesis: Preliminary experiences in high-risk eyes from the Department of Ophthalmology of the University of Cologne (20)	
	Berthold Seitz, GER	Excimer-PTK and Keratoplasty in aniridia (20)	

## NEUROSCIENCE, LOW VISION ASPECTS, QUALITY OF LIFE

16:50 – 18:15	Irene Gottlob, UK	PAX6 mutation without aniridia in a large family and OCT observations of foveal development in normal and congenitally impaired vision (20)	Irene Gottlob Barbara Käsmann-Kellner
	Nguyen Nhung, GER	Low Vision Care in aniridia patient: potential and challenge (20)	
	Barbara Käsmann-Kellner, GER	Ocular surface problems in aniridia – patients' experiences (12)	
	Rosa Sanchez de Vega (ES), Katie Atkinson (UK), Joeri van der Bosch (NL)	Living with aniridia – the view of affected persons (20)	

19:00	<b>CLOSED EVENT:</b> <b>LECTURER AND AWS / AE DINNER: JUST AT THE WATERSIDE OF EUROPE'S LARGEST INNER HARBOURS:</b> <b>DIEBELS IM HAFEN - WITHIN WALKING DISTANCE FROM THE CONFERENCE SITE</b>	
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**SUNDAY AUGUST 28<sup>TH</sup>, 2016**

**WAGR SYNDROME & WILMS TUMOR, METABOL. ALTERATIONS IN PAX6 HAPLOINSUFFICIENCY**

09:00 – 10:25	Kelly Trout, USA, <a href="http://www.wagr.org">www.wagr.org</a>	WAGR Syndrome: Guidelines for Clinical Management	(20)	Joan Han Norbert Graf
	Joan Han, USA	Role of brain-derived neurotrophic factor in obesity, intellectual disability, and impaired nociception in patients with WAGR syndrome	(20)	
	Norbert Graf, GER	Wilms-Tumor: treatment protocols and quality of life	(20)	
	Barbara Käsmann-Kellner, GER	Ophthalmological challenges in WAGR patients	(10)	
	Joan Han, USA	Sleep disturbances in PAX6 haploinsufficiency	(10)	
10:25 – 10:45	<b>COFFEE BREAK</b>			

**CLINICAL OPHTHALMOLOGY: CATARACT & GLAUCOMA**

10:45 – 12:00	Dominique Bremond-G., FR	Cataract in aniridic patients: special features and personal experience	(20)	Peter Netland Arne Viestenz
	Vadim Bondar, RUSSIA	Risks of Artificial Iris implants	(15)	
	Peter Netland, USA	Glaucoma and Cataract surgery in aniridia with an emphasis on glaucoma tube surgery	(20)	
	Arne Viestenz, GER	Secondary glaucoma in aniridia: anatomy of the anterior chamber angle and consequences for trabeculotomy	(20)	
12:00 – 13:00	<b>LUNCH AND POSTER DISCUSSION: BEHAEGEL ET AL: REGENERATION OF THE ANTERIOR CORNEA THROUGH A TRANSPLANTATION OF CULTIVATED LIMBAL EPITHELIAL STEM CELLS</b>			

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## CONCLUDING SESSION WITH QUESTION TIME FOR LISTENING PATIENTS & FAREWELL

13:00 – 15:45	Barbara Käsmann-Kellner Other Speakers: Tba	<b>Summaries of the scientific talks (in German)</b> <b>Options to ask questions to the attendant clinical and scientific researcher</b> <b>Zusammenfassungen der wesentlichen Ergebnisse der wissenschaftlichen Vorträge</b> <b>Möglichkeiten, den anwesenden Klinikern und Laborforschern Fragen zu stellen</b>	(90)
	Barbara Käsmann-Kellner & Denice Toews-Hennig, both AWS and Union members	Aniridia and soccer – Tribute & Thank you to 1. FC UNION Berlin, Soccer Club, 2 <sup>nd</sup> Bundesliga	(5)
	<b>Concluding remarks and thank you</b>		
	Denice Toews-Hennig, President Aniridia-WAGR Support Group AWS Germany		
	Barbara Käsmann-Kellner, GER, German Aniridia Center, Medical Advisor AWS Germany		
	New President of Aniridia Europe AE, Country tba		
	Gaelle Jouanjan, Geniris, and Dominique Bremond-Gignac, France: Information on European Reference Network ERN EYE and Announcement of the 4 <sup>th</sup> European Aniridia Conference 2018, Paris, France		

**WE CORDIALLY AND SINCERELY THANK OUR SPONSORS TO HELP MAKING THE 3RD EUROPEAN ANIRIDIA CONFERENCE A REALITY:**

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