



HSP Newslink

The Newsletter of the Hereditary Spastic Paraplegia Support Group
Registered Charity No. 1109398

Issue 36 – September 2015

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

Our gift to you



Some of the children at Dame Ellen Pinsent School

"I have had the honour and privilege to work for this wonderful school over the last 15 years. It is a Primary Special School in Birmingham for 135 children with moderate learning difficulties, but sadly I have to leave at the end of this academic year.

I was diagnosed with HSP just over a year ago, but it has progressed rather rapidly, and it has been decided that I now need to retire. You can understand my emotions as you read on - **these are my little stars** - I will miss them all so much!"

June Masding Region 7 – Midlands

Our Gift to you

From all the children at Dame Ellen Pinsent School

We have had a fun day in our school today! Our Head-Teacher, Mrs. Allen, told us that we didn't have to wear our school uniforms for one special day, but could choose anything we wanted to wear.

To be able to do this, Mrs. Allen asked that we each brought a little bit of money into school, and when it was all collected in, we could send it to a special charity.

We raised an amazing £165!!

Mrs. Allen then told us all about HSP, and how people can become very disabled with it, and that June, our Resource Technician has this condition.

We wanted you to receive our special gift, which we hope will be able to help everyone in some way with this disability.

With love from us all xXx

Chairman's Column

I'll begin my column with an apology. Unfortunately I am unable to attend the Region 4 meeting in Ashburton on October 3rd but David Harris has kindly agreed to say the necessary welcoming words and introduce our speaker. I believe I have a good excuse!! My elder daughter Jade is running the Bournemouth half marathon, raising money for HSP. Her boyfriend Theo is doing it with her and a good friend of my younger daughter is running the full marathon, also for HSP. I will be there with my family and some friends, cheering them on and trying to collect a few extra pennies. Jade has a page on JustGiving.

On 22nd May a great evening was enjoyed at my local pub, The Old Thatch. Following further fundraising, mainly as a

result of collection buckets on the bar, a local member was presented with a Power Chair. Live music and good food was enjoyed by all who attended. Management, staff and customers at The Old Thatch have been amazing with their support and I can't thank them enough.

There is also another pub down south fundraising for HSP called The Amberwood. You'll read an article later in this newsletter called Amberwood Adventures which details just one activity they have been involved with. Huge thanks to Gill McEwen and the team from The Amberwood for their support.

At the beginning of June I had the privilege to attend an international HSP meeting in Madrid. This was attended by the leaders of HSP organisations from Australia, USA, The Netherlands, France, Spain, Denmark, Italy, Switzerland, Sardinia and the UK. It was a pleasure getting to know everyone and improving relationships between our organisations.

Dr Rebecca Schule from the universities of Tubingen and Miami delivered a presentation titled: Establishing effective treatments – Patient registries and clinical trials.

Rebecca is involved in clinical work and sees patients with HSP. She is also very involved with genetic research.

She informed us of a new alliance team who will be involved with treatment and research of HSP. This team involves neurologists and researchers from several countries, including Germany, France, USA, Canada, Belgium, Austria and Professor Henry Houlden from the UK. The purpose of the alliance is advancing treatment in HSP and PLS. Its exciting news to have such a team working together for our cause. The individual HSP Support Groups will be able to help this alliance by assisting with the creation of a global HSP registry which of course will be useful for clinical trials when new drugs/treatments become available. For this reason we are constantly working on improving our

database and the help of John Mason with this has been invaluable. I will stress that no members details will be passed on to any other organisation without full consultation and permission.

At the meeting it was very interesting hearing from the individual Groups. An interesting highlight for me was from the Italian group and learning about HSP in Sardinia. There's a town in Sardinia called Nalvi with an exceptionally high concentration of HSP. The town has 5,000 residents but individuals are commonly seen walking with a spastic gait. It was suggested that individuals with HSP who struggle to come to terms with the condition sometimes visit Nalvi because HSP is so common there and it almost isn't seen as abnormal.

Bizarrely, an item on the agenda in Madrid was 'Potato Pants'. Many of you will now have come across the term 'Potato Pants', but for those who haven't, the following article titled 'Potato Pants' is written by the Austrian lady who first came up with the idea and explains all the thinking behind it. It was felt that Potato Pants events could become the 'ice bucket challenge' for HSP, an idea that appealed to the international Groups present. We are also considering developing a 'Potato Pants' logo which every HSP group adopts as a sub logo. This would show that we are all singing off the same hymn sheet and it would also generate further questions and discussion.

I have recently taken part in a Potato Pants event which was great fun and raised a few pounds. Some of you may have seen some of the pictures and videos on Facebook. I'm thinking of producing a further short video and nominating one or two of you to do similar.....be warned!

I can't end my column without once again thanking the latest very valuable addition to our committee, Adam Lawrence. Adam has not only contributed his regular column but also provided the excellent summaries of the presentations from this year's AGM

Ian Bennett

Potato Pants

So where did **Potato Pants** come from?

The idea that ALS, a rare disease that many people were becoming aware of because of the ice bucket challenge, was fantastic. At the same time, it was frustrating, because HSP is also rare. Here was the thinking: We are not famous. We don't have famous people pouring buckets of ice water over their heads for HSP so what can we do instead? How can we harness the same power?

During the height of the ice bucket storm (so July 2014), when every hour a new celebrity accepted and filmed their challenge, I saw my friend, Constanze Posautz, each day, for 10 days, because our daughters took part in a circus training camp. Each day, I became increasingly freaked out, in the sense of, we have to do something, what can we do? In the midst of these 10 days, I went hiking with my daughter and her friend. As a reply to their question about why I had to take a break every 5 minutes or so, I told them about feeling like I was carrying 10 kg. of potatoes on my legs. I told Constanze this. From that innocent line made while hiking in July, Constanze came up with the idea: "Let's have potato races at school." We work in a school. We have access to a huge community. That is harnessing power.

Cut to September 2, 2014, the first day of school. Constanze made an announcement to our faculty that we wanted to have potato races at our school festival, happening nearly exactly a month from that date.

It's fantastic to have an idea but how do you get to the end result: raising €6,000 for research and 350 participants having fun? You make a promotion video, you try out (and fail and laugh until your cheeks hurt) prototypes for the potato pants and you enlist the help of many people: the sewer of the pants, the obstacle course designers, the film editor, the M.C. who keeps the event going, the wranglers (no

horses though), the collectors, and of course, the participants.

Why is Potato Pants such a good idea?

- 1) I am writing this nearly a year after our initial event. It lives.
- 2) It's so much fun
- 3) It gives those who are wearing the pants a sense of what it might be like to have such heavy legs. You can liken it to a virtual experience although it's clear that at the end of the day, or the end of the race, you can easily take your pants off.

<https://www.youtube.com/watch?v=56XdjtrpZlo>

Potato Pants – Together against HSP

<https://www.youtube.com/watch?v=K80Q8BnU-V0>

Potato Pants – This was the run.

*Lori Renna Linton, Klosterneuburg,
Austria*

Adams Research Column

This issue's research column covers 2 things, firstly a couple of surveys you might like to complete, and secondly some recent news stories which have caught my eye. Most of the news stories are not directly HSP related, but I've included them because I found them interesting.

UK Rare Disease Survey

Rare Disease UK

<http://www.raredisease.org.uk> last did a survey which was published in 2010 and have one open now for completion by anyone who cares for, or is affected by, a rare or undiagnosed disease.

The survey takes between half an hour and an hour to complete, depending on how much information/detail you put in. The survey is open until September, so there is still time for contributions. Completing this survey adds another voice to the collection and may help shape policy/strategy in the future.

<https://www.surveymonkey.com/r/rduksurvey>

My own survey: modifications around the home

Members may recall I have previously surveyed about mobility, symptoms and medication. I am undertaking another survey this year, focussing on modifications made around the home which I would welcome members to spend a few minutes completing. People are welcome to provide as little or as much information as they want. I will collect answers for the rest of the year and report on my blog 28th Feb 2016. The link to the survey is:

<http://www.surveygizmo.com/s3/2310166/d1b822668a8e>

Dalfampridine/Ampyra

Back in April there was a new study reported which showed that Dalfampridine/Ampyra may be a treatment for people with HSP. A trial was undertaken in France with 12 people taking Dalfampridine/Ampyra for 2 weeks. 6 of these patients had improved mobility at the end of the study. The drug was well tolerated, and is currently approved for use for people with multiple sclerosis.

Non-invasive spinal cord stimulation

There was an article a few months ago about how it is now possible to deliver electrical stimulation to spinal cords by placing electrodes in the right place on the back. I'm not sure what the potential for this for HSP, given the degeneration along our nerves, but it might be one to watch out for.

<http://www.nih.gov/news/health/jul2015/nibib-30.htm>

Artificial neurons

On a similar theme, an artificial neuron has been created in the lab which has been shown to mimic a real one. The article talks about miniaturising and implanting these in the body in the future, with reference to neurological disorders. Again, I don't know the potential for HSP, but another one to

keep an eye on.

<http://www.kurzweilai.net/swedish-scientists-create-an-artificial-neuron-that-mimicks-an-organic-one>

Getting Fit

I read with some interest a blog on the BBC website about difficulties getting fit when disabled, which may be of interest to some readers: <http://www.bbc.co.uk/news/blogs-ouch-34071751>.

New Shoes

Lastly, Nike have recently launched a range of shoes for disabled people.

<http://news.nike.com/news/the-flyease-journey> This shoe was designed for people with cerebral palsy, but I wonder if its not the start of a new thing. It would be good for shoes to be designed with HSP in mind.

Congratulations!!

Neil and Caroline Brear
Saturday 13 June 2015



I would like to congratulate Caroline and Neil on their Wedding Day. Neil is a member of the HSP Group and the two of them have recently attended meetings in the North West.

Emma Saupe, Mark Tomlinson (Todd), Christine Snow and myself were fortunate enough to have been invited to share their day with them. Unfortunately Christine

was unable to be there but her thoughts and best wishes were. It was a lovely day and enjoyed by all.

Caroline was a bit concerned about what the weather was going to be like as the week had been lovely and forecast rain for their day. It was a bit dull before the service but when it came to the photos being taken the sun was shining and that made their day.

The next day they were heading off for their “Honeymoon” in the Dominican Republic, two lucky people.

I along with everyone else would like to wish them both all the best and many years of happiness together.

Debbie Best – Region 10

HSP Group Funds

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. Completion of one year’s membership is a requirement for applicants. If interested, please request a grant application form from a committee member.

Useful Information

Plymouth Research

Volunteers needed for research

We are looking for volunteers to take part in case studies investigating the use of insulation to prevent temperature related changes in people with Spastic Paraparesis.

If your walking is affected by cold temperatures you may be interested in taking part?

Case studies: Participating in the study will involve wearing insulating garments on your lower legs for a time period over the autumn/ winter 2015/16. After an initial visit to you by the research team you will complete questionnaires and wear an

activity monitor. Stamped address envelopes provided to send information in on a weekly basis.

For further information please contact:

Amanda Denton
School of Health Professions
Faculty of Health and Human Sciences
Peninsula Allied Health Centre
Derriford Road
Plymouth
PL6 8BH

Tel 01752 587995

Email: amanda.denton@plymouth.ac.uk

Amberwood Adventures

We would like to say a special well done to Gill McEwen, Business Partner at the Amberwood Inn and her group of adventurous friends who celebrated milestone birthdays by climbing and walking across the O2 in London at the end of June.

Gill raised £658.35 for The Amberwood Inn's local charity, Hereditary Spastic Paraplegia (HSP). The Amberwood Inn at Walkford on the Hampshire / Dorset border very kindly agreed to raise funds for HSP over a 12 month period. A huge well done to all involved!

If you would like to make a donation to Gill's chosen charity HSP, please visit www.justgiving.com/HSPGroup



Members' Letters

HSP - Work life balance

Work is crucial for anyone with a disability - not least somebody with HSP.

This view has been, effectively, endorsed by the Government, and by the academic Carol Black.

Professor Black, the Principal of Newnham College, Cambridge co-authored (with David Frost) a report looking at the levels of sickness absence.

In the report, delivered to the Department of Work and Pensions four years ago, the role of work was emphasised.

Fundamentally it increases a person's self-respect and boosts their income.

It also saves a great deal of money for the taxpayer.

The levels of mental illness, like depression were (and are) extremely high in Britain and they have become a major cause of not participating in the work-place.

This leads me on to another, more personal, point.

What if you want to return to the workforce but are only offered jobs below what you have been used to?

What does that mean for your self-respect and levels of depression?

I was diagnosed with HSP four years ago and, as a broadcast journalist, work became extremely difficult.

You have to move around a lot collecting interviewees or transferring from one broadcasting point to another.

After leaving the BBC the only jobs I could find were way below my perception of what I was worth.

That is not to denigrate in any way other jobs, but the reality is that my sense of self-worth was badly affected and it contributed to a severe depression.

In fairness to Professor Black she has stressed that a 'poor job' is worse than 'no job'.

But it seems to me this needs to be examined more fully.

There are many roles a person with HSP can fulfill and contribute to society as well as his or her own well-being.

But if it is important for coping with this, or any disability, to engage in work, it cannot be right that any work on offer might prompt depression, which is anyway a major part of the condition.

Phil Parry

Tramping in the Great Outdoors!

I am lucky to live near High Elms Country Park, Farnborough, in the London Borough of Bromley, North West Kent. It is an area where Charles Darwin and his friend John Lubbock used to admire the area's wildlife during the middle and latter 19th Century. A few years ago Bromley's Country Parks team were able to secure funding for the purchase of two 'Tramper' all terrain scooters. These scooters are available free to anyone who wants to borrow them. A circular route was marked out with gates which could be easily opened for Tramper use.



I used to walk the route before my mobility problem developed. It mainly follows a mixed deciduous woodland and an area of grassland. Much of the vegetation is controlled during the winter months by a flock of sheep which keep the grasses and sycamore shoots low. It is called the Orchid Bank because of the many orchids that grow there. Now I can access the site readily by Tramper. I have since found out that there is a national 'tramper' group –

www.disabledramblers.co.uk. I have not been on one of their 'tramping' days yet, but they seem a well organised team.

I love the countryside and am a very outdoor person. Indeed I used to manage an environmental education centre near Dartford for K.C.C. where I led walks for mainly primary children, taking them along and in the River Darent carrying out river studies and in the village looking at local history before I retired and my walking became difficult. Borrowing the 'all terrain' scooter at High Elms has given me a lot of joy and I recommend getting out in the outdoors to anyone.

Gary Cliffe

My Garden

Well, another summer has gone. The good hot days we did have I spent in my garden.

I have plenty of colour in the raised beds. There are Dahlias, Cosmos, Gladioli and Hydrangeas among some bedding plants. I also have a new perennial Salvia, the flowering stems are grey then blue appears. I would recommend this plant in any garden, they are gorgeous.



The rose bushes in my front garden have enjoyed the sun when we did have it. Masses of flowers. I won 2 first prizes for them at the local horticultural show.

Now I shall start planning for next spring. I think I shall include some Lillies in pots. I have started collecting seed heads and putting them in paper bags.

I couldn't have done gardening to look after the plants if I didn't have raised beds. I sit on the top and hoe. I am now 80 with HSP - I don't think I do so bad.

Barbara Jones .

Regional News

Beach Hut Opportunity

A fully accessible beach hut near Boscombe pier on Bournemouth sea front is available for members to use between October 11th and October 23rd. It is larger than a standard beach hut and has a gas ring for cooking. Next door there is a large disabled toilet/wet room which includes a shower. If you are interested in making use of this please contact Lorraine Saupe on 01425 627 410

Region 5 met at the Oaks Restaurant, Premier Inn, Norwich on 12th September.

We were lucky to have Rekha Srinivasan, Senior Physiotherapist at the Norfolk and Norwich Hospital, to come to give us advice. She told us about correct posture, walking correctly with our walking sticks and other helpful ideas. She said at the end of the afternoon that she would send us leaflets giving us suitable exercises.

All members enjoyed the meeting with lots of laughter too.

We are meeting again next March.

Barbara Jones, Region 5

Forthcoming Events

Region 4 get together

Saturday October 3rd 2pm onwards
The Dartmoor Lodge Hotel, Ashburton
Call Ian Bennett on: 01202 849 391

Region 9 Meeting

Saturday October 10th 2-6pm
Meeting Room
St Helens South Premier Inn
Eurolink, Lea Green, St Helens, WA9 4TT
Call Irena Pritchard on 01524 261 076

Afternoon tea Regions 1 & 2

Sunday November 15th 3pm – 6pm
The Clockhouse Milford
Call Jane Bennett on: 020 8853 4089

Colchester Meeting

Sunday October 11th 2.30pm – 4.30pm
Feering Community Centre
CO5 9QB
Contact Hilary Croydon: 01284 728 242

Region 7 Meeting Birmingham

Saturday October 17th 2.30pm – 5.30pm
Kenrick Centre in Harborne
Contact Penny Cohen: 07818288738
Email: pennycohen57@hotmail.com

HSP ANNUAL GENERAL MEETING Saturday 20th June 2015

Minutes

Apologies for Absence were received from Tony & Angela Barnicoat, the Slavin family, Simon Hubbard, Neil and Caroline Brear, John and Carole Moore.

The Minutes of the 2014 AGM were agreed to be a true record: proposed for acceptance by Caroline Begg and Penny Cohen.

Committee Reports:

Stephanie Flower welcomed new members to the meeting and hoped to be able to talk with them before the day's end. Help Line was still active with wide variety of call patterns - sometimes quiet weeks, sometimes several calls per day. She recommended members to use the Unite/Facebook site where a lot of members got support from others.

Dave Harris presented a summary of the groups status and recent activities:

- we have around 350 members spread out over most of the UK
- our main aim is to support our members - other HSP groups concentrate on research
- sharing information and communicating between members is a main activity

- a strong network of regular meetings is now established - thanks to volunteer organisers, many members can get to a meeting with less than an hours travel
- Group now over 25 years old - annual income varies from £4K to £12K, reserves last year £33K
- have given 50 member grants and three research grants up to end of 2014
- fundraising is increasing with more sources and often larger donations - thanks to all who contributed to group funds by whatever means.

John Mason presented the groups accounts for the year. Copies of the summary had been handed around to all attendees. John first gave thanks thanks to Sonya Mason for her assistance through the year and to Theo White as Auditor. After an opportunity for questions, adoption was proposed by Terry Duffy and seconded by Debbie Best. This was approved by the meeting with a show of hands.

Election of Officers:

Chairman, Ian Bennett was proposed by Mark Tomlinson and seconded by Stephanie Flower.

Treasurer, John Mason was proposed by Richard Williams and seconded by Della Brookman.

Secretary, Dave Harris was proposed by Caroline Begg and seconded by Terry Duffy.

Committee Member Simon Hubbard was proposed by Mark Tomlinson and seconded by John Mason.

Helpline, Stephanie Flower was proposed by Della Brookman and seconded by Richard Williams.

Committee Member John Moore was proposed by John Flower and seconded by Des Williams.

Committee Member Adam Lawrence was proposed by Penny Cohen and seconded by Della Brookman.

All the above appointments were approved by the meeting.

Any Other Business. No items were raised.

Speakers:

The AGM was followed by talks from two speakers:

- Prof. Henry Houlden, National Hospital for Neurology and Neurosurgery;
- Dr. Cahir O'Kane, Department of Genetics, University of Cambridge.

A Summary of the AGM Presentations

The differences between pure and complex HSP - Prof Henry Houlden

Professor Henry Houlden works at the National Hospital for Neurology and Neurosurgery, in London. His presentation covered three areas:

- **The differences between pure and complex HSP**
- **Genetics and genetic testing**
- **Shared network and genome sequencing**

Prof Houlden reminded us that HSP is generally described with increased stiffness, muscles becoming weaker and a slow progression. For pure HSP the three main areas are legs, bladder and the back. Pure HSP can also affect spasticity in other areas (including the upper body), but if there are other symptoms present then this becomes complex HSP. There is some variation in what symptoms might be expected, but these might include balance. A few videos were then shown with some examples.

If there is not a clear family history then an MRI scan of the brain and the spinal cord can help the diagnosis. For example, if there are some symptoms and compression in the spine this is not HSP and can be treatable. If the initial scan reveals that HSP is a contender, then blood can be sent for genetic testing, and tests on parents/siblings (etc.) can help in a better identification.

The scan would also show up the corpus callosum (a thick band of nerve fibers that connects the left and right sides of the brain allowing for communication: transfer of motor, sensory, and cognitive information). A thin corpus callosum occurs in some complex forms of HSP. For patients with complex HSP a repeat scan every 5 years would help to measure progress/change over time.

Some patients also experience problems with their feet - for example blisters and high arches (blisters can be a problem for people with HSP and diabetes

<http://www.nhs.uk/Livewell/fothealth/Pages/Diabetesandfeet.aspx>) Examination of these can lead towards a diagnosis.

Prof Houlden indicated that about 30% of HSP diagnoses are not genetic. Simply, not all of the genes which cause HSP have been identified yet. There are some 25,000 genes in the body, and currently (Feb 2015) 74 different HSP genes have been identified. Getting the correct diagnosis can give you confidence - there is confirmation of what you have, and the course of can be anticipated - including potential complications.

The most common pure HSP types with dominant inheritance are SPG4, which accounts for about 40% of cases, SPG3A which accounts for some 2-3% of cases and SPG 31 which accounts for some 1-2% of cases. The most common complex HSP types are SPG11, SPG7, KIF5A (also known as SPG10) and SPG 35. Prof Houlden did not give prevalence information.

Types of HSP which have a dominant inheritance pattern carry a risk of 50% of passing HSP to the next generation (assuming only one parent carries the gene). Each child has an equal risk of inheriting the gene. For types of HSP which are recessive, the risk of passing the gene to the next generation is tiny, unless both parents are from the same family. Tests can be done prenatally, by CVS (Chorionic villus sampling) at about 10.5 weeks or by amniocentesis at about 16 weeks - however both tests carry risks of miscarriage and/or infection. Alternatively, Pre-implantation genetic diagnosis can be used (like IVF) to check, however this needs funding and the permission of your GP.

Prof Houlden then talked about management of HSP, by reference to Spastin (SPG4). The main points were:

- Physiotherapy and orthotics
- FES (functional electrical stimulation)
- Baclofen
- Self catheter and Detrusitol (noting Detrusitol works best if the bladder empties fully)
- High walking sticks (these result in a more upright walking position)
- Hip and knee replacement later in life.

On FES, it was observed that relatively few people were using FES 3-4 years ago, and the uptake of this has increased more recently. FES is available for HSP in London, Sheffield and Salisbury.

Prof Houlden talked briefly about bowel issues. The effects are not predictable, and this cannot be used in differentiating between pure and complex HSP. Bowel issues can usually be treated.

Prof Houlden then talked about a potential link between HSP and dementia. There can be some cognitive problems later in the progression of HSP, including SPG4. Sometimes the brain has to focus on walking which it does at the expense of other processing. These problems may

also occur as a result of fatigue - which increases as it becomes harder to do more, or they may be a side effect of medication being taken. If there are mental problems with HSP then tools to help cognition may be of limited benefit. In this case it is better to treat the condition.

A European HSP network is (being) set up, with the objective of sharing information, particularly clinical problems, databases of patients and blood/MRI results. This can help research and the treatment of HSP. The network is looking for biomarkers for HSP which could track the progression of HSP, the effects on bones and/or the benefits of drugs. There is more on biomarkers here: <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3078627/>. The shared database of patients can be used to help find candidates to take part in drug trials. It is likely that drug trials will be targeted to specific genes, and it becomes necessary to find a sufficient sample to determine the benefits of the drug.

Lastly, Prof Houlden covered getting access to services. Your GP is able to give a referral, and appointments can be booked using the choose and book system. He advised seeing a specialist in clinic once every few years, noting that it can be difficult for a local neurologist to be knowledgeable of rare diseases, but there is more money available for rare diseases at the moment, and this is leading to increased awareness.

Insights into HSP from Drosophila - Dr Cahir O'Kane

Dr Cahir O'Kane works at the Department of Genetics at the University of Cambridge.

Dr O'Kane's talk was around the study of HSP in Drosophila - also known as fruit flies. The first aspect was why we do this. Firstly, they have a short development period, going from egg to adult in around 10 days. Also they are small, so one wall of the room at Cambridge can hold 15,000 flies. They have 4 of these rooms and there are several groups working with these flies.

Flies had a common ancestor with humans around 600 million years ago, which means that flies share about 80% of their genes with us, including the HSP genes. The team are able to breed paraplegic larvae, and as they age they lose the ability to climb. You can see this here: <http://labs.cellbio.duke.edu/kinesin/WTKHClarvae.html>.

These flies have axons in a similar way to humans. Nerve fibres or axons are typically up to 10,000 times longer than a cell body, and cell engineering is needed to maintain the axons. In HSP the longer axons breakdown first.

We have around 20,000 genes and the fly has around 14,000. Over 70 HSP genes have been cloned, and more continue to be found, around one every couple of months. Each gene disrupts a particular protein. Figure 2 here http://sp-foundation.org/file_download/510e252f-c9ec-484c-a90b-8f95372a499f shows which HSP genes affect which parts of the nerve fibre. Within this figure tubular endoplasmic reticulum (ER) can be seen, which forms a network within the cell. The axons in the fly have a similar ER shape.

Some of the HSP proteins have roles in the integrity of axonal ER in any organism. The research on the flies shows that if a fly is bred with the removal of one of the SPG/HSP genes there are mild effects, and with the removal of multiple HSP genes the ER becomes fragmented - and the nerve no longer functions as intended.

What does the future hold? It is possible that the functions of axonal ER could be altered by drugs, however at the moment it is not known which of these are important to maintain the axon.

The researchers have shown that it is possible to genetically reverse HSP in flies, but they are some way off being able to do this in humans in the embryo. Two of the issues putting this into practice are that there is a much larger risk of miscarriage, and that this is prevented by

biomedical regulations (ethics). The researchers don't know all about neurons yet - if the letters in the DNA were printed in a book the human genome would take around 2000 books to print it all. Each gene within the DNA is several thousands of letters in length.

Dr O'Kane mentioned the Tom Wahlig Foundation (<http://www.hsp-info.de/en.htm>) who research HSP and promote an international network of HSP researchers.

New Members

We welcome the following new members:

| | | | |
|---------------------------------------|---------------------------------------|--|--------------------------------------|
| Chris James Somerset Region 4 | Hugh Adlam Wareham Region 4 | Louise Larkins Cambridgeshire Region 5 | Robert Styles York Region 10 |
| Daniel Kitchener Herts Region 3 | Karen Webb Altrincham Region 9 | Paul William Wirral Region 9 | Steve Barister Luton Region 3 |
| Helen Osborne Wirral Region 9 | Kelly Atkin Portsmouth Region 1 | Rob Mulcahy Colchester Region 5 | Sue Kirkman Leicester Region 8 |

If you are interested in contacting the new members listed, please contact the relevant area co-ordinator, or the membership secretary, who can then supply the necessary contact details

Useful Contacts

| | Email | Telephone No: |
|--|--|---------------|
| Ian Bennett - Chairman 37 Wimborne Road West, Wimborne , Dorset BH21 2DQ | bravoechoonovember@btinternet.com | 01202 849 391 |
| 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 – Memb Sec/Reg 4 co-ordinator 37 Wimborne Road West, Wimborne , Dorset BH21 2DQ | bravoechoonovember@btinternet.com | 01202 849 391 |
| Simon Hubbard – Research co-ordinator | si@sjhubbard.com | 01908 508 866 |
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