pkd charity newsletter
Polycystic Kidney Disease
www.pkdcharity.org.uk Issue 10 - Spring 2011

Bio-resource bank could speed up PKD research

We are delighted to report on the PKD Charity's recent grant to Professor Pat Wilson for the establishment of a PKD bio-resource bank in the UK. Prof. Wilson, of the Centre for Nephrology, University College London said, "This award should greatly help speed-up PKD research in the UK and is the start of very exciting collaboration."

This award was only possible through the hard work and generosity of our supporters, for which we're very grateful. Tess Harris, PKD Chairman, comments, "The award meets our goal of funding PKD research in the UK, which has received minimal support from the Government. This unique bio-resource will become a crucial component in laboratory research – furthering our mission to improve the quality and duration of life for patients with dominant and recessive forms of PKD.

"The long-term success and acceleration of PKD research depends on well-characterized human PKD and age-matched normal control tissues, cell lines and animal models that faithfully recapitulate the genotypic and phenotypic characteristics of the disease, together with well-annotated PKD patient cohorts. The grant, which offers the potential for high research value at modest cost (approximately £32,000 over three years), will support a large existing collection started by Professor Wilson in 1993.

> **"This unique bio-resource will become a crucial component in laboratory research"**

The grant will also enable the establishment of a database compatible with those of other UK collections, such as St George's, the Renal Registry and Addenbrooke's. It is hoped that this will be the foundation for a PKD national registry that in turn will accelerate clinical trial testing by quickly identifying sufficiently large cohorts of PKD patients to produce definitive results.

Professor Pat Wilson

Prof. Wilson observed, "This award will facilitate high quality, translational PKD research projects in the UK by enabling us to provide project-ready, controlled sample sets."

Simon Greenwood (right) trekked to the summit of Mount Kilimanjaro, raising £1,565 (see page 6)

Welcome to the PKD Charity Newsletter

Tess Harris, PKD Charity Chairman

In this issue, we announce exciting news about our first significant UK research grant. We have articles on nutrition, insurance, kidney care plans and renal cysts and we celebrate our growing number of magnificent, big-hearted fundraisers.

I'd also like to welcome two new trustees to the charity, Judith Dignum and Edwina Graham, and thank all our newsletter contributors, readers and funders.

Tess Harris, Chairman

Calendar

30 May
Bupa 10K Run, London
More info: bit.ly/em8qLk

June
PKD Chairty Sponsored Walk, London
More info: tess@pkdcharity.org.uk

16 July
PKD Information Day, Royal Free Hospital, London
More info: bit.ly/PKDInfoDay

4 September
Magic Night, Radlett Centre, Hertfordshire
Email: joseph.arazi@sky.com

30 October
Great South Run
More info: bit.ly/em8qLk

Causes, symptoms and effect

The implications of testing for PKD

A reader writes...

Dear PKD Charity,

I enjoyed reading the article in your last issue on the pros and cons of having children tested for PKD. I did, however, wonder about the seemingly tentative advice:

"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, mortgage or future occupation."

In as much as insurance companies now do ask if there is a family history in new applicant questions, and often they will know the family line anyway, it also increasingly seems to be the case that children will be told, or informed another way, not necessarily by the parents. A doctor knowing the parent had PKD is bound to pick it up through a child's high blood pressure for example.

If the article is intimating a child's doctors conspire to elude honest procurement of insurance and the like I am surprised and hope, for them, that they have good indemnity! It would also, more worryingly, invalidate any claim on such an insurance, mortgage application etc.

Although in general I agree with the article and found it balanced, the use of 'may effect' is at this time misleading. 'Will certainly' is closer to the mark - if only through higher premiums or certain exclusions. Sadly, one can't exclude the possibility of being turned down altogether.

Yours,
Mr M Anthony

Detlef Böckenhauer of Great Ormond Street Hospital for Children NHS Trust responds…

This is a very controversial topic: the statement was referring to a situation where a person has no symptoms, but is at-risk of a disease that could have a serious impact on quality of life later on and for which there is as yet no proven treatment that could delay or prevent it. Should that person be tested or not?

Most doctors feel this decision should be made by the person him/herself. Some may want to know, some may not, and that should be respected. In children it is a bit difficult, as they may not be able to understand the complex situation and make an informed decision yet. Many doctors feel that with asymptomatic children diagnostic testing should be deferred until they are old enough to decide for themselves.

This is not a conspiracy to cheat on insurance companies. If they ask for family history that must be addressed truthfully. One could actually turn this around: if an ultrasound is performed and it is normal, then leaving out the at-risk of ADPKD on an insurance form would be cheating, as ultrasound can remain normal until 40 years of age. The situation is different if the child is symptomatic, for instance with high blood pressure. Appropriate work-up of high BP includes a renal ultrasound scan, which may then also reveal the diagnosis.

The situation would also be different if there was proven effective treatment available. Then early diagnosis and appropriate treatment would provide a benefit. But currently, in an asymptomatic child there is no clear benefit from obtaining the diagnosis.

Best wishes,
Detlef Böckenhauer, PhD, FRCPCH
Consultant Paediatric Nephrologist

The insurance implications of testing are complex and far reaching. We spoke to Sarah Angel at the Insurance Surgery (visit www.the-insurance-surgery.co.uk), a specialist broker with wide experience of dealing with PKD and other kidney conditions, for her advice.

Sarah told us that those diagnosed with PKD before the age of 20 would nearly always be refused life insurance. While insurers are not allowed to ask about genetic testing (a moratorium is in place as the only test approved by the Association of British Insurers to date is for Huntingdon's Disease), applicants are, as Mr Anthony says, obliged to submit family history and failure to do so would affect any potential pay out. If you have a family history and have not been tested (i.e. high resolution ultrasound and CT scan) then most insurers would decline life insurance and critical illness cover until you are aged over 50. A very few companies, however, do vary on this criteria and consider applications on an individual basis.

Sarah also explained that although life insurance is possible for those with a family history of PKD, premiums depend on age. Currently the basic increase in premiums for PKD ranges from 50 - 100 per cent. This figure, however, can vary significantly depending on each applicant's additional medical information and general state of health.

As Mr Anthony's letter suggests, insurance can be a difficult area. But, on a brighter note, Sarah was happy to report that over the last seven years she's seen a small but perceptible improvement in approval rates and terms as knowledge of PKD increases.

You can read more about insurance on our website. Although we can't recommend insurers, we have entered into partnership with two companies who specialise in high-risk insurance for people with pre-existing conditions. For every policy sold, they give us a donation at no extra cost to you. You can call the Insurance Surgery on freephone 0800 083 2829, or Freedom Insurance on 01223 454 290. They're very useful starting points but always contact a number of insurers to compare prices. Remember it's useful to have the following information when you call: your e-GFR, creatinine levels and blood pressure.

Getting personal

Tell us about your individual care plan

Lord Darzi's review of the NHS in 2008, which set out Government plans for NHS reform in England for the next ten years, suggested all 15 million patients with long-term conditions be given a personal care plan. A recent letter to NFK's Kidney Life, however, expressed concern that few Individual Care Plans (ICPs) had been produced for renal patients. The shortcomings of this are clear, as chronic health conditions invariably involve overlapping health issues. Pertinently, the letter also asked:

'How can we get ICPs developed by patients with support from renal staff? At the moment I get the impression there is a real risk they will be staff-run for patients. How can patients become empowered to take responsibility in both developing and applying their own ICPs?'

Below is Renal Tsar Dr Donal O'Donoghue's answer, and he'd particularly welcome feedback on this key issue. As Donal emphasises, ICPs are important not only because of overlapping health requirements, but because effective care planning is a lot more than simply giving an individual a standard booklet. It's a process that involves a dialogue between the patient, their family or carers, and the entire healthcare team.

Renal Tsar Dr Donal O'Donoghue responds...

'Care planning is part of ensuring individuals achieve optimal outcomes and meet the aspirations they set themselves. So the beliefs and values of the patient are every bit as important as the diagnostic skills and treatment options of the clinicians. Some years ago, in a kidney unit that will remain nameless, I was told everyone had a care plan. I was delighted and asked if someone could tell me more about it, and if it might be possible to pinch their ideas? I soon realised why my enthusiasm was being met with quizzical looks. I was handed a dialysis prescription chart on a poorly photocopied sheet... I think most people in the kidney world now appreciate that care planning is much more than a traditional treatment plan, but there's still concern that staff may not have enough time for it.

Care planning and the shared decision-making it needs to entail, does take time. In the case of dialysis, for example, patients need to come to terms with the diagnosis and prognosis, to think over and discuss their options. However, an informed and in-control patient supported by their various care teams has a better experience of care and better outcomes than if, as the letter says, 'care planning is staff-run for patients'. The result is often fewer outpatient visits, fewer complications, less anxiety and the need for less inpatient care. Investing in care planning definitely saves time and resources overall.

> **"Effective care planning is a lot more than giving an individual a standard sheet or booklet"**

Our challenge is to make the case for more face-to-face listening and talking time. We need to help lead a cultural change. People should be prompted to bring their questions to clinics and kidney units, to say up-front what they want and to go away with a care plan that reflects this. The question is, how do we make this the norm? And how do we make sure care plans are integrated? A good starting point is our own areas. For renal services that means an individual's plan has input from doctors, nurses, pharmacists, social workers and, where necessary, dialysis technicians or transplant surgeons. We also have to ensure that it meets each patient's priorities and personal goals.

The exciting news is that from April 2011, new arrangements for adults are being put in place in kidney services in England. Multi-professional outpatient visits will receive up to double the payment for physician-only attendances. This will help kidney units target staff to where they can add most value. It should result, for example, in more people receiving pre-emptive transplants and home dialysis.

I believe that the principles of effective care planning are in place, but we need patients to tell us where it's working so we can share their

insights. Ultimately, individuals in our care teams, and that includes patients, are the ones who will make care planning happen. I'm sure there will be some trial and error, but the quicker we get started the quicker we'll see the benefits.'

> **"Investing in care planning saves time and resources overall"**

Please do let us know your experiences – good or bad – so we can pass them on to Donal. Feedback is crucial if care planning is to be a success.

This article is adapted from one that first appeared in the winter edition of Kidney Life magazine, which you can download at: www.kidney.org.uk/k_life/download.html or from Donal's blog: http://renaltsar.blogspot.com

Recycle for PKD

Your old cartridges and phones can raise ££££s! Contact us for a freepost envelope - esther@pkdcharity.org.uk

PKD cysts explained by Professor Pat Wilson

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is characterised by the growth of numerous cysts in the kidneys, and sometimes in other organs. Here we describe what cysts are and how they form.

What is a cyst?

A cyst is a fluid-filled spherical structure lined by a single layer of tubular (epithelial) cells.

Where do cysts occur in the kidney?

Each normal kidney is composed of about a million functional units called nephrons. Each one of these segmented filtering unit structures has a glomerulus at one end attached to a long, convoluted, tubule. This tubule is lined by a single layer of specialised cells of 15 different types that carry out the complex fluid and ion reabsorption functions of the kidney.

In ADPKD, every segment of the nephron can develop cysts. These arise as out-pushings of the tube so that instead of a regular narrow tube, an expanded cystic portion is formed and soon pinches off from the nephron of origin (see figure 1).

How do cysts enlarge?

Once cysts close off from the nephron and become separated, they continue to enlarge by increased cell proliferation, fluid secretion and changes to the extracellular matrix (tissue which supports the cells) surrounding the nephron.

How does cyst growth affect kidney function?

As more and more cysts are formed, fewer and fewer nephrons can function properly. Fortunately the kidney has a large functional reserve capacity and does not need to use all nephrons at once. Therefore, it is not until about 60% of nephrons have stopped functioning (due to cyst growth) that the kidneys begin to fail (see figure 2).

Where else can cysts form?

The most common site, other than the kidney, in which cysts form is the liver, due to excessive growth (proliferation) of the cells lining bile ducts. This causes the liver to enlarge. Although fortunately liver functionality is not usually impaired, this can lead to pain, breathlessness and reduced mobility.

Normal nephron ADPKD Nephron

Figure 1: Diagram of a normal nephron (left panel) comprised of a glomerulus (to filter the blood) attached to a long nephron with specialised regions. During development of ADPKD nephrons (right panel), cystic outpushings can arise from every segment of the nephron. These close off from the nephron of origin early in the developmental process, rendering that nephron non-functional.

(Wilson, P., New Engl. J. Med. 350: 151-164, 2004.)

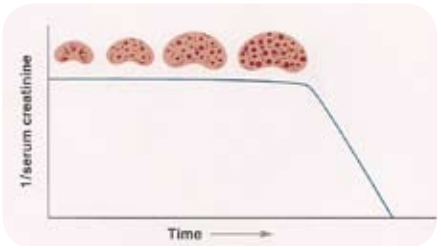

Figure 2: A typical pattern of renal functional decline in ADPKD patients, with respect to an increase in kidney size and cystic volume. Kidney function starts to decline after significant renal enlargement and approximately 60% cystic replacement of the nephrons.

(Wilson P and Goilav, B., Annu. Rev. of Pathol. Mech. Dis 2: 341-368, 2007.)

The importance of vegetable protein in our diet

By Jenny Tschiesche BSc(hons) Dip ION FdSc mBANT - Jenny is a fully qualified nutritional therapist and has extensive experience of PKD

What are plant proteins?

Plant proteins are beans, nuts, peas and soy products.

Why should I include plant proteins in my diet?

They're cheap and healthy – they contain low or trace cholesterol and saturated fat and more beta-carotene, dietary fibre, vitamin C, vitamin E, folate, iron, magnesium and calcium than animal protein. They also taste great and add texture to meals.

Anything else I should know?

Combine two or more vegetable proteins to make up a complete protein, for instance use lentils with rice or chickpeas with seeds (as in hummus).

Lentil burgers

Ingredients: 225g/8oz/1 cup red lentils, 600ml/1 pint/2½ cups water, 2 large carrots – grated, 1 large onion – chopped, 4 tbsp wholemeal flour, 1 tsp mixed herbs, freshly grated nutmeg, freshly ground black pepper.

Method:

1. Put rinsed lentils into a small saucepan. Cover with water.
2. Bring to the boil and put a lid on the pan. Reduce heat. Simmer gently until the lentils are cooked and the water has been absorbed (15-20 minutes).
3. Leave to cool
4. Preheat oven to 400ºF/200ºc/Gas Mark 6. Put lentils into a bowl, add carrots, onion, flour, herbs, nutmeg and pepper. Mix well.
5. Shape into eight burgers. Put on a floured baking tray and bake for 15-20 minutes.

Walnut pesto

Ingredients: 100g basil leaves, 10g flat leaf parsley, 140g walnuts, 5g toasted pine nuts, 10g Pecorino cheese, 250g walnut oil.

Method: Place a pan of water on the stove. Blanch the basil for about 1 minute, drain and place in iced water. Blanch parsley for 1 ½ minutes, drain and place in iced water. Put all the ingredients into a food processor and blend until the mixture forms a paste.

www.healthmatters360.co.uk

It's easier than ever to connect with others affected by PKD

The benefits of peer support within health are well-recognised. In the case of PKD, because it's still so little known, people really appreciate comparing experiences with others who have the same condition. It's often particularly important for people with relatively few symptoms who are unable to access the more established support for those on dialysis or awaiting transplant.

This is why we're expanding the range of our support until there is, we hope, something to suit everyone. You can be confident that whichever method you choose, it will provide the opportunity for those affected by PKD to share information, advice and experiences, and to offer and receive support.

A good starting point is a call to the PKD Charity helpline on 0300 111 1234 (local call rate). The volunteers can offer advice, answer questions and tell you if there's a support group in your area. These convenient local groups allow people to meet face-to-face and are very welcoming. You can ask about local groups by emailing: **info@pkdcharity.org.uk** - and we'd love to hear from you if you're interested in setting up a group.

For those with Internet access we run two thriving online groups. Our Facebook page (**bit.ly/958JOG**) is growing daily and is a good place to post questions or concerns. Our Yahoo group (**bit.ly/ggbwQy**) is another great place to meet others and share experiences. A long-running and very active forum, its members are a friendly bunch and a mine of useful information!

You can also email us with your questions (**info@pkdcharity.org.uk**). A number of medical advisors to the charity can supply general answers to clinical questions, although do always discuss any medical advice or suggestions with your doctor.

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'

Five key points from the PKD Charity International Scientific and Medical Conference

In September 2010 the first PKD Charity International Scientific and Medical Conference, organised by Tess Harris and Dr Anand Saggar, took place, marking the tenth anniversary of the PKD charity. 'PKD – More than a Kidney Disease' brought together over 100 participants from 16 countries to discuss the clinical, genetic and therapeutic aspects of ADPKD. The event aimed to promote greater collaboration between clinicians and scientists on PKD research, increase UK-based research and advance understanding. A full report and a series of webinars of the presentations is available on our website: bit.ly/PKDConf2010. Here are five critical points coming out of the conference.

1. Chronic Kidney Disease (CKD) is a priority in primary care but ADPKD, despite being the most common, genetic, life-threatening form of CKD, is still not well recognised. Initiatives are required to promote recognition and collect data.

2. A web-based, PKD-specific registry is urgently needed in the UK to provide high quality information, compare audit outcomes and facilitate recruitment to clinical trials.

3. The management and monitoring of children with ADPKD and the need to recruit younger participants for clinical trials, was highlighted.

4. ADPKD is usually associated with adult onset of symptoms with end-stage renal disease occurring in late middle age, but there is much phenotypic variability. Next generation sequencing and genome-wide association studies should identify allelic variants associated with disease severity, giving greater insight into disease progression and prognosis.

5. Data from drug therapeutic trials suggests combination therapies may hold promise and stresses the importance of early intervention to delay the progression of ADPKD.

The PKD Newsletter is produced by Jellymould Creative: writer Harriet Powney; editor Jo Johnson; designer Emile Facey.

PKD is on the run!

2011 is going to be a great year to put on your running shoes and raise funds for the PKD Charity. We have a whole host of running events for you to consider, from 10k runs to half-marathons and full marathons.

We have secured our first places at the London Marathon this year. On April 17th Delyth Wanklin and Simon Greenwood will take on this prestigious and gruelling event for the benefit of the charity. To sponsor them, please contact us or visit their Just Giving pages before or after the event: www.justgiving.com/Delyth-wanklyn and www.justgiving.com/SimonGreenwood03. This is a great opportunity to raise awareness about PKD and support the charity – so please dig deep.

We also have places available at the Bupa 10k, which will take place on the course of the 2012 Olympic Marathon on 30th May and the Great South Run (ten miles) on 30th October. Just raise a minimum level of sponsorship and you will

receive a PKD T-shirt and running vest, fundraising pack, PKD materials and ongoing support. You could also see your name and photo in a future edition of this newsletter.

If you are taking part in a running event this year, contact us for a fundraising pack, complete with a PKD running vest. Whether you are a novice, natural or expert, this is the perfect time to get running!

Contact us for more information about any of these runs: esther@pkdcharity.org.uk.

Christmas 2010 prize draw winners

- **First Prize - £500** - ticket number 05800 - from Aylesbury
- **Second Prize - £250** - ticket number 00851 - from Liverpool
- **Third Prize - £100** - ticket number 05926 - from Mirfield, Yorkshire

When shopping online don't forget to use the Easyfundraising shopping portal

It's free to use with over 2,000 high street retailers and top brands listed who give a donation to the PKD Charity on each purchase you make!

Just visit www.easyfundraising.org.uk/causes/pkd each time you shop.

A big thank you to our supporters

Once again our supporters have done amazing things to raise funds

As always we have been amazed by the amount of fundraising support we have received and here are just a few examples from the last six months:

Simon Greenwood trekked to the summit of Mount Kilimanjaro (see picture on front cover), raising £1,565. Linda Howell's social evening raised £1,660. Katie Leschorn did a Bungee Jump at O2 and raised over £400. Lindsey Bridgeman and several of her friends ran the Cardiff Half Marathon raising £1,162. Andrew Hartley's sponsored bike ride raised £880. Both Katie Peach and Stephanie Brumpton completed the Robin Hood Half Marathon raising £742 and £250 respectively. Kathryn Allen held a cake sale at work to raise £15. Jo Laing, Anna Siddle and David Anthony Junior all have collection boxes in their shops. Sue Searle and the Sir Robert McAlpine Company generated £1,770 from a fundraising evening. Zahra Sefiana ran the Royal Parks event in Oct 2010 and raised £655. Lisa Mantle and friends raised over £500 by completing the Lyke Wake walk (in North Yorkshire) in under 24 hours.

Katie Peach

Katie Leschorn

Thank you to everyone who has supported the PKD Charity by taking part in their own fundraising event or sponsoring someone else. To find out how we can support you with your fundraising, please contact our fundraising manager, Esther Wright. Email: esther@pkdcharity.org.uk.

Zahra Sefiana