

HSP Newslink

The Newsletter of the Hereditary Spastic Paraplegia Support Group

Issue 42 – October 2017

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

Jo and Abi run the Leicester 10K



Jo and Abi enjoying the pre race training

Abi and I have run together for around two years, and enjoy taking part in organised runs to give us a goal to work towards and motivation from knowing that we're raising money for charity in the process. Last year we ran Birmingham 10k to raise money for Macmillan, who cared for Abi's Grandad, and this year we were pleased to use the Leicester 10k as a reason to ask our friends to donate to the HSP Support Group.

The HSP Support Group has been a huge help to my Mum, who has HSP. It's offered her, and us, opportunities to learn about the condition and to meet other people in similar situations. As a rare condition I'm pleased to have been able to talk about it and raise awareness of both what the illness is and the great work that the HSP Support Group does.

With Abi having recently moved to Leicester and me living in Birmingham, we often trained solo but have been checking in with one another's progress. Having run a few 10k's before, we knew we could achieve the distance, so we were focused on doing it in a good time. I was very pleased to have shaved a couple of minutes off my personal best and ran in 53 minutes 21 seconds, making me the 66th woman across the line! Abi ran it in 58 minutes 32 seconds, and felt that she left it a bit too late to let out her final burst of energy, but ran close to her best time too.

We're so pleased to have raised so much for HSP Support group, and that it will be used to fund research and support those living with the condition. The current fundraising total stands at £645, and our fundraising page will remain open for the next few weeks, for any extra donations

to come in. To donate, and to see some pictures of us in training and on the day, go to <https://www.justgiving.com/fundraising/joanne-masding2>.



Abi and Jo proudly wearing their medals after completing the Leicester 10K

A huge 'Thanks' to everyone who supported us!

Love Jo & Abi xxx

Chairman's Column



Hello everyone. I am pleased to be writing wearing my new 'hat' as Chair of the group, having been elected at the AGM in July. The first and most important thing to do is to

thank Ian for his chairmanship over the past 7 years. I am very pleased that Ian is remaining on the committee as membership secretary and editor of this wonderful newsletter. Secondly, I'm looking forward to working with the committee over the coming years to make sure that the group can support you as much as it can. Thanks to John, Dave, Stephanie and Simon for carrying on the committee, and welcome to Della and Mike who have just joined.

It was really good to see so many of you at the AGM. I'm pleased that people make the effort to attend from all over the country both to hear the presentations made, talk with old friends and make new ones. As I said, I'm really keen to hear any suggestions for things that you think we should be doing more or less of. You can e-mail me at: chair@hspgroup.org or contact me through social media. Several members have made suggestions and these will be discussed.

The committee met in early September to discuss various aspects of the group, the most important of which is to bring the constitution of the charity more in line with what we do. We need to do this as fundraising is becoming a more significant part of our activities. The meeting was really productive and I'll be happy to share updates as we go.

My plan is to keep my chairman hat separate from the hat that I wear whilst writing my usual research column. You can read my report of the AGM presentations later in the newsletter, and it is also time for the launch of my annual on-line HSP survey.

Launch of my on-line survey

I'm pleased to announce that my 2017 on-line HSP survey is now open. This year I'm looking at:

- * How HSP affects people's jobs/occupations
- * Pain
- * Factors that affect walking
- * Wellbeing

The survey is a mix of my questions (jobs and walking) and standard questions for measuring pain and wellbeing. As per previous years I'll collect data until about the end of the year and analyse by rare disease day (end of Feb 2018). I'd be really pleased if anyone with HSP could take the time to answer my questions. You can access the survey through my blog at:

<http://hspjourney.blogspot.co.uk/2017/09/2017-survey-now-open.html>

Adam Lawrence

Editor's Column

Adam began his column by thanking me, so I'll begin by welcoming him. I've got to know Adam over the last two or three years and I've learnt enough about him to have confidence that he'll be an excellent chairman for our charity. He's very thorough and has already begun tidying up things that I was guilty of brushing under the carpet. Many of you will be familiar with the surveys that Adam has compiled and other work he has done for the newsletter. From this you will have clearly seen his attention for detail and passion for finding out more about HSP. He always has great ideas and he has the energy to follow them through. It's great for me to be quieter at committee meetings and I know that the work involved with AGM planning will now be shared. I welcome some of the weight leaving my shoulders and more importantly I welcome Adam to the role of Chair.

Adam was already a committee member prior to the AGM so had already seen how we operate and no doubt developed thoughts of areas where he can make improvements. However, we did take on two new committee members, Della Brookman and Mike Cain, and like Adam, I also welcome them aboard. They both bring ideas to the table and will have certain duties to work on. I have included an updated table of 'Useful Contacts' later in this edition.

I'm already busy organizing next year's Potato Pants festival. We try to make improvements every year and hopefully raise more money every year. I already have the music line up sorted out but there's plenty more to be

organized. I hope to see some of you there on June 2nd next year

I'm delighted to see the Facebook group; Hereditary Spastic Paraplegia's Unite, continue to be so well used. It now has nearly 1700 members and I have noticed people singing its praises recently. If you're not a member, I strongly recommend you join.

As many of you are aware, I am a trustee of another charity which is very close to my heart, called Flying Scholarships for Disabled People (FSDP). I was awarded a flying scholarship in 2005 and have kept involved ever since. Approximately 10 of our members have now benefited from a flying scholarship and many of them will agree that this really does change lives.

There is a link between the RAF and FSDP and because 2018 is the 100th anniversary of the RAF, FSDP are intending to award more scholarships than they've ever done so before. Currently 15 scholarships are the maximum number presented in a single year so next year there will be at least 16 and possibly more.

For this reason, if any of you are even slightly interested in applying, now is the time to do so. Closing date for application for a 2018 scholarship is November 30th this year, so although there's plenty of time to apply, you have to be hasty. For further information, either give me a call or visit

<http://www.fsdp.co.uk/>

Please go for it because this is a once in a lifetime opportunity.

I really appreciate any material that members provide for the newsletter. **Please continue to let me have any interesting stories or useful information which I can include for the benefit of our readers.** I was particularly interested in Barbara's account of her recent cruise, because I'm having my first cruise experience next month. I'll certainly let you all know how I get on.

Adam mentioned the AGM in his column so I won't repeat what he said other than agreeing what a great new venue we have found. Many thanks to June Masding and

Penny Cohen for their hard work in finding the venue.

On the subject of the AGM the committee recently agreed that it would be great to have a brief presentation from a member at future meetings. Amber gave a memorable presentation for us last year discussing her skiing and we thought it would be good to try to encourage similar. If any of you have interesting hobbies or anything else that you'd like to share with us, please let one of the committee know.

I'll finish by thanking everybody who's recently or currently involved with raising funds for HSP Support Group. You'll read about a couple of them in this issue but all efforts are much appreciated and funds raised are invaluable.

Ian Bennett

HSP Regions

In the early days of the HSP Support Group, a decision was made to divide the country up into various regions. The thoughts behind this were that each region would have a co-ordinator who would organize one or two meetings a year.

The committee have now decided to abolish the Regions and particularly their numbering system. Members are often confused which Region number they live in or what are the boundaries of a particular region.

Meetings will still be arranged by volunteers but will now simply be listed and identified by the town where they're located. For example, the meetings that I organize in the south west will no longer be referred to as Region 4 meetings, but Ashburton meetings.

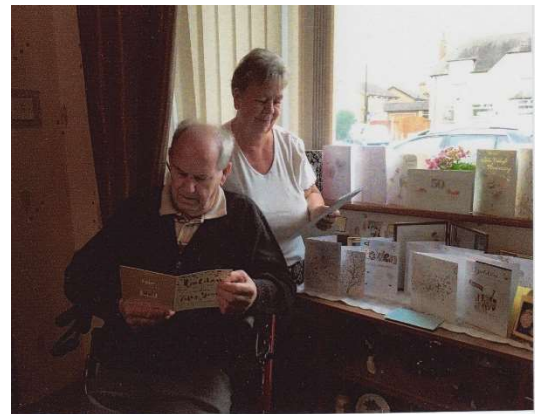
This enables volunteers to come forward and arrange an informal meeting anywhere in the country and when advertised in the newsletter, readers will hopefully find it much clearer where the meeting is located.

When I list the new members in each edition of the newsletter, I'll only highlight the town or county where they live and the Region number will no longer be listed.

Volunteers are still encouraged to come forward and organize local HSP get-togethers. The committee will always provide encouragement and support to help establish these important events.

Fundraising News

Congratulations and a big thank you



Many thanks to Marion and Gordon Jakeman who asked for donations rather than gifts, when celebrating their Golden Wedding Anniversary recently. On behalf of the committee and everybody connected with the Hereditary Spastic Paraplegia Support Group, I'd like to thank them for their generous donation of £350. Congratulations to Marion and Gordon from all of us for celebrating 50 years of marriage and we wish you many more happy years together

Ian Bennett

Model Railway funding

Burnham and District Model Railway Club design and manufacture model railway wagons. They have a great community support policy and they regularly produce a wagon where all the proceeds from sales are given to charity. As Chris James, a member of the HSP Group is also a member of Burnham and District Model Railway Club, we were fortunate enough to be selected as a charity that they'd support.

A wagon was designed and the proceeds from its sales were donated to us. On 23rd September I was presented a cheque for £521.82 by John Langley who was running a stall at the Dorset model railway exhibition.



John Langley and Chris James from Burnham and District Model Railway Club presenting me with the cheque

Many thanks to Burnham and District Model Railway Club for their generous support which I have told them will be used for research.

Ian Bennett

1000 miles for HSP



Stephanie, Stuart, Ian, Neil and Michelle getting in some extra miles on Bournemouth promenade

At the beginning of August, I had the great pleasure of meeting up with Michelle and Neil Brookes and their son Finnegan on Bournemouth beach to join them for a little stroll. It was also great to have the company of my good friend Stuart Montrose and his girlfriend Stephanie. Only last year, Stuart walked from Lands End to John O'Groats and in doing so, raised funds for our charity. Coincidentally, during Stuarts walk, he met Michelle and Neil.

As many of you will know, Michelle is walking 1000 miles over the course of the year, to raise funds for HSP. Michelle added ten miles to her total on the seafront at Bournemouth and it was a great excuse to meet her and Neil and have a good natter over lunch after our walk.

I felt a little inadequate in the company of Michelle and Stuart, bearing in mind the distances they walk, but with the great company of Neil, I managed over three miles in my wheelchair. I have to confess that we stopped at half way for some light refreshment.

This is a huge challenge that Michelle has taken on for HSP and amazingly, she has already raised £2800.

<http://uk.virginmoneygiving.com/MBrookes>

Ian Bennett

Petula's new Powerchair

We try to visit the Motability Road Show each year. 2016 was Silverstone Racing Circuit and this year Stoneleigh Park. The aim was to see what was on offer with lightweight portable powered chairs. More on offer this year and I rode around the venue on three different makes of chair. We took leaflets home on these chairs and I left it a while before deciding to go ahead as it was a big step for me as I did not want to make the wrong choice and feel I had wasted my money on something not suitable. After speaking to Ian my husband we felt the "Freedom Chair" was going to be most suitable with transporting it in the car, how comfortable it felt and ease of use.

I had previously been around my local National Trust property, Ascott House, Wing near Leighton Buzzard in Bedfordshire in my manual wheelchair with Ian pushing, hard work I believe! So with this in mind we suggested to Chris, the representative for "e-goes" (SMART E-MOBILITY) distributor for the UK and Ireland for the Freedom Chair that we would like to see how the chair would perform on the gravel paths and uneven ground around Ascott. Well after the second

attempt to arrange this, the first was abandoned due to holdups on the motorway, Chris arrived with all three of the models of the Freedom Chair, A06, the smallest, the A08 a larger model and then A08L, the L for larger wheels. I had thought that this would be the model for me so I rode on this one, Ian on the A08 and Chris on the A06. We set off in tandem and raised a few smiles from the people also visiting. We tried all the different surfaces and all three chairs managed them well, declines and inclines also. Two groups of people spoke to us about the chairs as they thought they would be ideal for members of their family or friends, so Chris was busy telling them about them. Also he did some filming for the company back in China, so I am on film somewhere apparently! We stopped off for coffee and cakes and I then swapped chairs and tried the A06, this was little higher and easier for me when transferring to another seat. We got back to the car and Ian set about putting them in the car, first the larger one, then the smallest and straight away we could see that the A06 was going to be the best all round. Lighter and smaller for Ian to handle into the car, and as I found that they seemed to all manage the terrain we decided to go ahead with the smaller chair with two batteries to give me approx. 16 miles between charging.



Petula in her new Freedom Chair

Ordering online (www.e-goes.co.uk) was straightforward, just completing their order form for the chair chosen and number of

batteries, plus their form to have it excluding Vat and delivery was arranged soon after.

Whilst this company were very helpful with my order and purchase of my chair I have since had problems contacting them regarding an item of paperwork. I would not like to promote this company if this issue was to continue but there are two other companies who stock this chair if anyone is interested: -

1. Uk-wheelchairs.co.uk
2. Spring Chicken Direct Ltd (GB)

Our first trip out was to Maldon in Essex, the excuse was as our water was going to be off most of the day so we thought it best to have a "day out". Ian remembered going there probably about forty years ago and liking the area so off we set. We finally arrived after traffic jams and losing our way, we decided lunch was needed. We set off together being able to chat which is something I had forgotten as when you are being pushed it is rather difficult to have a conversation also when your other half is a little hard of hearing. After lunch we travelled along the paths by the river which in some places were narrow and uneven but I felt comfortable and secure in the chair.

We found it invaluable on our week's self-catering in Scotland where I used it in the chalet and also on our walks around gardens, parks, shops, even RSPB nature reserves where their walks to bird hides are quite some distance and over muddy grass and dirt tracks, I did try to miss the puddles!

It now travels in the boot of the car with my manual one so dependent of where we are visiting I have the choice of either.

On the carer's view point, my husband's in this case.

Verdict on the Freedom chair is that the fold up design is very good and it should fit into most hatchback cars. It is well constructed, and by having the two batteries (lithium crystal) very lightweight, increased the range to about 16 miles. One or two issues with the joystick control bracket working loose, but it can be easily tightened. One word of caution, I feel that your upper body strength needs to

be reasonably good to lift it in and out of the vehicle.

All in all, I as the user and my husband feel this is a good powered chair that suits our needs at present, and we express our grateful thanks to the HSP Group for its grant contribution towards the cost of my chair.

Petula Baker

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 has recently been raised to £750. Completion of one year's membership is a requirement for applicants. If interested, please request a grant application form from a committee member.

SPG 11 Families

Our son, Tim, who is 24, has SPG 11 which is one of the complex, recessive SPGs so that he is the only member of our family to be affected.

SPG 11 is very similar to SPG 15 and they have a wide variety of additional symptoms. We find it has been very helpful to be in email or phone contact with other families who are living with the conditions. By talking to others we can all benefit from shared knowledge which means that we can support our young people more effectively.

We are in touch with six other families, so far, who are now also in contact with each other as a result. The affected members of the families also often talk to each other on Facebook.

Even the experts don't know how many SPG 11 and SPG 15 Families there are in the country – perhaps we can find out.

I'm sure there are other families out there! If you are part of an SPG 11 or SPG 15 Family and would like to be able to talk to other families for mutual support and to exchange

experience do, please, give me a ring on 01284 728242.

I would like to hear from you very much.

Hilary Croydon

Collect Stamps! Raise Funds for HSP!

Don't forget to collect your good used stamps for Phil Burton to sell to raise funds. Pass them on to branch meetings, the AGM or anyone who can get them closer to the Milford meeting where Phil can pick them up.

Members' Letters

Our Cruise to the rivers in France and Spain

On 10th June, my sister Shirley and I left for a two week cruise on Fred Olsen's ship Braemar. We left Southampton with our cases in our cabin and us feeling so excited. Our cabin was good and soon we had the unpacking and putting away done. As soon as the ship left Southampton we all had to go for the life-boat drill.

Later we went to the dining room for a tasty lunch and to meet four people on our table. The waiters were very helpful, Shirley and I both had a rollator and after seating us they put them in a safe place for us. That afternoon we made use of our rollators and wandered around the ship, so much to see and people to say hallo to.

We dressed up for dinner and once again we were looked after by the staff. The food was fantastic and the other guests on our table were good company. We went to the entertainment end of the ship after and watched a wonderful show. When that was over we went back to our cabin and bed.

That was the first day aboard Braemar. Over the next two weeks while at sea there was so much to do, including short mat bowls, bingo and quizzes. You could also just sit on deck

with a drink and watch people passing us doing their mile walk – four times around the ship. I did go round the ship once.

On leaving UK, our first port of call was Bordeaux. Then after a few days at sea we were in a very hot Seville, temperature 100F. So glad of our air conditioning on board. We did go by road on a trip around the city, not many people about, all indoors out of the heat. When the ship left Seville we were taken to Vigo and then to Rouen in France. Quite a lot of people left on a trip to Paris, a very early start for that.



Shirley and Barbara enjoying their cruise
I hope you like the details I have given of our holiday. We were so well looked after and met so many lovely people. Next year we are going on a Norwegian cruise, again taking our rollators with us.

Barbara Jones

Regional News

Barbara's Garden Party

Members mainly from the **Norwich** area met in my garden on 8th July for a 'Garden Party'. I had made a large Strawberry and cream cake the night before and with cakes brought in by

members, we all had an enjoyable afternoon sitting in the warm sunshine.

We were delighted to have the company of a new member, Mrs Diana Love and her husband. Mr Francis Pepper brought with him a foot vibrator to demonstrate and I showed off my new rollator that I used on my cruise.



HSP members enjoying Bararbara's garden party

I think we all enjoyed the afternoon. Lots of subjects were discussed with lots of laughter. Not once was BREXIT mentioned. Before my friends left a group photograph was taken. I hope we can do the same again next summer

Barbara Jones

Hitchin meeting

On 9th September 2017 we held a social get together meeting at our normal venue The Orange Tree pub in Hitchin, Herts.

We had a group of 10 HSP'ers that attended and we welcomed a new member to the fold.

Very relaxed afternoon, with everyone participating in various subjects, (not necessarily about HSP) but life in general.

The pub offered great food and beverages etc that members could purchase if they wished. Some had lunch, others a snack etc.

We have arranged the next social meet after Christmas, to break the doom & gloom feeling some people get after the big event excitement has passed. Saturday January 27th at the same venue.

Would be lovely to see/meet some of you there.

Della Brookman

Forthcoming Events

Morecambe get together

Saturday October 7th 2pm - 5pm
Midland Hotel,
Marine Rd W,
Morecambe LA4 4BU
Contact Irena Pritchard on: 01524 261 076
Or Mike Cain on: 0161 456 7531
Email irena.pritchard@btinternet.com

Birmingham Meeting

Saturday 14th October, from 12 - 3pm
The Kenrick Centre,
Mill Farm Road, Harborne,
Birmingham, B17 0QX
Contact Penny Cohen: 07818 288 738
Email: pennycohen57@hotmail.com

Colchester Meeting

Sunday, October 15th 2.30 - 5pm
Feering Community Centre
Feering,
Essex,
CO5 9QB
Call Hilary Croydon: 01284 728 242

Ashburton Devon Gathering

Saturday October 28th 2pm onwards
The Dartmoor Lodge Hotel, Ashburton
Call Ian Bennett on: 01202 849 391

Milford Afternoon tea

South/South East

Sunday November 12th 3pm – 6pm
The Clockhouse Milford, GU8 5EZ
Call Jane Bennett on: 020 8853 4089

Hitchin Social Meeting

January 27th 2018 1pm - 4pm
The Orange Tree Public House
100 Stevenage Rd, Hitchin, SG4 9DR
Contact Della Brookman: 07710 637 941

Stockport get together

Saturday March 17th 2018 2pm – 5pm
Quaker Meeting House,
2 Cooper Street
Stockport, SK1 3DW
Contact Irena Pritchard on: 01524 261 076
Or Mike Cain on: 0161 456 7531
Email irena.pritchard@btinternet.com

Potato Pants Music Festival

Saturday 2nd June 2018 from 2pm - 11pm
High Mead Farm, Ham Lane, Ferndown,
Dorset, BH22 9DR
Request disabled parking (much closer)

New Members

We welcome the following new members:

Anthony Sothern
Essex

David Hood
Glasgow

David Buxton
Cardiff

Ayshe Ufuk
Prestwich, Lancs

Jo Currass
Hampshire

Bernard Franckel
Surrey

Joan Gaskin
Leeds, West Yorkshire

If you are interested in contacting any of the above new members, please contact the membership secretary.

Useful Contacts

	Email	Telephone No:
Adam Lawrence - Chairman 18 Mayflower Court, Staple Hill, Bristol, BS16 5FD	adam@idios.co.uk	07834 505 161
David Harris – Secretary 18 Lodmoor Avenue, Weymouth, Dorset, DT3 5AF	dave@vadcar.org.uk	01305 772 286
John Mason - Treasurer 18 Sandringham Road, Trowbridge, BA14 0JU	john@jrmason.demon.co.uk	01225 761 788
Stephanie Flower - Help-Line 37 Alexandra Road, Great Wakering, Southend, SS3 0HN	FSPgroup@aol.com	01702 218 184
Ian Bennett – Membership Sec / Newsletter Editor 37 Wimborne Road West, Wimborne , Dorset BH21 2DQ	bravoechonovember@btinternet.com	01202 849 391
Simon Hubbard – Research co-ordinator	simonjameshubbard@gmail.com	01908 508 866
Jane Bennett – South / South East co-ordinator	luckyjane82@hotmail.com	020 8853 4089
Della Brookman – Trustee & Regional co-ordinator	della_brookman@msn.com	07710 637 941
Penny Cohen – Birmingham Regional co-ordinator	pennycohen57@hotmail.com	07818 288 738
June Masding – Birmingham Regional co-ordinator	june_masding@hotmail.co.uk	01214 445 095
Mike Cain - Trustee	michaelcain123456@yahoo.com	0161 456 7531
Irena Pritchard – North West Regional co-ordinator	irena.pritchard@btinternet.com	01524 261 076
Hilary Croydon - Colchester area co-ordinator	tohiti@btinternet.com	01284 728 242
Barbara Jones – Norwich area co-ordinator	babs27833@gmail.com	01603 423 267
Liz Redmond Neurogenetics Nurse	Redmond.liz@uclh.nhs.uk	02034 484 487

Summary of 2017 AGM Presentations

Overview of Genetics Service - Dr Nicola Cooper

Dr Cooper gave an overview of the genetics service offered at Birmingham Women's and Children's Hospital, although much of what she said is good relevant information. She began outlining that one in 17 of the population are likely to have a rare condition in their lifetime, and genetic testing can help to identify the cause and progression of such diseases.

There are four main aspects of the care that they give:

- 1) Giving information on what genetic testing is - how it can (with a definitive result) be used to guide treatment and give an assessment on potential outlooks on life, and can give information on if things are/can be passed to children and the level of risks for different aspects and provide information for the family.
- 2) Outlining the different choices available - there are tests for individuals, and tests can be done on children and during pregnancy.
- 3) Providing support for the whole family - the results of a test can affect more than just the person being tested.
- 4) Help families make the choices that are right for them.

In the wider sense diagnoses can be made in a number of different ways. The person's family tree can be examined, there can be a number of physical examinations or investigations, and

sometimes there is a genetic test available.

There are three types of genetic inheritance, and there are examples in HSP of all three types. Each gene that person has is a pair of genes, one from their father and one from their mother.

If a condition is 'dominant' then the gene for that condition needs only to be in one of those pairs for the person to have that condition. (SPG4 has dominant inheritance). With dominant conditions there are no skipped generation and each child has a 50/50 chance of inheriting from their affected parent.

If a condition is recessive, then the person needs to have inherited the gene from both parents. (SPG11 has recessive inheritance). If a person has one copy of the recessive gene, then they are a 'carrier' of the condition but are not affected by it. Recessive conditions can skip generations as people can be carriers. If both parents are carriers, then the chance of a child being affected by the condition is 1 in 4.

Lastly X-linked inheritance where the gene for the condition is on the X chromosome. These generally affects males as they only have one copy of the X gene, being XY). Females are XX, and are usually unaffected by the condition but can carry it. (SPG1 has X-linked inheritance).

Further to inheriting genes from our parents there will also be some genetic changes within us. Dr Cooper said that each person has around 60 genetic changes which are not in either parent. Such changes could lead to HSP with any form of inheritance.

Dr Cooper then went on to talk about diagnostic testing. This can be used for a person affected by a condition. The testing can firstly identify what the condition is, and then what type it is. Predictive testing can similarly be undertaken for unaffected relatives. Having a test can help with planning, career choices, life decisions and that kind of thing.

With predictive testing there will always be a look at personal history. This will help understand the personal circumstances for the person and level of support that they would have. One of the biggest factors is that having a test changes perspectives - you move from "might have" a condition to "will have" a condition. Part of the personal history is getting a feel for how this might affect someone psychologically. Having a result can be beneficial in terms of planning for the future, but could also have a disadvantage in that you may have to declare that you have a condition when applying for a mortgage, for example. She noted that a clinical examination is always a snapshot of a person, it cannot tell you how you will be in the future.

Dr Cooper described two different types of genetic testing. Until relatively recently genetic tests were done using "Sanger Sequencing" which looks at one gene at a time. Now such tests are done with New Generation Sequencing (NGS), where a panel of different genes are looked at simultaneously. Gene panels tend to have ~40 different genes in them.

One issue with panel tests is that interpreting the results can be difficult to do. Some of the results back are not clear. Other results may come back showing changes, but it is not always clear that those changes are giving rise to the effects being observed. Some people are affected by more than one rare condition,

and it may be difficult to identify what is going on from panel test result, especially if some of those conditions are similar. The age of onset and rate of progression of HSP have influences from other factors. Some of these factors will be genetic and others will be environmental.

Dr Cooper described genetic changes as being like a key. The genetic change can happen at any place in the gene and the effect of the change depends on where it happens. If we liken a gene to a key, then if the change occurs in the part of the key which you hold then the key will still open the lock perfectly. Some change may be small, and that might be like a slightly wrong key which you can get to open the lock by giving it a bit of a wiggle - and that all might be OK. When the change is bigger or occurs in the blade of the key then the lock may not be operable with that key.

If you are in the UK and do not have a genetic test result for HSP then you may be eligible for the 100,000 Genome Project. Talk to your neurologist!

Current HSP Research

Prof Andrew Crosby

Professor Andrew Crosby gave a presentation on current HSP research.

He began by giving an overview of some of the HSP characteristics. HSP is described as being "heterogeneous" - but what does this mean? Simply, it means "variable", genetically in this context. There are 73 different HSP genes identified, of which about 40 have been confirmed in follow up studies covering several families. Prof Crosby speculates that there will be hundreds of HSP genes in the end. There is also variability within one variation - he mentioned Silver Syndrome (also known as SPG17, which

inherits dominantly) which he described as HSP plus hand muscle wasting. First symptoms are usually observed in teenagers. One example mentioned had a parent who was normal at 48, but it is not known why.

Background information: Our genes are responsible for producing proteins which have jobs to do in our body. The proteins are made from the DNA in our genes, although they have to go through several steps to do this. Should a gene be faulty there may be a problem with the proteins that are produced. Neurological conditions are often referred to as "upper" where the brain and/or spinal cord are affected, or "lower" when the nerves between the spine and the muscles are affected. Some motor neuron diseases may affect the upper, lower or both sections. HSP is a motor neuron disease.

By considering all motor neuron diseases together provides a bigger family of conditions and knowledge of one genetic alteration may help all motor neuron diseases, and the more confident researchers can be of finding a genetic route for changes.

Prof Crosby described the Amish community, who live in the Pennsylvania and Ohio/Indiana areas of the USA. They originated from the Swiss/German borders and two waves of migration happened, in 1737 and 1815. The Amish population keep good genealogical records and tend to marry within the existing communities. There are 4 types of HSP in the Amish which are not found elsewhere. Given the records they can trace the current population back to the original migrants, and one person out of a couple carried a recessive form of HSP. SPG20 is one of the types found in the Amish. In this type one C in the DNA

becomes an A, the result of which is that no protein is made.

There are 13 HSP genes which are known to feature in at least one other condition. Drugs for other conditions with similar nerve problems could be looked at for treatment trials.

The work that Prof Crosby is doing at Exeter is to try to develop a blood test for HSP. Such a test may be able to prevent other clinical tests being done. If a test can identify a gene which is different then this can give information on: what has gone wrong, opportunities to improve the molecule, and help to develop a treatment.

Although HSP is a neurological condition there is a biochemical process. In order to develop a blood test, it is a question of identifying the pathways that are affected. Such a blood test would look for biochemical signals and, if successful, may be able to test whether people might develop HSP.

The issue with genetic testing is that some parts of DNA are more susceptible to change than other parts. Genetic tests on two people with the same genetic mutation would not, for example, prove that they are related to each other (they may be related some generations back). Tests will show a number of changes, but it is not always clear which change gives rise to HSP. With analysis of family trees this can help, and if a genetic change is identified to cause HSP with certainty, then this can be added to an HSP panel test.

The Caucasian population has been studied more than other populations and so there is more certainty on which genes cause which conditions. Genetic tests from people from other backgrounds are more difficult to interpret as there is less data available.

HSP Falls Study Results

Rebecca Chapman

Rebecca is completing her dissertation at Plymouth University, looking at the characteristics of falls and predictors of falls in HSP. She gave us an overview of the results obtained so far.

Rebecca outlined her approach - One of the main problems identified by a patient group last year was falls. This a self-reported study, i.e. participants in the study report things that occur to them rather than being quizzed about things. The study is a two stage approach. Participants firstly describe details about themselves and recall any falls that have happened in the past, and for the following three months participants record falls and send details in to Rebecca. These stages are the retrospective stage and the prospective stage. Rebecca had feedback on the approach through the HSP group meetings in Ashburton, Devon.

There was an initial trial with 5 participants, and the members of the group were recruited to take part. There were around 70 who expressed an interest, with 59 participants in the retrospective study and (at the time) 47 completing the prospective study. Rebecca gave us details looking at the results of the retrospective study.

The balance was 28 female and 31 male, with an average age of 60 (standard deviation 14 years). On average participants had had HSP for 25 years (standard deviation 17 years). 15 participants have SPG4 and 7 have SPG7.

Two thirds of people have fallen at least once, and just over half of people had fallen more than once (32 people). On average there have been 2 falls per person. 86% of falls have occurred indoors, but Rebecca didn't look at the

proportion of time spent indoors and outdoors. Of the indoor falls 21 were unable to get up unaided. 2/3 of people got a family member to help them up, 1/6 of people used someone external to help them up, and 1/6 used both family members and external help. Of those using external help 3 called a paramedic to help them get up.

Around two thirds (64%) have injured themselves with falls. Whilst most injuries are mild, and most are on the hip, around half injured themselves in multiple locations.

Rebecca looked at the data given by participants to examine possible predictors of falls, with the most likely ones being age and use of crutches. Most participants were aged between 55 and 65 with an average age of HSP onset of 40 - i.e. there has been some mobility impairment due to HSP.

It is known that some medication makes people drowsy. There was an average of 4 medications per person. The results were that this is a possible predictor, but were not statistically significant.

Co-ordination was also examined, as participants are frequently need to use their arms to help sit/stand, but again, these results were not statistically significant.

Looking further at the detail, falls indoors were often associated with everyday activities - cleaning and using the stairs. People on crutches tended to be more mobile than others, and younger.

Looking at the future, issues could be helping people to develop a falls strategy, giving both patients and family members falls training, and investigating falls aids. Rebecca mentioned paraladders and there are various youtube videos of people using this.

How does this study help?

- * It provides evidence of falls with HSP, and the report should open access to existing therapies
- * It sets out a strategy for improvements and training to reduce the risk of falls (i.e. to stop falls happening in the first place)
- * It helps people look at changes they can make - perhaps balance training or modifying doses of medications to alter the balance between stiffness and the number of falls
- * It gives evidence that people need to be taught how to get up, or aids to help themselves to get up.

Rebecca noted that the average NHS charge for an ambulance is £1200, so giving aids or teaching for people to get themselves up, which would reduce the number of ambulances going to help people, could be a cost effective for the NHS.

Update to PARCC study Prof Jon Marsden

Prof Jon Marsden gave a brief update on happenings with the Physical Activity in Rare Conditions Collaboration (PARCC) study.

Readers can read my blog post on the initial meeting back in January 2017 here: <http://hspjourney.blogspot.co.uk/2017/02/physical-activity-in-rare-conditions.html>. Jon said that the group comprised Huntingtons Disease (HD), Spinocerebellar Ataxia (SCA), Muscular Dystrophy (MD), Progressive Supranuclear Palsy (PSP) and, of course, HSP.

The researchers leading the work are experts within these conditions and associated symptom relief (e.g. physiotherapy). There are many

similarities in the symptoms of these conditions, and the approach is to develop an approach which works on these symptoms.

They are aiming to use the various support groups to map the different practices, working out what is done and how it is done. They are investigating potential physical activity rehabilitation options to deliver outcomes, working out how they will measure those outcomes, and working out how they would implement those options.

Jon referred to Rachel Chapmans falls study, wondering if they could look at walking style to reduce the risk of falls. It might be possible within the PARCC remit, or it may be for a different study.

Living with the enemy Robin Paijmans

The last presentation at the AGM was called Living With The Enemy: Psychology of Chronic Conditions, by Robin Paijmans. Robin is a psychologist looking at human behaviour, and how changes in behaviour affect life.

With chronic conditions there are both physical changes and mental changes. There are often different tools which can help to cope with the physical changes, however the issues around changes in mental are that these require a change in the way that we think. With chronic conditions there are three questions:

How do I cope?

How does my family cope?

How do professionals cope?

Robin observes that medical professionals are often compulsive problem solvers, they want to fix things, and often with chronic conditions there are no cures or

solutions, which presents a problem for the problem solver.

Generally, people deal with problems either by moving towards the problem or moving themselves away from the problem - the approach/avoid.

When we visit healthcare professionals we ask questions like: Will they know about my condition? Can I trust what they say? Will they help me? Are they behaving appropriately (listening/giving attention/etc.)?

The healthcare professional may have questions of their own: Will the patient know how I feel? What will I do? What if I don't know what to do?

Robin then described a "brain hack" which people may be able to use at times that they are not feeling happy. It is a mindfulness technique. I'll write the points as a list of bullets:

- Pick something which is worrying you
- Choose a number between 1 and 10 to represent how much this worries you (1 is perfectly OK)
- Imagine the issue as an object in the room/space that you are in. Think how it looks:

What colour is it?

What shape is it?

What size is it?

What texture does it have? (e.g. rough/smooth)

What temperature is it?

How heavy is it?

Where is it in the room/space that you are in?

- Now imagine moving the object to a place outside the room/space.
- Now imagine moving it a couple of miles away.
- Choose a number between 1 and 10 to represent how much this worries you (1 is perfectly OK)

The second number should be smaller than the first number, and you have mentally shrunk the problem. Note - if you cannot mentally move the object away from you then try changing its colour/size/weight/texture instead.

Robin discussed values, in that these values are a compass heading to guide you towards things that you want to do/achieve/have. The values themselves are not the destination. However, some things that we do to move away from discomfort can also move us away from our values. Once you have identified your values and being working towards them this can give you the strength to face threats. There are lots of things which we can do every day to reinforce our values.

Robin mentioned two books:

Feel the fear and do it anyway by Susan Jeffers and Living with the enemy by Ray Owen.

Thank you to Adam Lawrence for these very thorough and accurate accounts