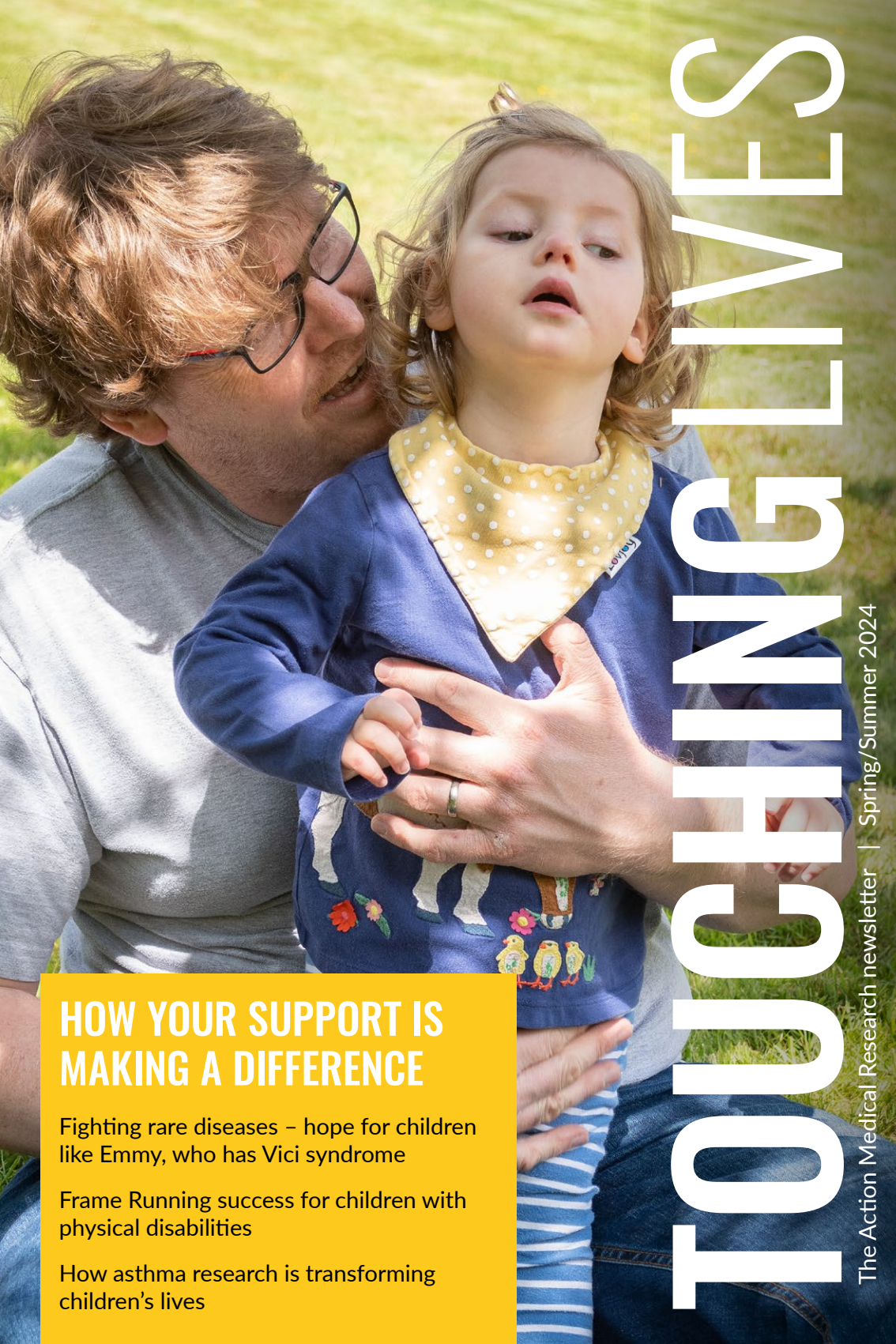

TOUCHING LIVES

The Action Medical Research newsletter | Spring/Summer 2024

HOW YOUR SUPPORT IS MAKING A DIFFERENCE

Fighting rare diseases – hope for children like Emmy, who has Vici syndrome

Frame Running success for children with physical disabilities

How asthma research is transforming children's lives

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IN THIS ISSUE

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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WELCOME

It's always wonderful to be able to share how medical research has transformed the lives of young people. And in this issue you'll meet Sophie, Ian and Christian, who have all benefited hugely thanks to the success of research funded in recent years – something that all supporters should feel incredibly proud of.

But at the same time, you'll also meet little Emmy, who has a life-limiting rare disease for which there is currently no treatment or cure. Across the UK there are thousands of families, like Emmy's, living under the shadow of rare diseases. We truly hope that new research into Vici syndrome, and other rare diseases, will one day have the same life-changing impact for families. With your ongoing support, we can fund more vital work, bringing hope to more families.

Fundraising comes in many forms, from events like our Race the Sun challenge or running a marathon, to taking part in our FIGHT BACK Friday Lottery (which has another exciting Spring Superdraw coming up!). However you support us, we are grateful to every single one of you.

Thank you.

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

75%
of rare diseases start in childhood

AROUND 95%
of rare diseases lack an effective treatment

NEWS

FIGHTING FOR FORGOTTEN CHILDREN

Funding for research into rare diseases is limited – but the impact rare diseases can have on families is enormous. Research brings hope to these forgotten families.

Our rare disease research campaign has now raised almost £600,000 towards research that offers hope for future treatments and cures. Hope for families like Emmy's, who features on our cover. We spent some time with Emmy and her family, making a short film to show why this work is so important.

You can watch the video here **action.org.uk/Emmy**

RESEARCH UPDATE

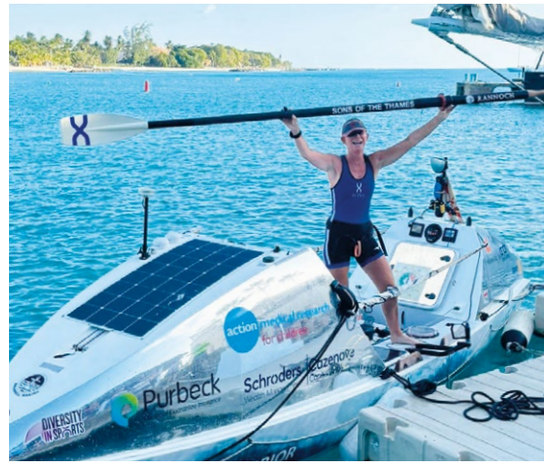
NEW RESEARCH

With your help, four new projects to help children fighting rare diseases are now underway. Co-funded with our charity partner LifeArc, this amounts to almost £1 million of vital new research.

These teams are striving to develop urgently needed treatments for some truly devastating diseases – including steroid-resistant nephrotic syndrome, which causes kidney failure, and diffuse midline glioma, the most lethal form of childhood brain cancer. Another aims to create customised face masks for children with facial differences who need ongoing breathing support at home.

ATLANTIC TRIUMPH

Adventurer and endurance athlete Linda Blakely has raised more than £100,000 for our research into rare diseases after completing an awe-inspiring solo row across the Atlantic Ocean. Linda, who celebrated her 50th birthday at sea, battled against challenging weather and tides – and even saw her 12-foot boat capsize in pitch darkness just two days in. We are so proud of her amazing achievement and so grateful that she chose to support Action.



FIGHTING FOR CHILDREN LIKE EMMY

Two-year-old Emmy has Vici syndrome, one of the most severe multi-system conditions that can affect children. There is currently no cure or effective treatment for this rare disease. Sadly, most affected children do not live beyond the age of five.

"Our beautiful girl is blind, tube-fed and unable to walk or talk, but her personality shines through," says mum Ellie. "She is calm and content, but with a brilliantly stubborn determination. She is very communicative once you learn her cues and responds to all kinds of therapies and inputs, and has an especially close bond with her sister."

Emmy was born on her due date in 2021 and from her earliest days, Ellie felt there was something wrong. Emmy found it incredibly difficult to feed and as the weeks went on, Ellie and husband Jon's fears grew. Emmy didn't begin to follow or track anything with her eyes, and she didn't begin to smile.

At three months old, Emmy had a chest infection and when she was admitted to hospital by the lead paediatrician, Ellie seized the chance to explain that they had serious concerns.

> **"We were blown away by such a catastrophic diagnosis"**

"I gave the doctor a long list of all the things that were terrifying me," says Ellie. "I explained that I didn't think Emmy could see, that we thought she had global developmental delay, that she wasn't smiling or interacting and couldn't grasp."

An MRI scan found significant changes in Emmy's brain. She was diagnosed with septo-optic dysplasia, affecting the optic nerves – and as Ellie feared, this meant that she couldn't see. She was also diagnosed with a condition affecting movement and muscle tone on one side of her body.

But it wasn't until Emmy was 18 months old that the most serious diagnosis of all was made – when genetic tests showed she has Vici syndrome.

While Ellie and Jon knew Emmy faced serious health issues, they were 'completely blind-sided' by the news.

"We were blown away – we really didn't think there could be anything so catastrophic wrong. She'd seemed to be thriving again," says Ellie. "We had managed to keep her out of hospital and felt we understood how things were working for her."

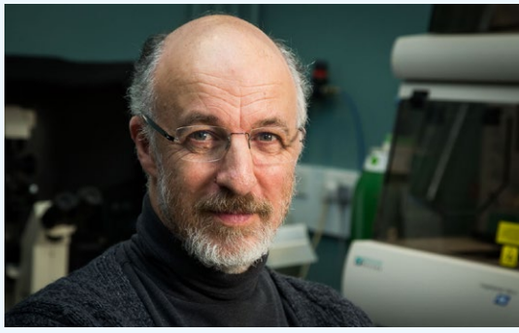
Research to find vital drug treatments

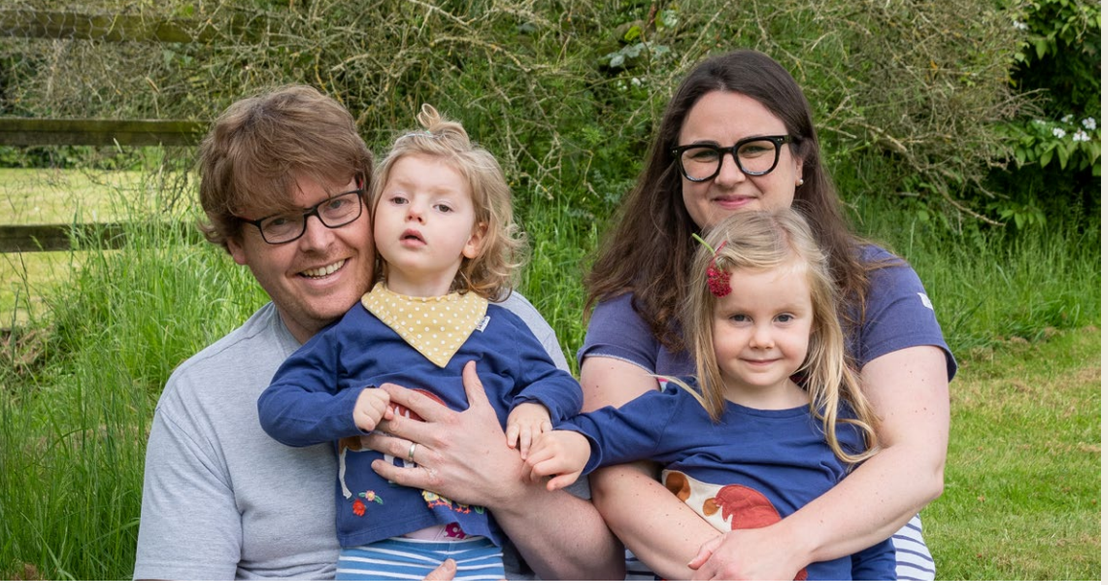
Your support is funding work to find drug treatments that could help reduce the impact of this devastating, life-limiting condition.

Researchers at University College London have recently discovered that patients with Vici syndrome have problems with their mitochondria, the energy-generating powerhouses in cells. They are now testing existing medicines that may help to restore mitochondrial function.

This approach could also be beneficial for children with other rare diseases which involve similar processes.

> **"This could help to identify new treatments that could transform the lives of children with Vici syndrome and their families"**
>
> Professor Michael Duchen





“We hope we’ll have longer with Emmy than the worst-case predictions”

Vici syndrome is incredibly rare and the doctor who initially shared the shattering news admitted they had no training or specialist knowledge they could offer. The family were emailed what Ellie describes as ‘a horrendous PDF, written for doctors’.

Sadly, the condition causes a very wide range of symptoms. These usually include recurrent infections due to an impaired immune system, heart problems, delayed development, deterioration of the nervous system and seizures.

Shortly after her diagnosis, Emmy developed epilepsy. “Then we began to truly understand that this was a very serious diagnosis,” says Ellie.

Because Emmy’s condition is so complex, her care involves multiple medical teams covering many different specialties.

“We’ve totally re-designed our lives – partly because we’ve had to but also because there’s nothing more clarifying, in terms of what really matters, than having a little person like Em around,” says Ellie.

Thinking about the future is very daunting. “We hope we’ll have longer with Emmy than the worst-case predictions, and that things will be easier and happier,” says Ellie. “I hope for the same things that all parents hope for – health, happiness and longevity but within a different framework I guess.”

“The research being funded by Action could be incredibly significant,” says Ellie.

THANK YOU!

Together, we can fight rare diseases.

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

PREVENTING PREMATURE BIRTH

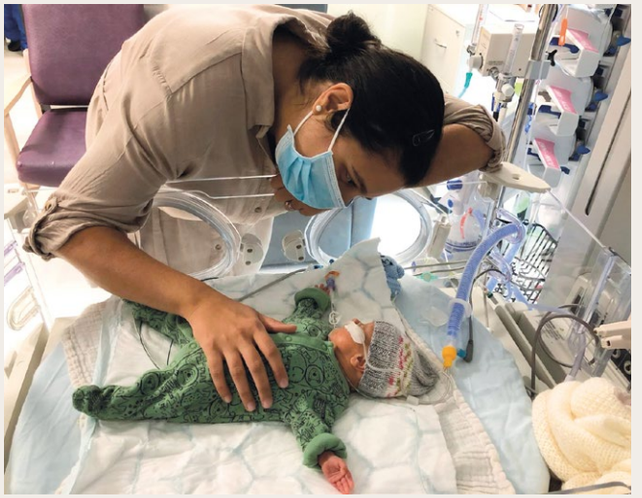
The causes of preterm birth remain poorly understood but an infection may be involved in forty per cent of women who experience an unexpected early labour. New research could help to prevent this in the future.

Babies born too early face an increased risk of disability and lifelong health problems – and sadly, more than 1,000 UK babies die each year after being born too soon.

Although the causes of preterm birth remain poorly understood, evidence suggests that in some women bacteria can pass into the womb, triggering inflammation and in turn increasing the risk of early labour.

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

With co-funding from Action and Borne, a team based at University College London aims to develop an innovative antimicrobial therapy to boost the body’s



natural defences in women who are known to be at higher risk of premature birth.

As well as providing a physical barrier, the cervix also produces specialised antimicrobial proteins to help fight infection. Dr Ashley Boyle and Professors Simon Waddington and Donald Peebles aim to boost this effect in cervical cells, which they hope will help stop bacteria from entering the womb.

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.

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

Dr Ashley Boyle

WHY I SUPPORT ACTION

As a teenager, Sophie was one of the first young people to benefit from life-changing brain surgery, guided by a pioneering technique called deep brain stimulation. Sophie shared her amazing story at our latest Dine with Davina fundraising event.

SOPHIE LENNOX

Q Can you tell us a bit more about how you came to get involved with Action?

A My life was more than likely saved, or at the very least given back to me, by work funded by Action. Also, when I was seven, my younger brother Tom died of a rare muscle disease that yet has no cure. So my family know only too well the difference medical research can make. It's been my pleasure to share my story at charity events, like Dine with Davina – there was a time when I wouldn't have thought it possible.

> **"I was having up to 20 seizures a day. Normal activities were out of the question"**

Q Can you tell us a bit more about your epilepsy?

A I had my first seizure during a run at school when I was 11. At first, everyone thought it was a one off. But within six months I was having up to 20 seizures a day.

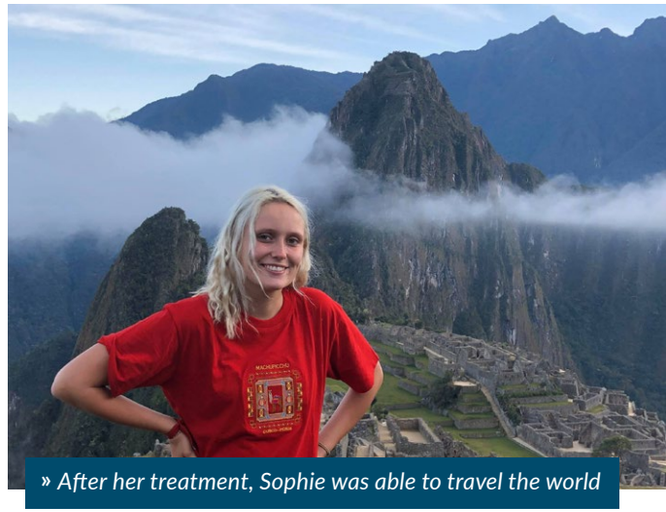
I tried many different medications, often with unbearable side effects, but my seizures became more aggressive. I began to lose my balance and would often end up crashing to the floor. Even when I was taking double the adult dosage of medication, I still had at least three or four seizures a day.

Q This must have had an enormous impact on you and your family...

A It did. The risks I was exposed to daily were endless. I fell in front of moving buses, crossing the road, down flights of stairs and often had seizures mid-lesson at school. My parents were on constant alert and life as a teenager felt very restricted. Normal activities like swimming or cycling were out of the question. I couldn't even have a bath safely. As you can imagine, my ability to learn also suffered dramatically.

Q Can you tell us a bit more about the surgery you had?

A I was diagnosed with focal cortical dysplasia (FCD), which is like a birthmark in your brain, and offered surgery. But I was told there was only a 55% chance it would work and that I could lose the function of my right arm. Then we found out about a new technique being trialled, where doctors were using deep brain electrical stimulation to pinpoint the exact area in the brain that needed treatment. This work was being funded by Action and Great Ormond Street Hospital Children's Charity.



» After her treatment, Sophie was able to travel the world

> **"There are not enough words to describe the impact that research funding provided by Action has had on my life"**

» Sophie in hospital for brain surgery

The results were amazing – I haven't had a seizure since, and here I am, seven years later. I've achieved things I feared I'd never be able to. I went away to university, completed my degree and a Masters. I've travelled around the world, jumped out of planes, white water rafted... things that would have never been possible without this surgery.

Q What does supporting Action mean to you now?

A The charity is currently funding more new research that could mean more young people with the type of epilepsy I had could benefit from life-changing surgery. There are not enough words to describe the impact that Action has had on my life. Thank you to all who donate. You really do change lives.

FRAME RUNNING
IS HELPING CHILDREN LIKE IAN TO SHINE

Action funding has helped to develop a new sport for young people with cerebral palsy and other conditions that limit mobility. This is already having a positive impact for children like 12-year-old Ian, pictured.

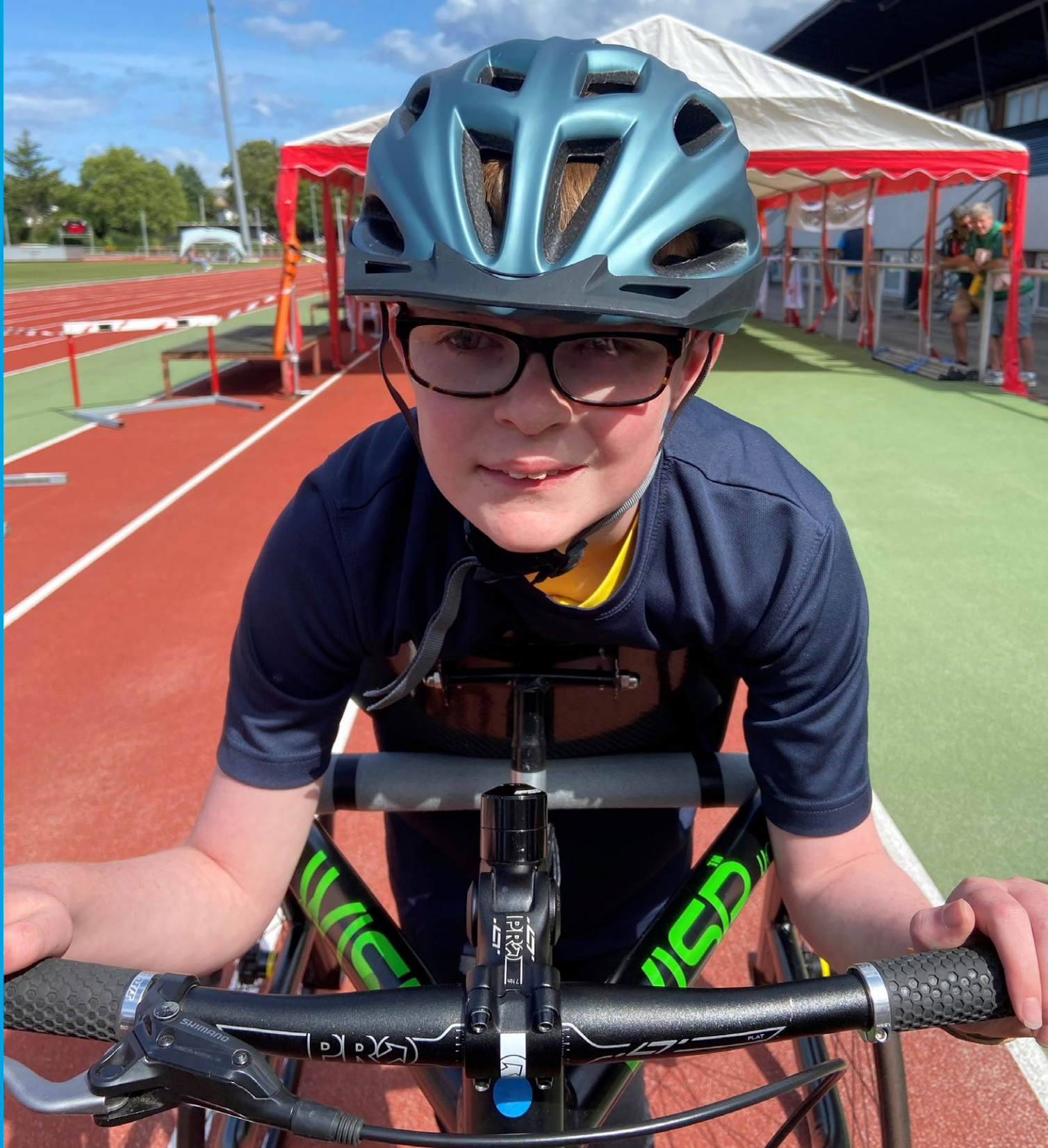
Ian has always loved trying out new things, but with cerebral palsy affecting one side of his body, he faces difficulties with muscle strength, stiffness, coordination and balance. He uses walking aids and, part of the time, a wheelchair.

Frame Running involves propelling a three-wheeled running frame with a saddle, body support and no pedals. In 2019, Ian joined a group of children taking part in a pilot study to assess if this encouraged children to increase their physical activity, and if it had a lasting positive effect on their health and wellbeing. This research was jointly funded with the Chartered Society of Physiotherapy Charitable Trust and led by Dr Marietta van der Linden and Dr Jennifer Ryan.

Today, Ian continues to enjoy all that Frame Running offers – and has twice represented Scotland at the annual international competition for Frame Runners, winning a string of gold medals.

"Frame Running gives children like Ian physical independence and a sense of achievement"

Ian's mum, Sheena



A NEW APPROACH TO TREATING ASTHMA

Action funding helped to support a groundbreaking clinical trial, which showed that personalising asthma medication, according to genetic make-up, can improve treatment and quality of life for children and young people – and the results are creating impact across the world.

Asthma causes coughing, wheezing and difficulty breathing – and is a leading cause of emergency hospital admission in children and young people.

When the condition is managed well, children can lead a full and active life. But when it's not, it can severely limit their ability to play, run and be active, and affect attendance at school.

Unfortunately, evidence was emerging that one of the most commonly used asthma controller medications, called salmeterol, was not equally effective in all children. This means that some would not see the expected benefits and continue to experience symptoms, including asthma attacks.

> **"Action's funding was critical in supporting the first-ever clinical trial of this kind of precision medicine for children's asthma"**
>
> Professor Somnath Mukhopadhyay

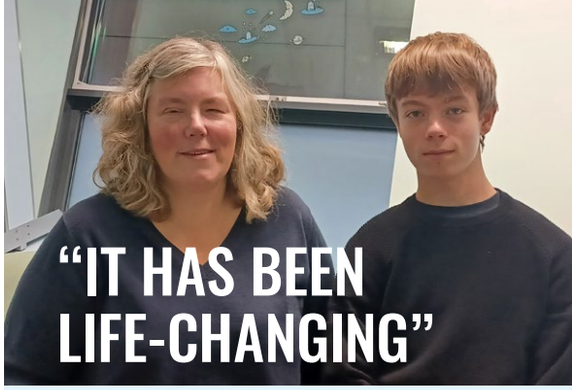
With Action funding of more than £270,000, an expert team, led by Professor Somnath Mukhopadhyay at Brighton's Royal Alexandra Children's Hospital, ran an exciting clinical trial, involving 240 children aged 12 or over. The first of its kind, this trial tested prescribing treatment based on genetics – an approach known as personalised medicine.

AROUND 1.1 MILLION
children in the UK have asthma

A cheap, simple saliva test was used to determine children's genetic make-up. They were then prescribed either salmeterol or another drug called montelukast, according to the results of the test. The research showed that a personalised approach could significantly improve quality of life.

In the UK alone, an estimated 150,000 children with asthma have the genetic make-up that means salmeterol is likely to be less effective. As a result of this research, life-changing care is now being provided for some of these children. The hope is this will extend to help many more, worldwide, in the near future.

We are grateful to the many trusts and foundations who helped us support this research, including The Henry Smith Charity.



"IT HAS BEEN LIFE-CHANGING"

Christian was one of the children who took part in the clinical trial. Changing his treatment, based on his genetic profile, has finally brought his asthma under control, giving him freedom to enjoy life with his friends.

Fifteen-year-old Christian had struggled with his asthma for years and the highest doses of medication didn't seem to help. His mum, Catherine, said it felt like they'd run out of options. "We were at the doctors' surgery all the time," she says.

Christian says his new treatment has made a big difference to his life. "I can now participate in sports that I previously struggled with, like football, badminton and going to the gym – things that would have affected me a lot before. It's allowed me to do lots of things with my friends that I would have struggled with before."

> **"It has been a total game-changer! It has had such an amazing impact"**

"It's been life-changing," adds Catherine. "We are so grateful to all the charity supporters who helped make this work possible and feel so lucky that we've been able to benefit from it."

PROTECTING BABIES AND CHILDREN AGAINST MENINGITIS

With your support, researchers have identified new and emerging strains of bacteria that are responsible for most cases of pneumococcal meningitis in young children.

Pneumococcal meningitis mostly affects babies and toddlers and sadly can have devastating consequences. Up to one in seven lose their lives and a quarter are left with severe after effects, including sight or hearing loss, seizures and learning disabilities.

In the UK and Ireland, vaccines now protect against the most common strains of pneumococcus bacteria. But little was known about the risk, severity and outcomes of meningitis caused by new and emerging strains of bacteria.

With Action funding, Dr Godwin Oligbu and his team at Public Health England analysed cases of pneumococcal meningitis. They collected samples and data from affected children to identify bacteria strains involved and compare symptoms, treatments, and outcomes.

> **"This study will inform future vaccine policy and treatment"**
>
> Dr Godwin Oligbu

Dr Oligbu says: "This study will inform future pneumococcal vaccine policy. It will inform policy makers of the predominant types of bacteria which are now causing meningitis and fatalities. It will also help provide guidance for paediatricians to better manage the condition."

OUR LATEST RESEARCH

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

HELPING CHILDREN WITH LIVER DISEASE

With your support, researchers are developing gene therapy for a rare disease that causes liver failure. Children born with progressive familial intrahepatic cholestasis type 3 eventually need a liver transplant. This is major surgery and the need for new livers far outweighs the number of donors. This research could lead to a safer and better treatment option, transforming lives and sparing children from the long waits and the risk of complications associated with liver transplants.

TREATING RARE HEART CONDITIONS

Arrhythmogenic cardiomyopathy is an inherited form of heart disease, where the heart muscle cells don't stick together properly, and the walls of the heart become weak. Some children show no symptoms but can still be at risk of life-threatening complications. Dr Sara Moscatelli, of Great Ormond Street Hospital, aims to identify early signs that can help predict how the disease will develop in children. This could lead to personalised approaches to treatment, helping to prevent sudden and unexpected death in children and young people.

FIGHTING FATAL BRAIN TUMOURS

Diffuse midline glioma is the most lethal form of childhood brain cancer. It will sadly prove fatal for nearly 90% of children within 18 months of being diagnosed. Current treatments do not work against this type of tumour, which grows in the midline between the two halves of the brain.

Researchers at King's College London aim to develop an urgently-needed new and non-invasive treatment approach. This will use gene therapy combined with focused ultrasound to deliver highly targeted treatment, personalised to each patient's tumour.

This project is co-funded with LifeArc.

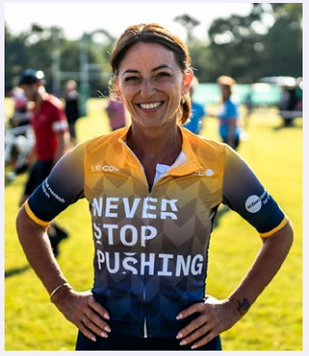
RIDE FOR LITTLE LIVES

Our cycling season has sprung into action again, with rides across the UK and beyond. All that pedalling helps to power vital research for babies and children!

Our RIDE series has something for everyone with some of the most scenic cycling the UK has to offer. Whether you're a new or a seasoned sportive rider, our routes range in distance and difficulty.

You could conquer the upcoming RIDE Castle in Kent (5 May), enjoy the more forgiving terrain of RIDE Suffolk Sunrise (19 May) or RIDE The Dales in stunning Yorkshire (2 June).

There's also the chance to join TV's Davina McCall at Davina's Big Sussex Bike RIDE on 2 June. Davina will set you off on your way, then ride the route to try to catch you up!



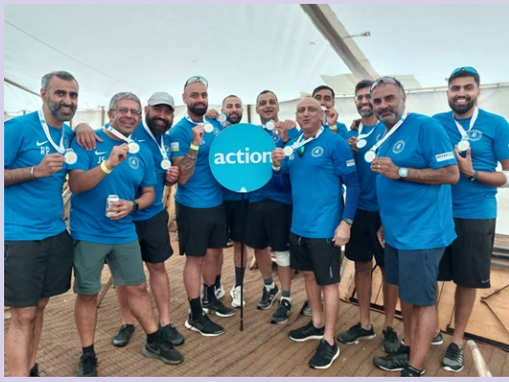
Scan the QR code to find out more or visit **action.org.uk/ride**

RACE THE SUN TEAMS TRIUMPH

2023 saw our biggest Race the Sun season yet and an amazing effort put in by the teams taking part in these triathlon-style challenges.

Among them were a team of 12 from Bradgate Bicycle Club, Leicestershire. Led by Nim Gill, they took part in the Lake District Off-Road event. They totally smashed their fundraising target, raising almost £12,500 to win the top fundraising team award for the event. Plus they were so much fun to have around on the day, really adding to the atmosphere.

A special mention also has to go to a team of four siblings, Gaagy's Gang,



who flew in from different countries to take part in Race the Sun in Keswick. They raised more than £5,500.

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

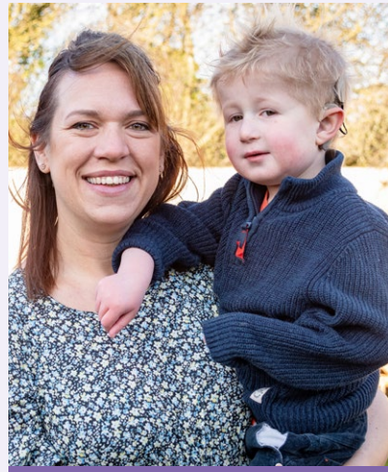
MARATHONS WITH MEANING

Among our brilliant Action runners in the TCS London Marathon this month will be teams inspired by two very special children.

Ellie Hudson is mum to Finley, one or our previous cover stars, who has a rare condition called Diamond-Blackfan anaemia. This means that six-year-old Finley currently needs a blood transfusion every three weeks. Ellie is running with friends Max Thorne, Mark Stephenson and Brad Lemmon.

Damian McConnochie and David Spurway will be running TWO marathons in a month in memory of Damian's step-son, Rohan, who would have been 21 this year. Sadly, Rohan suffered from a very rare and progressive mitochondrial disease for which there was no cure. He eventually lost his sight and died when he was just 10 years old.

Good luck to these and all our 2024 runners.



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

FROM BALLROOM TO BIKE

Fresh from competing in *Strictly Come Dancing*, Paralympic cycling legend Jody Cundy CBE was among guests at our annual Champions of CycleSport Dinner. The event was generously supported by Garmin, BDO LLP and Lifeplus, and raised an incredible £280,000. We're looking forward to doing it all again this November. Find out more at **action.org.uk/champs**

NEW CATALYST RIDE

We're excited to be launching a new industry bike ride this year. Catalyst is for professionals from the bioscience and pharmaceutical sector. We're partnering with BiotechBikers for the ride in Italy in September. Our other events include Raptör, with the insurance trade, and Ziggurat with partners in construction. All raising vital funds for research.

AMAZING ALLIANCE

Alliance Healthcare raised more than £20,000 for Action in 2023. This included a summer garden party, choosing us to benefit from their annual football tournament and having teams take part in Race the Sun.

FANTASTIC FUNDRAISING

SCOTLAND SPARKLES

Our two biggest fundraising events in Scotland, Killer Heels and Cocktails, held in Glasgow and Aberdeen, are always key dates on the Action calendar.

Each year, these popular events attract some of Scotland's most glamorous and influential ladies for a fabulous afternoon of food, fun and fundraising.

They are managed by two talented, hardworking volunteer committees, and we are hugely grateful for their dedication to making them so successful.

In 2023, they raised a net profit of over £80,000 to help fund more much-needed research. Details of 2024 social events can be found at **action.org.uk/social**

COULD YOU BE OUR NEXT BIG WINNER?

action medical research for children
FIGHT BACK FRIDAY LOTTERY

Christmas feels like a distant memory, but the run up to the festive season saw some fantastic prizes being won in our FIGHT BACK Friday Weekly Lottery!

One lucky supporter from Leamington Spa scooped a Rollover Jackpot of £10,500. While Dan (pictured), from Burgess Hill, was our Christmas Superdraw winner of £500 plus a luxury hamper.

Dan says: "Supporting Action Medical Research in the lottery draw is a wonderful way to help raise money. It was an added bonus to win the Christmas Superdraw – we very much enjoyed our prizes over the festivities."

There's another chance to WIN £500 and a luxury hamper – and up to £5,000 in our Rollover Jackpot – in our Summer Superdraw, which takes place on 5 July. Buy your chances now **action.org.uk/superdraw**

WORDSEARCH

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	M	I	R	E	R	V	R	S	E	R	E	S	B	B	E
G	N	I	N	N	U	R	E	M	A	R	F	S	L	I	M
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E	M	N	L	Z	E	X	N	B	A	S	T	H	M	A	M

1. Emmy
2. Vici syndrome
3. Epilepsy
4. Success
5. Frame Running
6. Asthma
7. Medicine
8. Meningitis
9. Liver
10. Bike ride
11. Running
12. Winner

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

Draw date:
5 July 2024

Just £1 a chance!

WIN up to £5,000 in our Rollover Jackpot!

WIN £500 and a luxury hamper worth £100!

Worth £60

Hurry... enter before 24 May and you could also win a box of Deli Delights in our Quick Reply Draw!

Plus... for every 12 entries you could win a mini hamper in our Free Gift Draw.

Play now!

action.org.uk/superdraw

Already playing the lottery?
You'll be automatically entered into 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