

Aniridia Europe E-Newsletter. January 2021

Aniridia Europe General Assembly 2020

Aniridia Europe AE is the European Federation of 13 National Aniridia Associations and representatives in many European countries. Officially based in Norway, AE was created in 2011, with the purpose to promote research on this rare eye disorder and associated syndromes, to bridge professionals and patients and to improve care and quality of life of people with aniridia.

Aniridia Europe held its online General Assembly in July 15th 2020, where the activity report 2018-2020 and the financial report 2018-2019 were approved, as well as the activities and the budget for 2020-2022. Representatives from 11 full member associations, with the right to vote, participated in the meeting. In this event, a new board was elected. Please see the new board at <https://www.aniridia.eu/about-us/trustees-2020-2022/>

A long way to go: our wishes for 2021

In 2021 Aniridia Europe will reach 10 years since its foundation, and 18 years since the very first contacts started in Paris in 2003.

Today, Aniridia Europe is a federation that gathers 13 national associations and more than 20 groups or reference persons in Europe, and has contacts with aniridia representatives worldwide. In the past years, our community has organized four scientific conferences with researchers and clinicians from all over the world, and the next one will be held as a digital event in summer 2021. Guidelines for clinical management and tools for sport practice and social and school inclusion have been developed and disseminated. We are currently involved in European projects of scientific research, network building and social inclusion. Through our close collaboration with the scientists, we have learned more about aniridia as a condition, being much more than an eye disease, and this is critical for the development of new therapies and treatments



Very little of this would have been predictable or even imaginable in 2003 or in 2011.

Still, we experience today many criticalities: lack of financial support, human resources based only on voluntary work, a mosaic of countries with different healthcare systems and legislation, to which we can add the systemic difficulties that we all face as citizens at national and international level, that have a hard impact in the field of rare diseases. Diseases that came out of the shadows only a couple of decades ago and that risk to be overshadowed again by the great problems of our time.

We have to be aware that every small step forward requires a lot of determination and energy, that there is still a long way to go to improve the quality of life of people with aniridia, and try not to be



pointed if our expectations are not met as fast as we would like. This is why we must be resilient and united, support each other, extend our collaboration with scientists and institutions and build the next generation of younger people that will continue our work.

I am confident that we can do it, because our community has grown up and we can count on people with knowledge and skills, good will and smart ideas, that share a vision of a better future for our fellows and our children. I am honored to be part of this.

In the next months, our priorities will be to follow up with the projects already going on (COST Action Aniridia-Net and Erasmus+ A school for all) and with the incoming ones, as well as to support Aniridia Network in the organization of the 5th European Aniridia Conference. We are also planning to hold some online meetings to share information and strengthen our community.

I wish us all that the new year 2021 will be full of harmony and achievements

Barbara Poli

President of Aniridia Europe



EUROPEAN ANIRIDIA CONFERENCE 2021



Make a note in your calendar now: You are invited to the next European Aniridia Conference 30 July to 1 August 2021. It will be held online and hopefully in London too.

As in the past, it will share scientific knowledge to prevent sight loss and deal with aniridia's effects. It brings together patients and the world's top experts to upskill the clinical, research and aniridic communities. It will also focus on the lives of people affected by aniridia.

The event was postponed in summer 2020. We are now arranging to run it primarily as a virtual conference. There may still be an associated physical element, perhaps at Moorfields Eye Hospital, depending on the pandemic situation. The new agenda will be just as, if not more exciting than the original! Although the valuable face to face interaction may not be possible, compared with previous conferences, we hope the internet will help to have more sessions, speakers, attendees and translations

We especially want people from small and eastern European countries to join in to build supportive networks. Scientists at the start of their careers are also particularly encouraged to get involved to increase research collaboration and understanding about aniridia. If that sounds like you, contact us with your ideas for making it a success for you and your peers. Write to: conference@aniridia.org.uk or to request a chance to present a poster or possibly even a session fill in the Call For Papers form. Latest news at: <https://2020.aniridiaconference.eu/news/>

Please pass this invitation on to anyone who might be interested. If you haven't already, register your interest to get news as it becomes available.

<https://aniridia.org.uk/conference/>



BREAKING NEWS

AAK-INSIGHT PROJECT ON ANIRIDIA KERATOPATHY, GRANTED BY EUROPEAN UNION!

The application entitled 'Aniridia – novel therapeutic tools to treat or prevent progressive corneal opacification (AAK-INSIGHT)' was submitted by a consortium of researchers from several countries, led by Prof. Neil Lagali (Linköping University, Sweden) and Prof. Daniel Aberdam (CNRS, Biologie, Inserm-Technion, France)

The AAK-INSIGHT project will last 3 years and will have a budget of 1.4 million Euro. Its overall objective is to find a drug able to induce the production of PAX6 protein in the eye and cornea; the drug will be tested in cells and eye models of aniridia.

Among the scientists involved are Prof. Dominique Bremond-Gignac (chair of Aniridia Europe Scientific Committee), and the teams of Prof. Barbara Käsmann-Kellner and Prof. Nora Szentmary in Homburg (Germany), Prof. Martin Collinson in Aberdeen (UK), Dr Jo Zhou from the Netherlands, and Prof. Ruth Ashery-Padan from Israel. Some of these investigators belong to our Scientific Committee, or are in contact with it.

Aniridia Europe will be in the Patient Advisory Board of the project, as well as our member associations Gênis, Aniridi Sverige and Asociación Española de Aniridia.

Please, see <https://www.aniridia.eu/2020/12/28/aak-insight-project-on-aniridia-keratopathy-granted-by-the-european-union/>

COST ACTION CA18116 ANIRIDIA-NET

Aniridia: Networking to address an unmet medical, scientific and societal challenge. Acronym: Aniridia-Netpt



These four year program has among its purposes, to Improve aniridia management through evidence-based research, harmonized clinical protocols, pooling/sharing of samples and models and consensus activities. There have been two Management Committee and Working Groups meetings in 2020.

- Lisbon. 27-28th February. University of Lisbon, Institute of Molecular Medicine.

- Online meeting on 26th October.. A highly successful meeting that gathered ophthalmologist, researchers, industry and patients' organizations members from 22 countries. to present the current progress and to stimulate a closer collaboration.

The different Working Groups have met online over the year to coordinate, share tasks and update information. For ex. in WG 1, authors and topics have been decided, in WG6, a pilot questionnaire has been developed to be tested, in WG 7 a calendar with pictures and drawings made by children with aniridia has just been published.



You can see more information about the working groups, publications and meetings at: <https://aniridia-net.eu/>

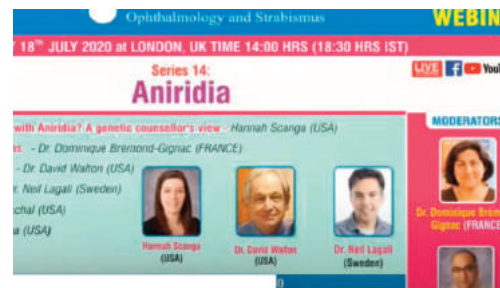
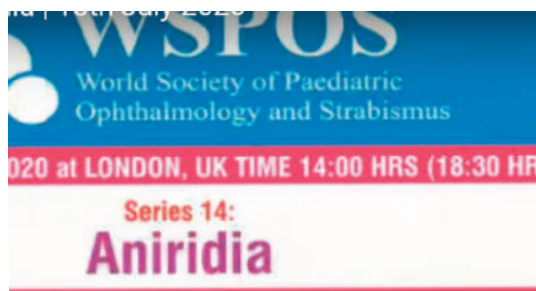
A new successful @AniridiaNet scientific mission at Oslo University by Dr. Gerard Boix Lemonche in PetrovskiLab. This mission focused on the production of bioengineered 3D corneal scaffolds and serves the basis for future collaborations. More at: [@COSTprogramme](https://bit.ly/3fnqIVe)



WEBINAR ON ANIRIDIA AT THE WSPOS CONGRESS

Online Webinar at WSPOS (World Society of Pediatric Ophthalmology and Strabismus), by Pr. Dominique Bremond and Pr. Neil Lagali.

There were over 5000 live attendees during the webinar, from around the world. Patients and doctors are invited to watch the video <https://www.youtube.com/watch?v=V-irFyEJt9>



SESSION ON ANIRIDIA AT THE GERMAN OPHTHALMOLOGY CONGRESS

There was a session on Aniridia in the Annual Congress of the German Ophthalmology Society, 10 October 2020, with the Title: "Congenital Aniridia – new clinical, genetic and molecular insights from patient cohorts in 5 European countries" Program below:.

Chair: Nóra Szentmáry (Homburg/Saar), Neil Lagali (Linköping)

"Aniridia is no prerequisite for Aniridia - broad phenotypic spectrum in PAX6 Syndrome". Barbara Käsman-Kellner (Homburg/Saar)

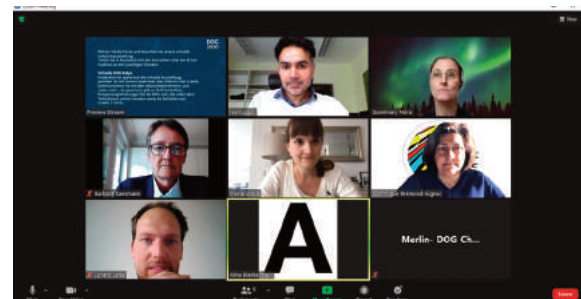
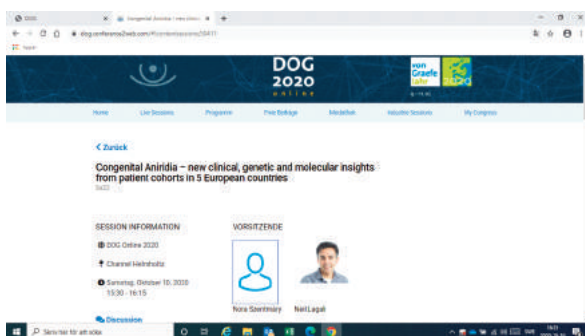


“Limbal stem cell deficiency in aniridia”. Maria Notara (Köln)

“Lessons from transcriptional analyses of ocular surface cells in congenital aniridia. Lorenz Latta (Homburg/Saar)

“Aniridia-associated keratopathy: origins, phenotype, genetics and prognosis based on European cohort studies”. Neil Lagali (Linköping)

“OCT in a French cohort of congenital aniridia”. Dominique Bremond-Gignac (Paris)



EUROPEAN REFERENCE NETWORK FOR EYE DISORDERS ERN-EYE

ERN-EYE, in its last meeting in October 2020, 15 new affiliated members of ern eye (European network for rare eye diseases) were approved-

- Spain: Hospital Universitari de Bellvitge, Barcelona, Hospital de Sant Joan de Déu, Barcelona, Hospital Clínic I Provincial de Barcelona, Barcelona, Hospital Universitario Cruces, Barakaldo (Bizkaia), Hospital Universitario Clínico San Carlos, Madrid, Hospital Universitario La Paz, Madrid,, Hospital Clínico Universitario de Valladolid, Valladolid.

- Austria: Styrian KAGES University Hospital Graz, Graz, Kepler Universitätsklinikum Linz, Linz, Center for Inherited Retinal Disorders and Neuroophthalmology, University of Vienna, Vienna,

- Slovenia: Eye Hospital, University Medical Centre Ljubljana, Ljubljana.

- Slovakia. W National Institute of Children's Diseases, Department of Paediatric Ophthalmology of Medical Faculty Comenius University, Bratislava.

- Malta. Mater Dei Hospital, Msida.

Most of them are specialized in Anterior segment of the eye.

New full members of ern eye (selection is still in progress) will be known in 2021. Thanks to Pr Bremond-Gignac and Geniris/ AE patient organisations, European guidelines for congenital aniridia is definitively validated as an objective of ERN-EYE



UPDATE ON ANIRIDIA RESEARCH IN RUSSIA.

Tatyana Vasilyeva

1. Molecular biologists of the Research Centre for Medical Genetics in Moscow have been investigating how changes in aniridia patients derived DNA (mutations) influence PAX6 mRNA splicing and abundance, as well as PAX6 protein synthesis efficiency in cell culture. A way to find the answer is to use computational prediction analysis, minigene assays, as well as luciferase tests and other techniques that could help to visualize consequences of primary nucleotide sequence changes. PAX6 mutations investigation at genome, transcriptome, proteome levels allows to reveal mutation functional mechanisms and further study of the possibility of its correction.
2. Concerning well known fact about an extremely high frequency of chromosomal rearrangements associated with congenital aniridia last year we carried out several studies and published two articles on the topic. Firstly, while analyzing the origin of identified in patients deletions we established they arose preferentially on the paternal alleles. PMID:32708836. Secondly, we reported a detailed information on the diagnostic of a sporadic case of congenital aniridia caused by pericentric inversion inv(11)(p13q14) associated with a 977 kb deletion in the 11p13 region. PMID:32948199.
3. Also, last year we eventually published the results of our statistical analysis of aniridia genotype-phenotype correlations, which allowed us to distinguish a milder phenotype in the carriers of PAX6 3'-cis regulatory region deletions. PMID:32467297Eud

ERASMUS+PROJECT. LOOKING OUT FOR A SCHOOL FOR ALL



Presential meetings were planned for this year, but Covid 19 made us take new coordination strategies and alternatives to go on with the development of the project. .We managed to hold the Third Transnational Meeting in Florence on February 21st, but the rest of the meetings had to be online, as well as the dissemination of the results and our participation in both Albinism Day (June 13th) and Aniridia Day (June 21st).

In Florence, advances on two intellectual products were presented. "Educational game: ict tool for the inclusion of students with visual impairments in the classrooms" and "Virtual training course for teachers and early education centers for the inclusion of students with visual impairment in their classrooms" and the advances in the new website, that was presented in July.

We assessed the challenges for the future multiplier events and decided to ask SEPIE for online multiplier events meetings and to postpone the deadline of the project to mid 2021, that was finally approved.

The virtual meeting was held on July the 17th., where control mechanisms were adopted, to keep quality and impact standards.for the dissemination of the project and its results.Please, see: <http://www.schoolforall.eu>



ANIRIDIA RUSSIA



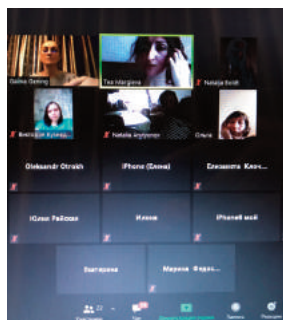
Celebrating of International Aniridia Day - June 21st

We created two events for the International Aniridia Day :

1. The International Art Competition for children "Drawing on the theme of Aniridia" 31 children from different countries sent to us 35 lovely pictures. All the participants and winners of the competition received the certificates.



2. Another event was the recording of short videos on the topic of aniridia in different languages . We received 51 videos from 19 different countries in 26 languages. Then from those videos we created one congratulatory video - <https://youtu.be/DdKA13k2i8A> All the participants got the certificates.



Celebrating of International WAGR syndrome awareness day - November 13th

On November 13, at the International WAGR syndrome Awareness day we had a Zoom-meeting in the format of Q&A between Russian-speaking WAGR families and doctors of the Russian Aniridia Center : neurologist, nephrologist, endocrinologist, neuropsychologist, vaccinologist and microbiologist from Research Centre for Medical Genetics Russian Academy of Science.

ANIRIDIA SPAIN



Aniridia Online Conferences

The Spanish Aniridia Association has launched a series of online events:

- On Aniridia Day the Association held a live conference with the participation of four professionals, Miguel Teus Univ. Alcalá. Madrid "Glaucoma associated to aniridia: diagnosis and and clinical/surgical treatment", Marta Corton, geneticist IIS-FJD. Madrid "The relevance of the genetic study in aniridia, Juan Álvarez-T Barraquer Clinic. Barcelona "Cataracts and ocular surface in patients with aniridia, which are the most suitable solution?" and Miguel Guzmán "Low vision in aniridia.





- In October, weekly live talks were made to discuss issues related to low vision and aniridia such as accessibility in public transport, early stimulation, embryonic genetic selection or acceptance and normalization of visual impairment. Both are available on AEA youtube channel. <https://www.youtube.com/channel/UCSKWgHNrtDasDTW0XhwYEXA>

Workshope in schools: “We are not invisible, we have low vision”

This year, the AEA has hold some workshops in different schools presenting Low Vision and Aniridia to kids. The children play some roles and videos are presented, so that they understand what this pathology is.

After a few months of uncertainty, with the collaboration of the teachers, the association has been able to reinvent the original campaign (face-to-face) to be able to carry it out online and safely for the study More information in Spanish at www.aniridia.es

ANIRIDIA IN FRANCE, MAGHREB AND BENELUX



French Funding and participation in aniridia research

In 2020, GROUPAMA 62, Euler Hermès and GÊNIRIS teamed up for fund a 5 years research project of Pr Brémond-Gignac on the anterior and posterior segment, the ocular surface and the tear film in the aniridia patient.

AFM TELETHON supports aniridia and GÊNIRIS for a new French research program.

GÊNIRIS participates in the patient advisory board of “AN INSIGHT” European consortium project, initiated in France by Pr Aberdam and Pr Brémond-Gignac. Selected by EJP-RD 2020, it will be funded for 3 years with 1.4 million euros. The aim of the project is to validate in vitro and in vivo three chemical molecules that Pr Aberdam's team has identified. The objective is to demonstrate their ability to correct keratopathy associated with aniridia.

Pr Brémond-Gignac. President of GÊNIRIS SC, has developed an Ataluren eye drops with the team of Necker Hospital and has filed for a patent. This is an innovative lead for the first treatment of eye drops in corneal complications due to congenital aniridia.

GÊNIRIS has funded more than 30ke at French Solidarity Heores Race for future research.



ANIRIDIA ITALIA

Aniridia Italy is writing some funny stories, in which the characters are the Aniridiex, the superheroes with black eyes and a coloured heart. Each boy and each girl is a superhero with a specific super power. This is a drawing that represents the young group of Aniridia Italy



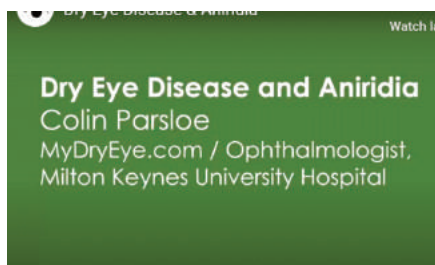
ANIRIDIA NETWORK UK & IRELAND



Aniridia Network Online Conference 2020

This year's Aniridia Day was more important than ever. The absence of our usual conference, due to pandemic restrictions, made it vital to find another way of connecting aniridia patients with one another, and with those who support them.

We therefore held a special online conference, providing an afternoon of free, informative presentations and discussions about aniridia. Anyone was welcome to attend from all over the world, especially patients with aniridia and their relatives in the UK and Ireland. Watch recordings of the event on Youtube.



Latest news: Gene.Vision website about aniridia launched in UK

<https://www.aniridia.eu/2021/01/06/gene-vision-website-about-aniridia-launched-in-uk/>