Wales Council of the Blind

Perspectif

Supplement no. 2

Support for specific eye conditions

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Support for specific eye conditions.

As John Sanders pointed out in his article in Roundup no. 29, *Owning Up*, it helps to be better informed about your sight condition for a number of reasons: getting the right monitoring and treatment; knowing you are not alone and that there are other people who can share information and experiences; and being able to explain things to employers, friends, family and others.

There are organisations out there that offer support, helping you to connect to others with the same condition, or provide information and advice. This edition of Perspectif gathers together a great many of these and gives a few articles from organisations to give a picture of the kind of support available. They may not relate to your circumstances exactly, but they help to illustrate what can be done.

Alström Syndrome - Peas in a pod!

Alström Syndrome is an ultra-rare disease affecting one in a million. Twins Katie and Hannah are even rarer!

At 4 years old adorable twins Katie and Hannah were diagnosed with one of the most rare and complex genetic conditions in the world, Alström Syndrome.

Hannah was very ill for the first year of her life. Parents Ian and Julie were devastated to learn that their little baby Hannah was experiencing heart failure. The family were told Hannah had a slim chance of survival and she may need a heart transplant in the future. But with dedicated care from Julie and Ian and the staff at the hospital, Hannah's health slowly began to improve until she could finally go home.

Both babies had poor vision and an extreme sensitivity to light. After seeing many consultants, the family realised the severity of Katie and Hannah's sight loss and in 2010 at the age of 4 they were both finally diagnosed with Alström Syndrome.

Hannah and Katie's Mum, Julie, says "Alström Syndrome UK (ASUK) coordinate specialist multi-disciplinary clinics at Birmingham Children's Hospital, where we receive specialist advice and treatments to enable the girls to lead as normal a life as possible. We get the chance to see all the clinical specialists in one place and meet other parents with children with AS. The condition is so rare that you are extremely unlikely to meet another parent that understands what the syndrome is like, which is why it is so helpful and supportive to get together. Katie and Hannah are now 11 years old and living life to the full. The girls have a fantastic bond, supporting each other through everything. We embrace every day and every challenge with the determination to fight for our precious girls"

Support for families
ASUK provides support for the whole family and organises family events; weekends away, day trips to themes parks and for teens the unforgettable driving experience! As we learn more about the complexities of the condition, ASUK raises awareness amongst clinicians and academics and brings together professionals and families to share their knowledge and experience.

What is Alström Syndrome?
Alström Syndrome is an ultra-rare genetic condition which can cause progressive blindness, loss of hearing and can lead to heart and kidney failure, type 2 diabetes, liver dysfunction and associated problems. The symptoms arise at different stages making diagnosis very difficult. It is an individual condition and not everyone will get all the symptoms, even amongst sibling's symptoms can vary widely.

ASUK endeavour to raise awareness, conduct pioneering research and enable better treatments and monitoring through the AS multi-disciplinary NHS clinics. During these clinics patients are monitored regularly and receive the most up to date treatments and advice to help manage the condition.

This is our story
Families and clinicians developed an animation to raise awareness, aid diagnosis and offer an insight into the condition as well as highlighting what ASUK do. It's at www.alstrom.org.uk/awareness-animation-is-launched/ We welcome any feedback and hope you will share this. Early diagnosis can make a big difference to children and families.

Behçet's Syndrome Society

Behçet's syndrome (or Behçet's disease) is named after a Turkish professor of dermatology. The cause of the condition is not known. A major disturbance of the immune system has been demonstrated in laboratory tests, but this is not thought to be the underlying cause. Some authorities have suggested an infective cause, but this has never been convincingly confirmed. It is known that there is a strong genetic predisposition.

Most common features of Behçet's are mouth ulceration, genital ulceration, eye involvement and arthritis and arthralgia. Up to 70% of patients with Behçet's disease will get eye involvement, and current estimates from around the world indicate that severe visual impairment occurs in 25% of involved eyes. Accordingly, the detection and treatment of eye involvement is vital.

The main ocular involvement is where the inflammatory process spreads inside the eye, causing uveitis. Other possible complications of the inflammation or its treatment include cataract (clouding of the lens of the eye), glaucoma (a rise in the pressure inside the eye which, if not controlled, leads to irreversible changes in the optic nerve and blindness), and the formation of new abnormal blood vessels in the retina and iris (which have a propensity to bleed and thus cloud the sight). Late changes include detachment of the retina, low pressure in the eye and eventual shrinkage of the eye, but by this time the eye has usually lost all useful vision.

The Behçet's Syndrome Society was formed in 1983 and exists to provide support and information to people with the condition and their families, together with those who are seeking diagnosis.

The Society provides a helpline, both telephone and email, where members and non-members can make contact with us. helpline@behcetsdisease.org.uk or 0345 130 7329.

We produce four newsletters each year, which include patient stories, fundraising, event and research news etc. We can provide one-off personal grants to members. We support and encourage peer support groups around the UK.

The Society holds an Annual Conference for patients and their families around the UK.

In 2016, we held the first Behçet's Family day in the Midlands and this year we held a family weekend in Cumbria and a family day in Northern Ireland.

In conjunction with various specialist doctors, we have produced 17 Factsheets on Behçet's, which are available on-line and on request in the post. We also have other leaflets and posters to help others learn more about Behçet's and to help raise much needed awareness of this rare disease.

We support, facilitate and fund research into Behçet's and have a comprehensive website www.behcets.org.uk

Birdshot Uveitis Society - My Journey with Birdshot

Birdshot chorioretinopathy, usually called birdshot uveitis, is a rare form of chronic posterior uveitis. It affects both eyes, is progressive, usually painless, and potentially blinding if not treated. It is thought to be an autoimmune disease: the body's immune system attacks the eye tissues, causing inflammation. The name 'birdshot' comes from the scattered white spots - small scars from inflammation - seen on the retina. The spots resemble the pattern of birdshot pellets fired from a shotgun at a target. Birdshot can be hard to diagnose, as the spots may not be clearly visible at first.

It is officially a rare disease. No-one knows how many people in the UK have birdshot: maybe around 600.

Birdshot is usually treated with medication taken by mouth (corticosteroids and immunosuppressant medicines) because eye drops do not reach inflammation at the back of the eye. Treatment aims to control inflammation and put the condition into remission, but this is tricky: the inflammation can flare up and it can take years to achieve remission.

Here is the story of Birdshot Uveitis Society (BUS) member Julie who lives in North Wales.

“My journey with birdshot chorioretinopathy began in January 2015. I woke up and could see a small circle and floaters in my right eye. My optician’s practice referred me to my general practitioner. I was seen there as an emergency. My GP dilated my eyes, and she wanted me to be checked at a hospital eye clinic immediately.

“Things moved very quickly. Within hours, I was having multiple blood tests, a chest X-ray, and my eyes were examined by several doctors. I was very frightened and worried greatly about what lay ahead.

“My work and personal life were disrupted by week-after-week of eye clinic appointments. The hospital was 30 miles away and, with eyes dilated at each appointment, I had to rely on friends to drive me there. My treatment during that time - different types of eye drops - did not improve my eyes.
“Eventually, I was referred to a tertiary eye centre for further investigation. After a fluorescein angiogram, I overheard a discussion about my condition and heard the word ‘birdshot’ for the first time. I went online for information and found the BUS site.

“I then spent the next four weeks in limbo: upset and worried till my next appointment, when my consultant confirmed the diagnosis. He told me that each birdshot patient is unique. It would be trial and error to find the correct medications and dosages for me.

Since then, my treatment journey has been difficult: juggling various medicines and their side-effects - prednisolone, mycophenolate, ciclosporin – with very little improvement. Ciclosporin in particular made me physically unwell.

“I have now been fortunate to be prescribed Humira (adalimumab), recently approved for use in England and Wales. I have been on fortnightly self-administered injections of Humira since September 2017. I have my life back both mentally and physically. My eyesight is improving with each dose and I no longer feel so ill.

“Throughout my journey I have had fantastic support from fellow ‘birdshotters’ on the BUS Facebook group who understand how I feel and what I see.

“However, my most important source of information is the BUS website, which tells you everything you need to know about birdshot, its treatments, side effects, and more. It helped me to get through the endless round of hospital appointments and tests. For anyone newly diagnosed, I would highly recommend checking it out, to know you are not alone on your birdshot journey.”

BUS is a charity and support group offering advice and information to people affected with birdshot uveitis.

BUS organises ‘Birdshot Days’ for people who live with birdshot and the professionals who treat it. If you are interested in attending any future Birdshot Days, please contact:
BUS, PO Box 64996, London SW20 2BL, birdshot.org.uk, info@birdshot.org.uk

The Childhood Eye Cancer Trust.

The Childhood Eye Cancer Trust (CHECT) is a UK charity dedicated to helping people affected by retinoblastoma (Rb), a rare form of eye cancer that affects babies and young children.

We first began as a parent support group founded in 1984 by a group of individuals whose children were receiving treatment at St Barts Hospital in London. Today CHECT has a staff team of eight helping to support anyone in the UK affected by retinoblastoma, raising awareness of the condition with both the general public and healthcare professionals to speed early diagnosis, and funding research into retinoblastoma.

So what is retinoblastoma? Retinoblastoma, or Rb as it is called for short, is an aggressive eye cancer which affects babies and young children under the age of six. It can affect one eye or both. It is a rare cancer, with around one child a week being diagnosed in the UK. Survival rates are good at around 98%, but around 70% of children will lose at least one eye in order to save their lives. Even when eyes are saved, there may be severe visual impairment. Some individuals also have an increased risk of second cancers later in life, as well as the possibility of passing the genetic form of the cancer onto any future children.

There are key signs which parents should be aware of, which can indicate the presence of retinoblastoma in their child's eyes. In most cases there will be another, less serious cause of the symptoms, but it is crucial to make sure the child's eyes are checked by an optician or GP as soon as possible if any of these signs are noted.

The two main symptoms of retinoblastoma are a squint and a white glow in a child's eye, seen in dim lighting or when a photo is taken using a flash. Healthcare professionals should carry out a red reflex test on any child showing these symptoms in order to rule out cancer. This is a simple, non-invasive test which is done using a medical torch in a darkened room to check the retina at the back of both eyes. If eye cancer is suspected, an urgent referral should be made (within two weeks), according to the NICE guidelines. Other symptoms include a change in the colour of the iris, a red, sore or swollen eye without infection and an absence of red eye in one pupil.

Children who are diagnosed with retinoblastoma are treated at one of two specialist Rb centres in the UK: the Birmingham Children's Hospital; and the Royal London Hospital.

CHECT is a small charity which plays a big role in the lives of people affected by Rb. In the last three years:

- 100% of families in the UK were offered support following a diagnosis of retinoblastoma and throughout their child's treatment.
- We provided over 2,000 hours of support to around 525 families each year.
- More than £8,500 was given in grants for 40 families facing financial difficulty because of the expenses related to their child's treatment.
- In feedback 100% of people who responded said CHECT support helped them in their experience of dealing with eye cancer.

What does CHECT do?

When a child is diagnosed, it can be a distressing and frightening time for everyone, and CHECT is there to help, providing one-to-one support at hospital clinics, over the phone and via closed Facebook groups. As a rare cancer, most people affected by Rb won't know anyone in their local area in the same position, so we arrange regional meet-up days for children and adults to share experiences and make new friends, as well as having a fun day out. We also provide information and put members in touch with other

organisations that can help: from organising respite holidays to sourcing adaptive technology.

As mentioned earlier, we are also committed to reducing the current delays in diagnosis experienced by many families, so we work hard to raise awareness of the signs and symptoms with the general public, as well as those healthcare professionals most commonly consulted by parents with concerns about their young child's eyes: GPs, health visitors and opticians. Finally, although we are a small charity, we do fund a small amount of crucial research into retinoblastoma, from understanding more about the disease, to exploring new treatments, and understanding the impact the condition has on those affected.

Angharad's story

When Angharad Price, 19, was photographed in the bath by her mum at just nine months old, her right eye was white in the photo instead of the typical red. Just one week after seeing the GP, Angharad had her right eye removed by surgeons after being diagnosed with retinoblastoma.

"I was diagnosed with Rb when I was nine months old and I had my right eye removed in what I remember to be as 'The Blue House' in St Bartholomew's Hospital, London. I am so fortunate that my mum and dad managed to spot it early – who knows where I would be without them! I remember regular visits to London with my parents and Grandma and I always felt welcomed and truly cared for."

Angharad was only technically discharged from hospital when she was 16 years old as she still had to regularly attend for check-ups every six months to a year to make sure the cancer hadn't returned. "I didn't think anything of it at the time really – I didn't know any different so I just got on with it.

"Luckily, Eluned, an orbital prosthetist in Rookwood Hospital, Cardiff who fitted my very first artificial eye still treats me now and I have built a strong friendship with her as a result. I still go to the hospital a minimum of twice a year to have it cleaned, polished and checked for fit. A couple of years ago, Eluned sent me to the most amazing place in Hemel Hempstead, to ensure that the colour of my artificial eye was a perfect match! It was incredible to see how the eyes were made and especially how they were made for use in the Harry Potter films.

“I feel that having my eye has made me more determined in life and show that nothing can hold you back. I have many close friends who accept me for who I am and have never judged me for it. However, I remember an incident when I was eight when a girl asked me to take my eye out, which I did because I did not know any different as I took pride in keeping it clean. Let’s just say her reaction was not what I was expecting!

“The only thing that it’s really affected is my field of vision and my ability to play some sports. However, I play netball and hockey and love skiing!”
In an amazing twist of fate, Angarad is currently studying medicine at St Barts Hospital in London – the same hospital where her eye was removed to save her life 19 years ago. Once Angharad has completed her five-year degree she hopes to specialise in paediatrics or obstetrics, which she says was influenced by her own experiences as a child.

For more information about retinoblastoma and CHECT visit www.chect.org.uk.

The International Glaucoma Association.

About Glaucoma
There are an estimated 64 million people with glaucoma worldwide and an estimated 600,000 people living with the condition in the UK today, half of whom are as yet undiagnosed. It affects two per cent of people over the age of 40, rising to 10 per cent over the age of 75.

It is the most common cause of *preventable* blindness, yet many people are unaware that glaucoma often has no symptoms in the early stages. But, if left untreated glaucoma can lead to serious loss of vision, with up to 40 per cent of sight being permanently lost before the effects are noticed by the individual. Once sight is lost it cannot be recovered.

The good news is that with early diagnosis, careful monitoring and regular use of treatments, the vast majority of people will retain useful sight for life.

What is Glaucoma?
Glaucoma is a group of eye conditions in which the main nerve to the eye (the optic nerve) is damaged where it leaves the eye. This nerve carries

information about what is being seen from the eye to the brain and as it becomes damaged vision is lost. This results in misty, patchy or missing vision in places.

Glaucoma eye tests

The IGA believes that everyone should have regular eye health checks, at least every two years (or every 1-2 years for over 40s). Glaucoma tests are quick, simple and convenient. A visit to your local high-street optician is all that is needed to see if you are at risk of glaucoma. There are three simple tests which include:

1. Looking at the appearance of the main nerve in the eye, called the optic nerve
2. Measuring the pressure in the eye, often referred to as the air puff test
3. Checking the field of vision. In Scotland there is a fourth test which measures the corneal thickness

Who is at greatest risk?

- Anyone over the age of 40
- Close relatives of a family member with glaucoma
- People with diabetes
- People who are very short-sighted
- People of black African, black Carribbean or South East Asian descent
- People who are long-sighted

How is it treated?

Most people with glaucoma are given eye drops to lower their eye pressure but in some cases tablets, laser treatment or surgery is appropriate. More information on glaucoma, the treatments and research which is being carried out can be found at www.glaucoma-association.com. The website has an active Forum, moderated by the IGA helpline staff. The helpline is led by an ophthalmic nurse and staff are available Monday to Friday to answer specific concerns and can be contacted on 01233 64 81 70. The IGA also has a Facebook page and Twitter account @Tweetiga.

About the IGA

The International Glaucoma Association is the charity for people with glaucoma. Established over 40 years ago, it raises awareness, promotes

research related to early diagnosis and treatment, and provides support to patients and all those who care for them.

The IGA provides services for people with glaucoma including a helpline (Sightline), free patient information through hospitals, opticians, and GP surgeries, patient support groups, awareness campaigns in June and September, and working in partnership with UK and European professional bodies to prioritise eye health, vision and glaucoma.

The Association is also a research funder and works jointly with the Royal College of Ophthalmologists, College of Opticians, Royal College of Nursing, Fight for Sight, UK and Eire Glaucoma Society to provide research funding into the treatment, management and detection of glaucoma.

It is run by a board of trustees which represents ophthalmologists, optometrists, ophthalmic nurses and people with glaucoma, and is funded entirely by voluntary contributions.

Macular Society Support Groups.

Madeline Roberts, Milford Haven Macular Society support group Secretary, tells us about her experience with the group.

Milford Haven's Macular Society support group was started on 1st October 2015 by an octogenarian after she was diagnosed with dry AMD. Having run a drama group for the local blind society, when she had sight, she knew the power of focussing on the things people with sight loss are capable of rather than the emphasis on what they cannot do.

Our regional manager for Pembrokeshire, Adele Francis, came to our very first meeting and explained about the Macular Society and its services – counselling, befriending and Skills for Seeing, to name a few – and impressed upon us how important support groups are, especially as most people over 65 being diagnosed with AMD can become isolated and depressed by the experience of sight loss.

Despite being much younger than 65 I experienced isolation especially at the fact that the way I saw was unique to me. Thinking back now, I don't

know how I found the strength to continue working while receiving treatment. I suppose denial willed me on: if I didn't admit there was something wrong then there wasn't anything wrong. But the truth is that I couldn't cope at all. I went on sick leave and was then made amicably redundant. Depression had started. What do you do when at 25 you can no longer do your dream job? When you understand but cannot accept that your diminished sight has taken away your ability to drive? When you have a general sense of worthlessness?

The best thing I ever did was contact the RNIB. The emotional counselling helped me more than words can say. I was able to give myself permission to be angry and upset about my failing sight and that it took my life (as I had known it anyway). My counsellor encouraged me to join a social group for people with sight loss run by the Council's sensory loss team, to get me engaged with life again. So I joined the V.I. ladies group in October 2010. With my growing 'I have nothing to lose' attitude I joined Pembrokeshire Disabled Bowlers club in January 2011. I doubted that I would like it as I saw bowls as an old person's sport. I was surprised, as it turned out I am good at it, having won medals representing Wales in tournaments for the vision impaired, gold in 2014, and bronze in both 2015 and 2017. In 2014 the secretary of PDBC and I became the first vision impaired bowls coaches in Wales.

All the help I have received, in one form or another, over the past nine years has made me remember that (in my old life) all I wanted to do was help people. And so back to the inaugural Milford Haven Macular Support group meeting…

Owing to my background in administration, the group leader and regional manager asked me to take on the role of group secretary. I was wary thinking that I wouldn't do a good job, but they instilled faith and encouraged me. I have been in the post for about two years and realise that the qualifications and skills I had learnt when sighted aren't for nothing.

My confidence was being rebuilt and so jumped at the chance of going through the Macular Society's Gadget Guide and Skills for Seeing training.

A Gadget Guide visits groups and demonstrates the things that are available to help people with sight loss. There are basic low-tech items

such as bump-ons and brightly coloured LED torches that cost as little as £1, high-tech pieces of equipment such as tablets and CCTV video magnifiers that cost hundreds and even thousands of pounds.

A Skills for Seeing trainer visits people on a one-to-one basis. An initial informal assessment is carried out to identify any remaining vision someone has. Then by teaching techniques that require practise, the trainee could indeed learn how to read again, to watch TV and recognise faces. However, the strategies and techniques are not suited to everyone.

There is a certain sense of job satisfaction when people come to our group and we see that it makes a noted difference to their lives. For example, I invited a gentleman in his late-nineties to come to our Macular group and speak about his time in the opera. It was hoped that it would encourage him to become a member. (The onset of age-related Macular Degeneration meant he didn't want to do the things he liked – he was possibly becoming depressed – and he was becoming isolated.)

He regaled us with tales of his touring days singing in shows such as HMS Pinafore and The Importance of Being Ernest. He treated us to a few lines from the Drinking Song in the Mikado; when he sang he seemed to become more alive. It sparked a conversation about the members' love of opera and indeed musical theatre. He now comes to every monthly meeting, he takes care of his appearance and joins in discussions.

The Milford Haven Macular support group meet on the first Thursday of the month at 2-4pm. We have a speaker or an activity each meeting. To give you an idea: we had a visit from someone at the Guide Dogs charity giving a 'show and tell' about the various tactile pavements etc. We felt the textures and types of tiles and were told the differences between them. Wiltshire Farms Foods came and gave a tasting session. We invited locally based Low-Vision assessors to explain about the service and show us the various aides that are available through the scheme, free on long-term loan. We had a paper craft session – although it was debatable as to how many Easter chickens we had actually made, but we laughed the whole way through. Then our members wanted to share tales from their lives; we had wonderful tales of a passion for cycling leading to the Tour de France, snippets from a life in Africa to one-man sailing trips around Europe.

In our refreshment break the group have a chat; views on what's been in the news, the most recent show they saw at the theatre. Someone commented on having difficulty in putting a plug into a socket as they couldn't see where the pins were meant to go. There was a wave of murmuring "Oh yes me too, it really annoys me because logically I know how to put a plug into a socket, but I can't see to do it."

Having recently done the gadget guide training I said "that's something that bump-ons can help with. A bump-on gets put onto the socket above the top pin hole, then put one on the top of the plug and line the two up". Members also share if they find a low vision aide to be helpful e.g. someone was debating whether it was worth buying a talking watch and three people all at once said "yes most definitely I wouldn't be without mine".

On occasions our members share about their forthcoming treatment, it's nice that they feel we have created a 'safe place' to voice anything they mightn't share with loved ones and the following meeting members will ask how they got on.

The camaraderie that has formed within the group led us to take part in an awareness church service in March 2017, all doing our bit to explain the condition, how it affects us and that there are things we can still do. We gave readings and chose hymns that were appropriate to the theme. It was very well received and, pardon the pun, it opened the congregation's eyes.

Milford Haven Macular Society support group's membership has doubled since it began two years ago, proving the need for it. We all look forward to going to meetings as we have become a close-knit group who share tips and the not so good things. In its way the group is alleviating isolation, building people's confidence and a little at a time proving to ourselves that not being able to see isn't the end of the world; that we are capable of more than we think we are and despite the age range of our members, sight loss brings us together.

Macular Society help line: 0300 3030 111

The Norrie Disease Society.

The Norrie Disease foundation was set up in early 2017 by three mums who's sons have Norrie disease and whom shared the same vision to promote vital new research and provide more support to families by further strengthening the Norrie network.

Norrie disease is a rare X-linked genetic condition which causes boys to be born blind or with severe sight impairment. Secondary symptoms can include progressive hearing loss, cognitive impairment, autism and delayed development. We were in contact with around 20 families in the UK with Norrie Disease and that number has now increased to over 30.
Saturday 13th May saw the culmination of a year's hard work and preparation as the Norrie Disease Foundation launched at University College London, Great Ormond Street, Institute of Child Health.

Around 70 guests gathered to celebrate the start of what promises to be an exciting journey for the Norrie Disease Foundation.

The launch was opened by Professor Maria Bitner-Glindzicz, a clinical and molecular geneticist, with a special interest in deafness, from UCL Great Ormond Street Institute of Child Health.

We are extremely excited that a research project at Great Ormond Street Institute of Child Health, University College London has begun to understand more about Norrie Disease. The project will focus on studying developmental changes in the ear and the eye in Norrie disease, so that we understand better how we might treat the problems that arise.

We have also held three community meet ups, two in London and one in Birmingham.

It was a wonderful opportunity for the community which ranged in age from 19 months to 34 years to get to know each other and share their experiences of living with Norrie disease and what has and hasn't worked for them.

Teddy, who has Norrie Disease attended the meet up with his parents Racheal McIntyre and Lee Ellerby. Rachael said this about how The Norrie Disease Foundation has supported her family.

‘Finding out I was pregnant with my first child was such an amazing feeling, feeling our son move and grow every day I couldn’t wait to meet him. Finally the day came and on 24th March 2016 we met our son Teddy and he really was beautiful. We were soon to learn that things weren’t quite right and we were about to begin a journey that we could never have anticipated. Teddy was diagnosed with Norrie Disease at a couple of months old and we felt so lost and uneducated. This feeling was soon to be lifted when we came across The Norrie Disease Foundation. This charity completely changed our lives and gave us the reassurance and confidence we were looking for, to be in touch with other families and see older boys with Teddys condition strive and succeed in life gave us answers we had been searching for. It has been a huge benefit to us to be able to ask other parents and families for their opinions and support in what would have been the hardest journey of our lives. We thoroughly enjoy meeting members of the Norrie Disease Foundation and without them we would feel so lonely and lost. Many people comment on how well we have coped with the news of Teddys condition but this is only down to the support network around us so I owe all my thanks to those as without them this would be a very lonely journey.’

We hope that the success The Norrie Disease Foundation has shown so far continues into the future.

https://norriedisease.org.uk/

Directory.

The services listed below are all on the Perspectif Portal at **www.wcb-ccd.org.uk/perspectif/**. The organisations delivering each service are given in brackets after the name. Also, the definitions of the various eye conditions are taken from the glossary at Perspectif. The sources of the definitions are given in each entry in the database.

The following defined eye conditions are located above the support services as far as possible. There are some areas of overlap.

For the contact information, please either go to the **Perspectif** site or contact **Wales Council of the Blind** on **029 20 473954**.

Macular Degeneration
Loss of central vision due to damage to the macular. Most common in older people (AMD) but can occur in younger people. There are ‘wet’ and ‘dry’ types.

Age-related macular degeneration (AMD)
An eye condition that affects a tiny part of the retina at the back of your eye, called the macula. It causes problems with central vision, but does not lead to total loss of sight.

Stargardt's disease
Causes some of the cells on the macula to stop working leading to problems with central vision, detailed vision and sometimes with colour perception.

Befriending - (Macular Society)
The Macular Society's befrienders make regular calls and enjoy chatting. You choose what you want to talk about - it could be about macular disease, your favourite hobbies or TV programmes, or just the weather! Particularly suitable for people unable to join a support group. The Society aims to find you a befriender who is local and shares your interests, experiences or type of macular disease.

AMD Alliance support - (AMD Alliance International)
The AMD Alliance offers information about age related macular degeneration such as guidance on treatment, questions to ask, living with the condition and your rights.

Macular Society Skype Groups - (Macular Society)
Skype groups give members the opportunity to talk to others affected by macular disease, without leaving home. People of all ages can take part in video or voice-only calls.

Macular Society local support groups - (Macular Society)
The Macular Society has local self-help support groups across the UK. The groups vary in size and style but all offer invaluable practical and emotional support to people with macular disease from people with macular disease.

Macular Society helpline - (Macular Society)
Being diagnosed with macular disease can leave you and your family feeling isolated and uncertain about the future. But, there is no need to face macular disease alone; the Macular Society specialist helpline team provide free information, guidance and advice to anyone affected by central vision loss whether it's you, a friend or a family member.

Aniridia

Aniridia is a rare congenital eye condition causing incomplete formation of the iris. This can cause loss of vision, usually affecting both eyes.

Befriending scheme - (Aniridia Network UK)
Befriending scheme for people with aniridia who are members of Aniridia Network UK - an opportunity to talk to someone who has experience of the condition.

Aniridia support - (Aniridia Network UK)
Aniridia Network UK provides a positive, supportive network of families, individuals and professionals in order to share ideas, experiences and give emotional support. It provides information to health professionals, patients and their families, about aniridia, the related conditions and their implications for the patient.

Albinism
Albinism is a comparatively rare, genetically inherited group of conditions which results in a reduction or complete lack of pigment (colour) in the skin, hair and eyes of people with the condition. This can result in pale skin which burns easily in the sun, virtually white hair, very severe short-sight and photophobia (a severe sensitivity to light).

Albinism Fellowship - (Albinism Fellowship)
Provides a website resource, on-line discussion forum and telephone support for people affected by albinism.

Alström syndrome
Alström Syndrome is an ultra rare recessively inherited genetic disorder, which means that both parents will carry the gene although probably be unaffected themselves. ASUK know of around 80 families in the UK who are affected, this figure could be higher due to delayed diagnosis and the rarity of the condition, many may still be undiagnosed. The condition is characterised by retinal degeration, hearing loss, obesity & insulin resistance.

Alström syndrome support group - (Alström Syndrome UK)
Provides support for those affected by Alström Syndrome, their carers and professionals working with them. This includes a forum, help with gaining access to medical assessments with doctors who have expertise in Alström Syndrome, quarterly medical screening clinics and a newsletter.

Anophthalmia
Anophthalmia is a medical term that is used to describe the absence of the globe and ocular (eye) tissue from the orbit.

Coloboma
A gap in part of the structure of the eye, caused when a baby's eyes do not develop properly in pregnancy.

Microphthalmia
Microphthalmia literally means small eye. Children may be born with one or both eyes, small and underdeveloped. Some children may be blind, but others may have some residual sight or light perception.

CHARGE Family Support Group Events - (CHARGE Family Support Group)
The priority for the Group is to create opportunities to bring families together. If you are interested in attending an event or helping to organise one in your area, contact the group.

Activities for young people - (Micro and Anopthalmic Children's Society (MACS))
MACS organises sailing trips and adventure weekends for young people with MACS conditions.

Eyeless Trust grants - (The Eyeless Trust (merged with Royal Blind Society))
The Eyeless Trust offers modest grants for families and children with anophthalmia, microphthalmia or coloboma. These are arranged through The Family Support Worker, and each year a large number of families benefit from a wide range of needs. Grants can be used for practical help, recreational activities and holidays.

MACS family helpline - (Micro and Anopthalmic Children's Society (MACS))
Telephone helpline offering advice and support to parents of children born without eyes or with under-developed eyes.

ICAN support group - (International Children's Anopthalmia and Micropthalmia Network (ICAN))
Provides a forum for parents of children with anopthalmia where issues can be discussed. Also has information about the treatment of the condition and medical research. Some activities (such as conferences) are US-based.

Grants from MACS - (Micro and Anopthalmic Children's Society (MACS))
MACS gives grants to cover the cost of activities or equipment needed by children born without eyes, or with underdeveloped eyes. You must have been a member of MACS for at least 6 months to apply.

Family support workers - (The Eyeless Trust (merged with Royal Blind Society))
Family support workers spend time with each family according to their needs and ensure that they have all the services that they need in coming to terms with the emotional, financial and practical issues associated with having a disabled child. Also help older children with independent living skills.

Behçet's Syndrome

A chronic condition caused by disturbances in the body's immune system and around 70% of patients will experience inflammation of the eye.

Behçet's Society Helpline - (Behçet's Syndrome Society)
A telephone helpline for members which is available between 9.00 am and 8.00 pm on weekdays and between 10 am and 6 pm on weekends and bank holidays (see website for full details). It is run by volunteers so answerphones may be used at certain times. Callers who leave a message are contacted as soon as possible at the Society's expense.

Best's disease

Best's disease, also known as Best's vitelliform macular dystrophy, is a hereditary form of progressive macular dystrophy first identified in 1905.

Beat the Best - (Beat the Best (Facebook group))
A Facebook group for people with Best's disease.

Birdshot uveitis
A form of uveitis where oval spots can be seen on the retina.

Birdshot Uveitis Society - (Birdshot Uveitis Society (BUS))
BUS supports patients with Birdshot, particularly when first diagnosed and provides up-to-date information about the condition and treatment options to help empower patients and ensure they have access to the most appropriate treatment possible.

Charles Bonnet syndrome
A condition where people who have lost their sight experience hallucinations.

Esme's Umbrella Help and Information Service - (Esme's Umbrella)
Telephone helpline for people affected by Charles Bonnet syndrome (visual hallucinations).

Macular Society buddies - (Macular Society)
The Macular Society can put you in touch with a Treatment Buddy who has already had treatment, for information and reassurance. If you are experiencing visual hallucinations (Charles Bonnet Syndrome) due to sight loss there is a buddy service where you can talk to someone who has also experienced visual hallucinations for information and reassurance.

Coat's Disease
A progressive condition of the retinal capillaries which occurs in children and young adults, usually males.

Coats disease community - (Coats Disease Community)
An online community where people can discuss issues related to this condition.

Deafblindness
A person is deafblind if they have a combined sight and hearing impairment that causes difficulties with communication, access to information and mobility.

Deafblind UK Information and advice line - (Deafblind UK)
Our free Information & Advice Line offers support and guidance to deafblind people, their carers, family and friends, as well as professionals working with people who have a loss of both hearing and sight. It also provides a service to Deafblind UK's members, who each receive a phonecall three times a year for a general chat and to remind them of the support available through the service. The Information & Advice Line can also connect you to the relevant team or staff member at Deafblind UK.

Peer Support & Social Groups - (Deafblind Cymru)
Deafblind Cymru's peer support & social groups provide those with both sight and hearing impairments plus their carers an opportunity to meet people and make friends. At regular meetings, all members are encouraged to get involved in the decision-making process to determine what activities the group should do, often including social outings and arranging meetings to enjoy guest speakers on subjects of interest.

CIB Dual Sensory Support Group - (Cardiff Institute for the Blind (CIB))
The club meets at Jones Court, Womanby Street, Cardiff on the 3rd Friday of every month fron 10.30am - 12.30pm. Anyone with a sight and hearing impairment are welcome. Volunteers will be on hand to assist if need be.

In Touch befriender service - (Deafblind Cymru)
Volunteer befrienders will regularly visit people with combined sight & hearing loss, helping them to do more of the things they enjoy. This could include a visit to the local cafe, leisure activities such as rambling, help with information technologies, assistance with simple DIY tasks or a cup of tea and chat - whatever the individual wants. The volunteers do not provide a personal care service so will not be able to help with activities such as lifting people or giving medication.

Deafblind support - (Sense Cymru)
Specialist one-to-one support for deafblind people through both communicator guides and intervenors, and outreach support to children, families and deafblind adults in their own homes.

Usher syndrome
Form of deafblindness where people are born deaf or hard of hearing and start to lose their sight (with retinitis pigmentosa) in the school years.

Sense Usher Service - (Sense UK)
The Sense Usher Service specialises in the field of Usher syndrome and supports people with Usher, their families and professionals. Support covers advice, assessments, training, mentoring and professional support.

Degenerative Vitreous Syndrome (DVS)
The spontaneous occurrence in the aging vitreous of opacities (floaters) that substantially interfere with activities of daily living.

Degenerative Vitreous Community - (One Clear Vision)
An on-line forum for people with DVS, the term used to describe substantial cloudiness in the ageing vitreous humour which interferes with the activities of daily living.

Diabetes/diabetic retinopathy
Diabetes can affect the eye in several ways, the most common being problems with blood vessels in the eye.

Peer support network - (Diabetes UK Cymru)
The Peer Support network connects people living with diabetes, giving the chance to share information, experience and support. It is run by a team of volunteers, each with experience of managing and living with diabetes. You can talk by email at any time, or phone during opening hours.

Dystonia
A range of movement disorders that can affect the eye.

Dystonia Society helpline - (The Dystonia Society)
The telephone Helpline is available Monday to Friday from 10:00 to 16:00 to deal with questions about dystonia from both members and non-members, and from healthcare professionals. Helpline staff are not medically qualified and the Helpline cannot offer advice about an individual's treatment or provide any type of diagnosis. The service can however provide support and information on many aspects of dystonia and its management

Dystonia Society Forum - (The Dystonia Society)
An on-line forum where people can discuss any aspect of dystonia.

Fuch's dystrophy
A form of corneal dystrophy which occurs later in life.

Fuch's dystrophy information - (Fuch's dystrophy website)
A website containing information about the condition, and links to a support group for Christians with the condition. Scope is unclear, but seems to be American. Note: website currently inactive.

Glaucoma
A group of eye conditions which cause optic nerve damage and can affect vision. Glaucoma damages the optic nerve at the point where it leaves your eye.

Glaucoma Support Groups - (International Glaucoma Association)
The International Glaucoma Association organises patient support groups across the UK. The concept of the groups is to allow patients to meet their health care professionals in a relaxed atmosphere, away from the time restricted atmosphere on outpatients, so the condition and treatment can be discussed in more depth.

Sightline - (International Glaucoma Association)
Telephone and email helpline for practical help and advice about glaucoma and emotional support. The line is open Mon to Fri, 9:30 am to 5:00 pm. After office hours, an answerphone is in operation, where you can leave a message and they will ring you back.

Keratoconus

A form of corneal dystrophy.

UK Keratoconus Self Help and Support - (UK Keratoconus Self Help & Support Association)
A membership organisation for people with keratoconus. The website has information about the condition.

Keratoconus discussion forum - (UK Keratoconus Self Help & Support Association)
An on-line forum where people with an interest in the condition can raise and discuss issues.

Laurence-Moon-Bardet-Biedel (LMBB)

An inherited genetic condition. Effects can include vision impairment caused by rod-cone dystrophy, often diagnosed as retinitis pigmentosa.

LMBBS support - (Laurence-Moon-Bardet-Biedl Society (LMBBS))
LMBBS can help by putting you in touch with other families who are facing similar problems, by passing-on advice which has helped others in the day-to-day management of their children, and by sending information which will help professionals in medical and educational fields to understand more about our special children.

Marfan syndrome
A disorder of the connective tissue which can affect the eyes.

Marfan Association UK - (Marfan Association UK)
Provides information and a range of literature about Marfan syndrome. Members are also invited to various events organised by the association.

Neurofibromatosis
One of the most common neurological conditions, caused by a single gene mutation. May cause eye growths.

Neurofibromatosis support services - (The Neuro Foundation)
Membership gives access to advisors with information about neurofibromatosis. There are also local groups in some areas, where you can share information.

Norrie Disease
Norrie disease is a rare X-linked genetic condition which causes boys to be born blind or with severe sight impairment. Secondary symptoms can include progressive hearing loss, cognitive impairment, autism and delayed development.

Information for people with Norrie Disease - (The Norrie Disease Foundation)
The website contains information for families affected by this condition.

Nystagmus
A continuous uncontrolled to and fro movement of the eyes.

Nystagmus Network helpline - (Nystagmus Network)
The helpline can answer questions on what it's like to have nystagmus, on treatment and research, on DLA, on education and employment, on driving and discrimination, in fact anything you can think of related to nystagmus, whether congenital (early onset) or acquired (late onset). Or you can just phone for a chat with someone else who has nystagmus.

Retinitis pigmentosa
Changes to vision which may include difficulties in dim vision/dark, and loss of peripheral vision.

RP Helpline - (RP Fighting Blindness)
This telephone Helpline is for people with Retinitis Pigmentosa, their relatives and anyone who needs to know more about RP and how to cope with the difficulties it presents. Questions can also be sent via email. This service cannot provide diagnoses or counselling, and any information given is intended only as a guide.

UK Care Pathway for RP - (RP Fighting Blindness)
An on-line resource giving information about the care of retinitis pigmentosa. There are sections for patients, parents, family & friends, teachers, employers and health professionals.

RP Telephone befriending service - (RP Fighting Blindness)
The RP Fighting Blindness Telephone Befriending Service (TBS) links people who would welcome a friendly phone call from a trained person with knowledge of dealing with RP, with a team of volunteers willing to share their experiences. The TBS offers a social link on a more personal and longer term basis than the Helpline, with regular calls being made by the volunteers for as long as they are welcomed.

South Wales RP Fighting Blindness Group - (RP Fighting Blindness)
RP Fighting Blindness volunteers run a number of Local Groups in towns across the UK. These enable people with RP to socialise and support each other as well as raise funds and run events on our behalf.

Retinoblastoma
A fast-growing eye cancer of early childhood.

Support for parents of children with Rb - (Childhood Eye Cancer Trust (CHECT))
The Childhood Eye Cancer Trust (CHECT) is there to help you through the difficult period of diagnosis and treatment of your child and beyond, offering support throughout their teenage years and into adulthood.

Sjorgrens syndrome
A disorder of the immune system. White blood cells attack the body's tear and saliva glands, which reduces the amount of saliva and tears produced, causing a dry mouth and dry eyes, along with other related symptoms.

Sjorgren's Syndrome helpline - (British Sjogren's Syndrome Association (BSSA))
A telephone helpline for advice about this condition.

Wolfram syndrome
Wolfram Syndrome is a rare genetic disorder which is also known as DIDMOAD syndrome after its 4 most common features (Diabetes Insipidus, Diabetes Mellitus, Optic Atrophy and Deafness). Everyone with Wolfram Syndrome will have optic atrophy at some stage, which means that the optic nerve wastes away and causes colour blindness and gradual loss of vision. Wolfram Syndrome is very rare and affects around 1 in 770,000 of the UK population.

UK Wolfram Syndrome support group - (UK Wolfram Syndrome Society)
An on-line forum providing information and support to families affected by the condition. Some information is also available to download.

Additional Conditions and Syndromes

Blepharitis (eyelid problems)
An inflamation of the eyelids which can make the eyes feel irritated or itchy, but rarely causes serious eye damage.

Cataract
Cataracts are formed when the lens inside the eye gradually changes and becomes less transparent (clear), making vision mistier.

Cerebrovascular disease
Cerebrovascular diseases are conditions caused by problems that affect the blood supply to the brain. One of the most common types is stroke, which can cause loss of vision in one eye or on one side of the field of vision

Chorioretinitis (choroid retinitis)
A form of uveitis where the inflamation affects the back of the eye.

Colour blindness
Effects can vary: there are extremely rare cases where people are unable to see any colour at all, but most can see as clearly as other people but are unable to fully see red, green or blue.

Congenital cataracts
Cataracts which are present at birth (rather than as part of the ageing process).

Corneal dystrophies
A group of eye conditions affecting the cornea.

Dry eye
Caused by a problem with the tears which leaves eyes feeling dry, scratchy and uncomfortable.

Flashes/floaters
Flashes of light/small dark spots or strands which move. Generally harmless, but can be sign of retinal detachment

Giant cell or temporal arteritis
A condition affecting the arteries which can cause serious sight loss if untreated.

Hemianopia
An effect of stroke where one half of the visual field can be lost.

Keratitis
An inflamation of the cornea.

Light sensitivity (photophobia)
An inability to cope with glare which can be associated with another eye condition, or a side-effect of certain drugs.

Macular hole
A small hole in the macular - different from macular degeration. Causes problems such as straight lines appearing wavy.

Monocular vision
Blinding or removal of one eye due to accident, injury or disease.

Myopia (high degree)
A chronic, degenerative condition which can create problems because of its association with degenerative changes at the back of the eye.

Ocular pemphigoid
An autoimmune condition which affects the eyes by causing inflammation of the conjunctiva which is the thin layer covering both the white part of the eye (the sclera) and the inner surface of the eyelids. The inflammation causes redness, discomfort, grittiness and dryness of the eyes.

Optic atrophy
Damage to the optic nerve.

Posterior vitreous detachment
A change in your eye (such as floaters) which is frustrating but does not normally cause sight loss.

Pseudoxanthoma Elasticum (PXE)
Condition affecting the macular in one or both eyes, causing problems with central vision.

Punctate inner choroidopathy
A type of 'white dot syndrome' characterised by small yellow/white dots or lesions which are visible in an examination of the retina.

Retinal detachment
Occurs when the retinal layers separate from the retinal pigment epithelium - must be treated urgently.

Retinal vessal occlusion
A blockage in the blood vessels of the eye that can cause sight loss.

Retinopathy of prematurity
Generally associated with premature birth - before development of the eyes is complete.

Sarcoidosis
A condition where abnormal lumps or nodules form in one or more parts of the body, which may include the eyes.

Squint in childhood
A problem when the eyes stop working together as a pair and therefore do not look in the same direction.

Stickler syndrome
A genetic progressive condition, which affects the body's collagen (connective tissue). Possible impacts on the eye include cataracts, glaucoma & high risk of retinal detachment.

Stroke-related eye conditions
Stroke can result in visual field loss, blurry vision, double vision and moving images.

Thyroid eye disease
A condition of the soft tissues such as the fat and muscles surrounding the eyes which is characterised by a period of inflammation and engorgement of these tissues, followed by a healing response.

Toxoplasmosis
Toxoplasmosis is a common infection from which most patients make a full recovery. However in more serious cases it can spread to the eyes causing loss of sight.

Uveitis
An inflammation of the middle layer of the eye (the uvea)

What is the Perspectif Supplement?

This new series of supplements to **Roundup** quarterly features an area of the services listed online at WCB's Perspectif Portal. Each edition includes an article or two about the service area in question from people who are active in providing the service. It presents a way for WCB to highlight the work done by third sector, local authorities, health boards and others in providing support to blind and partially sighted people and make that work better understood and more widely known.

Perspectif is WCB's online database of services and support for people with sight loss in Wales. If you know of any errors or services not listed, please get in touch to let us know.

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