



ANIRIDIA EUROPE

**Aniridia Europe Meeting
Universitätsklinikum des Saarlandes
Klinik für Augenheilkunde
Homburg/Saar (Germany)
1st and 2nd of June 2013**

Report

This event was jointly organised by the Eye Hospital for Ophthalmology of the University Hospital Saarland, Homburg /Saar, the German Aniridia Association, AWS Aniridie-WAGR Selbsthilfe e.V. and The European Federation of Aniridia Associations, Aniridia Europe, with the aim at bringing together people with aniridia from Germany and other European countries and at empowering them by providing them relevant with information on their disease and about the different situation in the different European countries, as well as showing them the potential of working together under a strong umbrella group. This is why several parallel meetings and conference were organised over those two days: a German meeting, an Aniridia Europe representatives meeting and a Scientific Symposium and the possibility of an ophthalmologic examination with low vision advice in the Eye Clinic.

June 1st – Morning: Parallel meetings: German meeting and European meeting

European meeting:

Aniridia Europe President, Rosa Sánchez de Vega, briefly introduced the meeting then gave the floor to the participants that presented the situation of their own country and of their national association, when existing.

Around the table there were people from Spain, Germany, Italy, United Kingdom, Switzerland, Belgium, Russia, Bulgaria, Denmark, Sweden, Norway.

Dr. Rasha Sameer, a medical doctor from Egypt, asked to participate in the meeting; this was welcomed by the group. Then the scheduled presentations were exposed.

Rosa Sánchez de Vega talked about *Aniridia Europe in 2013*, presenting the history, objectives, activities and future perspectives of the federation, in the frame of EU Rare Diseases policy and ongoing projects such as patients' registries.

The group discussed the possibility of building a European registry for aniridia patients and it was said that at present there is a need to wait for the results of some important developments on this issue at EU level; in the meantime, it was decided to build a contact list of patients in order to be ready when there will be a concrete opportunity of building a registry.

Barbara Poli, AE Secretary, presented *Protocols and Guidelines for Congenital aniridia: How and*

why, explaining the characteristics of these documents and relating on the two existing experiences: the Spanish *Protocol on the management and care of aniridia patients* and the Italian *Guidelines for the Management of congenital aniridia*.

Neven Milivojevic explained the policy chosen by Aniridia Europe of slowly but surely building a long term financial and organizational sustainability. He strongly advised participants to build an association in their countries because our previous experiences prove that a formally recognised organisation can have a stronger voice towards medical professionals and healthcare institutions. Some raised the problem that their doctors discourage them about forming an association, because they think that it more appropriate to rely on the existing organisations, such as the national blind unions. AE Board made clear that Aniridia Europe and aniridia national associations are neither in opposition nor in competition with the blind unions or any other existing association. Instead, our objective is to focus on the specific needs of aniridia patients connected to the specificity of the disease (knowledge, management and research on the disease), while we cooperate with other associations and institutions on all the matters we have in common. It was suggested by Rosa Sánchez de Vega to include a chapter on this kind of advocacy in the document that is being developed on *How to start a national association*.

June 1st – Afternoon



The participants moved to the Auditorium to attend the Scientific Symposium on aniridia together with the German patients. About 65 people attended the conference.

Prof. B. Seitz, director of the Clinic and Denice Toews-Hennig welcomed the participants and thanked Aniridia Europe for doing its best to combine both the European and the German meetings. Denice also thanked Dr Seitz and Dr. Kaesmann for their involvement and for hosting the event. She introduced herself not only as representative of the German Association and Aniridia Europe, but also as mother of a 4 year old child with aniridia. She still has many concerns and many questions on the management of the disease. The lack of information available and the difficulty to find competent doctors on this disease, motivated her to organise this event, specially when she met Prof Kaesmann. “I consider this conference as a starting point for a better future, for a common work, also at European level. I hope that from now on, doctors will share their research findings to offer the best possible treatment for people with aniridia” Mrs. Toews-Hennig added.

Rosa Sánchez de Vega presented Aniridia Europe to the German participants, that was nicely translated into German by Dr. Kaesmann.

Gabriele Feigl, BBS, from the Institute for blind and visually impaired in Nürnberg, talked about the *Fundamentals and Opportunities of Sight Training*. “Early sight training should always be performed after a diagnosis of a visual problem”, Mrs Feigl stated. She reported the way they offer support to families at their home, primary school, kindergarden or wherever is necessary. The goal is to

develop self management and self confidence in children, contributing to their happiness and autonomy. She shows how other senses can support the lack of vision.

Prof. Dr. Barbara Käsmann-Kellner, Paediatric Ophthalmology and Low Vision. Department of Ophthalmology. University of Saarland. Homburg, talked about *Clinic of Aniridia, simple and complicated histories and their non-operative treatment - Genetics of aniridia and recent studies on aniridia*. "In Aniridia all the structures of the eye are affected. According to statistics, there are about 800-1000 people with aniridia in Germany. Aniridia has no cure and glasses do not improve sight, aniridia is a complex disorder". Dr. Kaesmann explained.

Dr. Kaesmann reports on some of the complicated associated conditions that have a severe impact on these patients' sight, apart from the lack of iris, like, small optic nerve, cornea clouding and high pressure, that causes irreversible damage on the retina. She does not recommend artificial iris implants, since they may cause glaucoma, scars and fibrosis, but other surgeries like eye lid ptosis and nystagmus could be performed with more or less success, since there is no direct intervention in the eye.

Prof. Dr. B. Seitz, *The surgical treatment options in congenital aniridia: glaucoma surgery; cataract surgery (removal of the opaque lens of the eye), surgical possibilities in corneal scarring and corneal opacities*.

Dr. Seitz presented the different problems in the cornea with aniridia and the different surgical procedures. "Cornea is very delicate and get easily irritated, causing harm in these patients", this is why he does not recommend contact lenses.

"Preserving sight is our mission, then only surgery should be performed as late as possible and only when really necessary", he highlighted.

Dr. Seitz and Dr. Kaesmann-K. ppt presentations will be available at: www.anirdie-wagr.de



June 2nd – Morning

The morning was divided in three sessions.

- **First session:** there were some short presentations:

1) Neven Milivojevic explained that RD associations very rarely meet the criteria for getting funds; at the moment only the Spanish and the Norwegian associations receive some funding from the State.

Aniridia Europe received some funding from Eurordis in order to organise meetings to build the federation; this is a very important support, but it cannot ensure ordinary functioning.

AE has a sponsorship agreement with a private company, HumanOptics; the agreement makes clear that we do not recommend in any way to use their products as AE does not have the capability to offer direct medical advice or guidance.

This amount is not very large and we are keeping it to make a reserve to guarantee the financial sustainability of the 2014 Conference.

At the moment, this is the only money we have and it would be very good if full and affiliated members could help AE in finding philanthropy sponsors.

As for regular small tax-deductible gifts: the laws for tax deduction are those of the country of the giver, so the possibility to deduct a donation has to be investigated in every specific case.

2) Katie Atkinson talked on behalf of Jenny from UK and Kery from Spain, who propose to create a European group on WAGR. There is already an International WAGR 11p13 group in USA and every year they organise a meeting, but it is always in America and this is too far for people from Europe. The idea is to have a meeting in Europe too.

Jenny and Kery wish to create a group, to promote research, to cooperate with other organisations such as AE, to have a protocol, and to create a scientific panel. Participants are required to transmit this information on their national networks through mails, websites, social networks and so on.

3) Elena Tsoneva e Ognian Petkov explained that the Bulgarian association has applied for a project directed to build empowerment for families affected by aniridia. The partnership ensured by the Norwegian association will give the project more possibilities to be accepted.

4) Barbara Poli gave some information about the state of the art of the 2nd European Conference on Aniridia, scheduled in Italy in September 19-20th 2014: 2 days in Italy, including 4 sessions : 3 for professionals, 1 open to patients.

Doctors from European countries, Japan and USA will be invited as speakers

Main Topics are: Conservative serum, prevention, new possible approach, children.

- **Second Session: Editorial Staff meeting.** Some members of the Editorial Staff (Espen Vangsnes, Katie Atkinson, Barbara Poli and Denice Toews-Hennig) left to have a meeting on the website, that is at the moment undergoing revision.

- **Third session:** The rest of participants stayed with the President and the Vice-President

A paper with FAQs Frequent Asked Questions was delivered, answered by some members of AE Scientific Committee, that will be progressively updated by contributions of doctors or speakers at Oslo Conference 2012.

The participants shared their concerns and looked for solutions at European level, expressed their expectations and requirements to Aniridia Europe:

1. **Awareness.** All agreed that Awareness campaign on aniridia is very necessary, since very few people have heard about this disease and even doctors have not had the opportunity to see a patient with aniridia. Some (Belgium) proposed to disseminate information at universities and hospitals so that national doctors are more knowledgeable on this disorder. Dr. Rasha (from Egypt) suggests developing a brochure to be delivered in hospitals, universities, schools, etc. It is also important to invite doctors to our conferences and promote workshops on aniridia in professional conferences. It is important to have basic information flyers or ppt format to present it at universities, hospitals in every country.
2. **Treatments and coverage.** In some European countries autologous serum is not accessible or available (Italy and Denmark). It was suggested to get information on this, to find out

where it is available and where it is not, as well as getting information on the availability of other treatments for aniridia in each country. Neven proposed to address this issue to AE Scientific Committee, and also to find out if the European Medicines Agency EMA has approved it.

3. **Aniridia Europe Forum** to share experiences and support each other. Ognian (Bulgaria) explained the added value of a European forum, compared to facebook, as it is a closed group and keep the record of questions and answers.
4. **Making a European list of people with aniridia**, as a first step for a European Patients' Registry with Aniridia, trying to collect and store private data, but protecting private data confidentiality, with previous consent form to be shared or sharing them with a code and ensure the only access to data by a person responsible for this. The larger number of patients with aniridia we collect, the more interest from researchers we will get.
5. **The need for a common European Medical Card**. Many participants proposed the idea of developing a card with medical information on aniridia. Many have the concern that their children or themselves, when unconscious at an accident, may be mis-diagnosed as dead, because of his/her big pupil. In some Nordic countries are working on it. Some proposals like a card with a bar code were raised, as well as simple stickers designed for rare diseases, for emergency, for necklacse or wallets. They recall that the Red Cross, for ex., has also such things, like small lockers.
6. **Education info**. Some parents expressed the need of a common basic booklet for schools, that could be translated into different EU languages..
7. **Advocacy for patients' rights**. Aniridia Europe is expected to become a lobbyist (Belgium) at European and National level about the problems/barriers people with aniridia, to have treatment expenses reimbursed and facilitate the social integration, like low taxi payment for people who cannot drive, for ex. visual impaired.... AE is also perceived as a platform to get involved, develop or participate in European projects.
8. **Check if "Aniridia" is listed** as a rare disease at National level. Neven suggested the participants to check if "aniridia" is in the National list of rare diseases, because many of social services are based on the list.
9. **European Guidelines on Aniridia**. All the participants stood out the importance of a European Protocol or Guidelines on aniridia, that should cover medical and social information, medical literature, projects, etc.
10. **Positive opinion about Aniridia Europe** and this kind of European meetings. Most of the participants were happy to be able to meet other people with aniridia from other European countries and appreciated to feel part of a group, to break up isolation, particularly in countries where there is no association yet

There was a brainstorming on how everyone could contribute and support Aniridia Europe, to go forward and become stronger

Belgium (Joeri & Colle) and Denmark (Susan) are trying to develop a National association in

their country and check if “aniridia” is listed.

As it was mentioned before, some are willing to contact universities and give presentations on aniridia for students.

Switzerland (Urs) will try to contact people with aniridia in his country (although he is aware that the challenge of speaking 4 different languages in his country and guesses that depending on their language they could already feel supported by the already established associations in Germany, France and Italy. He will try to put an advertisement in these websites.

- Rosa promised to make AE ppt presentations available for the community to be adapted to National situation, different scope or targeted audience and to be translated into the correspondent language. **Didier** offered his knowledge on projects development for support and advice.

Russia (Gala) insists in involving as many doctors as possible in conferences on aniridia and offers her support to get sponsors from Egypt or Russia, she will contact doctors from different specialities in Russia and suggests organising a meeting in Egypt.

Bulgaria will work on seminars, brochures, on Aniridia Europe forum, with a previous registration. **Ognian** invites everyone to register. He asks the others for support for marketing the forum at National level. He will prepare a pack of information and send it to the group, to be widely disseminated and spread out.

Norway (Soelvi). The Norwegian association seems to be, currently, the most sustainable, financially speaking, that is supported by the government.

The final session gave the participants the opportunity to wrap up and to express their willingness to work together on aniridia research projects at National and European level. Everybody expressed their congratulations and thanks to Denice Toews-Hennig for the organisation of the meeting and for her warm hospitality.

