WELCOME TO THE 4TH EDITION OF THE ALKAPTONURIA SOCIETY NEWSLETTER

A STORY OF HOPE
By Bob Gregory
Founder and Managing Director, AKU

Watch the wonderful DVD of Bob’s story which is very moving, but filled with hope. Log on to the website www.alkaptonuria.info and follow the link.
Share this with friends and family.

EUROPEAN RARE DISEASES CONFERENCE 2010

Dr Nick Sireau and Dr L Ranganath both attended the European Rare Diseases Conference 2010 in Krakow, Poland, see Nick’s report.

Latest News

PATIENTS EVENT
We will be holding another Patients Event 4th September 2010. Come and join us in Liverpool and have a great day out.

AKU PATIENTS
2010 has been another exciting year for us. Our AKU Patients database is still growing within in England and Worldwide.

AKU PATIENT’S EXPERIENCES
We have asked some of our patients to write an article of their experiences with AKU, you will also see that a patient from the USA has kindly sent us an article for our newsletter.

FACEBOOK
Serge Sireau of our French Group has now set up a Facebook Page

MEDIA UPDATE
Fox News, USA, have interviewed one of patients, Shane Blamries, see the article in this issue.
INTERNATIONAL NEWS

We now have an International News Section within the newsletter, as more and more AKU sufferers are now being identified throughout the world.

AKU Support Groups have now been set up in France, Italy and we are hoping to set up a North American and Canada group. This has been successful in networking with other AKU sufferers.

Also Serge Sireau, President of ALCAP (French AKU Society) has set up a Facebook Discussion Group, why not go and join the group and join in the discussions on www.facebook.com Why not download photos, make friends with other sufferers, discuss any issues you may have with each other, help each other to live life to the full with AKU, you need not feel isolated with this disease, someone is out there willing to help, so don’t delay join today.

The USA Support Group has been recently founded and a few of the members have met and are in regular contact. Below is a photo of Sabrina Drake and Denise Higgins from California.

Professor Annalisa Santucci, Italy, has set up an AKU Society – AIMAKU. To date there are about 11 patients identified and we have a doctor, Dr Enrico Selvi, who is also involved in the Society.
We have been asked to be involved in an American health programme, Mystery Diagnosis. This program tells the stories of patients experiencing rare and unusual diseases - and strange symptoms - whose medical cases have baffled doctors, and who’ve endured various twists and turns on their roads to diagnosis. Featuring cases of little-known diseases has proven to be an effective way to raise awareness and visibility for patients (including those not yet diagnosed), their caretakers, and the medical communities around them. A number of our American Patients have shown interest in being involved in this venture

If you are interested in getting involved you can contact: Leah Rubin-Cadrain, True Entertainment, 601 West 26th Street, Suite 1336, New York, NY 10001
(P) 212.763.3659 / (F) 212.763.3700

If you know of anyone within the media – newspapers, television, radio, magazines – local, regional, national - who would be interested in doing an article on AKU, please encourage them to contact the AKU Society at info.alkaptonuria@info

Fox News has recently interviewed one of our patients from America, Shane Blamines. His article makes very interesting reading.

As a result of this article interest in Alkaptonuria is growing, especially in the USA.
LIVING WITH AKU
BY SABRINA DRAKE, USA

I joined the AKU family in March 2010. I had the classic dark urine as a baby in 1971. A Dutch doctor thought it could be AKU, but it was never definite or put on my chart. My urine had cleared up when I was 7 years old, so my parents thought I was well. We moved to the U.S. in 1981 when I was 10 years old.

Around age 26, my right ear seemed to have a green-blue spot in it. Last year my left ear turned blue. I was in a RN program, and a very observant student noticed it. My sister had noticed the little grey stripes in my eyes the summer before, being a make-up artist paying attention to fine detail. My sister remembered my dark urine as a baby and adding the three clues, I pretty much knew I had AKU. My geneticist, Dr. Nenes, diagnosed me with AKU and Ehlers-Danlos Syndrome III (hyper-mobility), confirming my suspicions.

I am 39 years old, feel great, and want to keep the symptoms of AKU at bay as long as I can. I am also hoping for a cure for all of us. I play the lottery regularly! I take NTBC and Tyrex II along with the recommended low protein diet accompanied with taking NTBC. A little worried about long term side effects of the NTBC, but then I get to worry about AKU’s symptoms on a daily basis, so I am willing to gamble a bit.

AKU has completely changed my life style. I dropped out of the RN program. Nursing is hard on the body as well as the program itself, and my new goal is to stay out of the hospital, not work myself into it. I swim, ride a wonderful pony that we bought shortly after my diagnoses, hit the gym a lot to get on the stationary bike as well as the elliptical. I walk and hike with my Jack Russell terrier and appreciate each day I feel fantastic.

AKU has made me live life to the fullest each day and be optimistic about a cure. I can’t wait until this little nasty genetic disorder is eradicated! We are only missing one enzyme, but it causes such destruction. Just because it takes a long time for the symptoms to show does not mean it is not significant. Awareness, funding, and a cure (at least treatment!) are what it is all about.

Sabrina Drake USA

Calling All International Patients

Please let us have your stories and experiences of everyday living with AKU. Send your stories to:

Alkaptonuria Society
c/o Nick Sireau
109 Paget Road
Trumpington
Cambridge
CB2 9JG

E-mail to nick@sireau.net
SHANE BLAMIRES, USA PATIENT

Shane Blamires, a 38-year-old from Lewiston, Idaho, has black urine disease. It's a rare, genetic condition, and fewer people have heard of it than can pronounce its clinical name – Alkaptonuria.

On the other hand, its informal name, black urine disease, is self-explanatory. Just ask Shane Blamires. "It's kind of gross I guess, but ... it turned the urine color black. And that's the acid; it oxidizes in the air," he said.

Alkaptonuria (AKU) is a metabolic disorder in which patients can't fully digest an acid called tyrosine because of an enzyme deficiency they are born with. This causes 2,000 times the normal amount of homogentistic acid (HGA) to build up. HGA is then converted into a black pigment in a process known as ochronosis that can turn tissue, muscle, cartilage, bone – and yee, even urine – black.

Beyond the shock of one’s urine turning black, which only happens when it’s left to stand, patients commonly have bluish-black discoloration in the ear and the whites of the eye, and experience joint and back pain.

"This HGA damages the body and the main issue is early severe arthritis," said Dr. L. Ranganath, a leading expert on AKU based at the Royal Liverpool University Hospital in England, and a trustee of the AKU Society, a patient-led support network that raises awareness about the rare disease.

Shane, a member of the AKU Society, was diagnosed in 2006 shortly after having spinal fusion surgery, during which his surgeon was shocked to find the disc he was removing was black.

"It was really dramatic. I think it just totally astounded the surgeon," said Dr. Jeffrey Butler, a clinical rheumatologist at the University of Washington, who Shane was referred to after the operation.

Butler diagnosed him just months later, but it was Shane who introduced the idea that it could be AKU by researching his symptoms online.

"Shane's been a real go-getter. He's done a lot of the leg work and that's been a big help to me," said Butler, adding that Blamires is his only patient with the rare disease.

AKU is more common in certain countries, but occurs in one in a million U.S. births, according to the EMedicine website.

Consequently, many doctors have never seen it, so it is often misdiagnosed as arthritis.

"Every doctor I go to, I have to explain to them what it is; what I'm doing about it," said Blamires, adding that his doctors have largely taken his lead on how to treat his disease. "It's not the best way of doing medicine, but it's kind of the only thing that's out there."

Although the disease is genetic, most patients aren't diagnosed until their 30s or 40s when the majority of symptoms – joint pain, ligament and tendon ruptures, kidney and prostate stones, leaky heart valves leading to heart failure, and kidney damage and failure – begin to show.

"The major issue for all patients is the constant back and joint pains from around age 30 onwards affecting quality of life – work, family and leisure," Ranganath said. "Osteoarthritis-like joint damage is 100 percent certain for all patients."

Shane had his first joint replacement surgery of the knee in January 2009. But a little further east in Pesquen Isle, Wis., another AKU patient has had seven joint replacement surgeries before the age of 60, and one more last year at the age of 61, making it eight in total.

"It's a cartilage degeneration disease really," said Nick Williams, a retiree who shrunk three inches in his late 40s. "It's like a sponge in the sink; it's real good for a real long time, and then one day you pick up the sponge and it falls to bits in your hands. And that's really what happens to cartilage."

Nick has had each of his ankles, knees and hips replaced – his left hip was replaced twice because the plastic failed in the first one – in addition to his right shoulder, all in the span of 14 years.

Unlike Shane, Nick was diagnosed when he was a baby.
“In those days, they didn’t have throwaway diapers, and when my parents first changed and washed my diaper, it ended up having a dark stain that does not come out, even with bleach,” Nick said.

His parents were worried so they brought him to the doctor. At the time, doctors didn’t know AKU was a metabolic imbalance and that the HGA in his urine is what caused the deep stain. But they were able to provide a roughly accurate diagnosis because they knew of the research done on it as early as the 1890s, which led to it becoming the first disease ever to be identified as inherited.

But if this early warning sign is missed, the disease isn’t noticeable in the years in between infancy and middle-age. Fast forward 40 years, and Nick found himself slowly losing mobility. It started with difficulty climbing stairs. Then, the disease began to spiral out of control.

At 41, he had four kidney stones in five months. Then he ruptured his right and left Achilles tendons at 42 and 44 respectively.

Joint replacement and pain medication are the only proven treatments for the floodgate of symptoms that hit many AKU patients in middle-age.

“The thing is you really don’t treat Alkaptonuria like it’s Alkaptonuria, you treat the symptoms that manifest themselves,” Nick said. “You do a joint replacement, you do a valve replacement; you do whatever is necessary, as if it were some other disease because it manifests itself like a joint failure.”

Clinical trials are being done to find a treatment that addresses the disease. The National Institutes of Health recently concluded a trial of an enzyme blocker called nitisinone to neutralize the effects of HGA. Neither Shane nor Nick were included in the trial of 58 people with AKU, despite applying. But Shane said he has been taking the same enzyme blocker used in the NIH study for four years.

Although the drug containing nitisinone was effective in blocking the excessive amounts of HGA in both the trial and Shane, it would never be able to repair the damage already done.

“Once the integrity of the joint has been violated, you get this progressive problem where there’s arthritis that we can’t really reverse,” Butler said. “Even if you’re able to block the process, the arthritis does progress.”

For Shane, the hope is that the nitisinone drug will stop the disease’s progression before it begins to affect his heart.

Although AKU isn’t fatal, it can have a drastic effect on quality of life.

“I can still golf, which is fine by me. But a lot of other things like just doing basic yard work or things like that where there is a lot of lifting or bending, I just can’t do it anymore. And a lot of times – I have two younger girls – I can’t even pick them up,” Shane said.

Another factor AKU patients have to deal with is the isolation felt because there is no one who knows exactly what you’re going through.

“I felt all alone when I first had it,” Shane said. “I remember my wife just crying for nothing; nobody to talk to, nobody knew anything about it.”

That’s where organizations like the AKU Society come in.

“It was set up to raise awareness about AKU, help patients, and fund research to find a cure,” said Nick Sireau, who has two children with the disease and helped found the AKU Society in 2003, along with Ranganath and others.

Even though Williams doesn’t hold any illusions that a miracle drug will restore his youth and allow him to do cartwheels down the driveway anytime soon, he is hopeful that medicine will catch up with the world’s first-known genetic disease.

Both Ranganath and Butler share Williams’ hope, and want to see further clinical trials. But in order to prevent the effect of early osteoarthritis, Butler stresses the need to begin treating it early.

“The hope would be that if you could initiate this at a very young age before any damage or any deposition of the HGA has happened; that you might be able to prevent the arthritis from developing,” said Butler, adding that gene therapy may also have potential as a future treatment.
My name is Simon Laxon I am 44 years old and married with 2 daughters. I was diagnosed with AKU when I was a couple of weeks old. Originally the Drs told my parents that AKU was a harmless genetic disorder that caused no problems in younger life but would cause the cartilage in my ears and nose to turn a blue colour, as I got older.

As a youngster I was physically active in sport. I played Rugby and threw the Javelin for my school and hometown. As I grew older I played squash, weight trained and did martial arts achieving a Black belt 1st Degree in Kung Fu.

I left school at 17 and worked in the car industry, doing heavy lifting and climbing in and out of engine bays, until 1995. I then worked for a company that made and fitted hydraulic tubes and fittings. Again I was lifting and carrying heavy weights, until 1997 when I collapsed one day at work with severe pain in my lower back.

I spent the next 4 years trying to get my Drs to understand and treat me for the problems caused by AKU. It wasn’t until I travelled to the NIH, to take part in clinical trials of Nitisinone, that my Drs eventually realised that AKU was the root of all my problems. Nowadays all of my Drs are fully aware of AKU and treat my symptoms accordingly.

Hopefully, in my lifetime, there will be a cure found for this destructive disorder so that people won’t have to go through what we have. I personally am willing to help out in any way I can, whether it be spreading the word or taking part in clinical trials. If by my partaking in these trials helps just one person in the future then what we have done is not in vein.

Simon Laxon

1. When did you first become aware of symptoms?

I started to get lower back pain around the age of 21 but it wasn’t until 1997 that I realised something was not right. The back pain became much more frequent and severe.

2. How long did you have to wait for a diagnosis?

I was originally diagnosed in 1966, when I was a couple of weeks old. At the time the Drs didn’t know much about Alkaptonuria and told my parents that it was a harmless genetic disorder. I was then re-diagnosed in the late 80’s, whilst under investigation for a Duodenal Ulcer. I was told that my Hospitals records had been destroyed in a fire at the hospital in the late 70’s, and as a result no information existed before then. This time I was also told that all about the destructive nature of the Arthritis and what lay ahead for me.

3. What were your first thoughts on learning of this diagnosis?

At the time, it didn’t have much impact on me as I was young and virtually symptom free. I was doing martial arts and weight training, as well as other physical sports, so felt fit and healthy. It wasn’t until 1997 that I realised how this disorder was going to basically tear my world apart and destroy my life. I lost my job, through back pain, and became depressed.
4. Have your feelings / experience of the condition changed over time?

Definitely - At first I felt that I was alone and that nobody understood what I was going through. As time went on I found that the Drs were becoming more understanding and through the help of the NIH and the AKU Society they were able to understand the clinical nature of Alkaptonuria and how best to treat the symptoms, of the many other disorders I would get as a by product of having this.

5. What is the biggest problem for you about living with this condition?

I suppose the biggest problem for me, personally, has been not being able to lead a “normal” life. I haven’t been able to support my family financially, which has meant the burden is now on my wife’s shoulders. We struggle to make ends meet and don’t have any social life because of this.

As a father, I haven’t been able to play outdoor games with my children and felt for my children on school sports day as they knew I couldn’t take part in any of the “dads” races.

At 44 years old, on the outside I look fairly healthy but on the inside I feel like I am 100 years old. I feel that the cruel hand of fate took these things away from me.

6. What helps you the most?

Knowing that I have the support of family and friends and knowing that there are organisations, like the NIH and the AKU Society, who are willing to listen and do something about trying the help the AKU community.

7. How does it affect family members and friends?

I know that it affects each person in a different way. Close family have to put up with a lot of stress both physically and mentally. I know from personal experience that it will push relationships to breaking point. Parents will feel guilt at having given this destructive disorder to their children. Friends will find it hard to understand why you can’t do tasks anymore that they may find easy to do. You may find that some friends can’t cope with this and turn their back on you, whereas other friends will stand by you whatever happens.

8. How did you find out about the Alkaptonuria society?

I became friends with Robert Gregory over 12 years ago, through Alkaptonuria message board sites, and knew that he had planned to set up a website for people with AKU. He and a few other concerned people decided to set up the Alkaptonuria Society whose original aim was to provide people with Information on the disorder.

SIMON LAXON
WEST MIDLANDS

IF YOU WOULD LIKE TO HAVE AN ARTICLE PRINTED IN OUR NEXT NEWSLETTER PLEASE CONTACT US.

Send an e-mail to info@alkaptonuria.info
My name is Ann Kerrigan and I am 51 years old and was diagnosed with Alkaptonuria late December 2009. For me, the diagnosis was a relief because it had taken over four years to reach that point. So I have not been living with Alkaptonuria for long but to put things into perspective here is my journey to diagnosis and where I am now.

Initially, my problems started in my early thirties with severe pain in my lower back. I went to a chiropractor who x-rayed and diagnosed osteo-arthritis and indicated this was very unusual for my age. I followed this up with a second opinion from my doctor and it was confirmed but without any attempt to determine the cause. However, life continued as normal with the use of painkillers and no heavy lifting until 2005 when my knees became extremely swollen and painful after a night out bowling. Coincidentally my eyes started to show pigmentation around this time although my ears changed colour around 2003. I had mentioned this to my doctor but probably not at the same time as we were dealing with my joints so the connection was never made.

In May 2007 I had a left knee arthroscopy and was informed afterwards that my knee was covered in dried black blood and I was sent to a Haematologist to investigate blood disorders. However, I did not have any blood disorders and a total knee replacement followed in October 2007. After the replacement the surgeon diagnosed Pigmented Villonodular Synovitis (PVNS), which is a rare disease that affects the knees or hips. At this point I was relieved to have a diagnosis and concentrated on physiotherapy and exercise to regain mobility. Unfortunately, my knee was still incredibly painful one year on and I underwent a revision in February 2009. Around this time my shoulders also became painful and there was no real improvement in my left knee so I returned to my doctor to find new answers and this led to my correct diagnosis last December.

This has been an interesting year so far and also informative since I joined the AKU society and spent three days in Liverpool in February as part of the evaluation study. For me this was a very positive experience and very reassuring to meet people who understood my disease and removed my feeling of isolation. As a result of the study a lesion was identified on my chest and with further investigation it turned out to be a large mass under my breastbone, which needed to be removed. Therefore, in June I had a median sternotomy, which went very well and thankfully the mass was benign and turned out to be related to Alkaptonuria. Since then I have made an excellent recovery and at the end of August I am having a right knee arthroscopy with my new Orthopaedic Surgeon in Oxford. I have to say it's been a difficult year but at this point I am very optimistic because I finally know what I'm dealing with and I have the right consultants looking after me. So now my hope is to regain my mobility and resume my career.

ANN KERRIGAN
BRISTOL
I'm someone like you, living with AKU. So far AKU has not been to bad to me, in fact since I have been in my wheelchair (outdoors mainly) I've met some lovely people and have done an Disability Access course and can now use my wheelchair experience to help other wheelchair users.

My joint degeneration has slowed considerably since I have been using my wheelchair and pain is at minimum. I am not suggesting everyone gets a wheelchair, only pointing out the benefits I have gained from it.

I have had 3 replacements 1 hip and 2 shoulders. AKU had affected most of my moving joints and my lower back has fused together and is in a bit of a curve. My ears have a constant ringing in them which is getting worse but I have got used to it. My hearing is only affected a little bit so I am lucky. I have been told I have some kidney stones so I suppose I will have to get them zapped at some stage.

So far my heart is ok, but I know it is probably only a matter of time before it starts to give me trouble. I am aware that as a female I have this disease strongly than men. I wonder why this is.

I have been a participant in the NIH trials in America and may yet join the trials in Liverpool, but at the moment my energy levels are quite bad. I am recovering from a kind of ME condition. I have found that forgetting about AKU as much as I can is the way forward for me. And my life is busy with 4 children, 2 grand children and some voluntary work. I try to think positively but that can be hard at times. I am a spiritual person and that helps me a lot. Warm Wishes to you all.

LIZ LINNANE
SOUTHERN IRELAND
I am Gary Houlden, 43 years old, from Bromley in Kent. I was diagnosed as having Alkaptonuria as a baby, but my parents were lead to believe that this was a childhood illness which I would grow out of. A few years ago at work I had an accident and injured my shoulder. When I went to my doctor about this, he asked how I was coping with my "condition". It took a moment to realise that he meant Alkaptonuria. I decided to find out more about it, finding the Alkaptonuria Association on the Internet. Since then, I have attended the 2008 patient’s event at Liverpool and been to Liverpool Hospital for the research project.

I work in a school as a Technology technician/teaching assistant. This does involve cutting up materials and moving machinery and equipment, but I use common sense and generally know my limits. The school have been very supportive since we have found out more about AKU, if I need help moving anything the site management team always help out. When I was asked to come to Liverpool for the clinical evaluation the school did not hesitate to give me the time off to attend.

On a day to day basis, I am not really affected by AKU, I get achy some mornings, but that passes. I have had a few days off from work as a result but this is rare. I have found that having a hot bath in the morning eases the aching. I am not on prescribed pain killers, but occasionally take Neurofen.
I was diagnosed with Alkaptonuria (AKU) when I was still a babe in arms in 1962. At the time my Parents were told that AKU was a genetic condition ‘At the other end of the spectrum to Albinoism’ (whatever that means) and that in later life I should have a ‘sedentary lifestyle’. At the time a lot of my family worked in the Coal Mines, not the best place to start to achieve a ‘sedentary lifestyle’. Fortunately I had enough academic capability to go to University to study Chemistry and went on to receive my Doctorate in 1992. For the past 12 years I have been working in the Pharmaceutical industry and have been married to my wonderful wife Cindy for nearly 18 years and we have 3 teenage children.

Like most AKU sufferers, I started having aches and pains in my back when I was in my mid 30’s and I put this down to me getting old. Eventually the pain got to such a level that I saw my GP and he prescribed some painkillers. When they didn’t work, he prescribed further painkillers and a course of physiotherapy. It was at this stage that I mentioned that I had AKU and could the pain be due to that. Unfortunately, when I went to University my childhood medical notes had been lost so he had none of my history. He wrote to a Consultant Geneticist to enquire about AKU and the reply was straight out of a Biochemistry textbook, word for word, implying that AKU was a novelty but nothing to worry about. I struggled through for a couple of more years.

In 2000 the pain had increased to such a level that I needed to see a GP again. By this stage I had moved jobs, moved house and, more importantly, moved GP’s. Also I had done some research into AKU myself, trawling through the medical journals for the limited information available, and realised that my pain was probably due to AKU after all. Fortunately, Cindy had returned to work as a District Nurse, after having a career break. Her knowledge of the GP’s in the Surgery meant that we were able to approach the one that would be least likely to dismiss the AKU scenario. In fact, when we saw the GP and told him what we knew, he freely admitted that he had not even heard of AKU, but crucially acknowledged that between Cindy and myself we knew what we were talking about. The extra stroke of good fortune is that, of all the Consultants he could have referred me to, he referred me to Prof Cox at Addenbrooke’s Hospital in Cambridge, who immediately took an active interest in my case and has looked after my health needs ever since.

Cutting a long story short, between 2000 and 2007 my health got increasingly worse. I had had several bouts of severe pain during this period. I attended a 3-week Back Pain management course in 2004, run by the Occupational Therapy Team at Addenbrooke’s and sponsored by the Rheumatology department. Fortunately I was in a job I loved which required me to sit at a desk most of the
time. It did involve quite a bit of travelling, especially to the Research sites in Belgium and Slough. I had spent increasing amounts of time working from home when the pain was too bad, even though I was already on morphine. By the summer of 2007 I was receiving Higher Rate Mobility Disability Living Allowance and was using a wheelchair to get around more and more. In 2008 I formally started working from home 4 days a week, with the other day travelling.

Things came to a head in December 2008. I was on one of my frequent trips to Belgium and fell in the shower, banging my back on the side of the bath. Typical me, I struggled on with the pain and managed to get back to the UK. Things were that bad the following morning that Cindy took me to A&E to get me checked out. Although no bones had been broken, I was signed off work for several weeks. As the accident happened on work business, a referral to the Occupational Health Team (Occupational Health) was automatically triggered and a meeting was set up for February 2009. To complicate matters, by the time of the meeting I was already booked in to have my left knee replaced. The Occupational Health Doctor took one look at my notes, at my condition and the fact I was going to have a knee replacement and recommended that I should be signed off for the month leading up to the operation, to get myself prepared. He also suggested that I should think long and hard about whether I should continue to work. At the time I thought this was a bit drastic.

The Knee Replacement went well, the tissue being donated to the AKU Research Team in Liverpool. I spent 3 months recuperating and a further 3 months easing myself back into work, with no travelling. I finally went back full-time in September. I lasted 6 weeks! By the end of October I was getting tired very quickly and was unable to do a full weeks work, even though I was just sitting at a desk. I’d had several episodes of being off sick and it all came to a head when, at the November review with Occupational Health, they recommended that I was not fit for work and arranged for a taxi to take me home. My GP took one look at me and signed me off for 2 months initially. I’ve not been back since. I had a final assessment with Occupational Health in Feb 2010, and the recommendation was that I was not fit for work and should be disabled out. The key observation of the Doctor was that:

“... I have a box of tissues on my desk because I normally have to tell people that they are fit for work, when they don’t want too. You are very unusual, you obviously want to continue working, but are in no fit state to do so.”

Like most people my thoughts went immediately to how to support my family. I consider myself very fortunate as my employers pay into a Permanent Health Insurance scheme, set up specifically for those that cannot continue working through injury, illness or disability. The scheme pays a percentage of your salary until you are either fit to return to work or reach state pension age. This coupled with the state benefits I am entitled too ensures that, although things are very tight, we still have a roof over our head and food on the table.

What aren’t well cared for are the mental and emotional challenges I have had to undergo throughout this period. I have always been an outgoing person, enjoying camping, hiking and all things relating to the ‘Great Outdoors’. Also I have always had a keen interest in helping out in the community. Over the past 5 years I have been the Chairman of the Annual Town Show, a Chair of Governors for a local school, a Cub Scout/Group Scout Leader and a member of the local Rotary Club. I’ve had to give all this up. At times my heart tries to persuade me that I can still do anything, this is usually on a good day. Normally my head has to overrule my heart. As
I cannot even dress myself fully, how am I supposed to go traipsing across the moors, peg out my tent? Fortunately I am usually a ‘Glass half full’ sort of person and with the full support of my family have evaluated what is important in my/our life and what is a nice to have. What I can actually do/not do and crucially, is there a way to adapt things to my needs? For example, when I could no longer manage in a tent, we bought a caravan, enabling the family to still enjoy the great outdoors. Where possible I use an electric scooter. The only issue is that suddenly I become a moving clothes-horse and the dog wants to climb aboard when he’s feeling lazy.

I always look back to the Pain Management Clinic I attended at Addenbrookes in 2004. The key take home message from this course was to manage your pain not let the pain manage you. To achieve this you need to set yourself realistic goals/challenges on a day-to-day basis for two key reasons. Firstly, to avoid the ‘Boom and Bust’ cycle, do not try and do too much on a Good day as more often than not the next day will be a Bad day where you feel you cannot do anything and secondly, it is human nature to feel good when you have achieved a task.

One chapter of my life has just come to a natural end and more importantly, a new chapter is just about to start. Where will it take me? Who knows, watch this space.

DUNCAN & CINDY BATTY
SUFFOLK
I have lived in Hampshire for over 30 years, with my wife, having moved from Hertfordshire. Our eldest daughter, husband and their young son live in Northamptonshire and our younger married daughter lives in Somerset. After being treated for Hodkinsons Cancer twice, we were delighted that she is expecting our second grandchild at Christmas.

I was diagnosed with AKU when I was in my “teens” and for many years attended a metabolic clinic at a London hospital. My main contact with the NHS now is through my GP and the orthopaedic department at my local hospital in Portsmouth. My back and neck are completely “seized” and I have had nine major joints replaced. (Both ankles, knees, hips, elbows and a shoulder), and this year I had a partial right foot fusion carried out. Plans are in place to have my other shoulder replaced followed at some stage by a total fusion of my left foot. I am the “Bionic Man”!

I was a Baker/Pastry Cook by trade and have not been in paid work for many years and have been registered disabled. I enjoy reading, TV/Radio and tending our small garden.

I am an Assistant County Commissioner in the Scouts and a member of the Education Appeals Tribunal Service. I am a Magistrate and chair adult and youth courts, a role I very much enjoy. My wife is a member of our local church and is the leader of the “Mothers Union” branch.

Of course AKU has had a great affect on mine and my family’s life. My mobility is restricted which at times make things very difficult. I am not a person that enjoys “Group Therapy” but the AKU Society has helped me and those people I come in contact with, particularly the NHS, DWP and friends understand Alkaptonuria more.......
NEWS UPDATE

THE AKU SOCIETY HAS A FACEBOOK GROUP

The AKU Society has set up a Facebook group dedicated to providing support and information for Alkaptonuria patients.

To access the group, log into Facebook and type Alkaptonuria into the search box.

![Facebook](https://via.placeholder.com/150)

This is a great way to keep in touch with AKU patients, their families and friends from all over the world. Sharing each others’ stories, and become friends through the forum. Reduce your sense of isolation speak to other sufferers. Get your friends and family to join as well and get them involved in discussions. Even let your G.P. know that we have a Facebook Group, encourage them to join as well. Why not go to Facebook now and join the group, download photos, and chat. We are waiting to hear from you. To date we have 84 members on the Facebook site.
The next Patient’s event is to be held at the Liner Hotel, Liverpool

Further information is available on the AKU website: [www.alkaptonuria.info](http://www.alkaptonuria.info)

At the event patients are able to meet other AKU sufferers, the Medical Team and the AKU Trustees, ask questions and get them answered. They are able to learn more about the research being carried out at Liverpool University by Professor Gallagher and his Team.

It is a nice, warm relaxing day in a friendly environment. Family and Friends are all welcome as well.

Several of our patients will be talking to the group about their experiences of AKU.

There will be four workshops throughout the day, which include:

- Clinical Evaluation
- Patient Support
- AKU Research
- Disability Awareness

The Conference will start at 10 am and will conclude at 5 pm. If you are still interested in attending please contact Bev Hebden by Tuesday 31st August 2010 on 0151 706 4387 or bev@alkaptonuria.info

Unfortunately due to funding we are unable to assist with payment for travel or hotel accommodation.
The Department of Clinical Biochemistry and Metabolic Medicine at the Royal Liverpool University Hospital in collaboration with the AKU Society of the UK have conducted a clinical evaluation study on AKU individuals. This study involved bringing people with AKU (22 in all when we are finished; 15 completed so far) to Liverpool from around UK for a fully comprehensive clinical and investigational examination. The clinical study included history taking, physical examination, as well as specialist evaluation by ENT, Ophthalmology, Radiology, Nuclear Medicine, Physiotherapy, Rheumatology, Dietetics and Metabolic Medicine consultants. This study has for the first time allowed us a better understanding of how healthy people with AKU are in the UK. All patients also filled in a series of questionnaires that are helping us to understand how AKU develops. This will not only help us understand the condition but also help to plan better treatment and when to start such treatment.

The results from the study were given to patients on the CD for their record. The group results will be presented and discussed at a patient workshop in Liverpool on 4th September 2010.

All the patients who came to Liverpool for the clinical evaluation met up with all specialists including myself. Patients were able to discuss their health issues in person. As a result of the evaluation one person was referred for urgent further heart tests and treatment. Another patient was found to have a tumour in chest that required open chest surgery. Patients have also gone away understanding more about the treatment options for AKU that are now available and what we hope to have in place in the future both because of these studies and others we will conduct in the future.

The subjects for the study were recruited from those identified by the National Lottery funded identification project. In addition to establishing a register for people with AKU in the UK, we have also become aware of how healthy these individuals are. The clinical evaluation study has only been made possible because of the National Lottery funding support.
REPORT BY NICK SIREAU
CHAIRMAN

We've just got back from attending the European Rare Diseases Conference in Krakow, Poland, from 13-15 May. It was fantastic and extremely useful (For a full programme, go to the Eurordis website: http://www.eurordis.org/)

Below I highlight the key connections we have made. I will be following up many of them over the coming months.

Key points that came out of the conference for AKU:

- The European rare diseases community is developing fast, thanks to pressure from patient groups and emerging policies at EU and national levels. Unfortunately, the UK is lagging behind somewhat, although pressure is building there.

- Many patient groups have pioneered promising avenues of funding and research that we need to learn from (take a look at www.behcets.org.uk)

- There is strong interest from the pharma industry for research into rare diseases and we need to build connections there.

- Building a network of research teams across the UK and the EU is crucial if we want to develop the multi-disciplinary approach and find the patient numbers for effective trials.

- There is potential for research funding from the EC and the MRC, although it will take time and lots of lobbying and proposal writing.

Overall, we came away feeling very positive about prospects for AKU so long as we put the time, effort and resources into building our network and reputation and scaling up our research. Opportunities for connections and funding are definitely available.

Poster Presented at Conference

Identifying Alkaptonuria (AKU) in the Population at Large: Challenges and Solutions

L. Ranganath1, B. Hebden1, J. Hignett2, J.A. Gallagher3 and N. Sireau4

1Department of Clinical Biochemistry and Metabolic Medicine, 2Obesity Biology Unit. 3Obesity Biology Unit. 4Department of Human Anatomy and Cell Biology, University of Liverpool
Sadly this is the last newsletter that the Alkaptonuria Information Centre will be producing due to the closure of the Information Centre based with the Royal Liverpool Hospital.

Bev and Jan have enjoyed their time at the Alkaptonuria Society and will miss speaking to the patients and their families. They both also enjoyed meeting patients at the various events that have been held over the last three years, especially when patients came into the Royal Liverpool Hospital for the Clinical Evaluation with Dr Ranganath and his team.

Bev and Jan would like to thank all our patients for their co-operation over the last three years in providing information to us, which has helped us greatly in research. They have also both been attending Medical Exhibitions promoting the Alkaptonuria Society, and in some cases educating the Medical Profession on the signs of AKU. The work that both Bev and Jan have done has gone a long way to the success that we are today. See some of their achievements below:

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**Raising awareness with the Medical professionals**

- **We have reached 46,500 GPs throughout the UK in the GP mail out**
- **We have identified an additional 24 patients via the GP mail out**
- **Attending Medical Conferences, namely**
  - **GP Conference**
  - **British Orthopaedic Conference**
  - **Rheumatology Conference**
Reducing the isolation factor

- This project has had a huge impact on reducing the isolation factor
- All AKU sufferers know about the AKU Society and have a point of contact
- We have identified 500+ patients worldwide and have been in regular contact with many
- We have met AKU patients during the clinical evaluation
- We have spoken to patients via telephone interviews
- We have met patients who attended the AKU Patient Conferences in 2007, 2008 and 2010
- Family members and carers have a better understanding of Alkaptonuria
- Producing a Newsletter on a regular basis
- Updating the AKU website
- Setting up a Facebook group
A BIG THANK YOU TO AKU PATIENTS FOR:

- Participating in the clinical evaluation programme
- Donating their tissue from operations for AKU research
- Participating in telephone interviews
- Completing numerous Questionnaires
- Fundraising

ALKAPTONURIA INFORMATION CENTRE

WEB SITE: www.alkaptonuria.info

BEV HEBDEN – PROJECT MANAGER
bev@alkaptonuria.info

JANET HIGNETT – ADMIN ASSISTANT
janet@alkaptonuria.info

Please note that as from 24th September 2010 the above Information Centre will be closed please do not e-mail any information to Bev and Jan after that date. All queries should be e-mailed to

info@alkaptonuria.info