HOW YOUR SUPPORT IS MAKING A DIFFERENCE

Helping children like Henry, who has a rare heart condition
New research into causes of premature birth
Big breakthrough in the fight against juvenile Batten disease
WELCOME

We have lots of new research to share with you, plus some exciting updates on work your support has already helped make possible. I hope you’ll enjoy seeing how you are helping to save and change children’s lives.

For children suffering from rare diseases, treatment options can be severely limited, or just not there at all. Rare diseases are also often overlooked and underfunded in terms of medical research. We know that research brings hope to families and with your support, we can do even more to help. Read more on pages 12 and 14 about progress already being made for two rare conditions.

This year, we are also celebrating 50 years of our Research Training Fellowship scheme. Funded with your support, this important scheme helps train the medical researchers of the future, and many go on to become experts at the top of their fields – as you can see on pages 10-11.

This issue also features some amazing Action fundraisers. From rowing the Atlantic to taking part in (and winning!) our FIGHT BACK Friday Lottery, we are grateful to every single one of you who supports us. Thank you.

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

NEVER

SHINING A SPOTLIGHT
ON RARE DISEASES

Across the UK, thousands of children are living with rare and devastating diseases that have no cures. And, tragically, 30% of these children will lose their lives before their fifth birthday.

That’s why we’ve launched a new campaign to help raise much-needed awareness – and raise crucial funds to support urgent new research into rare diseases that cause so much suffering for children and their families.

We are striving to develop treatments and cures. But we can’t do it alone. Medical research gives hope – hope for new treatments, hope for a cure.

With your help, we will NEVER give up

NEW RESEARCH

Thanks to your support, recent months have seen us award funding for 11 new research projects. These include work to test a new brain imaging technique to help children with epilepsy and research to better understand how labour is triggered, which you can read about on page 7.

There are also three new studies to help children with different rare diseases, including hypoplastic left heart syndrome and Vici syndrome – a devastating, life-limiting disease that currently has no treatment or cure.
New research to develop tests to save lives

With your support, researchers aim to develop new tests that can identify children with HCM and predict the severity and progression of the disease. Led by Dr Juan Pablo Kaski at University College London, the team has recently developed a blood test for adults. They will now see if this test also gives accurate results in children, including those who are known to be at risk but have yet to show symptoms.

The researchers also aim to identify new biomarkers in the blood and urine that are specific to children. This project is jointly funded with LifeArc.

"Our ultimate goal is to improve diagnosis, monitoring and treatment – and prevent the disease from progressing"

Six-year-old Henry is a happy, energetic boy who loves running around and playing football with friends. When you look at him you wouldn’t know that he’s harbouring a serious heart condition.

But when he was just two days old, Henry’s parents received the shocking news that he had hypertrophic cardiomyopathy (HCM), a condition that causes a thickening of the heart muscle. Although it is rare, it can cause sudden death in children and young people.

Estimates suggest up to 1,000 children in the UK have HCM, and there may be many more undiagnosed with few or no symptoms. The thickening of the muscular wall of the heart makes their heart muscle stiff and less efficient at pumping blood. This can lead to symptoms such as shortness of breath, chest pain and palpitations.

"Hypertrophic cardiomyopathy is usually hereditary," explains Henry’s mum, Kayleigh. "But we haven’t got it in our family so Henry’s situation is very rare. We know we are incredibly lucky that the doctors detected it, and this way it can be monitored closely."

As a precaution, Henry’s older sister Georgina also has annual checks: "We don’t know what the future could bring, it’s a huge worry that it could be picked up in our daughter too," says Kayleigh.

Living with two serious heart conditions, Henry’s earliest years were fraught with fear for Kayleigh and husband Dan. "We were in a constant state of worry – as we didn’t know what we were dealing with," she says.

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Helping children like Henry

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"We are incredibly lucky that Henry’s heart condition was detected – and not discovered at a catastrophic time later in life"

Henry was born via emergency caesarean at 37 weeks as doctors had detected another condition, supraventricular tachycardia, which can cause the heart to suddenly beat much faster. It was through being treated for this that doctors realised he had hypertrophic cardiomyopathy as well.

"Our ultimate goal is to improve diagnosis, monitoring and treatment – and prevent the disease from progressing"
As he has got older, Henry and the family have adapted well.

“Although we don’t want him to feel different, we’ve had to make sure he is cautious. Henry is aware of his condition and if he feels that something isn’t quite right he knows it’s important to take rest and let someone know,” says his proud mum.

Kayleigh used to feel very nervous when Henry ran around: “I had nightmares about him taking part in sports day at school as we simply don’t know how far he is able to push himself,” she says. “It feels awful to think we’re holding him back, but the thought of something happening to him is worse than missing out on some activities. He is very understanding and takes everything in his stride.”

“I don’t think we’ll ever get out of the habit of checking for defibrillators wherever we go”

Henry’s care is led by Dr Juan Pablo Kaski, who is also leading Action-funded research – and Kayleigh says the family is very grateful that Henry is monitored by ‘a brilliant team, who are leading vital work to help improve understanding of HCM in children’.

“This research brings hope to families,” says Kayleigh. “We hope that as Henry grows up his path, and that of others affected, will become steadier and easier.”

“I don’t think we’ll ever get out of the habit of checking for defibrillators. You hope you’ll never need to use one, but reassurance is everything when you’re living with a condition like this.”

Dr Victoria Male and her team, based at Imperial College London, have identified a new kind of immune cell in the lining of the uterus, whose number and activity increases during labour. These cells switch on genes that activate the local immune response and help the waters to break.

Around one in every 13 UK babies is born prematurely – before 37 weeks of pregnancy. The earlier a baby is born, the higher their risk of death or serious complications that can leave them with lifelong disabilities.

Many premature births happen when women go into labour too soon, often for no apparent reason. Researchers do not yet fully understand the mechanisms in the body that control labour.

“By understanding how these cells are involved in labour, future studies might be able to target them to reduce rates of premature birth”

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With co-funding from Action and Borne, the team now aim to determine whether these cells trigger labour. They will compare cells taken from pregnant women at full term, who are either not in labour or who are in early labour. They will also then investigate whether these cells are involved in preterm labour.

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Dr Victoria Male
My sisters Jade and Jordon lived very short lives, full of love and care.

When Jordan arrived two years later, it came with some shock that she also had PEHO syndrome. Only five children in the UK had this diagnosis at the time, including Jade and Jordan.

What was the best aspect of rowing the Atlantic?

It’s raw and pure peace amongst the crashing waves. It makes you find strengths you never knew you had, pushes your boundaries, and gives you time to think about what is really important. The sunsets and sunrises are like you’ve never seen, the night skies like a planetarium, the ocean, the bluest you’ve ever seen. It’s truly beautiful, harsh but beautiful.

What was the worst aspect of rowing the Atlantic?

For me it was getting injured. I fell on the boat quite early on, leaving me with concussion and two huge black eyes. It made me feel quite unwell for many days and, apart from a doctor on the phone, there is no instant medical help. I know this was scary for my crew.

Lack of sleep is tough too. The most we got at any one time would be 90 minutes, but due to alarms or the weather I would often be up during my off shift too.

How did it feel at the end of the challenge?

The end is amazing, with your loved ones all there waiting for you. The welcome was fantastic – people shouting, yachts blowing horns, music playing, flares burning. It’s quite overwhelming. We missed Christmas, new year and birthdays with our families, but we get to do that next year. The most important thing is we were able to help families not so lucky as us. That makes the difficulties and hardships all worthwhile.
Since 1973, we have run our Research Training Fellowship scheme, helping to train and develop some of the most promising doctors and scientists early on in their research careers. We are proud that some of the leading children’s researchers today were Action Research Training Fellows, people like . . .

PROFESSOR MANJU KURIAN
Who identifies and strives to treat the genetic causes of life-limiting brain diseases in children.

“I’m passionate about working to better the lives of children affected by neurodevelopmental conditions. Without Action, I wouldn’t be where I am today”

PROFESSOR BOBBY GASPAR
Who has helped pioneer and continues to develop life-saving gene therapies for rare diseases.

“My Action Research Training Fellowship set me on the road to my academic and clinical career. The ultimate prize is seeing children get well”

PROFESSOR SIR ANDREW POLLARD
Director of the Oxford Vaccine Group, who led the trial to test the Oxford/AstraZeneca vaccine for COVID-19.

“My Action Fellowship was critical. It gave me a portfolio of skills which I continue to use today”
We're delighted to share the exciting impact of vital research funded with your support – work that has led to a significant discovery and the clinical trial of a new treatment to fight this devastating rare disease.

Juvenile Batten disease affects the brain and over time causes severe and extremely distressing symptoms. Children with the condition seem healthy and develop typically in their early years. But when they reach primary school age, they start to lose their sight and develop epileptic seizures.

The faulty gene that causes juvenile Batten disease, called CLN3, was discovered around 20 years ago. It gives the body’s cells instructions on how to make a protein (also called CLN3). But nobody knew what this protein did.

Without this vital piece of information, finding new treatments proved extremely difficult.

In 2015, with nearly £200,000 in funding from Action, Dr Emyr Lloyd-Evans and his team at Cardiff University embarked on a journey of discovery. By unlocking the mysteries of this disease, they hoped to open up the potential for new treatments where there were previously none.

Making a major breakthrough, this work revealed, for the first time ever, what CLN3 does – it acts as a transporter for potassium within the bodies’ cells. Knowing this, allowed the team to explore whether certain existing drugs might reduce or even reverse the effects of this protein malfunctioning. They identified two potential new treatment strategies.

This initial work has now progressed further, resulting in a clinical trial of a new treatment for juvenile Batten disease – with results from the Action-funded work providing vital evidence to support the planning and approval of this trial. This work brings much-needed hope to families affected by this cruel disease.

The diagnosis left us numb,” says Dee. “You think it can’t be happening, it can’t be real,” adds dad Jody.

All three children have slowly lost their mobility. Their sight is severely impaired and other symptoms have included pain, behavioural problems and stammering. Izzy is also now affected by dementia. Sadly, the family know that a cure won’t come quickly enough for them but they are hopeful for others in the future.

We’re excited about the opportunities to translate our discoveries into new treatments”

Dr Emyr Lloyd-Evans

“We just hope that in the future other families won’t have to go through what we have”

In 2015, parents Dee and Jody were devastated to discover that three of their four children had juvenile Batten disease. Identical twins Toby and Corey were 11, their sister Izzy just six years old.

In their early years, all three children had seemed healthy and well. But in primary school the boys’ eyesight began to deteriorate. Doctors were initially baffled. Then the children began to suffer seizures.

Twin brothers Toby and Corey and their sister Izzy, top right, all have juvenile Batten disease.
**Finding New Treatments for Spinal Muscular Atrophy**

Action funding has helped researchers to show, for the first time, that combining another drug with existing treatments could further improve symptoms of this rare condition.

Spinal muscular atrophy (SMA) leads to muscle wasting and loss of nerve cells. There is currently no cure and until recently there were no effective treatments.

When Sophia, pictured, was diagnosed with the disease as a baby, she was not expected to live beyond her second birthday. Thankfully, Sophia has benefited from new gene therapies. But while these new treatments make a huge difference and are saving lives, sadly they do not improve all symptoms.

With funding from Action and SMA UK, Dr Melissa Bowerman and her team at Keele University have successfully tested a new drug approach. Their previous work had shown that certain hormones involved in regulating the body's metabolism are too high in SMA.

Early laboratory work has now shown that using an existing drug that targets these hormones can improve SMA symptoms – and can be especially effective when combined with a current gene therapy treatment.

Much further work is needed but this gives hope of an enhanced treatment for children with SMA in the future.

Sophia’s dad, Gennadiy：“Families like ours rely on research to keep discovering new treatments and optimising existing ones”

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**Nerve Repair in Children with MS**

Multiple sclerosis (MS) affects the brain and nerves. It can cause a wide range of symptoms, including problems with vision, movement, sensation or balance. Dr Jonathon Holland, of Addenbrooke’s Hospital in Cambridge, is studying nerve repair in children with MS to see if they could benefit from potential new treatments currently being tested in adults.

This Action Research Training Fellowship is jointly funded with the British Paediatric Neurology Association (BPNA).

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**Vitamin D and Child Health**

New Action funding is supporting a follow-on study to see if taking extra vitamin D during pregnancy benefits children right up to their teenage years, improving bone and muscle health and reducing obesity. Led by Dr Rebecca Moon at the University of Southampton, this work could ultimately lead to a change in national guidelines benefiting future generations.

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

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**Treating Brain Cancer**

Medulloblastoma and high-grade glioma are types of childhood brain cancer that are often very difficult to treat. Children with these tumours face prolonged treatment, with severe side effects. Sadly, many with high-risk cancer lose their lives.

Professor John Anderson and Dr Laura Donovan, at UCL Great Ormond Street Institute of Child Health, are developing a new immunotherapy treatment. Called CAR T-cell therapy, it has already proven effective for treating some other forms of cancer. It’s hoped this will have fewer side effects and save more lives.

This project is co-funded with LifeArc, plus a donation from Help Harry Help Others Cure.
JOIN OUR FACEBOOK FUNDRAISING CHALLENGES

It's never been easier to get active and help us raise vital funds, at your own time and pace, in one of our new virtual challenge events.

Our Facebook Challenge, Run 55 miles in March, has just finished and saw people across the UK join us remotely to raise money in honour of the 55,000 babies born prematurely each year.

Our next month-long challenge takes place in June, when we’ll again be raising funds to help sick babies and children. The activity will be revealed soon so keep an eye on our virtual events page.

Whatever it will be, you can really make the challenge your own, setting yourself individual goals to suit your own lifestyle.

And our friendly Facebook Challenge groups allow participants to be part of our fundraising community, connect with one another, and share tips and encouragement. Though if you’re not on Facebook, you can still take part, just let us know!

To find out more about our upcoming challenges visit action.org.uk/virtual

GET ON YOUR BIKE THIS SUMMER

Our cycling season has sprung into action and our next event sees TV star Davina McCall back on her bike. She’d love you to join her!

Davina’s Big Sussex Bike RIDE is on Sunday 25 June, with four route choices to suit all levels – from a gentler 27-mile route for newer riders, right up to a hilly 100-miler for the super-keen!

Looking further ahead, our RIDE series also includes the Suffolk Sunrise (21 May), followed by the return of a new-look Vyking event in Yorkshire (4 June) and RIDE Essex (3 September). We conclude with the mighty RIDE Castle in Kent on 1 October. Find out more and sign up at action.org.uk/RIDE

FUNDRAISING NEWS

CYCLESPORT DINNER RAISES £267,000

Some of the biggest names on two wheels helped raise vital funds at our twelfth Champions of Cyclesport Dinner.

Supported by Garmin, BDO and Lucky Saint, the evening saw guests mingling with elite cyclists, including Alex Dowsett, Gee Atherton and Nico Roche, plus rising stars like Josh Tarling, Pfeiffer Georgi and Noah Hobbs.

One of the highlights of the night was when Sophie, pictured, spoke movingly of her battle with epilepsy from the age of 10, sometimes enduring up to 20 seizures a day. When she was 15 she had life-changing brain surgery, using a new technique that was developed with support from Action Medical Research.

For details of the 2023 event visit action.org.uk/champs
As we look forward to the TCS London Marathon in a few weeks’ time, we say a huge congratulations to all those who ran for Action in the 2022 event in October.

Among them was Richard Alexander, who raised over £3,500. Richard’s son James suffered a lack of oxygen during a traumatic birth and received cooling therapy to reduce the risk of brain damage – a breakthrough treatment that Action funding helped to develop.

Richard says: “Luckily James responded well and is now a happy and energetic one year old, thanks to medical research. We want to pay forward the good luck we had and help other families in the future.”

If you’re interested in running in 2024, please visit action.org.uk/running

**ALLIANCE RAISES £91,000**

We have had fantastic support from **Alliance Healthcare** over the last two decades. Staff at the pharmaceutical products company have really got involved, including entering teams for events like Race the Sun and marathons, holding family fun days and selling our Paddington™ pin badges.

**MARKING A MILESTONE**

Our 70th anniversary year concluded last month with a special event – held at London’s Mansion House, where our founder, Duncan Guthrie, held the charity’s first ever fundraising event in March 1953. As well as celebrating research developments over the decades, the event also raised funds for new rare diseases research.

**LONDON CALLING**

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**MOTORSPORT MAGIC**

A return for the Celebration of Motorsports event in Belfast saw over £60,000 raised for Action, with personalities from the high-octane world of motor racing in attendance.

**NEXT TIME IT COULD BE YOU!**

Sharon from London was delighted to win the rollover jackpot in our **FIGHT BACK Friday Lottery**.

“I joined the Action Medical Research lottery to contribute to changing the lives of children,” she says. “I was speechless when I found out I had won £7,000 – I’ll be able to replace my old leaky roof now!”

If you’re not already playing, you can join our lottery today for just £1 a week for the chance to win our weekly prize of £500. You could even be our next Rollover Jackpot winner of up to £10,000!

**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.

1. Rare
2. Henry
3. Pregnancy
4. Atlantic
5. Fifty years
6. Fellowship
7. Batten disease
8. Discovery
9. Clinical trial
10. Vitamin D
11. Facebook
12. Rollover

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
Spring Superdraw
Draw date: 9 June 2023

WIN £500 cash and a £100 luxury hamper!

Don’t miss out... For every 12 entries you purchase we will give you an extra chance of winning a box of treats in our Free Gift Draw!

Other prizes include:
★ 30 runner up prizes of £5
★ Up to £6,700 in our Rollover Jackpot*

Win big and help save little lives – Play today!
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.

Quick reply draw!
Hurry! Reply within 14 days for a chance to win a SMEG espresso coffee machine worth £329.

*In our Rollover Jackpot you have the chance of winning a maximum of £6,700 and minimum £150

Terms & Conditions apply please see action.org.uk/superdraw for details.