



Aniridia Network UK Conference 2014

Session summaries

Introduction to Aniridia Network UK

By: Katie Atkinson - Aniridia Network UK Chairperson and Trustee

Introduction to Aniridia Network UK (ANUK) will set the scene for the day. It explains the basics of ANUK - why it exists, what its aims are and what it does to meet them. You'll be introduced to the people currently helping to make it happen and how your involvement as a volunteer, donor, fundraiser, or medical professional is critical to keeping it all going.

Keynote: Our Journey with Aniridia

By: Mark & Harry Westwood

There was no history of aniridia in the Westwood Family, but when Harry arrived their journey with aniridia had begun. This session is a personal account of learning to deal with Harry's aniridia and its implications from a family perspective.

Everyone's journey is different and their challenges of moving the family twice in the UK and growing up through the education system with several changes of school are discussed. By working to understand the condition, with support from a wide range of organisations, and by providing opportunities for Harry, they have grown as a family, had experiences that they might not otherwise have had and have learned a lot about themselves. They consider themselves to be fortunate, in that aniridia has given them as much as it has taken away.

Introducing the Cells for Sight Team and Our Work to Help Patients with Aniridia

By: Dr Alex Shortt PhD FRCOphth

Dr Alex Shortt will introduce the work of the Cells for Sight Transplantation and Research Programme. This is a group of [s](#)cientists and [c](#)linicians who work together at Moorfields Eye Hospital and the UCL Institute of Ophthalmology. The aim of the group is to develop new treatment for patients with blinding corneal diseases. The group is led by Professor Julie Daniels PhD and the clinical work of the group is led by Dr Shortt. In this session Dr Shortt will outline the challenges that arise in treating cataracts and corneal disease in patients with aniridia. He will also discuss recent advances that offer new hope to patients.

Developing a Human Model for Aniridia Related Keratopathy

By: Dr Victoria Tovell PhD

Understanding how a disease progresses is the first step towards identifying potential treatment strategies. In order to do this we ideally need a 'model' of the disease we are looking at. Generally animal models are used to look at disease progression to gain a better understanding of what is happening in vivo. Indeed scientists have developed a mouse model of aniridia by generating mice that are deficient of the PAX6 gene. These mice display characteristics similar to human aniridia and can therefore be used to investigate the role of the PAX6 gene in the progression of aniridia. However, differences between species mean that we cannot rely solely on data from PAX6 deficient mice. Dr Victoria Tovell therefore is looking into developing a human model of aniridia.

In their lab they can grow the different types of corneal cells from human donor corneas and use these cells along with tissue-engineering techniques to build a human cornea. Dr Tovell's project aims to turn the healthy corneal cells that they grow into aniridic cells by deleting the PAX6 gene and then using these cells to make a tissue engineered human model of aniridia. This will enable them to study how human corneal cells behave when PAX6 is deleted and might provide some insight into potential treatment ideas. In this session, Dr Tovell will talk about her progress with this project so far and the next steps she will be taking.

Stem Cell Treatment of Aniridia Related Corneal Disease

By: Dr Alex Shortt PhD FRCOphth

There are many different types of stem cells each with their own benefits and drawbacks. Stem cells taken from donor corneas or from the donors own mouth have been used to treat severe corneal disease in patients with aniridia. In this talk Dr Shortt will give an overview of what a stem cell is and what has been achieved so far in the treatment of aniridia.

Emerging Molecular Therapies for Aniridia

By: Prof Colin E. Willoughby

Professor Colin E. Willoughby's session will be a three part talk addressing the ocular features and common clinical problems in aniridia, the genetics of aniridia and new molecular treatments for aniridia. The first part of this talk will introduce some of the common clinical problems focusing mainly on aniridia-associated keratopathy, as this will be relevant to the evaluation of molecular therapies later in the talk. Prof Willoughby will also discuss iris implants, which is of significant interest to the aniridia community.

Part two of the talk will introduce the genetics of aniridia and, in particular, the underlying molecular mechanisms, as this will form the basis to understand the developments in molecular therapies. The final part of the talk will deal with the use of a drug called ataluren to treat the ocular pathology in aniridia. This was recently published by Prof Cheryl Gregory-Evans from the University of British Columbia, Vancouver in the Journal of Clinical Investigation. This is of great interest to patients and families with aniridia. The term START therapy was coined in this paper to describe a topical formulation of ataluren. The data and findings from this paper will be presented; and the potential role of ataluren or stop-codon read-through strategies in aniridia and genetic disease will be generally discussed. This is a very technical topic and Prof Willoughby hopes to make it palatable. The session will finish by introducing some early work being performed in Liverpool to manipulate PAX6 levels as a mutation independent therapeutic strategy.

Living with Aniridia – A Generational Perspective

By: Mary Cox

Mary Cox, her father, and her two daughters were born with aniridia. This year, 2014, is the centenary of her father's birth. In the past 100 years understanding and treatment of the condition has improved greatly. But people still have to deal with the consequences that aniridia has on their daily life. The focus of Mary's talk will be the impact that aniridia has had on her whole family, lessons they have had to absorb, changes they have had to accommodate, and some practical tips that hopefully will help others.

Guide Dogs - Sighted Guiding and General Mobility

By: Rick Allbrook

Rick Allbrook from Guide Dogs for the Blind (GDBA) will discuss during this session GDBA's role as a mobility charity and how they support people with sight loss to get out and about independently.

The services that GDBA offers are expanding and whilst guide dogs will always be their core service and account for 70-80% of what they do, GDBA now offers mobility to children, young people, and adults. They also have a strong campaigning profile around Dog Attacks, Streets Ahead and Talking Buses. Recently they developed a new service called MyGuide. This is a Nationally Accredited Programme of Training Sighted Guiding so that more people with sight loss can be offered assistance in the correct way. It is very important that all visually impaired people know themselves how to be guided correctly, so that more people with sight loss can set their own standards on how they want to be guided when offered assistance by members of the public.

Annual General Meeting

By: Sarah Hargraves – Aniridia Network UK Secretary and Trustee

The Annual General Meeting (AGM) is a formal meeting where the ANUK trustees will present their reports and update members on the previous year. These reports include the financial records of ANUK, fundraising activities and other activities designed to raise general awareness of ANUK. It is an opportunity to ask the trustees questions and put forward suggestions for future activities and fundraising.

Fundraising

By: Liz Atkinson - Trustee

An informative and lively session aimed at getting members involved in fundraising for Aniridia Network UK. Attendees will gain a better understanding of ANUK's need to reach our target of £4,500 by March 2015. ANUK appreciates your input and ideas on ways to raise funds, enthusiasm and help.

Question & Answer Session

By: Panel of experts and individuals affected by aniridia

Do you have a question that you always remember on the way home from your appointment? Is there a non-medical question that you've always wanted to ask, but there never seems to be the right time to ask it? The Q&A session is your opportunity to ask that burning question.

The panel will consist of an ANUK trustee, members of our medical panel, a parent and other ANUK advisers that are happy to answer your questions. The answer to your question will likely be helpful to someone else and may help them ask a question of their own.