pkd charity newsletter
Polycystic Kidney Disease
www.pkdcharity.org.uk Issue 9 - Autumn 2010

The PKD Charity turns 10

Well done! PKD Charity supporters, like Chris Ackroyd (pictured), have helped us almost double our income. Read more on page 5.

A very warm welcome to our Autumn 2010 newsletter, which we now send to nearly 1000 individuals and families, as well as to every renal unit in the UK.

In December 2010, we celebrate the 10th Anniversary of the PKD Charity, which was founded by Pam Hooley and Dr Anand Saggar. Anand is the Chairman of our Scientific, Clinical and Research Advisory Board, and we hope to bring you news in our next issue of some research initiatives we expect to be able to fund thanks to your support and generosity.

This issue is packed with lovely stories from our supporters, whose enthusiasm and energy generate the funds to publish this newsletter and all our other activities. We hope you will be inspired by the different types of fundraising we're involved with and consider having a go yourself. Contact Esther, our fundraising manager, for ideas and support.

As you will read, we have several local groups who are committed to raising money for the charity on a regular basis. Donations by standing order have risen hugely and we've included the form again in this issue for anyone thinking about supporting us in this way. Every penny counts and no amount is too small. Thank you all!

As a result of all your increased fundraising activities and donations, in the financial year to 31 March 2010 we nearly doubled our income. Download the report and accounts from our website if you'd like to read the details.

Many people ask us about ADPKD and children, in particular whether to test a child whose parent is affected. Great Ormond Street Hospital has produced an excellent leaflet which covers testing as well as symptoms and treatments. You can read an excerpt inside and find links to the full version.

The Renal Tsar Dr Donal O'Donoghue writes on page 3. Do let us know if you have questions that you'd like him to answer on anything related to kidney patient care in the NHS or the latest developments in transplantation and dialysis.

We're delighted to publish this newsletter during our first International Scientific and Medical Conference on ADPKD. Twenty-five of the world's leading renal scientists, researchers and doctors will present to nearly 200 delegates from 20 countries. Topics covered include the latest known science and genetics, worldwide research and clinical trials, current and future treatments and therapies, and the top issues in clinical management of adults and children.

Keynote speakers include British-born Dr Peter Harris, who was head of the European Consortium that identified the PKD1 gene in 1994 and identified the gene for ARPKD in 2002. After the conference, you can view selected presentations on our website and we'll be publishing summaries in the next newsletter.

Dr Peter Harris, Mayo Clinic, USA.

As always, if there is an issue that you would like us to cover in our newsletter, information leaflets, or on our website or at patient days, please email me on: tess@pkdcharity.org.uk. If you would like a copy of our information leaflets and can't use the internet, please phone our helpline on 0300 111 1234.

Finally, remember to follow us on Twitter and Facebook. Our Facebook page has over 700 fans – let's try to make it 1000 by the New Year! We can also link to your own pages if you're fundraising or posting on PKD. See the links on our website or go to www.twitter.com and www.facebook.com and search for us.

New hope for transplant patients

A study published in The Lancet could result in hundreds of lives a year being saved. Research has found that in patients undergoing their first kidney transplants, after five years there was no difference between kidneys from donors who were brain dead but whose hearts were still beating and those who had a controlled-cardiac death (people with irreversible brain injury where life support is withdrawn).

Until now kidneys from cardiac-death donors were believed to have less chance of success compared to brain-death donors. The findings suggest that the outcomes between the two are actually the same. However, it is important to stress that this does not include the many cardiac deaths that occur in emergency situations.

You can read more at: http://tinyurl.com/nhs-donation

The PKD Charity, Foxhill Close, Langwith Road, Scarcliffe S44 6TH
Tel 0300 111 1234 Email info@pkdcharity.org.uk

Advice from Great Ormond Street Hospital on ADPKD in children

Great Ormond Street Hospital has produced an excellent information sheet on ADPKD in children. They've very kindly allowed us to include excerpts below. You can read the full article on their website at: tinyurl.com/ADPKD-GOSH.

What are the symptoms of ADPKD?

Children can be affected in the same way as adults. ADPKD usually causes fluid-filled cysts to form in the kidneys, which press on the surrounding normal kidney tissue and stop the kidney working properly. Fortunately, if cysts are diagnosed, it is usually only a small number and kidney function in the vast majority of children is normal or only mildly impaired. Very rarely do children experience complications, such as increased blood pressure, pain, blood or protein in the urine or kidney stones. However, in a very small number of children, end-stage kidney failure can occur, requiring dialysis or transplantation. It is extremely unusual for children to have complications from ADPKD outside the kidney, such as aneurysms (ballooning of a blood vessel) in the brain. These aneurysms can potentially rupture, leading to severe bleeding inside the head. This complication has not been reported in children, but some experts recommend screening for it in late adolescence, if other family members have aneurysms.

What treatments are available?

There are no specific treatments that stop cysts developing in children at the moment, but there are trials of promising new treatments in adults, for which we await the outcome. Therefore, the only proven, safe treatment that we can currently offer to children is directed at specific symptoms, such as elevated blood pressure. High blood pressure not only accelerates the decline in kidney function but also affects other organs, such as the heart and blood vessels. In one study, half of all children with ADPKD have hypertension (high blood pressure). Moreover, enlargement of the heart muscle was already seen in children whose blood pressure was above average but still in the normal range. Yet this increase in heart size could be prevented with adequate treatment. It is therefore recommended that children of affected parents have their blood pressure checked once a year from about school age, when blood pressure measurements are more easily obtainable and accurate. If blood pressure is elevated, your child should be seen by a kidney specialist (paediatric nephrologist) to start appropriate treatment.

Should my children be tested?

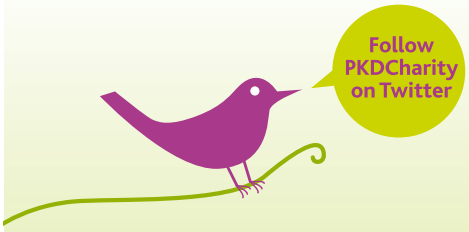
The decision to test your children for the disease should be carefully discussed within your family and with your or your child's doctor. The main argument against testing is that, in the absence of an effective treatment, establishing the diagnosis will provide no immediate benefits, but will burden your child with the knowledge of having a serious disease and this may affect his or her ability to obtain life or health insurance, a mortgage or certain jobs in the future. By delaying testing, you can let your child decide when he or she is old enough to do so, whether they wish to know the diagnosis. The main argument for testing is that it will provide certainty, so that children who have not inherited the disease can stop worrying about being affected and do not need extra visits to the doctor. Those shown to have inherited the disease can start to come to terms with the diagnosis. You will need to consider these arguments in order to find the right solution for your family. Currently, testing of children is not routinely recommended, but this would obviously change if effective treatment became available.

How can my child be tested?

If you want to have your child or children tested, there are different ways to do it.

Ultrasound: This is the most common method used as it is easily available. However, it can be misleading, as single cysts can occur in people without ADPKD. Moreover, the first cyst can occur as late as the fourth decade of life so the absence of cysts in children does not rule out the presence of the disease. In one study, 62% of children affected by ADPKD had cysts on renal ultrasound by five years of age and 78% by 18 years of age.

Genetic testing: This is the most accurate method. If a mutation in an ADPKD gene is identified in the family, the child can easily be tested for the presence of this mutation. In cases where no mutation is identified, something called linkage analysis can be done if two or more affected family members are available. This analysis uses markers on the chromosomal regions that contain the ADPKD genes to discriminate between the two copies to identify which one is likely to harbour the mutation and if the child has inherited that particular copy. Genetic testing for ADPKD is very labour-intensive and expensive due to the complexity of the affected genes and so is currently not performed routinely. This is likely to change in the near future as progress in the technology will make it much easier and cheaper to carry out these tests.



PKD Charity is using Facebook & Twitter!

Do you Tweet? Or use Facebook? If so, please follow us on Twitter and our Facebook fan page. You can follow the links on our website to get you started.

Twitter is a free service that lets you keep in touch with people through the exchange of quick, frequent answers to one simple question: What's happening? Join today to start receiving PKD Charity's tweets!

Join up now and follow 'PKDCharity'!

Your questions answered: Aneurysms in PKD – should we screen?

Below is an edited excerpt from Dr Donal O'Donoghue's blog. Donal (pictured above), the Renal Tsar for England, has kindly agreed to make this a regular column, so please do write in with any questions you'd like him to address.

Q: Hi Donal, I'm writing from outside a Parisian café, on my way back from Zurich after a weekend hosted by a big pharma drug company. An interesting couple of days. It was a facilitated open forum, with 13 transplant patients from Europe and the US, where they seemed to genuinely want to know what they might think of doing – above and beyond producing drugs – that would help patients. I was prompted to write by a post to the PKD email group that Tess Harris runs[1] which suggested it was standard procedure in France for dialysis-transplant patients to have a brain scan. I've been aware for some time that PKD patients have an above average risk of getting aneurysms in the brain. I'd never heard that dialysis patients in general had risks along these lines. I was just wondering what the attitude is to this in the UK. Best wishes, Andy Williamson, Chair, Guys & St Thomas' KPA

A: Dear Andy, I'm pleased to hear you're enjoying Paris and that the PKD email group is thriving. Inter-cranial aneurysm rupture is a rare but devastating complication of autosomal dominant PKD. It occurs in about 1 in 20 people with PKD without a family history but in up to nearly 1 in 5 people in whom there is a family history of rupture. This compares with about 1% in the general population. Rupture also occurs on average ten years younger in people with PKD; the youngest reported case being a six-year-old boy. Aneurysm rupture in both groups is associated with a combined mortality or severe residual disability rate of about 35-55%. However, treatment of aneurysms by either neurosurgical clipping or endovascular treatment also carries an unacceptably high combined mortality and severe disability rate of 21-34%. Some individuals make remarkable recoveries such as cyclist Alberto Contador who collapsed following an aneurysm rupture during the Spanish Vuelta a Asturias in 2004, but who went on to win the Tour de France in 2007, 2009 and 2010.

In the general population, size greater than 1cm, location and history of previous rupture are the major risk factors for rupture but in PKD the situation is less certain and it has been suggested that half of all ruptured aneurysms in PKD are under the 1cm threshold. Aneurysm formation and rupture also appears to run in families and there is some early evidence of genetic linkage and risk. Recent studies have concluded that the outlook for asymptomatic aneurysms in PKD is excellent. The only factor that seemed to correlate with development of new aneurysms was a history of previous rupture. Given this uncertainty we really don't know which patients we should be screening for asymptomatic aneurysms.

Patients who have suffered a previous rupture appear a higher risk group who require lifelong screening. For asymptomatic patients, the presence of a strong family history of rupture (at least two first degree relatives) remains the best predictor of rupture (it increases risk by 4.4 times) and the case can be made for screening this group. The evidence does not support routine screening of all PKD patients but all PKD patients should have good control of blood pressure, be advised about smoking cessation and limiting alcohol consumption to within health limits as these treatable and lifestyle factors are key determinants of rupture. In the future we may have genetic tests that help us assess risk but at the moment there isn't a UK-wide consensus on the approach that should be taken in people with PKD who might have asymptomatic aneurysms.

Best wishes, Donal.

Read the full blog at: renaltsar.blogspot.com where Donal is happy to receive comments or questions.

[1] **You can sign up to the group at: uk.groups.yahoo.com/group/PKD_uk/**

A reader writes...

Dear PKD Charity,

I am 62 years old, went for a pre-op prior to minor knee surgery only to be told I had high blood pressure. My GP ordered a scan as well as treated the BP and I was diagnosed with PKD. My doctor told me **nothing** at all about the disease, apart from the fact that it was inherited and I should tell my two children. He said I should arrange for a blood and urine test in three months, but there was no treatment.

I have enclosed a donation of £50 as a thank you for your clear, concise, readable booklet on ADPKD which I downloaded from the internet and found most informative.

With grateful thanks,
Jenny Y, Lewes

Thank you so much for your donation and kind words and we're glad you found our information helpful. It was disappointing to read about your experience with your GP. Unfortunately awareness amongst health professionals is not always as high as we'd like, and it's one of the areas we're working hard to improve.

With our best wishes,
Tess

Research round-up

There are now over 14 global clinical trials exploring therapies for PKD and many more basic research studies. Sadly we can't cover them all, but we've highlighted a few of particular interest below.

Heart disease, high blood pressure/hypertension and PKD

High blood pressure (BP) or hypertension occurs in approximately 40% of PKD patients by their twenties and is associated with decreased kidney function and increased kidney and left heart size, which can lead to congestive heart disease. It may surprise you to know that PKD patients are more likely to die from heart and vascular complications of PKD, rather than renal failure, so it is extremely important to maintain BP within a normal range.

As cysts grow, they compress blood vessels in the kidney which reduces blood flow. This leads to a series of events including the release of a protein called angiotensin II (AII). AII acts to restore blood flow and pressure to normal levels in several ways, including the constriction of blood vessels throughout the body. However, as the cysts increasingly prevent pressure and flow in the kidney, a broken feedback loop occurs, with AII continuing to be released. The result is high BP or hypertension.

"PKD patients are more likely to die from heart and vascular complications of PKD than renal failure"

The PKD Foundation in the US is collaborating on a major US government-funded study called HALT PKD. This is the first clinical trial to look at BP control and its effects on kidney and heart size in PKD patients. It is assessing the effectiveness of the anti-hypertensive drugs telmisartan and lisinopril on PKD progression and its cardiovascular complications in two different levels of kidney function. Due for completion in April 2013, we look forward to the results.

Other trials exploring BP control include an American study to determine the effect of pravastatin treatment on renal and cardiovascular disease in children and young adults with ADPKD, and a Danish study examining the effect of high and low sodium diets on kidney function and on hormones that affect the kidney. Two Japanese studies are also investigating the control of BP. One will evaluate the calcium channel blocker (CCB) candesartan in treating hypertension in ADPKD patients, and the other will compare combination therapy in patients whose BP is not controlled with ARB (angiotensin receptor blocker) alone. Either ACE (angiotensin converting enzyme inhibitor) or CCB will be added to their regimen and the results compared to those on ARB alone.

Somatostatin

Another area of research is somatostatin. Patients with ADPKD have a faster decline in glomerular filtration rate (GFR) than those with other renal diseases. Also they don't seem to benefit to the same extent from ACE inhibitor BP drugs. A reasonable explanation is that in ADPKD, progression is largely dependent on the development and growth of cysts and secondary disruption of normal tissue. Together with reducing BP and proteinuria, and limiting the effects of potential promoters of progression such as dyslipidemia, chronic hyperglycaemia or smoking, renoprotective interventions need to correct specifically the dysregulation of epithelial cell growth, secretion, and matrix interactions characteristic of ADPKD.

Evidence that specific receptors for somatostatin are present in kidney tissue gives rise to the possibility that somatostatin treatment might inhibit fluid formation and induce the shrinking of renal cysts. Somatostatin is a naturally occurring hormone in humans that binds to a receptor protein on the surface of renal cells and blocks cAMP (an intracellular second messenger) formation inside the cell. This reduces both fluid secretion and cell proliferation and results in decreased cyst and kidney size.

"Percent increase of total kidney volume was significantly lower in patients on somatostatin than those taking the placebo"

A recent six-month pilot study in Italy demonstrated the percent increase of total kidney volume was significantly lower in patients on somatostatin than on placebo. A long-term study is now documenting the efficacy of somatostatin treatment in preventing further increase or even reducing the total kidney volume and the renal volume taken up by small cysts. Due for completion in December 2011, the trial is comparing the effects on disease progression of a three-year treatment regimen using long-acting somatostatin or placebo in patients with ADPKD and normal renal function, or mild to moderate renal insufficiency. We await the results with interest.

With thanks to the PKD Foundation (www.pkdcure.org) for the above information. You can read about more global trials by visiting the Clinical Trials website at: www.clinicaltrials.gov and searching for 'polycystic kidney disease'.

Ciliopathy Alliance

We are pleased to announce the launch of the Ciliopathy Alliance UK (CAUK), of which the PKD Charity is a founder member. The Alliance was established in March 2010 to promote awareness, share knowledge and understanding, and encourage and facilitate research into diseases caused by defects in the function or structure of cilia. Cilia are small hair-like protuberances found on the surface of virtually every cell in the human body, including the kidney. Once thought to be vestigial organs, defective cilia are now understood to underlie a number of genetic conditions – of which one of the most important is PKD. The official launch of CAUK will take place in London on 29 November 2010.

Find out more at: www.ciliopathyalliance.org.uk

Fundraising

2010 has proved to be an excellent year for fundraising. From the weird and wonderful events you've organised through to the miles and miles of roads and tracks pounded by runners and walkers, we've been overwhelmed and delighted by your support.

World Kidney Day

March 11 2010 was World Kidney Day and many of you got involved by holding collections to raise funds and awareness for PKD or by organising your own events. Brian Stronge organised a Dog Racing Night raising more than £400 for both the PKD Charity and his local air ambulance. Meanwhile in the North West, Denise Thorp organised a collection with her friend Lesley Swinnerton at Marks & Spencer in Prestwich. They really wowed the shoppers raising an incredible £260 in just one day! Elaine Hanson held a collection at Barnsley football club on match day and, together with her husband, raised over £96.

Denise Thorp and Lesley Swinnerton

At Warlingham village primary school Debbie Sheppard's Year 5 class decided to raise money for PKD on World Kidney Day by asking pupils to wear their pyjamas to school for a donation of £1. Children could also pay an extra 50p if they wanted their teacher to wear pyjamas! Students chose the PKD Charity as Debbie's two-year-old son has PKD. On the day itself pupils and staff raised a magnificent £275.

Warlingham village primary school

Runners

Great Manchester Run

Once again this year, we had several runners in the PKD team running the Great Manchester Run on Sunday 16 May. As the biggest 10k in Europe, more than 40,000 runners took part, raising thousands of pounds for local, national and international charities. Taking part for their second consecutive year were mother and son team, Louise and Ryan McGealy. This year Ryan finished in just 49 minutes, with Louise just a few footsteps behind at 52 minutes. Their fundraising was boosted by family member, Edwina Graham, a long-standing supporter of the charity who herself is affected by PKD. Edwina wrote to all the local newspapers to help raise publicity which promoted complete strangers to make donations! Edwina's husband also took part, having secured his own place. In total the amount raised by the McGealy and Graham team was an incredible £897!

Louise and Ryan McGealy

Mr Graham

London Marathon

This year's London marathon saw no less than four runners taking part to raise funds for the PKD Charity. Matt Johnson finished in 4 hours and 32 minutes and raised a magnificent £1108.25 for the PKD Charity and the same amount for the charity S.P.R.I.N.G. Dean Fitzpatrick, whose father has PKD, raised £769 including Gift Aid and completed the 26-mile course in 3 hours 55 minutes. Eighteen-year-old Robert Hutchinson ran his first ever London marathon raising £325 and finished in 5 hours and 23 minutes. Robert's mother has PKD and has recently undergone a successful kidney transplant.

Chris Ackroyd entered this year's Paris marathon to raise funds for PKD. Chris raised over £1200 and achieved a time of 4 hours 9 minutes and 56 seconds in his first ever marathon. Other runners included Jo Neville who took part in the Reading half marathon in March, Fraser Henderson who entered the Aberfeldy Half Ironman Triathlon and Sam Clackson who ran in the South Downs marathon in June of this year and who has so far raised over £1000. Sophie Jones entered her local Race for Life, match funded the £410 she raised for cancer charities, and donated it to the PKD Charity in memory of her friend Jessica Neal. Andrew Holmes and Graham Williamson took part in Ireland's Dingle marathon and even entered the Newham 10k in March as part of their training. Donations are still coming in but so far they have exceeded their fundraising target of £1000.

Other Events

This summer it has certainly been true to say that what goes up must come down! Kairit Banks climbed up Snowdon on 2 July and came down again to tell the tale. She is currently collecting sponsorship from all of her supporters.

Charlie Douglas found a quicker way to come down, by bungee-jumping back in May, and has raised more than £1,500 so far. Charlie's mum has PKD and he was raising the funds on her behalf.

In other events, Madeline Freelove held a coffee morning at Hemel Hempstead church on 6th June and raised £110. Jane Brooker organised a dance festival to raise funds for PKD and Sheena Sefiani raised £320 from a fundraising event in her local deli in June. The Lockyer family held a garden party in memory of Peter Lockyer, one of the PKD Charity's founders and raised £955.

Taking to the water was Grant Devlin, who entered the world's biggest swimathon, raising more than £1600 and Lisa Mantle who took on the challenge of the Blue Mile swim and raised £335.

Taking to the streets were Mini enthusiasts Lisa-Marie Shepherd and

Lee Donaldson, who drove their mini from the West Midlands to Turin to re-create The Italian Job, raising over £1,200 in the process. Andrew Hartley organised a sponsored walk from Shropshire all the way to Aberystwyth and is still collecting his donations and no doubt nursing his blisters! Neil Whelpton ran, swam and cycled his way to more than £1,200 of donations for the PKD Charity in this year's London triathlon.

Lisa Mantle and a team of friends walked 40 miles across the Yorkshire moors in the dead of night raising over £500.

Anna Siddle of Mail Boxes Etc, together with her customers raised £67.76 in donations from a collection tin in her shop. As Anna said, "It's amazing how all the pennies add up!" Lily Wakelam has also been raising funds through collection boxes and from donations at her mother's funeral and plans to organise a sponsored bike ride next year. Francesca Lockett also got the cycling bug and organised a mammoth Coast to Coast bike ride at the end of April, raising an incredible £2620.80. Francesca organised the event in memory of her father Graham Lockett, who passed away in July 2008. She was supported by a team of other cyclists and support crew including:

Francesca Lockett, Simon Richardson, Alex Bradley, Daniel Bradley, Vicky Wellings, Keith Savage, Cordelia Johnson, Jason Holt, William Guest, Adam Guest (riders); Eileen Lockett, Steven Richardson, Sheena Richardson, Hannah Smith, Alex Zachariah, Dave Abel, Alex Barratt (support crew).

Regular readers will recall that in the summer of 2009, Jon Kennard organised a team of riders and their motorbikes to ride the length and breadth of the country. Jon worked exceptionally hard to organise the event and to fundraise, culminating in him donating the proceeds of £3007.50 to the charity earlier this year.

Our sincere thanks to everyone who has fundraised for the charity so far this year.

Forthcoming Events

October 2010 marks the first ever team entry in the Great South Run for the PKD Charity. Our team of 15 runners will be taking part in this landmark event to raise funds for us and we would love you to give them your support. You can visit www.justgiving.com/pkd to make a donation.

Kairit Banks - Snowdon

Emma Peach, Lee Ballard and Stephanie Brumpton are all taking part in this year's Robin Hood marathon in Nottingham, Lindsey Bridgeman and a group of friends are running the Cardiff half marathon, Zahra Sefinani is running in the London Royal Parks event in October and David Salt is getting in training for the Edinburgh marathon in 2011. Further afield, Kate Faver, currently living in California, is trekking the Himalayas in November and both Matthew Kromolicki and Simon Greenwood are independently climbing Mount Kilimanjaro this autumn.

Once again the PKD Charity is taking part in this year's Small Car Draw. Organised by the Foundation for Social Improvement, the draw gives small charities the chance to sell raffle tickets with a first prize of a brand new Mini. 95.2% of ticket sales go directly to the charity. Thanks to everyone who has bought or sold raffle tickets and good luck in the draw on 14 October.

In our last newsletter we asked you to become a Friend of the PKD Charity by giving a regular donation by standing order. We were absolutely overwhelmed by the response, with more than £2000 annually being pledged. If you would like to join our Friends scheme, simply complete the standing order form inside this newsletter and we'll do the rest.

Last autumn we announced our new Local Groups programme, enabling people around the UK who are affected by PKD to get together to share experiences, raise awareness and fundraise. One of the first to sign up was Amanda Craven who set up her own local group named "Helpkd" in the Bedford area. Amanda's husband, Mike, has PKD and sadly the family lost Mike's mum to PKD recently. This prompted Amanda to get together with her family and set up a local group. Fundraising plans include a 10k run, but so far this year Amanda has already organised a Fun Day in her local village to raise funds for the charity. She aimed to raise £1000 this year, but has already beaten this through the Fun Day, donations and collection tins. Amanda said, "Setting up my own local group has really given me the chance to make a difference to PKD, which is something very close to my heart." If you would like to join Amanda's group or find out more, you can contact her on helpkd@hotmail.co.uk. For more general information on setting up your own local group, please contact our fundraising manager, Esther Wright on 07825 882616 or email: esther@pkdcharity.org.uk. Setting up a group is really easy and we will provide everything you need from t-shirts, to collection tins, posters and leaflets.

Next ADPKD Patient Information Day

Saturday 16 April 2011

9:30am - 4:30pm

Guy's and St Thomas' Hospitals

London, SE1

- Put your questions to our medical experts
- Learn about the latest research
- Share your experiences with others

We're also planning to raise funds with an abseil down Guy's Tower the next day!

See www.pkdcharity.org.uk for more details.