Aniridia Europe’s General Assembly, Paris, August 24th, 2018

On August 24th, 2018, in Paris, Aniridia Europe held its General Assembly, that examined and discussed the state of the federation and elected the new trustees for the period 2018-2020, as well as the Scientific Committee, whose members were all confirmed.

You can find:
- the Minutes of the General Assembly at: https://www.aniridia.eu/minutes-from-the-general-assembly-august-24th-2018/?preview=true
- the new Trustees at: https://www.aniridia.eu/trustees-2018-2020/
- the Scientific Committee at: https://www.aniridia.eu/scientific-committee/

Aniridia Europe is a partner in two European projects:
- Erasmus+ project Looking out for a school for all: early educational inclusion for students with low vision

The project is focused on early educational inclusion for visually impaired children. It involves 6 partners from 3 different European countries. It is led by RedTree, a Spanish organisation that has a long experience in working with associations of people with visual impairment. The other partners are: Alba, the Spanish association for albinism, Aniridia Europe, Aniridia Norge, Aniridia Italiana and SmallCodes, an Italian company specialized in developing digital environments.

The project will last two years, from September 2018 to August 2020, and will include
4 transnational meetings in Spain, Italy and Norway.

The main objective will be to develop materials and tools to ease the inclusion of children with visual impairment in early education; for this purpose it is intended to design applications adapted to their learning as well as guides/handbooks on inclusion for the training of teachers and trainers of early education.

- **Aniridia-Net: Aniridia: Networking to address an unmet medical, scientific, and societal challenge**

COST (European Cooperation in Science and Technology) has decided to fund the program Aniridia: Networking to address an unmet medical, scientific, and societal challenge, whose aims are to build an excellent pan-European collaboration within the aniridia community and to improve the clinical management of the disease and advance research in cornea, stem cells, genetics, regenerative medicine, transplantation and many other fields.

The project involves an impressive group of 69 proposers from 24 countries.

Aniridia Europe is a partner with the specific task of representing the patients and playing an active role towards these objectives.

Information at: https://www.cost.eu/actions/CA18116/#tabs|Name:overview

Aniridia Europe would like to thank our sponsor PTC Therapeutics for supporting our cause and for its generous donation, essential to reach our goals.

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**PROFESSIONAL´S CORNER**

According to Dr. Juan Álvarez de Toledo, member of Aniridia Europe Scientific Committee, “physicians and researchers should address the following challenges in the future, to improve the situation of patients with aniridia:

1. –To find a trustful non-invasive diagnostic tests during early pregnancy, to check every pregnant woman. This should also be done with all possible rare disorders.
2. - To find drugs that could stop the corneal disease and glaucoma in early childhood, that could avoid any possible malformation in the limbus and anterior chamber
3.- To explore the possibility of achieving a successful iris transplantation.
4.- To study macular hipoplasia in children and how it evolves from birth birth to adulthood.
5.- To set which are the best surgical procedures, to treat every complication, associated to this disorder.

*In order to develop this kind of studies, a large cohort of patients is needed*”
Aniridia Connection! The 9th National meeting of the Italian Aniridia association, Padua, October 12th-14th, 2018

Under the domes and the steeples of Saint Anthony’s Basilica, Aniridia Italiana held its 9th national meeting, attended by more than 120 people, among which around 60 persons with aniridia.

Specialists talked about access to healthcare, genetics, clinical management of children and adults, ophthalmological, physical and cognitive rehabilitation for children with aniridia or WAGR syndrome. A lot of time was dedicated to questions and answers.

The Youth Programme focused on intergenerational dialogue. A meeting entirely dedicated to contacts and relationships, transmission of knowledge and experiences, confrontation between children, young people and adults, families and specialists, to seize all the opportunities offered by the network and to get involved without barriers and shyness. This was done with games, such as a sort of “speed dates”: people sitting in couples formed by a younger and an older person, face to face, having 3 minutes to answer to each other tough and tricky questions.

You can also see the exclusive interview performed by Matteo Castelnuovo with Charlie Cox, the English actor that has portrayed Matt Murdock, the blind superhero in Marvel’s Daredevil TV series at: https://www.aniridia.eu/2018/11/01/aniridia-connection-the-9th-national-meeting-of-the-italian-aniridia-association-october-12th-14th-2018/

Aniridia got gold and silver in paralympic fencing!

Milan, October 28th, 2018 -

Two wonderful women, and great champions, Veronica Tartaglia and Simonetta Pizzuti, conquered the gold and silver medal in the first national competition of paralympic fencing in Milan last October. After eliminating all their opponents, they played the final and Veronica won the match 10-8. Both members of the Italian aniridia association, they are deeply involved in promoting social inclusion for visually impaired people through sport. Veronica is currently in charge of the organisation of events in our association.
Nordic aniridia conference

On 7 to 9 June 2019 a Nordic meeting for aniridia patients and experts will be held in Hurdal near Oslo, Norway. The meeting is a result of a collaboration between Aniridia associations in Sweden, Norway and Denmark as well as Linköpings University Hospital in Sweden. At the meeting, a number of experts and practitioners from several Universities and Eye hospitals in Sweden, Norway and Denmark will be present.

IN 3 days, there will be formal and informal talks, presentation and socializing. Here members from both Sweden, Norway and Denmark are invited to come together to talk and share experiences and knowledge about aniridia with people from the other countries. The conference is to help create a partnership with cooperation between these countries in the future.

Eye research

The Norwegian eye-doctor Øygun Utheim, had her PhD presentation in November last year. The PhD was about limbal stem cell failure and aniridia. The Norwegian association supported this research study and patients with aniridia participated in.

Utheim has contributed to develop methods to a more safety storage and transportation of stem cells to treat the disease, in particular, eye surface injuries. As well as doing research on aniridia with a focus on the eye surface, she showed that eye inflammation, unstable tear film and loss of glandular tissue on the eye surface are more pronounced in aniridia, than in the rest of the eyes without the disease.
Focus on young people and families with Aniridia in Denmark

In Aniridi Danmark, we are currently working on expanding our group of members. At the moment we are especially focused on attracting young people and families with the diagnose. According to this goal, we organised an Activity Day in November 2018, where the activities were adapted to this group. It was great to meet the three families who participated, two of them were new, as well as other members.

In the beginning of 2019, we have contacted DBSU (the Danish National Association of Visually Impaired Persons), in order to recruit persons with the Aniridia diagnose. Our hope is to gather a number of young people who can participate in the youth activities at the upcoming Nordic conference in Norway in June 2019.

Aniridia in the Bulgarian Ophthalmology Conference

The conference "Novelties in Ophthalmology 2018" was held in Riu Pravets, Bulgaria on November 23-25th, 2018. The main focus of this scientific professional forum was on "Cataracts". It was addressed in a modern and innovative light, in all of its many aspects: diagnosis, treatment, complications, prophylaxis and rehabilitation.

Cataracts and glaucoma are common in most patients’ eyes with aniridia, with their own features and characteristics in every individual with aniridia.

The reports that specifically discussed the aniridia were presented by Assoc. Prof. Anna Popova ("From the Symptoms of Aniridia to Gillespie’s Syndrome"), by Maya Atanasovska et al. ("Molecular Genetic Studies to Bulgarian Patients with Non-Syndromic Aniridia"). by Maya Atanasovska, a genetic scientist, under the leadership of Prof. Draga Toncheva, with a dissertation work on Aniridia.
Other reports "Genomic Diagnosis of Hereditary Eye Diseases ...." and "Dysmorphic Syndromes in Ophthalmology", were presented at the Children Ophtalmology session, this one available for free at: http://nt-cmb.musofia.bg:8080/jspui/bitstream/10861/1431/3/popova-1-30.pdf?fbclid=IwAR1BtKw3Pg
SrUBdV-FoK9YM6SgFtBJtG_b3XVoNqO147S_YLRUULHB6e

Associate Prof. Anna Popova’s article "Glaucoma and WAGR Syndrome", was published in the "Bulgarian Forum Glaucoma" magazine, No2, 2018.

Technical aids grants 2018

Last year 2018, the Spanish Association of Aniridia, launched a call for technical aids for its full members. These grants were endowed with 4,000 €, most of these funds were raised with the sale of the A.E.A calendars. "With + flavor and stars 2019". 10 families were beneficiaries. The official presentation of the calendars took place at NH Eurobuilding in Madrid on December 3rd, with the collaboration of the Óscar Velasco, a well known kook, with two Michelin stars, from SantCeloni Restaurant, who cooked wearing low vision glasses, to simulate how people with aniridia can see and the the difficulties they have to face in their daily life.

94th Congress of the Spanish Ophthalmology Society SEO.

The AEA attended this SEO Congress where we had the privilege of learning from the experts and professionals who gathered there from September 26 to 29th 2018 in Granada.

It was a unique opportunity for the Spanish Association of Aniridia AEA, to disseminate information about aniridia and associated pathologies, the association, its role and tasks carried out for people with low vision, among professionals, students, teachers and experts who met at this annual event, as well as to continue learning and sharing information about ophthalmological pathologies. In addition, we also had some meetings with the industry labs, to obtain tears and information about new products related to dry eye.
Meeting of the the Scientific Committee of the Interregional Support Center for patients with aniridia "Iris"

The Russian Scientific Committee met in August 2018 and decided to write Clinical guidelines for congenital aniridia for the Russian Federation, as well as to start a general Register of patients with congenital aniridia in Russia. For more information:

Presentation of a PhD degree on Aniridia

Defense of the dissertation for PhD degree on biological science by Tatyana Vasilyeva, a member of the Scientific Committee of Aniridia Europe and member of the SC of the Interregional support Center for patients with aniridia "Iris", on the topic "Genetic and Phenotypic Features of Congenital Aniridia in the Russian Federation."


ANIRIDIA DAY, JUNE 21st

Aniridia Community celebrated Aniridia Day on June 21st in 2018 for the second year. Many activities were organised in different countries to raise awareness on aniridia.

It is a time to coordinate activities and improve understanding of the rare genetic eye condition. In 2018, we improved understanding of aniridia using international webinars and by describing aniridia sight on social media. More information at https://aniridiaday.org/

You are invited to participate in next Aniridia Day 2019. Join us to celebrate our day.
Abseil raises €1200 for aniridia

James in the UK was sponsored to abseil 80 metres down the Orbit tower. He split the money raised between Moorfields Eye Hospital and Aniridia Network.

James remarks that there are brilliant teams investigating possible treatments to preserve or improve the sight of people with aniridia at Moorfields Eye Hospital.

The event was organised by Moorfields Eye Charity. The weather was beautifully sunny. The Orbit is the equivalent of 26 storeys and is in Olympic Park in London. James said: "I nervously leaned back, then stepped off the edge. It tense at first and then excellent fun. It was an awesome view and so quiet when hanging in mid-air. Such a big thrill!"

A massive thank you to everyone who so generously sponsored James. In total, he raised £641 for research into aniridia at Moorfields Eye Hospital and £328 for Aniridia Network activities.

Aniridia Network Conference: 1 June 2019 in Birmingham, UK

The next big event in the UK will be a chance to hear from, and ask questions to people interested in and affected by aniridia.

There will be presentations about aniridia and related subjects from doctors and patients as well as the charity’s annual general meeting. Lunch and a creche will be provided.

Everyone is welcome to attend, especially Aniridia Network members from UK and Ireland.

Make a note in your diary now. More details and tickets booking will be available soon at aniridia.org.uk/conference.

To get an idea of what it will be like find out about the Aniridia Network Conference 2018 including videos of the presentations.
National and International meeting on aniridia in France

The 4th European Conference on Aniridia, Iris and Corneal Developmental Anomalies took place on 25th and 26th August 2018 in Paris, France. Organized by the French patients organisation Géniris, the Center of Rare Diseases in Ophthalmology OPHTARA, in Paris and Aniridia Europe, it brought together 130 patients and 75 health professionals from all over the world, who networked and discussed about care and research on genetics, systemic aspects, associated syndromes and cornea. Other associated events on 24th August 2018 took place too in Paris: international families and youth meetings, general assembly and scientific committee meeting of Aniridia Europe.

On Saturday 16th March 2019, there will be the national aniridia families meeting in Paris, France. Organized by Géniris, this is the opportunity for people to exchange knowledges and experiences all together and with clinicians and researchers. More information here: www.geniris.fr

GENIRIS and AFM TELETHON support researches on aniridia

In 2018 and 2019, GENIRIS and AFM TELETHON decide to support with 85 000 euros Pr Aberdam project on aniridia «MODELING OF HUMAN ANIRIDIA AND GENE THERAPY APPROACHES ». His group has designed an unique cellular model that recapitulates in vitro ARK. By the use of Crisp/Cas9 technology, a nonsense mutation found in aniridia patients has been inserted into one allele of the PAX6 gene into human limbal stem cells. It reproduces the PAX6 haploinsufficiency characteristics of anidiria. This cellular model allowed identifying altered function in these mutated cells. Remarkably, these defects were all corrected by treatment of the mutant cells with recombinant PAX6 protein. Moreover, we identified 2 small compounds by high throughput screening that rescue defective cell migration and proliferation. These drugs are already used for severe human depression and thus could be repurposed for ARK treatment provided that they will be validated in vivo.