action medical research for children

Research Review 2018

Saving and changing children's lives

Welcome

At Action Medical Research we fight for children's lives, funding groundbreaking research to help find answers. We funded 16 new projects in 2018 and today have £11m of research underway across the UK, involving 260 top researchers.

We make this investment confident that the research we fund makes such a positive difference for children and their families.

Last year we saw the impact made in improving treatment for children affected by dystonia, helping them to walk and go to school.

Exciting progress also included: a breakthrough in understanding juvenile Batten disease, with two potential new treatment strategies that could lead to a clinical trial this year; a new app to help teenagers with ADHD; and a new eye-pointing scale for severely diasbled children.

In the 45th year of our Research Training Fellowship scheme, which develops future research leaders, we funded our 177th Fellow, researching gene therapy for mitochondrial disease.

We also launched our BORN TOO SOON campaign, shining a spotlight on an important area of our work, premature birth. And we worked in partnership with other charities, launching a relationship with LifeArc to fund up to £1m of research this year to develop treatments for children with rare diseases.

Our achievements are only possible with the support of many individuals, groups and organisations. We are grateful to everyone, not least our hardworking network of local committees and volunteers, as well as those who kindly remember Action with gifts in their wills.

We're on a mission to save and change children's lives through medical research. Please join us and help fund more research that could make such a difference.

Julie Buckler
Chief Executive
Action Medical Research

Contents

Helping babies born too soon

Millie and Scarlett's story

In 2018 our BORN TOO SOON campaign was launched, shining a spotlight on the devastation caused by premature birth and the complications that can follow. Millie and Scarlett's story highlights why this is so important.

Millie and her twin sister Scarlett were delivered by caesarean section 28 weeks into a very difficult pregnancy. They had been affected by a condition called fetal growth restriction, which meant they were growing dangerously slowly. Babies with this condition often need to be delivered early, so it is also a cause of premature birth.

Scarlett arrived first, weighing just one pound 6oz, followed by Millie weighing a tiny one pound 4oz. To their parents Emma and Dan's immense relief, both girls cried as soon as they were born. But both were incredibly vulnerable, and too small and fragile to be held. They needed help to breathe and were not yet ready for milk, so were given fluids by tube.

Millie, the smaller twin, was stronger and was soon able to take tiny amounts of expressed milk, given by syringe down a tube. Scarlett sadly couldn't, and she also continued to need a great deal of support with her breathing.

Around 60,000
babies are born prematurely every year in the UK

Then, at three weeks old, Scarlett's already fragile health deteriorated rapidly. She developed necrotising enterocolitis (NEC), a life-threatening bowel condition.

When it became clear that Scarlett could not survive, Emma and Dan had to face the heartbreaking loss of their tiny daughter. "They told us we could hold her until they switched off life support," says Emma quietly. "We held her in our arms, on a cushion as she was so fragile."

Then, terrifyingly, Millie too became seriously ill with NEC. She would need a life-saving operation – surgery that was performed immediately on the ward, rather than delay by moving to an operating theatre. "It was the worst wait ever," recalls Dan. "The eventual knock on the door was so scary."

Thankfully the news was positive. Surgeons had successfully removed a small amount of bowel, repaired it, and given Millie a stoma bag to manage her body's waste.

After three and a half months, Millie was well enough to go home and her stoma was later reversed. She is now a happy six-year-old who loves school and is doing incredibly well.

> "We are so blessed to have our Millie, but saying goodbye to Scarlett was just devastating"
>
> Millie and Scarlett's dad, Dan

Emma and Dan are now passionate about medical research to help babies like Millie and Scarlett. "If research wasn't done, doctors wouldn't have the knowledge that they do now, and the ability to treat tiny babies who are born prematurely or get conditions like NEC," says Dan. "NEC comes so quick, it's a shock. We just hope research will find a way to stop it or catch it sooner."

New research to protect premature babies

Action funding is supporting research to find new ways to protect premature babies from a serious bowel disease, necrotising enterocolitis, and sepsis.

Every year in the UK around 10,000 babies are born before 32 weeks of pregnancy. Up to 4,000 of them develop either necrotising enterocolitis (NEC), the serious bowel disease that Millie and Scarlett had, or sepsis, a life-threatening complication of infection.

After the first week of life, these two conditions are the biggest threats to very premature babies, although they can also strike babies born after 32 weeks. NEC is also the most common reason for emergency surgery in newborn babies.

Despite their impact, NEC and sepsis are still poorly understood but they are thought to be linked to imbalances in the community of micro-organisms living inside a baby's gut.

Breast milk is known to reduce the risk of NEC and sepsis in premature babies, so researchers at the University of Northumbria in Newcastle are studying it to better understand which components provide these protective effects and how they work to support babies' inexperienced immune systems. They also want to determine how these protective components are carried in the milk.

This could shed new light on how to prevent and treat these illnesses in the future. It could also have implications for the handling and storage of breast milk in special care baby units.

> "Too many lives are being lost and babies who do survive often grow up with life-changing disabilities"
>
> Lead researcher, Dr Darren Smith

Steps Forward

Predicting the risk of premature birth

In 2018 researchers reported back on exciting progress made towards developing a test to identify women at higher risk of going into labour too soon.

Previous Action funding had already helped discover that women who lack white blood cells at the opening to the womb were more likely to give birth prematurely. These cells are a key part of the body's immune defences.

15 million babies worldwide are born prematurely every year

Further Action funding allowed Professor Nigel Klein and his team at the UCL Great Ormond Street Institute of Child Health to continue this work. They aimed to simplify the method of detecting cells so that it could be developed into a test for use during early pregnancy.

The team identified four biomarkers that when absent or reduced in cervical fluid greatly increase the chances of delivery before 34 weeks of pregnancy if left untreated. They are now seeing how well this new biomarker test performs on samples taken from 400 women.

Thank you to Dangoor Education for generous support of this project.

> "I had no idea how serious pre-eclampsia could be. Awareness and research are so important"
>
> Jacob's mum, Rachel

Jacob was born 10 weeks early after Rachel developed pre-eclampsia

Steps Forward

Shedding new light on pre-eclampsia

Pre-eclampsia endangers the lives of both mother and child. Research funded by Action made national headlines in 2018, showing that it could be linked to the mother's heart function prior to pregnancy.

Pre-eclampsia is diagnosed by high blood pressure in the mum-to-be and can become life-threatening. Sadly around 1,000 UK babies die each year as a result, mainly because they have to be delivered prematurely.

With Action funding awarded in 2016, researchers at Imperial College London, led by Professor Christoph Lees, tracked the health of more than 200 women before and during pregnancy. All were outwardly healthy, with normal weight and blood pressure. However, differences were found in the pre-pregnancy heart function and circulation of those who went on to develop either pre-eclampsia or another complication called fetal growth restriction.

Although technically within normal range, these women's hearts pumped less blood per minute than those who had uncomplicated pregnancies, and their blood vessels were more resistant to blood flow, causing blood pressure, before pregnancy, to be higher.

Future research could now look at the potential for screening to identify those at risk, and whether lifestyle changes made before pregnancy can help.

Cerebral palsy

Helping children like Alice

Like many two-and-a-half-year-olds, Alice loves to be the centre of attention. But sadly she faces a lifetime of challenges as her movement is severely impaired by cerebral palsy.

When Alice was born two months prematurely, an ultrasound scan showed no signs of brain damage. All had seemed well, but at her six week check-up, the consultant delivered unexpected and devastating news – Alice had cystic masses on her brain, indicating severe and irreversible damage.

Alice was diagnosed with a condition called periventricular leukomalacia. Cerebral palsy is the most common symptom and Alice's body and all her limbs are affected.

Despite initial fears that she wouldn't, Alice can now speak and her intellect is not affected. "She is a bright and happy child but she gets so frustrated because she can't walk, crawl or even sit up without help," says her mum, Jenny.

> "Anything that gives us more ways to help our children and help families to manage is really important" Alice's mum, Jenny

Family life now centres around physiotherapy and various appointments. "But without all of this help, Alice wouldn't be where she is now," says Jenny.

Research projects to help children with cerebral palsy

Action funding is developing new treatments and therapies to give children the best possible quality of life.

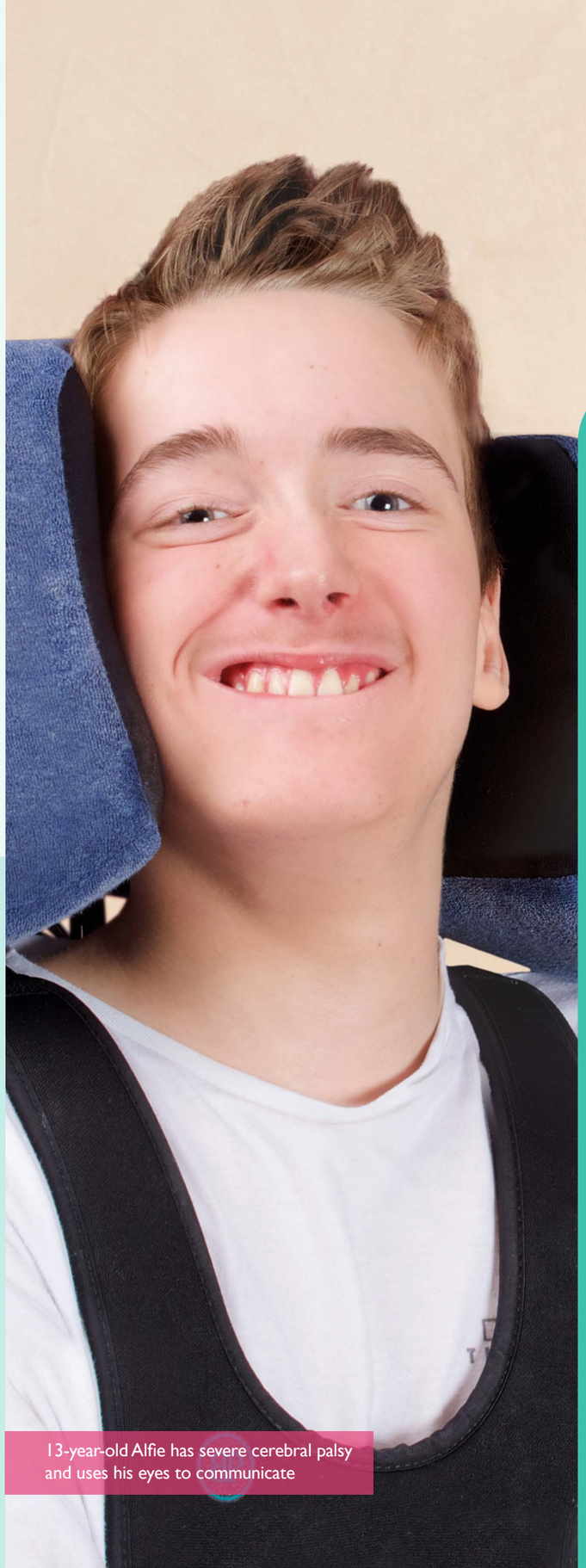
Cerebral palsy is the most common serious physical disability in children and is caused by brain damage that occurs before, during or soon after birth.

At Newcastle University, Action-funded researchers are developing and testing a child-friendly, wrist-worn device and smartphone app to help children with hemiplegic cerebral palsy, which affects one side of the body. The hand and arm are often severely impaired by this, so children favour their other limb, worsening the weakness. This device aims to increase activity in the affected arm to improve strength and movement.

Action is also funding a UK-wide study that aims to ensure children with bilateral cerebral palsy, affecting both legs, receive the best possible treatment. Led by a consultant orthopaedic surgeon, this team is assessing the effectiveness of an operation called single event multi-level surgery, used to correct muscle and bone deformities and help children's walking.

And researchers at Queen Margaret University, near Edinburgh, and at Brunel University are testing ways to increase exercise for children and young people who cannot walk independently or propel a wheelchair. This research team is assessing the feasibility and benefits of RaceRunning, which uses custom-built running bikes.

Cerebral palsy affects around **2,000** babies born in the UK each year



13-year-old Alfie has severe cerebral palsy and uses his eyes to communicate

> "We are delighted that the eyePoint scale is making an immediate and direct impact"
> Dr Michael Clarke

Steps Forward

New eye-pointing scale for severely disabled children

Communication can be incredibly difficult for some children and young people with cerebral palsy. Tested with Action funding, the eyePoint scale is now helping medical professionals and families to better understand how well children can use their eyes to engage with the world.

Severely disabled children are often unable to speak, nor can they point, reach or press buttons. But some can use their eyes to indicate what they want – a technique called eye-pointing. Knowing how well a child is able to use this skill can prove vital in providing the right kind of support.

In 2016 Action Medical Research and Great Ormond Street Hospital Children's Charity jointly funded Dr Michael Clarke and his team so they could test a new eye-pointing classification scale.

This work successfully showed that the scale was easy to use in real-life clinical settings, by different people, and gave consistent, reliable results.

The eyePoint scale is now available to download for free online, or via an app. It has been accessed by people in more than 35 countries and translated into five languages. And although it was originally developed for children with severe cerebral palsy, it has also proved helpful for doctors working with children who have other complex disorders.

Rare diseases

Fighting for children like Matilda

Matilda has Niemann-Pick disease type C, a rare and life-limiting neurodegenerative disorder. There is currently no cure and treatments are incredibly limited. Research means absolutely everything to families like Matilda's.

Matilda is a vibrant and creative little girl who loves reading and writing – and her current major passion, baking. "Obviously we're biased but she is an amazing little person, a sunshiny personality who's full of laughter," says her mum, Georgina. "But we've also discovered she is incredibly stoic and strong – which she doesn't always see in herself."

75% of rare diseases affect children

Poignantly, for a little girl so full of smiles, it was an unusual reaction to laughter that first raised concerns about Matilda. "She started falling over when she found something ridiculously hilarious," Georgina recalls. "She would literally collapse and was hurting herself as she did it – really hurting herself."

Sadly, it was the start of a journey that would lead to a life-changing diagnosis. Georgina noticed that Matilda, then aged five, was falling in a very specific way, similar to a puppet having its strings cut. "She'd lose all muscle tone and crumple straight down," she says.

Medical investigations followed and the diagnosis, when it finally came, was shattering. "My world crumpled in on itself when the doctor said I'm so sorry," says Georgina.

Niemann-Pick C is a devastating rare genetic disorder, which causes unwanted cholesterol and other fatty substances to build up inside cells, causing nerve damage. Over time, affected children develop major problems coordinating movements, such as walking, and experience a decline in their intellectual ability. Sadly, many do not survive into adulthood.

Following the shock diagnosis, the family felt overwhelmed by anger and fear. "I was just furious that this could happen to such an amazing little girl and about how cruel life is," says Georgina.

Matilda too was very angry, and incredibly afraid: "It really affected her mental health – being different, the only one in her world with NPC. One minute she was a regular kid in her class, the next she was having frequent hospital appointments," says Georgina.

Helping Matilda understand her condition and treatment has been a heartbreaking challenge for Georgina and her husband John, but they are incredibly proud of how their daughter has coped: "She has astonished us all. She is our little fighter," says Georgina.

Treatment has at times been gruelling, with Matilda initially enduring a lumbar puncture every two weeks to administer drugs as part of a clinical trial. Although her symptoms are currently relatively mild compared to others with NPC, Matilda's vision is affected and she has some memory and cognitive impairment. Her future is very uncertain.

Her parents major hope is that gene therapy will be developed in time for Matilda to benefit – something that could cure NPC. In the meantime they support any research that could help reduce symptoms or slow down the damage caused by the disease.

"We're so grateful that there is now more research and development. It's such a rare condition but there's hope that things can change," says Georgina.

> "This disease will try and take away abilities that Matilda has, so we need to fight against it. We need to try and find a treatment and a cure"
>
> Matilda's mum, Georgina

Testing a treatment for Niemann-Pick disease type C

Action is supporting important laboratory tests to find out if an existing drug, already used to treat multiple sclerosis (MS), could be repurposed as a much-needed new treatment for NPC.

Approximately 95 per cent of NPC cases are caused by faults in the gene that codes for a protein called NPC1. Without this protein, the body's cells can't work properly. US-based research has unexpectedly found that an MS drug could boost levels of the protein, suggesting it might also help patients like Matilda.

Professor Frances Platt, based at the University of Oxford, is now leading crucial tests to see whether the drug really works to improve symptoms of NPC, and also to see if combining it with another drug, the only existing approved treatment, proves even more effective.

"We hope using both drugs together could prove a powerful combination," says Professor Platt. "Particularly because they each target different biological processes involved in the disease."

Action Medical Research, the Niemann-Pick Research Foundation and Niemann-Pick UK are funding this study, together with NPSuisse and Niemann-Pick de Fuenlabrada.

Matilda's mum Georgina says: "This collaboration is really, really important. Everybody in the NPC community has the same goal – we all want a better life and a future for our children."

> "New treatments are desperately needed that can slow down or stop the progression of this disease"
>
> Professor Frances Platt

Tackling drug-resistant leukaemia

Most children with T-cell acute leukaemia can be cured – but sadly some don't respond to treatment, or the cancer returns. A new approach aims to improve survival for these patients.

Leukaemia is the most common cancer to affect children and young people, and up to one in five children who develop the disease will have a fast-developing form called T-cell acute lymphoblastic leukaemia, or T-ALL.

Around 500 children and young people are diagnosed with leukaemia in the UK each year

Intensive chemotherapy can, for most children, defeat the disease. But not for all – and in around 20 per cent the cancer eventually comes back and is then almost impossible to cure.

With Action funding, researchers at Newcastle University are investigating a new approach that combines two drugs already used to treat leukaemia. Building on earlier work, the hope is that used together they can overcome drug resistance and kill cancer cells. If successful, this would help children for whom currently no other treatments exist.

"If our results look promising, we aim to test this drug combination in an international trial, selecting the children with relapsed T-ALL who are most likely to benefit," says Dr Frederick van Delft, who is leading the research.

> "We feel we have made huge strides forward for children with juvenile Batten disease"
>
> Dr Emyr Lloyd-Evans

Corey and Izzy both have juvenile Batten disease, as does their brother Toby.

Steps Forward

Batten disease breakthrough

Action-funded researchers have made a significant discovery that could quickly open up new ways of fighting juvenile Batten disease.

Children with this rare neurodegenerative disease develop typically in their early years before experiencing increasingly distressing symptoms. These progress over time and include sight loss and epilepsy. There is currently no cure and children sadly become severely disabled before losing their lives at a young age.

The faulty gene that causes the disease was discovered 20 years ago. It gives the body's cells instructions on how to make a protein called CLN3 but, until now, nobody knew what this protein did.

With Action funding awarded in 2015, Dr Emyr Lloyd-Evans and his team at Cardiff University have now made a breakthrough discovery. They have found what CLN3 does and also identified two promising potential treatment strategies.

The team believe that certain epilepsy drugs that some affected children may already be taking could be more beneficial than anyone had realised – a finding which could rapidly help others. And they've also tested another existing drug, approved for other diseases, with promising early results. This could lead to a clinical trial in patients in the near future.

Fighting infections

Protecting babies like Ella from life-threatening lung infections

Bronchiolitis can leave babies struggling to breathe. With Action funding, researchers are using the body's natural immune defences to fight RSV, the virus that usually causes it.

When baby Ella arrived six weeks early with no complications, her parents Steph and Nick counted themselves extremely lucky. But at four weeks old she fell terribly ill with what initially seemed to be a simple cold.

Bronchiolitis is a type of chest infection which can cause congestion in the lungs. It's very common in children under one but very young babies and those who are already more vulnerable are at greatest risk.

It's often caused by the respiratory syncytial virus (RSV), a virus that in older children and adults causes mild, cold-like symptoms. But for babies it can be much more serious.

When Ella initially developed a runny nose, Steph wasn't too worried. But a week later her chest started to sound crackly. Then Ella suddenly stopped breathing. "She went grey, pale and limp – like all the life went out of her," recalls Steph. "It will haunt Nick and I forever."

Ella was rushed to hospital where doctors struggled to stabilise her – she was now fighting for her life. Needing the highest level of care, Ella was transferred to the Evelina London Children's Hospital and spent the next eight days on a ventilator.

Worldwide, more than
100,000 children under five
lose their lives to RSV each year

Led by Dr Donald Davidson, researchers at the University of Edinburgh have discovered a naturally-produced substance in the body that they think protects against RSV. But it's thought that very young babies, especially those born early, don't yet have enough of this substance, called cathelicidin, in their noses.

The team want to find out when babies begin to make cathelicidin, track its levels as they grow and see how it influences the risk of severe RSV infection. The hope is that if it could be boosted, it could help prevent RSV.

> "We saw so many other babies with bronchiolitis in hospital. We were very lucky"
>
> Ella's mum, Steph

Autism and ADHD

Scans to predict symptoms of autism and ADHD

Action funding is helping doctors study the role of two chemicals in the brain that could indicate which babies are at greatest risk of developing autism or ADHD.

Autism spectrum conditions and attention deficit hyperactivity disorder (AHDH) are common, and having one or both of these can make life much more difficult for children and their families. Children with autism typically have difficulties communicating with and relating to other people, while those with ADHD have short attention spans, find it hard to concentrate and are often restless and impulsive. Both conditions can seriously affect education, employment chances and quality of life.

ADHD affects up to
5 in every 100
children in the UK

It is already known that these conditions can run in families. But it's not known exactly what it is that makes a baby more vulnerable to developing them.

Dr Tom Arichi and his team at King's College London believe the answer could lie in understanding the balance between two chemical messengers in the brain – known as neurotransmitters. They are using sophisticated brain scanning techniques to safely and precisely measure levels of these two chemicals to see if this could allow earlier prediction in babies known to be at risk.

This could enable families to get support sooner and help develop new treatments.

Steps Forward

An app to help teenagers with ADHD

ADHD is the most common behavioural disorder in the UK, and young people with the condition are known to also face a significantly greater risk of developing depression.

Despite its potentially devastating effects, such as self-harm or even suicidal behaviour, depression often goes unrecognised and untreated in this group of young people. The symptoms of ADHD can also make it harder for them to reliably monitor their changing thoughts and feelings, making it difficult to engage with common therapies for depression, such as cognitive behavioural therapy (CBT).

With Action funding, a team based at King's College London's Institute of Psychiatry have worked with teenagers who have ADHD to develop and test a new mobile phone app. Called MoodmApper, this user-friendly digital diary prompts users to rate their thoughts and feelings several times a day. It is designed to be easy to use and give an accurate picture of mood and any mood swings in a real-life context.

As well as indicating signs of depression, the app makes it easier for teenagers to self-monitor and recognise how they're feeling, making treatments like CBT more likely to succeed.

Making an impact

Transforming the lives of children with severe dystonia

Action funding has helped to improve a surgical treatment that can dramatically improve quality of life for children suffering from uncontrollable muscle spasms.

Dystonia is a serious and unpredictable movement disorder in which abnormal signals from the brain trigger uncontrollable, sometimes painful, spasms. It can cause repetitive movements and parts of the body may be twisted into unusual positions. Growth and development of muscles and bones can also be affected, leading to deformities.

Children with dystonia can find all aspects of life difficult, including walking, speaking and eating. It can even become life-threatening. Some suffer ongoing spasms requiring heavy sedation and long hospital stays.

In 2012 Action awarded funding to a team led by Dr Jean-Pierre Lin, a consultant paediatric neurologist at London's Evelina Children's Hospital. He sought to improve how a surgical treatment called deep brain stimulation (DBS) was used to treat children suffering from severe dystonia. The method involves the insertion of electrical wires into specific areas of the brain, through which electrical pulses are delivered via a battery to control the spasms.

With Action funding, the team took highly specialised scans to make maps of children's brains. These new images helped to more clearly identify different areas which could be treated with DBS. They also helped doctors decide when, or not, it was a suitable treatment option, and informed neurosurgeons of the best area to insert wires.

These developments have improved the use and chances of success of DBS, and the techniques are now being used to help dramatically improve children's lives.

Children are already benefiting and for most there has been a clear and lasting improvement in their symptoms. Some have had astonishing results, such as being able to walk independently within two to four years.

Edward's story

Edward is now a happy, energetic seven-year-old. He's in mainstream school and runs around in his walking frame. "He plays football twice a week," says his dad, Martin, proudly. "And comes home with bruises where he's fallen over, chasing his friends."

But four years ago, Edward's life was very different. He had been a healthy baby but at around 16 months old things took a terrifying turn for the worse. His parents, Martin and Charlotte, noticed he was struggling to stand up, so took him to a GP.

Sadly, Edward's condition went rapidly downhill. Within a few months the toddler, who had previously started walking and saying his first words, could no longer lift up his arm to take a toy. He could no longer sit up, and then no longer eat.

He now had severe symptoms of dystonia and needed 'huge volumes' of medication to keep him comfortable. "If he wasn't medicated or asleep he was in rigid posturing, in pain and crying," recalls Martin. "To sit him up, he'd need his waist, legs, arms and head strapped into place. He was in incredible discomfort, and being fed through a tube. It was a terrible time."

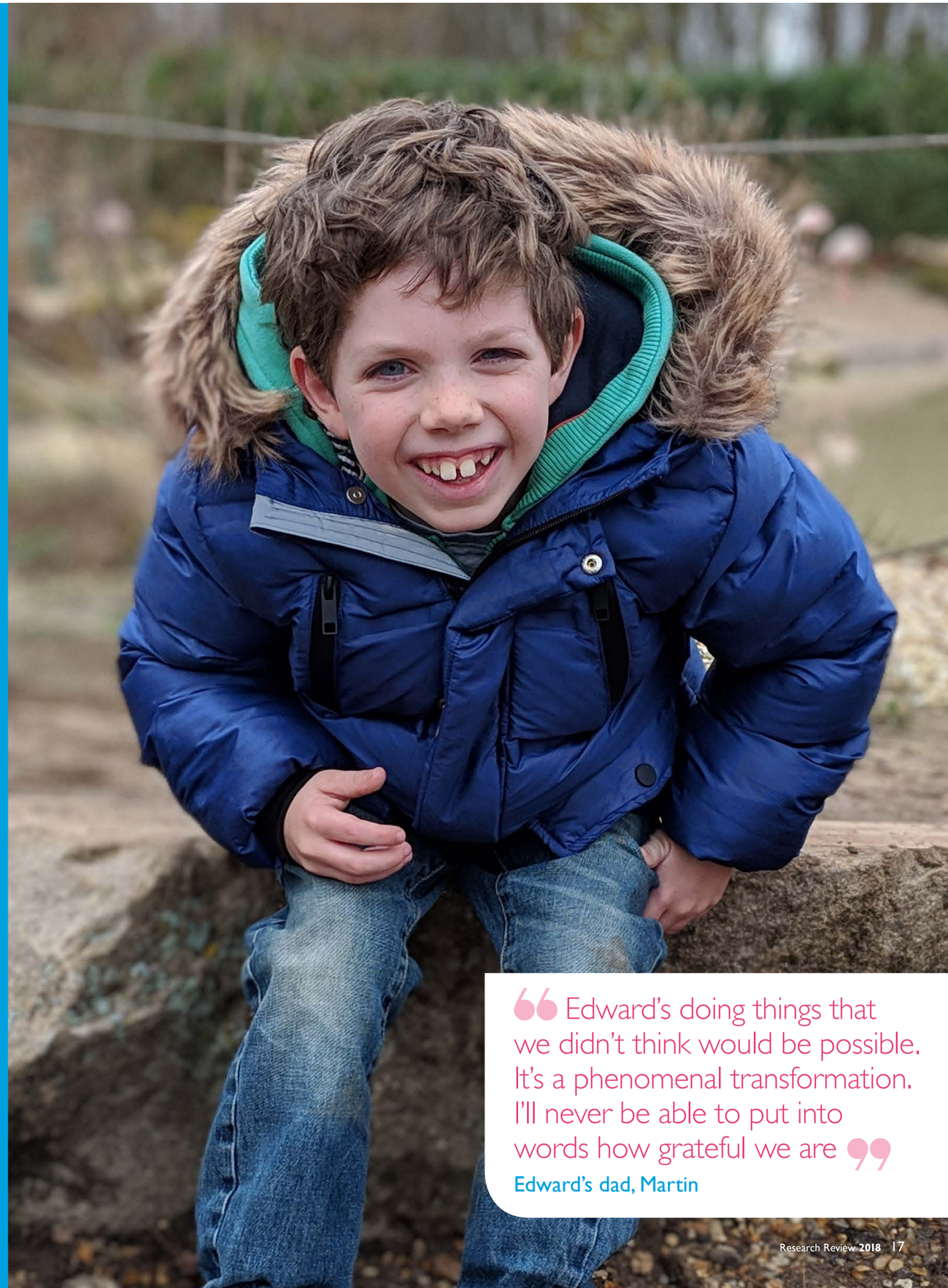
Dr Lin's team first saw Edward just after his second birthday and, after many tests, decided he was a suitable candidate for DBS surgery. This took place just after his third birthday, the earliest it could be done.

The initial measure of success set was for Edward to be able to reduce his medicine and play. Being able to sit up, eat and play with his younger brother was, says Martin, their ultimate goal.

Within a day of the operation Edward's medication was halved. "We saw immediate improvements, followed by more gradual improvements over time," explains Martin.

"The original goals were blown right out of the water. Edward has had one of the best responses to surgery, whilst also having one of the worst manifestations of the condition. It's absolutely amazing."

It is estimated that in the future
several hundred
children could benefit each year



"Edward's doing things that we didn't think would be possible. It's a phenomenal transformation. I'll never be able to put into words how grateful we are"

Edward's dad, Martin

45 years of developing leaders in children's research

2018 marked the 45th year of our Research Training Fellowship scheme, developing some of the UK's brightest doctors and scientists as future leaders in children's research. Some of today's leading lights in children's medicine were Action Research Training Fellows. And our more recent Fellows are all set to go on to equal their success.

A groundbreaking neurologist

Paediatric neurologist Professor Manju Kurian received her Research Training Fellowship in 2008, allowing her to embark on her first piece of research. She has gone on to establish her own research group at the UCL Great Ormond Street Institute of Child Health, carrying out groundbreaking work to diagnose and treat children with severe neurological disorders.

With her Action funding, Professor Kurian, then Dr Kurian, investigated the genetic basis of two devastating, life-limiting conditions affecting the brain and nervous system in very young children. This led to the discovery of one gene linked to early onset epilepsy and one that causes a Parkinson's-like movement disorder. She also developed genetic tests, meaning families could be given a faster, more accurate diagnosis and answers where previously they had none.

> "Without Action, I wouldn't be where I am today"

Professor Manju Kurian

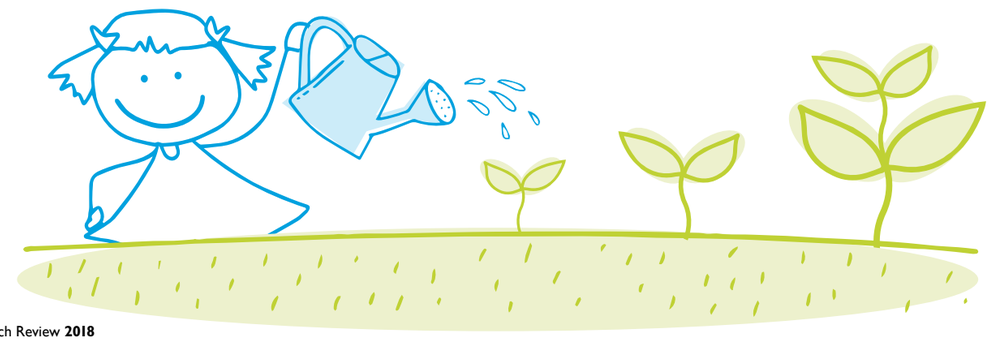
Developing MRI scanning for children, then and now

An Action Fellowship awarded back in 1987 helped Professor Margaret Hall-Craggs to develop the use of MRI scanning for children at Great Ormond Street Hospital. MRI can be used to investigate and diagnose a huge array of conditions, helping doctors to make fast and accurate treatment decisions.

Professor Hall-Craggs is now internationally recognised for her expertise – and in 2018 was awarded new Action funding to develop specialised MRI scans to help children with juvenile idiopathic arthritis. These colour-coded scans aim to make it easier for doctors to more accurately assess inflammation in the joints, and in turn make the very best treatment decisions for each affected child.

> "The Action Fellowship enabled me and another worker to start the use of MRI at Great Ormond Street Hospital"

Professor Margaret Hall-Craggs

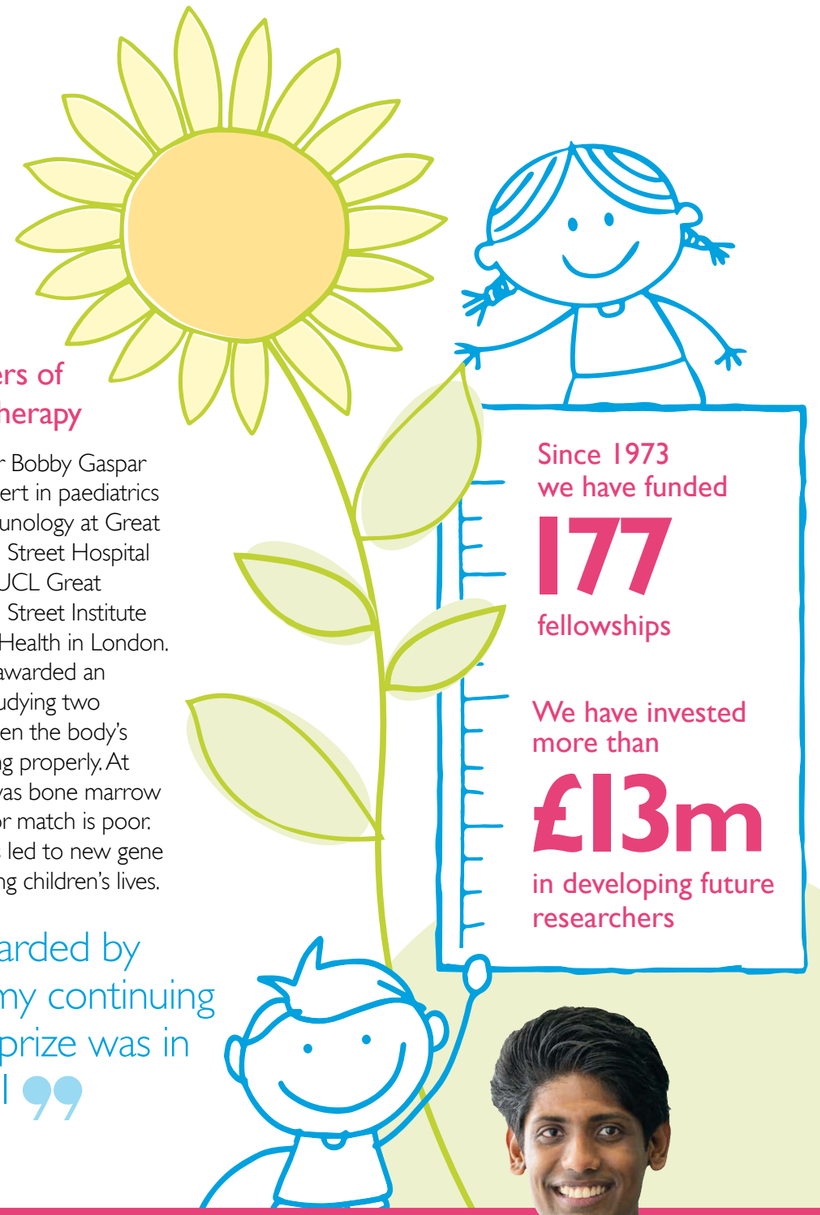


Pioneers of gene therapy

Professor Bobby Gaspar is an expert in paediatrics and immunology at Great Ormond Street Hospital and the UCL Great Ormond Street Institute of Child Health in London. He was awarded an Action Fellowship in the early 1990s, studying two life-threatening disorders that occur when the body's immune system is absent or not working properly. At that time, the conventional treatment was bone marrow transplant, which carries risk if the donor match is poor. Professor Gaspar's pioneering work has led to new gene therapy treatments which are now saving children's lives.

> "The fellowship awarded by Action was crucial to my continuing studies. Ultimately the prize was in seeing children get well"

Professor Bobby Gaspar



New in 2018

Dr Nandaki Keshavan was awarded a Fellowship in 2018. He hopes to develop the first gene therapy treatment to help children with a form of mitochondrial DNA depletion syndrome. In its most severe form, this rare condition starts in early infancy and has catastrophic effects on the liver and brain – affected children sadly often do not live to see their second birthday. Dr Keshavan hopes to be able to correct the faulty gene that causes the disease. If successful, this work would transform lives and could lead to similar treatments for children affected by other related disorders.

Photo: David Brunetti

Research grants awarded in 2018

Action Medical Research is funding more than 60 projects across the UK, 16 of them awarded in 2018. **The next medical breakthrough could be on your doorstep.**

Autism spectrum conditions and ADHD – early diagnosis in babies at risk

Lead researcher: **Dr T Arichi**
St Thomas' Hospital and King's College London

Birth asphyxia – predicting long-term effects

Lead researcher: **Dr B Vollmer**
University of Southampton, University of East London, University of Liverpool, King's College London, University Hospital Southampton

Cerebral palsy – can regular exercise improve health and mobility? *

Lead researchers:
Dr M van der Linden and Dr J Ryan
Queen Margaret University, Brunel University, University of Gloucestershire and University of Edinburgh

Cerebral palsy – hand and arm rehabilitation

Lead researcher: **Dr A Basu**
Newcastle University, Newcastle upon Tyne Hospitals NHS Foundation Trust, Evelina London Children's Hospital, Guy's and St Thomas' NHS Foundation Trust and Northumbria University

Cerebral palsy – investigating surgery to help walking

Lead researcher: **Mr T Theologis**
Oxford University Hospitals NHS Foundation Trust and University of Oxford

Cystic fibrosis – developing a new type of gene therapy **

Lead researcher: **Professor S Hart**
UCL Great Ormond Street Institute of Child Health and University College London

Infection prevention – antibiotic use and resistance in critically ill children

Lead researcher: **Dr N Pathan**
University of Cambridge, UCL Great Ormond Street Institute of Child Health, Great Ormond Street Hospital, London, St George's, University of London and Wellcome Trust Sanger Institute, Cambridge

Juvenile idiopathic arthritis – developing scans to assess joint inflammation

Lead researcher:
Professor M Hall-Craggs
University College London

Leukaemia – reversing drug-resistance if the cancer comes back

Lead researcher: **Dr F van Delft**
Newcastle University and University of Glasgow

Mitochondrial disease – investigating gene therapy

Lead researcher: **Dr N Keshavan**
UCL Great Ormond Street Institute of Child Health and Institute for Women's Health, University College London

Niemann-Pick disease type C – testing a potential new drug treatment ***

Lead researcher:
Professor F Platt
University of Oxford

Pneumococcal meningitis – improving diagnosis and management

Lead researcher: **Dr G Oligbu**
Public Health England, London, St George's, University of London and St George's Hospital, London

Preterm infants – protection against necrotising enterocolitis and serious infections

Lead researcher: **Dr D Smith**
University of Northumbria at Newcastle and Royal Victoria Infirmary, Newcastle upon Tyne Hospitals NHS Foundation Trust

Respiratory syncytial virus infection – harnessing the body's natural defences ****

Lead researcher: **Dr D Davidson**
University of Edinburgh

Spinal muscular atrophy – evaluating drug treatment

Lead researcher:
Dr M Bowerman
Keele University

XIAP deficiency – developing gene therapy

Lead researcher:
Dr C Booth
UCL Great Ormond Street Institute of Child Health, University College London

Locations of current research funded by Action

* Jointly funded with the Chartered Society of Physiotherapy Charitable Trust

** Funded together with the Cystic Fibrosis Trust

*** Action Medical Research, the Niemann-Pick Research Foundation and Niemann-Pick UK are funding this study, together with NPSuisse and Niemann-Pick de Fuenlabrada.

**** Also supported by The Chief Scientist Office (CSO) Scotland

Summarised financial statements

for year ended 31 December 2018

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 24 April 2019. The full financial statements, on which the auditor, Buzzacott LLP, gave an unqualified audit report on 3 May 2019, was submitted to the relevant statutory bodies, including the registrar of companies, on 21 May 2019.

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 2018.

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 may be obtained from Martin Richardson, Vincent House, Horsham, West Sussex RH12 2DP.

Signed on behalf of the trustees

Phil Hodkinson
Chair

Income and expenditure	2018	2017
	£000s	£000s
Net incoming resources		
Donations and legacies	2,686	2,666
Investments	44	95
Total net incoming resources	2,730	2,761
Outgoing resources		
Medical research projects	2,642	3,013
Medical dissemination	798	758
Net outgoing resources	(710)	(1,010)
Net (losses) gain on investments	(785)	321
Net movement in funds	**(1,495)**	**(689)**

Balance sheet	31 Dec 18	31 Dec 17
	£000s	£000s
Fixed assets		
Tangible	333	351
Investments	11,609	13,549
Total fixed assets	11,942	13,900
Current assets	1,328	1,575
Current liabilities within one year	(5,309)	(5,474)
Liabilities falling due after one year	(2,195)	(2,740)
Total net assets	5,766	7,261
Representing:		
Unrestricted funds	5,766	7,248
Restricted funds	-	13
Total funds	**5,766**	**7,261**

Who's who 2018

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

President
Field Marshal The Lord Guthrie GCB LVO OBE DL

Vice Presidents
Stephen May
Richard Price
The Duchess of Northumberland

Trustees
Phil Hodkinson, Chair
Luke Bordewich, Honorary Treasurer
Esther Alderson
Professor Sarah Bray BA MPhil PhD FMedSci
Professor David Edwards MA MBBS DSc MRCR FRCP FRCPCH FMedSci
Kathy Harvey – appointed April 2018
Professor Nigel Klein BSc MBBS PhD MRCP FRCPCH – retired October 2018
Nick Peters – retired November 2018
Val Remington-Hobbs
Professor David Rowitch MD PhD ScD – appointed October 2018
Richard Wild

Scientific Advisory Panel
Professor Nigel Klein BSc MBBS PhD MRCP FRCPCH, Chair – retired October 2018
Professor David Rowitch MD PhD ScD, Chair – appointed October 2018
Professor Graeme Black OBE DPhil, FRCOphth
Dr Claire Booth MBBS MSc PhD MRCPCH – appointed November 2018
Professor Clare Bryant BSc PhD BVetMed
Professor Inderjeet Dokal MBChB, MD, FRCP, FRCPCH, FRCPath, FMedSci
Professor Alicia El Haj FREng FRSB FEAMBES – appointed November 2018
Professor Jonathan Grigg BSc MBBS MD FRCPCH – appointed November 2018
Professor Catherine Hawrylowicz PhD
Professor Mark Johnson PhD MRCP MRCOG
Professor Samantha Johnson PhD CPsychol AFBPsS
Professor Fenella Kirkham MB BChir MD FRCPCH
Professor Scott Nelson PhD MRCOG
Dr Adam Shortland BSc PhD MIPEM CSci
Professor Shiranee Sriskandan FRCP PhD – appointed October 2018
Professor Michael Taggart BSc PhD

Thank you

We are hugely grateful to the many individuals, companies, trusts and foundations who have so generously donated 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.

Thank you too to those who have made provision for Action in their will and to those, named here, who left us a legacy gift in 2018.

Supporters

Airport Parking & Hotels
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Dangoor Education
Garmin
Glider Technology
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Grafton Group
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Liberty Specialty Markets
Mace Foundation
Milton Damerel Trust
Next Retail
One Stop
Oso Foundation
Paddington at Paddington Station
Price Bailey
Quai Administration Services
Rouleur
Rusk & Rusk
Sir William Coxen Trust Fund
Skerritts Consultants
Willmott Dixon

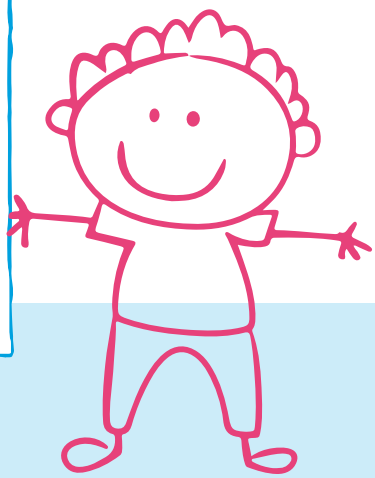
Gifts in wills

Betty Clark
Christine Cook
Yvonne Cowell
Alexandra Edgcomb
Sylvia Graucob
Geoffrey Feldman
Anthea Gardler
William Gillespie
Ronald Greenwood
Sheila Hemphill
Betty Keene
Lucy Lawrence
Margaret Lindner
Edith Lundy
Mary McClelland
John Mitchell
J W Prescot
Mildred Price
Percy Richardson
Beryl Russell
Joyce Shankie
Elisabeth Spaven
Dorothy Stephenson
Frederick Tuckman
Mr and Mrs H Vernon
Katie Wildridge

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

Thank you

With your support we fight for medical breakthroughs for some of the toughest fights our children face



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