TOUCHING LIVES

The Action Medical Research newsletter | Spring/Summer 2025

HOW YOUR SUPPORT IS MAKING A DIFFERENCE

Preventing premature birth – helping families like Elsie's in the future

Research for children during the COVID-19 pandemic

Fighting rare diseases – progress made and new research funded

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IN THIS ISSUE

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

Please send all communications to:
cairey@action.org.uk
or Touching Lives
Action Medical Research
5th Floor
167-169 Great Portland Street
London W1W 5PF

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

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WELCOME

In spring 2020, the world was gripped by the medical emergency of COVID-19. Schools and shops were closed. Many everyday activities curtailed. And medical research was suddenly very much in the spotlight. We couldn’t have navigated through the pandemic without it.

In this issue, we take a look back at how Action, with the generous backing of so many supporters like you, quickly adapted to fund urgently needed COVID-related work – and how, as this research now concludes, it has helped to make a difference.

We also have lots of new research to share, including projects to treat some devastating rare diseases. Our cover story however, featuring Elsie and her family, highlights an area that sadly isn’t uncommon. Premature birth is the single biggest cause of neonatal death and illness in the UK. With your help, we hope to change this.

As we head into warmer months, there are many great ways you can support us through our events. Or there’s our fantastic Summer Superdraw – which could be a win-win all round, if you’re lucky!

However you choose to support us, thank you so much.

Clare, Editor
cairey@action.org.uk

AROUND 55,000
babies are born too soon each year in the UK

3.5 MILLION
people are affected by a rare disease in the UK

YOU MAKE IT HAPPEN

NEW RESEARCH PROJECTS FUNDED

Thanks to your support, we were able to fund 14 new medical research projects in 2024. Already underway, this work will help babies and children affected by a range of conditions.

It includes research to improve neonatal care and to prevent brain injury in babies born prematurely. There are also studies that aim to help children with epilepsy and autism. Plus research to protect children who are already critically ill in hospital against lung and chest infections.

NEWS

RARE DISEASE CAMPAIGN MILESTONE

Our rare disease research campaign has now raised more than £1.2 million. This amazing total is making vital research possible – work to develop potential treatments and cures for some of the most devastating conditions that can affect children.

This includes new research to help boys with Duchenne muscular dystrophy, which you can read about on page 7. Plus work to treat two rare neurodegenerative diseases, which you can find out about on page 15.

Thank you so much for your support.

Danny, who has Hunter syndrome

ELSIE'S FIGHT FOR LIFE

Elsie weighed just 600 grams when she was born extremely prematurely. Her parents, Debi and Chris, were warned that her chances of survival were 1,000 to one.

As her sister had previously given birth prematurely, Elsie's mum Debi was understandably concerned that she too might be at risk. These fears intensified when around 23 weeks into her pregnancy, Debi began leaking amniotic fluid. Within a few days, it was confirmed that her waters had broken, and she was found to have an infection. "Whether the infection caused the rupture or vice versa remains unclear, but it became evident that our baby needed to be delivered immediately to prevent further complications," recalls husband Chris.

> "A day that brings most parents their greatest joy was the start of our most challenging journey"

Elsie was born at 23 weeks and five days, with the medical team offering little hope of survival. But despite the grim odds, tiny Elsie proved resilient, defying expectations from the start. She would spend seven months in neonatal intensive care, each day fraught with uncertainty.

Being born so early meant Elsie faced many serious setbacks. At just 10 days old she needed an emergency operation after a feeding tube perforated her stomach. As her lungs were so underdeveloped she battled severe chronic lung disease. Life-saving steroids used to treat this then led to a thickening of the heart muscle. Elsie also endured retinopathy of prematurity, an eye condition that required surgery.

These complex conditions demanded round-the-clock care and constant adjustments to her treatment plan. "The NICU became our new world," says Chris. "Elsie was so fragile; we needed to be constantly vigilant. We practically lived at the hospital during this time – our lives were completely flipped upside down."

Thankfully, despite such a traumatic start, Elsie is now doing extremely well. She needed 24-hour oxygen support for the first two years of her life and was fed through a feeding tube, but she is now totally off oxygen and eating normally.

"It's not always easy, but every day she's getting bigger and stronger," Chris says. "We feel so lucky."

Research to prevent pre-term labour

It's thought that infection may be a trigger in around 40% of women who experience unexpected early labour. With your support, researchers based at University College London aim to develop a treatment to boost the body's natural defences in women who are known to be at higher risk of premature birth.

> "We hope this will ultimately lead to clinical trials in women at high risk of giving birth too soon"

Dr Ashley Boyle, pictured below

This could lead to a new treatment to reduce the risk of early labour, helping to save tiny lives and prevent the serious health issues that prematurity can cause.

This research is co-funded with Borne.

Elsie is now walking and talking, milestones that give her parents great joy after previously being told she would likely never crawl.

> **“Having come through this, it’s important to us to do anything we can do to prevent this happening to others in the future”**

But ensuring she stays well remains a daily challenge. Elsie is still highly vulnerable to viruses and infections. She has been re-admitted to hospital eight times. “We have to be really careful, as every virus can cause more damage to her already fragile lungs,” says Chris. She also has an adrenal gland insufficiency, which requires daily steroids and close monitoring.

“She is too vulnerable to go to toddler groups or childcare yet, so we have to be creative with how she can be around other children. But her resilience is undeniable, we’re so proud of her and she brings us so much joy,” smiles Chris.

Thinking about the prospect of preventing preterm birth, Chris says: “This means so much to us. Those months in hospital, watching the person you love most fight for their life, while you can do very little to help, not to mention the ongoing challenges that follow, make it a deeply painful experience. No family should have to face this.”

SAVING TINY LIVES

Premature birth is the single biggest cause of neonatal death in the UK. With your help, we can change this.

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

A NEW TREATMENT STRATEGY FOR DUCHENNE

With your support, researchers are investigating whether combining the latest gene therapies with existing medications could improve their effectiveness. This could help to unleash the full potential of these cutting-edge treatments.

Duchenne muscular dystrophy causes severe and progressive muscle weakness. It almost always affects boys and there is no cure. By their early teens, most will need to use a wheelchair – and sadly most only live into their 20s or 30s.

The condition is caused by a faulty gene which means the body doesn’t make enough, or any, of the protein dystrophin, which is essential for keeping muscles healthy.

New gene therapies aim to restore sufficient levels of dystrophin in muscle cells, reducing symptoms and slowing disease progression. But so far, their effectiveness has been limited.

Previous research has suggested that combining these therapies with an existing type of drug could help to further boost dystrophin production.

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

Now, with co-funding from Action and LifeArc, researchers will test this approach on patient cells. Led by Professor Francesco Muntoni at UCL Great Ormond Street Institute of Child Health, they will study various combinations to determine which works best.

> **“Currently, at least one in three boys with DMD could benefit from this treatment approach, but that number is likely to increase”**
>
> Professor Francesco Muntoni

WHY I SUPPORT ACTION

As Head of Marketing at Garmin, Jack Fairbrother was already aware of and working with Action as part of a longstanding charity partnership. But this relationship became much more personal when his son Lando was born two years ago.

JACK FAIRBROTHER

Q Can you tell us a bit more about how you first got to know about Action?

A Garmin was already supporting Action when I joined the company, primarily through cycling events, like the annual Garmin RIDE OUT in the New Forest, and by donating great prizes for some of the charity's big social events. But in 2022, the connection took on a whole new meaning for me personally. I am immensely proud that as part of my job we support this charity.

> **"We were told that our baby needed to be born right now"**

Q Tell us a bit more about your son Lando's arrival.

A At our 20-week scan, we were told that our baby had a serious heart defect and a kidney condition. A few weeks later, we were told that for him to have a chance of survival, he needed to be born immediately – as in right now. It was terrifying. Lando was born 11 weeks early, weighing 2lbs 4oz. We lived in neonatal intensive care for three months, while incredible doctors and nurses helped him grow stronger.

Q Can you tell us about the surgery Lando had?

A Lando needed to have emergency open heart surgery before he had even reached his due date. By the time he was 18 months old, he had spent five of those months living in intensive care. He had been through four major operations. No child should have to go through that, but our story is a positive one.

Q How is he doing now?

A He has more challenges to come and will need further surgery, but Lando's doing really well. If you met him, you would just see him as any other kid – but with a badass scar! He is fearless, cheeky and full of life.

> **"Without charities like Action, doing the work that they do, my son simply wouldn't be here"**

Q What does supporting Action mean to you now?

A Action's past research helped to develop ultrasound scanning in pregnancy, which saved Lando's life. It's supported premature babies and helped children with congenital heart conditions. When I say that the work Action does makes a difference, it comes from a place of truly understanding that it does. Without charities like Action, doing the work that they do, my son simply wouldn't be here.

Jack is now getting ready to run the London Marathon. We wish him the very best of luck!

CHAMPIONS OF CYCLESPORT

Jack also shared Lando's story at our latest Champions of CycleSport Dinner. Here more than 500 guests joined some of the world's greatest riders, both legends and current, to help raise more than £290,000.

We're hugely grateful to Garmin, BDO, Lifeplus and ERDINGER Alkoholfrei for their support of this event.

For details of the 2025 edition visit action.org.uk/champs

PROGRESS MADE
FOR CHILDREN LIKE PADDY

Your support has helped researchers to identify a shortlist of promising chemicals for developing a drug treatment for a very rare and severe type of epilepsy.

There are currently no effective treatments for *KCNT-1* related epilepsies. Children like Paddy, pictured, whose story has previously featured in *Touching Lives*, endure repeated seizures every day. They are often left with severe disabilities, and many lose their lives at a young age.

Dr Jonathan Lippiat and his team combined computer analysis with laboratory work. This meant they could screen millions of potential chemical compounds before carrying out tests on those predicted most likely to be effective.

Around 35 of the chemicals tested in the lab showed promise as a potential treatment. “Our key outcome is a toolkit of compounds, providing a range of starting points for developing treatments,” says Dr Lippiat.

These results show that a combined approach to drug discovery, saving both time and money, has huge potential – and is one that could be applied to many other rare diseases, which are often neglected by the pharmaceutical industry.

> **“This could ultimately lead to a safe and effective medicine to help control seizures and prevent brain damage, offering hope to children and families”**

Dr Jonathan Lippiat

FIGHTING COVID-19

RESEARCH TO HELP BABIES AND CHILDREN DURING THE PANDEMIC AND BEYOND

Action has always risen to the medical research challenges of the day – and in 2020 a new health crisis emerged. In response, we launched our COVID-19 children's research appeal and funded research in this crucial new area.

In spring 2020, medical research was thrown firmly into the spotlight – and relied upon to deliver the life-saving breakthroughs needed to control and treat the new virus that was causing COVID-19. But while the global research effort was unprecedented, there was a lack of work focusing on babies, children and young people, and the impact of the pandemic on them.

Our broad remit, funding research across a wide range of diseases and conditions, allowed us to respond very quickly.

With your support, during 2021 and 2022 Action invested more than £1.1 million in seven studies to investigate the impact of COVID-19 on pregnant women, babies and children, and to help young people suffering from the debilitating effects of long COVID.

These projects were designed to provide vital information that could have an immediate impact, as well as help to fight future pandemics. They enhanced our understanding of the virus, identified risk factors for severe disease, and supported the development of new guidance.

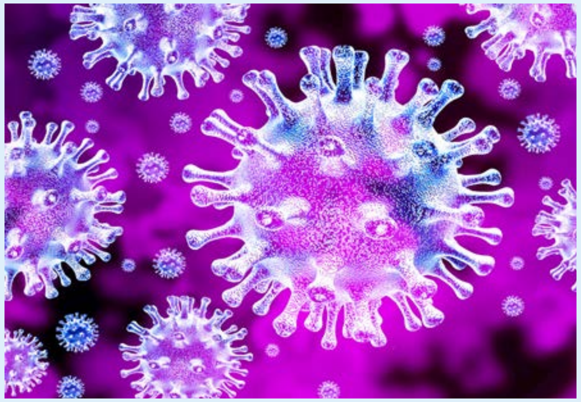
> **"Action funding was critical during an unprecedented time in medical science"**
>
> Professor Kirsty Le Doare

7 RESEARCH PROJECTS

FOR BABIES, CHILDREN AND YOUNG PEOPLE WERE FUNDED

This research has now helped to:

- Understand differences in immune responses between children and adults.
- Understand infection in pregnant women and their babies, including advice for vaccination.
- Identify risk factors and develop specialist tests to help prevent severe forms of COVID-19 that can, sadly, affect some children.
- Assess the impact of the pandemic on children with a newly-diagnosed brain tumour – with recommendations made for managing cancer services during any future periods of disruption.
- Identify young children who may be at risk of delayed social or emotional development following early-life exposure to COVID-19.
- Develop a new online treatment programme to tackle long COVID in young people.
- Improve understanding of how long COVID affects the brains of children with ongoing symptoms.



INVESTIGATING IMMUNE RESPONSE

Children with COVID-19 are usually much less likely than adults to become seriously ill – but in 2020, nobody knew why. Funding from Action helped to answer this key question.

Researchers, led by Dr Marko Nikolić, Dr Kerstin Meyer and Professor Sarah Teichmann, found that children have a much stronger immune response in the nose, helping to fight off the virus straight away, before it spreads to other body parts. They also have a weaker immune response in the bloodstream compared to adults – protecting them from immune-related damage to organs, as seen in severe disease.

> **"Our findings could be used to help identify which children and adults are at greater risk of severe illness"**
>
> Dr Marko Nikolić

This research, and follow-on work that the team has carried out since, has given crucial insights and also helped to identify potential avenues for preventing and treating severe cases of COVID-19.

HELPING MUMS AND THEIR BABIES

At a time of such great uncertainty, Action-funded research also led to much-needed guidance and reassurance for pregnant women.

Led by Professor Kirsty Le Doare, this work aimed to determine how much of a risk COVID-19 posed for pregnant women and their babies. It saw women screened at 10 different hospitals across England.

It found that the virus itself did not appear to be transferred to babies during pregnancy or breastfeeding. It also showed that antibodies against COVID-19 could pass from mother to baby, offering a level of protection against the virus to newborn babies, whether through vaccination or by natural infection.

These findings fed into guidance for pregnant women and new parents, published by the UK Health Security Agency and the NHS.

YOUR SUPPORT MAKES RESEARCH HAPPEN

We began when polio was a threat in the 1950s.

Almost 70 years later, we were still there to help children during the COVID-19 crisis.

We couldn't have done this without your help.

HELPING CHILDREN WITH LONG COVID

Action funding helped to develop online support for affected young people.

Long COVID can leave children with ongoing breathlessness and feelings of anxiety or low mood many months after their initial infection.

Dr Samatha Sonnappa and her team at the Royal Brompton Hospital in London set out to develop an online programme to help.

With input from several young people suffering from long COVID, they developed modules including breathing techniques and coping skills, along with bespoke videos.

"The grant from Action allowed us to bring together a team of specialists, alongside teenagers, to design and then deliver a new intervention. This simply would not have happened without this vital funding," says Dr Sonnappa.

RESEARCH UPDATES

IMPROVING PREGNANCY CARE FOR CHILDHOOD CANCER SURVIVORS

Thanks to advances in treatment, many children now survive a cancer diagnosis and eventually want to start families of their own. But it had been suspected that women who received a bone marrow transplant as a child or young adult were at greater risk of experiencing pregnancy complications.

With funding from Action and Borne, Dr Melanie Griffin and her team have worked to better understand this. Their study confirmed that those who had bone marrow transplants, particularly when combined with total body irradiation, were at higher risk than usual of giving birth very early and having smaller babies.

The team are now sharing their findings so that national guidelines can be updated and those affected can access more specialist support.

This project was generously supported by The James Tudor Foundation.

TACKLING TWO NEURODEGENERATIVE DISEASES

Niemann-Pick type C1 and Wolfram syndrome are rare, life-limiting diseases that both involve the gradual loss of nerve cells. Currently, there is no cure for either – and treatment options are extremely limited.

Dr Sovan Sarkar and his team at the University of Birmingham are testing existing therapies, already used for other health conditions, that could help slow progression of symptoms. They are using nerve cells and 'mini-brains' – generated from patient skin cells grown in laboratory dishes – to assess two types of treatment. If the results are encouraging, the team plans to advance the new treatments to clinical trials with patients as soon as possible.

This research is co-funded with LifeArc.

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

RIDE
CASTLE 20 YEARS

This year sees us celebrate the 20th anniversary of the RIDE Castle event. Join us to conquer this classic cycling challenge and help save little lives.

One of the original events in our RIDE series, this continues to be one of our best-loved UK bike rides. Setting out from the historic ruins of Tonbridge Castle, the routes take in some of the most beautiful Kent countryside.

As ever, there's a mix of rolling terrain and some challenging climbs, with four route options to suit all abilities, from the Cool choice of 36 miles right up to the Epic of 126.

This year's ride, on 11 May, will also see some extra post-ride activities to celebrate the past 20 years.

Other UK bike rides coming up include another old favourite, RIDE Suffolk Sunrise (18 May), which celebrates its 20th anniversary next year, plus RIDE the Dales (29 June) and RIDE Essex (31 August).

For more about cycling events, scan the QR code or visit **action.org.uk/cycling**

TRACK ATTAQUE

NEW for 2025, this 12-hour cycle challenge will be held at the iconic Herne Hill Velodrome, venue of the Olympic track cycling back in 1948. On 14 September, teams will vie to complete as many laps as they can between 9am and 9pm.

EVENTERS SHINE IN RACE THE SUN

Our triathlon-style challenges saw more than 1,550 people bike, hike and canoe for Action last summer. And there's more to come this year!

Superhero costumes are optional but everyone who takes part in these events is a hero to us.

Among them in 2024 were the New Cross Neonatal team. Father and son Mike and Henry took on the Brecons event, inspired by Henry's experience of being born too soon – he is now at university, hoping to become a doctor.

The Pen & Ball team also had personal experience of premature birth. They joined us at the Keswick off-road event – and made some brilliant videos.

Plus we again had some great corporate teams, including Alliance Healthcare, APH, Touchstone and Garmin.

Find out more about Race the Sun at **action.org.uk/sun**

CLIMB THE CAPITAL

Join us for a vertical test of endurance that takes in three iconic London skyscrapers.

Back for 2025, our Climb the Capital fundraising challenge is not your average fun run – it's a heart-pounding stair climb, ascending 475 metres!

Participants conquer the Cheesegrater, the Scalpel and the Walkie-Talkie buildings – that's more than 3,180 steps across 125 floors – before finally being rewarded with breathtaking views from the Sky Garden.

We need intrepid people to join us for this year's exhilarating event, being held on Saturday 4 October.

Find out more at **action.org.uk/capital**

KEVIN’S BIKE CHALLENGE

In response to his stage four cancer battle, long-time Action supporter Kevin Griffiths has launched a bold new fundraising challenge. Following colon surgery, avid cyclist Kevin can no longer sit comfortably on a saddle. So he devised Cancer My Arse! The idea is for participants to ride as far as they can without sitting down.
Find out more at **cancermyarse.co.uk**

FOOD AND FUNDRAISING

Brighton-based restaurant group **Gingerman**, run by Ben and Pamela McKellar, has supported Action for many years. The charity is promoted in their four popular venues, and we currently receive a £1 donation from every table of customers that eats, as well as counter-top collections.

RIDE RAPTÖR

This year sees the fifth edition of our bike ride for the insurance industry. This has now raised over £1 million, with 2024’s event raising more than £350,000 – the most yet. It has been superbly supported by founding partner **Ki**, along with main partners **Brit** and **Lloyds**, and a growing list of top names in the sector.

DOUBLE DAVINA FOR 2025

The last Dine with Davina ladies’ lunch was our best yet, raising a fantastic £78,000. And this year we’ll be doing it twice!

Hosted by the amazing Davina McCall, a tireless Ambassador for Action, our brand new Essex event, held on Friday 20 June, will join the long-running Southampton edition, which takes place in November (Friday 7). Our new venue will be the gorgeous Hylands Estate in Chelmsford, as recommended by Davina herself! Join her for a day filled with fun, inspiration and making a difference.

Tickets are on sale now. To find out more about both Dine With Davina events, please visit **action.org.uk/davina**

SUPPORTER STORY

THREE TIMES THE FUN!

Kay Prestney and friend Sam Harty took on not one but two Race the Sun events last year – and they’re back for the new Cheddar Gorge challenge this summer.

“We loved the idea of an adventure triathlon – something fun and different, held in breathtaking scenery, while raising money for a worthy cause,” says Kay. “We felt so proud of ourselves for finishing and delighted to know that our efforts mean more research can be carried out to find treatments for rare diseases. A close friend’s son has Wolfram Syndrome, a very rare disease that gets little funding. We wanted to do something to help and are thrilled that Action is now funding new research for this condition.”

To find out more about Race the Sun visit **action.org.uk/sun**

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.

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

1. Elsie
2. Premature
3. Birth
4. Duchenne
5. Potential
6. Therapy
7. Infection
8. Virus
9. Pandemic
10. Cancer
11. Wolfram syndrome
12. Castle ride

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**

Summer Superdraw

WIN up to £4,000 in our Rollover Jackpot!*

JUST £1 a chance!

How would you spend £4,000?

More chances to WIN than ever before...

55 guaranteed prizes!

1st PRIZE	£500
2nd PRIZE	£250
3rd PRIZE	£100
4th PRIZE	£50
5th PRIZE	£25

Plus 50 Runner up prizes!

Worth £65

Draw date: 27 June 2025

Hurry... enter before 16 May for an extra chance to win a bottle of Bollinger!

Plus...
for every 12 entries you could win £50 NEXT vouchers in our Free Gift Draw

NEXT

PLAY NOW! action.org.uk/superdraw

Already playing the lottery?
You're already in the Superdraw! Increase your chances of winning by buying extra entries for just £1 per chance.

action medical research for children

FIGHT BACK FRIDAY LOTTERY