



Aniridia Network UK

Welcome to Meeting 2016

- Eat lunch and visit the stalls
- Post on social media about today

Use **#ANUKconf**

Twitter: @AniridiaNetUK

- Sessions begin here at 1.15pm

Please silence mobile devices



Housekeeping

- Fire exits
- Toilets
- Break at 3pm
- Folder contents and slides are online
- Quiz

Thanks

Morning activity volunteers

- Wheels For Well Being
- Lois Turner and Change Foundation
- Guiders and drivers

Share

Post on social media about today

Use **#ANUKconf**

Twitter: @AniridiaNetUK



Aniridia Network UK

Keynote: PAX6 - The Other Effects

By Miss R Jane Leitch

Consultant Ophthalmologist

Web: aniridia.org.uk

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Email: info@aniridia.org.uk

Patron: Veronica van Heyningen, CBE

Registered charity, HMRC reference: XT26830

Aniridia



Miss R Jane Leitch
Consultant Ophthalmologist

PAX 6

- Pax 6 Gene
- Pax 6 protein

Across the species

- Zebra Fish to Humans

Where is PAX 6 Protein

- Role in eye and brain development
- outside the brain -pancreas

What does the PAX 6 protein do

- cornea
- lens
- iris
- retina

Cornea

- lack of stem cells
- problems with corneal surface in later life
- problems with corneal repair if damaged or trauma

Lens

- early onset cataract

Absence of iris

- sensitivity to light
- abnormalities in focus
- glaucoma

Retina

- macular hypoplasia - poor vision

Why nystagmus

- associated with poor vision and macular underdevelopment

Pax 6 and the brain

- role in brain development telencephalon-thalamus pituitary pineal cerebellum and spinal cord
- double dose of PAX6 abnormality structural problems with brain development

Pax 6 and pancreas

- Role in insulin metabolism
- issues with weight control and diabetes

Point /small mutations

- Aniridia in families
- different mutations can give different problems
- Different families different problems

Large mutations

- WAGR complex and Wilms Tumour association

New diagnosis of aniridia

- important to identify if WAGR or point mutation
- genetic studies

Growing up with poor vision

- school
- family life
- transition to adult hood and independence
- maintaining and preserving vision

Management

- correction of refractive error
- photochromic/dark glasses
- cataract - treatable
- glaucoma - managed
- corneal surface issues - managed

Thank You



Aniridia Network UK

Mobility as a Profession and Person with aniridia

By Deborah Reynolds

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Starting university with aniridia

By Harry Westwood

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Aniridia Network UK

Aniridia Network UK Annual General Meeting 2015

By ANUK Trustees

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Agenda

- [ANUK AGM minutes 2015](#) to be agreed
- Matters arising.
- Reports & Accounts.
 - Questions to officials
- Appointment of charity trustees
- Any other business



Aniridia Network UK

Annual Report 2015/16

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Events

- There were 2 excellent meet-ups for patients and parents. The first in Newcastle was hosted by Kelly and served as a warm up for the later conference. The second in London after Sight Village brought several old and new members together, overseen by Katie and James. Both led to new friendships and fruitful conversations.
- The Aniridia Europe Youth Camp held in summer 2015 was extensively publicised. One adult and 8 young people (4 with aniridia) went from the UK for 10 days of activities in and around Venice.



Conference 2015

- Hugely successful. Over 70 adults and children attended the very accessible venue. The event included an exhibition, food, creche and a raffle
- 89% said they were satisfied.
- Sessions:
 - Consultant Ophthalmologist Mr Michael Clarke
 - Aniridia: Check your knowledge by James and Jenny
 - The cornea: in health and disease by: Prof Martin Collinson
 - Developing a human model for aniridia related keratopathy by: Dr Victoria Tovell
 - Befriending with Lyn, Mary and Olwen
 - One Vision - European Youth Exchange by Tim Broome and Lois
 - VI tennis by: Wendy Glasper -
 - EHC Plans by: Clive Matthews
 - Services for families, children & young people by Wendy Sainsbury, Blind Children UK
 - WAGR families convention

Befriending and Enquiries

- In the past year we have created 30 buddy relationships
- Personal stories from two buddy relationships presented at 2015 conference
- 52 of our members have befriended someone since the service began
- Enquiries team has answered around 10 enquiries by email/phone this year as well as contributing to many discussions in Aniridic Family and other Facebook groups

Education Advice

- Three requests for pupil passports which are also available on our website
- Development of passport for children aged 0-3
- Development of guidance for practitioners working with children 0-3
- Advice and information given to two students from Leeds University doing a presentation on teaching children with aniridia
- Attended day conference at NatSip on Education and Health Care Plans (EHCP)
- Liaised with Living Paintings for 2016 Conference
- Liaising with RNIB to source information on counselling support for children and young people

Patron's Progress

- Helped with introductions for organizing the September 2015 ANUK Annual Conference in Newcastle, but was, sadly, unable to attend the meeting. Will be at May 2016 meeting!
- Met with some ANUK members at informal London meet-up in November
- Gave several talks during the year including to ophthalmologists in training and in celebration of Marcelle Jay, one of the earliest pioneers for eye genetics at Moorfields, as well as recent research talks in Berlin and Paris. Always talk about different aspects of PAX6, the major aniridia gene. Have accepted invitation to Aniridia Europe meeting in August in Germany.



With Marcelle Jay,
pioneer of eye genetics

Patron's Progress

- Worked with colleagues Sanjay Sisodiya and Mahinda Yogarajah on a scientific paper, which will be published soon, about how having aniridia and *PAX6* mutations alters some aspects of brain structure that can be observed by MRI (Magnetic Resonance Imaging).
- The *PAX6* mutation database continues to be maintained from Edinburgh: it documents over 900 independent aniridia mutations
http://lsdb.hgu.mrc.ac.uk/home.php?select_db=PAX6
- Colleagues in Edinburgh, in parallel with scientists in France, have identified a new gene, *ITPR1*, mutated in a rare form of aniridia, Gillespie Syndrome, where the unusual iris phenotype is associated with poor motor control and mild learning difficulties.
- Although retired, still working in different scientific areas, for example on a macular malformation of the retina with a new PhD student at the Institute of Ophthalmology (Moorfields).
- Attend some scientific meetings: met young scientist working on how brain adapts in people with visual impairment. Learning about possibilities of gene editing.
- Sit on several committees, including one promoting diversity (gender and racial balance, disability equality) in science. Ideas extend to all areas of life and work.

Fundraising: Highlights

- A big donation of £1 500 received in October from the Fairy Queen Dream Team cyclists who took part in the Prudential London 100 cycling event again this year for what is probably final time. Many thanks to Elly Chapple, family and friends for facilitating this.
- Thanks to Ceryl Roberts for the £100 donation from proceeds of a nursery auction .
- Raffle and T-shirt sales at our 2015 Conference raised over £300. Thanks to Kelly for all her hard work on this
- Number of one off donations from supporters such as £60 from Michaela Halsey received in December. Thanks to her and others who have contributed.
- Donation of £200 worth of audio recording equipment from Katie's colleagues at Dassault Systemes UK.



Fundraising: Regular Income

We have had fewer big events this year so income is lower. Big events can provide a real boost to fundraising but the number can vary year to year so regular income is also important.

- Regular monthly giving from supporters continues and is a vital income stream for us.
- Give as You Live continues to give us a small but steady amount of income from on-line shopping.
- Newly launched PayPal Giving Fund brings together donations made via PayPal and EBay for Charity. Positive start with £300 donation (thank you mystery donor!) and could be a growth area for us.

Liaison/Public Relations



- We had one of our blog posts about a member's experience with SEN republished as an article in RNIB Insight magazine.
- We advertised the Same But Different project. As a result two members with WAGR, Faith and Mari were featured in beautifully written articles with delightful photographs. These were extremely popular on social media.
<http://www.samebutdifferentcic.org.uk/new-blog/?tag=aniridia>
- The #irisselvie campaign from Rare Disease Day 2015 was featured on the JustGiving Blog as ["4 Brilliant Social Media Campaigns You've Never Heard Of"](#).
- Tracy took 400 ANUK leaflets to an ECLO seminar and handed them out, thereby distributing them around the country.
- Andrew Edwards represented ANUK at the Genetic Alliance UK Annual General Meeting.
- Katie represented ANUK at the NCVO Trustees Conference and the Genetic Disorders Leadership Symposium.

Aniridia Europe

- Aniridia Youth Summer Exchange
- Preparations for the 3rd European Aniridia Conference 27th-28th August 2016, Duisburg, Germany
- Participating in EURODIS project to develop a European Reference Network for rare eye diseases



Membership Statistics

Category	2012/13	2013/14	2014/15	2015/16	Year on year increase			% of estimated affected people in the UK
					2013-14	2014-15	2015-16	
familial aniridia	83	114	134	143	37%	18%	7%	
sporadic aniridia	73	94	116	135	22%	23%	16%	
aniridia but origin unknown	87	87	84	88	0%	-3%	5%	
Total with aniridia	243	295	330	366	21%	12%	10%	26%
Traumatic aniridia	0	2	2	2	200%	0%	0%	
WAGR	22	31	34	36	41%	10%	6%	54%
None/unknown	187	237	265	316	27%	12%	19%	
Total	452	565	635	720	25%	12%	13%	

Note: Increases in familial and sporadic cases may be due to clarification of type from unknown

Membership Statistics

- It has been estimated that there are 1344 people with aniridia in the UK.
- Only a fraction of people with aniridia in the UK have contacted ANUK. Of course many more may have accessed our information without us knowing.
- We have gained new members this year but the trend of a slower rate of increase each year continues.
- New members are usually families with new-born cases of aniridia.
- We may be reaching a plateau, at least based on our current recruitment methods.
- James continues to manage membership. His diligent research and persistence has led to several new affiliations.
- We need a dedicated Membership Officer to recruit and retain members.
- James and Keith have made some improvement to our database, including:
 - Ability to show members on a map
 - Adding a category and data for ECLOs
 - Facilities to manage volunteers
 - Upgrading to latest software

Website and Email

Website

- Usage of our website has increased:
- Visitors: 8217 (+16%)
- Views 15,637 (+12%)
- Blog posts: 20 (-17)

Email broadcast

- 10 campaigns, mainly relating to events
- 366 subscribers
- Between 30-50% open rate
- Mainly read using mobile devices, predominantly iOS

Social Media

- The growth of our social media has continued but slower, in-line with membership.
- We trialled livestreaming sessions at Conference 2015 with some success.
- Monitored and promoted mentions of “aniridia” online.
- Actively engaged in Aniridic Family Facebook group, including posting on a member’s behalf to protect their privacy.
- James currently manages our social media but we really need an outgoing, enthusiastic Social Media Officer to build on our excellent base.



Site	Followers											
	2012/13	2013/14	2014/15	2015/16	Year on year increase							
					2013-14		2014-15		2015-16			
Facebook	383	531	607	705	148	39%	76	14%	98	16%		
Twitter	282	416	517	597	134	48%	101	24%	80	15%		

Charity registration

- At our last AGM members voted to close existing organisation and create a new one to allow us to become a Charitable Incorporated Organisation (CIO), and register with the Charity Commission.
- The trustees are working towards this goal.

Restructure

- We reorganised how the now smaller trustee board works.
- Each trustee has an area of responsibility within which they carry out the work and/or recruit and manage teams of volunteers to do so.

Management Roles

- James
 - Chief Executive
 - Head of Communications and Membership
- Katie
 - Head of Services and Information
- Ben
 - Head of Finance and Funding
- Martin
 - Head of Volunteering and Development

Financials

2014/15

Accounts 2015/16

Income	
Conference Tickets	£2,494.75
Conference Stands	£182.00
Fundraising	£3,354.60
Other Income	£571.11
Total Income	£6,602.46
Expenditures	
Conference Costs	£435.00
Conference Venue	£1,606.90
Administrative	£276.64
Professional Services	£99.00
Total Expenditures	£2,417.54
Net income	£4,184.92

Notes

- Conference income is high as it includes a transfer from PayPal of ticket sales for 2014, 2015 and 2016 events
- Fundraising income has declined
- Up to £2,000 is earmarked for rebates Summer Camp participants pending European grant administration

Volunteers and trustees

2015-16

Volunteers 2015/16

Thanks to all our volunteers for their work this year:

- Mary Cox
- Jenny Langley
- Tracy Wilson
- Lyn Buller
- Heather Phagan
- Keith Spink
- Liz Atkinson
- Kelly Graham

Trustees 2015/16

Thanks to trustees for their work over the year:

April-September:

- Chair: Katie Atkinson
- Treasurer: Ben Rendel
- Secretary: Sarah Hargraves
- Trustees
 - James Buller
 - Jenny Langley
 - Liz Atkinson
 - Chris Moore
 - Mary Cox
 - Craig Anyon

October-March:

- Chair: Katie Atkinson
- Treasurer: Ben Rendle
- James Buller
- Martin Fleming

Questions to trustees?

Elections

Elections for Trustees

Nominations

- Katie Atkinson
- Ben Rendle
- James Buller
- Martin Fleming

- Others?

Any other business



Aniridia Network UK

Volunteering

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Volunteer roles

- Trustees
 - Chair, Treasurer, Secretary and Trustees
- Conference & Events
- Fundraising
- Membership
- Social media
- Volunteer coordinator
- Communications:
 - Content, Social Media, PR, AV. IT, Membership/Data
- Liaison:
 - Aniridia Europe, AFI, IWSA, VI organisations
- ... various odd jobs

Make a pledge

In the next 12 months I will help ANUK financially by:

- Donating a raffle prize
- Setting up a regular donation
- Holding a fundraising event
- Doing a sponsored feat
- Including ANUK in my will

Make a pledge

In the next 12 months I will do this for ANUK:

- Write a blog post
- Take on a role
- (Help) Organise an event
- Become a buddy
- Up-skill in X so I can do Y

Make pledge

In the next 12 months I will help ANUK by doing something using my knowledge of:

- Communications & PR
- Design
- Events
- Finance
- Research
- Policy
- Fundraising
- Education
- Benefits and rights
- Retail
- Governance
- IT
- Membership
- Administration
- Customer service
- Disability/VI
- Hospitals/Medienne
- Law

Quiz

- Top prize is £25 M&S voucher
- Assistants are available
- £2.50 to enter at the Partylite stall

Stalls

- Stem cell researchers
- Ophthalmology consultation
- Partylite
- Arts Coaching Training
- Blind Children UK
- Nystagmus Network
- Action For Blind People – Online Today
- Aniridia Network UK
- International WAGR Syndrome Association

Refreshments

- tea and coffee
- victoria jam sponge
- chocolate peanut brownie
- coffee & walnut cake
- fresh fruit salad

Sessions restart at 3:45

- Blind Children UK + Nystagmus Network
- Life, campaigning, politics & aniridia + Living Paintings demonstration

Post on social media about today

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Aniridia Network UK

**Life, campaigning, politics &
aniridia**

By Emily Brothers

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Aniridia Network UK

Living Paintings demonstration

By Maria Storesund

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Aniridia Network UK

Blind Children UK

By Jenny Holder

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PRESENTING BLIND CHILDREN UK SERVICES

Jenny Holder

Specialist Family Support Officer

Part of the Guide Dogs family

OUR KEY SERVICES



- FAMILY SUPPORT
- EDUCATION
- ACCESS TECHNOLOGY
- CUSTOMISE BOOKS
- HABILITATION
- ACTIVITIES

WHO DO BLIND CHILDREN UK SUPPORT



- Blind Children UK supports children and young people between the age range 0-25 and their parent/carer
- We offer support and advise to parents from the point of diagnoses, right through to the age of 25

- We support the parent from the point of diagnosis in helping to prepare the child/young person through transition thus enabling them to become independent.

SPECIALIST FAMILY SUPPORT SERVICES



WHO ARE WE

- Specialist Family Support Officers are keyworkers. We are trained, qualified and have experience in this specialist field.
- We use a person centred approach to identify the tasks to be addressed in the areas relating to visual impairment, health, welfare and social needs

- Emotional support
- Early diagnosis, e.g explaining terminology, attending hospital visits with families should the need arise; once diagnoses is made we send information on specific eye condition
- Linking with other families

- Advise and support parents on professional meetings they may need to attend such as Team around the Child/Common Assessment Framework/Child in Need
- Advise on certificate and registration
- Housing support
- Advice on any aspect relating to Family Support



- Referral to CWDT or liaise with the team on families behalf
- Referral to specific eye condition groups
- Benefits support
- Referrals to other team within Blind Children UK/Guide Dogs services



Over the years we have supported many parents of children/young people with Aniridia right from the point of diagnoses whom I am sure have found our services to be invaluable.

WHY FAMILY SUPPORT



- We offer a listening ear
- We have a clear understanding
- We are sensitive
- Offer emotional support
- Key working
- Build a relationship right from the start by being open and honest
- Being there in times of need
- Empowerment
- Positive outcomes

ACCESS TECHNOLOGY

Our Access Technology Team works directly with young people and their families to give advice on choosing the appropriate technology. This allows the child/young person the support they need to gain skills and greater independence



Blind Children UK help children and young people access a wealth of useful technology to include: computer equipment and software; braille devices; multi sensory toys and more

Part of the [Guide Dogs family](#)



- Services are available up to the age of 25 across the whole of the UK, with any degree of visual impairment.
- Advice can be given on equipment and software for both home and school
- Arrange demos with supplier and may possibly offer funding



- We ask for 10% towards the cost of the equipment
- The primary reason has to be loss of vision and families will need a professional to support their application

Criteria and all other details are on the website, however contact must be made at the first instance via the Access technology team as they carry out an eligibility check

Any queries contact: 08007811444 or email services@blindchildrenuk.org

EDUCATION SPECIALIST

The logo for Blind Children UK features a stylized blue hand with fingers in green, purple, and orange, positioned to the left of the text 'blind children UK' which is stacked in three lines.

Specialist Education Support Service is an advisory phone service which aims to empower parents through independent, impartial advice, guidance and information on the Special Education Needs (SEN) system and statutory education entitlement.

- We work with Parents and Young People (YP) to understand the SEN process
- provide impartial information and advice to make informed choices
- advise of statutory entitlement
-

- help incorporate parental and YP's views wishes and aspirations into the EHCP process
- liaise with School, LA and other agencies etc. to achieve the best for the Child/young person with Vision Impairment.



- Information on specialist education provision - schools, curricular access etc.
- Clarifying complex terminology
- Completing forms
- Signposting to other appropriate organisations - Action for Blind People, IPSEA etc.

ACTIVITIES



The Blind Children UK Recreational Events team organise a programme of larger family events throughout the UK. These family events take place at important family times of the year like Easter, Summer and Christmas parties and include audio described pantomime visits, exclusive use of sensory playgrounds, zoo visits with tactile experiences and many more.

- Working closely with the family support team, we also focus our two family weekends to be specialised for families whose child/ren has had a recent diagnosis of a vision impairment.

- Working with Guide Dogs, our Family Guide Dog Experience days at the GD training schools, give families the opportunity to learn about what it could be like for their child to own a guide dog in the future.

- Our events give children, young people and their families the chance to take a break and meet others in similar situations. Activities have a “whole family” ethos to include sighted siblings and offer an opportunity to share experiences in a relaxed environment.



- Our events also offer access to our key services through the presence of some of our children's service team including family support, education and habilitation staff at events.





- Our events also offer access to our key services through the presence of some of our children's service team including family support, education and habilitation staff at events.



- Events are advertised on the Blind Children UK website www.blindchildrenuk.org and social media pages.

HABILITATION SPECIALIST



Habilitation is a process of helping children and young people with a vision impairment. Children/young people with vision impairment often need to be taught the basic skills and concepts which their sighted peers develop naturally.



These are life changing skills which are taught by qualified Habilitation Specialist, who work alongside local authorities and organisations to make this possible.

Areas of support include:

- Handling money
- Preparing food
- Interacting with others
- Work with babies to develop early movement
- Develop overall independent living skills

CUSTOMISE BOOKS

The CustomEyes service produces large print books for children and young people with visual impairment or dyslexia. We support children and young people across England, Northern Ireland, Scotland and Wales. Each book can be customised in the font size, spacing, page colour according to the child/young person's need



- Our books open up a whole world of imagination and learning, by enabling visually impaired children and young people to read the same books as their sighted friends and classmates.
- Books available for you include picture books, fiction, non-fiction, Oxford Reading Tree, GCSE and A Level English texts and textbooks, plus revision guides for Key Stages 2, 3 and 4.



- We have over 4,000 large print books with new titles every month which includes fiction, non fiction and educational textbooks
- Books can be purchased by children/parents for the cover price of the original print copies.

- Our books are tailor made to suit each child or young person in the best:
 - § font
 - § font size
 - § spacing
 - § paper colour

- § binding (for example spiral binding so books lie flat)
- We supply both school libraries and local authority libraries with standard large print for their visually impaired members and can also customise large print books to suit.

IN CONCLUSION



- All of our key services are assessed on it's own merit and services are offered on individual need and circumstance.



Aniridia Network UK

Nystagmus Network

By Daniel Lewi

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Questions & Answers

Panel

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Thank you

- Trinity Centre
- Tinies
- Chefs
- Stallholders
 - Commercial
 - Charities
 - Medical
- ANUK Trustees

Special Thank You

- Volunteers
 - Guiders and volunteers
 - Speakers
 - Organising team
Mary Cox & James Buller

Say what you think

- Post comments and photos on social media. #ANUKconf
- Feedback by email or the survey sent by email

Social Evening

- All are welcome
- Hosted by *Martin and Susan*
- The 'Whispering Moon' Wetherspoon pub
- 10 minutes walk, opposite station.

- Please leave promptly

Guides

- Ask for and offer help getting to the pub and station

**Hope to see you at a future
event!!**

Organised by you?