

Nail Patella Syndrome(UK) is a charity that provides information and support to those whose lives are affected by this condition and also for the use of the medical profession.



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An introduction from the Chair

Submitted by Carol Dobbins

First of all, let me say how happy I am to be announcing the fact that NPS(UK) is an active charity within the United Kingdom to help increase awareness and raise funds for this rare genetic condition. At this moment in time, we are eagerly awaiting our official stamp of approval from the Charity Commission which hopefully will happen when this first newsletter edition goes to print..

We are an affiliated charity working alongside NPSW (Nail Patella Syndrome Worldwide). Our main aims are to find as many people with this condition as possible and to provide them with as much information as we can, to support and work with the medical profession in assisting with questionnaires or studies that may be carried out. We also need to show our support to the wonderful people within the medical profession who work tirelessly for the betterment of NPSers across the globe. We need to raise as much public awareness as possible to educate and provide information that is much needed within the medical journals. There still is very little written within the medical journals and very often people are mis-diagnosed or have not received treatment due to uncertainty about the condition. We also need to raise as much funds as we can to provide support and assistance to many. The money has been raised in so many different ways in the past, including leg waxing in a Pub (my poor husband will never forgive me!) My son, Stephen, who also has Nail Patella Syndrome, has attended St. Leonard's Primary School for two years and they have supported us by organising a fundraiser and raising over £400 for our cause. I have raffled mountain bikes, had friends run the Glasgow Marathon, and I must say that every occasion has been just incredible.



These guys raised over £700 for us all by running the Glasgow Marathon. They are incredible!

Ronnie, Steven and Graham, we can't thank



Here is a picture of the Nursery Staff at Saint Leonard's School. They decided to organise an event to help with our needs.

Alison, Nicola, Margaret and Stephanie, we can't thank you enough.

So what's inside?

| | |
|--|---|
| Front page | 1 |
| Introduction | |
| What is NPS? | 2 |
| UK Chester conference planned for August 2003! | |
| Meet our treasurer and secretary. | 3 |
| Read our financial report | |
| Ideas needed! | |
| Back page | 4 |
| A report from the 2002 Pittsburgh conference. | |
| Contact details | |
| Interesting sites | |

What is Nail Patella Syndrome?

Nail Patella Syndrome, otherwise known as Fong Disease, Turner-Keiser Syndrome and Hereditary Osteo-onychodysplasia (HOOD) is a rare genetic disorder that is usually apparent at birth. It affects approximately 1/50,000 newborns and is autosomal dominant.

Although the symptoms and physical characteristics may vary, characteristic abnormalities tend to include improper development (dysplasia) of the fingernails and toenails; absence (aplasia) and/or underdevelopment (hypoplasia) of the kneecaps; underdevelopment of certain bones and/or webbing of the skin at the bend of the elbows; and/or abnormal projections of bone from the upper (superior) portion of both sides of the hipbone (bilateral iliac horns) Children can be born with club foot (talipes) or develop curvature of the spine.

In addition, some individuals within certain families may have increased fluid pressure within the eyes (glaucoma). Open angle glaucoma, without appropriate treatment may cause increased narrowing of visual fields and eventual blindness. Other ocular abnormalities may be associated with NPS.

Approximately 40% of individuals may develop renal abnormalities also. These are usually displayed as proteinuria and/or haematuria.

UK Conference planned for Chester 2003

The early stages of preparing for our first NPS (UK) conference are underway.

We have decided that we should use a location that is easily accessible for as many people as possible and we have decided that Chester would be a good choice. At this stage, we are considering as many options as possible with regards to location, cost and facilities. We would be looking for a venue that will accommodate for people with mobility problems also.

I have contacted the local newspaper here in East Kilbride to try and obtain a good deal of interest in what we are doing and why. They will arrive tomorrow and I will give an interview to the reporter and some more "ugly mug" shots will be taken.

I will discuss the fact that the conference is so vital in allowing many people the opportunity to learn about this condition. We are hoping that many from the medical profession will also attend and discover all the work that has gone into this condition over the past few years.

I, personally feel that one of the most vital parts of these conferences is meeting with others who share Nail Patella Syndrome. It was one of the most emotional moments in my life when I finally got the chance to share my life experiences with others

Right now, I am liaising with a lady who works as a conference co-ordinator in Chester and she has been a great help. She has sent a brochure and we are gradually narrowing it down to a small few that we can check out.

The date we are looking at is Saturday 16th August 2003. The talks will likely continue throughout the day. There will likely be an entrance fee of approximately £10 to try and cover much of the cost in organising this event. I will be able to confirm the list of speakers in the next newsletter, which should be in April, but for now, I am able to confirm that Dr. Elizabeth Sweeney and Dr. Iain McIntosh will be gracing us with their knowledge.

If anyone wants to contact me with any information or questions with regards to the conference planning, please feel free to on my details at the back of the newsletter.

An introduction to our treasurer

Shirley Raynor, an accountant from Bath, is the treasurer for NPS(UK). She comes from a family with a long history of Nail Patella Syndrome. As far as she knows, her great grand-mother was the first in the family to have this condition. Both of Shirley's children have NPS also.

A huge thank you must go to you for all your help with the setting up of this much needed charity here in the UK.

An introduction to our secretary

Dr Elizabeth Sweeney MRCP DRCOG, is a Consultant clinical Geneticist based in the Royal Liverpool Children's Hospital, Alder Hey. She, very kindly, agreed to become our third trustee member of NPS(UK), and became our official secretary. She has researched a number of people with Nail Patella Syndrome and her work within the NPS community has been outstanding.

We are eternally grateful for your support.

NPS(UK) Financial report

Submitted by Shirley Raynor

Welcome to our first Newsletter, and the very first financial report. I am thrilled to be able to tell you all that we have a little over £1150 in our bank account at the moment, the majority of which has been raised or donated by NPS sufferers, their friends and families. Thank you all so very much for all your hard work and generosity. However, we still need to work hard at this— our planned conference in Chester next year is the target we are working towards. Once we have chosen the Hotel, we will be able to put together a budget of the costs that NPS(UK) itself will have to meet, and then we will know exactly how much we need to raise. Watch this space!!

More good news is that having raised more than £1000 means that we can push ahead with our Charity Commission registration— we are eagerly awaiting the reply that will tell us that we are a fully fledged charity. Having this registration will give us a lot more scope for fundraising, so it is something we need to do very much. I hope that by the time I come to write the next financial report we should have our Charity Registration number proudly displayed on the front of the Newsletter.!

Another benefit for us of registering is that we can claim tax from the Inland Revenue on donations made by UK taxpayers. All it will mean is a simple declaration by the giver that they pay UK tax, and we will be able to claim back £28.20 for every £100 donation we receive. Hopefully this is another source of income for our fund that we will be able to tap into. I will give more details of this in our next newsletter.

Fundraising ideas ?

For the past three years, I have enjoyed a wide range of fundraising events, ranging from simple raffles to larger events including the Glasgow Marathon. Each one has been a lot of fun and has generated a great amount of interest from the newspapers and also raised a good deal of cash .

I now appeal to you!

If anyone has any ideas, comments, suggestions, please feel free to contact me at any time on the details at the back of the newsletter. When we have our official registered charity status, I have a much wider scope to raise funds. I would also like to ask anyone who may know someone who would run a Marathon for our cause to get in touch and we could pay their registration fee in return for them raising as much money as possible for NPS(UK).

The 2002 conference in Pittsburgh

Submitted by Shirley Raynor

When I decided to take my two children to the NPSW conference in Pittsburgh earlier this year, I really didn't know what it would be like. It was such a long way, an 8 hour flight; and EXTRAORDINARY visa bill– would it be worth it? If you want the short answer– it's "YES!!!" If you'd rather have the long answer, then read on.....

I won't bore you with the details of our flight, or the rather eccentric US customs lady who insisted on giving us tourist information and then took my apple away (no food to be brought in!???) Our arrival in the beautiful city of Pittsburgh was without a hitch– and as soon as we arrived at the Holiday Inn, we were ready to crash out– the time difference is 5 hours, and by 7.30 pm we were all in bed! I do vaguely recall answering the phone to a couple of other conference attendees who had just arrived and wanted to say hello, but I don't think they got much sense out of me! Of course, we were awake at 5 am, and I can tell you now that US morning TV is just as exciting as UK early morning TV– and I think we were among the firstcomers for breakfast...and that is when it all got exciting!

The next thing we knew, there was Julie-Anne Pickett, (a Huddersfield girl, now married to an American) with her lovely family, and we were soon comparing knees, elbows and fingernails. Julie-Anne is the first non family NPSer I ever met, and wow! It really was a special moment. We spent the morning with J-A's family, and later on met Gary (oh, that Alabama accent!) and all had a late lunch together, before getting back to the Hotel for the pre-conference get together. It was so exciting to meet, at long last, some of those people who I felt I already knew so well, we were soon comparing feet, hands and even backs!! The children had very quickly made friends, it was especially moving to watch the small crowd of boys, all around the 10-12 year age group, happily chatting about their Gameboys and occasionally comparing elbows.

The next day, Friday, was the day of the conference, and what a lot of work had gone into the organisation. As we checked in, we were allocated time slots for the various tests that needed to be booked– the glaucoma tests, orthopaedic consultancy appointments, bone density tests and MRI's for those who needed them. Other tests were there to be taken advantage of, as and when, - blood and urine tests for kidney function, dental examinations etc... On top of all this there was a full programme of talks by each of the specialists, all of whom explained their own area with great thought and care. I know I learnt a lot from these sessions, and in particular found very interesting the explanation of how the faulty gene which causes NPS can be tracked during foetal development, so that they

can show how it affects different parts of the body. An extraordinary amount of work had gone into these presentations, and they were all well received.

By the end of the day, we were pretty tired of being prodded and poked about, and were ready to head back to the Hotel in the minibus laid on for us. Those of us who had just had our glaucoma checks still had dilated pupils from the eyedrops, and the sun was so bright outside that they had to lend us special sunglasses to wear on the bus.!

The next day was time tabled with a number of group sessions where we could go to discuss a particular topic with other NPSers. The topics included ADD and ADHD (a common NPS trait!), NPS exercise routines, and many others. We were all pretty exhausted by the end of the day, especially the children, and a large number of us went out to enjoy ourselves together in Pittsburgh.

So, what did we, as a family, gain from our trip? We made some wonderful friend, who I know we will keep in touch with for many years. We had a wonderful (but brief) holiday. We were given some excellent advice from the consulting doctors about Rosie's knees, and we were able to bring home a copy of her MRI scan– not easy to obtain here, on the NHS. We all had tests that will not only help us in knowing how NPS affects us personally, but will be of use in furthering specialist knowledge of this condition, and therefore improve treatment for all future NPSers. Oh,...and Rosie bought some Gap Jeans for \$7!!!

Contact details and useful sites

Any correspondence, donations, letters can be sent to:-
NPS(UK) / Nail Patella Syndrome UK,
PO Box 26415,
East Kibride,
Glasgow
G74 1YW
Scotland

Alternatively email me on dobbinsek@btinternet.com
Phone me on 01355 241277

Join many of us on <http://www.OurNPSgroup@yahoogroups.com>

Nail patella Syndrome website with useful information can be found on www.nailpatella.org

