

touchinglives



the action medical research magazine • autumn 2003

Sickle cell – the unpredictable disease

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Plus:

4 new fellowships awarded! • London to Paris Bike Ride success • A day in the life
– Hurler syndrome • Helping children with the world's deadliest toxin

From the Editor

New name, new logo, new image. The last three months have been a particularly busy time here at Action Medical Research. But what an exciting time to be involved with Action Medical Research! We're transforming the way we go about communicating who we are to the world. We've got a story to tell, and it's vital that the largest possible number of people get to know us and the vital medical research we make possible.

I hope you'll agree that Touching Lives looks great! And inside you'll find the same great articles, outlining the valuable work done by Action Medical Researchers across the country. And watch out for some new features from guest writers.

We've also taken the opportunity, as you would expect, to refresh our website. Log on to www.action.org.uk (note our new web address) to see the new design for yourself.

All that remains for me to do is to welcome you to the Autumn 2003 edition of Touching Lives. I hope you enjoy it.

Best wishes

Daniel Sartin
Editor

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Action Medical Research is one of the country's leading medical research charities. For over fifty years we have been funding pioneering research into a wide range of illnesses and conditions.

By pushing back the boundaries of science and enabling medical breakthroughs such as the polio vaccine, ultrasound scanning and the hip replacement operation, we play a crucial role in fighting disease and disability and improving the health and quality of life of everyone in the UK.



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Spotlight falls on name change

The Action Medical Research spotlight fell on The Royal College of Surgeons in Lincoln's Inn, London, on September 16th for the official media announcement of the Charity's new name.

Ninety guests including VIPs, health journalists, Trustees, volunteers, celebrity supporters and researchers attended the reception. The Charity's President Lord Guthrie made the announcement of the new name explaining that although our name is changing, the Charity's remit to fund innovative, life-enhancing medical research remains the same.

Head of Communications, Andrew Proctor said: "The focus of Action Medical Research is very

much on the future with a great deal of enthusiasm to move forward and build on our great track record. Our new name and revitalised logo will help build awareness of the Charity's work and, in turn, this will help our fundraising."

After the announcement, the Harding Award for an outstanding contribution to medical research was presented to Professor David Delpy, Vice Provost of University College London. Making the presentation, Neil McIntosh, Professor of Child



Charity staff celebrate the name change

Life and Health at the University of Edinburgh highlighted Professor Delpy's valuable research into physiological monitoring. In particular, he has worked on the development of non-invasive techniques for monitoring the chemical processes and functioning of the brain in newborn infants.

Successful results from stroke project

Severe constipation and bowel leakage are common and distressing problems faced by people who have had a stroke. Such bowel problems dramatically impact on the quality of life of both patients and carers and are a major factor in referral for long-term care in nursing homes.

Thanks to funding from Action Medical Research, the first rigorous trial has taken place to compare a structured programme of assessing and treating bowel problems in stroke patients with routine care. The £67,000 study was based at St Thomas' Hospital in London in conjunction with King's College Hospital, London and Orpington Hospital in Kent, and was headed by Dr Danielle Harari. Dr Harari said: "We predict that our research will change practice both in the way bowel problems are managed in people with stroke,



Dr Danielle Harari (left) and Linda Lockwood (research nurse on the project) with the booklet

and in the way we provide health education and promote self-management in older people with long-term disabilities. Action Medical Research is to be thanked for having the foresight to support this type of research, where the aim is to find strong scientific support for good clinical practice in vulnerable patients."

The treatment programme consisted of a single meeting with a nurse who assessed the patient clinically, recommended bowel medicine and pelvic muscle strengthening treatment where needed, and educated the patient and carer regarding lifestyle habits that relate to the bowel (diet, fluid intake, exercise, toilet habits) with provision of a booklet.

The results of the trial showed that even up to six months later, these patients clearly benefited in terms of bowel habit being restored to 'normal' in comparison with a similar group of patients receiving usual care only. Treatment patients were much more likely to alter their diet and fluid intake to control their bowel problem even up to 12 months after receiving the educational booklet.

Free copies of the booklet are available from Action Medical Research.

Folic acid may combat cleft lip and palate

Action Medical Researchers have found that pregnant women who take folic acid supplements may be doubling the health benefits for their unborn babies. The B vitamin could help lower the risk of infants being born with cleft lip and

palate, as well as reduce the chances of them developing spina bifida.

Action Medical Researcher Professor Robin Winter and colleagues at the Institute of Child Health, London, have identified changes in a

particular gene which may increase the risk of children suffering a cleft lip and palate. The gene, known as MTHFR, plays a vital role in regulating the body's supply of folic acid.

Read more at www.action.org.uk/folic

All the latest medical and fundraising news from Action Medical Research

Pre-eclampsia makes headlines

Recently, the BBC reported the results of a study which indicated that taking the anti-oxidant vitamins C and E early in pregnancy halved the rates of pre-eclampsia in women with a high risk of developing the condition.

This is exciting news, not least because pre-eclampsia is such a common complication of pregnancy, affecting one in 10 pregnant women at least mildly. For one in every 50 pregnant women it causes serious complications. The

devastating condition causes a pregnant woman's blood pressure to rise to dangerously high levels and can prove fatal to both mother and baby. The cause is unknown, there is no effective treatment, and in severe cases doctors have no choice but to deliver the baby early.

Action Medical Research is funding a three year study at Oxford's John Radcliffe Hospital, headed by Professor Christopher Redman, to examine whether shedding of tissue from the placenta into the mother's circulation could lead

to pre-eclampsia. They are investigating whether a shortage of oxygen in the placenta causes increased shedding of debris and if treatments (including anti-oxidant vitamins) can protect the placenta when oxygen supplies run low.

The work of Professor Redman and his team is laboratory-based, using placental tissue taken from consenting mothers. The project, due for completion in early 2005, should further our understanding of the causes of this devastating condition and may lead to new treatments.



Action 100's sizzling success

Over 300 cyclists braved the hot weather on August bank holiday Sunday to complete the challenge of the Action 100 Bike Ride, hoping to beat last year's sponsorship total of £32,000. Cycling a demanding 100 miles in one day, the riders left Bristol and Bath from 6am and followed the scenic A4 route to finish at Chobham in Surrey.

Amongst the most adventurous riders were a Bristol postman cycling in uniform on his work bike, an amputee who rode an extra nine miles to get to the start, four tandems and one participant cycling in flip-flops!

After the event Jennifer Reid, Community Fundraising Manager for the area, said: "The turnout was excellent and we are really grateful to the cyclists and those who volunteered and worked tirelessly throughout the day."

Future rosy for Touching Lives

Action Medical Research is to get its very own rose, to be called 'Touching Lives'. For every plant sold the Charity will receive a generous donation of £2.

The rose came about through supporter Josie Maudlin who is a neighbour of specialist rose growers Chessums. The family firm grows 5 million roses over 220 acres.

Fiona Oomes, Community Fundraising Manager for the East of England, helped in the choosing of the rose, "The decision was unanimous. The bloom has a lovely scent and

is gold with a hint of pink on the ends of the petals. In fact, it is absolutely stunning!"

"The rose is a sturdy bush rose of moss type – when open it has lots of small petals in the middle. It is also disease resistant, repeat flowering and keeps its colour."

Plans are afoot to launch the rose at the Hampton Court Flower Show in July 2004. Then the rose will be distributed through retail outlets across the country as well as through a reader offer in its eponymous publication Touching Lives.

Website redesign launched

Log on to www.action.org.uk

To coincide with the announcement of the Charity's new name, on September 16th Action Medical Research launched its redesigned website. The new design has brought the Charity bang up to date, and with a design that the site's Editor Daniel Sartin believes will stand the test of time. "Our new design is fresh, simple and highly attractive. I believe that it compares very well against other sites in the charity sector, and will be a pleasure for the Charity's supporters, and people who want to find out more about us, to use," he told Touching Lives. "We have a new, shorter web address, and we've also taken



the time to make some improvements to our online sponsorship, media and research sections." It's easier than ever to donate online, and the Charity has maintained its rigorous approach to site security and confidentiality. If you'd like to give your feedback on the changes, please email webmaster@action.org.uk

New email newsletters launched

If you'd like to receive a new, quarterly email newsletter from Action Medical Research, log on to www.action.org.uk and submit your email address. We aim to keep you up to date with the very latest news from the Charity between issues of Touching Lives.

The unpredictable disease

An Action Medical Research project has been looking at the biggest cause of stroke in children: sickle cell disease (SCD). Our medical press officer, Louise Brown investigates for Touching Lives.

SCD is called the unpredictable disease because you never know when the sufferer will have what is termed a 'sickle cell crisis'. Blood cells are not round but crescent shaped in people who have SCD and this causes problems with circulation leading to blocked blood vessels and damaged organs. This is what causes the crisis of acute pain, centred particularly in the chest and back. Such crises occur in most patients with SCD at some point in their lives, and can last from a few hours to a few days. Some people only have one crisis every few years while others may have many and require hospitalisation to receive pain relief.

Repeated crises can cause damage to the kidneys, lungs, bones, eyes and the central nervous system. Symptoms of SCD include joint pain, fatigue, rapid heart rate, delayed growth and puberty, susceptibility to infections, ulcers, jaundice, and bone and abdominal pain.

This terrible disease affects an estimated 10,000 people in the UK.

Brain injury

On an Action Medical Research project, Professor Faraneh Vargha-Khadem and colleagues from the Institute of Child Health in London have been following a group of around 100 children and babies with SCD to track their development and monitor the problems they

experience. The children are being compared to those from similar backgrounds who do not have SCD.

Professor Vargha-Khadem told Touching Lives: "The Action Medical Research grant helped us follow the children from the original group as they grew up and allowed us to monitor a number of babies throughout the first year of their life. What we found was that some of the children had developed specific learning problems, and that different techniques could be used even with babies to monitor changes in mental activity in association with SCD.

"The goal of our research was to use a variety of techniques to identify those children with SCD who are at high risk of stroke. For example, by examining the velocity of blood flow, it is possible to identify those children who are showing abnormal levels of blood supply to the brain. Such techniques allow the identification of brain regions that are vulnerable to imbalances in blood supply. We can then treat patients at risk by medication, or in some cases surgical intervention. Additionally, by examining those who have already suffered stroke, we get an idea of the range of problems that can develop and relate these to the location and extent of the brain injury. At present, there is no cure for SCD and so it is vital that affected children are monitored to help avoid crises."



Above: **Louise Brown**



Above right: **Professor Faraneh Vargha-Khadem**



William loves basketball, but has to be careful not to overdo any exercise

Sickle cell disease can often cause damage to the frontal lobes of the brain. These frontal lobes play an important role in short term memory, attention, and the ability to plan and multitask, so children with damage to these brain areas often find it difficult to maintain their attention and to concentrate. Over time, these problems can lead to reduced learning ability and interfere with the normal development of intelligence.

Professor Vargha-Khadem explained: "The learning problems associated with SCD are often diagnosed once the child has reached

adolescence when the difficulties have become instilled and have caused psychological damage as well. We have devised techniques to identify children who would be at risk of such problems with the aim of intervening early during the educational process to help remediate learning difficulties. This approach is new. In so doing, we hope to minimise adverse effects so that sickle cell-affected children can have the best start in life."

William's story

William Kamara is sixteen. Diagnosed with SCD when he was just six-months old after a routine test, William has been participating in the research project.

William's mum, Melrose, a Teaching Assistant and also Chair of the Greenwich Sickle Cell Support Group, talks of the difficulties of having a child with SCD.

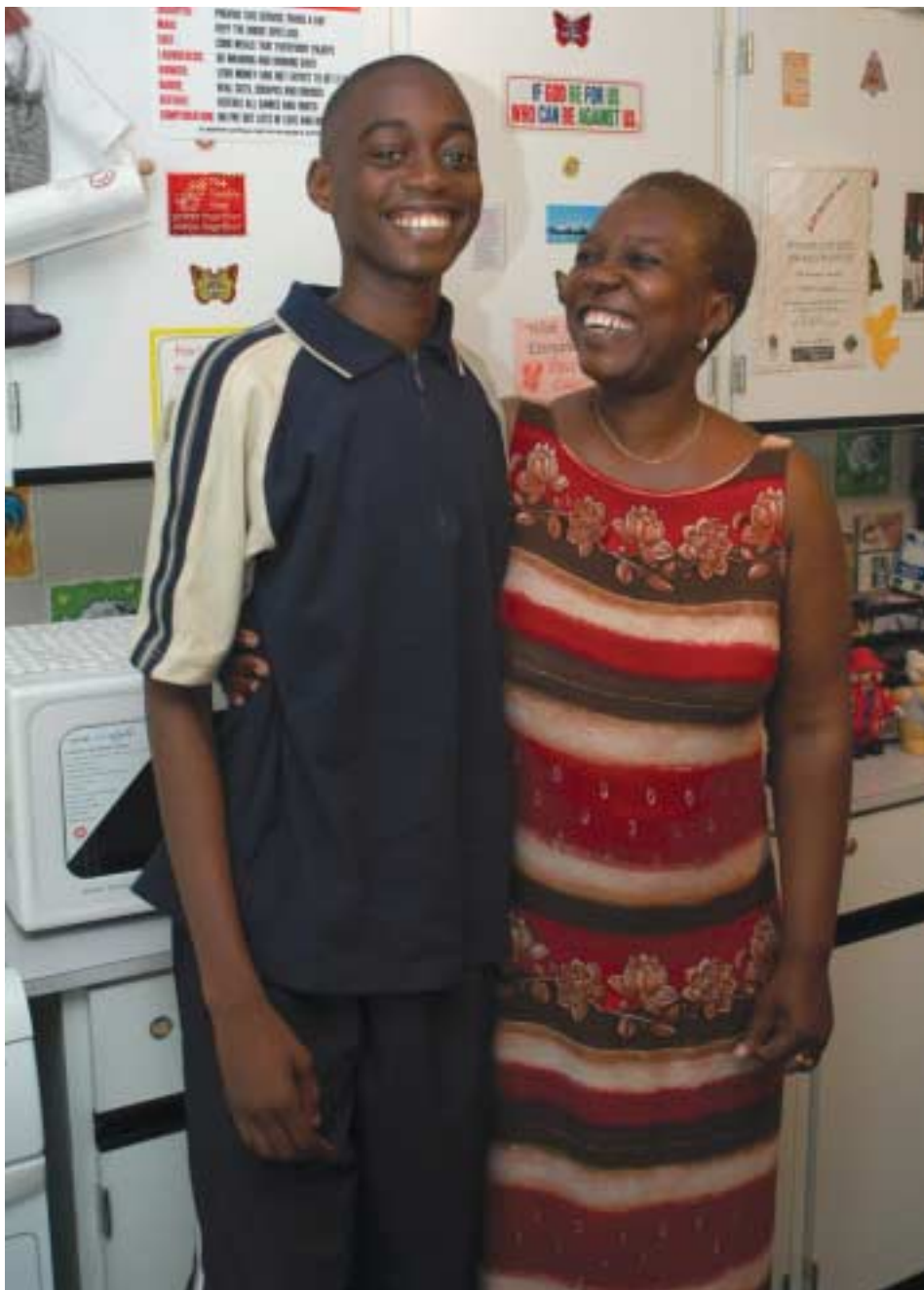
"You are always fearful when you have a child who has SCD. William has spent a lot of time in hospital and there have been many complications. You never know when he is going to have another crisis. Sometimes he balloons up and gets terrible pain in his legs, chest or stomach and then you must get him to hospital so the doctors can administer morphine. When he was younger I think he deliberately used to wait until the middle of the night to get sick and get me out of bed!

"He has to take his medication every day – penicillin to ward off infections and folic acid for his blood, and he will have to do this for the rest of his life. William has to go to three different hospitals: one looks after his breathing and neurology, one treats the ulcer on his foot, and another tests his blood. I make sure that he keeps all his appointments!

"There are lots of areas of William's life that are affected by SCD, and this also affects the rest of the family. His sister used to get so upset to see him in pain and his aunt couldn't believe it when she saw him have a crisis and just swell up before her eyes.



Melrose will fight for her son's best interest



Mother and son make a formidable team

'A devastating disease'

"It's a devastating disease because people can die so quickly if they're not taken to hospital. It is vital that schools are told if a child has the disease and what to do if a child is having a crisis because their life could depend on it.

"I have fought for William since he was young and will carry on fighting for him and others who are in the same situation. There is a stigma attached to the disease so we need much more education about SCD, which is why I run groups and seminars so people can find out more, and talk and help one another."

William loves his computer. After sixth-form college he hopes to study I.T. at university. "I can manage OK now as I'm not really sick any more. I haven't had a crisis for about two years now, but I know that I have to look after myself. My mum nags me about eating and drinking enough and wearing a hat to keep warm, and lots of other things too!

"There is a girl in my class who has SCD and we talk about it sometimes. I'm lucky my friends are supportive of me when I'm ill. Having the disease means that I am not allowed to do certain things like swimming or visit a farm because of the risk of infection, and there is a limit to how much exercise I can do too.

"I sometimes fall asleep at school because of the lack of oxygen to my brain, which the teachers don't like very much! And concentration is hard sometimes and this is another thing associated with the disease. Because you miss some school my mum arranged for me to have tutors at home so that my education doesn't suffer too much. But you just have to deal with it. I'm glad that everything is OK at the moment and I hope it stays that way."

For more information on SCD visit www.action.org.uk/scd

Helping children with the world's deadliest toxin

It wouldn't seem immediately obvious that children might be helped with a potentially lethal toxin, but an Action Medical Research project has proved just that.

Dr Peter Moore is Senior Lecturer and Consultant Neurologist at the Walton Centre for Neurology and Neurosurgery in Liverpool and has been using botulinum toxin to help children with cerebral palsy.

Yes, that's botulinum toxin – the same stuff that people inject into their faces to reduce wrinkles! Although it is best known as a cosmetic treatment, botulinum toxin is used for all sorts of medical conditions. It is effective in relieving some kinds of pain, as well as easing different sorts of muscle spasms, and stopping excess sweating and salivation!

Supporting children with cerebral palsy

Up to 100,000 children in the UK have cerebral palsy, and Action Medical Research has a proud record of funding projects to help them.

Cerebral palsy is neither an illness nor a disease but is a description of a physical impairment that affects movement. It mostly occurs as a result of failure of part of the brain to develop either before birth or in early childhood. This can be due to a blocked blood vessel, complications in labour, extreme prematurity or illness after birth. Infections during pregnancy and early childhood can also cause cerebral palsy.

Children with cerebral palsy often have powerfully and uncontrollably overactive muscles. Dr Moore's project helps such children by making their leg muscles become temporarily

“We are really pleased with our results. No trial has looked so thoroughly at whether it was safe to treat children with botulinum toxin over long periods...”

weaker and thinner when they are injected with the botulinum toxin. Having the treatment means the children experience less pain, discomfort and deformity, and they can control their muscles more easily.

Depending on the severity of their problems, the children received injections in up to five leg muscles. The large calf muscle was often treated, because when this muscle is stiff it forces the child to teeter on tiptoe. Other muscles commonly treated included the hamstrings, which can make the children walk with bent knees, and the inner thigh muscles that pull the knees together and make the feet cross over each other.

Trials

Earlier trials tested the effect of just a single set of botulinum injections, and results showed the

therapeutic effect tended to wear off after three or four months. However, no one knew whether repeated sets of injections might give a longer lasting or even permanent benefit and it was not known if they were safe.

While undergoing the treatment some children were able to walk, or at least walk better. For others, physiotherapy was improved or they were able to manage without ankle splints. And simple everyday things like getting dressed were easier.

Dr Moore, who worked closely with Alder Hey Children's Hospital in Liverpool, told Touching Lives: “We are really pleased with our results. No trial has looked so thoroughly at whether it was safe to treat children with botulinum toxin over long periods. It was very reassuring that we detected no long-term side effects after so

Sausages, not so lovely sausages

The first person to have the idea of using botulinum toxin as a treatment was a German physician and poet called Justinus Kerner over 200 years ago. He discovered that it was the cause of a special kind of food poisoning, which was most often associated with eating sausages! He worked out that this 'sausage poison' reduced excessive activity in the nervous system. The Latin word for sausage is 'botulus', so another German doctor called Muller named this kind of food poisoning 'botulism'.



many treatments, especially in such young children who are still growing, as safety is of paramount importance in any treatment.

Thrilling findings

"Now that we know that single sets of injections are helpful we are looking at our results to see if extra sets continue to help although any benefit derived is limited to the usual duration of the injection cycle. Our future work will aim to show the best ways to use the botulinum toxin and identify the children who will benefit most.

"We are thrilled with our findings but the results really belong to the children and parents who took part in the study. They were wonderful throughout and we are most grateful to them and to the paediatricians and orthopaedic surgeons in the North West who all supported us enthusiastically. We hope that many more children with cerebral palsy can benefit from this groundbreaking Action Medical Research project."



Dr Peter Moore

How it works

Botulinum toxin works by stopping the nerve endings from sending chemical signals to the muscles to make them pull. Doctors select those muscles that are overactive, and carefully inject them with the toxin. It takes a week or two to start working, by temporarily poisoning the nerve endings within those muscles. Botulinum is one of the world's most powerful toxins, and these injections administer only a few millionths of a gram. At time of writing there is a limited service using botulinum toxin in some NHS centres in the UK. The Government has yet to decide whether, and by how much, to expand capacity, but it is hoped that the treatment will be made widely available in due course.

How it was discovered

A new column that aims to shed light on some of the medical advances we now take for granted

Ultrasound scanning



In the UK an ultrasound scan is a routine examination for a pregnant woman, usually performed at both twelve and twenty weeks

The fuzzy black and white image of a baby in the womb produced by ultrasound scanning is now familiar to everyone. For expectant parents it provides an exciting first glimpse of their child. But, more importantly, ultrasound is a crucial medical tool for doctors to check that a child is healthy, and that allows any problems to be caught early on.

In fact, ultrasound also has many other less well known benefits, such as helping tissue repair where muscles and ligaments are torn or strained, as well as many other therapeutic uses. So not only can it help doctors to detect problems, it can actually help solve them too. Projects funded by Action Medical Research have been at the centre of the development of ultrasound into the lifesaving tool it is today.

How does it work?

Ultrasound scans are based on completely harmless sound waves, which travel through the body, through skin and muscles, and get reflected back when they encounter harder objects, like bone and cartilage. The harder the object, the stronger the reflection. The reflected sound waves are used to create an image on a screen, showing exactly what's going on inside the body.

When was it invented?

While the rudiments of sound wave technology can be traced back to the early nineteenth century, it was the two World Wars that provided particular impetus for developing the use of ultrasound, primarily to detect flaws in the metal hulls of large ships and to aid submarines' navigation. In the 1940s, work began on using ultrasound as a medical diagnostic device. Then in 1955 an Action Medical Research funded doctor, Professor Ian Donald, began to pursue the potential of the technique in the field of obstetrics. Despite initial scepticism from many quarters, Professor Donald's work went on to change the face of pre-natal medicine.

What next?

New advances are still being made in the use of ultrasound – a team at Imperial College London have developed 4D scanning, which not only shows the baby in three dimensions, rather than the flat image we're used to, but aims to produce the image in 'real time'. This will enable more effective detection of genetic abnormalities, such as heart conditions, reducing the need for invasive and risky amniocentesis. Building on the pioneering research of Professor Donald and his team, work goes on to give us an ever clearer picture of pregnancy.

Fundraising forum

Why I support Action Medical Research

Karen Jankel has been a volunteer Trustee for eleven years and is MD of Paddington and Co.

What do you do to support the Charity?

As a Trustee, I am involved in all aspects of the Charity's work including strategy and finance – in terms of responsibility the buck stops with the Board of Trustees.

How did you first get involved?

When I started working at Paddington and Co 22 years ago, I became the main point of liaison between the Charity and Paddington himself. The Charity uses Paddington to help with fundraising in a variety of ways. I still look after Paddington's interests today – approving ideas for using him and hopefully coming up with a few ideas myself!

Why do you support Action Medical Research?

Although it is just one charity, because of the broad remit I feel like I am supporting several. Even without my Paddington connection I would support Action Medical Research.



What would you like to see happen with the Charity in the future?

To see it become better known and I hope the new name and identity will go some way to achieve this.

What aspect of our medical research is most important to you and why?

The work on hip replacement surgery and ultrasound are just two of the ways in which the Charity has touched my life. But its work on Motor Neurone Disease is particularly important to me because my mother-in-law died of this terrible condition and so any work investigating it has great personal significance.

What do you see as Paddington's role in the Charity?

As the mascot of Action Medical Research, he brings a light-hearted element to very serious work. He helps with publicity for the Charity as journalists love to photograph him at events and this helps with awareness.

One ted-tastic day!

It's been a fantastic year for Bring Your Bear. Schools, clubs and businesses all over the UK celebrated Paddington's birthday on June 25th by organising Bring Your Bear events to raise funds for Action Medical Research.

Over 650 organisations took part, holding activities such as teddy bear picnics, face painting competitions, treasure hunts, sponsored events and even a teddy bear 'Olympics' in order to raise money for Action Medical Research. Over £90,000 has been raised so far from these events – a magnificent result!



Special mentions go to the top three fundraisers for Bring Your Bear 2003:

- Lochpot Primary School in Aberdeenshire who raised the staggering sum of £1309 between just 63 children
- Winsor CP School in Beckton, London, which raised a wonderful £1205
- St Nicholas School in Kingsbury, London (pictured) whose 75 children contributed a goodly £925.

A big thank you to everyone who took part this year – whether you raised £10 or £1,000, every penny helps us fund vital medical research.

If you would like information on taking part in Bring Your Bear 2004, please telephone 01403 210406 or email byb@action.org.uk

Paddington badges on sale soon



Paddington Bear will soon be appearing in over 2,000 Lloyds TSB branches around the UK on behalf of Action Medical Research.

Taking the form of badges, our ever-supportive mascot will be gracing the counters from late November to late January making him an ideal stocking filler. The new "zip pull" badges look great, come in 5 different designs and can be

attached to zips on clothing or school bags, or even used as a key ring. With a suggested donation of £1, the Charity hopes to raise over £50,000 from the badge sales. Do buy one (or two!) when you see them.



As a charity, Action Medical Research is entirely reliant on voluntary donations. Here we profile news and views from our events and fundraisers

Bridging the gap

When our Bury St Edmunds fundraising group organised a bridge tournament to raise funds for Action Medical Research, they hit on a simple idea with huge fundraising potential.

The tournament took place between March and June this year, and followed a very simple format. A group leader and playing partner locate four other bridge pairs, each participant giving £10 to Action Medical Research. Each pair plays a session of three rubbers against the other pairs – four matches in total – entertaining in turn at their houses and providing a simple tea. Once all the matches have been played, the group leader sends all the scores to the organiser, who aggregates them to find the two prize-winning pairs with the highest scores.

Beginning as a local event, the tournament quickly gathered momentum with 140 bridge enthusiasts taking part from five different counties! In all, £2000 was raised for the Charity.

Next year's tournament is set to be even bigger and better as the organisers hope to expand the event across the country. If you, or someone you know, would like to take part in Bridge Tournament 2004, please ring 01403 210406 for more details.

Did you know?

Action Medical Research has a network of over 200 fundraising groups across the UK. Call 01403 210406 to find the nearest one to you!



Hidden story of bike ride's success



Action Medical Research press officer Rob Orme writes

In the early hours of a late July morning, 122 cyclists converged on Blackheath Common for the start of the London to Paris Bike Ride. This was double the number of cyclists who took part last year and is set to raise over £125,000 for vital medical research.

During the four-day ride the cyclists notched up 300 miles in the saddle en-route to Paris to witness the excitement of the final leg of the Tour de France.

The cyclists embarked on the ride for a variety of reasons... some for the challenge, others for a love of cycling, and still others for the closeness of their connection to Action Medical Research. Amongst the people who take part in our fundraising events there are often moving personal stories hidden away.

Take Trevor Gilby, who chose the ride based on date, distance, duration, and because it combined with the Tour de France. But, when Trevor looked at the Charity's work he realised Action Medical Research was a cause that really was close to his heart.

Trevor explains: "My brother and I were born sixteen weeks premature as identical twins, each weighing under three pounds. We were both put in incubators and the doctors, fearing the worst, suggested that we be christened immediately. They gave my brother the best chance of survival, so he was given my parents' preferred name, and me their next choice. After a week of fighting, sadly, my twin brother passed away, but I struggled on against the odds."

Maybe the survival instinct born in Trevor over 44 years ago ensured he would finish

the ride despite cycling much of the final day with only one pedal. Through his sponsorship, he is gratified to have supported research into premature babies, so that more children can now survive the traumas of early birth.

Trevor continued, "Had Action Medical Research been around much earlier, and been supported by such amazing people that I had the privilege of cycling and socialising with, then I would like to think that there would have been two Gilby brothers receiving their medals on the last night of the ride and not just one."

London to Paris takes place 21st – 25th July 2004. Telephone 01732 744031 or visit www.action.org.uk/l2p for an information pack.



Trevor Gilby had an uphill struggle

Research Training Fellowships – 4 new awards!

Each year we select some of the most talented young medical researchers in the country to benefit from the scheme. We fund their research and training for up to three years, giving them valuable time to gain vital research experience. Not only will these young researchers gain an opportunity to consolidate and refine their research skills, but in addition, the research our RTFs will be undertaking is vitally important in its own right. Here is your chance to meet them.



Checking markers on a tissue sample



Angela calibrates the Instron uniaxial testing machine

Angela Deakin

University of Strathclyde

Rheumatoid arthritis – influences on tendon function

Award: £89,831 over 3 years

Angela's route to medical research began with an engineering degree sponsored by Rolls Royce. "I then went on to work for them as a mechanical engineer for seven years," she told Touching Lives. "I spent a year working in the US, but the move back from the States presented me with a suitable opportunity to reassess my career.

"I looked at what my options were, and, given my background, medical or bio-engineering were obvious areas. I was attracted to the opportunity to improve people's quality of life, and to contributing something to society more generally. Having decided to make this career move, I did an MSc in Biomedical Engineering in 2001/02 – and my MSc research turned out to be a pilot study for my training fellowship."

Angela's research addresses a condition which is a predominant cause of disability in older people and afflicts about 600,000 in the UK. Amongst other problems, rheumatoid arthritis causes changes in the structure of tendons, which attach muscle to bone. These changes are thought to influence the mechanical properties of the tendon, which affects the efficient functioning of joints. "There are three elements to my research," Angela explains. "I'll be looking at the mechanical properties of tendons – how they transmit the load that's applied through muscles to bones. Alongside this, I'll be developing a mathematical model of these mechanical properties. I'll also be using a microscope to look at the structure and materials the tendon is made from, so that I can look at the relationship between what the tendon is made from, and its structure and mechanics – in both normal and arthritic tendons. "These three elements will go together to build a biomechanical computer model of the hand, enabling us to better understand why you get loss of tendon function with arthritis. This will help us to find out which therapies do and do not work, and why."

Angela explains how the day-to-day personal contact with a team of researchers contributes to the quality of the work she does. "I really wanted to do something that was part of an established team's research.

Investing in the future is crucial. At Action Medical Research we support the cutting-edge doctors and scientists of tomorrow through our Research Training Fellowship (RTF) scheme.

That way, you know that the work is going to be useful, because you're contributing to a wider project rather than working in isolation, where it might be harder to see the direction the work is taking. Working with a team of researchers gives you added motivation because you know other people are relying on you to get the work done. I enjoy that sense of working in a team and knowing that all of us, technicians and support staff as well as other researchers, are really interested in and committed to what we're doing."

Angela's experience in industry brings to the team an important and productive element. "Working in industry enables you to develop a really strong skills base, and the problem-solving and analytical skills you acquire are very different from someone who has come from a life sciences or medical background. It's the usual thing that two heads are better than one, especially when they bring two different perspectives. You can address a problem in various different ways and find more innovative solutions when you can bounce different ideas off each other."

Giles Kendall

University College London

Understanding brain damage in newborn babies

Award: £154,086 over 3 years

1 in 400 babies are born every year in the UK with some form of brain injury. Evidence suggests that infection, as well as lack of oxygen, may play a significant role in the brain damage that can occur during childbirth.

"There seems to be a relationship between infection, lack of oxygen, and a key molecule called 'tumour necrosis factor' in producing brain damage, but we're really not sure yet what the relationship is," says new RTF Giles Kendall. "It's a long 'pathway' where something affects something else which in turn affects something else." If successful, future therapies could be developed to block this molecule in babies at risk, thus helping to reduce the occurrence of such brain injury.

"I get real satisfaction from the difference I make to individual lives in my day-to-day work as a paediatrician. But research has a more wide-ranging impact – potentially making a difference to every doctor's clinical practice, and improving the lives of many more people."



Giles Kendall

Dr Wendy Stannard

University of Leicester

Pneumonia in children – the effects of virus infection on susceptibility

Award: £129,414 over 3 years

Every year the bacterium pneumococcus causes 1 million child deaths worldwide. There is evidence that infection by a virus that causes respiratory infections in children (called respiratory syncytial virus or RSV) may increase the chance of the child being infected by pneumococcal bacteria possibly leading to life-threatening diseases such as pneumonia and meningitis. It is thought that RSV increases the likelihood of pneumococcus infection by enhancing invasion of the bacteria through cells lining the airway into the body.

Wendy's objective is to understand how the virus makes the lining of the

airway more susceptible to such infection. Improving this understanding will enable future testing of therapeutic interventions to decrease the incidence and severity of pneumococcal disease – which, as Wendy explains, is ever more important in the face of increasing antibiotic resistance. "Over the last decade in particular, more and more attention has been paid to the fact that bacteria are getting increasingly resistant to antibiotics. Organisms such as pneumococcal bacteria used to be susceptible to antibiotics such as penicillin, but antibiotics are becoming less and less effective. So it's particularly important not only to find ways of preventing infection from the bacteria in the first place, but also finding other mechanisms to treat the infection."



Dr Wendy Stannard with her supervisors Professor Peter Andrew (left) and Professor Chris O'Callaghan

Dr Jane Warren

Imperial College London

Language impairment after stroke

Award: £90,155 over 2 years

It is estimated that one quarter of people who suffer a stroke have aphasia – communication problems affecting speech, understanding, reading and writing. Around 15,000 people a year in the UK suffer aphasia after stroke and, while some people recover language use spontaneously, others do not.

Using specialised brain imaging techniques, Jane aims to identify the changes in organisation of speech processing after stroke that are associated with successful recovery of language comprehension. "Increased bloodflow in the brain shows where there is increased nerve cell activity – so where the brain is working hardest. I'll be measuring bloodflow while a person is having sound played to them – some nonsense, and some words, to see what their brain recognises as speech and how the brain responds to it"

The ultimate aim of the research is to come up with a combination of drug and behavioural therapies to help people recover their language use. "Until we know more about what the brain does by itself to get better, we won't be able to help people who don't recover language after a stroke. But everyone is different – no two brains are the same, which is one of the amazing things about studying the brain, and one of the challenges."



Dr Jane Warren with a volunteer in the PET scanner

Growing your own knee cartilage!

To most of us growing bits of your own body sounds like something from a science fiction film. But at the Royal National Orthopaedic Hospital in Stanmore in London science fiction has become a reality.



Dr Akmal's working laboratory

Dr Mohammed Akmal is a surgeon who is also involved in developing tissue engineering techniques. With Action Medical Research funding, he and his team have successfully developed a technique to grow knee cartilage. Cartilage is a tough, fibrous connective tissue with excellent shock absorbing qualities. It is also found in other joints such as the ankle, hip and shoulders and provides a smooth protective surface for ease of movement.

Damage to cartilage in the knee usually happens when people are playing sports and they twist or fall badly. Around 10,000 people each year suffer from cartilage damage as a result of injury, often from football, tennis or skiing.

It has been long recognised that once damaged, cartilage fails to heal or renew itself due mainly to an absence of blood supply. In the worst scenario it can lead to osteoarthritis where the bones rub against one another – a hugely painful condition.

But total knee replacement is not a satisfactory solution in young people as the prosthesis is prone to infection and wear which may lead ultimately to more extensive and risky revision surgery. Current techniques to deal with cartilage damage involve creating bleeding within the joint which results in a blood clot on the surface of the bone. Under the stimulus of movement and weight the blood clot transforms into a form of primitive cartilage, but because it is not the same quality as the original cartilage it does not last very long but will provide some relief to the patient.

In the bioreactor

With only these inadequate options, the race was on to find a way to grow cartilage more effectively. The team at Stanmore found that such growth is best promoted by using a special device called a bio-reactor:



Dr Akmal (right) and colleague Dr Mike Wiseman examine pictures of cartilage

A small number of cartilage cells are harvested from a non-weightbearing portion of the knee joint during the patient's initial examination. These cells are then cultured in the laboratory to produce much larger numbers before being re-implanted into the defective area of the knee joint. Using your own cells for the operation has the advantage of lessening the risk of infection and preventing rejection. The cells grow within the bio-reactor in a compartment that holds special 'food' which the cells need to grow. The machine revolves constantly and this motion stimulates cells to grow in the best way as they receive the right amount of nutrients. Once matured the culture process produces new cartilage-like pellets which may be used for implantation. It takes up to two and a half weeks to grow enough cells to be transplanted and many millions of cells are needed to repair just a small defect.

Dr Akmal told Touching Lives, "This Action Medical Research grant has really helped us to better understand what cells do and thereby improve the techniques of growing them for the purpose of implantation. The traditional method of growing cells involves having them in static flat culture plates, but this means that cells fail to get enough food for normal development. Using the bioreactor enables us to successfully grow a much greater number of cells which are of much better quality and resemble those of normal joint cartilage.

"Having the best cartilage to work with means that patients should have a better outcome. And the cost of making the cells, the operation and the rehabilitation is about the same as having a total knee replacement. But the real advantage of using a patient's own cells in this way is that it is a far less invasive procedure and patients should be up and about much quicker.

"The technology that we have developed could also be applied to other body tissues and may

prove to be a critical step in human tissue engineering. Skin, kidney and liver cells can already be grown with great success and there is also progress in using a patient's own stem cells from the bone marrow. The potential to repair spinal discs in patients with backpain is now on the horizon, which we intend to investigate."

Patched up, plugged and transplanted

Richard Painter, an I.T. consultant from Hemel Hempstead, damaged his knee while skiing in 1989 at the age of 32. The injury did not immediately lead to any problems but three months later while he was playing squash his knee gave way.

He had his knee repaired in 1990 using a patch made of carbon fibre and one of the bleeding techniques described above. Richard was advised to continue to do all the sports that he

likes doing – squash, running and skiing. But five years later he was experiencing problems again.

He was told that the patch had worn out and that there was nothing more that could be done for him. His family has a history of heart disease and so exercise and keeping fit is very important to him. But Richard refused to accept no longer being able to play all the sports he loved – and the possibility of osteoarthritis – so he started to do some medical research of his own. He read up on a brand new treatment which involved growing the patient's own cartilage and transplanting this back into the knee. After some persistence Richard found a surgeon willing to perform the new procedure on him, despite the fact that he already had one failed operation behind him.

"I had my cells cultured and a few weeks later Professor Bentley of the Royal National Orthopaedic Hospital did the implantation surgery. It was quite an easy procedure, a lot quicker than the patch I had had before." Richard Painter was one of the first patients in the UK to undergo cartilage transplant surgery. Since his operation, the techniques to perform the surgery and culture the cells have much improved. Dr Akmal's bioreactor research will improve the quality of tissue used for transplantation and will ensure that transplanted tissue integrates much better into the knee.

"I'm thrilled with the results," said Richard. "It gets better and better with time and after five years I can now go skiing without any pain which is wonderful. I can also go running again and recently did four miles! I'm keen to maximise the use of my new knee!

"I think growing your own tissues for repair is the future for a lot of diseases. I am delighted with the results that I have had from the procedure and am very grateful to still be able to enjoy all the sports I love doing."



Richard Painter can now run without pain

Meet the Researchers



Here we get behind the white coats and find out about the day-to-day lives of our researchers. **Marianne Magnusson** is one

member of a team working on an Action Medical Research funded project investigating back pain at the University of Aberdeen.

How did you get to where you are now?

I trained as a physiotherapist in 1969, and continued with research, getting my PhD from Göthenberg University in 1991. In 1994 I left Sweden for the US and worked in Iowa, and I arrived in Aberdeen in 1998, where I'm now a senior lecturer in Ergonomics.

What might you do in an average working day?

My working life is very varied. Obviously I spend a lot of time teaching, and I have several research projects which I supervise and for which I'm the lead researcher. Up to a quarter of my working life is spent at an orthopaedic clinic, which is where I get to meet patients. I might also spend time writing scientific articles, or a chapter for a textbook.

What are the most rewarding aspects of your job?

I really enjoy the challenge of research. I like the process of coming up with an idea and its application, and designing a study. The really fun part, though, is analysing the data once you've done the study. I also love writing articles for publication – seeing your own words in print is very satisfying.

What would you be doing if you weren't a medical researcher?

Something I've always dreamt of doing is going abroad and working with an organisation like the World Health Organisation. Or I could be teaching at a different level, say working with students going into sports training, and using my expertise in that way.

What do you do in your free time?

My partner and I go away somewhere in Scotland almost every weekend. Since I moved here, I've been determined to make the most of living in such a beautiful country. Even if we don't go away, we go out and have a picnic somewhere. Also, I often go back to Sweden to see my children – my first grandchild has just been born so you could say that my next career move will be to be a good grandmother!

The Doctor's notebook



Dr Mark Porter writes exclusively for Touching Lives to address some of the most common health issues affecting people in the UK

A pain in the back

Low back pain affects about 80 per cent of all people at some stage of their lives. Back pain is more common in women and typically affects working adults between the ages of 20 and 55 and accounts for 12 million GP consultations a year! Interestingly, occupation seems to have little bearing on back pain, suggesting that most cases are not the result of a person's job.

The causes

95 per cent of cases of back pain are simple sprains and strains of the lumbar region (lower back) – known as 'simple acute back pain'. But causes can vary from arthritis or a slipped disc, to osteoporosis or cancer. It's the GP's role to differentiate between simple acute back pain and these more sinister causes.

The symptoms

Suspicious symptoms include pins and needles and weakness in the legs, incontinence (or problems passing urine), accompanying fever, weight loss, or any other indicator of general ill health. Cases giving particular cause for concern include those that don't settle within 4 – 6 weeks, where there is a history of violent trauma (e.g. a nasty fall or road traffic accident), or where the patient is under the age of 20 or over the age of 55.

The treatment

Bed rest and time off work are now generally frowned upon for the vast majority of cases of back pain. Evidence now suggests that an

immediate or early return to physical activity, including work, leads to a quicker recovery. X-rays are a waste of time in most cases of back pain and are only done if the doctor suspects an underlying problem. An X-ray of the lumbar spine gives a radiation dose equivalent to 65 chest X-rays and should be avoided unless it's likely to help clinical management.

There isn't much evidence to support the use of creams and sprays, but painkillers make life more comfortable and ease muscle spasm (as can low dose diazepam). Assuming sinister causes have been ruled out it makes sense to consider some form of manipulative therapy (such as osteopathy, chiropractic or physiotherapy). Thankfully time is a great healer and most cases will settle within a few weeks.

Prevention better than cure

The key to a healthy back is to avoid injury (make sure you lift and carry things properly!) and to ensure good tone and posture by exercising the core muscle groups around the midriff – the abdominal, back and pelvic floor muscles. Ask a physio or qualified trainer to show you how.

For more detailed advice and information contact Backcare (formerly the National Back Pain Association) on 020 8977 5474 or visit their website at www.backcare.org.uk



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A day in the life

Callum's story gives hope to others

At just 10 months old, little Callum Pollock had a bone marrow transplant. The donor was his seven-year-old sister Chantelle. Such desperate measures were needed to give Callum, who has Hurler Syndrome, the chance to live a life that otherwise would have been denied him.

Callum is now seven. He attends school, goes to Tae Kwondo, plays football – and is something of a celebrity in his home town of Blackpool.

With his mum Carla, dad Donnie, brother Donnie junior and sister Chantelle, he has recently been on holiday to Majorca, where a stranger approached his mum.

"I know you," she said. "You're from Blackpool – I recognise Callum."

Carla told Touching Lives, "It happens all the time, everyone seems to know Callum, no matter where we are. He's a bright, cheerful little boy who will talk to anyone. Sometimes, it's hard to believe we've been through so much as a family, and the hardest part is that we still don't know what the future will bring. Callum's condition is so rare that no-one can predict what will happen."

Although he was a bouncing 10 pounds at birth, Callum became a sickly baby, vomiting and apparently having difficulty breathing. After several trips to the doctor and the hospital, at



five months his family still didn't know what the problem was.

Desperate

"It was my mother-in-law who pushed me to make a stand and insist that we get some sort of diagnosis," said Carla. "I didn't know what to do but we knew that there was something very

wrong and he was becoming so ill that we were desperate.

"When I was told he had Hurler Syndrome and without radical treatment probably wouldn't live beyond nine I was devastated. We were packed into a tiny room at the hospital and I just couldn't take it all in.

"Then other doctors in the hospital started to come and look at him because they had never seen a child with Hurler before. He was the youngest ever diagnosed and the first case diagnosed in Blackpool, but I hated the fact that he seemed to be some sort of novelty. I can understand now why people wanted to look, but at the time it was terrible."

Carla was contacted by the MPS Society, a support group for families of Hurler children, but most of the literature she read about the condition painted a very gloomy picture.

A chance

She said, "When he was diagnosed, the consultant said he needed a bone marrow transplant quickly, even though he was only a baby. I wanted to know more about the condition, but didn't read anything about Hurler Syndrome being treatable. We were terrified, but decided that we had to give Callum a chance."

"Without the transplant he would deteriorate mentally and physically and die. The transplant itself carried a big risk – at first he was too weak to go through it and we had to wait – but we saw it as a chance, and we took it."

Big sister Chantelle was found to be the perfect match, and though Callum was in hospital for a year after the transplant, his life was transformed as a result.

"I started to see the Hurler symptoms being reversed," said Carla. "He was still very prone to infection and was in and out of hospital for several years, but he's at the stage now where he seems better able to fight off the chest complaints he used to get and I don't worry about him quite as much as I did. We take the view now that we all have to get on with life – whatever it brings."

Pressure

But having such a sick child inevitably put a huge amount of pressure on Carla, Donnie, who works as a painter and decorator, and their older children.

Carla said, "I was living at the hospital, but the other kids and all the family were just brilliant. I had worked right up until having Callum, but I've never been able to go back. I decided to enrol on a computer course when he started school, but the day I started I got a phone call to say he was ill. I think I was called home five times during the course."

Callum and his family have lost count of the operations he has had. Hurler children can suffer from curvature of the spine and a procedure to try and straighten his back has not been successful.

Callum's spine is the worst doctors have ever seen, curving in two different directions, and his X-rays have travelled the world as his consultants try to find an expert who can help.



The Pollock family. Sister Chantelle donated bone marrow to give Callum a chance

Carla said, "We're just getting on with things, and I'd like to think that other parents who have a child with Hurler Syndrome will be able to read about our experiences and see that there is a chance for them."

"We're not soft with Callum, we don't treat him much differently from the other two, and I think in many ways it was good that we had two children already."

"He still has some problems. The curves in his spine mean that he walks on his toes most of the time, and he suffers bad migraines, which seem to come on when he's excited about something."

"But we decided that we have to get on and really live our lives, even though we don't know what the future will bring. The bone marrow transplant has undoubtedly prolonged Callum's life, but his doctors can't say what will happen, or when."

"Callum's story is in the MPS literature now and that makes me feel better, because other mums and dads in our situation will be able to read that there really is hope."

New treatments under investigation

Action Medical Research has given a grant of £105,000 to a project at the Department of Haematology, Royal Manchester Children's Hospital, to investigate the modelling of new therapies to treat Hurler Syndrome.

Hurler Syndrome is a single gene disorder causing the deficiency of an important protein. It results in the build up of mucopolysaccharides – long chains of sugar molecules – that in most people are continuously broken down and recycled. In Hurler sufferers, these accumulate instead and can cause severe damage. Sufferers

have progressive mental and physical problems and rarely live beyond the age of nine.

Dr John Grainger is leading the research into new therapies. At the moment, Hurler Syndrome can be treated with bone marrow transplants – but this is limited by the availability of donors, toxicity and the problems of rejection.

He said, "The research is investigating the use of mesenchymal stem cells (msc) administered alongside genetically modified bone marrow cells. Mesenchymal stem cells can be derived from the affected individual and are able to develop into other tissues of the body. In addition msc can dampen down the immune reaction to bone marrow cells reducing the toxicity and the possibility of rejection."

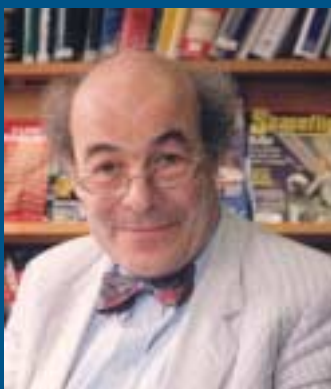
"To date we have established a protocol for isolating msc and demonstrated how they can develop into different tissues and for genetic modification. We have now moved into a transplantation model to investigate the potential to aid engraftment and establish the fate of transplanted cells."

About Hurler Syndrome

Hurler is the most severe of a group of three similar disorders. Hurler Syndrome was first recognised in 1919. It is rare, occurring in about one in 100,000 births, but because its cause is a recessive gene, it can suddenly appear in a family with no other known cases. Without a bone marrow transplant, Hurler children rarely live beyond eight or nine years.

The MPS Society supports families with Hurler Syndrome. Telephone 01494 434156 or visit www.mpssociety.co.uk

Jargon busters



TV scientist Professor Heinz Wolff explains some common – and some not so common – medical and scientific terms.

Sickle cell disease

Sickle cell disease is a disorder of blood composition where abnormally high levels of sickle haemoglobin cause red blood cells to become misshapen when they give up the oxygen they carry. This results in the characteristic crescent or 'sickle' shape. On re-oxygenation, red blood cells may regain their normal disc shape but they become weaker with subsequent sickling and unsickling. Repeated sickling-unsickling leads to early destruction of red blood cells. When the disease is active, the abnormal shape of the red blood cells interferes with their proper function.

Folic Acid

This is a vitamin essential to life. Since people cannot manufacture folic acid, external sources are necessary. Citrus fruits and juices; green leafy vegetables; beans; peas; liver; and whole-wheat bread are natural sources of folic acid. It can also be made synthetically. This is the origin of the folic acid dietary supplement.

Mathematical Model

Scientists and Engineers can obviously do experiments on the material with which they are dealing, but such experiments tell you what is happening, like a tendon breaking under stress, but little about the mechanism why it breaks. If you know something about the properties of the different materials which make up the tendon, you can have a shot at calculating what ought to happen, under different test conditions. Depending on the results of further experiments you refine the mathematical model until it allows you to explain your experimental results. You can then begin to forecast what would happen if you were able to change the properties of the materials, for instance to make a better artificial tendon. It is really no different from a business plan, which is a mathematical model of a business you hope to start. As experience is gained the model is adjusted to reflect the real world more accurately and so becomes a forecasting tool. The availability of computers has made the use of Mathematical Models very attractive, because the calculations involved in modelling can be very complicated, yet the "experiment" of varying some characteristic can be made in minutes.

New hope for coeliac sufferers

"When I was an undergraduate, it was thought that perhaps one in every 1,800 people suffered from coeliac disease" said Paul Ciclitira, Professor of Gastroenterology at St Thomas' Hospital, London, and leader of the research team.

"Now we know that one per cent of the population is actually affected – a huge number of people – and though we have come a long way since young children died because the condition went undiagnosed, there is still a lot for us to learn about the way in which it is inherited and how the disease process actually works."

Coeliac disease is a complete intolerance to wheat gluten, but because its primary symptoms are stomach upsets and gastric pain, it can go unrecognised for some time.

It tends to be hereditary; in fact the research completed by Professor Ciclitira and his team indicates that members of a family where one person is a known sufferer are many times more likely to have the disease than people from families with no known cases.


He said, "In our research we collected blood samples and checked for antibodies that are found only in people with coeliac disease. This was followed up by a small bowel biopsy, to confirm the presence of the disease. We used the blood samples to examine the DNA for genes associated with the development of the condition.

Screening

"In the UK there is no routine screening for family members, but our research indicates that it should be considered. In many other European countries screening of relatives is automatic, and here I think it could lead to



Healthy red blood cells (left) contrast with sickle cells



Wheat is a staple part of the Western diet – but it has a lot to answer for. Wheat flour, rye, barley and possibly oats, contain gluten (a type of protein) and are the cause of coeliac disease, which affects one in every 100 people in the UK. This sensitivity to gluten can cause inflammation and damage to the small intestine and symptoms can include abdominal pain, weight loss and in extreme cases, malnutrition.

The symptoms can be controlled by following a strict gluten-free diet – but Action Medical Research and Sainsbury's teamed up to fund a three year study into coeliac disease that could help its diagnosis and treatment.

earlier diagnosis, saving the patient a lot of discomfort and the NHS money in treatments.”

What is the actual process that causes coeliac disease? A particular peptide (a breakdown product of wheat gluten during digestion) has been shown to cause tissue damage in the small intestine of sufferers. This was demonstrated by the team's research, which has so far involved a number of volunteer patients.

A clearer understanding of this peptide and its action on the small intestine is likely to be critical in determining the exact cause of coeliac disease and its possible prevention.

Immune system

The study of genes associated with the development of the disease may unearth a way to work with the body's own immune system to 'block' the inflammation altogether and stop the symptoms completely.

A genetic test for coeliac disease may be developed that could replace the more invasive biopsy of the small intestine, which doctors currently rely on and which means that sufferers have to undergo a surgical procedure in hospital.

Just as exciting, the work could lead to a technique to genetically modify wheat, or another cereal crop, so that its flour does not affect coeliac sufferers.

Following a strict gluten-free diet inevitably means that things like cakes and pastries are off the menu. But a flour that does not contain gluten could mean that for the first time coeliac patients may safely be able to eat a sandwich made of something that resembles real bread!

Professor Ciclitira continued, “It has been a fascinating study, like being given the pieces of a jigsaw and having to put them all together. We

knew that wheat was the driver and that has allowed us to focus on the mechanism of the disease to find out exactly what's going on.

Good results

“We have had some very good results and it's important now that more research continues to build on our findings. For example, we investigated the prevalence of coeliac disease in other disorders of the immune system and found that four per cent of patients with conditions such as Lupus (a long-lasting disease of the immune system where the body attacks its own cells and tissues causing a wide range of chronic symptoms) also suffer from coeliac disease. This has never been reported before.”

Geoff Spriegel, Technical Director at Sainsbury's, is equally pleased with the project's findings. “An increasing number of our customers are asking for a wider range of gluten free foods. Sainsbury's is delighted to have supported this innovative project by Action Medical Research which is clearly helping to improve understanding about the causes of coeliac disease. We're excited by its potential to improve the food choice and product quality for sufferers.”

Help and advice is available from Coeliac UK on 0870 444 8804, or log on to www.coeliac.co.uk

Living with coeliac disease

Andy Bunce, a customer communications manager in the defence industry, has suffered gluten intolerance since being a baby – and has always had a restricted diet.

He said, “Ordinary bread, cakes and things like pizza bases are out. If I eat them I feel queasy and gradually start to get sick. I was originally diagnosed because I wasn't putting on weight as a baby and though I tried reverting to a normal diet when I was 17, after a couple of weeks I felt so ill that I had to go back to wheat-free.

“Things are improving for coeliac sufferers. I was at primary school in the late 1960s and I had to have special dispensation to take my own lunch rather than have school dinners.

“Back then, there was very little available for coeliac sufferers 'off-the shelf' and those gluten-free cakes and biscuits that you could buy in the shops tasted awful. Things are better now, food labelling is certainly



improving, though many gluten-free products still tend to be expensive.”

Andy, who lives in Preston, Lancashire, has three children but none are affected by coeliac disease. He said, “It seemed to skip a generation with me – I had a great uncle who is thought to have suffered from it – so we'll be keeping a close eye on our grandchildren, if and when they arrive.”

In the News



We receive press attention for both our medical work and our fundraising activities, and over the past quarter Action Medical Research's media coverage has been a real mix of the two. Here's a look at the best bits...

The summer was a busy time, with several research projects coming to an end and exciting results to publicise. The Times featured our pain scales project in its T2 supplement. Here, researchers developed a scale that can be used to help understand pain in children who are not able to speak and communicate the type and location of their discomfort. The article generated a lot of interest and positive feedback from the public.

Additionally, the Daily Mail published an article on the results of another Action Medical Research funded project which has found that taking folic acid supplements during pregnancy can reduce the chances of babies developing cleft lip and palate in the womb. The Charity is already renowned for its discovery of the role of folic acid in preventing spina bifida. Other publications also picked up on the story, including Essentials magazine.

Bring Your Bear 2003 has featured in hundreds of regional newspapers in Britain, with lots of photos of kids tucking into picnics and Paddington making surprise visits all over the country. A great way of introducing kids to our Charity!

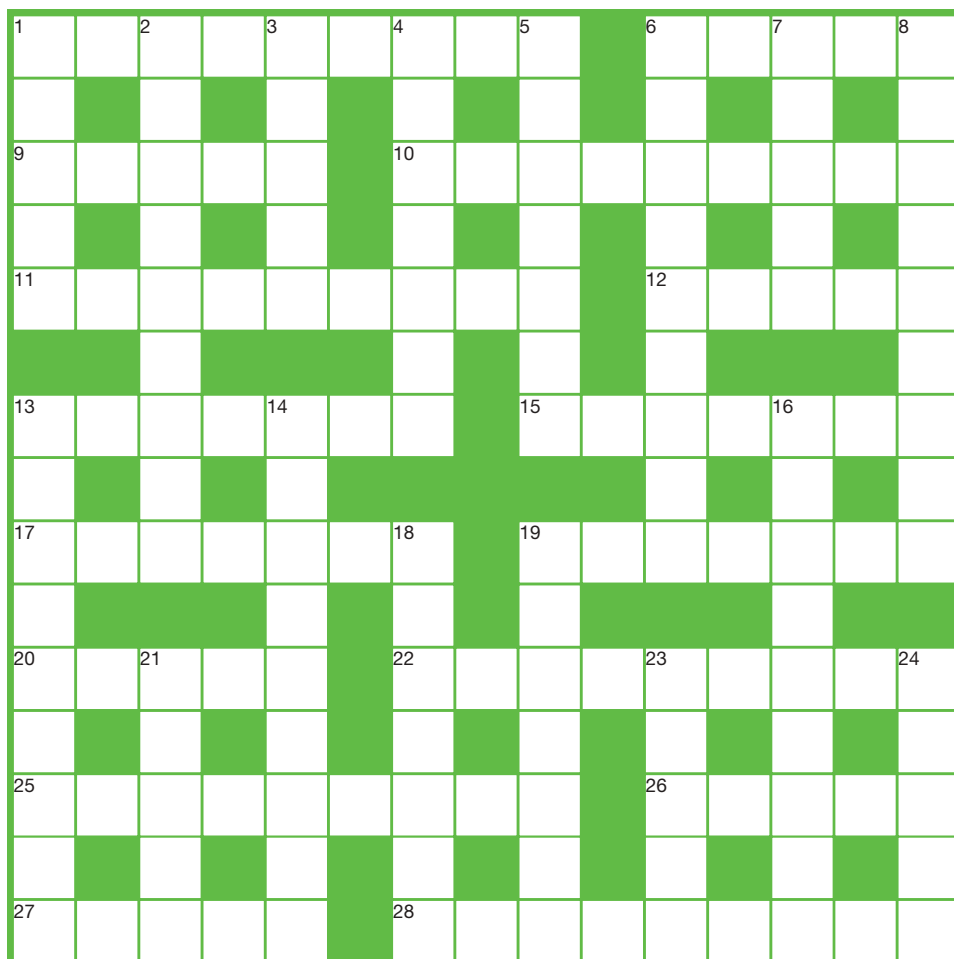
Action Medical Research's support of Professor Herman-Taylor's work into Crohn's disease was again in the spotlight recently when The Guardian reported his latest findings. He has come up with the strongest evidence yet that this distressing digestive disorder is caused by the same bacterium (MAP) found in cattle with the condition. The article highlighted that Action Medical Research has given almost £1 million pounds to the Professor over the last 17 years for his vital work.

Touching Lives online!

You may be interested to know that there is a PDF version of this magazine available online at www.action.org.uk. Click through to our news and media section. We'll be keeping an archive of previous editions there too.

Crossword 5

by Esmee



Some of the answers can be found within this magazine.

Across

1. Pursues (7,2)
6. How Richard walks before treatment (5)
9. Temporary doctor (5)
10. Callum's martial art (3,4,2)
11. Closes tight (5,4)
12. Regulator (5)
13. Starting point for bike ride (7)
15. Painter's sport (7)
17. Very busy (7)
19. 19dn commode (7)
20. Clear (5)
22. Pen for livestock (9)
25. Church in 23dn (5,4)
26. Fend off (5)
27. Gene regulating 1dn acid (5)
28. Where St Nicholas's school is (9)

Down

1. Kind of acid which may combat cleft lip (5)
2. Profitable (9)
3. Fiona the fundraiser (5)
4. Bag (7)
5. Machine for folding (7)
6. Nadirs (3,6)
7. Painter meant to be interpreted (5)
8. It emphasises the star (9)
13. Toxin which may help kids with cerebral palsy (9)
14. Senior undergraduate (5-4)
16. Unrehearsed (9)
18. Bird of prey (7)
19. Common (7)
21. Get hold of – a song (5)
23. End of Trevor's bike ride (5)
24. Vice Provost David (5)

For your chance to win a Parker fountain and ballpen set, send completed crossword to: The Editor, Action Medical Research, Vincent House, Horsham, West Sussex, RH12 2DP. Closing date for entries is 28th November.

Congratulations to Wendy Barlow from Norfolk who won the prize in our Summer 2003 issue.

Where the money goes!

Another opportunity to see how your donations to Action Medical Research are making a real difference.



Professor John McGrath

Do you take your teeth for granted? Most of us do, and beyond the occasional trip to the dentist hardly give them a second thought. And what about your hair? It might have unruly days, but be honest – does it ever really give you cause for concern?

There are some people who live with debilitating genetic disorders that affect their teeth, nails, skin and hair to such an extent that their whole life is affected.

Now a study funded by Action Medical Research has made important progress towards understanding more about a group of rare diseases affecting the growth and development of body tissue, resulting in improved treatment for sufferers – and potentially impacting on more common conditions such as skin cancer and even on the effects of ageing.

The two-year £88,000 project, led by John McGrath, Professor of Molecular Dermatology

at St John's Institute of Dermatology, London, focused on searching for genes that might have 'malfunctioned' in people born with inherited abnormalities of the hair, skin, teeth and nails.

This malfunction can lead to a range of conditions – 170 in all – known as ectodermal dysplasias. With each condition, the symptoms can range from mild to severe. Some sufferers cannot sweat because of the malformation of sweat glands, some may be born with missing fingers and toes, others need dentures and dental implants from childhood due to misshapen teeth, and some have fragile skin that easily cracks and bleeds.

No known cure

It's thought that as many as seven in every 10,000 births could be affected in some way and though the first ectodermal dysplasia syndrome was identified in the 1860s, there is still no known cure.



The devastating effect of ectodermal dysplasias on the skin

Things start to go wrong in the womb, when the baby develops its tissues and organs. The outermost layer of the developing child is called the ectoderm and it is defects in its formation that lead to ectodermal dysplasias. The ectoderm contributes to so many parts of the body – from the fingernails to the brain – that a sufferer can be affected in many ways.

And as well as the physical discomfort caused by the conditions, patients with highly visible skin and tissue disorders also have to put up with the stares of strangers, and the psychological impact can be profound.

Important findings

Professor McGrath's study has made some important finds, unearthing new genetic faults in several disorders; and about 30 of the known syndromes have been properly classified for the first time, which means that patients will be able to receive a more accurate diagnosis and better genetic counselling about how the disease can be passed on.

The research also discovered a new gene that is abnormal in individuals with another form of ectodermal dysplasia called Kindler Syndrome. It is a very rare disease, with fewer than 100 cases ever reported, and results in thin, fragile skin that blisters easily after trauma. Skin also becomes wrinkled and easily damaged by sunlight.

"Many of our findings will contribute towards the better management and treatment of a range of conditions that can make life very difficult for those affected," said Professor McGrath.

He added, "Our discoveries have opened up a whole new field of research that has relevance not only to the study of ectodermal dysplasias, but to wider fields in health research such as the process of wound healing, the risks of getting skin cancer and the nature of skin ageing.

"Though our remit was to focus on conditions that affect relatively few people, the spin-off research from our findings could potentially impact on millions."

This is just one of the valuable medical research projects we can fund because of your support. Thank you.