TOUCHING LIVES

The Action Medical Research newsletter | SAVING LIVES FOR **70 YEARS** | Autumn/Winter 2022

HOW YOUR SUPPORT IS MAKING A DIFFERENCE

Fighting for children like Paddy, who has a very rare form of epilepsy

Developing a new blood test for inflammatory bowel disease

Tackling peanut allergy in children

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action medical research for children

Action Medical Research is the leading UK-wide charity saving and changing children's lives through medical research.

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Touching Lives is also available to download at **action.org.uk**

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WELCOME

It's a pleasure to bring you your latest newsletter, showing how your support is helping to drive vital medical research forward.

In the UK, there are thousands of children like Paddy, our cover star, living with a rare disease for which there is no cure or very few treatment options. Did you know that there are more than 7,000 known rare diseases? 75% of these rare diseases affect children, with a third losing their lives before their fifth birthday.

With your help, we want to fund more life-changing research to give hope to these forgotten families and we will be working hard to do more on this over the coming months.

There are many different ways to support us, and you can read about some of these on pages 16-19, including our Autumn Superdraw, which gives you the chance to win some great prizes and help children like Paddy.

Thank you for helping us to save and change lives.

Best wishes,
Clare
Editor
cairey@action.org.uk

NB: *Our privacy policy has been updated. View the latest version at action.org.uk/privacy*

70 YEARS
of medical research

AROUND 55
projects across the UK

NEWS

NEW RESEARCH

We have recently announced £1 million of vital new rare diseases research, as part of our ongoing partnership with fellow medical research charity LifeArc.

Four new projects include developing a new treatment approach for children with brain cancer and using advanced genetic techniques to better understand and diagnose developmental disorders.

RESEARCH UPDATE

TWINS SUCCESS

Action-funded research has led to the development of an ultrasound-based technique, which has now been used to treat five pregnant women with twin-twin transfusion syndrome in a new Medical Research Council funded trial. The procedure is helping increase the chance of survival for affected twins by stopping the uneven flow of blood and nutrients between them. Further work will be needed to show how well the treatment works in a larger number of women.

CHRISTMAS CARDS THAT COUNT

Send good wishes and support Action this festive season! We have a great selection of cards available to order now. There are eight designs to choose from and packs of 10 cost just £4.95. Order online at **action.org.uk/cards**

HOPE FOR CHILDREN LIKE PADDY

Three-year-old Paddy has *KCNT1*-related epilepsy, a rare disease which has no cure and has left him severely disabled. Most devastatingly of all, children with the condition often don't live into double figures.

"Paddy is a beautiful little boy who is absolutely adored by everyone who meets him," says his mum, Catherine. "He has this strong presence in a room and brings so much joy. He will let us know if he isn't happy about something, especially if you put him down from being cuddled – that's his favourite thing."

Paddy came into the world 'like an absolute whirlwind' after a 32-minute labour, a baby brother for sister Nancy. "When they told me he was a boy, my life was complete," says Catherine.

"He was perfect and the first two weeks of his life were a breeze. He fed, slept and hardly cried," she says. "Although I had noticed some shaking while he was in his Moses basket, it was passed off as him adjusting to life outside the womb and it didn't seem to bother him."

But at two weeks old, everything began to change. "The screaming started – 15 hours a day, seven days a week. It was absolutely awful."

"Paddy has had thousands of seizures in his lifetime and they don't get any easier for us to watch"

After numerous visits to the doctors, nothing seemed to help, and Catherine instinctively felt that something was seriously wrong. "His eyes would roll and he didn't begin to smile," she says.

"He would just sleep for hours or scream. At eight weeks old, I took him to hospital after what was a noticeable seizure and told them I wasn't leaving."

Paddy had many blood tests, an MRI scan and lumbar puncture, and was given various medications – none of which worked. Catherine and husband James were told it could be months before they had genetic tests back.

The diagnosis, when it came, was more devastating than anyone could have imagined. *KCNT1*-related epilepsy is a very rare and severe neurological condition. It can leave children severely disabled. Many never walk or talk and sadly most do not survive into adulthood.

Research to find vital new treatments

Thanks to your support, researchers at the University of Leeds are working to identify chemical compounds that could form the basis of urgently needed new medicines for children with *KCNT1*-related epilepsies.

"Effective new treatments that can help to improve and extend lives are desperately needed"

Dr Jonathan Lippiat, who's leading this research, says: "Developing a safe and effective new treatment that can help control seizures and prevent damage to children's brains would greatly improve the quality of life for affected children and hopefully allow many more to reach adulthood."

Paddy's heartbreaking diagnosis changed the family's future in an instant. "I was so bitter and grieved for a boy that I had longed for," says Catherine. "Life was never going to be normal again. And to this day it's never been normal. But everyone's normal is different."

"Medical research gives us a sense of hope, not only for Paddy but for others in the future"

At his worst, Paddy suffered up to 50 seizures a day. Finding the right medications to help is a delicate balance. The family never know when something will stop working.

"Paddy's seizures don't get any easier for us to watch. We always ensure we are there to comfort him and hold his hand," says Catherine.

At three years old, Paddy still has the developmental age of a young baby. "He has hit no milestones, and this won't ever change. He makes his own little sounds and will let us know if he is unhappy or hungry. But his only form of communication is crying," explains Catherine.

Paddy's condition has a huge impact on family life. "It has put so many barriers in our way. Every day we have to plan and think about what we are doing or where we can go."

The family's ultimate goal is to keep Paddy out of hospital and as healthy, comfortable and happy as possible – to make his life the best it can be.

"Medical research gives us a sense of hope," says Catherine. "Not only for Paddy but for others. We understand that there may be no miracle cure but if this condition can be better understood then we will know we have helped future children."

THANK YOU!

Together, we can support more rare disease research.

To find out more scan the QR code or visit **action.org.uk/rare**

NEW INFLAMMATORY BOWEL DISEASE RESEARCH

Your support is helping to fund work to develop a simple new blood test to improve the diagnosis of inflammatory bowel disease and help select the best treatment for each child.

The two main types of inflammatory bowel disease (IBD) are Crohn's disease and ulcerative colitis. These long-term conditions cause inflammation of the digestive system, triggering distressing and debilitating symptoms.

"A child may experience painful stomach cramps, recurrent diarrhoea, weight loss and extreme tiredness," explains researcher Professor Jack Satsangi.

Diagnosis is generally made using endoscopy – an invasive procedure usually done under general anaesthetic, which can be upsetting for children.

Professor Satsangi's team, at the University of Oxford, has previously discovered specific changes in a child's DNA that can accurately tell if they have IBD. They now plan to use cutting-edge technology to create a blood test that can rapidly examine these and other genetic changes linked with the condition.

IN NUMBERS

MORE THAN

500,000

people in the UK have Crohn's or colitis

"We aim to develop a new test that can improve diagnosis and personalise treatment for children – improving outcomes and reducing invasive investigations"

Professor Jack Satsangi

WHY WE SUPPORT ACTION

Jenny and James began supporting Action after the loss of their baby son Elijah. With family and friends, they have raised almost £40,000 and inspired so much more, including our first Talisker Atlantic Challenge rowing crew, Team Elijah's Star.

JENNY AND JAMES

Q **Can you tell us a bit more about Elijah and how you became involved with Action?**

A Our third son, Elijah, was born very prematurely, at 25 weeks and three days. We were very lucky to have him, but his life was painfully short. After he died, having a focus, something positive to do in his name, was really helpful. And staff at Action helped us not feel anonymous – helped us feel like we were making a difference, and in turn that Elijah's life could still make a difference.

> **Elijah's life was painfully short**

Action remains a link to him and we have had some very special times because of it. We're still passionate about making life-changing outcomes for babies who are yet to be born.

Q **And Elijah was actually involved in an Action-funded study while he was in neonatal intensive care…**

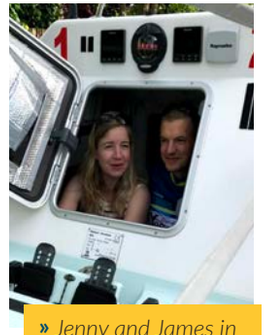
A Yes, he was recruited to a study while he was in hospital. Premature babies are known to be at risk of a serious bowel disease called necrotising enterocolitis. The project aimed to spot early warning signs. I come from a healthcare background and James a statistics one, so Elijah was always going to be involved in research if it could help. As it turned out, Elijah did develop NEC and deteriorated swiftly and significantly.

Q **You have been involved in many charity activities, but the Atlantic Challenge must have been one you weren't expecting!**

A Having the crew row across the Atlantic in Elijah's name blew our minds! The commitment and strength they channelled into the race, all with Elijah's name plastered across their chests, boats and flags, is still a little hard to fathom. It was incredible to wave them off, and to be introduced to people as Elijah's parents. They raised an enormous amount of money – it was a very moving experience.

Q **Ahead of the rowing challenge, you also appeared on BBC1's *The Repair Shop*. What was that like?**

A It was an emotional but beautiful experience.



» *Jenny and James in the Elijah's Star boat*

> **The thought that being involved might just move research forward and change someone else's path in life remains a strong motivator**

We were greeted with such warmth – and Lucia and Dom repaired our seascape painting, a family heirloom, to perfection. We came away feeling like they had gifted us a little more time 'with' Elijah and the programme also aired in Baby Loss Awareness Week. The painting is in our living room and is a beautiful reminder of the row and our love of the sea having lost our boy.

Q **Nine years on, what does supporting Action mean to you now?**

A We still love how the charity gives us a connection to Elijah. The thought that being involved might just move research forward and change someone else's path in life because of Elijah remains a strong motivator. We love to talk about him, we still want his short life to continue to be worthwhile.

FIND OUT MORE

Following in the Elijah's Star team's oar strokes, another crew of Action rowers will be taking part in this year's Talisker Whisky Atlantic Challenge in December. Find out more on page 18.

SAVING LIVES FOR
70 YEARS

PROTECTING CHILDREN FROM POLIO

As we look ahead to World Polio Day on 24 October, we remember our charity's roots. Action was founded in 1952 by Duncan Guthrie in his quest to find a cure for polio. Early research funded by the charity helped to develop the first UK polio vaccines, which have kept millions of children safe ever since.

Duncan Guthrie pictured in 1959 with children being treated at an orthopaedic hospital for the after-effects of polio.

PERRAULT SYNDROME PROGRESS

Action has helped to improve diagnosis of Perrault syndrome, a rare condition which causes severe hearing loss and infertility.

Perrault syndrome is linked to hearing loss, and sometimes accompanied by learning difficulties, developmental delay and nerve damage. Sadly, girls also experience reproductive problems, which may result in them being unable to have biological children of their own. While the condition was first identified in the 1950s, its underlying causes were still not well understood at the turn of the millennium, hampering both diagnosis and treatment.

As new laboratory techniques became available, researchers began to make progress exploring some of the genetic changes that can lead to Perrault syndrome. Professor Bill Newman and his team at Saint Mary's Hospital, which is part of Manchester University NHS Foundation Trust (MFT), began to identify genes that were linked to it.

> **"Our findings are already helping to improve genetic testing for children with hearing loss around the world"**
>
> Professor Bill Newman

In 2016, Action Medical Research, assisted by a grant from The Peter Stebbings Memorial Charity, awarded funding to help continue this important work – and by 2021, the team had identified four new genes which are linked to the condition, as well as different genetic changes within these genes. They also found a common mechanism showing how these changes affect and damage cells.

This work has informed new approaches to genetic testing and further research into treatments. It is already having clinical impact.

Genetic testing helps families to get diagnosed earlier and to access the right care and support sooner. For example, it can inform decisions about cochlear implants to restore hearing and allow early hormone replacement therapy for girls, or the choice to preserve their eggs to give them reproduction options later in life.

HELPING TO GIVE FAMILIES ANSWERS

Sisters Sabrina, now 22, and Alisha, age 10, both had hearing loss from an early age but it wasn't until they were seen by Professor Bill Newman's team that they were diagnosed with Perrault syndrome.

By this time, Sabrina was 16 and her family had become concerned when she appeared to have not gone through puberty.

The girls were first tested to see if there were any changes in genes already known to cause the condition – there were not, but changes in another gene were discovered. Since then, two other families have been found to have changes in this same gene – meaning the team could be confident they had a definite explanation for the sisters' health problems.

> **"Without this research we still wouldn't have known what was going on"**

Their father Ibrahim said: "It's still very hard, with the condition being so rare, making it hard to get the right support. But it's helped us to understand more and for Alisha there are more benefits, now that we have the history of what's happened with her sister and know why. If by taking part in the research we can help play a small part in helping other people in the future, then we are very happy to be part of that."

"We hope that Oscar and Isla are pioneers for a new treatment that will improve the lives of so many"

LIFE-CHANGING RESEARCH INTO PEANUT ALLERGY

Peanut allergy can cause life-threatening reactions. Action is funding a study into oral immunotherapy. For siblings Oscar and Isla, taking part in this trial, the results have been life changing.

Peanut allergy affects at least one in 50 children in the UK, caused when the immune system reacts abnormally to the protein found in the nuts.

Oscar and Isla knew from a young age to be very careful – with Isla having been hospitalised following an especially scary reaction after eating a peanut butter sandwich.

"Birthday cake at parties was never an option," says Oscar.

The siblings have taken part in research led by Dr Sharanya Nagendran of Imperial College London.

It involves introducing very small, increasing amounts of boiled peanuts under medical supervision, with the aim of desensitising the child's immune system.

"Two years on, they don't have to worry about eating food containing traces of peanuts," says mum Kirsty. "It really has changed their lives."

RESEARCH UPDATES

BABIES

USING AI TO HELP PREMATURE BABIES

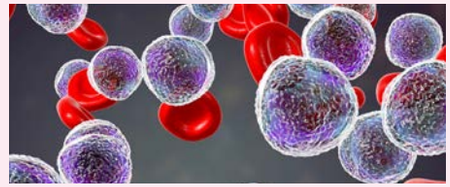
Sadly, more than a third of babies born extremely early, before 32 weeks of pregnancy, develop a serious lung condition or lose their lives within their first few weeks.

New Action Research Training Fellow, Dr T'ng Chang Kwok of the University of Nottingham is using artificial intelligence (AI) to identify which premature babies are at greatest risk.

His team will analyse routinely recorded data within the electronic medical records of many thousands of very preterm babies. The aim is to develop a new tool that can accurately recognise complex patterns within these data and guide personalised treatment decisions for these very vulnerable babies.

YOU MADE IT HAPPEN

TRIAL FOR NEW LEUKAEMIA TREATMENT



With your support, researchers have moved a step closer to developing a new treatment option for T-cell acute lymphoblastic leukaemia. This fast-developing type of cancer is diagnosed in around 100 children and young people each year in the UK.

With Action funding, Dr Frederik van Delft and his team at Newcastle University has been investigating whether combining two existing drugs could offer a new treatment.

Excitingly, based on the success of this research, and similar good results from other research groups, this new drug combination has now been included in an international trial due to start in the UK this year.

We are also very grateful to the Team Lewis Trust and other charitable trusts who helped us support this project.

Read more about research funded with your support at action.org.uk/research

PEDALLING TO PARIS FOR LITTLE LIVES

After a two-year pause due to the pandemic, our London to Paris bike ride made a triumphant return this summer!

Coinciding with the final week of the Tour de France, we had not one but two groups take on the four-day challenge, each following a different route. More than 100 riders joined our annual Action London to Paris trip, while another 50 took part in Raptör, our bike ride for folk from the insurance industry.

Together they are set to have raised almost £250,000 to help fund research to save and change children's lives.

Among those taking part in the ride was children's doctor Peter Bale, who said: "It's been a pleasure to ride from London to Paris with Action Medical Research – amazing charity, doing great things and as a children's doctor we absolutely need the research to make things better for babies and children moving forwards."

Fivos Valagiannopoulos, another rider, said he was very proud to have cycled for Action and for his nephew who is battling leukaemia.

Riders covered 300 miles and the final day saw them joined by cycling legend Sean Yates. They literally stopped the traffic to reach the Eiffel Tower together and complete their epic adventure!

Registration is now open for our 2023 event.

Scan the QR code to find out more about Action London to Paris 2023 or visit action.org.uk/L2P

EVENTS DIARY 2023

RUNNING

23 April TCS London Marathon

BIKE RIDES

2 April RIDE Wessex Downs

7 May RIDE Castle, Kent

21 May RIDE Suffolk Sunrise

4 June RIDE Vyking, York

11 June RIDE Davina's Big Sussex Bike Ride

19-23 July Action London to Paris

For places in more bike rides email **events@action.org.uk**

RACE THE SUN

10 June Brecon Beacons

8 July Coniston, Lake District

9 September Keswick, Lake District

23 September Snowdon NEW

We have events to suit all and would love you to join us! Visit action.org.uk/events

A SPECIAL EVENT

Liberty Specialty Markets generously hosted a reception for Action at their Walkie Talkie building offices in London. Around 80 guests were able to meet researchers whose work is currently being funded. They also heard a very moving speech from Sally (pictured), mother of Danny who has Hunter syndrome.

CORPORATE SUPPORT

We're delighted to have been chosen as a charity of the year for specialty (re)insurance group **Chaucer**. They have supported us with a £40,000 donation. Thank you also to **Touchstone Underwriting**, who raised more than £2,500 at their charity golf day in July.

CYCLESPORT RETURNS

We're already wheely excited about our Champions of CycleSport gala dinner this November! The event is supported by **Garmin**, **BDO** and **Lucky Saint**, with a guest list that boasts a host of cycling stars. Find out more at **action.org.uk/champs**

SUPPORTER STORY

RARING TO ROW

Following the success of team Elijah's Star, a new crew have taken up the gauntlet to row across the Atlantic Ocean this December. Team Raring to Row will take on the 3,000-mile Talisker Whisky Atlantic Challenge, raising funds to support our rare disease research.

Kevin Watkins, Adam Siggs and Gemma Best are training hard, motivated by the tragic loss of two of Gemma's siblings. Her sisters Jade and Jordan lived very short lives full of love and care but both sadly passed away very young due to a rare neurodegenerative condition.

Gemma will inspire us all when they set off on their big adventure, as she's also set to become the first female rower to cross the Atlantic twice.
Find out more at **raringtorow.com**

Congratulations to Nikki and her gorgeous family

SUPERDRAW WINNER!

Nikki from Brentwood was the lucky winner of our 70th Anniversary Superdraw, winning £500 and a luxury hamper. Nikki was on her way to her hen weekend when she received the good news, making the win extra special!

"I support Action because I believe the work they do makes a huge difference to children. As a mother of three myself, I will always do my small part to help where I can. The prizes are honestly an added bonus and I was so grateful," she says.

Want to be lucky like Nikki? You could win £1,000 in our Autumn Superdraw. Buy your chances today and also be entered into our Quick Reply Draw to WIN £50 Marks and Spencer vouchers.

Enter by 4 November. To find out more scan the QR code or visit **action.org.uk/superdraw**

WORD SEARCH

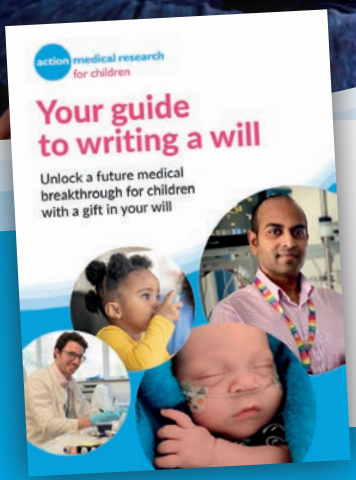
Complete the word search and let us know which word is missing. Send us your answer for a chance to win a £15 National Book Token.

A	N	G	U	Y	G	R	E	L	L	A	T	U	N	A	E	P
C	Y	C	L	E	S	P	O	R	T	N	H	Z	N	P	G	A
T	E	R	J	M	O	T	S	E	E	G	D	F	F	X	N	D
I	E	F	H	H	E	Y	C	M	Y	E	Z	C	S	H	I	D
O	X	R	N	T	Z	O	T	L	U	A	R	R	E	P	W	Y
N	I	Q	S	D	I	A	G	N	O	S	I	S	I	A	O	C
G	T	T	U	B	E	Z	S	P	A	O	U	N	B	B	R	X
S	S	S	E	R	G	O	R	P	I	W	W	O	A	F	C	M
H	A	S	T	I	F	Z	I	Y	O	D	X	E	B	M	Q	Y
R	A	B	S	Q	U	A	E	I	P	C	D	L	N	R	Y	F

1. Action
2. Paddy
3. Hope
4. Treatment
5. Perrault
6. Progress
7. Diagnosis
8. Babies
9. Peanut allergy
10. CycleSport
11. Rowing
12. Superdraw

Please send your answer to **editor@action.org.uk.** Entrants must be 16 years or over. Terms and conditions apply, for details visit **action.org.uk/wordsearch**

Unlock a future medical breakthrough for children with a gift in your will



Find out more and write your will for free – order your free guide today at

action.org.uk/willguide

The next big breakthrough in children's health could be possible thanks to a **gift in your will.**