



HSP Newslink

The Newsletter of the Hereditary Spastic Paraplegia Support Group

Issue 55 – August 2023

HSP & FSP – Hereditary Spastic Paraplegia,
Familial Spastic Paraplegia & Strumpell-Lorrain Syndrome

Power to the Powerchair



I would like to share my journey with you following the purchase of a PowerChair, in the spring. It has completely changed my life. The onset of my condition was 27 years ago; my walking has progressively deteriorated and I've been using two sticks now for a couple of years. The restrictions outside the home had become arduous to say the least. All outdoor activity required strenuous effort, which was difficult

and tiring. My days out with friends were shortened, as my slow speed could have a detrimental impact on the pace of the day. I suspect there were occasions when I wasn't necessarily considered for some days out. This no longer need be the case, as I am now capable of moving along at some pace. I really appreciate being in this better position.

I did a lot of research before making my choice of aid. I often attend HSP monthly zoom meetings, which I find invaluable and extremely informative. We share our challenging experiences and offer advice, which can be healthful and beneficial. I asked the participants on the zoom meeting their thoughts on PowerChair V Mobility Scooter. I was most grateful of their experience and profound knowledge. I decided the PowerChair was the correct aid for me.

It was essential the chair wouldn't be puzzling to adjust, as I have poor balance. I chose one which can fold without dismantling fixtures. I purchased a bag which fits over the back of the chair; this has two pockets which house my sticks. The bag is big enough to hold many things when I go shopping and to the gym. It also has a compartment for a packed lunch and drinks. In addition, I changed my car to an SUV which meant I could have an automatic hoist fitted. This is a fabulous apparatus; I hook two straps on either side of the chair and it is then transported safely into the boot.

Acquiring this has been life-changing. My quality of life has increased enormously. Galleries are accessible to me for the first time in years. I can enjoy exhibits I find of interest at my leisure. I now delight in my days out. Trips to the theatre have become a joy as I'm no longer struggling to get around.

I am a keen shopper; one of the most enjoyable aspects of using my PowerChair is having freedom to browse through the shops at my pace, which had become impossible for me. Now I am looking forward to attending The Edinburgh Tattoo in August and a holiday to Italy in September.

I chose a PowerChair, which I can also use as a rollator. I believe it is extremely important to exercise and whilst weight bearing exercise maintains muscle strength and growth, cardiovascular fitness is equally important. I go out walking short distances with my chair and when I get tired, I simply sit on the chair and use the power.

Be The Change That You Wish To See In The World

Jacqueline McLaughlin

Chairman's Column

Hello to all our members and others reading. I'm writing this column during the school summer holidays after our AGM and the first two presentations. This is the first newslink of 2023, so there have been quite a few different events since I last wrote. We had our group social at Tally Ho! in October 2022, which was good and allowed people to meet and chat with each other informally. On a personal note I have recently started taking a small daily dose of Baclofen.

AGM and trustee update

We held our AGM virtually again this year, and asked members their views on this with the voting. There was a majority of people seeking virtual over face-to-face, and whilst I like the idea of hybrid we would need to find an economical way of doing this. I have written summaries of the first two presentations we had after the AGM which are elsewhere in this newsletter. You can watch the AGM and other videos on our YouTube channel. <https://www.youtube.com/channel/ukhspsupportgroup>.

To remind you of our trustees following the AGM. John Mason stood down, and Shanake Amarasinghe was elected, taking over the treasurer role from John. Juan Vazquez Ruiz was also elected for the first time. Mitesh and I were re-elected and there are no other changes, giving us seven trustees this year. We welcome Shanake and Juan into the group. We have agreed to schedule our trustee meetings this year, and they will be on 20th September, 15th November, 17th January, 20th March, 15th May, with the 2024 AGM on Saturday 13th July.

Welcome to new members

I am always pleased to hear about new members joining us. I am happy to welcome them to our friendly supportive community, hoping they don't feel quite as lonely or isolated as they did before finding us. Remember that the key elements of our community are this newsletter, our meetings and our website.

Support our fundraisers

Several of our members or their friends and families are taking part in different fund raising and awareness activities in the next few months. If you're able to make it to Ian's potato pants festival, you should! <http://potatopantsfestival.co.uk/>. Also, please let people know about these activities as more sponsorship is always welcome: Emma is dog sledding in Sweden - <https://www.justgiving.com/fundraising/emmasarcticadventure>, Jason and Gavin are trekking to Everest Base Camp - <https://www.justgiving.com/page/jasonmain>, and Tom is running in his Viking gear again - <https://www.justgiving.com/page/tomvikingmarathon>. Remember to let us know about any other activities so we can publicise them.

Looking for help

As you know we are a small charity, and we rely on people giving us their time to achieve all of our work. Our website has a page about how people with or without a connection to HSP could help the group: <https://hspgroup.org/want-to-be-able-to-help-the-group/>. I am keen to hear from anyone, so do drop me a line.

My HSP survey

As many know, I run an annual survey on my HSP blog and interested readers can keep an eye out on my blog for my 11th survey which I hope to launch in October, once I've got my questions finalised and translated into six other languages! <https://hspjourney.blogspot.com/>. My 11th Survey will include questions on symptoms, and I am interested in answers from anyone who answered my 1st survey in 2013 or my 6th survey in 2018, so I can see how symptoms have changed over five or ten years.

Adam Lawrence

Editor's Column

It's been a long wait for this edition of Newslink and I apologise for that. I'll try to get another issue distributed this year, but I can only produce one if I have some interesting content, so please keep the articles coming in. I'd particularly like to hear from members who've not contributed an article before. Thank you to those of you who've submitted content for this edition.

Since the last edition I have had the delight of another addition to the family, on July 17th my third grandchild was born. I now have two granddaughters and the latest, Elle, was the best 60th birthday present I could have had. Two days after she was born, I turned 60 and thanks to all members who sent birthday wishes.

Since the last issue, the most exciting change of my circumstances is that I'm now a member of Poole Athletics club. This enables me to enjoy my latest hobby which is

wheelchair racing. Look out David Weir, here I come. I train every Sunday and currently do about 10,000m around a local athletics track. I'm hoping to enter a 5km race in October and the long term ambition is to do a marathon.

The Potato Pants Festival is fast approaching, in fact it's touch and go as to whether I'll finish this Newsletter before the festival takes place on August 26th, but I'm trying hard to achieve this. Final preparations are taking up a lot of my time at the moment, but all seems to be falling into place. I have 11 musical acts ranging from solo artists to a six man headline band, performing on two stages. There'll be the usual Potato Pants races providing loads of fun, but also giving people an awareness of what it may be like to have HSP. There will be some stalls and as always, food and drink will be available throughout the day. There will be a raffle and tombola, a playground for the children and the site is very accessible with accessible toilets. There is free on site parking and disabled parking is close to the stages. Mobility scooters will be available to use

during the day if required. Let's hope the weather improves and August ends with a mini heatwave. There are plenty of tickets still available either on the gate or by using the following QR code:



I was very honoured to be awarded with the David Pearce award for raising awareness of HSP at our AGM. I believe that this was mainly because of my involvement with the Potato Pants Music Festival. There certainly aren't many people in my neck of the woods who haven't heard of HSP on account of the festival.

I look forward to seeing some of you at future events, and remember, please make my job easier and keep those articles coming in.

Ian Bennett

Trampers

what a great and inexpensive treat!

www.countrysidemobility.org

Hopefully the picture shown can say so much more than I can articulate here – so rather than try to do that I'll give a few notes to outline 'what they are, plus where and how to hire them' – there are many choices.

What are they? – a very sturdy motorised mobility scooter, capable of considerable off-road mobility, with a powerful battery providing good range too. They can take you over terrain that would be too much for either your own wheelchair, or powered attachment.

How difficult are they to control? – not difficult at all. Before your first outing, you will be given an easy-to-understand explanation of the controls, and asked to give a brief demonstration that you have understood / have appropriate control. For rentals after that, you will be asked to do the brief demonstration before setting out.

Do you have to be a member? – at Countryside Mobility you can take out either a temporary membership, or sign up for a full

year (easily renewable thereafter). A look at the website is the best place for you to see / understand these and the differences between the two. I initially took out a temporary membership, but now simply renew the inexpensive full membership each year.

Another benefit for those of you 'unsure / not needing a wheelchair yet' – this is a great way to get you to places that you probably wouldn't otherwise go, and hopefully gives you a 'useful insight / experience' of how good 'a little bit of extra mobility can be'.

How much is it? – The introductory 2-week membership is a very modest £2.50, and a full year is just £10. The cost of each rental over and above the membership cost varies from site to site, but is still very modest in return for the enjoyment it brings. Many rentals are £0.00 nil, or an optional voluntary donation. Where there is a charge this should be no more than the equivalent of £2.50 per hour, or equivalent for longer sessions. It's very modest, for the huge enjoyment these trips provide.

Where are they? - Locally to me, they are at Woolacombe North Devon, Bridport Dorset, Lifton Gorge Devon, and Durlston Country Park Dorset, but I have had as much enjoyment at several other sites too, and there are many more on my 'to do list'. Take a look at 'Locations' on the website above.



Andy using a Tramper to access the beach in North Devon

A useful bit of advice –book in advance – a few days notice sensible too!!

Countryside Mobility was founded in the Westcountry, but the number of sites over

wide-ranging locations has grown considerably. Similar organisations have also been set up in other parts of the country – some are listed on the Countryside Mobility website – but I’ve Googled ‘Tramper Hire’ and found a number of sites around the UK.

Good luck – hope you enjoy as much as I do. Send your pictures to Ian for future newsletters too!

Andy Barrett

Artificial Intelligence

AI is in the news these days. There are different opinions, the more extreme ones involve robots putting us out of a job or turning on the human race and that will be the end of us. So I thought it was about time that this particular Luddite found out what AI could do.

Young Son (I have always referred to him as Young Son although he is no longer young at 47 and is my only son) has been using ChatGPT, an application for AI that was launched at the end of last year. It is a chatbot and “it is notable for enabling users to refine and steer a conversation towards a desired length, format, style, level of detail, and language”.

He has used for a few months and is very impressed with it. He believes the application will be able to build a software application if you state what you want it to do. That would put some programmers out of a job for certain. The chatbot can also create text and, to my disbelief, images. So I sent Young Son a painting I had done and he asked the chatbot to improve it to suit his living room better. You can see the AI version is FAR BETTER than mine so my painting career is in the balance now.



Young Son has also used AI to create questions to test his daughters on their school homework. Now that is a good use of technology, it has always been difficult for parents to challenge their kids learning but now they can with confidence.

So I thought it was time to sign up for this new technology myself.

Signing up is easy, put ChatGPT into Google and you fill in a very simple form to register. After that you can start a conversation. The term conversation is important, it is what makes the app very useful. When you ask Google a question you get a lot of links as a reply. If you ask another question Google does not retain any knowledge of your previous question. The AI chatbot does, so responses get more focussed just like a conversation with a human being.

I tried a few questions relevant to HSP. I list them below so that you can see for yourself whether the answers were any good. Personally, I think they are. They are well structured, accurate as far as I can tell, well balanced and include good advice.

The great advantage is that you as an individual can ask questions that are relevant to you. Rather like a Help Line that knows everything there is to know without having a sales pitch.

How do they do it? I have no idea; I just think the results are amazing.

Why do they do it? There is no money involved - yet. But I presume that when the finished product is available it will make money as a Google replacement and that we will be bombarded with targeted advertising. But in the meantime let’s enjoy this brief ad-free holiday. I would be interested to hear of other members’ experience.

John Mason

Specialist Centres for HSP

In the UK, there are several specialist centres and clinics that focus on rare genetic conditions, including neurological disorders like HSP (Hereditary Spastic Paraplegia). Here

are a few examples of specialist centres in the UK that may have expertise in HSP:

- **UCL Queen Square Institute of Neurology:** Located in London, this institute is internationally renowned for its expertise in neurology and neurogenetics. It has specialized clinics and research programs focused on various neurological conditions, including genetic disorders.
- **Oxford University Hospitals NHS Foundation Trust:** The Oxford Centre for Neurosciences is affiliated with the Trust and offers comprehensive neurology services. They have a neurogenetics clinic that may provide specialized care for individuals with HSP.
- **The National Hospital for Neurology and Neurosurgery:** Located in London, this hospital is a renowned centre for neurology and neurosurgery. They have specialized clinics and services for various neurological conditions, including genetic disorders.
- **Great Ormond Street Hospital for Children:** This hospital in London specializes in pediatric care and has expertise in treating a range of genetic and neurological conditions. They may have clinics or specialists dedicated to pediatric HSP.
- **Sheffield Diagnostic Genetics Service:** This service, based in Sheffield, offers diagnostic testing and genetic counseling for various genetic conditions, including hereditary spastic paraplegias. They work closely with local clinicians and specialists to provide comprehensive care.

Please note that the availability of services and specialists may vary over time, and it's essential to consult with your primary care physician or neurologist for the most up-to-date information on specialist centres near your location. They can provide guidance on referral processes and help you access appropriate care for your specific needs.

Amazing opportunity at The Calvert Trust

Some of you may remember our former Chairman David Pearce. On the front of the August 2006 newsletter David wrote an article about how much he enjoyed a stay with the Calvert Trust. For those of you who don't know, the Calvert Trust is an organisation with three locations around the UK, where they provide many exciting challenges for disabled people. These challenges include horse riding, kayaking, sailing, archery, cycling and abseiling to name but a few. I can remember David telling me what an incredible experience this was and he'd be delighted to know that some of our members have recently discovered for themselves how much fun and uplifting a Calvert trust break can be.

Sadly, David Pearce passed away in 2018 but he very generously left us some money in his will. Some of this was used for research but we wanted to use some for a purpose that we knew David would fully approve of. For this reason, we decided to pay half the cost of a Calvert Trust experience for any member. This is limited to one holiday per member every two years. We have allocated a certain amount of money for this and when it's gone, it's gone.

Please check out the Calvert Trust's website and if you like what you see, get yourself booked in. The HSP Group will refund half the cost.

For more information on this contact Ian Bennett

bravoechoonovember@btinternet.com

Fundraising News

Thanks to all who have raised funds for us using Easyfundraising.

If you ever shop online and are interested in supporting the HSP charity, please register for this or get in touch. Use the following link and follow instructions:

<https://www.easyfundraising.org.uk/causes/hspsupportgroup1/>

We are also registered with Amazon Smile so you can now raise funds for the group when shopping at Amazon. To use this go to:

<https://smile.amazon.co.uk/ch/1181539-0>

and that will help you select the group as your charity. The difference from normal Amazon shopping is that you need to remember to do your shopping from

<https://smile.amazon.co.uk/> so that the donations come through.

Covid meant that we all stayed indoors for far too long. It is time to get out there again and think of ways to get some exercise and in doing so, maybe raise some funds for HSP. If you are running in an event for HSP, we will be very happy to provide you with an HSP Support Group running vest.

Ian Bennett

HSP Group Grants

Funds are available for members to apply for financial assistance with the purchase of mobility aids or equipment that may improve quality of life or simply make life easier.

The maximum grant available is currently £1000. Completion of one year's membership is a requirement for applicants. If interested, please request a grant application form from the Secretary Dave Harris, details at the end of this newsletter. There is also an online form available via our website.

Members' Letters

Hello from New HSP Member – Philip Bungay

Hi my name is Philip Bungay I live in Rochester in Kent (Rochester is part of the Medway Towns of Rochester, Chatham & Gillingham – and although geographically part of Kent it is a Unitary Authority named Medway). I have been married to Louise for 33 years this October and we have x3 grown-up children; Emily (26) – a Shepherd on a farm near Rochester, William (24) – an electrician in Maidstone & James (19) – still at home. I took

early retirement in 2019 after a 29year long career in Whitehall (Ministry of Defence).

I have recently joined the HSP Support Group (joined in 2022 but didn't pay my subs until 2023), but my story goes back to 2012, if not before!

In 2012 I was diagnosed with Parkinson's Disease by a neuro consultant at Medway Maritime Hospital in Gillingham, Kent. Fast forward to 2019 and I volunteered for the 100,000 genome project – *at the end of 2019 ie just before the COVID pandemic, it was not until the end of 2020 that my consultant, now at Kings College Hospital at Denmark Hill in South London, that I received the result – genetic testing had shown that I had HSP. So, lots of unanswered questions – still! 'have I got both Parkinson's and HSP7', apart from the medication I take for Parkinson's is there anything extra I should be taking?' etc.*

Before I retired I became a leading member of the civil service disability group, which encouraged me to work together with other neurological support groups in Medway.

Together with the Chair of the Medway Branch of the Multiple Sclerosis Society (I was then the Lead Coordinator of the Parkinson's UK: Medway Working Age Group) we formed the Medway Neurological Network (www.medwayneuro.org.uk), this network now has 14 members including Ataxia UK, Dystonia UK, The Alzheimer's Society, Motor Neurone Disease Association, MSA Trust, Parkinson's UK and I can now represent HSP.

The aim of the Medway Neurological Network (MNN) is to be the voice of those affected by neurological conditions in Medway. Its mission is to defend and improve the quality of health and social care services available to those with neurological conditions, campaign for improved disability services and facilities; promote & provide support services/therapies/research projects to improve health & wellbeing; raise awareness of neurological conditions in Medway.

The MNN achieve their Aims & Mission by:

- Managing the [Medway Skylarks](#) singing group for all neurological

conditions – an evidence-based programme initially provided by the www.canterburycantatrust.org.uk.

- Running a series of drop-in groups or Neuro cafes in Medway – we work with Jaspers Community Café in Rochester to provide a Neuro Café each Friday of the month, including on the 3rd Friday a drop-in for Rare Conditions, St Augustine’s Church in Gillingham to provide a drop-in on the 4th Wednesday of the month & with the Walter Brice Neuro Rehabilitation Centre in Hoo St Werburgh to provide a drop-in on the 3rd Tuesday of the month.
- The MNN also have a campaign officer (me) – we campaign on National issues (ie the PIP process); national issues that are administered locally (ie Bus Passes issued to working age disabled people that can only be used after 9am) and local issues (i.e the Passport to Leisure scheme that doesn’t recognise fluctuating conditions).

In addition to being the Co-Lead of the MNN, I’m also a member of the Parkinson’s Centre for Integrated Therapy in Canterbury. The Director of the centre is Professor Wilkinson from the University of Kent – the MNN are currently in negotiation with the Parkinson’s Centre to establish a centre in Medway, which will be a resource for ALL neurological conditions.

I am also a member of the South East Advisory Group to the Disability Unit in the Cabinet Office, along with being a member of Parkinson’s UK’s South East Research Interest Group.

Philip Bungay

Zoom Meetings

Zoom Meetings for all HSP’rs

First Wednesday of every month

These meetings began during lockdown because we felt that it was important to give people the opportunity to get together and keep in touch with one another.

I volunteered to host them and have really enjoyed doing so. Attendances vary from about a dozen people to 40 and these people are always spread all around the UK. Since lockdown attendances have understandably dropped a little but a wealth of information has been shared. The list of topics discussed is far too long to print, but mobility aids, disability benefits, exercises, holidays, different types of HSP, symptoms, HSP specialist locations, and medication are regularly discussed.

New friendships have also developed which is always a highlight for me and on a few occasions, we’ve had people attend who have never come across anyone else with HSP.

These meetings begin at 7pm and usually end at around 9pm but people are free to come and go as they want. We have people who love to talk, and we have others who love to listen and learn. Both types are very welcome at these meetings.

Invitations and reminders are sent out to members by email.

I look forward to seeing regulars and new faces at future meetings. If you’d like further information on these meetings you can contact me by email

bravoechoonovember@btinternet.com

Ian Bennett

Zoom Scotland 21 May 2023

John circulated all members in Scotland

Some said they would come and some sent apologies

In the end we had an interesting discussion about how the fund that is supposed to exist for Scots to travel to centres in England can be accessed. It seems one exists to allow those on the islands to travel within Scotland for treatment.

It seems common that knowledge of HSP is low, and that different Health Boards have different approaches

There is also a shortage of clinical neurologists

The English Govt have published their strategy and a couple of trials In SE England and Merseyside are being done for neurology, and presumably the Scottish Govt are waiting to see the result as to locating their centres or using those in England.

We decided that a regular Zoom will be held for all people in Scotland and we will decide if/where physical locations will be as interest demands.

For your interest

In Scotland, there are 4 main regional Clinical Genetics Departments:

- Edinburgh(South East) Clinical Genetics Department.
- Glasgow(West) Clinical Genetics Department.
- Aberdeen(North) Clinical Genetics Department.
- Dundee(East) Clinical Genetics Department.

Mike Cain

Birmingham Support Group Meeting Saturday 15th April

It was great to be able to see everyone again at our first 'face to face' meeting since Covid.

All 16 of us (including 5 people who hadn't been to this support group meeting before),

enjoyed a super buffet lunch and an informal chatty afternoon.

We have already booked up the next meeting for Sunday 15th October 12-15.00

June Masding and Penny Cohen

Forthcoming Events

The South West

November 18th 2pm

The Dartmoor Lodge Hotel, Ashburton

Contact Ian Bennett: [07941 535 282](tel:07941535282)

A meeting for all members

Simply a natter to catch up with old friends and maybe make some new ones

The first Wednesday of every month 7pm, online using Zoom

Zoom details are emailed to all members or

Contact Ian Bennett: 01202 849 391 or bravoechoonovember@btinternet.com for info

Birmingham/West Midlands

October 15th 12.00pm – 3.30pm

pennycohen57@hotmail.com or

june_masding@hotmail.co.uk for details

Colchester/Essex

October 22nd 2.30pm – 5.00pm

Feering Community Centre

Contact Hilary Croydon for further information Tel: 01284 728 242

tohiti@btinternet.com

Milford

Sunday November 19th 2pm – 5pm

The Clockhouse Milford

GU8 5EZ

Call Jane Bennett on: 020 8853 4089

New Members

We welcome the following new members:

Ruth Gammon Shrewsbury	Phil Arnold Liverpool	Stephanie Samson Jersey
Mario Gauci Birmingham	Stephen Ledwith St Helens	Sam Lawrence Epsom
Michelle Rowberry Birmingham	Deborah Holt Peacehaven	Tracy Askham Skipton
Paul Hartley Huntingdon	Michalis Kontopodis Leeds	Emily Ryan High Wycombe
Darren Ralph Shrewsbury	Mark Cowan Midlothian	Matt Hodgson London
Karen Winter Canvey Island	Susan Baylis Doncaster	Zoey Hemmings Kent
Luke Scudder Tewkesbury	Paul Zollo Templecombe	Philip Bungay Rochester
Nick Garrett York	David Karl Frome	Hayley Boase Plymouth
Isobel Morgan Bracknell	Richard Walker Colchester	Roseann Reilly Glasgow

Useful Contacts

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Kevin Mills – Wales meetings host	kevinmills944@gmail.com	01874 622 727

2023 AGM - Diagnostic Odyssey

On Saturday July 22nd Ela Curic gave a presentation to the group about her student voice essay. Ela began by introducing herself, she is a 5th year medical student in Sydney, Australia, who has included rare diseases and genomic testing in her studies. She entered the Beacon (formerly findacure) and Medics 4 rare diseases student voice competition. Ela's essay was shortlisted in the 2022 competition.

She chose to answer the question on the diagnostic odyssey by comparing and contrasting two different people/rare conditions, and looking at the wellbeing impacts of the diagnosis journey. Ela interviewed our chair about HSP and Laura, who has Bethlem Myopathy. Ela gave a brief overview of HSP and Bethlem Myopathy, noting that HSP has a prevalence of about 3.6 in 10,000 and Bethlem Myopathy of 0.77 per 10,000. There are about five times as many people with HSP than Bethlem Myopathy.

Getting a diagnosis for a rare disease can improve the pathway to managing the condition, and many have a genetic basis. The challenge for many is that they cannot get a diagnosis easily, they go through a series of different appointments where they have to explain their situation again, without getting a clear answer. This affects their mental wellbeing. Overall the diagnosis often takes over five years. The traditional approach for diagnosis is the doctor recognising common symptoms and using their knowledge to identify what has happened. For rare conditions this approach cannot work, leading to a succession of disjointed appointments without progress. Ela then went on to consider three different aspects of the diagnosis journey:

Navigation

Often there is no clear route for diagnosis of a rare disease. People often visit 8 different clinics, 4 with their doctor/general practitioner, and 4 with specialists of one kind or another. Given that rare conditions are not well known it is common for people to start with a misdiagnosis, and in that typical journey, there are 3 different misdiagnoses.

For people whose condition affects their mobility they have the added burden of travel between these different clinics, which can be difficult at times. All of these factors - many clinics, misdiagnosis and mobility challenges have negative effects on wellbeing.

However, once a diagnosis is reached there are positive effects. A correct diagnosis means that treatment options, exercises and other measures are done in the knowledge that they are the right things to do, and the diagnosis can open up access to different benefits and services.

Uncertainty

Rare conditions have additional uncertainties above commonly experienced conditions. Once a diagnosis is reached this can help re-frame the different symptoms and experiences that people have experienced, allowing them to realise reasons why they have felt different.

Diagnosis can also reduce the uncertainty of the future, by being more certain about what is coming in the future, and allowing people to plan for this. Some people will be able to use this in the context of reproductive issues to help them make decisions about having children. Others will use this when deciding on treatments and assessing their progression, although this is often difficult to do and there will still be some uncertainty here. People with rare conditions often have a lower health related quality of life, and some of this is due to uncertainty which is independent of their diagnosis.

Connection

Before people get a diagnosis they often feel disconnected from their usual lives. Getting a diagnosis offers an avenue for them to connect with other people. Connections with people is important. For conditions with support groups, this can be one route to connect with others that are similarly affected. There are also groups of people who do not yet have a diagnosis, which gives another route. Psychological support during the diagnosis journey, and beyond, is beneficial to all.

What does this all mean?

Ela concluded her essay considering the experiences of the people she interviewed. The role of the diagnosis is not always clear, with different people drawing different aspects from this. The main aspect is that having a diagnosis can help people to understand the impacts of the condition on their lives. For some people the diagnosis helps plan the future, but it is important to realise that many rare conditions do not have treatment options available. Also, the diagnosis offers routes to connections with others. All of these factors are helpful to people's wellbeing.

You can watch this presentation on YouTube: <https://www.youtube.com/watch?v=82bgjNPo8E>

2023 AGM - Spastin recovery in HSP

On Saturday July 22nd Cinzia Rinaldo gave a presentation to the group about the work that her research group has been doing in Italy. The group part-funded this research, and the presentation was to inform members about their new findings.

She outlined the different people involved in her laboratory and their partners/collaborators. There have been two different aspects of the project, which focuses on the SPG4 type of HSP, where the genetic mutation is in the Spastin gene. Where spastin is affected by HSP its effectiveness is reduced, and therefore it is unable to do its job within cells as well.

- Firstly, they are seeking to increase the levels of Spastin by reducing its degradation. They note that this is a possible therapeutic approach for HSP. They have been testing the drug MLN4924.
- Secondly they are trying to identify biomarkers for SPG4 to make it easier to detect when drugs are working.

Spastin's job within the cells is to cut microtubules. Most people affected by SPG4 have one copy of spastin which functions properly, and one copy which does not. Therefore, the microtubule cutting is not done as effectively in those with SPG4. The approach for the study is to target recovering the level of spastin in the cells to allow it function better. Their study seeks to answer three questions;

- How are the levels of spastin regulated within the cell?
- How can the levels be increased?
- How can the recovery of spastin levels be detected?

Regulation

They identified a pathway which regulates spastin, and this pathway can be modified by drugs. There is an inhibitor which can be used to block the degradation of spastin within the cell. Their research also explored all of the different factors which can elevate the levels of spastin, allowing other pathways to be identified in the future.

The drug MLN4924 is currently being trialled as a cancer treatment, and has been demonstrated to block the degradation of spastin in cells. They are hoping to re-purpose the drug for use in HSP.

Increasing levels

They have been testing MLN4924 using animal models of HSP. With the HSP mouse, this doesn't usually have any copies of spastin. When the mouse has one copy of spastin the HSP effects are small and occur late in its life. A mouse with no spastin is not a good choice to test methods to recover spastin.

However, the fruit fly model of HSP has spastin, and it is easy to detect the changes in the fly as a result of changes in spastin. Cinzia showed photos of fruit flies, an un-affected one, one with the spastin mutation, and one with both spastin mutation and the treatment. They demonstrated that the levels of spastin are recovered using the treatment.

Detecting change

The team have developed a non-invasive cell imaging method to be able to detect the effects of spastin (or lack of) in blood cells. The team can use the imaging to identify both cells which are affected by lack of spastin, and the effects of the drug being trialled. This work has been published:

<https://onlinelibrary.wiley.com/doi/10.1111/ene.15756>

Next Steps

She concluded by describing the next steps for their team. They want to extend their cohort of people affected by HSP to be able to consolidate their biomarker results and potentially develop more. They are going to investigate the spastin elevating drugs and their impacts on cells. They want to develop prognostic and predictive methods for tracking spastin in non-neural cells.