ANIRIDIA EUROPE

LOOKING OUT FOR THE ANIRIDIA COMMUNITY

Aniridia Europe E-Newsletter. January 2020

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 are currently 13 National aniridia associations and one support group. Please, see https://www.aniridia.eu/in-your-country/

(The articles have been published as they arrived, first arrived, first served)

COST ACTION: ANIRIDIA-NET

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

1st COST Action Aniridia-net.eu #CA18116 meeting. September 2019. Paris

The 1st COST Action Aniridia-net.eu #CA18116 meeting was held last September 12-13th 2019 in Paris with a wide multinational participation of stakeholders. Aniridia-net.eu is a pan-european network of researchers, ophthalmologists, Aniridia Europe patients' representatives, industry and special interest groups, funded by the European Cooperation in Science and Technology (COST) program, to improve aniridia clinical management and promote innovative research for its diagnosis and treatment.

To tackle these challenges, six working groups were brought together in Paris to discuss leading new research and embark upon bottom-up multidisciplinary and collaborative networks, to learn, share and stimulate aniridia-focused clinical practice and research.

Among other important goals, this Action will focus on the harmonization of clinical management guidelines, the development of multicentre clinical studies and tissue banks, the evaluation and translation of regenerative medicine therapeutic strategies and the promotion of patient-driven research.

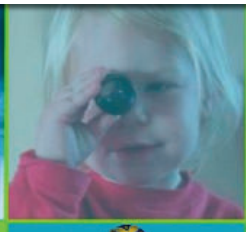
Next meeting will take place on February 27-28th,2020 in Lisbon, Protugal

#COSTProgram, #aniridiaawareness

#aniridiaresearch, #aniridia

ANIRIDIA EUROPE

LOOKING OUT FOR THE ANIRIDIA COMMUNITY



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

Second transnational meeting. 14 September 2019, Sandefjord (Norway).

In this second transnational meeting, hosted by Aniridia Norway, the partner entities met to show and evaluate the progress made in the project.

The participating organizations presented the different activities and tasks carried out so far, especially focused on the expected results in the project and with especial emphasis on the development and coordination progress of two intellectual outputs: the Virtual Training Course for teachers and early education centers, as well as the Educational Game:ICT tool, both for the inclusion of students with visual impairment in the classrooms.

The project's schedule was revised and the intermediate evaluation of the project was carried out on-site.

SAVE THE DATE:

The 5th European Abniridia Conference, organised by Aniridia Network, with the support of Aniridia Europe, will take place in London, UK, on August 14-16th 2020. Please, see more information at: https://2020.aniridiaconference.eu/

ANIRIDIA EUROPE

LOOKING OUT FOR THE ANIRIDIA COMMUNITY

ANIRIDIA RESEARCH

Aniridia study project in Russia is gradually becoming a medical technology

According to Pr. Tatyana Vasilyeva, it embraces the efforts of dozen of experts:

Ophthalmologists and geneticists have finished the work on the medical recommendations on aniridia patients' diagnostics and eye therapeutic and surgical care. This year neurologists and endocrinologists are planning to collect and analyze data on neural and endocrine status of aniridia patients and develop recommendations on treating usual for aniridic patients neurologic and homeostatic problems.

Latest publications on the Aniridia study project in Russia.

On the genotype-phenotype correlations in WAGR syndrome patients.

We assessed the involvement of the neighboring to the PAX6 and WT1 genes from deleted 11p13 chromosome regions in the patients with and without Wilms' tumor. Reliable confidence was obtained for the LMO2 gene, which is significantly more often deleted in patients with nephroblastoma. Thus, our study presents genetic evidence that the development of Wilms tumors in WAGR syndrome patients should be attributed to the deletion of WT1 and LMO2 rather than WT1 only. doi:10.1093/hmg/ddz168

On the established genotype-phenotype correlations in aniridia patients.

Based on the results of detailed clinical examination and occurrence of clinical features in a large cohort of aniridia patients (155 people), we undertook statistical analysis and revealed significant relationships between a particular type of PAX6 mutations and aniridic eye phenotype. Only patients with 3′-cis-regulatory region deletions develop a distinct and milder phenotype, without nystagmus, keratopathy, or macula hypoplasia Phenotypes of the patients with all types of point pathogenic variants in the PAX6 gene and large chromosomal rearrangements excluding 3′-cis-regulatory region deletions do not differ significantly. doi.org/10.1038/s41431-019-0494-2

On the functional reassessment of the PAX6 VUS.

Functional analysis of several PAX6 sequence variants with unclear clinical significance including deep intronic variants and synonymous substitution, allowed to reclassify them into pathogenic mutations disrupting splicing. doi:10.1038/s41431-018-0288-y

Previous publications:

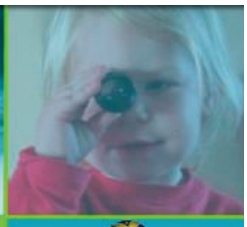
AX6 Gene Characteristic and Causative Role of PAX6 Mutations in Inherited Eye Pathologies doi:10.1134/S1022795418090156

Molecular analysis of patients with aniridia in Russian Federation broadens the spectrum of PAX6 mutations. doi:10.1111/cge.13019

Clinical and morphological manifestations of aniridia-associated keratopathy on anterior segment optical coherence tomography and in vivo confocal microscopy. doi:10.1016/j.jtos.2017.07.001

ANIRIDIA EUROPE

LOOKING OUT FOR THE ANIRIDIA COMMUNITY



ANIRIDIA RESEARCH

Genetic diagnosis will bring us one step further towards improved clinical management of aniridia.

Ophthalmologists at the Saarland University Medical Centre, including Prof. Barbara Käsmann-Kellner and Prof. Berthold Seitz, have found specific variations of the primary gene that causes aniridia (also called PAX-6), that may account for the different subtypes and severity degrees of this devastating eye disease. This novel finding will improve disease characterization and diagnosis, facilitating treatment decisions, as well as future research in patients suffering from aniridia.

Hereditary aniridia is a rare, severely impairing disease, principally caused by one (or several) mutations or alterations in the PAX-6 gene; however, patients present with a wide range of clinical manifestations, varying progression rates and with diverse prognoses. These wide variations hinder clinical management and make impracticable a general guideline for treatment. Hopefully, this new discovery linking specific mutation type to the clinical picture will enable accurate genetic information, to assist the ophthalmologist to reach a more precise picture of the progression and consequences of the disease, to enable better and more personalized treatment choices to be applied.

A group of children and adult patients diagnosed with aniridia at the Saarland University Medical Center in Homburg, Germany, have participated in this study by providing blood samples as well as the clinical examination data, that have made this discovery possible. An international team of researchers led by Dr. Neil Lagali from Linköping University in Sweden, has been involved in this investigative work, that has involved researchers from Sweden, Poland, Norway and Germany, under the support of EU COST action ANIRIDIA-NET #CA18116 (https://aniridia-net.eu).

A wide variety of PAX-6 gene mutations had already been described, but the investigators found nine new mutations not previously reported. Most interestingly, the association of the type of gene alteration with the clinical features and progression of vision loss in the disease, led to the identification of different subtypes of the disease, which is the key finding of this piece of research that may lay the foundations for a better clinical management of aniridia patients.

The full article: Lagali N, Wowra B, Fries FN, Latta L, Moslemani K, Utheim TP, Wylegala E, Seitz B, Käsmann-Kellner B. PAX6 Mutational Status Determines Aniridia-Associated Keratopathy Phenotype. Ophthalmology. 2019 Sep 28. https://www.aaojournal.org/article/S0161-6420(19)32101-3/fulltext

Acknowledgment: The work described here was in part supported by the European Union's COST Program, under COST Action CA18116, ANIRIDIA-NET (www.aniridia-net.eu) as well as by funds from the Dr. Rolf M. Schwiete Foundation in Mannheim, Germany and the Norwegian Association of Aniridia – Aniridi Norge. The work was also made possible by the cooperation of the German Aniridia Association (AWS Aniridie-Wagr e.V)

ANIRIDIA EUROPE

LOOKING OUT FOR THE ANIRIDIA COMMUNITY

ANIRIDIA FRANCE AND BENELUX

Regional meeting on aniridia

Two Regional Aniridia Families Meeting took place on 12th October 2019 in Nantes and on 30th November 2019 in Lyon. 42 patients and families could share experiences and knowledge with each other and health professionals.

Two other Regional Aniridia Families Meeting would be held in Clermont-Ferrand and Toulouse on 2020.

International meeting on aniridia

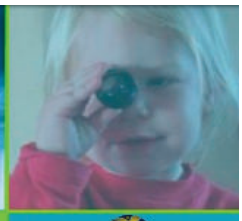
GÊNIRIS and one of its Scientific Committee Members were present at the AFI Families and Professional Meetings in August 2019 near Cincinnati in United States. Pr Aberdam has presented an update of his project «Modeling of human aniridia and gene therapy approaches».

Gêniris at WSPOS/ESCRS. Paris.

GÊNIRIS and the President of its Scientific Committee were present at WSPOS Subspeciaity Day in September 2019 at ECSRS/ Paris. Pr Brémond-Gignac was organizer of this meeting in France and chairperson of "Paediatric Ocular Surface Disease" session where 2 update about aniridia management were made.

ANIRIDIA EUROPE

LOOKING OUT FOR THE ANIRIDIA COMMUNITY



ANIRIDIA SPAIN

Aniridia meeting at the 95th Congress of the Spanish Ophtalmology Society

On September 26th, the Spanish Aniridia Association attended the 95th Congress of the Spanish Ophthalmology Society and co-organized the meeting “Aniridia, its problems and solutions”, together with Professor Jorge Alió, an International wellknown ophthalmologist.

This congress brings together annually more than 2,000 ophthalmologists and presents the latest studies and findings in the field.

This Aniridia Scientific Session was divided in:Genetics, , Glaucoma; Ocular surface and Low vision:in Aniridia.

Other awareness and fundraising activities.

The Spanish Association, together with Juan Tamariz Magicians Shcool, held a Humor and Magic show in Alcrocón, Madrid, on November 19th with more than 400 attendees

A calendar with some of the most famous cooks in Spain and children with aniridia was also developed.

ANIRIDIA NORWAY

Course on disability. Oslo, 18-20 October 2019

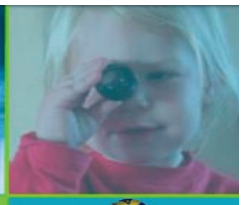
Aniridi Norge held last year a course in Oslo, to debate about the aspects of living with a disability, as well as to find out how that disability will impcat or affect one´s identity and in which degree. Albinism association was also invited, to discuss these issues and give perspective on the various challenges, techniques and knowledge towards looking at life and living with a disability.

Conclusion:There is no recipe for a good life, but the course created reflection and awareness which, again, can lead to meaningful changes.

Aniridia Norge also hosted the 2nd transnational meeting of the European educational Erasmus+ projec:School for all. Please, see the second page of this newsletter

ANIRIDIA EUROPE

LOOKING OUT FOR THE ANIRIDIA COMMUNITY



ANIRIDIA RUSSIA

The Russian Aniridia Center is now involved in Aniridia-Net

Inter-regional Support Center for Patients with Aniridia reports that:

1. The Russian application as a Near Neighbor Country / MC Observer has been approved by the COST Association. From October 2019, doctors and geneticists at the Russian Aniridia Center are involved in the Aniridia-Net COST Action!,

2. The Russian Federal Aniridia Center moved, from July 2019, to the Central Clinical Hospital of the Russian Academy of Sciences

ANIRIDIA NETWORK

European Aniridia Conference

European Aniridia Conference. 14-16 August 2020, London, UK

Tickets will soon be on sale for this event combining excellence in aniridia research, clinical practice and patient engagement.

By sharing the insights of those developing treatments and living with aniridia, the goal is to upskill each other, prevent sight loss and deal with its other effects.

Professionals from around the world such as: ophthalmologists, researchers, vision scientists, and geneticists will be the primary delegates. People who have aniridia and their relatives are also encouraged to come, especially to the later family sessions as part of the annual Aniridia Network conference. Everyone is welcome.

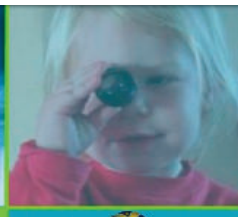
Come for clinics with top consultants at Moorfields Eye Hospital and tours of UCL Institute of Ophthalmology. Then 2 days of presentations and networking to grow understanding of how aniridia affects the eye, brain, pancreas etc.

Spend time visiting the many tourist sites in London too.

Find out more about European Aniridia Conference http://2020.aniridiaconference.eu/

ANIRIDIA EUROPE

LOOKING OUT FOR THE ANIRIDIA COMMUNITY



ANIRIDIA NETWORK

Aniridia Network held meet-ups in Edinburgh and London,

For their first ever event in Scotland, trustee Elean-Burke welcomed 11 people to an informal get-together.In the south, we held a social gathering Kensington, London. A group of 9 people, hosted by trustees James and Dave, met for drinks and dinner. Both were a lovely evenings of socialising and shared stories of life with aniridia.

The next meet-up will be in Dublin to celebrate Rare Disease Day, Saturday 29 February.

Consultation on artificial iris implants

The UK healthcare system has been creating guidance for doctors on the use of iris implants. Aniridia Network publicised this and inputted what we know about the subject. The finished recommendations will be published shortly.

Cornea disease drug research

Scientists in Paris and Tel Aviv have found two, already approved drugs that can increase PAX6 protein production in cornea cells with aniridia mutations. Now a team in Aberdeen will be testing the effectiveness of these drugs on the vision of aniridic mice. They hope it will reduce, reverse or prevent cornea disease for multiple types of mutations. More about the cornea disease drug research https://aniridia.org.uk/2020/01/06/testing-if-drugs-can-improve-cornea-regeneration/

ANIRIDIA DENMARK

Meet-up in autumn 2019

Aniridi Danmark repeated the Activity Day celebrated in 2018, however, this time the entire family with a person with Aniridia was invited the whole weekend, October 5-8th 2019.. More than 20 children gathered and spent a great weekend with, both professionals´ presentations and lots of physical and creative activities for young and old.

In 2020 Aniridi Danmark will celebrate their five year anniversary, with a big annual meeting at Fuglsangscentret in Fredericia on 6th-8th March 2020. This will again be an event for the entire family with both presentations and family activities