action medical research for children

Research Review

Saving and changing children's lives

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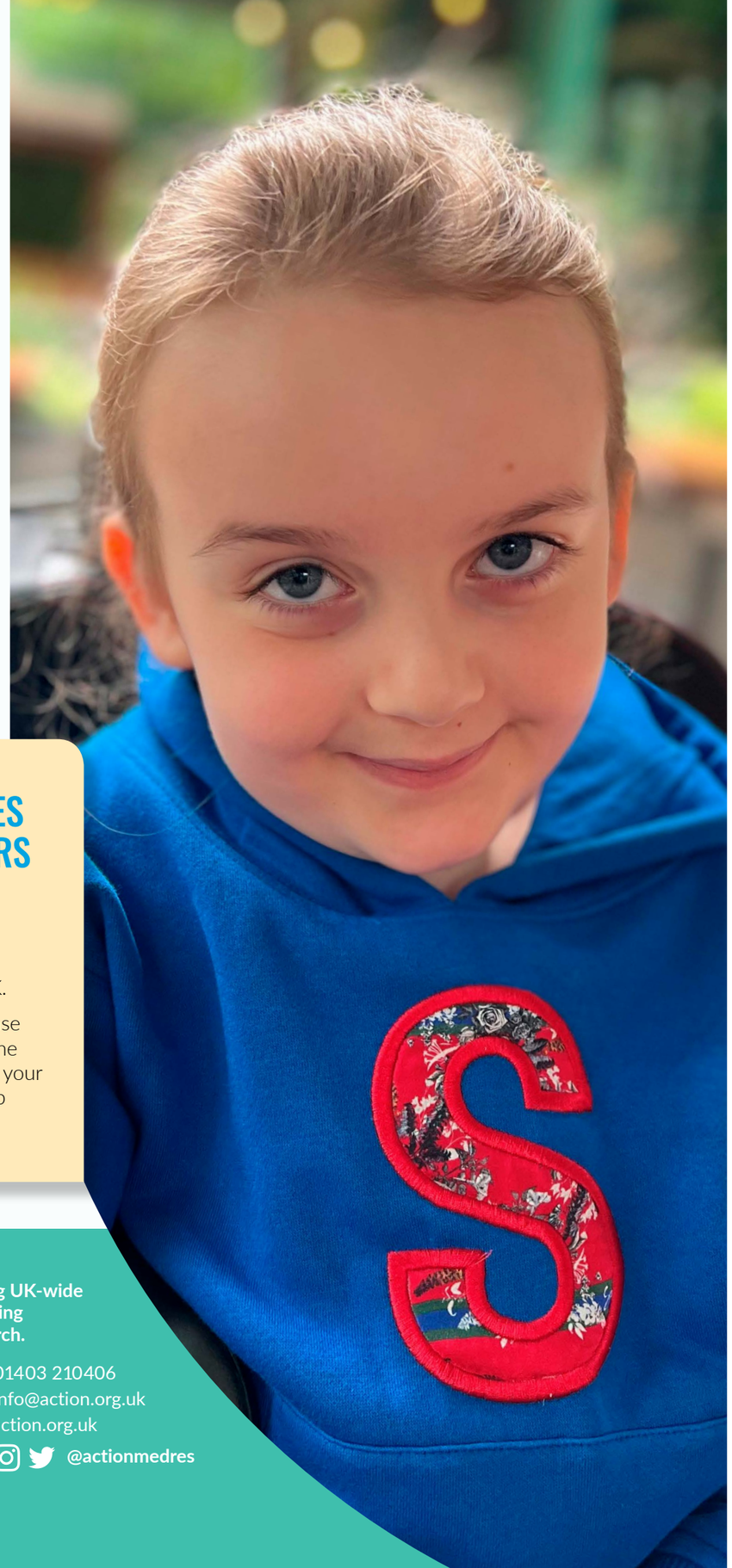

SAVING LIVES FOR 70 YEARS

Our work began in 1952 when the UK faced a deadly disease, polio, and we helped to develop the first polio vaccines in the UK.

Today we continue to fight disease and disability, funding some of the best research in the world. With your support, we can continue to help save and change children's lives.

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

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WELCOME

2022 marked our 70th year as a charity saving lives through medical research. As we funded more vital new research to help many more babies, children and young people in the future, we were proud to reflect on the many successes that Action Medical Research has made possible.

Our charity has been saving and changing lives since 1952, funding over £131m of medical research, in today's terms over £355m.

But there is still so much need for research to help many children and their families. Thanks to our amazing supporters and a successful year of fundraising, in 2022 we were able to invest in more new and important research to help save and change children's lives.

We awarded thirteen new grants across a range of conditions affecting children including brain cancer, epilepsy, preterm birth, congenital heart disease and multiple sclerosis. Our work together with other charities included a joint Research Training Fellowship with the British Paediatric Neurology Association and four new awards in partnership with LifeArc into children's rare diseases.

A new report evaluated the impact of our funding into juvenile Batten disease research leading to an exciting new clinical trial. This rare genetic condition affects the nervous system, with children developing epileptic seizures, losing their sight and the ability to move and speak, before sadly losing their lives in their late teens or twenties.

Other exciting progress included researchers moving a step closer to developing safer and more effective treatments for children with high-risk neuroblastoma and other solid cancers, as well as research to develop a new preventative treatment which could reduce the risk of babies being born with neural tube defects, such as spina bifida.

At Action Medical Research we save and change the lives of babies, children and young people by funding world-class medical research. The difference we make for children depends on the support of our expert scientific panel and many trusts, foundations, companies, individuals and groups. We remain truly grateful to each of our supporters including our network of hardworking local committees and volunteers, as well as those who so generously remember Action with gifts in their wills.

Supporting Action Medical Research makes breakthroughs possible. Please help us in our fight for children's lives.

Julie Buckler
Chief Executive
Action Medical Research for children

FIGHTING RARE DISEASES

Across the UK, there are thousands of children who are living with rare and devastating diseases that have no cure.

Launched in November 2022, our children's rare disease research fundraising campaign shines a spotlight on what is too often an overlooked area of medical research.

Our aim is to support more, urgently-needed, new research into conditions which cause so much suffering for children and their families.

There are 7,000 rare diseases – 75% of these affect children and, sadly, 30% of children with a rare condition will lose their lives before their fifth birthday. For families facing a life-changing diagnosis, medical research gives hope – hope for new treatments, hope for a cure and hope that no other family will have to go through the heartbreak of losing a child to a rare disease.

> "It's only with the support of charities like Action that research happens – and every new development brings hope"
>
> Professor Bobby Gaspar, a pioneer in gene therapy

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

It can also lead to progress in tackling wider health issues – uncovering vital new biological insights, or developing new medical techniques, that can go on to benefit many more people.

HOW WE HELP

Action has a long track record in funding research to better understand rare diseases in children and in helping to develop treatments and cures for them.

In the 1990s, the charity awarded Professor Bobby Gaspar a Research Training Fellowship to study two life-threatening immunodeficiency disorders. Professor Gaspar's pioneering work led to new gene therapy treatments and today he continues to work on life-saving gene therapies for children affected by rare diseases.

He says: "More needs to be done to help find answers and to develop new treatments and cures for these often devastating conditions. It's only with the support of charities like Action Medical Research that research happens. Every new development brings hope for families affected by rare and often life-limiting diseases."

HELPING TO DIAGNOSE RARE DISEASES

In 1985 Dr Michael Baraitser and Dr Robin Winter received funding from Action to help support their work developing a database of information on rare syndromes and genetic diseases. This became the Winter-Baraitser Dysmorphology Database.

This vital tool has continued to be updated and refined ever since and is now part of the London Medical Database, the most comprehensive resource of its kind. Today, it is part of the international Face2Gene library – a suite of apps that doctors can access on any device – and likely to be a first port of call when trying to help a baby or child with an unknown condition.

HOPE FOR CHILDREN LIKE EMMY

Two-year-old Emmy has Vici syndrome, a very rare and life-limiting genetic disorder. There is currently no cure or effective treatment and, 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 to try and move. She is very communicative once you learn her cues and responds to all kinds of therapies and inputs, and she has an especially close bond with her big 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. She 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.

"I gave 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, couldn't grasp."

An MRI scan found significant changes in Emmy's brain. She was diagnosed with septo-optic dysplasia, affecting the optic nerves – meaning, as Ellie feared, she couldn't see. She was also diagnosed with schizencephaly, 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. "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."

"We were blown away – we really didn't think there could be anything so catastrophic wrong. It wasn't until she developed epilepsy, soon after this, that we began to truly understand that this was a very serious diagnosis."

Vici syndrome is one of the most severe multisystem conditions that can affect children, causing a wide range of symptoms.

CURRENTLY, THERE ARE OVER

7,000

KNOWN RARE DISEASES

FIGHTING RARE DISEASES

"We were blown away by such a catastrophic diagnosis"

Emmy's mum, Ellie

Emmy's care is complex and involves multiple medical teams covering different specialities.

Thinking about the future is very daunting. Ellie says: "We hope we'll have longer with Emmy than the worst-case predictions, and that things will be easier and happier. Most of all, we hope that we have a lot of quality time."

"Really, I hope for the same things that all parents hope for – health, happiness and longevity but within a different framework I guess."

NEW RESEARCH

Action Medical Research is funding work to test drug treatments that could help children with this devastating disease.

Vici syndrome is caused by a gene mutation which results in a faulty protein that is key to a biological process called autophagy – a process that is essential for cells to survive and function properly.

Professor Michael Duchen and his team at University College London aim to better understand and target the cellular processes that go wrong, and test drugs that could encourage the repair and improved functioning of cells. This approach could also be beneficial for children with other diseases which involve impaired autophagy.

"This research could be incredibly significant," says Ellie.

GENE THERAPY FOR A RARE LUNG DISEASE

Lung surfactant deficiency can cause severe breathing problems from birth. New research aims to develop a life-saving treatment.

Lung surfactant is a complex mixture that lines the lung tissue and makes breathing easy. Without it, the lungs can collapse, leading to serious breathing difficulties.

Babies born with severe deficiency, caused by faults in the *ABCA3* gene, will struggle to breathe after birth. Sadly, their treatment options are limited and without a lung transplant, less than one in five children will live to celebrate their fifth birthday.

Led by Professor Deborah Gill, based at the University of Oxford, researchers aim to develop a new gene therapy that could be delivered directly into babies' lungs through a ventilator breathing tube. This could lead to a life-saving new treatment and could also help children with milder forms of the disease.

This research is co-funded with LifeArc.

> "This could transform the outlook for babies born with lung surfactant deficiency"

Professor Deborah Gill

> Steps forward

DEVELOPING IMMUNOTHERAPY TO TREAT RARE HIGH-RISK CANCERS

By refining therapies which are currently in clinical trials, researchers have moved a step closer to developing safer and more effective treatments for children with high-risk neuroblastoma and other solid cancers.

Around 100 children are diagnosed with neuroblastoma each year in the UK – and around half are classed as high-risk. These children, who are usually less than five years old, can need extremely intensive treatment lasting many years. Despite this, for some the cancer returns and sadly many lose their lives at a young age.

Action funding has helped Professor John Anderson, at UCL Great Ormond Street Institute of Child Health, to evaluate and refine a new treatment approach, called chimeric antigen receptor (CAR) T cell therapy. This has already proven effective for treating some types of blood cancer and uses a patient's own T cells, part of the immune system, to kill cancer cells.

This study, jointly funded with Great Ormond Street Hospital Children's Charity and Neuroblastoma UK, builds on previous work and Professor Anderson says: "We have successfully developed solutions that may be implemented into the next clinical trials of this cutting-edge immunotherapy. These refinements are designed to lead to more effective and kinder treatments in upcoming clinical trials."

SAVING TINY LIVES

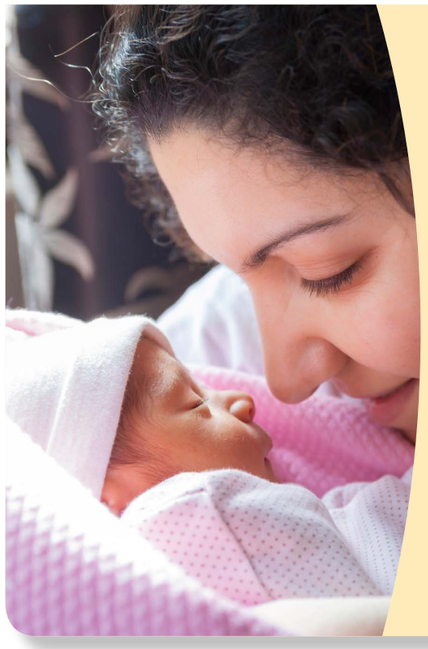
In 2022, we continued to fund new research that aims to give babies the best possible start in life.

Around one in seven babies born in the UK needs some form of special care as a result of a difficult birth, a life-threatening condition, or because they were born too early.

We continue to fund research that aims to reduce the high rate of premature birth, advancing the search for new treatments that could prevent or delay early birth.

Other new work aims to ensure sick and vulnerable babies receive the best possible care, helping to develop the latest techniques and technologies.

We also continue our heritage of investing in research to prevent pregnancy complications and improve child health for future generations.



SAVING LIVES FOR 70 YEARS

A HISTORY OF TRANSFORMING PREGNANCY CARE

In the 1960s, 70s and 80s Action supported research that led to developments that are now a standard part of pregnancy care in the UK.

We supported a 20-year programme looking at whether vitamin supplements taken before and during pregnancy could help prevent spina bifida. Today, taking folic acid is routinely advised and the number of babies affected by spina bifida in the UK has dramatically reduced.

Action also funded research that helped to develop the use of ultrasound technology to monitor a baby's development and diagnose problems before birth. Thanks to ultrasound, the death rate for babies at the time of birth is estimated to have halved.

VITAMIN D IN PREGNANCY

Vitamin D is already known to be essential for good health – new research could lead to a change in pregnancy guidelines to benefit future generations of children.

Our bodies make vitamin D from sunlight on the skin, and it is especially important for healthy bone and muscle function. But low levels are common in the UK, as many people don't get enough time in the sun.

Pregnant women are already advised to take a daily supplement to help protect against certain pregnancy complications. But research has shown that increasing this dose could offer additional, long-term health benefits for children as they grow up.

A previous study found that children whose mothers took additional vitamin D had stronger bones and muscle at age four, and less fat.

With Action funding, Dr Rebecca Moon and her team are continuing this work: "We plan to establish if differences seen at age four are still present at adolescence – a critical age for bone development due to puberty," she says.

Ultimately, this could lead to a change in national guidelines.

> Steps forward

PREVENTING DISABLING NEURAL TUBE DEFECTS

Research to further reduce the risk of babies being born with severe neural tube defects, such as spina bifida, has progressed towards a clinical trial which could have global impact.

Neural tube defects affect a baby's brain, spine, or spinal column. Taking folic acid during pregnancy helps to reduce the risk, but sadly some babies are still affected by these conditions.

Professor Nicholas Greene, at the UCL Great Ormond Street Institute of Child Health, has been investigating if taking a vitamin called inositol can also help.

The findings of this work are now informing the design of a new clinical trial to test how well inositol works, over and above the protection currently provided by folic acid. If positive, this could lead to a cheap and effective new approach within the next five years and have worldwide impact.

> "Our hope is that this will mean fewer parents receiving heart-breaking news about their baby"
>
> Professor Nicholas Greene

HELPING SICK BABIES

Babies born extremely prematurely are at risk of dying or developing a serious lung condition in their early weeks. New research is using artificial intelligence to identify which babies are at greatest risk and guide better treatment.

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

Medicines can be given to help prevent this. But these can have serious side effects, such as cerebral palsy – so doctors must balance the potential risks and benefits.

Dr T'ng Chang Kwok, of the University of Nottingham, aims to create a new diagnostic tool to support clinical decision-making for these vulnerable babies.

His team is analysing routinely recorded data within the electronic medical records of many thousands of very preterm babies. The aim is to develop an AI-based system that can accurately recognise complex patterns within these data and guide treatment decisions.

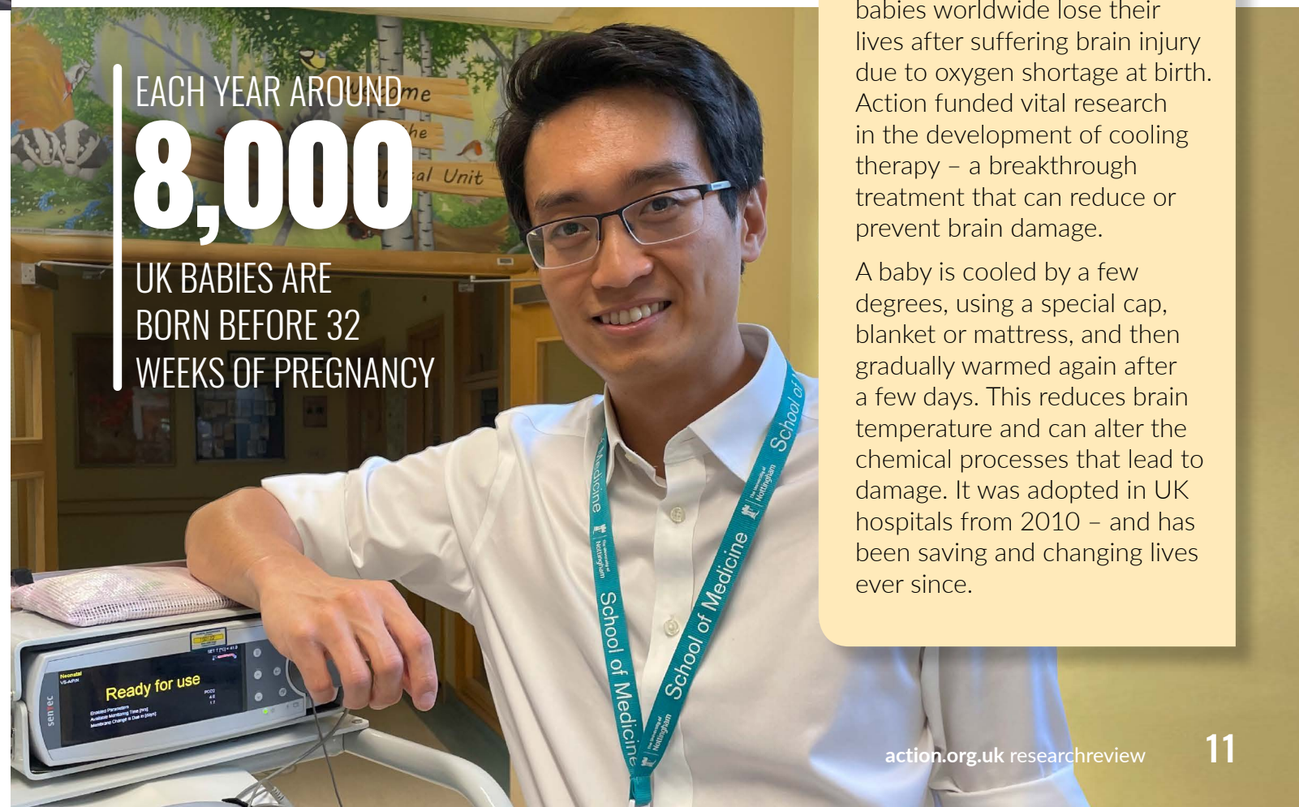
"Predicting who is most likely to benefit from treatment, and when, would give each child the best chance of a successful outcome," says Dr Kwok.

Thank you to the Albert Gubay Charitable Foundation for their generous support of this research.

COOLING THERAPY FOR NEWBORNS

Each year, almost a million babies worldwide lose their lives after suffering brain injury due to oxygen shortage at birth. Action funded vital research in the development of cooling therapy – a breakthrough treatment that can reduce or prevent brain damage.

A baby is cooled by a few degrees, using a special cap, blanket or mattress, and then gradually warmed again after a few days. This reduces brain temperature and can alter the chemical processes that lead to damage. It was adopted in UK hospitals from 2010 – and has been saving and changing lives ever since.



PREVENTING PREMATURE BIRTH

When Jack was born more than three months early, his parents were told he had a fifty-fifty chance of surviving. After such a traumatic start, he has made remarkable progress. But he has continued to face challenges as he grows up.

After a seemingly easy and problem-free pregnancy, Jack's arrival more than three months early was a sudden and terrible shock for Jenny and husband Matthew.

At 25 weeks, Jenny had started bleeding and was rushed into hospital. Though she was concerned, she could still feel Jack kicking so thought things would settle. "I thought that once the bleeding stopped, we'd be going home," she recalls. "But that didn't happen. I was already in early labour, and the doctors decided to do an emergency caesarean."

Jack was born weighing just 1lb 15oz. He was transferred straight away to another hospital to receive the specialist care he needed in neonatal intensive care. Jenny, who had been under general anaesthetic, was taken by ambulance to join him, 12 hours later.

"When you come round, you expect to have your baby with you," says Jenny. "But, of course, he wasn't there. When I did finally see him, his skin was transparent and he was so delicate I couldn't hold him."

Jack was extremely vulnerable and needed oxygen, help with his breathing and feeding tubes to survive. He suffered from a collapsed lung and brain bleeds in his first few days of life.

Then, when he was six days old, he developed necrotising enterocolitis (NEC), a life-threatening infection of the bowel. Jack was transferred again to another hospital for urgent treatment. "It was the most traumatic thing," Jenny remembers. "The staff said they'd never seen a baby so small."

Jack underwent life-saving surgery, during which a piece of his bowel was removed – the first of seven operations during his first six months of life.

Thankfully, he recovered from each setback and after four and a half months in hospital baby Jack was finally allowed home. "He was still on oxygen, so the canisters came too," says Jenny. "We were just so relieved to be home."

> "Without research, we wouldn't have medical advances and Jack probably wouldn't be here"
>
> Jack's mum, Jenny

AROUND

55,000

BABIES ARE BORN PREMATURELY IN THE UK EVERY YEAR

Jack caught up with his weight in his first year but has chronic lung disease due to the oxygen he needed in his early months. He has also experienced some developmental delay, both with his speech and physically, and has been diagnosed with mild cerebral palsy. The family do a lot of physiotherapy at home to help support him.

But Jenny says that nothing phases Jack: "We are so lucky to have him. There's no stopping him!"

Jenny is acutely aware of the value of medical research: "Without research, we wouldn't have medical advances – and Jack probably wouldn't be here," she says.

Jack's little sister, Alice, was also born prematurely, so finding out what causes premature labour is especially important to Jenny: "Anything that helps identify why women go into labour too soon is vital," she says.

NEW RESEARCH

A major barrier to preventing premature birth is that we do not yet fully understand how the onset of labour is triggered.

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.

With co-funding from Action Medical Research and Borne, the team now aims to determine whether these cells trigger full-term labour – and whether these cells are also involved in spontaneous preterm labour.

"By understanding how these cells are involved, future studies might be able to target them to reduce rates of premature birth," says Dr Male.

These findings could ultimately lead to happier outcomes for many babies and their families.

HELPING CHILDREN WITH DISABLING CONDITIONS

IMPROVING RECONSTRUCTIVE SURGERY

Living with a facial disfigurement can be especially difficult for children as they grow up. Action Research Training Fellow Mrs Cynthia de Courcey is working on an approach that could transform reconstructive surgery in the future.

Children with facial disfigurements, either because they were born with missing or malformed features, or affected by trauma, burns or cancer, often experience long-term emotional and/or physical problems.

While surgery is an option, existing approaches can be painful – often taking tissue from other parts of the body – or carry high risks of infection and rejection.

At Swansea University, Mrs Cynthia de Courcey is part of a team that is developing 3D-bioprinting technology. This would allow surgeons to create precise, patient-specific implants that are loaded with a child's own cells, helping to overcome many of the challenges associated with current procedures.

"This would revolutionise reconstructive facial surgery for children, having a positive impact on many young lives," says Mrs de Courcey.

This Research Training Fellowship is supported by the VTCT Foundation.

SAVING LIVES FOR 70 YEARS

A UNIQUE SEATING SYSTEM TO SUPPORT DISABLED CHILDREN

Over 30 years ago, Action funding helped to create the award-winning Matrix seat for severely disabled children. It is still in use today and more recent modifications mean it may even help to correct spinal deformity in children with conditions such as muscular dystrophy.

With support in the early 1980s, researchers developed the 'Matrix' system from prototype, through to development and clinical trials and on to international commercial use.

The versatile design can be fitted closely to each unique body shape. Its shape can also be changed as needed by adjusting the links which make up the web-like structure. This was not possible before and is particularly beneficial for growing children.

> Steps forward

NEW SCANS TO HELP CHILDREN WITH ARTHRITIS

Children and young people with juvenile idiopathic arthritis are set to benefit from improvements in their treatments and quality of life, following Action-funded research.

Juvenile idiopathic arthritis causes inflammation in the joints, leading to pain, joint deformity and disability. The latest drug treatments can help but they are expensive and can have serious side effects.

With Action funding, Professor Margaret Hall-Craggs and her team at University College London have developed new techniques, using MRI scans, to precisely measure how much inflammation is present in patients' joints. Treatments can then be adjusted, so that just the right amount is given.

The team has completed the largest ever study of whole-body imaging in young patients and shown that these scans could alter treatment and management decisions in up to a third of cases.

Professor Hall-Craggs says: "The impact of this Action funding has been to support work in adolescents who have a chronic illness. This is an area that is under-investigated and under-funded."

The Albert Gubay Charitable Foundation helped to support this research.

> "This is already helping to optimise treatment for young people with a chronic, lifelong condition"
>
> Professor Margaret Hall-Craggs

TREATING SEVERE EPILEPSY

New research is set to investigate if a new type of brain scan could enable more children to benefit from life-changing surgery.

Epilepsy affects around 70,000 babies, children and young people under 18 years old in the UK. Sadly, medications don't work for up to one in three, meaning their lives can be difficult and unpredictable.

Many children with severe, drug-resistant epilepsy have abnormal areas of brain tissue known as focal cortical dysplasia (FCD). Surgery to remove the affected tissue can be an option for some, but doctors can only consider this if the affected area can be detected using scans.

With Action funding Professor Chris Clark, of UCL Great Ormond Street Institute of Child Health, will investigate whether an advanced new scanning technique can help to improve detection of FCD.

"Surgery offers the possibility of a cure but unfortunately, some children may be missing out on the opportunity to have potentially life-changing treatment due to the current limitations of MRI scans," says Professor Clark.

> "This could open up the possibility of transformative surgery to more children"
>
> Professor Chris Clark

HELPING YOUNG PEOPLE WITH MS

Andrada was diagnosed with multiple sclerosis in 2022, aged 16. She is now taking part in new Action-funded research to help children and young people with this debilitating condition.

Multiple sclerosis (MS) affects the brain and nerves, and can cause a wide range of symptoms, including problems with vision, movement, sensation or balance. While no two people will have the same range and severity of symptoms, unfortunately the condition is lifelong and there is no cure – and while most are diagnosed between the ages of 20 and 40, the number of young people being diagnosed under the age of 18 is increasing.

Andrada was just 14 years old when she first began to experience symptoms of MS, which affected her eyesight: "I was seeing double, which was making it really hard to carry out simple tasks," she recalls.

Her concerned parents took her to hospital – and the family were shocked when doctors discovered that her optic nerve was paralysed. Andrada underwent three days of intensive treatment, and the following months involved many hospital visits and tests to investigate the cause – but the teenager would often leave frustrated and knowing nothing new.

Ambitious and determined, Andrada is a high achiever and has always performed well academically. But over time she started to feel increasingly unwell. Concerns grew about her frequent illness and following an MRI test, Andrada was referred to a specialist at Addenbrooke's Hospital in Cambridge. Here she received the diagnosis she and her family were dreading – she has MS.

As time moves on, Andrada is starting to experience extreme fatigue and migraines. "I'm currently studying for my GCSEs but struggling with the revision," she says. "I find it hard to remember information as easily as I used to. I feel like I am falling behind and not able to keep up as well as I used to. I am so grateful I have a supportive family, friends and school but I sometimes feel isolated."

"I was always a very energetic person, but now struggle with severe tiredness. I have more migraines too. They even make me faint sometimes. I always loved socialising and playing sports like tennis, but I have to be careful I don't expel too much energy and impact my studies and health," she says. "That said, I am determined not to let MS define me – I am trying to live with it as best I can and do everything possible to live a 'normal life'."

Speaking about the importance of research, Andrada says: "I am pleased to be part of this and want to help in any way I can. There is no cure for MS and research is so important to help people like me who are facing an uncertain future."

> "There is no cure for MS and research is so important to help people like me who are facing an uncertain future"
>
> Andrada

FIVE IN EVERY 100

PEOPLE WITH MS EXPERIENCE THEIR FIRST SYMPTOMS BEFORE THEIR EIGHTEENTH BIRTHDAY

NEW RESEARCH

With support from Action Medical Research, Dr Jonathon Holland, of Addenbrooke's Hospital in Cambridge, is studying nerve repair in children with MS to see if they could in the future benefit from new treatments currently being tested in adults.

MS is caused by the body's immune system mistakenly attacking a protective layer called the myelin sheath that surrounds and protects nerve cells in the brain and spinal cord. The body can repair some damaged myelin, but this can be impaired in people with MS. New treatments that promote the repair of myelin could help slow down disease progression.

"We suspect that this repair process is more effective in children with MS than adults, which would indicate that these new treatments might also be helpful for them," says Dr Holland.

This Research Training Fellowship is jointly funded with the British Paediatric Neurology Association.

MAKING AN IMPACT

Izzy, now aged 14, has juvenile Batten disease

BATTEN DISEASE BREAKTHROUGH

Action Medical Research funding supported work that has revealed vital new insights into this devastating and life-limiting rare disease. Crucially, this has led to clinical trials of a new treatment where previously there were none.

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.

They go on to experience problems with movement, and the loss of other mental and physical abilities.

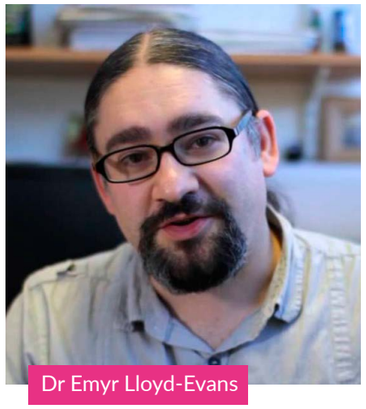
> “We’re really excited about the opportunities to translate these discoveries into new treatments”
>
> Dr Emyr Lloyd-Evans

There is currently no cure and no way to stop or slow down the disease. Tragically, children become severely disabled before losing their lives in their late teens or twenties.

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 previously there were none.



Dr Emyr Lloyd-Evans

Making a major breakthrough, this work revealed, for the first time ever, what CLN3 does – it acts as a transporter for potassium within the body’s cells. Having this knowledge 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 clinical trials of the first ever drug treatment for juvenile Batten disease – with results from the Action-funded work providing vital evidence to support the planning and approval of these trials. The first of these, in young people aged 17 or over, is already underway in the US and showing very promising results. A further trial, testing the treatment in a larger group of US patients, is due to start during 2023.

This work brings much-needed hope to families affected by this cruel disease.

A FAMILY’S FIGHT

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 and a few weeks later tests showed that their little sister Izzy, aged just six, was also affected.

“The diagnosis left us numb,” says Dee. “My first thought was, no – my babies are going to die.”

“You think it just can’t be happening, it can’t be real,” adds dad Jody.

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.

In the years since being diagnosed, all three children have slowly lost their mobility. Their sight is severely impaired and other symptoms have included pain, behavioural problems, night terrors, short-term memory loss and stammering. Izzy is also now affected by dementia.

Sadly, the family know that new treatments are unlikely to come quickly enough for them, but they are hopeful for others in the future.

> “We hope that in the future other families won’t have to go through what we have”
>
> Parents Dee and Jody

RESEARCH GRANTS AWARDED IN 2022

Action Medical Research is currently funding more than 50 projects across the UK, 13 of them awarded in 2022.

The next medical breakthrough could be on your doorstep.

Congenital heart disease – predicting the outcomes of heart surgery
Dr Adelaide de Vecchi
King's College London, Evelina London Children's Hospital, Guy's and St Thomas' NHS Foundation Trust, University of Leeds

Developing cutting-edge gene therapy for severe childhood epilepsy*
Professor Dimitri M Kullmann
University College London

Developing a new gene therapy for babies born with a rare severe lung condition*
Professor Deborah R Gill
University of Oxford

Developing a new treatment for high-risk brain cancer in children*
Professor John Anderson
UCL Great Ormond Street Institute of Child Health

Improving facial reconstructive surgery for children using tissue engineering**
Mrs Cynthia H K de Courcey
Swansea University

Multiple sclerosis – assessing nerve repair in children to find out if they could benefit from new treatments***
Dr Jonathon AA Holland
Addenbrooke's Hospital, Cambridge University NHS Foundation Trust, University of Cambridge

Understanding how labour starts – to uncover new ways to prevent spontaneous preterm birth****
Dr Victoria Male
Imperial College London

Preterm birth – reducing the risk of babies dying or developing a severe lung condition using a new AI-based tool*****
Dr T'ng C Kwok
University of Nottingham

Primary ciliary dyskinesia – searching for drugs to treat this debilitating, rare lung condition
Professor Chris O'Callaghan
UCL Great Ormond Street Institute of Child Health, University College London, University of Leicester

Severe epilepsy – could a new type of brain scan enable more children to benefit from life-changing surgery?
Professor Chris A Clark
UCL Great Ormond Street Institute of Child Health

Improving the genetic diagnosis of rare childhood developmental disorders*
Professor Matthew E Hurles / Dr Elizabeth J Radford
Wellcome Sanger Institute, University of Cambridge, Addenbrooke's Hospital

Vici syndrome – investigating drug treatments
Professor Michael R Duchen
University College London

Vitamin D deficiency during pregnancy – could additional supplementation provide long-term benefits for children's health?
Dr Rebecca J Moon
University of Southampton, Southampton General Hospital

* Jointly funded with LifeArc
** Supported by the VTCT Foundation
*** Jointly funded with the British Paediatric Neurology Association (BPNA)
**** Jointly funded with Borne
***** Supported by the Albert Gubay Charitable Foundation

Locations of current research funded by Action

A LASTING LEGACY

Gifts in wills are a vital source of income to us and we are incredibly grateful to those people who choose to show their support in this special way. A gift in your will could unlock a future medical breakthrough. It could help us find new treatments and cures that could change children's lives for years to come.

Thank you to those who have already made provision for Action in their will and to those who left us a legacy gift in 2022:

Laura Bisset
Ann Caldwell
Alan Chisholm
Marion (Edith) Collins
Peter Cursons
Margaret Eastwell
Gladys Fillary
Wendy Harrup
Gertrude Palmer
Joan Watson
Audrey Whaite
Peter Wilkinson
Marjorie Wurr

"Aiden is getting a chance at life thanks to Action's research. We want to ensure others get the same chance in the future, so we've decided to leave a gift in our wills."

Keith and Fleur's son Aiden was treated with cooling therapy after a traumatic birth, a life-saving treatment that Action funding helped to develop.

If you would like to find out more about leaving a gift in your will to Action, please contact Gill on **01403 327413**, email **legacy@action.org.uk** or visit **action.org.uk/giw**

SUMMARISED FINANCIAL STATEMENTS

for the year ended 31 December 2022

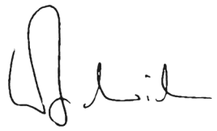
Report by the trustees on the summarised financial statements

The summarised financial statements below are extracted from the full trustees' annual report and financial statements, which were approved by the trustees and signed on their behalf on 27 April 2023. The full financial statements, on which the auditor, Buzzacott LLP, gave an unqualified audit report on 10 May 2023, was submitted to the relevant statutory bodies, including the registrar of companies, on 16 May 2023.

The auditor has confirmed to the trustees that, in their opinion, the summarised financial statements are consistent with the full financial statements for the year ended 31 December 2022.

These summarised financial statements may not contain sufficient information to gain a complete understanding of the financial affairs of the charity. The full trustees' report, financial statements and auditor's report can be viewed on the Action Medical Research website at action.org.uk/FS

Signed on behalf of the trustees



Luke Bordewich Chair

Income and expenditure	2022	2021
	£000s	£000s
Net income		
Donations and legacies	2,097	1,899
Investments	(31)	(11)
Charitable grants	468	–
Other income	–	4
Net income before charitable activities	**2,534**	**1,892**
Charitable activities		
Medical research projects	2,930	2,114
Medical dissemination	716	669
Net expenditure	**(1,112)**	**(891)**
Net gains on investments	850	1,051
Net movements in funds	**(262)**	**160**

Balance sheet	31 Dec 22	31 Dec 21
	£000s	£000s
Fixed assets		
Tangible	283	302
Investments	12,113	12,496
Total fixed assets	12,396	12,798
Current assets	1,678	1,186
Current liabilities within one year	(5,721)	(5,530)
Liabilities falling due after one year	(1,811)	(1,650)
Total net assets	6,542	6,804
Representing:		
Unrestricted funds	6,542	6,804
Restricted funds	–	–
Total funds	**6,542**	**6,804**

THANK YOU

We are always hugely grateful to the many individuals, companies, trusts and foundations who so generously donate to the vital work supported by Action Medical Research.

We would also like to express our thanks to the organisations listed for their contributions and involvement with the charity.

Supporters

Alison Hillman Charitable Trust
Alliance Healthcare
Anne and John Walters Charitable Trust
AON
BDO
BGC Partners
The Big Give Trust
Boongate Kia
Brett Group
Bridgehead Software
Brit Insurance
Chaucer Group
The Chyah Davies Foundation
Day Group
Finning
Garmin
The Gingerman Restaurants Group
Grafton Group
The Hobson Charity
Hospital Saturday Fund
Ki
Lendlease
Liberty Specialty Markets
Lucky Saint
Maylim
Milton Damerel Trust
Next Retail
Oso Foundation
Paddington at Paddington Station
Peak Scientific
P E Lennard Charitable Will Trust
The Peter Sowerby Foundation
The Rooney Foundation
Rosetrees Trust
Rouleur
Scrap Car Comparison
Smeg
Touchstone Underwriting
TSML
Willmott Dixon

WHO'S WHO 2022

Former Royal Patron

HRH The Prince Philip Duke of Edinburgh KG KT OM GCVO GBE ONZ QSO AK GCL CC CMM (deceased)

President

Field Marshal The Lord Guthrie GCB LVO OBE DL

Vice Presidents

Phil Hodkinson
Stephen May
Richard Price
The Duchess of Northumberland

Trustees

Luke Bordewich, Chair
Richard Wild, Honorary Treasurer
Esther Alderson retired July 2022
Professor David Edwards MA MBBS DSc MRCR FRCP FRCPCH FMedSci
Kathy Harvey
Karen Last
Professor David Rowitch MD PhD ScD
Professor Stephanie Schorge PhD appointed October 2022
Rajat Sharma appointed November 2022
Richard Stoneham-Buck

Scientific Advisory Panel

Professor David Rowitch MD PhD ScD, Chair retired September 2022
Professor Stephanie Schorge PhD, Chair from October 2022
Professor Claire Booth MBBS MSc PhD MRCPCH
Professor Yanick Crow MBBS MRCP CCST PhD FMedSci retired May 2022
Professor Jonathan Grigg BSc MBBS MD FRCPCH retired May 2022
Professor Alicia El Haj FREng FRSB FEAMBES
Professor Muzlifah Haniffa BSc MBBCh MRCP PhD FMedSci from March 2022
Professor Catherine Hawrylowicz PhD retired February 2022
Professor Samantha Johnson PhD CPsychol AFBPsS retired October 2022
Professor Ed Johnstone MBChB PhD MRCOG from October 2022
Professor Andrea Nemeth BSc MBBS DPhil (Oxon) FRCP
Dr Adam Shortland BSc PhD MIPEM CSci retired October 2022
Professor Sarah Stock BSc MBChB PhD joined Feb 2022 and retired May 2022
Professor Shiranee Sriskandan FRCP PhD
Professor Michael Taggart BSc PhD retired December 2022

Scientific Advisory Panel for joint Action Medical Research/LifeArc project grant applications on rare disease research

Professor David Rowitch MD PhD ScD, Chair retired September 2022
Professor Stephanie Schorge PhD, Chair from October 2022
Professor Claire Booth MBBS MSc PhD MRCPCH
Professor Paul Brennan PhD
Professor Yanick Crow MBBS MRCP CCST PhD FMedSci retired May 2022
Professor Alicia El Haj PhD FREng, FRSB, FEAMBES
Dr Thorsten Forster PhD
Professor David Gray PhD retired May 2022
Professor Jonathan Grigg BSc MBBS MD MRCP FRCPC retired May 2022
Professor Muzlifah Haniffa BSc MBBCh MRCP PhD FMedSci joined March 2022
Professor Catherine Hawrylowicz PhD retired February 2022
Professor Samantha Johnson PhD, CPsychol, AFBPsS retired October 2022
Dr Andy Merritt PhD CChem Joined July 2022
Professor Andrea Nemeth BSc MBBS DPhil (Oxon) FRCP
Dr Adam Shortland BSc PhD MIPEM CSci retired October 2022
Professor Michael Taggart BSc PhD retired December 2022

THANK YOU

action medical research for children

We're the leading UK-wide charity dedicated to funding vital research to help sick babies and children.

A member of the: amrc ASSOCIATION OF MEDICAL RESEARCH CHARITIES

Registered with FUNDRAISING REGULATOR

FSC www.fsc.org MIX Paper from responsible sources

Front cover photo: Ben Rector