



## Cystinosis Foundation UK Conference The Chairman's Summary

### Thank You All For Attending

Thank you to all who came along to the Manchester Conference Centre on the 24th September - we had an excellent attendance. As an organisation, it is one of our basic aims to provide education about the condition and its treatments, so this was the main motivation for the event. It is your participation that gives us the encouragement to keep doing what we do. There hadn't been a UK conference since Nottingham in 2004, so we were well overdue in organising another.

### The Presentations

In the morning, after the initial opening of the conference, we split into two groups – one for adults, one for children.

The group for adults was given a presentation by consultant nephrologist and clinical director of the University Hospital Birmingham, Graham Lipkin. The main focus of this talk was on the issues around the transition of children into adult care. The need to look at the development of the care adults with Cystinosis receive was also raised and this will be addressed under the umbrella of the UK Renal Registry.

The children's group had a presentation from William van't Hoff, consultant paediatric nephrologist at Great Ormond Street Hospital, London. This presentation focused on the basics of what the condition is and the current treatments, all very important information for new families coming along to these events. The presentation utilised material from a new book that has been jointly produced by the Cystinosis Foundation (USA) and Orphan Europe, with advice from William van't Hoff himself.

In the afternoon we heard from Raptor Pharmaceuticals and their chief medical officer, Patrice Rioux. This focused on the development of their enteric coated drug called RP103, which could be used as an improved replacement for Cystagon™. Raptor's work has had some trials, the results of which were presented. The Raptor team travelled from America to attend our conference, so we owe them particular thanks for the huge effort in making that journey. Accompanying the Raptor team were researchers who conducted interviews with individual families and made an online electronic survey available for people to complete. The information they have gathered will hopefully help with the development of improved drug treatments in the future. Thank you to all who made the extra effort to participate in these interviews and surveys.

Following the initial sessions, Professor Roz Anderson, of the University of Sunderland, talked about her team's research into "pro drug" therapy. Lisa Frost, whose PhD in this area we are currently funding, was also in attendance. This work at Sunderland has progressed extremely well and shows real promise of producing significant improvements in treatments for Cystinosis. However, these are some time off in the future. The presentation from Professor Anderson also covered her work studying proteomics, which is funded by the Cystinosis Research Network (USA). In addition, she spoke a little about gene therapy work that she had seen presented at another event, which she felt should be highlighted to us all. This work had previously been presented at the 2010 international conference in Italy and appears promising. Genetic research has been on-going for other conditions for some time. However, this is not a quick or easy path to follow. Prof Anderson was keen to stress that the various research programmes should be able to work in partnership together, and not in competition, and our event allowed these researchers to meet, hear about each other's work and develop professional relationships.

We then heard from Dr Patrick Harrison of University College Cork, who is involved in genetic therapy research. Dr Harrison explained just how small the defective gene parts he is researching are, and just how difficult this type of research is. As a comparison, 20 years has been spent on researching cystic fibrosis gene therapy and it still has not been cracked. His work is funded by the Cystinosis Foundation Ireland and the Cystinosis Research Foundation (USA).

A short Q&A panel session closed the very busy day and our brains were all a little tired after absorbing so much information in such a small space of time. The time constraints of a one day event do mean that attendees are bombarded with information and have little "free" time during the day. However, we really feel this was a worthwhile event.

We must make a final note of thanks to the two organisations that helped fund the conference. First, the Martin Currie Charitable Foundation, who we were introduced to us through our new member, Neil Hutchison. We hope they will be on-going supporters of the Foundation. Secondly, Orphan Europe, who turned up with a strong team of representatives and had a small stand at the conference. They handed out copies of the new booklet on Cystinosis aimed at children and the new board game developed by Cystinosis Foundation USA and Orphan Europe.

Will we do this again? The answer is “yes, definitely”! However, it may not be for a couple of years as there is an international conference expected next year in France.

Thank you again to everyone that came along. It was good to meet you and we look forward to seeing you again at the next conference.

**Roy Forsyth**

## Fundraising report

September 2011

Only 9 months after I reported that our research funds appeal had raised over two hundred thousand pounds, it is incredibly pleasing for me to be able to report that we have now just topped the two hundred and sixty five thousand pounds mark – at exactly **£265,208.04**. The appeal hoped to raise just one hundred thousand to support research of whatever sort into better treatments or understanding of cystinosis – so to continue raising funds in this way is fantastic.

At this moment, we have already paid out over one hundred and ninety four thousand pounds and reserved another fifty six thousand for future years, with several other projects under consideration.

The first major area we have funded has been the Prodrug project at the University of Sunderland, led by Dr Roz Anderson, which is testing new drug formulations which it is hoped will deliver the benefits of cystagon directly to cells, so avoiding the most obvious side effects and also requiring much lower doses. We have also recently bought some equipment for Roz, a HPLC unit, which was vital for the work and replaces a very old unit that they were managing to keep working. The total funded so far or committed to Sunderland is £199,943.

The second major area we have funded is work at the Robert Gordon University in Aberdeen, led by Dr Don Cairns. This research is looking at an alternative form of cystagon that can be administered in different ways and we have sponsored this work, paid for a Vacuum Oven, a Refrigerated Centrifuge and supported promotion of their research. The total funded so far or committed to Sunderland is £36,111.

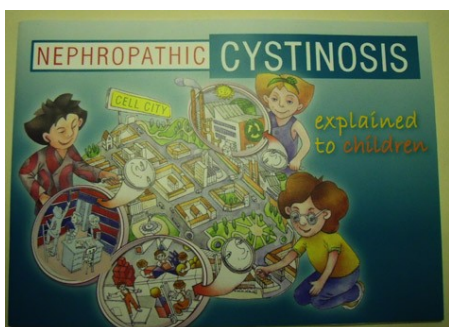
On behalf of the Foundation & the Trustees, can I take this opportunity to again say a huge thank-you to everyone that has raised funds of whatever amount in whatever way – the range of projects is stunning, from sponsored bike rides, walks, marathons, golf events, quiz nights, bridge events, cake making, tea towels, knitted animals – the list is almost endless and continues to grow.

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 raised our appeal target to £300,000 last year and are already halfway to that new target thanks to all our supporters efforts.

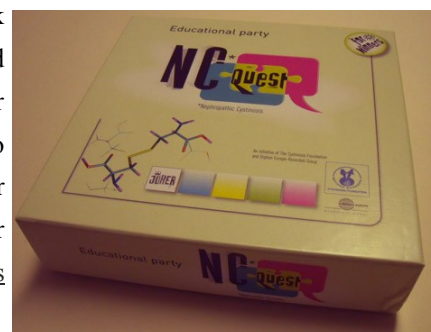
Gareth Jackson

## Game and Booklet

**We're pleased to announce Nephroathic Cystinosis Quest, a board game about Cystinosis.** The game, is aimed at those 7 years old and above, but can be enjoyed by all ages. We have a limited supply of games courtesy of the Cystinosis Foundation USA and Orphan Europe. The game is free, except for postage costs, which we can advise on at the time of



purchase. Please note there is a limited stock and sales are on a first come, first served basis. In addition, we have a small number of books that explains Cystinosis to children. Again, the book is free, except for postage costs. If anyone would like either the game or book, please [Contact Us](#) directly via the website.



## Events since last newsletter

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 and tell us about it as well for use in future newsletters and for the website.

### **Singapore Half Marathon**

On December 5th 2010, Chris and Erin Neesham ran the Singapore Half Marathon and successfully raised close to £3,000 for the Cystinosis Foundation UK. We are hugely grateful to them both for running through the gruelling heat and raising such a marvellous amount. This will help us continue funding research now and in the future.

### **The Folkestone & Hythe Orchestral Society's Annual Summer Concert**

A concert was held on Saturday 9th July 2011 at the Saga Pavilion, Sandgate. The Society elected the Cystinosis Foundation UK as their supported charity, for which we are very grateful. Vice president and violinist, Derek Kemp, along with his daughter and fellow violinist, Katie, highlighted the Foundation to the Society after Martin (Derek's son) informed them about Elliott Mace (his friend's son) who had been diagnosed with cystinosis in 2009. John Terry travelled to Folkestone and gave a talk to the audience about the charity. The orchestra played Dvorak's 7th Symphony. A total of £725 was raised from a raffle, a donation from the orchestra and donations from the audience.

### **Marathon Efforts**

Martin Kemp, who ran the London Marathon on the 17th April 2011 in aid of the Cystinosis Foundation UK managed to raise a total of over £3,000. The team managed to get "spotted" by Radio 5 and gave them an interview about cystinosis raising awareness. Thank you "Team Kempy"



"Team Kempy" being interviewed with Radio 5

### **Sheffield Sponsored Walk**

On 9th April 2011, a bumper 11 mile sponsored walk took place in Sheffield in support of the Cystinosis Foundation UK. The walk was organised by Mark Hewitt, who wanted to support his best friends, Paula and Paul Ingham, whose 4 year old son, Harley, was diagnosed with cystinosis. A whopping £8,576 has been raised by the 60 people who took part in the 11 mile walk and we'd like to thank each and every person who was involved the organisers, the walkers and all those who donated gifts, the use of facilities or offered sponsorship. This is a tremendous amount of money that will go towards cystinosis research, whilst the walk itself is probably the largest participation event ever organised for the Foundation.

### **Charity Football Match**

The Cystinosis Foundation UK would like to thank all the players and members of the 118th and 92nd Boys Brigade Old Boys football teams for raising £550 that will help fund cystinosis research. The teams turned out for a charity game on May 14th, with the proceeds being split between Cancer Research UK and the Cystinosis Foundation UK, as members of both teams have links with those affected by cancer and cystinosis.



### **Sky Dive**

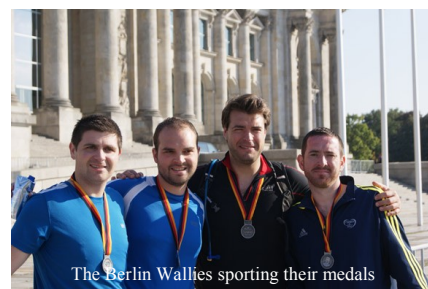
A sky diving teacher and a week of school fundraising events has raised over £3,000 for the Cystinosis Foundation UK! After hearing how several members of one of her student's families has cystinosis, teaching assistant Di Lakin decided to support the family by helping

to raise money for the Cystinosis Foundation UK. First up was a sky dive for Di and her daughter, Nicola (pictured), followed by a full week of fundraising events at Di's school, Derby Moor Community Sports College.



### **Berlin Wallies**

A huge congratulations to the Berlin Wallies who completed the Berlin Marathon on Sunday 25th September and raised a whopping £7,057.40 for the Cystinosis Foundation UK in the process. The team, consisting of Neil Hutchison, Duncan Farrar, Dan Norbury and Charlie Tulip all completed the marathon in times ranging from 4 hours 19 minutes to a heroic 5 hours 36 minutes from one of the team whose lower left leg gave in after 10 miles.



The Berlin Wallies sporting their medals

### **Hair raising Challenge**

A big well done to Jessica Kemp who, challenged her friends to sponsor her £200 in return for which she would shave off her golden locks. Within 2 days of announcing her hair-raising challenge, Jessica has succeeded in raising over £250 and, true to her word to ensure her friends were not short-changed, took a razor to her hair. Her partner Darren also had his chest waxed and a total of £470 was raised.



## **Garage Sale**

Congratulations, and a big thank you, to Amy Mace who managed to raise £220 from her garage sale on September 4th. Amy participated in the Capel-le Ferne garage safari. Participants were able to buy a map of various garage sales taking place in Capel-le Ferne (near Folkestone, Kent) to visit at their leisure over the day. All proceeds from the sale have gone to the Cystinosis Foundation UK, as Amy's son, 3 year old Elliot, has cystinosis. Elliot also took part in the day, handing out leaflets about cystinosis to prospective buyers. (pictured below)

We are very grateful to Amy for her fundraising efforts, as well as her mum, Christine, and auntie, Denise, who both assisted in organising the stall. A special thank you must also go to Elliot for his part in the day. Well done to you all!



## **Sock Puppets**

We are grateful for the efforts of Corinne & David Yates, who have been busy creating sock puppets in order to raise money for the Cystinosis Foundation UK. However, the pair have not only given up their time to create and sell these cheeky little creatures, but they pay for the raw materials, so 100% of the money raised goes straight to the Foundation.

*"They make great toys for children but also adults seem to like them for their cars or desks."* says Corinne, whose grandson Edward has cystinosis.

*"They are all made out of one or a pair of new socks. Our contribution is that we pay for the socks, stuffing, etc. and then make and sell them. Whatever anyone pays, 100% will go to the Cystinosis Foundation"*

The Yates family launched "The Sock Drawer" website in August. This all came about when Edward's nana, having sold many sock creatures to raise funds for the foundation, wondered what to do with all the photos she had of them. Edward's Uncle Pat set up the site [www.thesockdrawer.co.uk](http://www.thesockdrawer.co.uk),



which offered a peek at the creatures and a monthly draw to win one of four. Over three months it raised just over £300 including gift aid.

The draw is now closed but the site is open for donations and for ordering sock creatures. So please feel free to take a look. Edward's nana and grandad have the sewing machine at the ready!

## **Grandmother's swim**

Ethne Wright (grandmother of Darcy Benford) successfully swam 200 lengths of the pool in Esporta Belfast on 5th October 2011 in a time of 2 hours and 50 minutes. This was quite a challenge as she only learned to swim 6 years ago. She did this to help fund research into improvements in the medication of cystinosis. A total of £2500 was raised including £200 which Darcy raised through sponsors for her Nanna from her school.



## **Run the DAM**

On October 16th 2011 Simon Whittle and Dave ran the Amsterdam Marathon and raised £3281.20



## **Continued Support**

The Foundation would like to thank Elizabeth Forsyth (granny of Lena Forsyth) for all her sterling work over the last eight years.

In total this year she has raised over £5100 from events such as a concert, car boot sales, craft sales, distributing quizzes to local shops, who sell them on behalf of the charity, and selling the quizzes herself. She has attended numerous car boots selling items she has been donated and to craft sales where she sells the knitwear she makes, with the help of her husband Jimmy.



## **Cycle in Memory of Acacia**

Acacia Dawn Robinson was only 4 months old when, tragically, severe heart complications ended her life prematurely in hospital in Brisbane Australia. As a mark of gratitude for the support the family received, Acacia's grandparents, Michael and Julia Robinson, decided to raise funds for an Australian charity Heartkids and a UK based children's charity by cycling 173 miles around East Anglia. The lives of Michael and Julia had recently been touched by two children with cystinosis, so the Cystinosis Foundation UK received a donation of £700 from Michael and Julia's cycle in memory of Acacia. We would like to take this opportunity to thank Michael and Julia for supporting the Foundation, whilst offering our deepest sympathies to the family for their loss of Acacia.



## Orphan Europe UK Ltd

**Orphan Europe were one of our sponsors at the Cystinosis UK conference held in Manchester in September this year for whom we were very grateful . This article explains who they are and what their involvement is with the Cystinosis community.**

### **Serving the Unmet Clinical Needs of patients with Rare Disorders**

Many of our members will be familiar with the name of Orphan Europe, who provide CYSTAGON® (cysteamine bitartrate) capsules in varying doses, licensed for the treatment of nephropathic cystinosis. But what is the history & mission of this company?

Orphan Europe was founded in 1990 with the aim to provide treatment for patients with unmet medical needs suffering from rare diseases. The UK subsidiary was established in 1996.

Today Orphan Europe provides nine orphan products to patients all over the world with the help of 130 medical, scientific and marketing specialists in over 15 countries, including the United States and the Middle East. Orphan Europe has developed excellent local & international relationships with clinicians, researchers, patient groups, health authorities and partnering companies. The company enjoys an unmatched understanding of the European regulatory process required to gain approval for the marketing of orphan drugs and its entire infrastructure, is adapted to serve the particular requirements of niche patient groups.

Orphan Europe is evidently committed to the improvement of knowledge about rare diseases among health care professionals. This is exemplified by the Orphan Europe Academy that provides unique, independent, high-level education on rare diseases.

### **Expertise in Orphan Drug Research & Development**

Standard clinical trials are not feasible for orphan drug development. Patients exhibit a wide range of heterogeneous symptoms, often needing individually tailored treatment regimens. The use of placebo controlled trials is not ethically possible and standardized treatments are not available for comparison. Since the very few patients are scattered all over the world, it is virtually impossible to bring them together in one trial.

Working closely with physicians, Orphan Europe designs tailor-made clinical programmes that define the key evaluation criteria for each drug, involving a unique external network of experts who support and cooperate closely with the company.

### **Regulatory Affairs**

In 2009, Orphan Europe received its 7th market authorisation in Europe.

Building a marketing authorization file with very low patient numbers requires specialist knowledge and a solid working relationship with regulatory authorities. Orphan drugs undergo the same rigorous review processes as other pharmaceutical products. They therefore need to meet the same high standards of quality, safety and efficacy.

Orphan Europe now has 21 years of worldwide regulatory experience with regard to orphan drug designation & provision, all Orphan Europe products being supported by a comprehensive pharmacovigilance & medical information facility.

Orphan Europe personnel are always conscious that patients are waiting for treatment.

## Cure Cystinosis International Registry



The Cystinosis Research Foundation in collaboration with the Cystinosis Foundation UK and other key Cystinosis leaders and organisations around the world have launched a patient-based international Cystinosis registry.

The Cure Cystinosis International Registry ([www.cystinosisregistry.org](http://www.cystinosisregistry.org)) will provide an opportunity for those with Cystinosis to be involved in research that will help develop and test new therapies and develop a cure for Cystinosis. The registry resource will connect the stakeholders in the Cystinosis community – the scientists, researchers, clinicians, pharmaceutical companies, patients and families – and provide them with resources that have never been available in one place before. The Cure Cystinosis International Registry is dedicated to improving the lives of those affected by Cystinosis and to finding a cure for Cystinosis.

The Cystinosis Foundation UK is a supporter of the registry and we encourage all those with Cystinosis in the UK to register to help improve the quality of information in the database, which will aid researchers and professionals in their work.

The information provided on the Cure Cystinosis International Registry has been developed with direction and advice from the Cure Cystinosis International Registry Medical and Scientific Council, a group of experts in Cystinosis, and the Cure Cystinosis International Registry Advisory Board.

All the information in the registry can only be accessed by approved professionals who cannot see which individuals have provided their data.

**Contacts**      **Email** : [advice@cystinosis.org.uk](mailto:advice@cystinosis.org.uk)      **Website** : [www.cystinosis.org.uk](http://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](http://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](http://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!

## Hot off the press wrist bands for sale



We have a new 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.....

DISCLAIMER: Information provided, either verbally or in written form, is purely of an advisory level and we recommend that you consult your doctor before making any decisions regarding the future of the patient concerned.