



Alström Syndrome UK

Strength for today, hope for the future

Alström Syndrome UK (ASUK) is a charity who provide information, support and advice for individuals affected, their families and carers and professionals. Alström Syndrome is a very 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. Our mission is to raise awareness, conduct pioneering research and enable better treatments and monitoring through the AS multi-disciplinary NHS clinics. Further information about our work can be found at www.alstrom.org.uk

Welcome to the Winter 2020 ASUK Newsletter

An update from Ann Chivers, Chief Executive

Hello One and All,

Firstly, I hope you and your family and friends are keeping safe and well. Usually I like to start our biannual newsletter with some upbeat news on what families are getting up to, but at the time of writing we are all at the beginning of the second lock down and I expect many of us may be feeling anxious at the prospect of spending lock down during the cold days and nights. So, I want to update you on what we have been doing to support families at this time and share with you some of the activities that the AS community has been involved in.



Ann Chivers, Chief Executive

At the start of the pandemic we contacted every family, known to us at ASUK and where needed, helped with signposting and navigating social services and other local services to make sure everyone could get their groceries, meds, appointments with GP's where needed, and offered a listening ear to those who were very anxious and confused about the guidelines. During the next phase, we have focussed on getting clear information out to families and presented webinars on a range of topics including managing diabetes and healthy hearts and minds. If you missed any of these webinars you can watch again on our website. For those of you who did join, thank you for your pertinent questions and as always, our gratitude to all the clinicians and professionals who gave their time to answer worries and concerns.

Virtual Clinics

As we have moved to getting used to virtual meetings, many of you will have attended your annual clinic online or by telephone. We know this isn't an ideal situation but I hope for those of you who have attended a virtual clinic that you have gained some small comfort from meeting with Tim and Tarek and teams at the QE and Birmingham Women and Children's hospital, all of whom are working extremely hard in very difficult circumstances to advise, support and help us all cope.

Global Alström Conference.

I hope by now you will have seen updates about the first ever Global Alström Conference taking place early in December. We are working closely with people around the world to bring everyone together, young and old, their families, clinicians, scientists, researchers and anyone in the Alström world, to learn more, share experiences, connect and support each other at this critical time. I look forward to seeing you there.

Meanwhile please take some time to complete the patient registry, this is a major investment of time and funds to build a bank of knowledge and information that we hope will benefit people with Alström.

At a more local level, we are pleased to see so many of you sending in recipes and ideas for the Alström Wellbeing pack, Carol and Marie have been cooking, testing out and enjoying many of your creations and we're hoping to be able to send you a small thank you in the post in December called; **Stay Fit, Stay Healthy, Stay Happy.**

You can find more recipes and cooking ideas on the website where Melissa (one of ASUK's Trustees) and others are demonstrating their fabulous cooking skills – it's better than Bake Off! You'll find other resources, top tips and ideas on managing well-being on the ASUK website too, as well as an up to date Covid support section. You'll find ideas and top tips, about taking care of our mental health, such as giving ourselves permission to say things are not ok, when you feel overwhelmed

<http://www.alstrom.org.uk/family-support/>.

Maybe this is a time for reflection and asking ourselves what we can do to help others? A time to come together and support each other, even if that is just a 'phone call asking someone, if they are okay?' Our lives are usually so busy that perhaps this is an opportunity to stop and smell the roses, well the wet leaves at the moment, but you get the idea!

Finally, a big thank you to all our funders, everyone who continues to send donations, the Doctors, Nurses and professionals who support our Alström community and last, but not least the ASUK team who are awesome!

See you at the conference, keep safe, keep well, keep going!

Best for now

Ann

Ann Chivers
Chief Executive

Around the world in two days!

Registration is open!

In true 2020 style – Let's bring the Alström global community together for the first ever, virtual Family and Professional conference.

We will tour the world, with insights from AS experts and inspiring personal presentations from families.

For the first time ever, not only will it be virtual, but it will be a fantastic opportunity to hear from scientists and researchers from China, Italy and here in the UK. There will also be time to network and chat together in our interactive sessions – how exciting!

Let's connect, learn together and be empowered...

Friday 4th December 2020 – Scientific Symposium, providing research and clinical updates

Saturday 5th December 2020 – AS Global Conference, bringing the Alström community together to share experiences from AS members and AS experts.

You will need to register to attend the event.

Pop along to the ASUK website for further information and to register
<https://www.alstrom.org.uk/as-conference-register-now/>

AS Global Conference



Register now!

Patient Registry – don't delay, enrol today!

With the world suddenly finding themselves in lock-down, let's take this opportunity to break out of ground hog day and complete the ASUK Patient Registry. This is an ideal time to get all your medical information and medications all in one place.

We have two new videos, featuring Ann and Kerry who answered your Patient Registry questions:

<https://www.youtube.com/watch?v=PhstDDBWtBQ&t>

Or you can check out their shorter version in this podcast:

<https://www.youtube.com/watch?v=9529wvuSQ3Q>

Knowledge is power!

The ASUK Patient Registry enables you to provide your health information in an easy and straightforward format by completing questionnaires about your health in a safe and secure way. This information will be available (anonymised) for researchers and clinicians worldwide to learn more about Alström Syndrome and develop treatments into this rare condition. It is free to join for both you and professionals. This information will help build a worldwide picture of Alström Syndrome, improving knowledge and understanding of this rare and complex condition for everyone on the planet!

You have the potential to make a real difference to everyone living with Alström.

How to join?

Find the CoRDS Connect website and complete the CoRDS activation form, it is quick, secure and easy to do, just go to the website below:

<https://cordsconnect.sanfordresearch.org/BayaPES/sf/screeningForm?id=SFSFL>

Don't delay, enrol TODAY!

A big thank you to those who have already completed the registry. Why not volunteer to help others? If you can offer any support to help others to complete the registry, please get in touch with Catherine.

Nigella eat your heart out!

Due to receiving Children in Need funding, we have been able to bring together a Wellbeing pack of resources with your favourite healthy recipes and top tips to stay healthy and happy. Over the past few months, we have enjoyed receiving your families favourite recipes. These have now been brought together into the family friendly 'Stay Fit, Stay Healthy, Stay Happy' pack. Thank you to everyone who has contributed. We can see people have had fun creating and cooking the recipes together. We would love to receive your photos of your kitchen creations and your bake-off masterpieces. We will be sending these in the post in early December.

Find the recipes and fantastic cooking videos on the ASUK website

<http://www.alstrom.org.uk/stay-fit-stay-healthy-stay-happy/>



Horsing around! Alex, shares her story...

As many readers will know, I've been horse obsessed and riding from about the age of 5. Over the years, the obsession that I had with horses has only gotten stronger and stronger. In 2018, after years of riding in riding schools, I began to loan a horse from a friend of mine. I was over the moon with this, as it enabled me to do all the things I loved, mucking out, grooming and generally just spending time around horses. I loaned him for 3 years, before I decided that I wanted to take my riding to the next level.

I began to look for opportunities to start competing within the discipline of dressage. My loan horse wasn't a suitable option and so I began riding with the local riding for the disabled (RDA) group. I thought that this would be the perfect opportunity, to gain experience within a setting that was used to supporting disabled riders. Over the years I've come up against a lot of barriers, and although I always found a way around them, I was hesitant to join the RDA. I always believed that they were only aimed at children and didn't have anything to offer me. I joined a local group in early 2019 and thought that I would be competing in no time. However, I was wrong.

The group I'd joined were lovely – the volunteers and the horses – however I soon quickly realised that they had neither the horses nor the capacity to be able to help me progress to competitions. It was suggested to me that I joined another group, which was further away from my house, but they were able to give me private one to one lesson and had experience supporting blind riders. I was dubious about what they would be able to offer me, due to my previous experience but decided to give it a go. My first lesson was so different from my previous experience of RDA, that I was quietly hopeful that maybe this could work. I began having lessons regularly and my skills and confidence grew from strength to strength.

When I joined, the centre was aware that I wanted to compete. Once they'd seen how I could ride we started working on a dressage test, with the aim of attending a qualifying competition at Hicksted in May 2020. This was an RDA specific competition and if I qualified, I would go to the championships at Heartbury in July 2020. Sadly, in March Covid-19 put a stop to my hopes of competing at Hicksted as the competition was cancelled. I was extremely disappointed, as my coach and I had put in so much hard work to get ready for the competition. The riding centre was also closed due to Covid and I stopped riding for almost 6 months. I wondered, if I would ever get back to riding and was extremely anxious. My mental health suffered, due to not being able to do both the exercise and the stress release that riding gives me.

In late July 2020, I was able to start riding again. With no sign of competitions taking place anytime soon, I spent some time enjoying riding again and going on some wonderful hacks around the countryside.

In early October, during one of my lessons, my coach mentioned to me that a website called Dressage Anywhere were running some online competitions and there was an RDA specific class. She asked if I would be interested in entering and after some discussion, I said yes. During our next lesson we focused on perfecting the test – thankfully it was a test I had already learnt for Hicksted – and the following lesson I was due to perform the test. The test would be filmed and then uploaded

to the website, for marking by an exclusive dressage judge. On the day of the test, I was extremely nervous and was unsure how I would perform in the test. Upon arriving at the riding centre, I was told the horse I had practised the test on was not available, so I had to switch to a different horse. Thankfully he was a lovely 15hh cob called Bomber, who I have spent a lot of time riding, however this change made me even more anxious. I can only ride accurately, because I have the markers around the outside of the arena called for me by a sighted person. I use these calls to know where I am in the arena. As well as by counting the horse's strides between each marker, and in each gate, I am able to perform each movement in accordance with the test. As I was riding a different horse, we had to do a quick recalculation and readjustment as Bomber is a bigger horse, with a bigger stride than my previous horse. After this was done, the test began.

I entered the arena at the A marker and had to proceed down the centre line and halt to salute the judge, before moving off to perform the rest of the test. Riding in a straight line is a particularly difficult skill when you are visually impaired. You get lost in the space and don't have any landmarks to use as a guide. After riding down the centre line the rest of the test went reasonably well – so I was told – I felt I made lots of mistakes but for a first time experience I was reasonably happy with how I had performed.

The video was reviewed and submitted online, and I had to wait for the results. I was again nervous about how I'd done. I was expecting a low mark as I have convinced myself that I hadn't done very well. I logged onto the Dressage Anywhere website a week later and the results were in. I opened the score sheet with bated breath and began to read my scores and comments. I was overcome with shock as I read that I had achieved 6's and 7's out of 10 across the board, for all the movements I'd performed and got some great comments. I got a total score of 70.56%! I couldn't believe it. I clicked through to the scoreboard where the placing was uploaded and when the page came up, I was delighted and shocked once again to see that I'd come in 1st place!

All the hard work that I'd put in, all the hard work my coach had put in had finally paid off. We were both delighted with my score and are now beginning to work towards moving up to compete at the next level.



The Cochlear Implant process, the ups and downs

Chris and his Mum Carol, share their journey...

After several years of waiting to be considered for a Cochlear Implant and many more years of feeling isolated, this journey shows the ups and downs of life before and after having a Cochlear Implant. The frustrating waiting game for surgery, the lessons the medical teams learned along the way and finally the astonishment as the day arrived when he was able to hear the birds singing, the rumble of his cane on the floor and the trickle of water, sounds that had not been heard for a lifetime.

What is a Cochlear Implant?

A Cochlear Implant (CI) is a device which attempts to replace the function of the damaged cochlea by electronically stimulating the auditory nerve to produce a sensation of sound. CI consists of internal and external components. The internal component is inserted during an operation that lasts approximately 2 hours. The internal part is called the receiver or stimulator or electrode array. There are 3 external components. A transmitter coil, a speech processor and a microphone that is part of the speech processor. The external components allow the internal component to receive sound and are generally fitted around 4 to 6 weeks after surgery. This is referred to as the 'switch-on'. It can take several months for the ear to retrain and make sense of the sounds.

The whole process can take up to two years from once you are referred, until you have a successful implant.

It's also worth mentioning that once referred, there is no guarantee that you will be accepted onto the Cochlear Implant programme. For this case study, the process took several years before a multi-disciplinary team at the Cochlear Implant centre, made the decision to go ahead with surgery. Even on the day of Cochlear Implant surgery, there were still some questions raised that may have stopped the surgery from taking place.

Background information

Chris was diagnosed with Alström Syndrome and registered blind at the age of 5. He has a profound hearing impairment, now wearing digital hearing aids in both ears. In more recent times, he became aware that there were elements of everyday conversation that he could not hear. In noisy environments, such as supermarkets, restaurants and crowds of people, he could not hear the person talking at his side. Outside, he struggled to hear traffic noise, one-to-one conversations and in windy conditions, this made any directional sound clues a real challenge. He became unsure of his surroundings and had to stop using his long-cane mobility skills to walk independently, even on familiar routes to familiar places. He became aware that the digital hearing aids, now working at their maximum capacity, were not providing him with enough audible information to carry out everyday tasks. Socialising with friends, talking on his mobile phone and shopping tasks became more and more frustrating as conversation and communication became increasingly difficult.

Audiology equipment

Chris explored the options of stronger digital hearing aids and other equipment, with the help of his local NHS audiology department and was told that he was indeed using the strongest digital aids available. After talking with other people with a severe hearing loss, Chris found the use of a Roger Pen to be beneficial. The device looks like a pen and works like a microphone, sending sound to the hearing aid via a Bluetooth connection. A small 'shoe' is fitted to both hearing aids. The Roger Pen can be worn around the neck on a cord with a magnetic clasp for individual conversations or placed flat on a surface for Round Table discussions or it can be used directionally to pick up sounds in any direction. This equipment worked well for 12 months, although it had become clear within this period that a long-term solution needed to be explored.

The Cochlear Implant referral process

The Ear Nose and Throat (ENT) Consultant had remarked that there was a definite change in Chris's behaviour. He had noticed that he had become withdrawn, frustrated and angry at life. The Consultant suggested that it may now be time to try to re-engage the Cochlear Implant team services to explore the possibilities of this surgery. The Consultant made the referral to the Cochlear Implant Centre, in April 2018. Two years previous, in 2016, Chris had met with the Cochlear Implant team, however, at that time, even though he had a severe hearing loss, his word recognition was still too good to be considered for an implant. Before this, Chris had been referred to the adult Cochlear Implant Centre in 2007, where his initial assessment of his hearing was it was too good to be considered for a Cochlear Implant. The referral process, this time, produced an early appointment for a 2-hour Cochlear Implant assessment.

The 'word recognition' test

Chris, had a series of hearing tests including a word recognition test. He was asked to repeat words and phrases from a male and female voice recording. The outcome would decide if his hearing was impaired enough to qualify for the next step on the Cochlear Implant programme.



He didn't have to wait long for the word recognition test results. Listening to the male speaker, he scored '90% keywords correct' using bilateral hearing aids and '84% keywords correct' from the female speaker. The right ear showed a marked reduction in accuracy but overall, he did not meet the criteria to be considered for a Cochlear Implant. The official response remarked that his word recognition, even with his profound hearing loss, was still better than a typical Cochlear Implant user. The Cochlear Implant team have a strict threshold and even though the deteriorating hearing and sight-loss was considered, he still did not meet the criteria. This was devastating news for Chris.

The local ENT consultant was also disappointed with the news. Another hearing test revealed further deterioration and the Consultant asked if it would help if he wrote to the Cochlear Implant team again. He felt that it did not serve Chris's best interests to delay the Cochlear Implant surgery process, given that there was clear evidence that there was further significant deterioration in hearing. The Consultant revealed that he knew 3 of the 4 surgeons and felt that it was worth asking for a reconsideration.

After a further 8 weeks, the Cochlear Implant team wrote to Chris inviting him to a review meeting where he could repeat the word recognition tests. This time, with much relief, he was now considered to be a potential candidate for surgery.

Attending a review meeting, at the Cochlear Implant centre, was a very stressful time. Nothing was certain. The future was unpredictable. Chris was not hearing one-to-one level conversations, even in the quietest of rooms. He could no longer have a one-to-one conversation with anyone outside. Family and friends could only converse with him by raised voices. Socialising with friends had become so painfully frustrating that it was easier to decline invites and isolate himself. He resorted to text messages to communicate, via his Braille sense machine. His independence was greatly impaired, his social isolation increased, and his mental health declined daily.

The next step: physical suitability

It was welcome news that the Cochlear Implant team had reviewed all the information and accepted the Chris as a candidate and they now needed to explore if he was still physically suitable. An appointment arrived for a CT scan followed by a further consultation with the Cochlear Implant team. The CT scan presented physical issues as Chris had increasing kyphosis (curvature of the spine) in his neck and upper back area. The CT scan operator did not want to provide a wedge or sponge block to support his neck, stating that he would only be lying down for 5 minutes. Chris said this would cause him severe pain and would like a support during the CT scan. Reluctantly, the CT scan operator rolled up a pillow and attempted to use this as a support. It was a painful process; his anxiety and stress levels were not helped by this process. The second scan on the same day was an MRI scan where the adult male had a 20-minute scan to look closer at the cochlea within the inner ear.

Shortly after the CT scan, one of the Cochlear Implant team discussed the various Cochlear Implants available and their suitability. Chris was keen to explore the latest technology including MRI compatibility, Bluetooth connection to his iPhone, streaming options and other devices. Chris was given a glossy folder with the available options. He asked if there was a Braille copy or a word document that he could access via email or a website where he could access the information. Chris was told that there was not an accessible option available for a blind person. He took the brochures home and I read them to him. This took a few sessions so that all the information and options could be explored and discussed. This was to ensure that he was able to make a choice, based on his own health needs and personal preferences.

By this time, Chris had attended 16 appointments. He had no remaining useful hearing and all communication was via his Braille sense machine and a Qwerty keyboard. The deterioration into profound hearing loss/deafness had a slightly better transition as his support staff had all been trained in the use of this technology, in preparation for further hearing loss and for times when his hearing aids could not be worn. It was hard to accept that his hearing had to deteriorate to this level before a decision to go ahead with Cochlear Implant surgery could be made. The next 2 appointments involved further hearing and word recognition tests to determine and confirm the previous tests. Further discussions with the team resulted in a decision that he could be considered for the next stage of the process; surgery.

Preparing for surgery, medically and emotionally

Chris, was told the waiting list was approximately 3 months and he could expect surgery to take place within this time. In preparation, it was important to discuss the procedures including anaesthesia, communication with nursing staff post-surgery and the recovery time. People with Alstrom Syndrome are at risk of post-operative hypoxia where oxygen levels can decrease after a general anaesthetic. At this stage, Chris asked for a letter of support from Dr Hiwot at the Inherited Metabolic Disorders service at Queen Elizabeth Hospital, Birmingham. It was important to discuss with Chris exactly what needs would, or could, be met during his time in hospital.

One of the challenges of a hospital stay, involved how staff at the hospital could communicate with Chris, someone with sight and hearing loss in a hospital environment. There was no guarantee that staff would be experienced in caring for someone with a dual sensory loss. It was important to create a hospital support plan for staff, so that all Chris's needs were met. This took many hours of discussion, exploring all the possible challenges. A final care plan was created with Chris, a copy of the Alstrom Syndrome Medical Handbook was given to the team and discussed with the CI co-ordinator. After careful consideration, the CI co-ordinator decided that the best option for Chris was to have a familiar supporting adult with him throughout his hospital stay. Chris was relieved at this prospect and felt less anxious about his forthcoming surgery. The hospital care plan would be added to the notes for the attention of all clinicians and nursing staff.

Chris, wanted to find out other adults' experience of the CI process and if they had any top tips that would help him adjust emotionally. He contacted 2 other Alstrom patients and discussed their experience with them. One top suggestion was to prepare by repetitively listening to a talking book so that it would become very familiar to you. It was also suggested that it would be very useful to have flash cards. These were created with simple phrases, laminated on white card, with Braille overlaid so these were accessible to all.

Another top tip was to be prepared for frequent, lengthy appointments at the CI centre. Chris used these suggestions and replayed the Harry Potter talking books each day in preparation. He created his own set of flash cards to use in and out of hospital and at each CI appointment he took his own soft drinks and sandwiches.

The hospital admission and surgery

On the day of surgery, Chris had prepared himself well, with a hospital care plan in place and his familiar supporting adult, me, by his side, he felt confident that his needs would be met. The anaesthetist confirmed all the information provided by Dr Hiwot around post-operative hypoxia. The surgeon confirmed the type of CI to be fitted would be an MRI compatible one and finally he was able to change into a gown and pressure socks, in preparation for surgery. The one issue that had been overlooked was whether his neck could be tilted backwards enough for the intubation tube to be inserted. This was not the case and, at the last minute, intubation had to take place with a tube up the nostril and down the throat. It was uncomfortable for Chris and me to witness. Disappearing through the theatre doors, Chris's long awaited Cochlear Implant surgery was finally taking place.

Post-operative care

After 2 hours of surgery, Chris was transferred from theatre to the Acute Medical Unit (AMU) for close post-operative observations. He had his head bandaged and the canula had been left in place, to deliver IV fluids. It was necessary to monitor his blood-pressure and oxygen saturation (SATS) through the night. Chris had been given routine anti-biotics after surgery to prevent infection. He was in a great deal of discomfort and complained of severe head pain. Analgesics were given to relieve the pain and soon he was able to have some sandwiches, use the toilet and eventually have some restful sleep.

At this time, there were no plans in place for me to remain with Chris and the care plan had been ignored. As nursing staff were unable to use the Braille communication equipment and Chris was unable to communicate effectively, it was important that I be allowed to remain with him overnight to prevent any issues. A bed was eventually found at 1am.

During the night, I discovered Chris was bleeding through the bandages and alerted staff. It took some time for the bleeding to stop and the wound was re-dressed, and more pressure applied. It was a distressing time. The flash cards came in very useful to communicate with Chris and reassure him.

Discharge from hospital

The following day, after an x-ray and a visit from the surgeon, Chris was discharged to recover at home. An appointment was made to attend the hospital, one week after surgery. At this appointment, Chris had an x-ray to check that the implant was still in the correct position. The soft tissue would take 4 to 6 weeks to heal. Further appointments were scheduled for the 'switch-on' or Initial Programming, for Audio Rehabilitation and for further CI Programming.

Coping with isolation: what helped

During this time, Chris relied solely on his Braille sense machine to communicate with everyone. It was important to take time to communicate in this way to limit the isolation he felt. It was important to remember that previously good independence skills were reduced, and reassurance was needed to encourage any independence. Every part of the day was planned and discussed through his Braille sense machine and a backup Perkins Braille machine was used when technical issues occurred with the Braille sense machine. In order to be included in any activity such as shopping, or eating out, every detail needed to be explained via a Braille machine as there was no effective communication once leaving home. The 'flash cards' were tailored to his own needs and really helped communication when outside as these could be easily handed. These can be made in advance and tailored to each persons' own needs.

Here are some examples of the flash cards: Are you ok? Are you in pain? Would you like a drink? Have you eaten? Have you taken your medication? What would you like to do today? How can I support you today? Would you like to go out for a walk? We need to leave here now. There is a problem but it's being taken care of for you. It's ok, you're doing great. I'm leaving the room now. Back in a few minutes.

During this period, Chris was quiet and experienced great isolation. He found comfort in creating with clay and his pottery sessions became the most important part of his week. In these sessions, he could relax.

Switch on: let the magic commence

The process of programming the processor is commonly known as 'switch on'. The processor is worn like a hearing aid and is attached to the programming equipment by wires. The Audiologist has a screen view of 12 different frequencies, and each is raised in turn until Chris could hear sounds. As each frequency level is raised, the electrode array is stimulated and, on this occasion, Chris started to experience pain and the programming had to be stopped. The process was repeated after a further 14 days and was more successful. Chris was given an array of equipment that accompanied the CI, all neatly packaged in a rucksack. The equipment included a toolkit for changing the various parts of the processor and lead, a hearing aid sanitiser box, a waterproof cover for the processor and an interchangeable rechargeable battery pack with charging unit. He was given a box of disposable batteries to use and a contact number to call if there were any issues. It was suggested that he try to experience as many sounds as he could tolerate and to ask someone if he didn't recognise a particular sound.

On leaving the CI centre, Chris was able to hear the rumble of his long cane rollerball on the pavement and traffic noises. He could hear a distant aircraft flying overhead. He listened to LBC on the radio on the journey home in the car, for the first time in years. He said we all sounded like 'mechanical chipmunks' and laughed each time I spoke. He was told that the quality of the sound would improve the more he listened to different voices and sounds. Thankfully, the headaches didn't return, and he started to build his 'sound library'. It was wonderful to be a part of this journey and each new day brought more sounds into his life.

Rehabilitation: building the sound library

Further programming sessions and adjustments continued weekly, for 6 weeks, at the CI centre, until the frequency levels matched his hearing loss. Hearing tests proved that the implant was working well. Chris was shown how to use a streamer device where he could receive and make phone calls using his iPhone. The voice would go straight into the processor so that the incoming voice was private. This was an amazing addition as voice calls had proved to be troublesome since switching on the processor.

There were so many wonderful moments of witnessing Chris hearing sound for the first time. One memory that sticks in my mind is being asked to come into the bathroom quickly as Chris had picked out a sound that he didn't recognise. It was the sound of the water, from the shower head, hitting the bathtub below. He had never heard the sound of sprinkling water, hitting a surface, before now. I later caught him listening to the water turned on full, as the water splashed from the taps into the handwash basin. He remarked how noisy the birds were in the morning and that there were several magpies in the tree, in a neighbouring garden, that he didn't know were there! Truly remarkable!

Equipment failures: how he adjusted

There were some issues with the processor within the first 2 months. Using the streamer to take phone calls, caused a surge in power and the disposable batteries would fail and this created a fault causing the processor to stop working. The CI centre were swift to respond and reprogrammed another processor. This was posted out by next day delivery to lessen the impact. When the second processor failed, Chris returned to the CI centre for a full investigation. The audiology repairs team were brilliant. Their technical knowledge meant they could identify which part of the equipment was failing. After 3 processors failed, with the same fault, the CI team suggested a meeting with the manufacturers to find a way forward.

The meeting with the manufacturers Medel – took place at the CI centre and included the technical manager from the CI centre as well as the Senior CI Programmer and Chris and I. The Medel salesman was very helpful and even though he could not resolve the issues on the day, agreed to take the faulty processors back to their German factory to be analysed further. Initial tests revealed that the act of using the streamer device, also made by Medel, drained the disposable batteries and caused a fault however, if the disposable battery unit was changed for the rechargeable battery unit, the processor continued to work with the streamer. This has continued to work well however; the rechargeable batteries are fiddly to fit in the charging unit as they have no distinguishing marks. This makes recharging the batteries very difficult when you are visually impaired. Chris has not been able to successfully recharge these batteries independently, so he is looking for another solution. The full report from Medel has not been received to date and emails remain unanswered. The next appointment at the CI centre is a 6-month check-up.

The lessons learned

Chris would ask people considering the CI process to be patient with the process. He was not accepted onto the CI program initially. Ask lots of questions and don't give up if you believe that your hearing is so poor that you no longer enjoy everyday communication. Remember that the CI is designed to hear better speech and an aid to communication. Music may sound very different from what you listened to previously. Persevere when trying to listen to sound with the new CI. It does get better. The more you wear the processor, the clearer sounds become. Listen repeatedly to the talking book that is familiar to you. Don't give up.

What's been possible: MRI scan

Chris, had his first MRI scan, with his MRI compatible CI, as part of the Alstrom Syndrome clinic in February 2020.

The future: sounds and socialising

Chris, was unable to hear his metal music with his CI, to begin with. It was suggested that he listen to music where there was one musical instrument so either a guitar or keyboards with no vocals and then gradually introduce songs with a combination of 2 then listening to a combination of 3 and so on. This has worked well as the brain adjusts to isolated sounds through the CI before it can build a sound picture of lots of instruments. Chris emphasises that it takes lots of repetition and daily listening to his music to begin to make sense of it.

To date, seven months after the initial programming of the CI, Chris reports that his metal music is beginning to sound like it did before the CI was fitted. This is great news for him as his biggest fear was that he would not be able to recognise his favourite bands and music. He says it's been a worthwhile journey as he can now enjoy conversations inside and outside of his home. Socialising has become a pleasure again and he is keen to re-establish himself at open mic nights. He also plans to resume bass guitar music lessons. Plans are underway to safely attend an indoor metal music festival in 2021!

University Hospital Birmingham have useful information on their website about cochlear implants, including patients videos

<https://www.uhb.nhs.uk/cochlear-implant-videos.htm>



A BIG THANK YOU to Chris and Carrol for sharing their personal journey with us.



Guide to having your flu vaccination (jab) during the coronavirus pandemic

1

It is important to have any vaccines such as your flu vaccination, during the coronavirus pandemic.



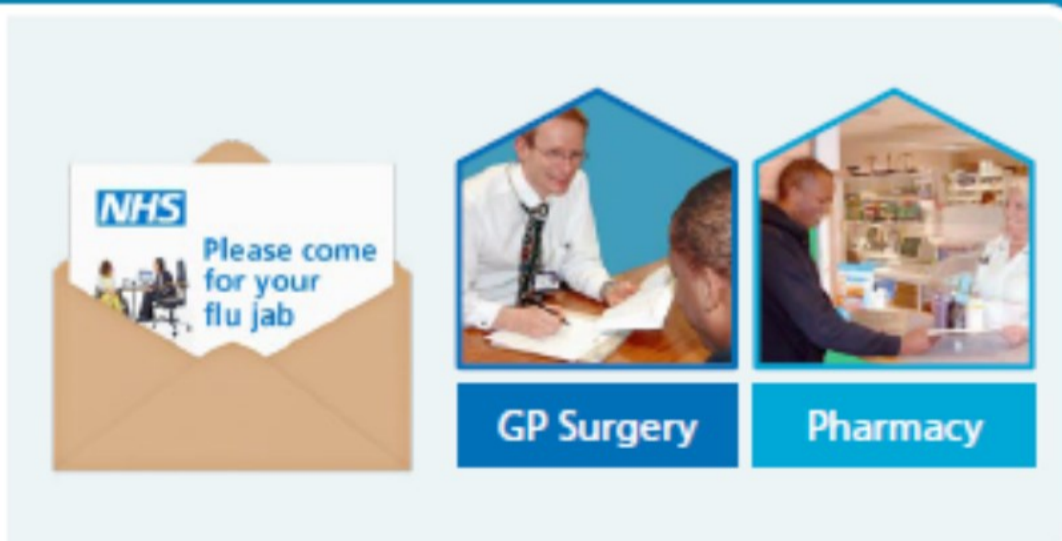
2

You should not have your appointment if you are feeling unwell or think you might have coronavirus.



3

You will have a letter telling you to go to your doctors surgery (sometimes called your GP) or to your pharmacy.



About your appointment

- 1** Your appointment may be in a different place from usual. Please check the address and make sure you go to your appointment at the right time.



- 2** People travelling on public transport and visiting health services are meant to wear a face covering although some people don't have to.



- 3** You may have to follow arrows or footprints to show you which way to go. Everyone will work to keep you safe while you have your flu jab.



- 4** Make sure that you wash your hands with soap and water or hand gel after you travel and when you get home.



- 5** Your nurse may be wearing some protective clothes called PPE. For example:

A mask



An apron



Gloves



A visor
(plastic face covering)



This is
to keep
everyone
safe.



To find out more information about your flu vaccination (jab) you can go to this leaflet and why you need to have it:

www.gov.uk/government/publications/flu-leaflet-for-people-with-learning-disability

First walks alone, you can do it!

Kathryn shares with us, her independent travels since restrictions came into force.

I was very anxious about setting off for my first walks on my own, with my guide dog and without my mum shadowing us. With time, this gradually went, as I learnt to trust myself and built up my confidence about completing routes, I know I can do, without getting lost or taking a diversion. Most of the time, if this happens, I try to retrace my steps and sometimes this works and at other times it doesn't. I make sure I have my mobile phone to call for a rescue from my mum in the car. I monitor my alertness and energy levels daily. On days where I am feeling tired, or want a walk where I can think, or do not have concentration, then I choose an easy short walk. On days, when I can concentrate, I choose a longer more challenging walk. I have two long walks, two medium walks and two short walks that I can do myself. My guide dog Emerald loves finding poles, the post-box and bus shelters so we try to do these on our walks. I find Saturday is the busiest day and Monday is the quietest day.

My first bus journey

My guide dog Emerald and I, set out after I had double checked I had her harness, lead, gentle leader, poo bags, clicker, kibble, my mobile phone, purse, bus pass, key, mask and hand sanitiser with me. I make sure we stay away from people and if they are close, from their noise, we stop and wait a short time before continuing along our route. I harness walk Emerald between my house and the bus stop which is a ten-minute walk. At the bus stop, I hand sanitise using my non-alcoholic spray due to my alcohol allergy and put on my small pink mask and once adjusted I leave the mask alone. I do not have to wear a mask due to my asthma, but I can tolerate it for short periods, and it protects both myself and other people. I get out my bus pass and get Emerald to sit and wait for the bus. Once it pulls up, we get on it, as I ask Emerald to find the step and ask for our stop. I get Emerald to find the seat and then I sit down and tap the side of the bus and say come, sit, down, stay and get her to lay in front of me or beside me. At the other end of our journey, we get off the bus. I sanitise my hands again. If the place, we are going to, say the doctors' surgery which is close to the bus stop then I leave the mask on, as it's only a five-minute walk away.

At the doctor's surgery, they have a buzzer and the doors open. You can then walk into the reception area, where there is a table to keep you away from the counter and be dealt with there and then. If you have an appointment, you go into the waiting room and get called to go and see the doctor and then walk out again. Once I am walking back home, I take my mask off and carry it or put back in its plastic bag and wash it at home. I do this, as walking back there are stairs to climb and a very hilly road and I cannot breathe with the mask on.

My first taxi journey

Before I could book this appointment, I had to complete some paperwork and send my contact details. Before I left home, I washed my hands and put my mask on, checked that I had everything and was ready to go. I phoned the Taxi and booked our journey to my massage therapist. It is a 15- or 20-minute journey between our two houses. Emerald jumped in the taxi and laid down nicely and the journey was good. We arrived and because I just had to walk up her drive and climb two flights of stairs, I kept my mask on. Then we had to complete some paperwork and get my temperature checked before sanitising my hands. I had my treatment done and paid for it. I had to keep my mask on the whole time. My massage therapist now has plastic wrap over the chair, and she must open the window. She also must clean around everywhere and change the towels and bedding she uses. I sanitise my hands and then I get a taxi home. I was glad to take my mask off at last!

Village trip

I walked to the village. Near to where we live to go to the pharmacy and Peaches, a beauticians. I walked down with Emerald on harness and I was not wearing my mask. I stopped to hand sanitise and put my mask on before I went into the pharmacy. I did what I needed to do and left. At present only two people at a time are allowed in the pharmacy. I then proceeded to Peaches as they are on the same street, because I was early, I stood outside in the sunshine. I still had my mask on. I went in and had my treatment done, paid and came out. I crossed the road and at the bottom of the stairs I needed to climb, I took off my mask as the walk home was uphill.

Trip to the hairdressers

I went to the hairdressers and another pharmacy. This time I took the bus and wore my mask on the bus, walking to the hairdressers and in the hairdressers itself. They washed and cut my hair around it. I paid and left. I went into the pharmacy with my mask on and they had tape where you could not go up to the counter directly. The staff love Emerald. Once I was done, I left still wearing my mask as we had to cross the road and then get the bus home. Once home I took off my mask, washed hands and washed the mask.

Hospital visits

My mum and I went to my first hospital appointment. We made sure we had our masks and hand sanitiser, letter and shoes and insoles. We took the car to the Hospital. When we arrived, we put our masks on and went into the reception area where we sanitised our hands and we were asked whether we had any covid-19 symptoms. We waited and then my name was called, and we went into the room where I had my insoles fitted into two pairs of shoes. Afterwards, the person walked out with us and we left in the car. Once in the car I took off my mask. My mum and I took the car to a different hospital for my Ear, Nose and Throat (Ent) appointment. We arrived in the car and put our masks on, before going into the hospital and once inside we sanitised our hands. We went upstairs which said on them, keep to the left-hand side. I would not have known this without my mum there. We went to the reception desk and they gave us surgical masks to wear so we swapped masks. I sat down and my mum had to sit two metres away from me. I got more hearing aid batteries and elbows with my yellow book. Hearing aids are composed of the actual hearing aid which is the top bit with the battery compartment, mode button and volume switch. I have three modes on mine, the normal mode, t-loop mode for listening to the television and an FM mode for my direct lead headphones. For these headphones, you need different shoes which is what holds the battery compartment at the bottom of the hearing aid. The hearing aid then has piece between it and the tubing which is called the elbow. They are different shapes depending upon who makes your hearing aid. The tubes are different sizes too. The tube attaches to the mould which goes into your ear. Some hearing aids have filters that need changing every month. After this, I was called, and we went into have my ears suctioned to remove the wax. Then once done we walked out of the building and took off our masks and went home.

Trip to the dentist

I recently visited my NHS dentist for a check-up. I walked down with Emerald on the harness. Once close to the door I put my mask on. I went into the reception area and booked in, before going upstairs to the waiting room. When I was called, I went into see the dentist. I took off my bags and made Emerald lie down away from me. The dentist helped me into the chair, and we talked through our masks. I took off my mask only when he examined my mouth and then put it back on again. Once I had my prescription, I headed downstairs, got it stamped, gave them a list of my medications on repeat prescription and got another appointment for three months' time. Once outside, I took off my mask and walked home.

Lessons I have learnt

- ♦ Ask a bus driver to radio control if they do not know the stop you are getting off at. They should know the route and the names of the stops on that route.
- ♦ Wearing a mask is ok for short periods.
- ♦ Hand sanitising is necessary and important.
- ♦ Do not be scared to try something new but take it slowly and when you're ready.
- ♦ Do not do something new on a day when you are very tired, lack concentration or feel unwell.
- ♦ Do go to your GP with other things that are not related to Covid-19. For instance, I had a stomach bug that needed treatment. I went to my GP and she checked me out using full protection of mask, apron and gloves.
- ♦ Be aware of tape and screens blocking your proximity to people behind counters.
- ♦ Be aware that you may not hear people clearly if they are wearing a mask or behind a screen.
- ♦ Be prepared to repeat what you are saying.
- ♦ If you can get things online do so as it is quick, easy and mostly straight forward.
- ♦ If family members can help you out, then let them do so.
- ♦ I would not have got through this pandemic without priority Tesco slots (I had to ring Tesco directly to arrange these) and prescription deliveries from my local pharmacy.
- ♦ Be prepared to order your medicine online and for it to not always go well.
- ♦ A pharmacy outside your local area will not deliver.
- ♦ Be prepared to spend your time learning online platforms like Zoom, Microsoft Teams, Go to meeting and WebEx. Accessibility features may be limited.
- ♦ Make sure you phone friends and family more frequently.
- ♦ Look after your physical and mental health properly.

Stay safe, Stay well.

It's beginning to look a lot like Christmas!

For more than 20 years, the elves and fairies at RNIB have helped Santa with hundreds of letters – they love opening the mail and reading about what children have been up to and the things they would like for Christmas. They help Santa send out his replies in each child's preferred reading format. The format can be in Braille, large print, audio CD or in a 16pt email.

If you are quick there is still time to get your letters to Santa, you have until the 1st December 2020 if you would like a letter in the child's preferred format or you can get an email reply and the deadline for this is the 21st December 2020.

There is a form you can complete online and upload your letter or alternatively you can send your letter by post to Santa Claus, RNIB, Midgate House, Midgate, Peterborough PE1 1TN. If you do send a letter by post, please make sure you include your child's first and last name.

National Braille week

In October, we celebrated National Braille week and we were delighted to hear the progression of the Lego Braille bricks.

The Lego Foundation is teaming up with charities around the globe including RNIB to promote the importance of learning through play.

The organisation's latest venture is Lego Braille Bricks, a new concept of play based methodology that aims to teach braille to children who are blind or have a visual impairment. RNIB are the sole distributor of Lego Braille Bricks in the UK.

Each brick in the Lego Braille Bricks toolkit retains its iconic form, but unlike a regular Lego brick, the studs are arranged to correspond to numbers and letters in the Braille alphabet. Each brick shows the printed version of the symbol or letter, allowing sighted and blind children to play and learn together on equal terms. This ingenious combination of features opens a whole



For more information and to complete the form, head across to the RNIB website.

www.rnib.org.uk/information-everyday-living/family-friends-and-carers/resources-parents-blind-or-partially-sighted/



new world of playful learning that teaches children Braille in an enjoyable and tactile environment.

With thousands of audio books and computer programs on the market today, fewer young people are learning Braille. Yet, individuals with blindness or impaired vision all over the world rely on Braille to work, study and enjoy their daily lives to the fullest. This simple but highly practical educational tool could teach a range of skills needed to thrive.

We would love to hear your experiences if you have used these bricks.

ASUK offer a help-line during office hours and an answer phone service at all other times where calls are promptly returned by our experienced Family Support team;

Please call 07970 071675 / 01709 210151



Meet the ASUK team

Please get in touch if you have any articles or suggestions for our next edition, Catherine is looking forward to hearing from you!



Kerry Leeson Beevers
National Development Manager
Monday-Friday
Email:
Kerry.leeson@alstrom.org.uk



Jane Biglin (South)
Senior Family Support worker
Working days are Tuesdays and Wednesdays
Email: jane.biglin@alstrom.org.uk



Asya Choudry
Community Engagement Manager
Monday, Wednesday and Thursday
Email:
asya.choudary@alstrom.org.uk



Carrol Birchall (Midlands & North)
Senior Family Support worker
Monday-Thursday
Email:
carrol.birchall@alstrom.org.uk



Marie McGee
National Transition Coordinator
Part time
Email:
marie.mcgee@alstrom.org.uk



Catherine Lewis
Office Manager
Monday - Friday
Tel: 07970 071675
Email:
Catherine.lewis@alstrom.org.uk



Steve Scoffield
Finance Manager
Works one day a week usually on a Wednesday
Email:
steve.scoffield@alstrom.org.uk

Registered Office address: 4 St Kitts Close, Torquay, Devon TQ2 7GD

For all enquiries please get in touch on **07970 071675**

Please visit our website www.alstrom.org.uk to keep up to date with our latest news.

ASUK has a dedicated You Tube page <http://www.youtube.com/alstromsyndromeuk>

Please keep up to date with Alström Syndrome UK via twitter [@AS_UK](https://twitter.com/AS_UK) and also via our dedicated Facebook page www.facebook.com/alstromsyndromeuk please click 'like' to show your support and spread the word!

You can donate securely online, visit our Just Giving page to make a difference today, <https://www.justgiving.com/alstromsyndromeuk>

