



TOUCHING LIVES

HOW YOUR SUPPORT IS MAKING A DIFFERENCE

Giving hope to children like Digby, who has Duchenne muscular dystrophy

Meet the researcher tackling a rare brain condition

Progress made – helping to develop gene therapy for Rett syndrome

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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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or Touching Lives

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Registered charity: England and Wales
no. 208701; Scotland no. SC039284

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WELCOME

It's been a busy year so far, and we have lots of news to share with you! From the latest research funded, to progress being made – these pages are full of examples of how your support is helping to make a difference.

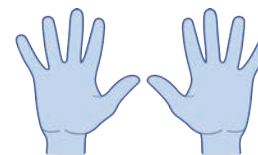
Medical research is a journey. Some researchers, like Dr Eva Ioannidou (who you'll meet on page 10) are just setting out. Aiming to unlock crucial answers that will take them forwards towards potential new treatments. In other cases, like the research into Rett syndrome that we funded over 15 years ago, life-changing medicines are finally within reach. These breakthroughs don't happen overnight. Small steps, over time, lead to big breakthroughs – and none of this happens without your support.

There's still more vital work to be done. More journeys to begin. More critical steps to take. That's why we hope you'll explore other ways to get involved. From exciting events, to seasonal shopping or even considering leaving a gift in your will, there are so many ways you can help us – and help children and families who are waiting for the next big breakthrough.



Thank you for all your support.

Clare, Editor
cairey@action.org.uk



10 NEW PROJECTS

funded so far
this year



£320

can fund a pioneering
research project for a day



YOU MAKE IT HAPPEN

10 NEW RESEARCH PROJECTS TO HELP BABIES, CHILDREN AND TEENS

So far this year, we've invested more than £2.3 million in new research, and it's all thanks to you.

With your ongoing support, exciting new studies are underway across a range of important areas.

Co-funded with Borne, two projects are tackling the causes of premature birth – including one to develop a much-needed treatment to prevent early labour that is linked to infection or inflammation in the baby's mother.

We're also supporting work to protect babies at risk of brain injury due to

lack of oxygen at birth. While a further study is using AI technology to improve pain monitoring in babies after surgery, helping to ensure the tiniest patients get the best possible care.

Other research will help children with cystic fibrosis, which you can read about on page 9.

Plus a pilot study is investigating if a widely available food supplement can help to relieve symptoms of polycystic ovary syndrome (PCOS) in teenagers.

None of this would be possible without your support.

RESEARCH UPDATE

RARE DISEASES, REAL PROGRESS



Funding for research into rare diseases is limited – but the impact these conditions can have on families is enormous. Which is why, with your support, we continue to fight for new treatments and cures.

Crucial progress is being made – as you'll see in this issue. Past research funded by Action has helped to pave the way for clinical trials happening right now in Duchenne muscular dystrophy on page 8, and Rett syndrome on pages 14-15.

We're currently funding 24 different research projects to help children with rare conditions. Thank you for helping to make life-changing work happen.

WATCH EVA'S STORY

HOPE ON THE HORIZON

Eva has a rare condition that causes progressive liver damage – her mum describes it as 'living with a ticking time bomb'. A transplant is the only current cure, but it's risky and donor livers are scarce.

We're funding research that offers real hope for a new treatment in Eva's lifetime. We spent some time with Eva and her family, making a very short film to show why this work is so important.

Scan the QR code to watch it now, or visit action.org.uk/videos





FIGHTING DUCHENNE FOR CHILDREN LIKE DIGBY

Boys like 10-year-old Digby face a relentless decline in their physical abilities due to this life-limiting condition. But with your support, Action is funding groundbreaking research that gives hope to families living with this devastating diagnosis.

Digby's journey with Duchenne muscular dystrophy began when he was just two years old. His mum, Lisa, had noticed he wasn't reaching key developmental milestones. He was late to walk, unsteady on his feet, and struggled to sit up without first rolling onto his tummy. "He was always falling, but at the time we thought it was just clumsiness," she recalls.

Then came some shocking news. Around this time, Digby's older sister, then 11, had been experiencing ongoing muscle pain and fatigue.

After many tests, she was found to carry the gene for Duchenne. Although Duchenne typically only affects boys, some female carriers can show milder symptoms too.

"Time with Digby is precious, so we are creating memories for him to help us keep going when the condition gets worse"

All the family was then tested – and Digby, the youngest, was diagnosed with Duchenne muscular dystrophy.

Boys with Duchenne do not produce enough, or any, dystrophin. This is essential for healthy muscles. Without it, muscles weaken and waste away. Over time, this leads to severe disability and, sadly, paralysis.

It was a devastating shock, made even more frightening by Lisa's knowledge of the condition. "I'd had a cousin on my father's side who'd had Duchenne and tragically passed away at 21. I knew straight away what it was – it was absolutely terrifying."

The diagnosis has had a profound impact on the family. Lisa gave up work to care for Digby full-time. They've also renovated their home to accommodate his wheelchair and medical equipment.

But there has been a glimmer of hope. Just two months after his diagnosis, Digby was accepted onto a medical trial. While this new treatment isn't life-extending, it aims to slow down the progression of the disease. Lisa believes it has made a difference to his mobility and quality of life.

In the UK, around
100 boys
are born with
Duchenne muscular
dystrophy each year



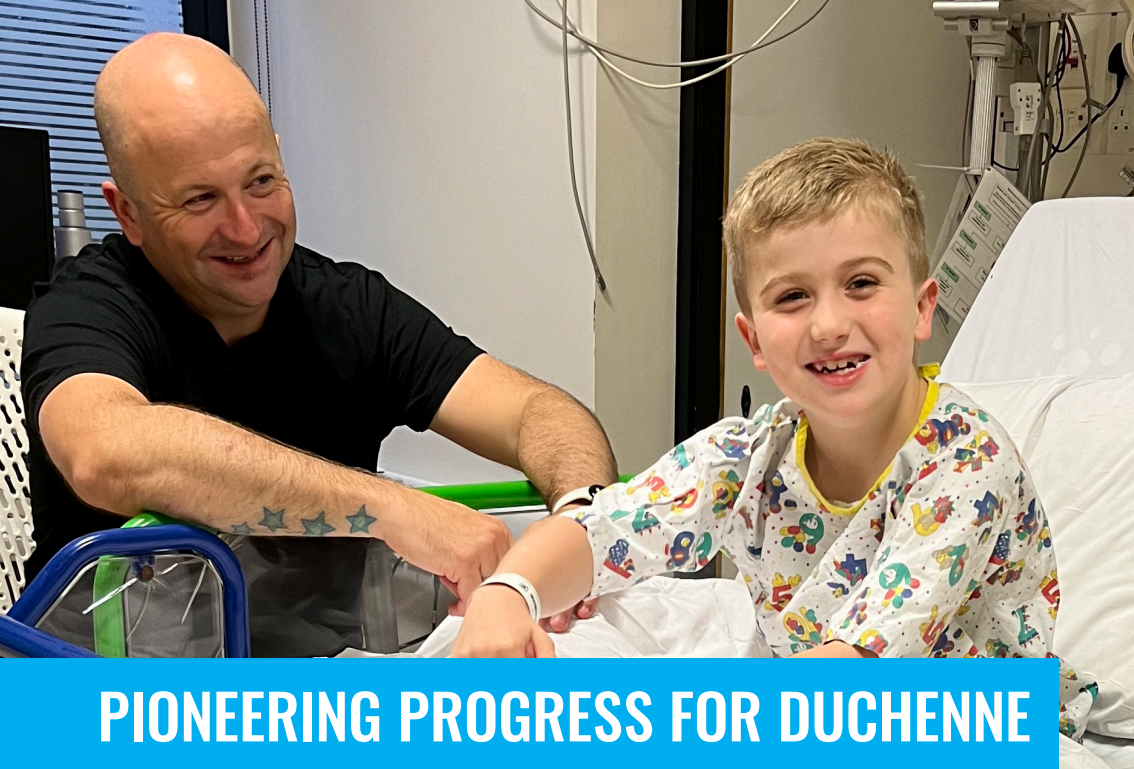
"It's a huge commitment, an infusion every week, but Digby is an absolute trooper," says Lisa. "While we take each day at a time, we're incredibly grateful that he's still got his mobility at this point and leads as normal a life as possible. But we know this can change overnight. The feeling that our son is on 'borrowed time' is heartbreaking."

"We are so grateful to the research teams investigating new ways to treat Duchenne," says Lisa. "Research gives families like ours hope."

Thanks to your support, we continue to fund vital research to help children like Digby – read more overleaf.



>> Digby was diagnosed with Duchenne when he was a toddler



PIONEERING PROGRESS FOR DUCHENNE

Action helped to lay the foundations for gene therapy breakthroughs that are now helping children like Digby. Back in 1997, we funded early research by Professor George Dickson to test gene therapy for Duchenne muscular dystrophy in the lab. This vital proof-of-concept work helped spark the clinical trials that are now offering hope today.

“It all has to start somewhere and I think it’s a tremendous legacy for the charity to be proud of”

Professor George Dickson

NEW RESEARCH NOW

Supercharging gene therapy

At UCL Great Ormond Street Institute of Child Health, Professor Francesco Muntoni is testing whether combining gene therapy with an existing medicine could make treatments even more effective. This work is co-funded with LifeArc.



Protecting fragile bones

At the Royal Hospital for Children Glasgow, Dr Sarah McCarrison is developing a tool to predict spinal fractures in boys with Duchenne. The aim is to personalise treatment for those at greatest risk, while sparing others from the potential side effects of taking unnecessary medicines.

JOINING FORCES FOR CHILDREN WITH CYSTIC FIBROSIS

With your support, we’re delighted to be continuing our joint efforts with Cystic Fibrosis Trust, striving to drive change and improve the quality of life for babies and children.

Though there have been improvements in medicines and treatments in recent years, cystic fibrosis (CF) remains an incurable and life-limiting condition.

Children with CF experience a build-up of thick sticky mucus in their lungs, digestive system and other organs. This can make it hard for them to breathe and digest food and leaves them very vulnerable to serious chest infections.

Two co-funded research projects are underway.

Dr Rebecca Dobra’s team at Imperial College London is working with young children who have an inconclusive diagnosis. They aim to find ways to predict who will go on to develop CF or a related condition. This could reduce anxiety for families, improve early care and ensure timely intervention for those at risk, while avoiding unnecessary tests for healthy children.

And Professor Guy Moss and his team at UCL aim to develop effective new drug treatments to help

Cystic fibrosis affects around

3,800

babies and children in the UK

more children live longer, healthier lives. While groundbreaking new medicines (called CFTR modulators) are life-changing for many, they don’t work for all, leaving around one in 10 children in desperate need of other options.



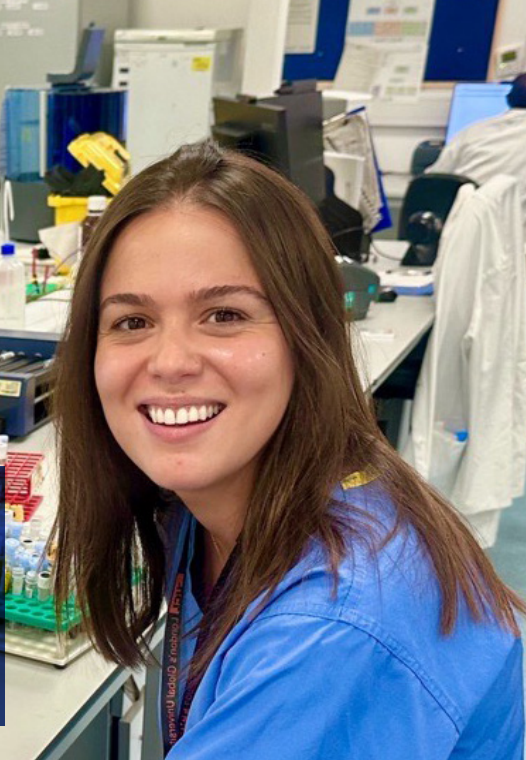
WORKING TOGETHER

Since 2014, Action and Cystic Fibrosis Trust have worked together to invest more than £1.2m into much-needed research. Together, seven projects have been funded, helping to boost understanding, improve treatments and diagnosis, and protect children like seven-year-old Sophia, pictured.

MEET THE RESEARCHER

DR EVA IOANNIDOU

Our Research Training Fellowship scheme helps to train and develop talented doctors early on in their research careers. Dr Eva Ioannidou is one of our newest Fellows. She is investigating the underlying causes of a very rare and devastating brain condition called Rasmussen's encephalitis.



Q When and why did you decide to pursue a career in medicine?

A The honest answer is that medicine has always felt like a part of me. I can't recall a time when I considered any other path. For me, there was only ever medicine. What drew me in, and still keeps me grounded, is the mix of science, ethics and real human connection. Medicine is problem solving with purpose – using scientific innovation to make a difference in someone's life. I've never regretted choosing this path.

“Medicine is problem solving with purpose – using scientific innovation to make a difference”

Q At what point did you decide to focus on paediatrics and why?

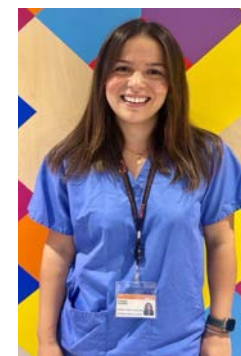
A Seven years ago, during a summer elective at Great Ormond Street Hospital, I attended a complex epilepsy clinic led by Dr Marios Kaliakatsos. Seeing the complexity of the cases, the almost supernatural resilience of the children and their families, and the thoughtful, deeply human approach of Dr Kaliakatsos inspired me. I knew I wanted to focus on paediatric neurology. Now, as I begin a PhD in the same department with him as a supervisor, it feels like things have come full circle.

Q What inspired you to study Rasmussen's encephalitis?

A My interest in this rare disease began with the stories of previously healthy children whose lives changed overnight. Children who were thriving and then suddenly facing seizures, neurological decline, and the prospect of life-changing brain surgery. Rasmussen's encephalitis is an example of a disease where epilepsy and neuroimmunology meet, yet these fields have often remained separate. I was struck by that gap and driven by a need to bring them together in a meaningful way.

Q What do you hope your research will achieve?

A I hope it will help us to better understand the immune mechanisms underlying Rasmussen's encephalitis and how they contribute to seizure activity. By identifying early biomarkers, I aim to support earlier diagnosis and pave the way for less invasive treatments. More than anything, I hope this work will offer children a chance at a different trajectory – one that doesn't end in life-altering surgery.



I hope to create a support group for families, offering some clarity and connection in the uncertainty they face. I also see this as part of a broader journey that will open doors to understanding other epilepsies, where answers are also needed.

“

In rare disease research, every step forward is hard-won. I hope my work will offer children a chance at a different trajectory”

Q What does Action funding mean to you?

A In rare disease research, every step forward is hard-won. Progress depends not only on scientific drive, but on belief. Action's funding is a powerful vote of confidence in a field that's often overlooked. It allows me to train with a world-class team and focus entirely on the research, without compromise. It means I can build something that reaches beyond the lab, engaging with families and helping to shape a future with fewer unknowns.

FIND OUT MORE

You can read more about Dr Eva Ioannidou's research on our website. Scan the QR code or visit action.org.uk/rasmussens



Dr Ioannidou is an Action Medical Research and British Paediatric Neurology Association Research Training Fellow

PROTECTING SLEEP AND BRAIN DEVELOPMENT IN PREMATURE BABIES

An Action Medical Research study has shown the importance of sleep cycles on brain development for babies born prematurely.


In the second half of pregnancy, a baby's brain undergoes rapid development and at the same time distinct sleep cycles emerge. But babies born too soon are cared for in an environment that's very different to the womb.

Frequent medical procedures, bright lights and loud noises can interrupt natural sleep cycles. This could have a significant impact on healthy brain development.

At Cambridge University Hospitals NHS Foundation Trust, researchers developed a non-invasive imaging system to study sleep cycles in babies and track blood flow in the brain. This study was the first to demonstrate wearable technology for cot-side neuroimaging in newborn infants.

Led by Professor Topun Austin, the team found there were significant differences in functional brain connectivity between neonatal sleep states. This has now led to a follow-on study, with the hope of developing a clinical system to promote healthy sleep cycling in these vulnerable babies.

Professor Austin says: "Sleep, although something all babies spend a lot of time doing, is very under-researched and being able to demonstrate a connection between sleep states and brain development provides an important evidence base to focus on protecting sleep in the newborn infant."



"This could have a major beneficial impact on the way infants are cared for in neonatal units"

Professor Topun Austin



GENE THERAPY GIVES HOPE TO GIRLS WITH RETT SYNDROME

» Professor Sir Adrian Bird

Rett syndrome is a rare and severe neurological condition. There is currently no treatment or cure, but clinical trials are beginning to offer families new hope – and early Action funding helped pave the way to the exciting progress now being made.

Imagine watching your baby develop normally, only to see those precious early skills and milestones slowly slip away. This is the heartbreaking reality for families affected by Rett syndrome.

It's caused by a fault in a gene that's critical for normal brain functioning. Symptoms start subtly, usually around six to 18 months after birth, and worsen over time, leading to severe physical and learning disability.

In 2009, Action Medical Research awarded over £180,000 to support the search for new treatments for this devastating condition.

“Action played a part in developing the story towards clinical trials, for which the charity deserves significant credit”

Professor Sir Adrian Bird

Professor Sir Adrian Bird and his team at the Wellcome Trust Centre, at the University of Edinburgh, had previously generated early evidence that symptoms of Rett syndrome might be reversed if healthy gene function could be restored.

With Action funding, they took the next step. They used viral vectors – harmless viruses modified to carry healthy genes – to deliver a corrected version of the faulty gene into brain cells, and showed that this could successfully reverse Rett symptoms in the laboratory.

In collaboration with researchers in the US, they then showed that injection of their vector into the bloodstream could give more effective results.

This work, published in 2013, generated initial proof of principle that gene therapy held promise to treat, and potentially even cure, Rett syndrome. It also laid the foundations for further advances, including a ‘mini-gene’ approach that was also effective and easier to deliver.

Excitingly, the first clinical trials of gene therapies in patients were launched in 2023 in the US and Canada and have since started recruitment in the UK. One of these has adopted the mini-gene approach pioneered by the Edinburgh team. Initial trial results show promise, bringing the possibility of life-changing treatment closer than ever before.

This research was supported by a generous grant from The R S Macdonald Charitable Trust.

THANK YOU!

Thanks to supporters like you, research backed by Action helped to lay the foundations for new treatments that could change the lives of families affected by Rett syndrome.



For families like five-year-old Mollie's, gene therapy for Rett syndrome can't come soon enough.

“We're so close to getting to where we need to be, but Mollie's getting older by the day,” says her dad, Mark. “Even though she's only five, you can see how much this has taken away from her. I've never heard Mollie say daddy, never seen her crawl or walk, never had her put her arms around me. Skills she did have, like the ability to sit and play with toys, have gone.”

“We're blessed that Mollie has a sister, Lily, who is three. But she wants to play and doesn't understand why Mollie doesn't move or speak. It's heartbreaking to watch one child live the life the other should have too.”

“In simple terms, gene therapy could set Mollie free”

Mollie's dad, Mark

“For us, seeing Mollie where she is now, just being able to regain basic skills would be huge. We are so excited and buoyed by all the clinical advancements. Many families have suffered for much, much longer than we have. But change is now within reach,” says Mark.



PREDICTING AND PREVENTING ASTHMA IN YOUNG CHILDREN

With your support, researchers have developed a new approach to treat wheezing in pre-school children, which could help to prevent asthma and long-term lung damage.

Pre-school age children account for three quarters of all childhood hospital admissions for acute wheezing – and for some, it can be the start of lifelong breathing problems.

Researchers at Imperial College London and the Royal Brompton Hospital aimed to find a way to predict which young children who'd suffered attacks of wheezing were most at risk of developing asthma by school age.

Led by Professor Sejal Saglani, they followed affected children and combined clinical data with laboratory experiments and computer modelling.

“This funding was critical in making an advance”

Professor Sejal Saglani

This found two main risk groups. The first have allergies and high numbers of blood cells called eosinophils. The second group are not allergic but have bacterial infections in their lungs.

These findings mean doctors can now use simple tests, like skin prick allergy tests and finger prick blood samples, to help predict asthma risk.

Researchers also identified three common bacteria linked to future asthma, and are now testing whether nose and throat swabs, like COVID tests, could be used to spot these early and treat with antibiotics to prevent long-term harm.

OUR LATEST RESEARCH

FIGHTING SIGHT LOSS IN CHILDREN

Dominant optic atrophy is an inherited cause of blindness that causes progressive and irreversible sight loss from early childhood. It's caused by gradual degeneration of the optic nerve, which carries visual signals from the eyes to the brain.

With Action funding, researchers at the UCL Institute of Ophthalmology hope to identify key genes involved in the death of optic nerve cells. This could help to uncover new treatment approaches to slow down or stop sight loss in children.

NEW STUDY TO HELP AUTISTIC CHILDREN

Many autistic children find social interactions challenging, finding it difficult to understand others or to express their own emotions. With your support, researchers are studying brain activity during social behaviour in children with autism.

The team, at King's College London, are combining virtual reality with advanced MRI scanning to create an interactive, engaging experience that's fun and feasible. They hope to uncover differences in social brain processing. This could lead to more targeted support to help children to navigate the social world.

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



A BREATH TEST FOR CRITICALLY ILL CHILDREN

Every year, millions of children worldwide are hospitalised with chest infections. In the UK alone, around 6,000 are admitted to intensive care. Although many infections are caused by viruses, antibiotics are often given first because current tests for chest infections are slow and unreliable.

Researchers at Imperial College London are developing a rapid breath test to accurately detect bacterial infections. This would be quick, safe



and painless, and could be repeated to track a patient's progress. It could transform care, reduce unnecessary antibiotic use, and ensure critically ill children receive the most appropriate treatment at the right time.

BIKE, HIKE AND PADDLE RACE THE SUN 2026



Join us on one of our triathlon-style team challenges – raising vital funds and shining a light on children's medical research.

Our Race The Sun events see participants bike, hike and paddle against the clock in some of the UK's most stunning landscapes – from the

beautiful Lake District, to the rugged Jurassic Coast.

We're now looking for contenders to take on one of five epic events planned for next year. These include a new location. The dramatic clifftops of the Seven Sisters, on England's south coast, will provide

a breathtaking backdrop, with stunning sea views, quiet lanes, meandering waterways and a few cheeky climbs to keep things interesting!

You can compete as either a team of two or four. The event works brilliantly for groups of family or friends, or as a company team builder.

Whether you want the challenge of setting a fast time, or just an exhilarating adventure for a great cause, Race the Sun is a fundraising challenge not to be missed!



To find out more and sign up, scan the QR code or visit action.org.uk/sun

EVENTS DIARY 2026

RUNNING

26 April TCS London Marathon

CYCLING

10 May RIDE Castle, Kent

17 May RIDE Suffolk Sunrise

TBC May GARMIN RIDE OUT

(sign up to join waiting list)

7 June RIDE The Dales

28 June RIDE Tommy Godwin

Challenge **NEW**

5 July RIDE Wessex Downs

5 July Maratona dles Dolomites

30 August RIDE Essex

RACE THE SUN

25 April South Downs Way (road) **NEW**

13 June Lake District Off-Road

27 June Jurassic Coast Off-Road

11 July Cheddar Gorge (road)

5 Sept Lake District (road)

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



DAVINA DAZZLES IN ESSEX

Dine With Davina Essex, held at Hylands House in Chelmsford in June, raised an amazing £41,000. A fantastic result for a brand-new event. The day saw 160 ladies come together for a fabulous afternoon of fun and fundraising with Davina and MCs Nina and Saski (pictured). We'll be doing it all again at the long-running Southampton edition in November.

SUPPORTER STORY

MARATHON MAGIC

A huge thank you to the 70 people who ran for Action in this year's TCS London Marathon. Together, these wonderful folk raised more than £200,000.

Among them was Helen West, who had first been due to run in the 2020 event, cancelled due to COVID-19. Helen's connection to Action deepened in 2022 after the heartbreaking loss of her first daughter, Lilly. She was born at 29 weeks and weighed 2lb 12 ounces, but due to an infection and underdeveloped lungs lived for just six hours.

Helen raised over £5,500. She says: "The day was tough, emotionally and physically, but it was amazing. I'm delighted that I managed to raise so much money for research to help families in the future."

RAPTÖR RIDE RECORD!

Our bike ride for the insurance industry saw 85 cyclists pedal from Sussex to Roubaix via historic French battlefields. Now in its fifth year, the event goes from strength to strength and this year raised more than ever – an amazing £400,000. Thank you to **Ki, Brit** and others in the sector for all their support.

HEALTHCARE HEROES

We've once again had amazing support from **Cencora Alliance Healthcare**. This year saw the company take part in and sponsor our new **Race the Sun Cheddar Gorge** event, where staff raised more than £15,000. Whilst most took on the whole event, others showed solidarity by completing the walking section with their colleagues.



To find out more about running for Action visit action.org.uk/run

FUNDRAISING NEWS

RIDE CATALYST

Catalyst, our newest industry bike ride for those working in the biotech and pharma sector, saw 24 cyclists head from London to Bruges. We were kindly supported by **Appleyard Lees LLP** as lead partner for this first event, as well as **EBD Group** and industry networking group, **BiotechBikers**. Covering 250 miles across three days, riders contended with both a heatwave and strong headwinds, as well as some iconic Flanders cobbles climbs!



HEAGE GARDEN PARTY TRIUMPH

For almost four decades, the Heage Committee, in Derbyshire, have held an annual Garden Party for Action. Hosted by Jean and Glyn Fardoe, each year sees the whole community come together to enjoy a variety of activities, stalls, food and live music.

This year's event raised almost £6,000, taking the total that's now been raised over all the years to more than £100,000! This is an outstanding achievement for a small village and we are so grateful to all involved.

NIGHT OF CHAMPIONS

Top names in cycling will again come together for Action at our Champions of CycleSport Dinner in November. This gala evening offers cycling fans a unique chance to dine alongside legends such as Stephen Roche, Sean Yates, Sean Kelly, Alex Dowsett, Dani Rowe MBE, and Pfeiffer Georgi (pictured). We're incredibly grateful for the generous support of sponsors Garmin, BDO and Erdinger Alkoholfrei, with additional backing from Vires Velo.



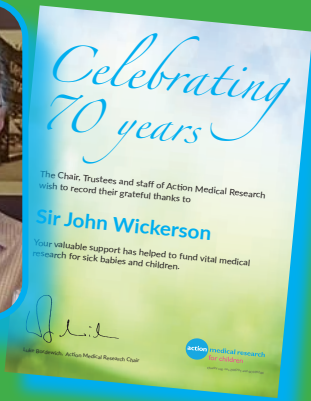
Find out more at action.org.uk/champs

THANK YOU SIR JOHN

At our recent AGM, we honoured the work of Sir John Wickerson LLB, who has helped the charity for more than 70 years.

A former President of the Law Society, Sir John has been involved with the work of Action since its earliest days, back in the 1950s, when he'd just started his legal career. During his training, he worked under the supervision of Tony Dumont, the charity's then solicitor and a friend of our founder Duncan Guthrie.

Sir John eventually took over as the charity's solicitor, and in this role negotiated the very successful Paddington Bear contract. He was also a member of our Finance Committee and then its chair.



In his retirement, Sir John served as a charity trustee from 2006 to 2014, and he remains an active member still today.

Sir John says: "I am delighted at the work that has been done over the years and impressed that it continues. I would like to thank all those who are involved for the work that they have done."

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.

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

- 1 Digby
- 2 Duchenne
- 3 Cystic fibrosis
- 4 Encephalitis
- 5 Neonatal care
- 6 Rett syndrome
- 7 Gene therapy
- 8 Preventing
- 9 Asthma
- 10 Running
- 11 Garden party
- 12 Christmas

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

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NEXT



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action.org.uk/superdraw

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Draw date: 5 December 2025

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"We used a local solicitor who was highly professional, and this gave us reassurance throughout the process. We would recommend this service to everyone. Having witnessed the profound impact of rare diseases first-hand, we chose to leave a gift to Action Medical Research so that their vital work could continue to benefit future generations."

**Lynne and Andy,
Action supporters**

Please note that all will-writing partners will be acting only in your interests and on your instructions.

We also recommend that you get independent legal advice.

Action Medical Research is a registered charity: England and Wales no. 208701; Scotland no. SC039284