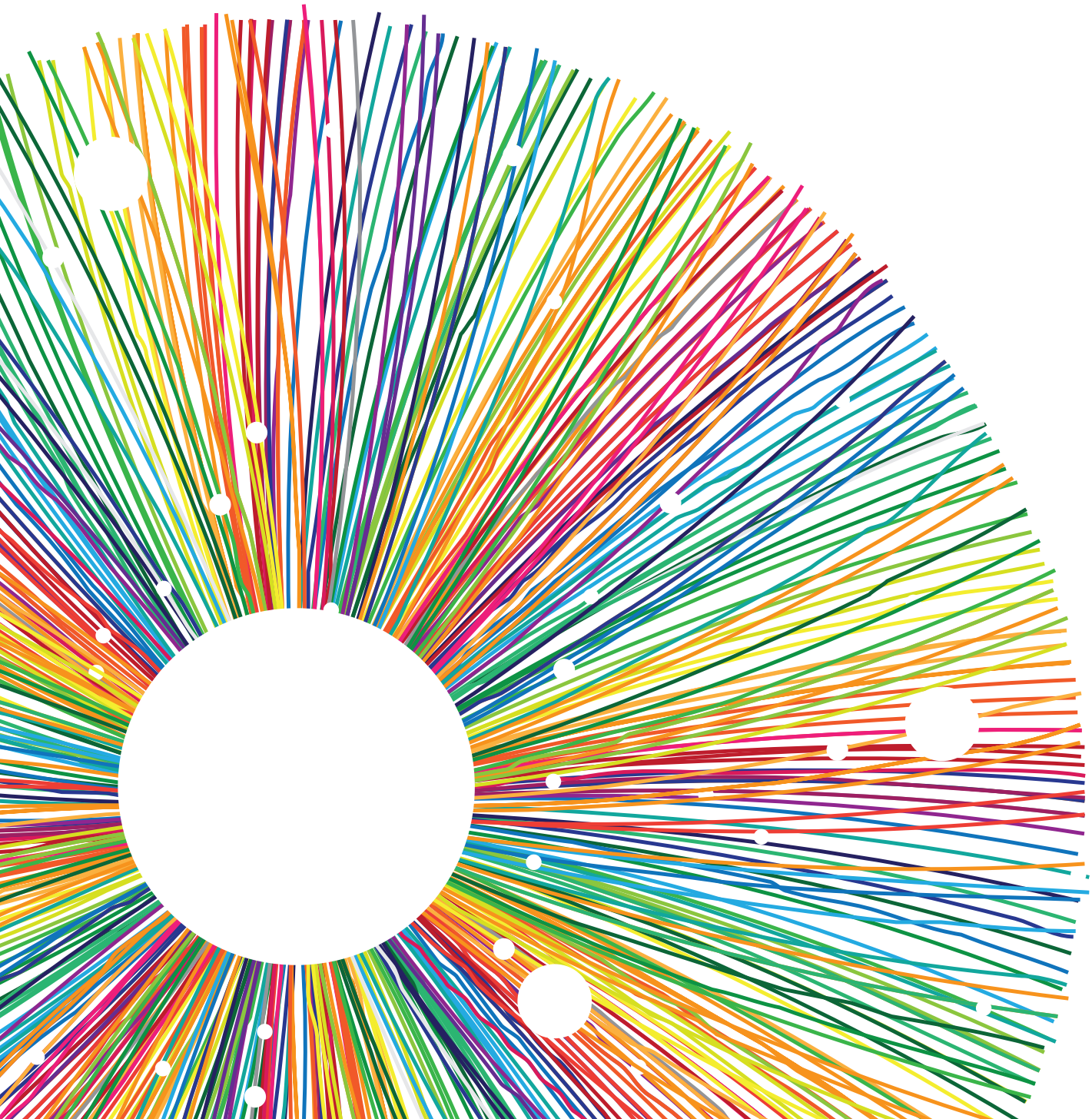
NEURO KEY

An Alliance supporting people with neurological conditions

FRIEDREICH'S ATAXIA

'Unlocking minds and opening doors'

Open Door series by Neuro Key



Contents

Foreword

Since inception Neuro Key, the working name of the Charity, Tees Valley, Durham and North Yorkshire Neurological Alliance, has harnessed lived experience as an educational tool to improve service delivery and professional understanding of not only the common threads between neurological conditions but also the complexities. From enabling participants to be fully included in research, delivering lived experience seminars, making sure people can respond to surveys and strategic consultations or supporting people to lead the operational direction of the Charity, we are passionate about our social purpose.

We promote a culture of mutual exchange to broker common purpose across all neurological conditions. More importantly, we do not define by the deficits a condition may impose on people's lives. We concentrate on the value of lived experience to professional knowledge and skill, the contribution to the evidence base and for people themselves to share and learn from each other. By supporting people to be more confident in knowledge sharing and cooperation across social divides, we sustain a well-informed neuro community to improve self-management skills, neuro-literacy and social capital. In addition, we reduce post-diagnosis isolation and fear.

Grounded in psychosocial principles of managing a life-long condition, impairment or disability, we foster collaborative working partnerships to respond, support and sustain a high customer service focus. We encourage creative problem-solving and efficiencies to reduce risk or crisis and preserve psychological well-being. We strive for excellence with agreed standards and outcomes brokered by the very people who need skilled, knowledgeable services.

Our Open Door series of booklets has been developed to acknowledge the challenges of living with a neurological condition that is misunderstood, subject to media-driven or political myths and is neither prevalent nor the focus of national publicity campaigns.

Friedreich's Ataxia is a rare, progressive genetic neurological disease that causes movement difficulties, loss of sensation and impaired speech. The disease is also known as spinocerebellar degeneration. The disease causes damage to parts of the brain and spinal cord which degenerate over time resulting in awkward, unsteady movements and impaired sensory functions. One third of people with the disease have problems with their heart.

The following stories are all real life narratives written by parents of or young people living with Friedreich's Ataxia. We have adopted fictional names to protect their identities.

NEURO KEY

January 2021

Amelia's Story

Hi, my name is Amelia and I am 18 years old. I got diagnosed with Friedreich's Ataxia on 6th November 2018, I was 16 years old.

Let me tell you about my road to diagnosis.

It all began when I was 11. I had just started secondary school (2013) which was already a really hard time as I was getting bullied, the typical kids saying rude things. When I was eating, food started getting stuck halfway down my food pipe, which I referred to as choking. Every time it happened I was panicking so much it wouldn't pass, so instead I threw up. After throwing up, I would be so scared of eating I wouldn't eat anything else.

I mentally couldn't cope with the information I found looking in to ataxia

My mum took me to my GP and they did a bunch of bloodwork and referred me to paediatrics at the hospital nearest me. I had always been slim and quite tiny. I always got sick throughout my life too. The paediatrician did bloodwork first and then some physical tests, such as testing my reflexes which I don't have. He then referred me for a barium bowel test. This test was negative. There were no lumps or anything during my scans to make them worried.

He came to the conclusion that I had an eating disorder and so I got referred to CAMHS to talk to someone to help me. When I first went, my mum and I fought with them that I didn't have an eating disorder and that there was something else going on. Eventually we gave in. To us they were the professionals. They said that because of the bullying it had caused ARFID (Avoidant/Restrictive Food Intake Disorder).

I attended CAMHS until I was 14. Over this period of time where my weight was going up and down, I became quite clumsy. I started to trip over my own feet and was unsteady. They blamed this on my low weight, telling me once I was a healthier weight, these issues would rectify themselves. By age 14, I moved to a new school and I was a healthy weight and the choking episodes still happened, but not as often as I had learned how to cope and manage.

He then asked, "What do you want to do?"

To which I replied, "I wanted to travel independently."

He followed by saying, "The independent part might be difficult but at least you don't want to be a dancer."

I stopped attending CAMHS at this point and just focused on school as I had started my first year of GCSEs in 2016. I went for an MRI in early 2017 but they found nothing unusual. So I continued on with school. In late 2017, my mum took me back to my GP as my walking was still getting worse and I had shaky hands. The second I walked down the corridor she knew there was something wrong so she referred me back to paediatrics.

I saw a new paediatrician in October 2017 and on my first appointment he thought that I may have stunted my growth during the weight loss so he wanted me to come back 6 months later as it may have corrected itself and I was referred to an ENT who found no issues. So, in April 2018, I went back to see him again. This time he got me to walk up and down the corridor. Instantly he asked me to come back into the room so that he could talk to my parents and me. He also tested my reflexes, telling my parents that he wants me to get an urgent MRI as I have an ataxic gait, while he goes and researches some things.

My parents and I spent time googling 'ataxic gait' which led us to find out what ataxia is. We were panicking as we had no idea what was to come. I went for the MRI a week before I began my final GCSEs. There was already so much stress around the exams to add on the health stress, it was a lot to deal with mentally. During my exams I had to go back for a second MRI as I had twitched during the first one.

I mentally couldn't cope with the information I found looking in to ataxia and the bad things that come with some of the types. The paediatrician then met with me and my parents the day before my final GCSE and my 16th birthday, which were on the same day. He got me to come into the hospital the following Monday for observations overnight and testing.

At first they just took bloods, checked my balance and everything else I was used to at this point. I then got told that I was going to have to get a lumbar puncture.

A lumbar puncture terrified me.

I got numbed up for this and while the numbing cream was beginning to work, they came back in to take more bloods. They came back right before the lumbar puncture and told me they were going to do it the next morning instead.

I then spent the night in the hospital and the following morning I had multiple students come and do their rounds, all pointing out this ataxic gait. My paediatrician then came to tell me and my parents that they were not going to do the lumbar puncture, instead a physiotherapist was going to come round to see me then I could leave. He told us some of the bloodwork was being sent for genetic testing and that it would take 3 months until I got the results. As I just turned 16, I was moved to an adult neurologist for my results.

There was zero support offered, no follow ups or counselling.

The physiotherapist came round and said "I am sorry that you are now disabled but there are some groups you can join, such as wheelchair basketball." She did a few tests before telling me that I need to use a walking stick, which I refused as any normal 16 year old would. So she gave me crutches and that's what I left with.

continued...

I spent my summer waiting for the appointment for my results. My appointment came in September when I had just begun my first year of A Levels. I went to the hospital with my mum expecting to finally have answers, he didn't have them. He told us he would have to take my bloods again as they weren't sent through to him. He did all the usual exams, walk in a straight line, reflexes, touch my finger touch your nose and he made me close my eyes as he moved my big toe up or down and I had to say what direction it went. Nobody had ever done this test, but I thought it was easy. When we left after being told that it would be 3 months again, my mum informed me I got almost all of the directions wrong.

He then asked, "What do you want to do?"

To which I replied, "I wanted to travel independently."

He followed by saying, "The independent part might be difficult but at least you don't want to be a dancer."

My mum got a call 3 weeks later to say that my results were there.

This was it, 6th November 2018.

As my parents and I walked into the room, the neurologist told them that he wasn't there to talk to them as I am the patient. What came next was a complete shock. He said, "As I suspected you have Friedreich's Ataxia (FA). There are some heart conditions that are fatal, but we will keep an eye to your heart and hope you aren't one of them. There is no cure or treatment. Do you have any questions?" I was silent.

He then asked, "What do you want to do?"

To which I replied, "I wanted to travel independently."

He followed by saying, "The independent part might be difficult but at least you don't want to be a dancer."

I stayed silent. My mum was crying and my dad was angry. They asked for support groups to which the neurologist told them that there aren't any but we could find some ourselves on Facebook. That was it. We left.

I do wish there was a way to explain it easier to friends and family as they don't understand it at all.

We drove home in complete shock of how he acted. I shutdown completely, my mum told my nana and grandad while I sat there, numb.

There was zero support offered, no follow ups or counselling. I stopped going to school for months as mentally, the diagnosis affected me hugely. My parents and I went to a genetic specialist just to figure out what side of the family it came from so that others in the family could get tested.

As a way to tell the most of my friend group and peers, I posted a Facebook status a few days after being diagnosed. By then, my close family and friends knew as much as I did but I do wish there was a way to explain it easier to friends and family as they don't understand it at all. Teachers were the worst, some of them googled for information and others didn't so they were only going of what the neurologist sent. My school weren't personally helpful in offering support and understanding so I want to make sure there is as much support for people newly diagnosed in the future. If the school is not supportive, it can be difficult.

The neurologist referred us to the genetic specialist as he made it very clear that it could run in the genetics of my family.

This was terrifying for my parents as they already have me to deal with, but then to have my two younger sisters on top of it. Luckily, they're both okay.

I am now with a new doctor at a different hospital who has been amazing and I am so thankful to have him. Also, I am studying Law and Criminology with the Open Uni. I was determined on learning how to drive and I passed my test in March 2020. Eventually, I stopped being stubborn and got a walking frame in 2019, which has been the best decision I ever made.

In our house I walk around with no walking aids as our home is very small but hopefully in time we can get into a new home that will be wheelchair accessible. I have my own wheelchair which I use part-time. I can now manage the choking episodes and I have seen a speech and language therapist.

Mentally, I was referred for counselling right before Covid-19 so it is a long process which I am still going through. Every day is different, sometimes when I'm having a bad day I get very frustrated and I lose any motivation. However, with something like FA we are entitled to bad days, as long as we remember not every day is going to be bad.

> There was zero support offered, no follow ups or counselling. I stopped going to school for months as mentally, the diagnosis affected me hugely.

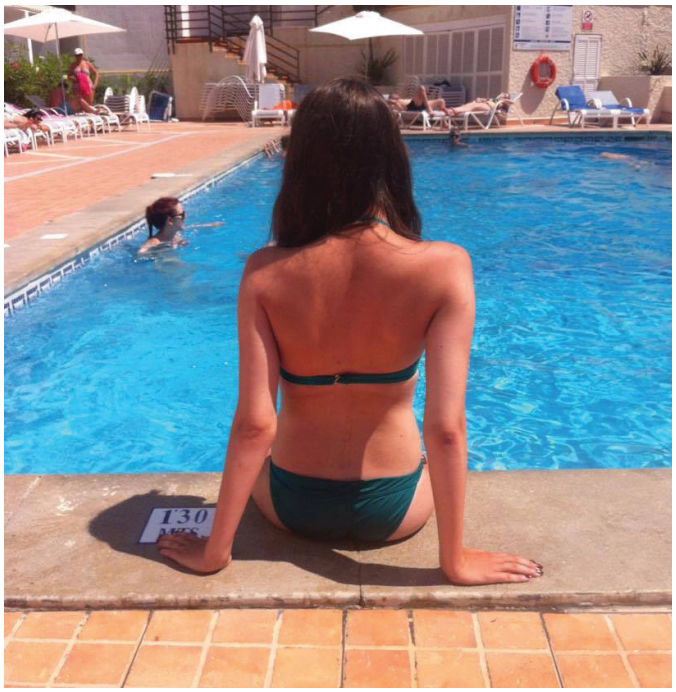
I wish healthcare professionals dealing with the diagnosis process could offer sympathy, ie simply saying you are sorry. Sympathy can go a long way to some people.

After my diagnosis, I joined some support groups on facebook and from there, saw posts sharing Ataxia UK. They have been very helpful and I am actually subscribed to their magazine. They are constantly raising awareness and right now my family are raising money for Ataxia UK.

The main goal is to raise awareness for FA. The more professionals that fully understand it, the better they may be able to address a patient.

> The main goal is to raise awareness for FA. The more professionals that fully understand it, the better they may be able to address a patient.

Lucy's Story



Our story began when Lucy was just 22 months old, and I thought there was something odd about the way she ran. I mentioned this to our Health Visitor who dismissed me as a hysterical mother. A GP took a similar attitude when we took Lucy to the surgery at about 3yrs, temporarily unable to walk. He claimed that 2 spoonsful of Calpol would rectify the problem, and believed that, as the youngest and very cute child in the family she was having us all on such as a pair of dressing up shoes had temporarily numbed her feet!

During the reception year at school Lucy's teacher had noticed how clumsy she was. Something we had also noticed in the ballet class. We were sending Lucy to ballet as she had profoundly flat feet, discovered by the man in the shoe shop, in the hope it would strengthen her muscles. She would always hold on to things as she crossed the class room and at home, we had noticed her reluctance to sit on a bicycle with stabilisers.

She felt she was falling off. She also always sat down to dress herself. At school, Lucy was tested for dyspraxia but as she could hold 12 commands in her head at one time dyspraxia was ruled out.

By this time the extremely flat feet were beginning to cause problems. We could not get shoes to fit comfortably and Lucy was eventually referred to a paediatric orthopaedic consultant, who suggested exercises for the feet and inserts to be made for her shoes. We mentioned her clumsiness and the consultant called the physiotherapist who, on watching Lucy walk, decided that she was hypermobile and would benefit from regular physio sessions. By this time, Lucy had started suffering from UTIs on a regular basis and was referred to a paediatrician at our local hospital. At the same time, her Year 2 teacher had noticed that she found handwriting particularly arduous and went to some lengths to get her referred for some Occupational Therapy, the regular physiotherapy sessions had stopped. The thinking was that her hypermobility was causing more problems than just the walking. In September 2007, Lucy saw the paediatrician with regard to the UTI problem. I seized the opportunity to tell her everything about my daughter and voiced my concerns about all these problems she seemed to be having. She listened politely, whilst looking at the clock all the time. Whilst she found 'my story' very interesting, she could only deal with the UTI/kidney issue at this appointment and referred Lucy for a kidney x-ray (with the dye etc).

> At school, Lucy was tested for dyspraxia

In October 2007, Lucy had an appointment with an Occupational Therapist. After 2 appointments and more than four hours of testing she decided that Lucy had an underlying condition that she could not determine. Lucy had ticked the boxes for so many different things up to a point, but at the final hurdle it would all fall to pieces. She said that she would make an appointment for her to see the same paediatrician as in September. Finally, I thought, someone was taking this seriously.

At the beginning of January 2008, still waiting to hear about a 2nd appointment with the paediatrician, I took Lucy and her

friend swimming. As she walked ahead of me I could see a very obvious spinal scoliosis which I somehow had never seen before. We went to the GP that evening who was baffled. I asked if it could be something to do with her hypermobility. To his anger and disbelief he said he knew nothing about this and for some reason had not been included in the loop from the hospital. He also retrieved a letter from the paediatrician at the hospital who, following the appointment we had in September, said she had given Lucy a thorough examination whilst at the appointment and all things, including her spine, appeared normal – hence she was referring her for the kidney x-ray. He faxed the paediatrician there and then and said he wanted her seen within 5 working days. She was.

...the regular physiotherapy sessions had stopped.

She apologised for not looking at Lucy's entire spine, as clearly a scoliosis of this degree would have been obvious in the previous September. At this point, she did 3 tests with Lucy – the usual ones fingertip to nose co-ordination, the reflex hammer tests and finally she asked Lucy to stand up and close her eyes – she started to fall backwards. At this point she asked Lucy if she would like to go and play with the toys whilst she spoke to us, her parents. She said she believed that Lucy was suffering from an ataxic condition that she could not diagnose. We had never heard of ataxia and she did very little to explain, other than it was something that affected the central nervous system and that there were many different ataxias, some of which were progressive/degenerative and some life shortening.

This information was only given prompted by questions from me. She said she would refer Lucy to a neurologist at the John Radcliffe Hospital in Oxford and that an appointment would be given fairly imminently, a matter of days. We left the hospital in a complete daze.

However, nothing was to go smoothly.

Six weeks later we were telephoned and asked to come to see the neurologist, this time without Lucy.

By the end of January we had still heard nothing from Oxford. I rang the consultant's secretary who informed me they had had no referral for Lucy. She took control of the situation, rang Reading where sadly the letter was still sat awaiting postage! Within 1.5hrs we had our appointment….for the beginning of March. The neurologist wanted several tests in Reading before our appointment – a brain MRI, a spinal MRI, an ophthalmology appointment and numerous blood tests. When we went to Oxford we had a 2 hour appointment. All the test results had been looked at, deemed normal and although there was evidence of thinning of the spinal cord covering it was still within the parameters of normality. He told us that in his opinion she was not ataxic (we did clarify this with him before we left the appointment) but had a peripheral neuropathy, fewer nerve endings than is normal particularly in her hands and feet and lower legs but it would not progress. Our daughter may have some trouble with co-ordination as she physically got bigger. She was tested by a neuro-physiologist who confirmed this. We were on cloud 9! He did however take extensive blood samples that day, just to rule out various other things.

Six weeks later we were telephoned and asked to come to see the neurologist, this time without Lucy. He said that the results of the blood tests showed that she had Friedreich's Ataxia, a progressive degenerative ataxia for which there was no cure, and advised me not to look on the internet but gave me some sheets he had printed out and said that the charity Ataxia UK could be very helpful. Lucy would continue to see him on a yearly basis.

continued...

> In about half an hour, the doctor who had given us such hope 6 weeks previously had completely shattered us.

The referral to the spinal clinic would obviously go ahead although she would not now be referred to the gait clinic as the diagnosis now explained her gait. He also said that he would need to refer her to a cardiologist as Friedreich's patients could be affected by hypertrophic cardiomyopathy, explaining that this was a serious heart condition, but not all patients' hearts were affected.

The diagnosis was not explained well to us both. In about half an hour, the doctor who had given us such hope 6 weeks previously had completely shattered us. He was very quiet and subdued, probably because he knew he'd made a big mistake. He told us not to look it up on the internet and did not give us enough information.

In June of that year, we went to see a geneticist who was amazing in comparison. She made us feel like we were the only patients she had, which is how it should be done.

As Lucy was only young, I understand why they asked for her not to be present when they told us the diagnosis but more help telling her would have been welcome. We were told to not sit her down and tell her, so instead we just answered questions she had whenever she had them. With hindsight, we should have told Lucy more because she forgot about asking these questions and grew up believing she'd found out by herself and we had never been honest.

Further appointments over the next 6-8 weeks showed that Lucy did indeed have the heart condition and the geneticist told us that, following DNA testing, we both carry the FA mutation. At some point in time, our other 3 children, half siblings to Lucy would need to be tested.

After a complete roller-coaster ride to diagnosis I have to say our treatment as a family, by the team at Oxford, was second to none, once we knew what we were dealing with. There was no help offered to telling our daughter's friends or teachers and after listening to other people's stories I think this would have been helpful.

In my opinion lack of any kind of family counselling or ongoing physiotherapy for Lucy are huge holes that I personally would like to see addressed.

> There was no help offered to telling our daughter's friends or teachers

George's Story

As a child I was fairly 'normal'. I was constantly playing or watching sport.

As a child I was fairly 'normal'. I was constantly playing or watching sport. There was nothing more important in my life than running around a field kicking, throwing, bowling or hitting a ball. Thinking back, the only slight anomaly was my reticence to riding a bike (without stabilizers). I remember seeing friends do it effortlessly, standing up on their pedals and doing wheelies, and being a bit confused why I struggled to stay on two wheels for more than twenty seconds. Something was slightly off with my balance, but I shrugged this off as a minor thing. Not everyone loves to ride a bike, right?

This initial nonchalant attitude was probably caused by the lack of similar visible stories, among people I knew or in the mainstream media. I vividly remember being upset one day after I'd walked home from school - in zigzags - and googling my wobbly symptoms. Maybe it was just my shoes? Or something I'd eaten? Google didn't show much, obviously wasn't as knowledgeable in 2007! There was definitely no sign on there of the neurological condition I went on to be diagnosed with – Friedreich's Ataxia.

I continued to struggle with my balance and coordination through the first few years of senior school – often avoiding certain busy staircases or corridors and turning up to lessons late as a result. I came within seconds of missing an exam because I just couldn't make my body run to the exam hall, with the teacher imploring me to do so as he was about to close the doors. The thing that really tipped me over the edge though was my cricket performances.

I continued to struggle with my balance and coordination through the first few years of senior school

continued...

GPs have little awareness, if any, of the condition.

After developing as a spin bowler for a couple of years, by 2009 it was hard work to even bowl the ball in the vicinity of the batsman. The coaches didn't know whether to laugh or shout, and neither did I. I walked off the pitch in the middle of my over once because I just could not bowl properly, overcome with embarrassment and confusion.

Thinking back, this was such a devastating time for me. Something definitely wasn't right and it was time to find out what it was.

In October 2009, I went to my GP with my equally worried Mum. After explaining my symptoms to him, he was visibly concerned, but also confused. He guided me through some exercises so he could see my wobbliness for himself. I particularly remember us laughing because I couldn't hop. I think the laughter was to conceal our unease. He clearly thought there could be something malignant going on.

My Mum remembers him quietly mentioning to her about suspicion of a brain tumour, but more tests would be needed – at this stage that was just conjecture. Given this and the fact I was just 15 years old, I understand why this wasn't mentioned to me. Also, because of its rarity and the subsequent dearth in knowledge of it, Friedreich's Ataxia was not brought up.

I've since been told by fellow Ataxians how it's often been a similar experience for them – GPs have little awareness, if any, of the condition. I think this is getting better but could definitely still improve to make detection and diagnosis that bit smoother, and ease the strain of patients and their families in an incredibly stressful time.

The next stage of my diagnosis was quite fast and well managed, especially compared to other people I've heard from since. This was probably due to me living in Liverpool, home of one of the best children's hospitals in the UK – Alder Hey. As I was just 15 at the time, this is where I went. I can't recall the precise details of each, but I had a few tests and scans over a couple of visits. Mum heard whispers from the neurologist of some 'unusual activity' and a mention of a word that sounded like 'taxi'. When we got home, she immediately trawled the internet with this clue in mind, paired with my symptoms. Amazingly, Mum found a condition called Friedreich's Ataxia. She was almost certain that was what I had. Confirmation of those fears didn't take long, as a short time later - December 2009 – my mum and stepdad were called into Alder Hey to be given my diagnosis by a neurosurgeon.

It was decided between Mum and Alder Hey that I wouldn't go in for this initial appointment to be told the news at the same time.

It was decided between Mum and Alder Hey that I wouldn't go in for this initial appointment to be told the news at the same time. I completely understand this. Mum wanted time to process what could be a crushing revelation, to be as strong as she could for me when I was told. February 2010, during my half-term break from school, was pinpointed as a good time for me to get the news – if there is ever a good time to be told you have a degenerative, neurological condition!

I had a label for it; something to understand, something to fight against.

Alder Hey decided it would make more sense for a neurologist to break the diagnosis to me, rather than a neurosurgeon. The neurologist apparently had more rounded knowledge of neurological conditions, although he admitted I

They did direct us to the Ataxia UK website

was his first Friedreich's Ataxia patient. On the day there were around seven or eight hospital staff in the room, along with Mum and myself. I knew it was something serious because of this. I think as I was given my diagnosis Mum was more tearful than I was. Maybe it took a while to sink in and I was a bit naïve and almost disbelieving about what it would entail. Although for some reason, there was an underlying calm emotion in me.

It's strange. For a couple of years leading up to that day, not knowing what was 'wrong', my head was all over the place. But when I got the news, while the room was tearful and sombre, relief took over for me. Finally I had an explanation for losing the ability to do things. It wasn't just me being stupid. I had a label for it; something to understand, something to fight against.

As I said, there were quite a few different hospital specialists present for my diagnosis. I think this helped. It was reassuring to know I had experts of different fields in my corner. They each introduced themselves and told me how they could connect with my neurologist to support me. I can't remember exactly (it was all a bit of a blur!) but they included an occupational therapist, a couple of physios, a speech and language therapist and an emotional support therapist. They too, like the neurologist, had never had an FA patient but they were definitely all aware of the magnitude of the condition.

There wasn't a leaflet or any written information on the condition given to us during the diagnosis – probably due to the rarity and lack of nationwide knowledge of FA – although maybe there was more to it than this; it was possibly a conscious decision to not overwhelm me with negative details.

Mum was already quite clued up on the condition, so a leaflet showing symptoms and how my life might look in the future would have been unnecessarily frightening for me at the time. They did direct us to the Ataxia UK website, for us to get information when we felt ready. Ataxia UK is the national charity for my condition. I didn't really want to know anymore at the time, so it took me a while to go on this. Mum had similar thoughts but did find it really helpful in the end and was grateful and relieved to see relatable stories and collect general advice on FA.

The support given to me and my close family after my diagnosis was very thorough. We were constantly in touch or had in-person appointments with the emotional support therapist. I think she also organised counselling for us through the hospital, for Mum and myself to attend either together or separately.

The emotional support therapist had a meeting with my mum, my friends and head of year in school, to talk about my condition.

As a student of psychology and counselling now, I'm extremely thankful for this support. When you're affected by a health issue, your mental wellbeing is vital, for you and also those around you. I'm probably more thankful for these interventions now than I was at the time.

Another really good aspect of the support was offered by the genetic specialists. Mum had a few appointments with them and was tested for the Friedreich's gene just prior to me being given my diagnosis, to be absolutely sure. My dad was also tested, as were my brothers.

This was important for their peace of mind as they had plans to start their own families.

continued...

The one aspect I think my diagnosis could have improved was more knowledge at GP level of Friedreich's Ataxia. Obviously it wouldn't be fair to blame the GP I saw individually – I think it is a systemic problem.

After figuring out the best way to tell my close family and me about my diagnosis, telling my friends and teachers, people I saw everyday was the next step. I think this is really important, as some people become isolated when they receive a diagnosis. But, this shouldn't be the case. I think the key is to be open and honest as early as possible, despite this maybe not being easy, to create an understanding and a bubble of support. This way you don't feel the need to hide things and it's easier to own the disability, rather than letting it control you.

Again, Alder Hey was great with this. The emotional support therapist had a meeting with my mum, my friends and head of year in school, to talk about my condition. They just went through the outline of it really; the main symptoms and what to expect. This meant everyone around me was suddenly a bit less curious and more empathic. School was definitely more bearable after this! Having that intermediary specialist voice between me and my friends was so helpful. It took the pressure off, having to tell people would have been difficult. I can't stress how vital this aspect of diagnosis is.

On the whole, I couldn't complain at all with how Alder Hey handled my diagnosis. They were very organised and professional. It can't be easy to break news like this to a 15-year-old, but they with so handled it professionally and offered so much support, significantly psychologically which I now realise was vital for me and those around me. Informing Mum of the diagnosis before me was wise I think, for reasons I mentioned earlier. This is an important point for any patients at a similar age or any younger than I was, in my opinion.

The one aspect I think my diagnosis could have improved was more knowledge at GP level of Friedreich's Ataxia. Obviously it wouldn't be fair to blame the GP I saw individually – I think it is a systemic problem. Or it was, hopefully now there is more knowledge of rare neurological conditions among GPs. I think this would make diagnosis that bit more efficient, and significantly less of a protracted emotional toll on patients.

Susan's Story

...she wanted high heeled shoes for parties, she's a pocket rocket at five feet tall, but she couldn't get any to fit. I promised her we would ask for a podiatrist referral for her high arches and maybe an insole would do the trick.

Everything in our lives, pre-diagnosis, was ordinary.

A small family unit, two working parents and a much longed for fifteen-year-old daughter. I was career driven and had high expectations for my daughter and she, for her part, complied. She was academic but also loved her theatre group. She had been a horse rider, a gymnast and a street dancer whilst growing up and by her 15th Birthday enjoyed a busy social life. I took pleasure in how independent and self-sufficient she had become.

I worked long hours and also cared for my mother who was exhibiting early signs of dementia. On reflection, I can remember that it was just as well that my daughter needed me less. My life was so busy and she was doing just fine navigating her way through her teens. She asked so little of me, when she complained a few times that she wanted high heeled shoes for parties, she's a pocket rocket at five feet tall, but she couldn't get any to fit. I promised her we would ask for a podiatrist referral for her high arches and maybe an insole would do the trick.

A few weeks later, I took a morning off work and we visited our local NHS Podiatrist. A lady who was lovely and very sympathetic to the shoe story agreed that she could make some insoles to compensate for my daughter's high arches. Before we left, the lady asked my daughter to walk so that she could check her feet from behind.

She turned to me and asked if I was concerned that her balance was a bit off? I wasn't and didn't answer. She then asked if she could refer my daughter to the Genetics Clinic in Newcastle. I agreed and she said she would be in touch when the insoles were ready.

We returned home and I watched my girl walk down the road to school from an upstairs window. She typically weaved almost in a zigzag along the pavement rather than in a straight line and what I previously thought of as her walk, potentially had another explanation.

continued...

Our family history had no sinister diseases, other than cancers and dementia in later years. I naively thought that if she had any major problems, we would have been aware of it long before she turned fifteen. I returned to work, not giving much thought to the appointment at the Genetics Clinic. After all, she definitely wasn't ill!

The appointment came through a few weeks later and we all attended the Genetics Clinic in Newcastle. My daughter, as independent as ever, went into her appointment alone. Her dad and I stayed in the waiting room. I remember seeing and reading a notice board regarding help and support groups for muscular dystrophy. When my daughter's appointment had finished, the doctor invited us all into the room. He asked if he could take some blood tests from all of us in order to rule out various conditions. Of course, we agreed. He explained he would probably check on my daughter in six months' time. His parting question (as we were leaving) was "Are you aware that she has scoliosis?" We weren't.

"Are you aware that she has scoliosis?"

We returned home and picked up our lives. Obviously, I researched scoliosis and was horrified that in extreme cases, it could result in the use of steel rods to correct the curvature but reasoned that hers could not be that bad as the doctor had not suggested any follow up.

A week later, I was working away for work and returned home quite late. In the post was a letter from the doctor at the Genetics Clinic. It informed us that there was a new appointment two days later for my daughter. It would be at our local hospital and the doctor would be attending with members of his team. It was too late to contact the clinic to get any further information. I felt uneasy and unsettled. I Googled muscular dystrophy and learned that there were many different types of which some I couldn't even pronounce. Some had links to YouTube clips of patients seemed impossible even to consider and bore no resemblance to our beautiful baby girl.

Predictably, the next two days passed very slowly. My daughter was unconcerned and a bit put out that she would miss a geography field trip at school. The date of the appointment was the 28th March 2012 at 2pm. It was then that our lives changed forever.

It was then that our lives changed forever.

We were invited into the doctor's room and I noticed that there were a couple of nurses present. We sat down with my daughter sitting closest to the doctor. He told us that they had discovered that she had a rare genetic condition with a funny name. Her dad laughed a little and I waited. He told her that the condition she had was called Friedreich's Ataxia, named after the scientist who discovered the gene. She stared at him blankly and we listened. I only remember some phrases – probably need a wheelchair - heart disease – slurred speech. I was struggling to breathe and remembering the YouTube clips. I wanted to scream but knew that would be the wrong reaction. I concentrated really hard on not making any noise. She turned to me and said "Mum. Why are you crying? It will be ok". I didn't realise that my face was wet. A nurse leant over and touched my hand and said, "There are benefits". I thought the woman was clearly mad! How could any of this be of any benefit? I asked her what she meant. She told me that they could arrange handrails in our home.

The doctor gave my daughter a leaflet to explain her condition and said that a nurse would visit us at home in a few days. The leaflet explained basic facts regarding Friedreich's Ataxia. It spoke of needing a wheelchair and early mortality, it gave none of us any comfort and actually I think my daughter put it in the bin when we got home.

We left. Our small family unit fractured, broken. It had only taken thirty minutes to destroy our world. Over the following months, we were sucked into a vortex of clinical appointments – Neurology, Orthopaedic, Cardio, Physio, Orthotics. It just

went on and on. Of course, I was able to carry out extensive research and learn about the condition.

I eventually came across Ataxia UK and FAPG (Friedrichs Ataxia Parents Group). I was off work for six months. I think on reflection, the manner in which we received the diagnosis resulted in a kind of PTSD, probably for all three of us. Certainly, it took many months before I lost the feeling of having a heavy bar sitting on my chest. Normal conversation and activities felt wrong and pointless and during this awful time, having to keep up a positive front to reassure the person it affected most.

We were sucked into a vortex of clinical appointments – Neurology, Orthopaedic, Cardio, Physio, Orthotics.

I have spoken to other parents over the years regarding their diagnosis experience. Some of them were asked to attend appointments prior to attending with their child. I think had this happened, although still devastating it would have given me the opportunity to be better prepared and stronger for my daughter. Of course the outcome remains the same but I feel on reflection as a family we needed time to process the information individually.

I feel on reflection as a family we needed time to process the information individually.

I think a factsheet signposting to Ataxia UK and other forums should be given to all of newly diagnosed families. The feeling of loneliness and isolation and generally being "apart" from the life you knew before is something that will stay with me for a long time.

Since that time eight years ago, I always try to reach out to any newly diagnosed parents. It's difficult with a prevalence of only 1 in every 50,000 so inevitably, no other FA families live close, but I cannot bear the thought that other families have to endure what we experienced.

Of course, no diagnosis of such a serious nature can be welcomed but I think clinicians need to establish a protocol and be mindful of the patient journey which has brought them to diagnosis. We were asked to attend an appointment with a specialist nurse at the school, without my daughter. The purpose was to explain to the teachers her diagnosis and provide them with information they may need whilst she was in their care. This was arranged within days of our diagnosis appointment. I think in principle this was helpful to the school but at that point I was really struggling to engage with anyone.

My only memories were of resentment that other people were discussing her and how best to help her. I just wasn't ready for practical solutions.

The services we needed for my daughter were immediately available – Cardio, Physio etc but there's one question we have never been asked...

"How are you feeling?"

Learning outcomes

	Service delivery	Relevant for
1	Present information to the person and parents mindful of the age of the young person and tailor language to be empathetic. Is information too technical or based on medical jargon? Is information age appropriate, written or digital, accessible or have implications of morbidity? A leaflet or website?	Genetic services Neurologists Specialist nurses Primary care - GP's and staff Occupational Therapy Orthopaedics Cardiology
2	Ensure consistency across services. Is there a protocol for informing children under 16years after the parents? Is there a follow-up appointment for the family to fully digest the diagnosis? Is genetic counselling automatically offered?	Genetic services Neurologists Specialist nurses Primary care - GP's and staff Physiotherapy Orthopaedics Cardiology
3	Identify the support needs of parents, communication or management difficulties. Is there signposting to local support systems?	Genetic services Neurologists Mental Health teams Specialist nurses Primary care - GP's and staff Voluntary Sector
4	Configure a multi-disciplinary, co-produced care plan to include access to early and regular psychological support.	Genetic services Primary care organisations Health psychologists Mental Health teams Physiotherapy Speech Therapy Orthotics Orthopaedics Cardiology
5	Initiate protocols for awareness training.	Primary care - GP's and staff Schools, colleges and Universities Voluntary Sector
6	Establish a review to take into account transitional needs of children to teens and teens to young adults, particularly for equity of access to education, adaptations or equipment needs.	Primary care Social Care Occupational Therapy Mental Health teams

From these narratives, the UK does not have a standard process for diagnosing people with rare neurological conditions like Friedreich's Ataxia. The way we are introduced to any life-long illness can have an impact on how we psychologically adapt, self-manage or direct our care when we deteriorate. Areas to improve patient outcomes have been identified.

What can we learn from these narratives?

How can services respond more appropriately?

Are the needs of significant others taken into account?

Acknowledgements

We are indebted to our volunteer research assistant, Georgia Hart for her drive, study and coordination of this resource to contribute to a much-needed evidence base. This booklet would not have been possible without Georgia's consideration and care. Sadly, one of these narratives was written by the parent carer after their daughter passed away. Parents never lose sight of something better for their children.

We hope these narratives can provide a legacy to inspire appropriate care pathways that support young people to achieve their goals and bring peace of mind to parents, families and the wider community.

Neuro Key
St Mary's Centre
Middlesbrough, TS1 2RW
T: 01642 641825
E: admin@na-tvdny.org.uk
W: www.na-tvdny.org.uk

Ataxia UK
12 Broadbent Close
London, N6 5JW
Office: 020 7582 1444
Helpline: 0800 995 6037
Office: office@ataxia.org.uk
Helpline: help@ataxia.org.uk

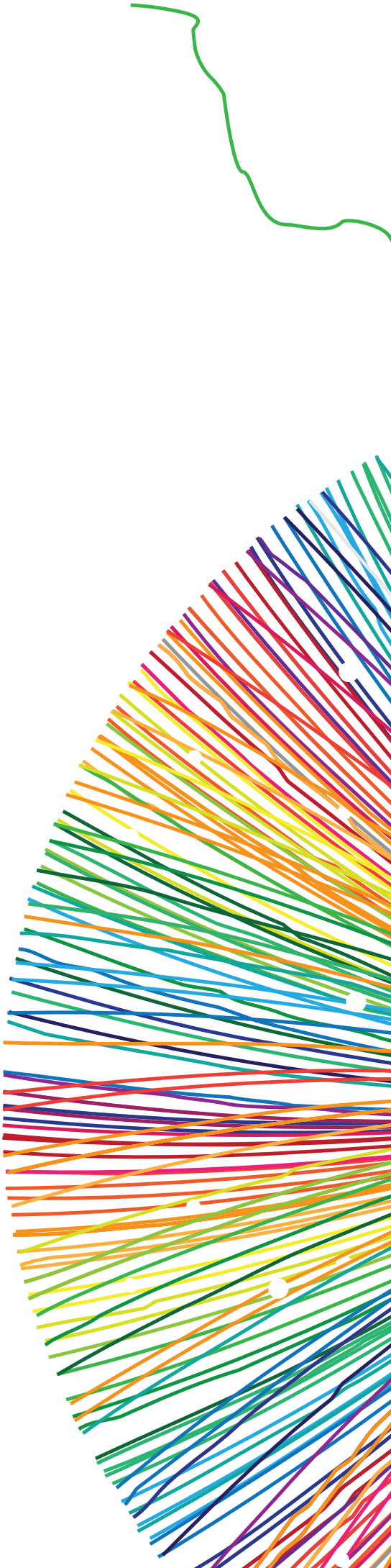
Notes

NEURO KEY

An Alliance supporting people with neurological conditions

Our Open Door series is collectively created to share our expertise and ultimately, benefit the whole neuro community. However, we need to sustain this Charity. If this resource has been useful for you, we would appreciate a donation to help keep the work going.

Scan the QR code with a smartphone and go to the donation page on our website.



Tees Valley Durham and North Yorkshire Neurological Alliance
Registered Charity No: 1119043

Design by www.nickballdesign.co.uk