Proteus Family Network UK Newsletter

Summer 2016

Well, summer is here, or so they say...and they must be right because it has been raining here for the past couple of days! Still,the forecast shows it brightening up by the weekend...here's hoping.

It doesn't seem 12mths since we got together at the Holiday Inn at Aylesbury and I seem to remember it was pretty wet then, when we visited the llama centre on the Saturday.

Quite a lot has taken place since then, with difficult and sad times and also times to celebrate.

Just a week after we took our leave of each other at Aylesbury we lost Michelle Gardiner, who suffered a pulmonary embolism quite suddenly one morning and never regained consciousness. In her passing she was able to help three people through organ donation. Michelle was a lovely young girl, who, at the age of eighteen, had her future ahead of her. She faithfully came to every family weekend along with mum and dad, Sarah and Neil. In recent years she was always helped looking after the younger children...she had a real flair for that...and everyone loved her. So many people, on hearing the news of her passing, said how caring and helpful she was. In her short life she touched many hearts and I for one feel blessed to have had her in my life.
We continue to hold Sarah and Neil in our hearts and thoughts.

💕💕*Our congratulations to Chloe Jones and Ricky Broughton who were married recently. Chloe first came along to a family weekend some ten years ago and since then has joined us at many weekends with mum Michelle and sister Amy. We wish both Chloe and Ricky a happy future together.*💕💕

We welcome new members Gary Leonard and Ben Clemson. With this Newsletter you will have also received an updated Family Directory...please check your details and let me know if there are any changes, thanks. We are also providing new information leaflets which now includes the glossary and a research update provided by Dr Veronica Kinsler, the research lead at the Institute of Child Health (ICH) in London, next to Gt Ormond St Hospital.

What have your committee been up to.....!

A few months ago we attended a research update day at Addenbrookes Hospital where Dr Robert Semple is leading genetic research and trials into segmental overgrowth conditions, including Proteus and KT.

In April we held our first Medical Board meeting of 2016 at GOSH. Since Prof. John Harper retired from his position with GOSH, Ms.Deborah Eastwood, Consultant Orthopaedic Surgeon has taken the lead of the Medical Board, although we are grateful to Prof.Harper for continuing to serve on the Board and support both the support group and research through the ICH. The meeting was well attended, with medical professionals from GOSH, the Royal Free at Hampstead and the research team.We are grateful to Dr Samira Syed for facilitating our meetings and to all who serve on the Medical Board who give their time and professional expertise to support us.

We had a good discussion around the possibility of routinely carrying out Doppler scans of limbs, particularly the legs, in those with overgrowth conditions, even if, for instance, the legs do not appear overgrown, as the vascular system can still be affected. We are looking at whether teenagers approaching transfer from paediatric to adult care could be scanned so that a complete picture is available ready for transfer. If this takes place and results indicate vascular problem previously unknown, it may be that routine scanning could be extended to include younger children.Further news on this to follow.

Research News

Recently, Sarah Gardiner, Sarah Rogers and myself attended a rare conditions support group research and networking conference at the ICH, led by Dr Veronica Kinsler. The day covered a number of rare conditions and it was good to be able to share experiences with other patient groups.We are looking to collaborate with other rare conditions support groups and will be networking at other conferences throughout the year....the next being the BAD ..British Association of Dermatologists ... In Birmingham in July.

Progress into identifying the genetic cause of overgrowth conditions is fast paced. The research team at ICH have been working in collaboration with the research team at Addenbrookes Hospital, Cambridge, led by Dr Robert Semple. If you were with us at our family weekend last year you will remember that we welcomed Rachel Knox who works alongside Dr Semple and Victoris Parker and who gave us an update on the Addenbrookes research.

Whilst the AKT1 gene was identified as being a main player in Proteus, recent research has identified the PIK3CA gene as being involved in some overgrowth conditions. Genetic testing for the PIK3CA gene has been taking place collaboratively through both the ICH and Addenbrookes. Results are interesting, for example, for those who have lived with a KT diagnosis and symptom wise satisfy the diagnostic criteria, the PIK3CA gene is currently being identified in only around 40-45% of those tested. It is clear that in the future the identification of overgrowth conditions will be much more narrowly defined and there will be no longer be simply a differential diagnosis of Proteus or KT, it will depend on the genetic mutation identified in those affected. Some of those

testing positive for the PIK3CA gene have had the opportunity to take part in trials of the medication Sirolimus, which is some instances has been shown to reduce overgrowth. If you wish to know more or to be tested for the PIK3CA gene contact the Addenbrookes research here:

http://www.overgrowthstudy.medschl.cam.ac.uk

News from Rare Disease UK

What is the real cost of your rare disease?
Managing diagnosed and undiagnosed rare conditions can be stressful and costly, for both families and the NHS. Although some rare conditions are managed through coordinated services (e.g. through multidisciplinary clinics or through a care coordinator), most conditions are not managed in this way.

We're carrying out a small study to look at the value of coordinating health care services for patients with rare and undiagnosed conditions. The study will identify and explore ways to measure different types of costs and benefits - to patients, family members and the NHS.

Currently, we are looking for patients or family members to test a 'patient diary' over a short period of time. Participants will be asked to record in the diary the different costs they face such as the time they spend coordinating their care each week, or the money they spend on travelling to medical appointments. We want to get feedback from patients and families to improve the patient diary, so that it can be used in future research projects.

If you would like to take part in this research or would like to ask further questions, please contact: Amy Simpson/ 0207 704 3141 or Dr Amy Hunter Please contact one of the research team if you would like this information in a different format or in the Welsh language.

If you are interested in the progress in the wider picture of research into rare conditions why not take a day to visit the Midlands Rare Disease Showcase in Birmingham...free to attend but you need to book a place.....

Remember to visit our members only Facebook page where we post information and news of events, etc, in between newsletters. If you would like to be a member of the FB page please drop me an email to let me know and I shall send you an invitation to join ..... remember though, it is for members only, to allow for sensitive discussions where needed.....

An invitation form the Proteus Syndrome Foundation - UK group: the PSF UK are holding a family get together over the weekend of 8-9 October in Bracknell. Dr Robert Semple, Dr Veronica Kinsler and Dr Susan Huson are expected to visit over the weekend. There may be subsistence for those living with a rare condition and their carers, but any other family or friends will have to pay the full price of accommodation. We are unable to tell you the cost at the moment, but if you are interested in knowing more please email the PSF UK Chair, Tracey Whitewood-Neal on traceywhitewoodneal@yahoo.co.uk

Our own family weekend get together will be held in Spring next year...details and booking forms will be out to you early in January 2017.

Do remember that we are happy to receive applications for a grant...for either specialist equipment or services, etc.
Either email myself on jean.harrison11@btinternet.com
or drop me a line at
10 Overhill Road, Stafford ST17 0QA

As always we are here on the support line...01785 661263.....9.00-22.00 365 days a year.....for a chat or just a listen. I know from experience what a relief it is to talk with someone who has gone through similar experiences to your own.....this is also the most positive feedback from our family get togethers.

We are also planning to meet with a Member of Parliament to discuss the financial implications of providing clothing for those living with an overgrowth condition which affects particularly the upper body. If you have problems sourcing clothing to fit, or have to have clothing ...shirts, jackets,etc.....made to measure, with the resultant cost, do let me know so we can use your experiences, alongside our own, in pressing for a specific allowance under PIP ... Personal Independance Payment....to allow for clothing to be provided.

I hope this newsletter finds you as well as can be....as ever there are always worries and concerns for our families and friends living with such rare conditions, but we live with the hope that the ongoing research and tremendous progress made in recent months, will one day provide the answer and hopefully, treatment for all rare overgrowth conditions.

Take Care and enjoy summer .... if it ever turns up!

Best Wishes

Jean Harrison
Chair Proteus Family Network UK.

Finding the answer .....supporting the need

Proteus Family Network UK Registered Charity No. 1098608