



Newsletter No.2

January 2010

As I sit here writing this newsletter, I am thinking back over the last few years about the determination I have needed to get where I am today. My son James is 13 and at age 6 was diagnosed with TRPS type I. After a normal birth and a straightforward 5-6 months things started to go a bit haywire. He failed his first few milestones – didn't sit unaided until approx 11 months old, didn't crawl until about 15 months old and failed his first hearing tests. If I had known what was ahead of me, I would never have believed that I could have coped with what was coming, yet here I am 12 years on with a gorgeous, witty son who shames me at times with the way he has coped with everything so far.

To date he has had 3 sets of grommets, plastic surgery to correct his ears and further surgery to repair a large perforation to his eardrum, a broken collar bone, 17 teeth extracted in one go, perthes like changes to his right hip, plus more visits to our local hospital for him to be prodded, poked and photographed than I care to remember. I have had to fight every step of the way sometimes with doctors on my side and sometimes not. I have spent hours on the internet reading everything I could to try to understand what lies ahead for my son and the short answer to my search is nobody knows. No two people with TRPS present the same – though many do share "common" problems and many do look very similar to each other.

If you have just received a diagnosis, don't despair - when James was diagnosed the only help I could find was our sister American site. They were fantastic, but couldn't answer many of the questions I had regarding our NHS systems as they were obviously in the US. But now with our site here in the UK, we can offer you the help and support you might need. From those early days back in 2003 we now have 26 people with TRPS types I and II registered with the group - many who are willing to offer advice and support if and when required.

James will also tell you that life hasn't been easy. He has coped with all the operations and the hospital appointments with barely a murmur (though he hates needles now). He has developed an excellent sense of humour and makes light of many of his problems. Rather than hide away hoping nobody notices that he is different to his classmates, James relishes a challenge and last year had a starring role in his school play (he currently wants to be an actor) and in December hosted our local Children's Hospital Christmas carol concert for the second year in a row.



James and a very rare snowy day in Plymouth

Dr Adam Shaw

Adam Shaw is an honorary consultant in clinical genetics at Great Ormond Street hospital and a researcher at UCL Institute of Child Health in London. After qualifying as a doctor, he trained in both paediatrics and adult medicine. Pursuing an interest in genetics he went on to research the long-term health needs of patients with Noonan syndrome. During this time he also worked in the NHS, caring for children and adults with a wide range of genetic conditions. He moved to Great Ormond Street in 2007 to set up specialist clinics for a range of genetic illnesses including Tricho-rhino-phalangeal syndrome, hoping to better understand patients' health needs and develop medical management guidelines. He is a member of Rare Diseases UK, a group campaigning for better provision of health care for sufferers of rare diseases, and he advises several patient support groups in the UK and Europe.

Professor Raoul Hennekam

Some of you will remember, if you attended our first ever Gathering for people and families living with TRPS at Legoland, Windsor, in July 2006, the speaker who came for the day and who gave a PowerPoint Presentation. Professor Hennekam had been recommended by our other great friend Dr Hermann Josef Ludecke, from Essen in Germany and who is the major researcher into the disorder and who established the only reliable and accurate molecular blood analysis test in the World and who liaises closely with many of our UK geneticists and laboratories in this country and Europe.

During a very happy and informative day, looking at slides of patients from Professor Hennekam's Amsterdam Clinic, it became clear to us how other people though abroad still had the familiar features of TRPS and whose lives were similarly affected. During a personal session, when he met and talked with all present in an individual way and took photographs, we all came to realise just how keen he was on his subject and how dedicated he appeared to answer questions clearly and helpfully and was enthusiastic to hear from those present as to what their hopes and needs were. It was during the question and answer session that the idea was born, not only of having a Support Group of our own established but importantly, everyone felt they wanted a place they could go, to discuss their emotional and medical problems with complete honesty, privacy and to receive empathy, understanding, and practical advice. Hence another ambition was born....that we could all attend our own TRPS Clinic and the Professor promised he would do his best to see if he could establish such a place. During the latter part of 2006 and onwards into 2007, this kind and sincere man, working on our behalf grappled with all the intricacies and frustrations of overcoming "red tape", ethical committees, funding issues and every kind of delay posed by NHS administration to make this become a reality!

Eventually, it was with huge delight we heard that Dr Adam Shaw had been appointed to head up the research programme, to meet with and record the details of every TRPS patient he possibly could, or talking with babies and children's' parents. This data will become invaluable in unravelling all the mysteries of what the disorder reveals in time. Dr Shaw, along with the Professor, has worked diligently since Spring 2008 discussing, recording data, photographing, arranging blood tests, and helping more people with TRPS than any other establishment in Europe....quite an achievement! Though taking a background role, and busy with his main work within GOSH, Professor Hennekam has always been on hand to guide and advise and to help deal with all the "officialdom" which besets the NHS. It was with great sadness therefore, that we have heard the news that Professor Hennekam has given up his post at the Hospital and has now returned to his native Netherlands, for personal reasons, to spend more time with his family and to take up his work again at the Amsterdam Clinic.

This group owes a great deal of gratitude to his kindness and sheer determination in helping us become established and raising the profile of TRPS in the scientific and medical community. We send him our sincere thanks and very best wishes for the future.

TRPS Support Group UK – The Story So Far

As most of you will know it has taken us nearly 6 years to get where we are today. From the first contacts on the US website back in 2004 through to the Legoland meeting and onwards to the website we are now building. My thanks must go mainly to Diane who has fought alongside me since the very early days to get us to where we are now. Caroline has also been invaluable with her "web experience" and

as I have another older son currently doing a computer graphics course at college I know that it is far more difficult than it looks!

Thanks must also go to Martin and Kim who have raised funds for the group. Martin and Kim raised the fantastic sum of £300 by completing the Moors the Merrier 26 mile run/walk last year.

Martin and Kim together Kelly and Danny are planning are also planning a bike ride later this year to raise further funds – our best wishes and support to them all.



2010 is now upon us and hopefully we will continue to grow. It would be lovely to hear from some of you – your thoughts and pictures could be incorporated into future newsletters. Please get in touch and let us know if you are raising funds or if you just want to share your story with others.

Are You Having Bone or Joint Surgery in the Near Future?

Researchers at University College London are hoping to better understand the problems affecting the bones and joints in people with TRPS. One way to do this would be to study the bone that is thrown away during joint replacement surgery. **We would be very grateful if anyone affected by TRPS who may be considering**

having joint surgery in the next 2 years would agree in principle to donating their discarded bone for research. If so please contact Dr Adam Shaw - a.shaw@ich.ucl.ac.uk.

Registering your interest now does **not** commit you to having surgery or donating your bone, but it will help us decide on the best way to carry out the research.

Thank you!

Sarah Neil
Chairperson of TRPS Support Group UK

