



Issue 17
May 2008

An introduction from the Chair Submitted by Carol Dobbins



As most of you will be aware from the last Newsletter, NPS UK have been working with GIG on creating a Family Route Map for those who live with NPS. This two year project concluded this year and the launch was held in London on the 5th March at the Wellcome Trust conference centre. Kath Lovatt and I travelled down on the 4th and shared a room in a hotel across from Euston station. We went out at night to meet with Melissa, the GIG communications manager and it was nice to catch up in an informal environment!

I was asked to be one of the speakers. It was a wonderful event with many interesting people in attendance, including pharmaceutical reps, medical professionals, sponsors, support group members and those who share rare genetic conditions also.

The Family Route Maps are absolutely wonderful and I think they will be an incredible source for many to disseminate throughout the UK on our visits to the hospitals, GP surgeries, and basically to "whoever really wants to read them", as Colin Clarke stated during the day. Colin is Karen Clarke's husband who shares NPS with us and he came along to the event also. I feel he summed it up well with his comment as that is exactly what we need to do. We need to be pro-active in our own care and also the care of others with NPS, by handing the information out each time we visit the doctors or medical profession. It was such a great support for me personally to see the familiar faces. Anna from GIG referred to us as the NPS Posse!!! Bill and Anne were, once again, showing their brilliant support for the charity and listened intently and participated in discussions on how we shall use this source of documented literature.

I feel it was a major triumph and I must confess to feeling a little sad that the project is completed, for now. I have let them know that we are interested in continuing with any further projects so I am sure it won't be the last dealings we have with GIG!

The Final report is now available to download in pdf version! It is a wonderful piece of literature and I am immensely proud to have been involved with the whole thing. I do believe it will be a great step in moving forward in gaining interest in NPS.

For information on the Route Maps and the Final Report document, please visit http://www.gig.org.uk/gig/familyroutemap.htm#FRM_results It would be great if you could continue to hand these out wherever you go e.g., GP surgeries, hospitals, physiotherapy departments, family members, etc...



GIG recently has been selected as one of the 10 Winners of the GSK Impact Awards this year. As part of there publicity GIG needed to provide a case study of work that it has carried out that has benefited its members. They have decided to use the Family Route Map as such an example, and as we took part in this exciting project, they asked me to give them a quote saying how I think having an FRM for NPS will benefit future patients that may contact us for help and information. This information is yet another way to promote our condition! (See below)

Most of GIG's member groups are small support groups for families with very rare genetic disorders, such as NPS UK, a support group for families affected by Nail Patella Syndrome (NPS). NPS is a rare genetic disorder that causes abnormalities of bone, joints, fingernails and kidneys. It is thought to occur in 1 in 50,000 people and due to the rarity of the condition, families often remain undiagnosed for several generations despite having been seen by doctors from a variety of disciplines. When families are diagnosed, they often experience extreme difficulty in accessing appropriate information, treatment and surveillance for their condition.

GIG recognised that this was an experience that was shared by families from many of our smaller member groups, and in April 2006 we started working on *The Family Route Map Project* to address the difficulties that families with rare genetic disorders experience in accessing information and services. We worked closely with six of our smallest, most under-resourced, member groups, including NPS UK, to produce a series of condition-specific *Route Maps* - guidance and advice to help families to access the information, treatment and services available to them. The *Route Maps* are now complete and are being used by families across the UK affected by the six rare disorders, including those with Nail Patella Syndrome. The founder of NPS UK described the completed *Route Map* for NPS as a "wonderful resource to guide individuals immediately to the professional services in their area relating to their specific complaint".

The Project has also developed a generic template that can be used by other Patient Support groups to enable them to develop their own *Route Map*.

Fundraising dates for the diary!

Jerry Crook has decided to take on a huge challenge to raise awareness and some funds for NPS UK. On the **10th May**, he will start on the edge of the South Downs in Sussex, and walk the 20km route! This charity walk is designed to offer enthusiastic walkers the chance to experience a breathtaking walk over the picturesque South Downs and generate funds for their chosen charity. I am incredibly touched that Jerry is willing to take this on. Tina, his wife, and all of us at NPS UK will be supporting you all the way! Let's just hope that the sun shines!



If anyone would like to sponsor Jerry for his walk www.just-walk.co.uk, please remember you can do this by using our Paypal account.

Alternatively, just send a cheque to NPSUK to the PO Box address.

Can I ask that if you know anyone who is willing to do something for a charity, to consider us? We really do need all the help we can get right now. My son, Stephen will be unable to take on any challenge this year as he will be undergoing surgery on his leg, so we will need to ask for as much help as we can get. He is upset that he will not be able to do the junior marathon, but if you know anyone who would like to take this or any other marathon on, please let me know.



Another brave soldier has stepped forward to offer his services to our charity. This man, Phil, is a neighbour of Lisa Ward (nee Threadwell) On the **22/23 June** he has decided to walk along the coast of Kent from Hythe to Herne Bay. It's carrying on from where they finished last year as they raised money for charity. The walk will be approx 55 miles and they are aiming to complete it in approx 24 hours - no sleep - just the odd break for a drink/sock change or two. He will get firm details of the actual route in the next few weeks - i.e. what town's we'll go through, checkpoints etc.

He will do the walk with 4 other guys from his work (RBS) and he has decided to raise some cash for charity whilst getting some blisters! RBS are great in supporting charity and last year they raised a significant amount- as RBS match what you raise. He has already committed himself to raising cash for a charity that they support in work; hospices for children and would love to raise as much as he can for whoever he can - and he would love to raise money for our charity as well. I will post some sponsor forms to him and a t-shirt to wear to raise some awareness. Lisa is keen in supporting him on our behalf, so I hope to update you on the results of this in our next newsletter in September.

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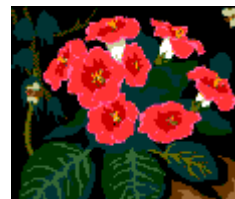


I received a lovely surprise when James Kelly, who's son lives with NPS, contacted our free phone line to let us know that his work, Hymans Robertson had raised some money for charities and that James was successful in

asking for some of the money to come to us. We received a cheque of £500. It was an incredibly kind gesture. James works in Glasgow, so I was eager to go along and say thank you in person. Many thanks to you James from us all at NPS UK.

Many thanks to those of you who continue to support our charity by using the collecting tins. This is a great way to generate funds.

Special thanks go to Paul and Julie Cummiskey, for placing the tins in their shop, Fresh n less in Rutherglen, Elders Bar in Tollcross and Bill and Anne Sneddon, who continue to send what they can. This helps with the day to day running costs of the charity and also goes a long way to making a difference! In the past few months, we have not been able to organise as many fundraisers as we usually would have due to a load of health problems, however, this money keeps us afloat and I can't thank you all enough for it!



Please continue to use the Everyclick.com service. It is an incredibly easy way to raise funds without needing to do a thing. Let's be honest, running a marathon is out of the question for many of us due to physical restraints, but we can do this simply. It requires no energy, and no money, just tell everyone you know to make Everyclick their home page on their computers at work and at home, add yourself as a supporter of NPS UK, and we continue to generate money for our funds!

Help us spread the word. Tell your friends about everyclick.com
The search engine that helps charity



Questions and answers

Welcome to our new Q & A. section!!! Please feel free to submit all your NPS queries and I am happy to put it to our knowledgeable professional bodies for their feedback. In this edition, I would like to introduce Dr Iain McIntosh, who has kindly agreed to answer all questions.



More about Iain...

Iain McIntosh has been studying the genetics of NPS since 1994. His curiosity was sparked by wondering what the connection might be between development of the knees, nails and kidneys.

His laboratory at Johns Hopkins University (Baltimore USA) worked to identify the precise location of the NPS gene (now known as LMX1B) on chromosome 9 and subsequently described over 100 of the known NPS mutations. By studying where LMX1B is working during development his group guessed that it may play some role in the nervous system. They went on to demonstrate that many patients with NPS have reduced sensation to pain and temperature in their fingers & toes.

He has also worked on the possible link between NPS and symptoms of Attention Deficit Disorder (ADHD). Prior to working at Johns Hopkins, Dr. McIntosh studied at the University of Edinburgh. Since 2006 he has been Professor of Medical Genetics at the American University of the Caribbean in St. Maarten.

"I have been asked to go into hospital earlier now with my neck as its is getting worse by the day. The question the hospital are asking themselves now is, whether the condition is linked with NPS, so maybe this would be worth asking in the news letter?? I have bulging discs in the neck and am going in to the neurological hospital on 28th May for surgical decompression of the discs." (Lisa, Essex)

"Neck pain. It is certainly possible that any skeletal problem COULD be due to NPS but it would be much more likely in the limbs than spine. There is no evidence than LMX1B (the NPS gene) plays a role in the development of the spine - only in the differentiation of some nerves within the spinal cord."

"I have a question - is there much info/research done regarding the weight aspect of NPS? i.e. it being hard to put on weight? I've lost quite a lot of weight and don't quite know what to do to try to bulk myself up - I know, lots of people would be jealous of me especially after having a baby, but I really am getting skinny! I spoke to my kidney doctor about it yesterday and he doesn't really know what to suggest apart from referring me again to the renal dietician to talk about supplements..."

Any bit of info would be gratefully received!" (Sarah, London)

"Weight gain/loss. It certainly appears that people with NPS are generally thinner than their peers. Why this is the case, is not clear. NPS patients certainly tend to towards poor development of some muscle groups (especially the quadriceps and biceps/triceps) and that will obviously affect weight. I am afraid I don't have a more definitive answer."

Hope those help. Always remember that not every symptom, ache or pain is due to NPS - the rest of us can get sore necks too!!!

Personal stories—Getting to know one another better. Have your say! Write today!

One idea that I have always wanted to pursue was a personal story page. I would love us all to get to know one another a little more. It is nice to put a face to a name and read the fact that there is so much more to us all than sharing a "disability". I am delighted to say that Laura has decided to be our first person willing to share a little bit about her life. Please keep sending me the stories, and pictures if possible.



A bit about me

My name is Laura and I have Nail Patella Syndrome. I sound like somebody at an Alcoholics Anonymous meeting, but perhaps this is because I am now starting to learn more about NPS and myself. Growing up I never thought of myself as a person with disabilities. I wasn't good at PE at school, but then neither were my friends. I certainly didn't have a Disabled Students' Allowance at university, but then I didn't need one.

On graduating with a degree in French, German and European Studies, I ventured across the Channel to start a career in the European Parliament. I worked for two MEPs during my two-year stint over there and in that time worked on various equality policy issues, but I was particularly taken by the disability lobby. I was determined to join the campaign to fight for the rights of people with disabilities. Why did I feel so strongly about this? I suddenly realised that anti-discrimination legislation could directly impact upon my life too. For the first time I started to declare I had a disability on job application forms. I admit that in the past I was ashamed to say I had a disability. Yet now I know that this can actually work in my favour.

For the last 2 ½ years I have been working for a disability charity. I have learnt a lot more about specific disability issues, but more importantly I have a lot more confidence in myself and my abilities. So much so that in September I will start the next phase of my career – I am going back to university to train as a secondary modern languages teacher. I am convinced that as a teacher with a disability I will bring more than just French and German to the classroom, but perhaps this is a discussion for another day!



Submitted by Kath Lovatt

Time flies – it’s not been long since the last newsletter and here we are yet again!

Back in March I drove down to London, picking Carol up from Heathrow Airport on the way and we made our way bravely into central London to attend the GIG Family Route Map Launch. Firstly I’m amazed we managed to find our way through the traffic, and we even ventured out at night and had a great meal out with the lovely people from the Genetic Interest Group (GIG) office staff, who were very hospitable. The next day Carol and I even ventured onto the underground and both being train virgins, I think we did exceptionally well to make our way to Oxford Circus! Just buying the tickets was an adventure on its own!

We made our way to the meeting, to see a few familiar faces there– it was great to see other NPSer’s at the meeting supporting this great project. Carol had to give a presentation on what NPS is and how the charity operates and how the project will help us, to which she did a great job. The rest of the presentations and group brainstorming on how the project could be developed was very interesting not only for the 6 charities it is supporting but for all charities across the country to help families, GP’s and genetic counsellors. All aspects of the chain were discussed. In my opinion, it will be a huge asset to us as Carol or myself are normally the first point of contacts either by phone or email where the patient has either self diagnosed or has been clinically diagnosed and quite often don’t know where to go from their diagnosis. The Route Map that has been produced is jam packed with useful links and information on what to do, along with support groups, geneticists etc – the list is endless, and has been compiled by people that attended the original Family Route Map Focus Group along with those that contributed at Chester last year. It is a godsend to any newly diagnosed case. All I can say is Bravo to all that contributed and a massive thank you to the staff at GIG for coming up with a fantastic project that will not only benefit NPS UK but has the potential to help every charity that supports a medical condition.

The 0800 line is still ringing away with requests for information and help, which is only a good thing. It always helps to talk about your experiences so never feel afraid to pick up the phone, no matter how big or small. I have had a lot of interest in DLA applications of late. I have had my fair share of experience with these so please don’t be afraid to ask for help if you need it, I am also informed that your local citizens advice are very well experienced in the forms if you need to talk to someone more professional. I also have had a great deal of email enquiries through the website and we get an enormous amount of hits, which goes up in volume every month. I personally feel the website is brilliant but I would like to see more things on there and would appreciate any suggestions – life stories and how NPS affects them; if you are newly diagnosed and want to share your thoughts – that would be great to see on there. Anyone that has had successful surgery regarding NPS – would also be good to see on there and we could even think about adding a database of UK consultants in area’s around the country as that is a very common question.. I would still like to develop a website especially for children and anyone that has experience of building websites and is willing to help please get in touch.

Till next time!!

Kath

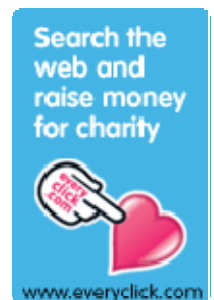


I would like to say a huge thank you to so many of you for helping with the Everyclick Charity Challenge. I am sure that many of you were clicking very hard as we managed to smash our target!!! To date, we have now earned £2,007.28 from this incredible organisation. Please continue to support us. The competition is now over, but as far as we are concerned, our challenge is to keep going and raise as much as we can! I hope to deliver the results of this competition in the next newsletter.

I have enjoyed working with the Everyclick team on many projects in the past few months. It was an honour for me to nominate Polly Gowers, the founder of Everyclick.com for the Beacon Prize for creative giving. I have my fingers crossed for this incredible woman!

I also want to say a huge thank you to those of you who have nominated me in Everyclick.com's competition to find Britain's Most Inspiring Fundraiser, I am very grateful. I am sure that there are many other people who share a similar passion and I feel incredibly humble by this. I could not do all this without the love and support that you give every day. I have a passion for our charity because I know that we work together, as a team, making a difference. I feel like the luckiest person alive as I have a wonderful NPS family..

I do not expect to win this competition, but I want you all to know how grateful I am that you felt the need to nominate me...whoever you are!!!!





Welcome to the Spring 2008 Finance report..

There hasn't been a lot of movement in our bank balance in the last 3 months, and we currently have just over £8000. I have just also received a very generous company donation of £500 from Hymans Robertson LLP, which was arranged for us by one of their employees. James Kelly. Thank you so much!

We continue to raise a steady stream of fund from the Everyclick website – thank you everyone who uses Everyclick as their home page and search engine. Remember Everyclick doesn't cost you anything at all, and creates income for NPS! Please try and use it, it does make a difference. The only costs we have had in the period are our BT phone bill for the Freephone line, £43, and the cost of ink for Carol's printer, £207, used to produce our newsletters..

We are now registered with the Charities Aid Foundation, which allows us to receive Give As You Earn donations. GAYE is a way of giving to charity direct from your salary – you get tax relief on your donation straight away, and the charity gets the full donation paid into their bank account. If you would like to know more about how you, your friends or your employer could be involved in this scheme, please let me know, or take a look at the website:

www.giveasyouearn.org

Thank you everyone for all your hard work so far in 2008, let's keep it going.

■■■ charities aid foundation

CAF

Make life simple with Give As You Earn

James Kelly, whose son lives with NPS contacted me to let me know about this scheme. James stated,

" I am in the process of setting up a "Give as you earn" donation to NPSUK. I thought that I would mention this as there seem to be quite good tax benefits for both companies and employees to donate to a registered charity."

If you earn a salary, or if your pension is paid via PAYE, making a commitment to giving doesn't get any simpler than this. With Give As You Earn – the UK's largest payroll giving scheme – you can give directly from your pre-tax salary, so money that would normally go to the taxman goes to charity instead. You just decide how much you want to give each month – it could be a couple of pounds, or hundreds or thousands – and who you want to give it to. Then your donation comes out of your pay packet before the taxman touches it. Your chosen charity benefits because they know they've got money coming regularly and can plan what to do with it in the long term.

You benefit because you don't pay any tax on the donations you make. If you're a basic rate taxpayer, a gift of £5 only actually costs you £4 – so you can pass the tax relief on to the charity.

[Find out more about how you can give more through Give As You Earn](#) Give As You Earn is incredibly flexible – you can give as much or as little as you like, in [four different ways](#), and change how you give and who you give to whenever you like.

To join Give As You Earn your employer will need to be running a scheme. For more information, please contact <http://www.cafonline.org/default.aspx?Page=7026>

If you have a question about Give As You Earn, please call them on 01732 520 019.

In January, I received a nice call from a local newspaper, the East Kilbride Mail, asking for permission to have my son, Stephen in the newspaper as the East Kilbride Icon! It came as a huge surprise, and Stephen accepted. He was chosen as he has managed to achieve his black belt in Tae Kwon Do despite his physical limitations.

The Newspaper article read:-

“Like the words “great” and “awesome”, the term “inspiration” is often used in print but is rarely reflecting of its true and accurate meaning.

However, in the case of young Stephen Dobbins from West Mains, there has never been a more appropriate example of someone whose determination can encourage others to overcome the odds. Stephen, 10, has ignored painful symptoms of the rare genetic condition, nail patella syndrome, to become an expert in the fearsome discipline of Tae Kwon Do. His success has amazed and impressed instructors in equal measure and culminated in the award of his black belt shortly before Christmas. Stephen’s family—mum, Carol, who also suffers from NPS, dad Keith and sister Nicola—are just as proud of his achievements. When he is not fighting his way to more accolades, Stephen follows his second passion, football—and more specifically Celtic—with just as much vigour.”



Upcoming surgeries

With so many of us, from time to time, having to undergo surgical procedures, I thought it might be nice to let those who are, know that we are all thinking of them. Last year, I faced a year of surgery and I know that personally, I found great comfort from all your kind words and well wishes.

On 28th May, Lisa Ward will be going in for some neurological surgery, Kath Lovatt is awaiting a date for knee surgery and Stephen Dobbins is also waiting to go in for knee surgery.

I would like them all to know that they will all be in my thoughts and prayers.



Please keep sending me all your questions for our medical section and stories for our “personal stories” section. I look forward to adding them to our next newsletter. You can send them by email or post.

Many of you have asked about help with benefits that you may be entitled to. It is always something that causes a great deal of confusion and I also have first hand experience of this in my own life.

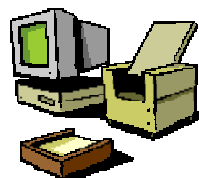
Here are some useful websites that provide some guidelines in where to go.

<http://www.bhas.org.uk/dla/index.shtml>

<http://www.youreable.com/forums/forum.jspa?forumID=1>

The sites are particularly useful for people who are looking for information on DLA.

Another useful site is <http://www.dwp.gov.uk/medical/med-prac.pdf>



Any correspondence, donations, letters can be sent to:-

NPS(UK) / Nail Patella Syndrome UK,
PO Box 26415,
East Kilbride,
Glasgow G74 1QX
Scotland

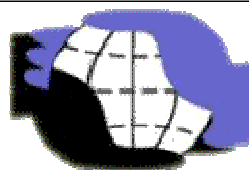
Look out for our next edition of the Newsletter in September 2008. If you have any photos or stories you would like me to add, please feel free to send them to me.

You can also use our free phone number of 0800 121 8298 to call us at any time.

For more information on Nail Patella Syndrome, visit:-

www.npsuk.org

www.nailpatella.org



NPS UK

Registered Charity 106621

Alternatively, email me Carol Dobbins on dobbinsek@btinternet.com

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