



Aniridia Europe E-Newsletter. July 2019

Aniridia Europe is a European Federation of National Aniridia Associations and individuals with aniridia, officially based in Norway, created in 2011 with the purpose of promoting research on this rare eye disorder and associated syndromes, bridging professionals and patients, to improve care and quality of life of people with aniridia.

We very happy to welcome two new National aniridia associations as full members in Ukraine and Lithuania, that makes 14 National associations and one support group. Please, see <https://www.aniridia.eu/in-your-country/>

ERASMUS+PROJECT. LOOKING OUT FOR A SCHOOL FOR ALL: early educational inclusion for students with low vision



First transnational meeting. 30 March 2019, Valencia (Spai

As it was the first transnational meeting, the partners (among them: Aniridia Europe, Aniridia Italiana and Aniridia Norge) introduced themselves and defined the intellectual outputs of the project and covered issues like visibility. The meeting was organised by RedTree, leader of the project and Aniridia Europe was represented by Rosa Sánchez de Vega and Lucía Mosquera. Soelvi Oesternvik and Torve Hauge on behalf of Aniridia Norge and Corrado Teofili on behalf of Aniridia Italiana.

The e-learning course "virtual training course for teachers and early education centres for the inclusion of students with visual impairment in their classrooms" was presented, as well as the educational game (App) "ict tool for the inclusion of students with visual impairment in their classrooms", its structure, characteristics and objectives were started to be developed. The partner entities shared their reality, experiences of people with visual impairment and the obstacles they face, thus enriching the debate.

Towards the end of the meeting, proposals were made regarding the dissemination of the project, that is, how to get the results to the beneficiaries: early education teachers and nursery schools, children with low vision (with Aniridia and Albinism) the population in general.

The next meeting will be in Sandefjord, Norway, September 14th 2019.





COST ACTION: ANIRIDIA-NET

Aniridia Networking to address an unmet medical, scientific, and societal challenge



Kick-off Management Committee Meeting. Brussels, 11-12 April 2019

The Management Committee MC, where Aniridia Europe was represented by Ivana Kildsgaard, met in Brussels, to present the objectives and deliverables to the participants.

The management positions were also elected: Chair Neil Lagali (SE), Vice-Chair Claus Cursiefen (DE), STSM Coordinator Thomas Ritter (IE), ITC Conference Grant Coordinator Sara Xapelli (PT), Training School/Workshop Coordinator Rozalia Hristova (BG), Science Communicator Juliana Martinez-Atienza (ES)

The different working groups & leaders were also appointed: WG1: Clinical Guidelines, Dominique Bremond-Gignac (FR). WG2: Clinical and Cohort Studies, Vito Romano (UK). WG3: Stem Cells and Regenerative Medicine, Stefano Ferrari (IT). WG4: Transplantation, Inflammation and Immunity, Davide Borroni (LV). WG5: Aniridia Models for Collaborative Research, Daniel Aberdam (FR). WG6: Patient-driven Research, Christina Grupcheva (BG).



ANIRIDIA EUROPE RESEARCH AWARD 2020 Cell-based Aniridia Research



Aniridia Europe and PTC Therapeutics signed an agreement, in which PTC will fund Aniridia Europe Research Award 2020 in the category "Cell-based Aniridia Research" with 7,000 €, that will be granted in the frame of the 5th European Aniridia Conference in the UK in 2020.

The application requirements, as well as the jury, will be published soon.

Thank you very much to PTC for its support to aniridia research.



ANIRIDIA EUROPE

LOOKING OUT FOR THE ANIRIDIA COMMUNITY



PROFESSIONAL'S CORNER

Statement from Dominique Bremond-Gignac. Head of OPHTARA Rare Eye Diseases Center



Ophtara Centre was accredited by French Health Ministry and accredited ERN-Eye (Europe) as a consortium. OPHTARA Center in Paris is dedicated to non-syndromic and syndromic Aniridia diseases. the unique multidisciplinary rare eye diseases center dedicated to care of aniridia patients. We follow more than 300 aniridia patients. We propose consultations and surgery with Hospital specialized Departments a high quality innovative ocular imaging platform and genetics NGS sequencing.

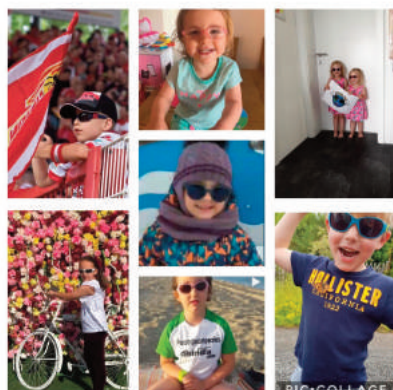
Dominique Bremond-Gignac MD, PhD, is a French physician, clinician, surgeon, researcher, Professor of Ophthalmology and Head of University Orthoptic Department in Paris, France, Head of Ophthalmology Department and OPHTARA Aniridia and Rare Eye Diseases center of reference at Hospital Necker-Enfants malades in Paris since 2015. She works on ocular diseases at INSERM UMRS1138 Research Unit, University Paris Sorbonne.

As Head of CLAIROP Research Clinical Center accredited by Europe EVI-CT we have clinical research with innovative therapies as gene therapy or new molecules. As President of Scientific Committee of Aniridia Europe and Geniris in France, we work all together to improve information, care, research and quality of life of patients.

ANIRIDIA DAY 2019



The purpose of the day was to show the world that, despite aniridia, great achievements can be reached: at school, at work and in the daily life. The Organising Committee worked very hard to raise awareness of our condition and to get a high participation in the campaign which was carried out on digital networks. We made it!



More than 350 people participated on Aniridia Day Facebook Group and near 200 on Instagram, where their pictures and videos with their stories were posted.

With the hashtag #CanDoWithAniridia. we sent a positive, full of energy message about living with Aniridia and WARG, linked with blindness and low vision.

Thank you to everyone who was involved in this Aniridia Day Campaign and we hope to grow next year.



PARTICIPATION IN EURORDIS GENERAL ASSEMBLY AND IN THE EUROPEAN PAEDIATRIC OPHTHALMOLOGY EPOS MEETING

Aniridia Europe was represented by Oana Simerea at Eurordis General Assembly AG, where a new board was elected, and at its Membership meeting in Bucharest, May 2019.

Oana attended to some relevant workshops, like the European Patients Advocacy Groups and European Reference Networks.

"This is a unique opportunity to learn from others experience, contact other associations and potential funders", said Oana.

Henrihs Kigelis, from Latvia, represented Aniridia Europe at EPOS conference in Rigga, Latvia. where we shared an information booth with Albinism Europe. Information about aniridia and Aniridia Europe was disseminated among doctors.



NORDIC ANIRIDIA CONFERENCE

The Nordic aniridia conference was organized under the COST action framework , that supported the researchers participation and the organisation of the event.

Over 100 persons from Norway, Sweden, Denmark and Finland got together for the Nordic Aniridia conference in Hurdal, near Oslo, Norway, 7th-9th of June

Expert doctors made presentations about aniridia research, inovation treatments in genetics and stem cells, as well as the newest treatments on aniridia related keratopathy and glaucoma.

After every presentation there was some time for questions and debate. This was really nice and made the conference go fluently forward, since the lecturers could make their whole presentation and patients had enough time for questions. The conference language was kind of mixture of Swedish, Danish and Norwegian.



We also had a panel discussion with all the lecturers and some workshops on social and other aniridia related health issues.

The smaller children and teenagers had their own programme

On Saturday evening we also had the honor to have Daniel Rosén, singing in every present Scandinavian language.



Aniridia Day 2019 celebration.

Many events were organised by the Interregional Support Center for Patients with Aniridia "Iris" in Russia, among them:

A flash mob, with "Creation", with the participation of Russian celebrities wearing sunglasses. More than 200 videos were recorded to support people with Aniridia.

Some families from Krasnodar region gathered to celebrate Aniridia day together and two Russian paralympic champions recorded a video about her achievements with aniridia.



Aniridia Center in Moscow, Russia

In February 2019, Aniridia Centre, in the National Medical Research Center for the Children's Health (Federal State autonomous institution of the Russian Federation Ministry of Health) in Moscow, started to work.



In this center, patients with aniridia (children and adults) will receive not only a full ophthalmologic examination, but also a professional check up of the endocrine and nervous systems, including sleep disorders. For patients with WAGR syndrome nephrologist, gynecologist and urologist examinations are also provided.

ANIRIDIA BELGIUM

The Aniridia Support Group in Belgium celebrated Aniridia Day

To celebrate Aniridia Day, an informal family meeting was held on Saturday June 22nd in the Parc du Cinquantenaire in Brussels. 3 french and 3 dutch speaking patients and their families were present. In total we were 14 persons.

After a lunch together in the park, it was play time for the children in the playground. Given the nice weather, it was an enjoyable getting together.





Annual Meeting of Families with Aniridia in Spain



The Spanish Association of Aniridia (AEA) organized the Annual Families Meeting with Aniridia on April 6th. This meeting has been always an opportunity for patients and family members to share their own experiences and concerns.

Young people with aniridia attended to activities such as "planting Iris" or talks on sexuality, social skills and genetics with different professionals. In the other hand, adults attended to a mindfulness workshop.

Both adults and young people enjoyed a cooking workshop by chef Eduardo Sánchez, who wore low vision glasses for cooking. Finally, the blind paratriathlete José Luis García, alias Jota, gave a motivational talk.

<https://aniridia.aniridia.es/xix-encuentro-anual-de-ninos-jovenes-y-familias-con-aniridia>

Aniridia Research was Awarded by FEDER Foundation.

As part of the celebration of the Aniridia Day, The Spanish Association of Aniridia awarded Marta Cortón, researcher at Jiménez Díaz Foundation, with FEDER Foundation research funds, for the project: "Genetic characterization of Aniridia in Spain through genomic and experimental studies".

The Spanish Federation of Rare Diseases (FEDER) recognized 8 research projects promoted by the associative network.

Original article: <https://enfermedades-raras.org/index.php/slider-home/12474-feder-reconoce-8-proyectos-de-investigaci%C3%B3n-en-enfermedades-raras-impulsados-por-el-tejido-asociativo>.





First French Guidelines on Congenital Aniridia in France and Benelux, third in Europe

The first French guidelines on congenital aniridia is published since 10th April 2019. It was initiated by OPHTARA-Necker Hospital in Paris, the French center of reference for aniridia and GENIRIS, the French patients association. An English version is planned. This is the third guideline on congenital aniridia in Europe after the Spanish one on 2009 and the Italian one on 2014.

Read more here : https://www.has-sante.fr/portail/jcms/c_2963463/en/aniridie

Geniris and French aniridia experts support Cost Action "Aniridia Net" project.

"Aniridia Net" is an European network to address an unmet medical, scientific and societal challenge on aniridia. This project is led by Pr Lagali from Sweden. GÊNIRIS and two of its Scientific Committee Members support this project as :

1) French representatives on "Aniridia Net" steering committee :

Pr Brémond-Gignac (French Aniridia Center of Reference OPHTARA-Necker, Pr Aberdam (Inserm), GÊNIRIS and Isabelle Petit (Inserm).

2) Working Groups : WG1 Clinical Guidelines leader, Pr Brémond-Gignac- WG5 Aniridia models leader, Pr Aberdam - GÊNIRIS participate in several working groups.

Aniridia in Czech Republic

There is no Aniridia Association in Czech Republic yet, but Michaela Kubizňáková is making some steps toward this goal and raising awareness on aniridia in this country.



- She has created a website for Czech community of aniridia to find the basic information
- She has been contacted by a few people who are connected to aniridia, mainly parents of children with aniridia. "All were happy for having some source of information as well as somebody with aniridia to keep in touch with and ask questions".
- She has translated, together with her eye doctor, Aniridia Europe Brochure, into the Czech language, that can be unloaded on the website.

The website link is: <https://www.aniridie.cz> Facebook page Aniridie Česko:

<https://www.facebook.com/aniridie/>



ANIRIDIA UK & IRELAND



Aniridia Network Conference: 1 June 2019 in Birmingham, UK

The UK Aniridia Network Conference took place on 1 June 2019 in Birmingham.

Speakers included experts in child development, education and life with aniridia, and the charity held their Annual General Meeting. Videos of all presentations will soon be available on the Aniridia Network Youtube channel.

Delegates also enjoyed the opportunity to engage with other patients, families and professionals, through workshops, a patient discussion group and an evening dinner. There were further social meetups on Aniridia Day, 21 June, in London and Dublin.



Preparations are now being made for next year's European Aniridia Conference, which will take place on 14-16 August 2020 in London. Offers to volunteer and fundraise to assist with the event will be very welcome. For details see <https://aniridia.org.uk/conference/>

New Aniridia Research Projects

Research into the growth of eyes with aniridia has begun in the UK, jointly funded by Aniridia Network and Fight For Sight, along with the generous donations of two families of children with aniridia.

Dr Mariya Moosajee, leading the project at Moorfields Eye Hospital, said: "The study will advance our understanding of PAX6 in early eye development. Plus it will provide proof-of-concept of amlexanox as a treatment for nonsense-mediated aniridia."

Dr Moosajee has also secured funding to study the medical history of 150-200 aniridia patients. This will help identify reliable outcome measures for clinical trials, and which patients may be more suitable for certain treatment.

She says: "I feel like there is a spotlight on aniridia-related research and I am very grateful for all the funding and support I have received to expand our knowledge boundaries."

Details of both projects are on the Aniridia Network blog - <https://aniridia.org.uk/blog/>





Eleanor Burke, representative of Aniridia Network in Ireland, met Lord Mayor of Dublin, to celebrate Aniridia Day.

"The Lord Mayor of Dublin, Mr Paul McAuliffe, honoured my quest to raise the profile of Aniridia by meeting with me at the Mansion House on 21 June 2019. Equipped with Aniridia merchandise we chatted about aniridia and Paul's own involvement in rare diseases". Eleanor said.

ANIRIDIA LITHUANIA



The National Aniridia Association "Aniridija LT" was established in Lithuania on June 12th 2019.

Association members and doctors meeting on the Aniridia Day



On the occasion of the International Aniridia Day in June 21, representatives of the association "Aniridija LT" held a meeting with doctors at the Kaunas Clinic. At the warm meeting, areas and opportunities for mutual cooperation were discussed. Head of Eye Clinic and other great doctors already joined our association as honorary members. "We have planned to work together for better conditions for Aniridia patients living in Lithuania and at the same time for better working conditions for those who care for them", Irma Byle stated.

ANIRIDIA UKRAINE



The organization "Aniridia WAGR-Sindrome Ukraine" started to work in May 2018.

On May 11th 2019, the first Ukrainian conference with ophthalmologists from all Ukrainian regions was organised / <http://aniridia.org.ua/node/57> /. There, the doctors discussed the questions concerning Aniridia.

Moreover, in April, the sponsor ZEISS agreed to help all the members of our organization with free lenses.

On Aniridia Day the families gathered together in the towns and shared their experience / <http://aniridia.org.ua/node/60>

