



Newsletter issue 21

November 2012

New Logo

Roy Forsyth

It's been a long road to announcing our new logo and not an easy one either, but here it is and we hope you like it. The design was trialled on a couple of tee-shirts at the Paris conference this year and received good feedback. Our new logo was developed by Dean Pannifer, a friend of CFUK Trustee Neil Hutchison. Dean, who is a graphic designer by day, kindly donated his evenings and weekends to develop the logo, which gives the Cystinosis Foundation UK an exciting new identity.

To explain it a little the logo obviously appears as a smiley face which we see reflecting the positive attitude of so many of the people with Cystinosis and their families. The logo also symbolises support, with one person offering support to another, we see that reflecting what we do as a charity and as a community within ourselves.

You will start to see this logo weave itself into the fabric of what we do and the old logo will be around until we use up our stocks of leaflets and any other materials.

The Trustees would like to take this opportunity to thank Dean for all his hard work and Neil for making this happen.

If you would like to use the logo for fundraising purposes, please get in touch with any of the Trustees.

Chairman's Report

Roy Forsyth

It has been a year since the last newsletter and I have to apologise for that, but hopefully from the information we share with you in here you will see it's been a very busy year and things are going very well.

Back in January the trustee group met to recruit some new members to our ranks. We felt that more people were required as the charity was growing and generally getting busier. As we do this in our spare time we are aware that our responsiveness could be improved. So I am pleased to announce that we now have three new trustees of the charity.

These are; Ranjit Seehra, from Derby. Neil Hutchison, from Edinburgh. Ami Froelich, from Preston.

Profiles for these and the rest of the trustees can be found on the website.

That for a brief time brought our little team up to eight trustees;

Roy Forsyth

David Benford

Neil Sugden

Gareth Jackson

John Terry

Jonathan Terry (The charity founder)

Sadly though John Terry bravely fought a brief battle with cancer this year and passed away in August. John had been an integral part of the charity from the beginning and was very well known and respected in the Cystinosis world. John was a man who didn't appreciate a fuss so with that in mind we haven't made any announcements on his passing other than this note here. He is a great loss to us as an organisation but we will continue to strive to meet the high standards he set in his work for the Cystinosis Foundation UK.

As a small charity we are really doing very well and we can only continue to work for those with Cystinosis with your help. Please keep fundraising that has been brilliant over the last year again. Please keep in touch through our website, or emails, or Facebook or through your contact with Jonathan. Tell us what you need from the charity it's what we are here for.

Just over 12 months ago I reported that we had reset our research funds appeal as it had raised exactly **£250,345.86**. When we first started the charity, we simply concentrated on survival, so it is outstanding for me to say that the appeal has now collected **£383,826.67**. This means that in just one year our supporters have run, swam, cycled, danced (and so many other activities) and in doing so have generated over **£130,000** !

Last year I reported that we had paid out over one hundred and ninety four thousand pounds and since then we have given another twenty five thousand pounds to the University of Sunderland.

These latest donations continue to fund work related to the Prodrug project, in particular for Pre-formulation studies on the lead Prodrugs that have been identified. The study is investigating the physical & biopharmaceutical properties of the lead Prodrugs which will give essential data for the development towards pre-clinical & clinical trials.

After this, the next development stage is work in manufacture and toxicity evaluation of the Pro Drugs, requiring funding of £125,000 which we have agreed to provide. Altogether these projects will advance the Pro Drug approach to an appropriate stage for an industrial partner to then fund these through clinical trials. This work begins in 2013 and runs through to 2014.

On behalf of the Foundation & the Trustees, can I again take this opportunity to say a huge thank-you to everyone that has raised funds of whatever amount in whatever way.

Even though we have raised this huge amount raised, we still know that the costs of bringing any new drug fully to market, having gained all necessary approvals, are very large. For this reason and because of how well the research projects are progressing, we continue to welcome funds to our appeal target which has had to be reset again to a stupendous figure of **£500,000**.

Gareth Jackson Treasurer



The Cystinosis Foundation UK are grateful for the continuing financial support of the
Martin Currie Charitable Foundation

Journey to a PhD

The first time I heard about cystinosis was as in an undergraduate lecture, when Professor Roz Anderson talked about the problems associated with cysteamine administration. Little did I know then what a big impact cystinosis would have on my life!

I initially chose the PhD on the development and testing of prodrugs for cystinosis as I thought that it would suit my practical skills as a pharmaceutical chemist perfectly; however, it soon became more than an abstract topic. Thanks to the excellent cystinosis community, I have had the privilege of meeting many people with cystinosis and their families and this has really impacted on my research. From speaking to patients first hand, I have been able to understand how an improved treatment would really benefit them. Undoubtedly, there is a lot invested in this project, not just financially, but also in terms of what a positive outcome could mean to patients. This has been a real motivation to work hard and ensure the research is carried out as thoroughly and carefully as possible.

Working towards a PhD was challenging and exciting. As a mum, it was sometimes difficult to juggle life with research and I have had to make sacrifices, but I feel they have been worth it. I am lucky to have lots of support, particularly from my husband and my supervisory team.

The University of Sunderland has been very encouraging and provided a positive environment and exceptional facilities. The Cystinosis Foundation UK has been an excellent sponsor; I am grateful for the funding and I feel honoured to have been involved in this project. However, although my PhD has now been successfully completed, my journey with cystinosis is not over. I hope to continue researching in this field and work towards improving the treatment of cystinosis, in particular, I am keen to see the prodrugs progress further, hopefully to clinical trials and future clinical use, and to use a similar prodrug approach to improving the treatment of ocular cystinosis.

Dr Lisa Frost



Picture of me working on cells in the microbiology lab.

Raptor Pharmaceutical UK Trial

The Cystinosis Foundation UK would like to announce that we are in discussions with Raptor Pharmaceuticals to assist with supporting the launch of a new phase III b worldwide clinical trial here in the United Kingdom.

The Recruitment period is planned from early to mid 2013. This is a long-term, open-label trial comparing RP103 and Cystagon®.

For information on the development of RP103 for Cystinosis please visit www.raptorpharma.com/RP103_cystinosis.html.

Details of the trial in the UK and how to apply will be released very soon.

Some Fundraising Events in 2012

It has been a year since our last newsletter and there has been an amazing amount of fundraising in that time, we would like to acknowledge some of these events. If yours isn't mentioned then please excuse us they all do matter but we have limits on space. To help us if you do have a fundraiser then write to us about it as well for use in future newsletters and for the website.

In May, the Impy Inc Fishing Competition was held at Greenacres Farm Fishery in Biddenden in Kent. The competition was organised by Barry Richards, who wanted to support his friend after hearing how his friend's son was diagnosed with cystinosis.



Among the runners at Kent's largest half-marathon this year was a team of 30 (yes thirty) hardy souls raising money for the Cystinosis Foundation UK. Justin Spencer, whose daughter Jessica pictured here, has cystinosis, pulled together this quite incredible band of runners, joggers and generally all-round good people.

Thanks also to Chris Conroy, as well as to everyone at Kings Hill Football Club and Paddock Wood Athletic Club.

Having won two places to take part in L'Étape du Tour, where amateur cyclists race over the same route as a stage of a Tour de France stage, Tom Barlow saw a fundraising opportunity for the Cystinosis Foundation UK and enlisted the help of an old friend, and keen cyclist, Stuart to join him on the ride. Tom and Stuart were cheered on by Seth, Tom's son who has cystinosis. Tom only bought a bike last October, so despite hundreds of training miles the Étape was a huge physical and mental challenge for both him and Stuart.

Well done to Mark Hewitt and friends (including Paul and Paula Ingram, parents of Harley) who repeated their successful 11 mile walk around Sheffield from last year, taking in all three major football grounds in the city. As you can see from the photo the rain didn't dampen the spirits of the Wallies.



Massive thank you to the joint team from Barclays and Source Developments who tackled the mighty Three Peaks Challenge in June. Starting at 5pm on the Friday at the foot on Ben Nevis, by 6pm, the following day, the bulk of the team were peeling off their waterlogged socks in the car park in Snowdon base camp. The conditions were nothing short of treacherous, particularly when climbing the highest mountain in England in the dark - well done all.

The Cardigan Challenge involved a 30k loop around London. Approximately 80 walkers completed the event with times ranging from 6 to 10 hours. They had an enjoyable, if slightly soggy, day in the process.

At the end of June the Cystinosis Foundation (USA), Cystinose France and AIRG France hosted the 7th International Conference near Paris. AIRG are an organisation for information and research into genetic renal conditions.

The event was very well attended both by delegates and Professionals, it was great to see a strong UK contingent there and thank you for advocating how well our own little conference had gone in 2011.

A welcome was held on the Thursday evening and then two full days of programme took place on the Friday and Saturday with a half day wrap up session on the Sunday. The wealth of professional knowledge in the room over that period was truly amazing. It is not the intention here to provide a detailed account of the event that can only be gained from attending the event but listed below are some key points.

Bone Marrow Stem Cell Transplantation and Gene Therapy.

Speaker, Stephanie Cherqui of the Scripps Institute, USA.

Dr Cherqui spoke in Italy 2 years earlier on this research work and it involves using a mouse model to show that transplantation of bone marrow stem cells expressing a functional CTNS gene could improve Cystinosis. Cystine has been shown to be depleted by 57% to 94% in all tested tissues. Quantities of “healthy” cells can be observed in all the organs. There is a proof of concept on this research for developing a bone marrow cell transplant treatment for Cystinosis. Some early clinical trials will begin in the USA initially with stem cells from a healthy sibling, although this is a difficult approach with high chances of rejection.

The long term objective is to develop a treatment where the patient’s own Stem cells are used.

Transition from paediatric to adult care

Speaker Nicholas Webb of the University Of Manchester Hospital.

Dr Webb spoke about the work they have carried out into improving the transition of patients from paediatric to adult care. Transition being the purposeful and planned movement with a chronic physical and medical conditions from child centred care to an adult system.

This should be distinguished from the actual transfer which happens on a set date.

Current practices across the UK aren’t applied consistently and there is no “best practice” to follow during this critical phase of a patients treatment. There is evidence to show a high frequency of transplanted patients having graft loss at or just after this transfer point. But where good transition planning is in place prior to the transfer this is significantly improved.

There is a basic key list of criteria that need to be met before the transfer over, these are copied below

Competencies to be achieved prior to transfer

Watson Pediatr Nephrol 2005 20 113-117 and IPNA consensus statement

Table 1 | An example of the competencies expected of a young adult being transferred to an adult renal unit



- I understand my condition and can describe it to others
- I know my medications and what they are for
- I can make decisions for myself about my treatment
- I know what the adult clinic arrangements are and who will be reviewing me in clinic
- I know how to make my appointments
- I can make my own transport arrangements to get to the hospital for appointments
- I know who to call in a medical emergency
- I am able to talk about my worries concerning blood tests and other treatments
- I know the dietary advice that I have to follow and the importance of activity
- I have appropriate knowledge about sexual health matters
- I have discussed alcohol, smoking, and drug issues



Key elements of successful transition

- Do not transfer at times of crisis (medical or family)
 - Take into account treatment plans of other medical teams
- Time transfer after major educational and social milestones (ideally complete school and puberty)
- Written transition policy agreed by all parties, including a process of consultation with patients, parents and their families



Key elements of successful transition

- Written transition plan completed by age 14y
- Clinical leads for transition in both paediatric and adult teams
- Young adults undergoing transfer during the transition process should have access to some form of **support service** e.g. 'young adult support workers'



Key elements of successful transition

- Transfer is commonly targeted for 18-21y in most countries
 - However important to avoid assumption that age correlates with cognitive or social maturity
 - Chronic illness will delay maturity process
 - Age at transfer dependent upon individual circumstances
- Formal transfer summary
- Option to participate in young adult clinic after transfer



Key elements of successful transition

- Education programme for patient and parent addressing medical, psychosocial and educational / vocational aspects of care
- Training programme in adolescent health and transition for paediatric and adult teams
- Evaluation, research and audit



Long Acting Cysteamine- Raptor RP103

Speaker Craig Langman of Feinberg School of Medicine, USA.

Dr Langman spoke of the recent trials on the RP103 developed by Raptor Pharmaceuticals. This trial was a randomized cross over trial to demonstrate the effectiveness of the RP103 drug in maintaining good white blood cell (WBC) cystine levels and was safe when compared to Cystagon®

The RP103 drug was shown to be “not inferior” to Cystagon® when taken every 12 hours and could maintain good WBC cystine levels associated with optimal outcomes in the disease.

A lower dose of drug is capable of maintaining these results and gastrointestinal side effects were still a factor.

In all the results were welcomed by professionals and we look forward to future UK trials.

Conclusion

Dr Bill Gahl of the National Human Genome Research Institute, spoke of the long term outcomes of people with Cystinosis and the treatments given for it in the past and today. The treatments we have today work and data shows that this is the case with expectancies of renal transplants being much later than they were. Yes there are other complications with Cystinosis that still need to be understood in greater detail. But the treatments that are available today work effectively albeit with intensive management.

There is hope for the future with the work that is on going world wide, “Beyond Borders” was the theme of the conference and we are a global community.

Contacts **Email** : advice@cystinosis.org.uk **Website** : www.cystinosis.org.uk
Helpline/Fax : 0118 941 4232 **Charity address** : 174 Corwen Road Tilehurst Reading RG30 4TA

Our on-line referral e-Store has now been rebranded as **Spend And Raise**

Before you make any purchase on-line, check out our site at www.spendandraise.com/cystinosis and if the retailer is listed there, click their link and any purchases you make can earn us money.

This costs you nothing and you may even find exclusive offers on our site, such as free delivery from Littlewoods Direct.

A referral donation is made by the retailer and costs you nothing – you just have to click the link first! And you have to click it every time, before you visit the retailer's site.

Remember: www.spendandraise.com/cystinosis



Please consider using spend and raise and pass it on to your friends. For a few extra clicks of your mouse and no cost to you you can help raise money for the Foundation. You may even save money through the exclusive deals on this site!

Wrist bands for sale



We still have a stock of wristbands for sale.

Please contact Jonathan Terry direct on the above contact details to be able to purchase them.

They cost £2 each plus postage and packing.



Mailing List Update

We regularly update our mailing list and wish to ensure the details we have for you are correct. You may no longer wish to receive this newsletter. You can download the newsletter from the website. If you wish to change your details, or be removed from the mailing list please complete the form below and return it to Cystinosis Foundation UK, 174 Corwen Road, Tilehurst, Reading RG30 4TA

Current details

Name.....

Address.....

Postcode.....

New Details

Name.....

Address.....

Postcode.....

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