

An introduction from the Chair Submitted by Carol Dobbins



First and foremost, I hope that you have all enjoyed a peaceful and healthy festive season!

Hasn't time flown since the last Newsletter?!

On January 27th, NPS UK will celebrate 5 years as an official UK charity. We are all very excited about this as during these years, we have grown from strength to strength. We are very much a team as we strive to generate publicity and funds for our charity to allow us to help those whose lives are affected by Nail Patella Syndrome.

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It has been a wonderful 5 years and I certainly look forward to many more in the future!



In September, 2007, Stephen Dobbins and his school friends, Rhys Jephcott, Darren Lloyd and Paul Mullen all

took part in the Great Scottish Run in the Glasgow Green. They all crossed the line safely and in great times, raising awareness and funds for our charity. We raised approximately £400 from this very difficult event and I would like to thank the boys for all their hard work from all at NPS UK!

Runners needed!!!

Over the years, we have realised that one of the best ways to raise awareness and funds is to have brave volunteers who are willing to run various marathons. All we need to do is find the volunteers who are willing to do this and I am happy to deal with the rest ie. publicity, t-shirts, sponsor forms etc...



There are all sorts of marathons organised throughout the UK now, so if you know of anyone who would be willing to do this for us, please get in touch. If you feel you can take on this challenge or know someone who will, you will need to register as soon as possible as the numbers are high nowadays.

Gail Duxfield, who lives only a half hour from the start line at the Great North Run has offered her home as accommodation for anyone here who is willing to attempt this one next year also.

Please ask around and make 2008 our marathon year!



Many thanks to so many people for sending their donations to our charity. One of the costs that mount up for us as a charity is the Newsletter costs and this can be as much as £120 every four months. This time, I am delighted to say that two dear friends of mine, Paul and Julie Cummiskey have placed our collecting tins in their own shop and have raised over £100 last year. This will pay the full amount for this edition of our Newsletter which provides relevant information about our charity and all the events from the past four months.

Thank you also to those of you who continue to raise funds in this way. We really need all the financial help we can to keep providing help and support to those who need it.

I am also very grateful to those of you who sent kind donations over the festive period. I received donations of £60 from kind neighbours also.

CONGRATULATIONS TO ELIZABETH, SIMON AND THEIR BEAUTIFUL NEW ARRIVAL, LUCY GRACE FROM ALL AT NPSUK.

One lady who deserves so many thanks for all she does for us is Elizabeth Sweeney. She works as a consultant clinical geneticist and supports all our work by coming to our conferences and providing up to date information on NPS.

She gave birth to Lucy Grace at 8.02 on the 26th October, 2007. She was 8lb, 8oz.



So what's inside?

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One of our speakers at our conference in August 2007 was a lady called Anna Allford who works for the Genetic Interest Group. Anna has been working on preparing Family Route Maps for various genetic conditions including Nail Patella Syndrome. We met up in London with some of our fellow NPSers to discuss this in a Focus Group Meeting in London. Anna attended our conference to gather as much information from those who attended as possible to assist with the Family Route Map. She, later, wrote an article for the Autumn edition of the GIG today magazine, which was publicised and therefore creating more awareness for this rare genetic condition.

Many thanks Anna!

GIG Today – Article Autumn 2007 - Anna Allford

Family Route Map Workshop at the NPS(UK) Conference, Chester 11th August 2007

Arriving in Chester to glorious sunshine I entered the lovely Conference hotel and found a throng of chattering adults and children already exchanging information and shared experiences. I had been invited to present a workshop to develop the Nail Patella Syndrome (NPS) Family Route Map and was expecting to have a small gathering in a room off the main conference hall. Instead, I was ushered into a hall full of people seated behind tables and was told there was about 70 people here as not everyone could make it! Slightly anxious I took my seat and listened to experts from the USA who were providing new and breakthrough information about ADHD (Attention Deficit and Hyperactivity Disorder) associated with NPS, and education and care for affected children and adults.



The enthusiasm, interest and genuine friendliness of everyone there was the spur for me to stand up and deliver a workshop originally intended only for about 10 participants! I had prepared a set of questions around both the NPS related issues and the previous outcomes of the combined focus groups we'd held across all six conditions that we are currently working with to develop condition-specific Family Route Maps for: Barth Syndrome; Gorlin Syndrome; Multiple Endocrine Neoplasia (MEN); Myotonic Dystrophy; Nail Patella Syndrome; and Syndromes without a name (SWAN). A generic template for other support groups to develop their own Route Map will also be freely made available through GIG at the end of the project.

The positive response from those present was overwhelming and by the end of the day the A3 sheets for each individual question had all been filled up with ideas, information and contacts. This provided me with sufficient information to develop the first draft of the NPS Route Map and also gave Carol Dobbins, Chair of NPS(UK) and her Committee plenty to think about in terms of what their members wanted and importantly, information about healthcare services accessed by members which had been helpful.

After a great lunch (and more new friendships being forged) the main Conference continued with further clinical information about the kidney and eye structural abnormalities associated with NPS and how surveillance is required to detect any complications in these systems. Additionally, many people present benefited from the detailed facts relating to the joint involvement and best practice when surgery is required presented by a Surgeon who also has a son affected by NPS. The Speaker discussing the molecular genetics of NPS put into context how the altered gene influenced the development of the affected systems and led to the way the condition presented in individuals and families.

Other events at the Conference included a children's activity group and the opportunity to have a professional manicure for free! Now many of you may wonder why on earth this luxurious pampering session was included as part of the Conference? Well, in addition to those who took advantage of it feeling totally relaxed and beautified, the name of the condition, Nail Patella Syndrome, indicates that the nails are involved and are often absent or misshapen, especially the thumb nail, exposing the nail beds or leading to pitting or discolouration of the nails. Many people, especially females, feel their self-confidence to be affected by this feature and had never considered having a manicure. Lisa, the Manicurist showed each person individually how to make the best of their nails giving them a huge boost in their confidence. Saturday evening saw the social events begin with Karaoke and a disco as I slipped away to spend the night in a spooky house (but that's another story).

On the 5th March, the Family Route Map will hold its formal launch in London. I am delighted to have worked together with GIG on this very positive project and I would like to thank those of you who have played a part in providing information for this. The article you are about to read will be included in the next GIG Today edition and I think it will be of interest to you as it explains in detail what the project has entailed.

The Family Route Map Project will conclude with formal Launch

We are delighted to announce that the Launch of the six condition-specific Family Route Maps together with the generic template and Final Report will be held at The Wellcome Trust Conference Centre in London on March 5th. We very much hope that the Support Group representatives and individuals and families who helped with the project will be able to attend as we aim to invite clinicians with an interest, the Department of Health and NHS decision-makers to join us in this launch and workshop.

Developing *Family Route Maps* as a Tool to help families with genetic conditions to access appropriate information and services in the UK was the primary objective for this groundbreaking and unique project. It was agreed that listening to the concerns of patients, families and carers was essential. Workshops were held for each condition with the aim to explore information and services currently available to these families as the first stage in the development of the Route Maps and also to ask them what they would like to see included. Additionally, an on-line questionnaire was made available through the GIG website with links to the websites of the six Patient Support organisations, and interviews with clinicians and other healthcare professionals with expertise and experience in these conditions were undertaken. In primary care in the UK we know very little, from the point of view of the patient, about caring for patients with rare genetic disorders and therefore primary care services were talked about in all of the patient focus groups. A focus group with General Practitioners (GPs) discussed the concerns and issues raised in the patient focus groups through a series of vignettes presented for discussion. From this collective information a template for a Family Route Map that could be used generically by other Patient Support Organisations was also created.

Seven important over-arching themes (below) were identified from the qualitative data with a number of sub-categories and these are discussed in the report in relation to current care and future possible developments:

Information, Communication, Diagnosis, Treatment and Surveillance, Education of Healthcare Professionals, Ethical, Legal and Social Issues; and Empowerment of patients, parents and carers.

Overcoming barriers

The patients, parents and carers involved in this project were by and large well informed as many had already been in touch with Patient Support organisations and some were themselves involved in providing support to others within one of the six charities representing the chosen conditions. However, it should be noted that during the course of the focus group discussions, people were actively exchanging relevant information and peer-learning took place. Newly diagnosed patients, the less articulate, less forthright and less resilient patients have an incredibly difficult time trying to get what they need or in some cases remain ignorant of the care they should be receiving because the current system of health and care services doesn't proactively seek to provide information in a timely and comprehensible fashion. Instead many people need to *ask the right questions* or for some finding relevant information is left to chance.

["It was only by chance that we came across the \[Support\] Group...it's really a case of who you know."](#) (Gorlin Syndrome focus group)

What we found out from the project

Findings from this project suggest that patients with rare genetic disorders are not given sufficient information about their condition, services are considered 'patchy' and some families are still not aware of, or accessing, NHS Clinical Genetic Services. Many patients report: delays in being diagnosed; difficulties accessing the treatment and surveillance they need due to a lack of knowledge about rare genetic conditions in the medical profession and to an absence of coordination and continuity of care; and little psychosocial support leaving patients and families frustrated and 'stuck' in the system.

["It felt like you were on your own!"](#) (Myotonic Dystrophy focus group)

Participants in the GP focus group were well informed of the risk factors and protocols for referral to a Clinical Genetics Unit for more common genetic cancers but for rare genetic disorders it appears less clear. Discussions between healthcare professionals, and individuals and their families about genetics-related information crucial for informed decision-making are often ad-hoc or absent, as those affected by genetic conditions move through their life-stages. Information through Patient Support Groups and the media (including the internet) fulfilled some of their needs and helped them ask questions of professionals in order to make informed choice which is core to service delivery in the NHS.

["I didn't realise my daughter could be a carrier"](#). (Barth Syndrome focus group)

Many people expressed dissatisfaction with the lack of co-ordination and continuity in their care, and were frustrated by having to constantly explain the condition and fight for what they need. They describe themselves as 'slipping through the net' with no-one taking responsibility for them, as everyone thinks that someone else is caring for them.

["It makes me feel a bit like I've got to fight for everything I need"](#) (SWAN focus group)

Continued overleaf....

It is apparent that not all patients with the same condition are receiving the same surveillance and/or treatment. This may be appropriate due to different manifestations of the same condition, but patients feel unsure about whether they are receiving the most appropriate care, as many healthcare professionals do not have knowledge and experience of these rare genetic conditions.

“...in terms of eyes, kidneys [surveillance]...I have nothing whatsoever.” (Nail Patella Syndrome focus group)

Importantly, even when lack of knowledge of these rare conditions by healthcare professionals was openly expressed to patients in a supportive way, participants felt there was an opportunity to learn together, valuing each others experience.

Those affected by the six conditions appreciate that it is not possible for healthcare professionals to know about every condition, but feel they should have the option to be treated by an expert, even if this involves travelling outside of their Primary Care Trust (PCT). People said that they would be prepared to travel in order to see an expert in the condition or to be cared for by a Centre of Excellence.

“Patients should be under the care of a specialist centre.” (Multiple Endocrine Neoplasia Disorders focus group)

Conclusion

Although this study focuses on six specific conditions, it is an important source of information about the patient experience of clinical management and commonalities such as the need for clinical guidelines are likely to apply to the majority of genetic conditions. This project has found that patients' needs are not being met and has generated suggestions for possible strategies that could be used to provide better care. A possibility favoured by patients is the option to attend a specialist centre, as this would ensure they are receiving the appropriate treatment and surveillance and allow for coordination and continuity of care. Patients currently find it difficult to identify experts within the field, so require more signposting, and experience difficulties with getting referral to these experts. Some people suggested that a further role for clinical genetics is to be part of the multidisciplinary care for patients with rare genetic conditions, providing information, coordinating care, and acting as a point of contact for any queries or concerns.

Another suggestion from some participants in one of the patient focus groups is that practice nurses in primary care could carry out the role of 'champion', coordinating care to ensure appropriate surveillance and treatment, and providing support to patients. It was felt that they did not need to have extensive knowledge of the condition, provided they knew what surveillance should be taking place, as they would be able to follow guidelines that had been put in place by experts. They are viewed as friendly and supportive, and it was felt they were ideally placed to be an advocate for patients who feel that their voices are often not heard.

“I think we all need an advocate.” (Myotonic Dystrophy focus group)

The development of Family Route Maps will give patients access to information that they may otherwise have had to wait months or even years for, due to the rarity of the conditions and the inconsistent information that is available. The Route Maps will also help the health professionals who work with these families to explain all the treatment options, where to go for further information and which other disciplines the condition requires contact with. We anticipate that the Family Route Maps will also help to educate patients and professionals and will raise awareness of the complexity of these conditions and what patients may need to know.

If you would be interested in attending the launch of this project or receiving further information on the work please contact Melissa Hillier on melissa@gig.org.uk or 020 7704 3141

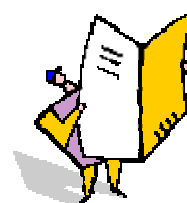
Anna Allford, Melissa Hillier

contact a family
for families with disabled children

Interesting factsheet

The winter guide: help with fuel bills and keeping warm' this factsheet has just arrived Contact a family office and at the time of writing is not yet on their website, but will be there shortly (see the link to the factsheet page below). This guide gives information about keeping fuel bills low, sources of financial help to pay bills and some practical suggestions around keeping warm in winter. If you would like to order a copy of this factsheet please call the helpline on 0808 808 3555. General link to their factsheet is:

<http://www.cafamily.org.uk/factsheets.html> .



NPS(UK) finance report

Submitted by Shirley Raynor



NPS UK Finance Report January 2008

Happy New Year to you all, and I hope that 2008 brings you everything you wish for.

We currently have just over £8000 in the bank. Since the conference we have not had a great deal of activity through our account – we have received £472 from sponsorship, fundraising and from the Everyclick website. Thank you so much to those of you who worked hard to raise this money, and who used the Everyclick website as their search engine. Remember Everyclick doesn't cost you anything at all, and creates income for NPS!

The only costs we have had in the period are our BT phone bill for the Freephone line, £49, and the cost of our PO Box number, £58.

Looking forward to the New Year, it would be great to hear about any fundraising events you have planned. If anyone is planning to raise money by sponsorship for a run or other event, please let us know so that we can help raise awareness for you via our website. And don't forget to ask Carol for a blank sponsorship form that you can use – this includes provision for sponsorship to be given under Gift Aid, which means that we can then claim an extra 28% of the amount donated direct from the Revenue.

Thank you everyone for all your hard work in 2007, let's keep it up for the New Year.



Recently I have been working with Everyclick.com to promote our charity in various ways. I have been communicating with their PR manager, Emma Walton, as we organised some publicity with the local newspapers and how Everyclick has raised so much money for us in the past. It made a great local story and many people were interested to learn how to use this in order to support our charity.

I have also been working with another member of their team on a fantastic opportunity for our charity called the "Charity Challenge"

Following Everyclick's successful poster campaign, they are delighted to announce that Clear Channel Outdoor is working with Everyclick to offer us the chance to win a national poster campaign on over 1,500 sites across the UK.

Entering the Charity Challenge has been very simple and could bring us huge benefits. No matter how big or small all charities entering the competition will be given:

1. The chance to win a national poster campaign
2. The chance to win advertising space on Everyclick.com
3. An Everyclick home page customised to promote NPS UK and provide desktop brand awareness for us every time our supporters log on

For more information about the challenge visit <http://charities.everyclick.com/charities/charity-challenge.htm> or call Everyclick on 01608 653026.

The challenge kicks off on 14th January and NPS UK has completed its registration. So, please, more than ever, make this your homepage and start searching using Everyclick. Ask all your friends and family to support us on this too. It costs nothing at all and benefits so many!



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



Personal stories

I would love to include in our newsletters some personal stories of life with NPS. I would love for you to send me details of what you would like to include. It could be an event relating to NPS that you feel would be beneficial for others in our NPS community to hear. If you have photos, I would love to include these too. It is always nice to put a face to a name!

I would like to start with a little story that I hope will provide a little hope for those who live with NPS and feel that they may be unable to participate in something they have always wanted to.

As many of you know, my son, Stephen, who is now 10 years of age, has taken part in Martial arts for three and a half years now! I always remember the first day he said he wanted to do this as I was terrified, fearing the worst and thinking that it would be impossible for him. Throughout the years of training and competitions, Stephen has had many successes, however, he has also endured a few mobility problems and pain too. His love for Tae Kwon Do was stronger though and on the 23rd of December 2007, Stephen became a TaeKwon Do Black Belt after enduring a six hour examination in Edinburgh, Scotland. His Master, who graded him on the day with the overall highest marks, said that he was extremely good. He was aware that his arms were not able to fully extend (Stephen's arms lie at approximately 90—110 degree angles) but he had adjusted his technique accordingly.

I have always said to Stephen that anything is possible if we truly believe and I think this is a great example of this.

Stephen is pictured in the front row, far right.



Inspirational stories

I am pretty sure that there may be other stories like this one from many courageous children and adults who have NPS. Please send them to me and I would be very happy to add them to our Newsletters.

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.

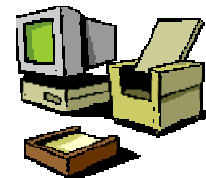
Thanks to one of our members, she has found a website that is very useful in providing some guidelines in where to go.

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

<http://www.youreable.com/forums/forum.jsps?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>



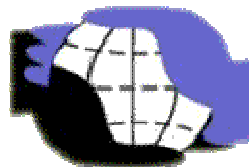
Any correspondence, donations, letters can be sent to:-

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 May 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
Phone me on 01355 241277