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Thank you to everyone who was interviewed for, or gave permission for their picture to be used in this review, as well as the many members of the UCL Institute of Child Health and Great Ormond Street Hospital staff who helped during its production.

Please visit [www.ucl.ac.uk/ich/research-ich](http://www.ucl.ac.uk/ich/research-ich) or [www.gosh.nhs.uk/research-and-innovation](http://www.gosh.nhs.uk/research-and-innovation) for an online version of this review.

UCL Institute of Child Health and Great Ormond Street Hospital for Children NHS Foundation Trust

Research Review 2014/15

Research Review  
2014/15  
The child first and always







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Cover: Two-year-old Benjamin  
on Caterpillar Ward.

Left: Four-year-old Hassan  
on Lion Ward.



## Director's report

The UCL Institute of Child Health (ICH) made an important contribution to University College London's (UCL) outstanding performance in the Research Excellence Framework (REF) 2014, a nationwide assessment of the quality and standing of research within UK universities.

It is hard to overestimate the importance to universities of the REF, which is conducted every six years. The results of the most recent REF were announced in December 2014, and in Clinical Medicine (where ICH scientists represented about a quarter of all UCL's assessed staff) 80 per cent of UCL's research was assessed as either world-leading (43 per cent) or internationally excellent (37 per cent). Staff from our Population, Policy and Practice Panel Programme were evaluated by the Public Health, Health Services and Primary Care Unit of Assessment. Here, 81 per cent of UCL's research was judged as either world-leading (46 per cent) or internationally excellent (35 per cent). In addition to the feedback for Clinical Medicine, we were specifically commended for our outputs and the impact of our research. This assessment is a critically important external endorsement of the outstanding quality of research at the ICH in collaboration with its clinical partner Great Ormond Street Hospital (GOSH).

Research needs to be funded and our academic and research staff work hard to prepare excellent funding applications for national and international funding bodies. I am pleased to report that over a two year period our successful research applications more than doubled from £22 million (applications submitted in 2011/12) to £50 million (2013/14). Of particular note was our success with government bodies (principally NIHR) where funding awarded increased from £2 million to £13 million.

In June 2015, we received a very valuable visit from an international External Advisory Board (EAB), who provided an external review of ICH's academic strategy. Broadly speaking, the EAB endorsed the new academic strategy, which they felt was already showing important benefits. They had some suggestions for further improvements and they were very excited by the potential for the cross-cutting theme addressing rare diseases. We will be working hard in the coming years to consider and implement the recommendations of this EAB.

One of the key drivers over the past year has been to enhance our collaboration with the pharmaceutical industry. It is very good news for children with rare diseases that many small and large companies are now committed to developing products that can provide novel and exciting therapies. We now have an active engagement with a number of companies with the overall mission of improving treatments for children.

There have been a great number of achievements by staff over the course of the past year. In 2013/14, I reported that nine of our staff had achieved promotions to full Professor, which I then felt was exceptional. Unbelievably, this year we have had another nine individuals who have achieved this academic distinction: Paul Brogan, Mario Cortina-Borja, Patrizia Ferretti,

Two-year-old Aimi on Eagle Ward.



Director’s report

Continued

Andreas Roposch, Lesley Rees, Juan-Pedro Martinez-Barbera, Waseem Qasim, Eleanor Main and Paul Winyard.

In addition, we have seven staff who have been promoted to Reader and one member of staff to Principal Research Associate. It is hard to single out any of these excellent individuals, but I would like to mention that it is great to see full-time NHS Consultant Lesley Rees being promoted to full UCL Professor. This shows that it is possible for people in demanding clinical jobs to achieve this very high position. Furthermore, it is a great pleasure to see that two of our new professors, Paul Winyard and Eleanor Main, have achieved this through their major contribution to teaching.

Our senior staff have also been honoured in a number of other ways. Professor Adrian Thrasher was awarded a Principal Research Fellowship by the Wellcome Trust, the most senior fellowship that the Wellcome Trust awards. In last year’s report, I talked about how unusual it is that we have been awarded a National Institute for Health Research (NIHR) Professorship in two successive years, to Professor Persis Amrolia and Professor Paolo De Coppi respectively. Quite exceptionally, we’ve repeated the feat again this year – one of the four awards presented nationally to NIHR Research Professors was given to Professor Waseem Qasim at the ICH.

Additionally, Professor Terence Stephenson became the Chairman of the General Medical Council, Professor Tim Cole was awarded the Bradford Hill Medal by the Royal Statistical Society, and Professor

Catherine Law was renewed as an NIHR Senior Investigator and achieved Fellowship of the Academy of Medical Sciences. Our staff were also recognised in the Queen’s Honours List with awards of CBE to Catherine Law and Rosalind Smyth, and OBE to Helen Cross.

It has been a superb year for the ICH, but we recognise that these achievements require considerable commitment and talent from our very dedicated academic staff. We have an outstanding group of individuals conducting research at the ICH in collaboration with GOSH and this report is a testimony to their huge contribution to improving the health of children.



**Professor Rosalind Smyth**  
Director  
UCL Institute of Child Health







## Chief Executive's report

Research is integral to Great Ormond Street hospital (GOSH). By improving diagnoses and treatments and finding cures for some of the most complex and rare conditions, our work can benefit children in the UK and worldwide.

GOSH has a world-class reputation for clinical excellence underpinned by world-leading biomedical paediatric research. Since being appointed Chief Executive in January 2015, I have found this reputation to be hard-earned and well-deserved, and I continue to be impressed by the quality of the basic and clinical research conducted by our committed and talented teams.

In this year's *Research Review*, you will read just a few examples of the groundbreaking research conducted at GOSH throughout the past year. Dr Stephen Marks, Consultant Nephrologist, gives us an insight into his work on improving treatments for children with end-stage renal failure. Together with his team, he has introduced a radical new approach to kidney transplantation and is building on that work by trying to understand how to reduce the number of organs that are rejected. You will also be able to read about Dr Kate Brown's excellent research into long-term outcomes of children after heart transplants. Her focus is now turning towards improving the way these children are looked after in the community to make sure that the best outcomes are achieved for all.

The research highlighted in these pages covers a huge range of specialties, from rare inflammatory conditions to the most aggressive brain tumours. Although these are varied, they have a common goal: to improve diagnosis and treatment of unwell children within and beyond GOSH's walls.

The impact of our research around the world has been borne out by new data from

Thomson Reuters, who we commissioned to analyse our publication output. This showed that, for the five years up to 2014, research papers at GOSH and the UCL Institute of Child Health (ICH) have the highest citation impact of any of the top five children's hospitals in the world. This is impressive and we should strive to continue supporting such high-quality research to maintain our place among the best children's hospitals in the world.

Our strong reputation also makes us an attractive proposition for collaboration with international research partners across the globe. By working closely together, we can bring forward more research breakthroughs and deliver improved outcomes for children all over the world. As we look to the future, we will endeavour to ensure that research remains a priority. By providing the right environment and support we can build on our strong foundations and move towards a truly integrated research hospital. This will mean aligning our activity to give every patient the opportunity to become a research patient. For some of our sickest patients research is their only hope. For others who do have a number of options, evidence has shown that if they are involved in research, they have better clinical outcomes.

In addition to the fantastic minds that we have at GOSH and the ICH, we are also fortunate to have some extraordinary facilities. The close proximity of the ICH – adjacent to the hospital – helps ensure that findings in the laboratory can be translated into patient benefit as quickly as possible.

The opportunities and benefits of our joint working will be amplified through the creation of our new Zayed Centre for Research into Rare Disease in Children. The centre, which is due to open in 2018, will house state-of-the-art laboratories alongside clinical space for outpatients, and will be a very tangible expression of GOSH's commitment to improving care for children through research.

With its combination of expertise and facilities, GOSH is well placed to make the step changes that the young patients we see so desperately need. To maximise the use of these resources, it is vital that we continue to work in partnership with the ICH and with Great Ormond Street Hospital Children's Charity. We are committed to working to ensure that all of our strategies are aligned and that together we can continue to make the discoveries and changes needed in child health.



**Dr Peter Steer**  
Chief Executive  
Great Ormond Street Hospital for  
Children NHS Foundation Trust

Nine-year-old Anthony  
on Elephant Ward.





## Division of Research and Innovation report

Great Ormond Street Hospital (GOSH) and the UCL Institute of Child Health (ICH) joint Division of Research and Innovation: Research is Core to Care.

In 2014/15, we have seen a continued growth in our research activity with more than 8,000 patients taking part in research at GOSH – 3,000 of these recruited to high quality collaborative projects on the National Institute for Health Research (NIHR) Clinical Research Network Portfolio. You can read examples of the impact our research is making throughout this report.

A recent analysis of publications from GOSH and the ICH (carried out by Thomson Reuters) demonstrates the quality and impact of our research. It reinforces our position as one of the leading children's hospitals in the world in terms of our research, with the citation impact of our publications (the number of times others cite our research publications) being twice the world average.

Our commitment to supporting clinical research has been acknowledged by the NIHR with three of our investigators receiving awards from the NIHR Clinical Research Network for their contribution to clinical research – Dr Ri Liesner as a Leading Commercial Principal Investigator, Dr William van't Hoff for "delivering above and beyond", and Professor Francesco Muntoni for "consistently delivering to time and target" and "first global or European patient".

We are extremely pleased, thanks to the leadership of Professor Lyn Chitty and colleagues, to be playing a leading role in the co-ordination of a network of hospitals participating in the Genomics England 100,000 Genomes Project. GOSH is the lead organisation responsible for co-ordinating the recruitment of patients through the new network that will form the North Thames Genomic Medicine Centre and, alongside other partnering London Trusts, will recruit participants to the project.

In our report last year, we noted the establishment of an External Advisory Board (EAB) with international members who are leaders in their fields to provide oversight for the performance and strategy of our

NIHR Biomedical Research Centre (BRC). We have now had our second meeting of the BRC EAB, chaired by David Williams, Chief of Paediatrics at Boston Children's Hospital, and Leland Fikes, Professor of Pediatrics at Harvard Medical School and Chief of the Division of Hematology/Oncology and Director of Translational Research at Boston Children's Hospital.

The EAB review focused on two of our themes: 'Novel therapies for translation into childhood disease' and 'Diagnostics and imaging'. The leadership of the BRC and that of the themes was commended, as was the level of collaborative activity with UCL and external partners supported through our BRC. The unique environment that GOSH and the ICH offers for developing the next generation of translational researchers was also noted. Through our BRC we are committed to developing clinical academics and have developed a comprehensive training programme. The leadership of Dr Kate Oulton has been acknowledged by the NIHR as an exemplar for supporting a clinical academic training programme for nurses and allied health professionals.

GOSH aims to be a leading paediatric research hospital, and in the past 12 months we have developed our five-year Research Hospital Plan. Our vision is for research to be integral to everything we do and to see our research drive continual improvement in our clinical activity. We want to learn from each and every patient we see to improve treatments available. Over the next 12 months, we will be working to expand our research capacity and capability. We will continue to ensure that we have systems and processes that facilitate rather than prohibit research and we will be building on our research communications for our staff, patients and families.

Patients and families are at the centre of our plan, ensuring that they are not

only informed about the opportunities to be involved in research but that they are also active contributors to our research programmes and work with us to develop our communication and education about research. The recent Nuffield Council on Bioethics report, which looks at the ethics of involving children in research, is a key report that highlights the importance of research with children. Moreover, the report states that if we want to continue to improve our understanding of childhood disease and provide healthcare based on best possible evidence, then research should become a key part of the NHS. Our staff commitment is vital in supporting and delivering our vision – moving research into everyday clinical practice is key to making research core to care.

We look forward to reporting further exciting developments next year.

**Professor David Goldblatt**  
Director of Clinical Research and Development  
Great Ormond Street Hospital

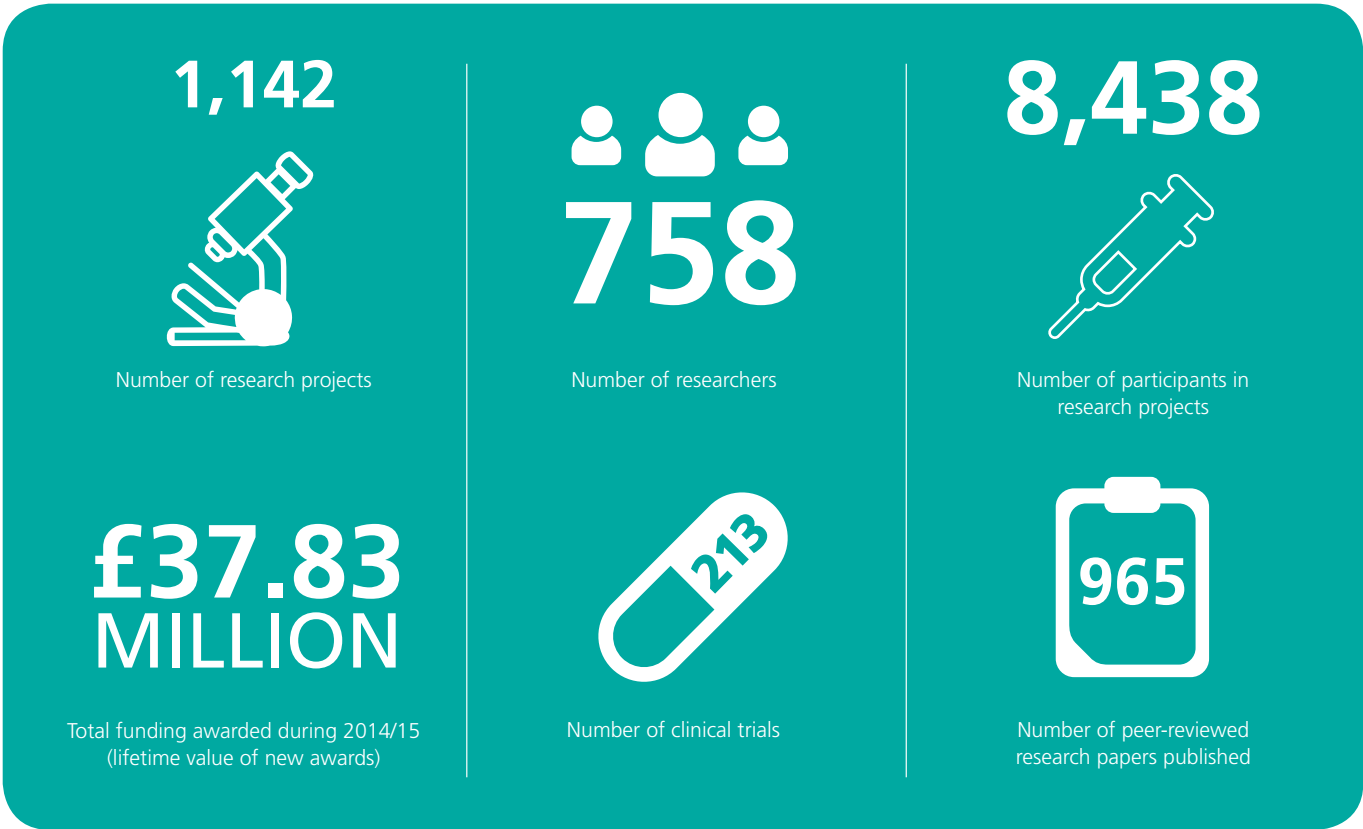
**Ms Emma Pendleton**  
Deputy Director of Research and Innovation  
Great Ormond Street Hospital





Our year in figures

During 2014/15, GOSH/ICH researchers collaborated with scientists and doctors all over the world







### Bilaal’s story

“When I was 14, my hands and feet started to ache and they went black and blue. It was really painful and my mum was worried, so we went straight to the GP, who referred me to the Royal London Hospital. When I got there, I was put on a morphine drip, but they decided that I was better off at GOSH. My dad drove me over as quickly as possible and they told me later that if I had got there much later then I could have lost a finger.

“Even when I got to GOSH, they initially found it hard to diagnose my condition. I had all sorts of different treatments, including chemotherapy, which made me really tired and gave me awful stomach cramps.

“Then the doctors took a small biopsy from my toe and found that I had a faulty gene called CERC1 that was causing my polyarteritis nodosa (a rare blood vessel disease where some arteries become swollen and damaged). I’ve got a lot better since having that diagnosis and the right treatment, and I’m really grateful to Professor Brogan for all his work.

“GOSH is so much nicer than the other hospitals I’ve been to! It doesn’t have that hospital smell and it’s nowhere near as hectic and rushed.”



### Professor Paul Brogan

“I did my pre-clinical training at the University of St Andrews before going on to do my clinical training in Manchester. One of my first medical jobs was in Hope Hospital, where I witnessed an adult patient die of a disease that we would probably now recognise as microscopic polyangiitis (a form of vasculitis). It was that encounter that first provoked my interest in vasculitis.

“When I first moved to GOSH, I was training in paediatric nephrology and I was inspired by Professor Michael Dillon and Professor Nigel Klein to do a PhD with them. After completing my PhD in 2002, I moved into rheumatology, which broadened my view and helped me develop further in the field of inflammation. Working across both GOSH and the ICH, I’ve been able to grow my research team, and to deliver in my current role as a senior clinical academic at UCL and consultant in the Rheumatology department at GOSH.”

### Professor Brogan’s research outlined in this report was supported by grants from:

- Arthritis Research UK
- Bupa Cromwell Hospital
- Butterfield Trust
- Great Ormond Street Hospital Children’s Charity
- The Lauren Currie Twilight Foundation
- National Institute for Health Research Biomedical Research Centre at Great Ormond Street Hospital for Children NHS Foundation Trust and University College London
- Novartis
- Roche Products
- Rosetrees Trust
- Vasculitis Foundation
- Vasculitis UK

### Improving diagnosis and treatment for children with rare inflammatory diseases

The immune system protects us from the bacteria and viruses that surround us. But certain diseases cause the immune system to turn on healthy tissue, causing a variety of symptoms. Professor Paul Brogan is working to improve outcomes for children with these rare inflammatory conditions.

The immune system is an extremely powerful force, able to protect children from the bacteria and viruses that are all around us. But certain autoimmune and autoinflammatory diseases cause the immune system to spin out of control and turn on healthy tissue. Children with these diseases – known as the autoimmune and autoinflammatory diseases – can endure a huge range of unpleasant symptoms – from the irritating (skin rashes), to the debilitating (blindness) to the overtly life-threatening (damage to major organs and heart attacks). Professor Paul Brogan is dedicated to improving diagnosis and treatment for children with these conditions.

“Many children who have these rare diseases will develop life-long complications or even die before they are given an accurate diagnosis,” says Professor Paul Brogan, Honorary Consultant in Rheumatology at Great Ormond Street Hospital (GOSH) and Reader at the UCL Institute of Child Health (ICH). “To make sure that children get the treatment that they need at the earliest possible stage, it’s vital that we work hard to develop better diagnostics to pinpoint the problem.

“Much of my research over the past seven or eight years has been to develop new genetic tests for the diagnosis of these diseases and to rapidly translate them into clinical practice on the NHS. When we started the fever clinic here at GOSH in 2002 there were only four genetic tests we could request for cases of suspected genetic inflammation.

“Since then, we’ve found that there are many more genetic inflammatory diseases – many that we weren’t picking up with the limited suite of tests that we had. One of our really big achievements in recent years has been the development of one test that looks for 113 different genetic diseases in one go. We’ve shown that this test can provide more rapid and accurate diagnoses.

I’d anticipate it being available for use by GOSH clinicians in the next 12 months and as a national service soon after that.

“What’s more, some of the genes that this test examines were actually recently discovered by young scientists in our research group. That just shows how discoveries in the lab can translate into patient benefit very quickly.”

Alongside his work in developing better diagnostic tests for children with rare diseases, Professor Brogan has also been pivotal in a range of clinical trials testing new treatments for these conditions. “In 2006, we set up the first clinical trials for children with a disease called ANCA vasculitis (a type of autoimmune disease),” explains Professor Brogan. “At that time, children with ANCA vasculitis were virtually always treated with cyclophosphamide, which was very effective at combatting the disease, but came at a cost. Cyclophosphamide is a strong chemotherapy drug commonly used in cancer treatment, and can have a range of long-term negative effects, including infertility and secondary cancers.

“We were able to get children involved in a trial alongside adults in collaboration with the team at Addenbrooke’s Hospital that showed we could use a gentler, less toxic drug and get the same benefits in terms of disease control.”

With encouraging results such as these, Professor Brogan has high hopes for the future. “I think the outlook for children with these diseases is very bright. We’ve got great researchers spanning basic and translational science and that really helps to drive innovation. By continuing to ensure that we encourage talent coming through and foster the cross-talk between the clinic and the lab, we’ll be able to ensure better outcomes for patients with rare inflammatory diseases.”





**Dr Kate Brown**

“I did my medical degree at Cambridge University, then I trained in paediatrics in London. From there I went on to Boston Children’s Hospital and the Children’s Hospital of Philadelphia to train in intensive care for children. While I was in the USA, I realised that public health training can help doctors to understand the wider health system, so when I came back to the UK, I took an MSc in public health, where I learned the epidemiological and statistical methods that I use in my work now.

“Shortly after, I started applying for grants and got funding from Great Ormond Street Hospital Children’s Charity to begin my work on the national audit data of outcomes for children after heart surgery.

“It’s a big challenge to keep doing research alongside my clinical work, because if you spread yourself too thinly, then everything suffers a little bit. But I think it’s important to have a foot in both camps so that I’m constantly reminded of what the important issues are for the patients.”

**Dr Brown’s research outlined in this report was supported by grants from:**

- Great Ormond Street Hospital Children’s Charity
- National Institute for Health Research

**Getting the best for children after heart surgery**

Great Ormond Street Hospital (GOSH) is the biggest heart surgery centre in the UK. Dr Kate Brown’s research investigates what happens to children in the short- and long-term after they have had major surgery for congenital heart problems.

“Recently, I was part of a team that looked at national audit data to see if survival rates from children having major heart operations had improved since the year 2000,” says Dr Kate Brown, Consultant Intensivist and Research and Outcomes Lead for the Cardiac Intensive Care Unit at GOSH. “We found that as practice has evolved and improved, the mortality rate for children within 30 days of an operation had fallen by about half.

“It’s fantastic that more children are surviving, but it also means that we have more children living with the after-effects of these very complex operations. We have a responsibility as a specialist centre to think about what happens later down the line when they go home.

“Together with the UCL Institute of Child Health, we’ve just finished another analysis of national audit data to understand what the biggest risk factors are for babies when they go home after major heart surgery. For example, we found that if an infant has a co-existing condition alongside their heart disease or has a long stay in intensive care after their operation, then those babies are at a greater risk of unexpected deterioration when they go home. They need to be watched more closely by their paediatric cardiac care network to make sure any complications are picked up and dealt with quickly.”

As part of this study, Dr Brown and her team interviewed a range of health professionals – GPs, health visitors, paediatricians, cardiologists and nurses – from every centre in the UK looking after children with heart defects to find out how these patients are treated during their care. They also spoke to a group of parents to understand how children are looked after when they leave the hospital. “Some of the parents who spoke to us had lost a baby unexpectedly

after going home from hospital,” says Dr Brown, “so they were extremely generous given what they’d been through.”

Dr Brown and her team are now analysing all their data to make recommendations for how care could be improved in the community setting. “We’re looking at whether we can improve the information that we give to families when they go home,” says Dr Brown, “so that they are better equipped to recognise and act when their baby is becoming unwell. We’re also looking at whether we can improve communications between the specialist centre and the centre in the community so that we can ensure that babies get the help they need in a timely way.”

Dr Brown’s research interests don’t begin and end with survival, however, as she goes on to explain.

“Because some of these young children have been very ill and their recovery periods tend to be very long, their overall development and quality of life may be negatively impacted. These children can go on to need extra help and support to do well at school and fulfil their potential.

“At the moment, we’re testing a screening tool called the Brief Developmental Assessment (BDA) in a large group of children who have heart disease. The BDA is designed for use in the NHS as a screening tool for nurses and doctors to pick up those children that might need extra help – whether that might be speech and language therapy, help with school work or physiotherapy – so that they can be referred for assessment. If we can work out a way to identify those young people that need extra attention as early as possible while they’re growing up, then we hope that might help more children grow up to lead fuller lives.”







**Professor Lyn Chitty**

“I was doing a PhD in the Chest Unit at King’s College Hospital when I decided I wanted to do medicine. I continued working as a half-time research fellow throughout my medicine degree. King’s was a really exciting place for obstetrics and gynaecology at the time, so I was inspired to take up this specialty.

“As part of my elective, I worked for six months in genetics with Robin Winter and came to GOSH every Thursday. During that time, Marcus Pembrey, Robin and I went to the Royal Society to hear a lecture, but it was full and so we went out to the park for tea instead! While we were sat there chatting, Robin and Marcus decided that someone needed to bridge the gap between obstetrics and genetics, and I would be the ideal person to do this.

“I then received a Medical Research Council research training fellowship and trained jointly in genetics (at GOSH) and fetal medicine (at King’s) before taking up an appointment at GOSH, the ICH and UCLH. I am now the only consultant in genetics and fetal medicine in the UK.”

**Professor Chitty’s research outlined in this report was supported by grants from:**

- International Fund Congenital Adrenal Hyperplasia
- National Institute for Health Research
- National Institute for Health Research Biomedical Research Centre at Great Ormond Street Hospital for Children NHS Foundation Trust and University College London
- Great Ormond Street Hospital Children’s Charity

**Revolutionising prenatal testing for genetic conditions**

Professor Lyn Chitty is leading a groundbreaking programme of research, which aims to make prenatal testing for genetic conditions safer for parents who are expecting a baby.

Accurately diagnosing genetic conditions in unborn children requires doctors to have a sample of the developing baby’s DNA for analysis. Until recently, the only way of getting access to the genetic material from an unborn child was to take a small sample from the amniotic fluid or from the placenta. But both procedures carry with them a very slight risk of miscarriage – between 0.5 and 1 per cent – which has been enough to dissuade some parents from taking a test. Professor Chitty’s work is helping change that, making the choice an easier one for parents.

“We now know that it’s possible to detect fragments of the baby’s DNA circulating in the mother’s blood,” explains Professor Lyn Chitty, Professor of Genetics and Fetal Medicine at the UCL Institute of Child Health (ICH) and Great Ormond Street Hospital (GOSH), and Consultant in the Fetal Medicine Unit at UCLH. “So instead of using an invasive test to see whether or not a baby has a certain condition, we can take a small sample of the mother’s blood, extract the DNA and analyse the genetic material to look for any potential problems. The work we’re doing is truly transformational – we’re making prenatal diagnosis of genetic conditions safer by reducing the need for invasive tests and that’s making it more accessible for parents.”

“The tests that we’ve been developing fall into two separate groups. The first can help diagnose children with genetic diseases caused by faults in individual genes, conditions like cystic fibrosis, Apert’s syndrome (a genetic disorder that affects the formation of the head and other parts of the body) and achondroplasia (a form of dwarfism). These tests are mainly applicable to families where there is already an affected child or where the parents know they are carriers for a particular condition. At the moment, we are the only accredited public service laboratory in the country offering non-invasive testing for these single-gene disorders, and we’re also getting referrals from across the world.

“The second group of tests help screen pregnancies for conditions like Down’s syndrome, where the baby has an additional chromosome. Our research has shown that this is a highly accurate test, but, unlike the tests we have developed for single gene disorders, we do need to confirm a positive test result by analysing cells from the amniotic fluid or placenta. However, because the new test is very accurate and safe, parents have been very keen to take it and we’ve been able to pick up more cases of Down’s syndrome before birth.

“At the moment, the non-invasive test for Down’s syndrome is only available privately, but we’ve done a big study to show that this could work in the NHS. We have presented the data to the UK National Screening Committee and they are now consulting on implementing this into the NHS.”

The benefits of detecting genetic diseases during pregnancy are clear, as Professor Chitty explains.

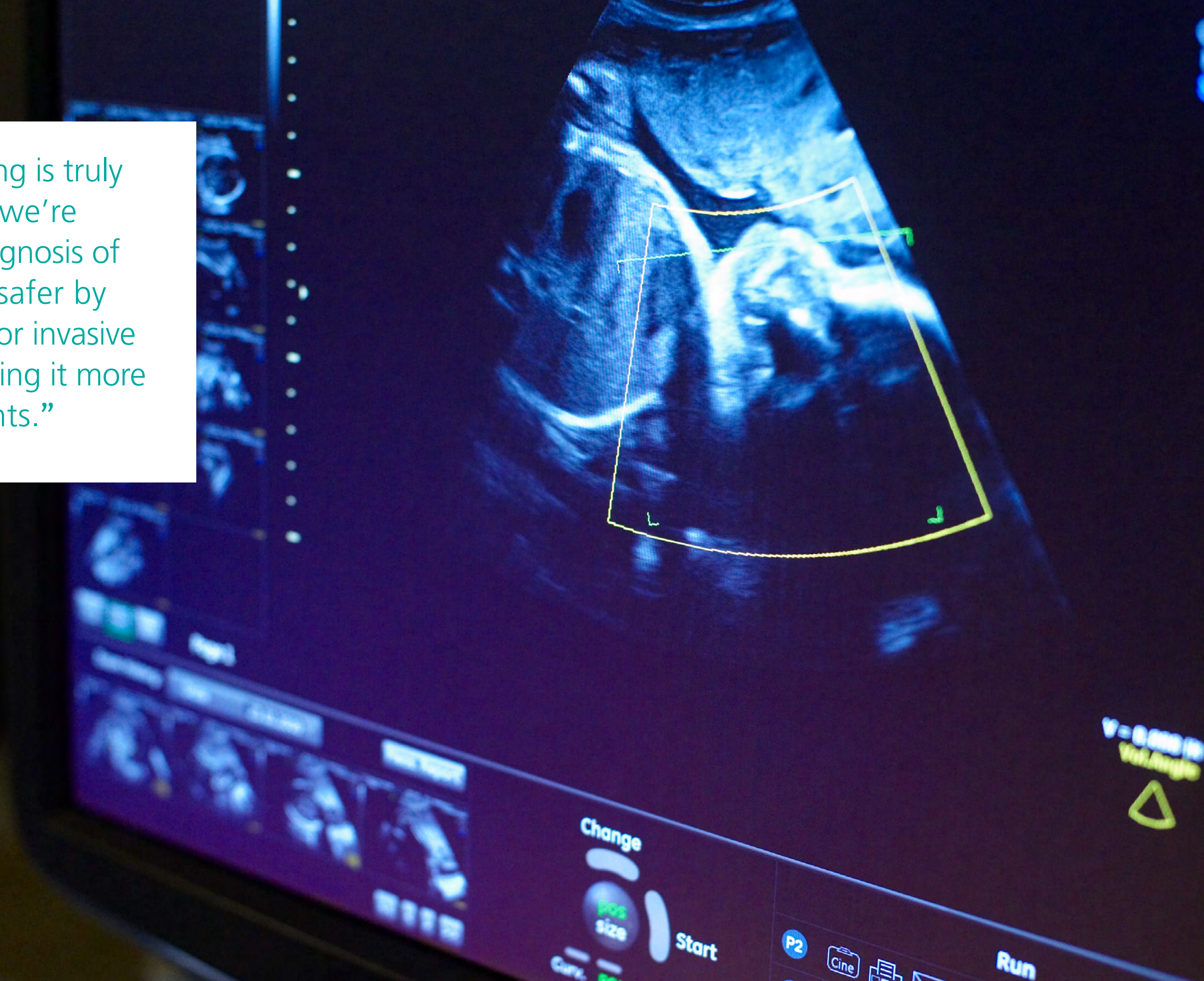
“Knowing an unborn child is affected by a certain genetic condition gives the parents plenty of forewarning to prepare them for the future and allows them to organise any early post-natal treatment that’s needed. There’s also lot of exciting work going on to develop pre-natal gene therapy for these conditions. In the future, we might be able to use gene therapy in utero to help change the course of a disease.”

Professor Chitty’s work also extends to post-natal genetics, where she is the clinical lead for North Thames for the 100,000 Genomes Project. The initiative, which began in early 2015, is gathering genetic information from thousands of people across England and linking it with their medical records to make connections between genetic patterns and certain diseases. Ultimately, this could lead to faster, more accurate diagnosis and more effective treatments for the people that need it. “The 100,000 Genomes Project stands to transform the NHS,” says Professor Chitty. “Genetics and genomic medicine is going to underpin a lot of healthcare in the future.”



“The work we’re doing is truly transformational – we’re making prenatal diagnosis of genetic conditions safer by reducing the need for invasive tests and that’s making it more accessible for parents.”

Professor Lyn Chitty







#### Professor Helen Cross

“After training in medicine in Birmingham, I specialised in paediatrics and subsequently paediatric neurology when I successfully applied for the Registrar post in paediatric neurology here at GOSH.

“I then undertook my PhD in evaluating new imaging techniques for presurgical assessment of children with epilepsy. After that, I was appointed Consultant in 1996, transferred to Senior Lecturer in 2000, promoted to Professor, and ultimately appointed to the Prince of Wales Chair of Childhood Epilepsy in 2008.

“In the meantime, I developed my national and international profile – I was on the guideline development group for the National Institute for Health and Care Excellence (NICE) guidelines for diagnosis and management of epilepsy, which were published in 2004, and clinical advisor to the revised guidelines in 2012.

“I’ve been heavily involved in the International League Against Epilepsy (ILAE) to enhance awareness of surgery for epilepsy and encourage international collaborations in research. In 2013, I was elected to the position of Secretary General of the ILAE, the first woman to be elected to the Management Committee in the organisation’s 100-year history. Earlier this year, I was recognised on the Queen’s birthday honours list, receiving an OBE in recognition of my contribution to epilepsy research and treatment.”

**Professor Cross’ research outlined in this report was supported by grants from:**

- Action Medical Research
- European Union
- NIHR
- Vitaflo

## Working towards better outcomes in epilepsy

Around one in 240 children in the UK live with epilepsy. Professor Helen Cross is dedicated to giving hope to these children when the default treatment does not work.

“At the moment, the typical treatment pathway for children with epilepsy is to try them on anti-epileptic drugs and see what happens,” says Professor Helen Cross, The Prince of Wales’ Chair of Childhood Epilepsy and Honorary Consultant in Paediatric Neurology at the UCL Institute of Child Health (ICH) and Great Ormond Street Hospital (GOSH). “Around 60–70 per cent of those children will have their seizures controlled by drugs. It’s that 30–40 per cent of children for whom the drugs don’t work that I am particularly interested in helping.”

Over the years, surgery has become an option for children with drug-resistant epilepsy. While not a surgeon herself, Professor Cross has been instrumental in developing the epilepsy surgery service at GOSH that was initiated in 1992, helping it grow to become one of Europe’s largest and best.

“My early research looked at developing various imaging techniques to assess children with drug-resistant epilepsy before surgery,” explains Professor Cross. “The idea was that if we could pinpoint the exact area in the brain where the seizures start, then surgery becomes a viable option where we ultimately remove that bit of the brain.”

Professor Cross’ research on the use of magnetic resonance imaging (MRI), and other techniques like single photon emission computed tomography (SPECT), has seen both become part of the routine clinical presurgical evaluation at GOSH.

“These days, I’m becoming more involved in the development of clinical trials,” says Professor Cross. “I think my biggest achievement in that arena is the trial that we did for the ketogenic diet.”

The ketogenic diet is a high-fat, low-carbohydrate diet that has been used to treat children with epilepsy since the 1920s. In 2008, Professor Cross and her team published results from the first large randomised controlled trial of the

diet, showing that children between the ages of two to 16 years of age who did not respond to anti-epileptics could have their seizures improved by following the diet.

“We really feel that our work put the ketogenic diet on the map,” says Professor Cross. “It’s something that’s been used for nearly 100 years, but we produced the evidence base proving that the diet definitively worked. After following up on these children, we now have more evidence to show that response to the diet is both fairly immediate and long-lasting – the positive effect is usually sustained after the children have come off the diet.

“In collaboration with Simon Heales at the ICH, we’ve been building on this work in the past year or so to look at what it is in the diet that is the ‘active ingredient’. We’ve known for a little while that the diet increases the levels of an organic molecule called decanoic acid. Now we’ve found that decanoic acid increases the activity of mitochondria, which we think might help reduce the tendency towards seizures. We’re now planning to start a clinical trial to see if giving children decanoic acid alone would be as effective for controlling seizures as the diet itself.

Professor Cross adds: “It’s a really exciting time for children with epilepsy: for so long we’ve had a blunderbuss approach – using anti-epileptics and hoping for the best. Now we’re gaining a greater understanding of the causes and that is giving us a real chance of targeted treatments, to improve the lives of children with epilepsy.”







**Dr Stephen Marks**

“I was a medical student in Scotland and did my initial paediatric training in Newcastle. After spending a year in Australia, I first came to train at GOSH almost 18 years ago with further training in paediatric nephrology in Toronto, Canada and carried out MSc and MD research at the UCL Institute of Child Health.

“I have led the kidney transplant team for over a decade now and continue to be involved in international multicentre randomised controlled immunosuppressive drug trials in paediatric renal transplant recipients. I have been the Chair of the NHS Blood and Transplant Paediatric Kidney Advisory Group since 2011.

“I lead the transition programme, and because we look after children until they are 18 years, I’ve cared for patients from when they were babies right up to when I have transferred their care to adult colleagues. I love seeing how we are able to improve the quality of life for our patients on dialysis after they receive a good functioning kidney transplant. It is an honour and privilege to look after children and their families for many years and see them throughout their childhood growing up into young adults for the future. If you came to my kidney transplant clinic, you would see a group of healthy children who can lead lives to the full.”

**Dr Marks’ research outlined in this report was supported by grants from:**

- Astellas Pharma Europe Ltd
- Kids Kidney Research
- Novartis

**Improving the lives of children with severe irreversible kidney failure**

Dr Stephen Marks is dedicated to improving outcomes for children with renal failure who require kidney transplantation.

From the moment we are born, our kidneys work tirelessly to filter the blood circulating through our bodies, removing toxic waste products and keeping us healthy. However, for children with severe irreversible kidney failure (chronic and end-stage kidney disease), this blood-cleaning system breaks down and they need kidney replacement therapy to survive.

However, some children with kidney disease can be deemed ‘untransplantable’ due to having high levels of powerful molecules in their blood, called human leukocyte antigen (HLA) antibodies, which reject new organs.

“Historically, children with high levels of HLA would not have been able to receive kidneys from living donors, but new techniques are starting to change that,” explains Dr Stephen Marks, Consultant Paediatric Nephrologist and Clinical Lead for renal transplantation at Great Ormond Street Hospital (GOSH).

Dr Marks’ multidisciplinary kidney transplant team at GOSH have collaborated with the transplant surgeons at Guy’s Hospital and King’s College London to introduce a new technique that allows previously ‘untransplantable’ children to receive a kidney transplant. The new method filters out HLA antibodies from the recipient’s blood before a transplant is carried out, markedly increasing the chances of a successful procedure.

Alongside this pioneering work, Dr Marks is also interested in understanding why kidney transplants eventually fail – whether after 20 hours or 20 years – and what can be done to make them last longer. He hopes to use this research as a platform to further improve treatment for children with kidney disease in the future.

“Transplantation is about the body understanding that the kidney is now part of you and not a foreign body,” says Dr Marks. “We use powerful drugs –

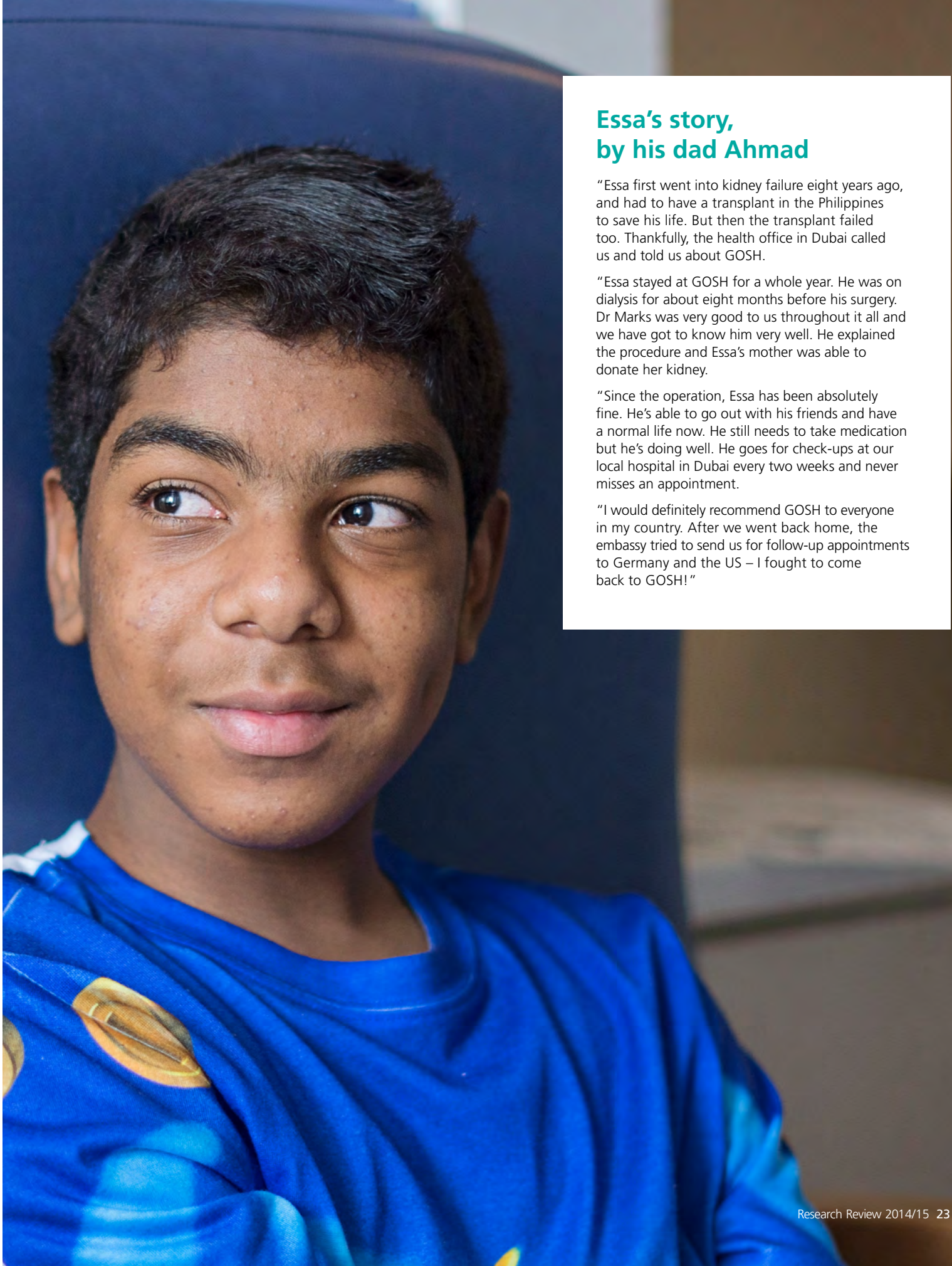
called immunosuppressive agents – to reduce acute rejection episodes where the body starts attacking the new organ. However, the most powerful of these drugs can actually damage kidneys, causing, on average, half of kidneys from living donors to fail before 20 years. But interestingly, there’s wide variation in terms of how patients react to a transplant – some children tolerate the new organ very well and actually require minimal or no immunosuppressive agents at all.

“We are now examining this immune response to try and predict what is going to happen to individual patients. If we look at a large cohort of patients, we can see that some of them develop antibodies that start attacking the new kidney, while others have few problems. We are now following these children up, looking out for biomarkers that will help us foresee a child’s immunological response.

“The future aim is to be able to predict how much immunosuppression a child will need before transplantation so we can tailor treatment to the individual, reducing the burden of immunosuppressive therapy where we can, and ultimately get the best outcomes for all.”

With such exciting research gathering pace, Dr Marks has a clear vision of the future:

“Our ultimate goal is to understand not only the genetic basis for children with kidney disease but the genetic basis for transplantation and how the children react to medication. This will require collaboration between different teams within UCL, nationally and internationally. I envisage one day we will look at factors in the child or teenager who requires a kidney transplant, match it with the immunological factors from the donor, and work out a personalised immunosuppression plan, which altogether will ensure better outcomes for these patients.”



**Essa’s story, by his dad Ahmad**

“Essa first went into kidney failure eight years ago, and had to have a transplant in the Philippines to save his life. But then the transplant failed too. Thankfully, the health office in Dubai called us and told us about GOSH.

“Essa stayed at GOSH for a whole year. He was on dialysis for about eight months before his surgery. Dr Marks was very good to us throughout it all and we have got to know him very well. He explained the procedure and Essa’s mother was able to donate her kidney.

“Since the operation, Essa has been absolutely fine. He’s able to go out with his friends and have a normal life now. He still needs to take medication but he’s doing well. He goes for check-ups at our local hospital in Dubai every two weeks and never misses an appointment.

“I would definitely recommend GOSH to everyone in my country. After we went back home, the embassy tried to send us for follow-up appointments to Germany and the US – I fought to come back to GOSH!”









### Georgina’s story, by her dad Simon

“Just after Georgina was born, the team looking after her noticed that something was wrong with her hip and immediately referred us to GOSH.

“It was a worrying time for us, particularly as we’d never heard of hip dysplasia. But we were very impressed with GOSH when we came for Georgina’s first appointment a month later – everything was explained carefully to us and everyone was very helpful. It was great that there were toys to keep Georgina’s older sister Alicia happy while we waited on the occasions she had to come with us.

“Professor Roposch explained Georgina’s condition. He said that it was a good thing that they’d found it early because it meant that surgery wouldn’t be needed. But she still needed to wear a Pavlik harness to

realign her hips, which made life at home a little more complicated, particularly changing and holding her. At first this seemed very difficult, but it was surprising how quickly we got used to it. It worked, and after a couple of check-ups, about four months later we went back to hospital. Professor Roposch said that Georgina’s hip had healed enough for the harness to come off – we were delighted.

“Georgina is now two-and-a-half years old and very active – she loves climbing all over the furniture at home and is learning to ride her scooter! We don’t know that we’re totally out of the woods yet, but it’s very comforting to know that we have Professor Roposch’s first-rate team at GOSH looking after us.”



#### Professor Andreas Roposch

“I have always wanted to go into an operative specialty and work with children. I did my MD in a paediatric orthopaedic department and it secured me an academic training post.

“After I qualified as a consultant at the University of Vienna, I went on a clinical fellowship in Toronto. Following this, I was fortunate to receive a scholarship in the Research Training Competition Programme, which was intended for clinicians wanting to undertake university-level research training. I read for a master’s in Clinical Epidemiology at the University of Toronto alongside a post-doc in Population Health Sciences under Professor James G Wright. This was a fascinating time as Toronto has one of the strongest programmes in clinical epidemiology in the world. There were so many bright people to learn from.

“My move to ICH and GOSH has put me in the best place in Europe to take forward my interest in academic surgery and patient-based research.

“It can be challenging sometimes for surgeons to keep high level research going. The clinical work can be physically exhausting at times, and to get the experience needed to take on difficult cases, one needs to do enough of them. I’m a strong believer in the surgeon-scientist model, though, and many professional bodies and learned societies are supporting a move towards more academia in surgery.”

#### Professor Roposch’s research outlined in this report was supported by grants from:

- Great Ormond Street Hospital Children’s Charity
- National Institute for Health Research
- Newlife foundation for Disabled Children
- Arthritis Research UK

### Optimising decision-making in orthopaedic surgery

Reaching a decision on the best way to care for a patient is an enormous responsibility. Professor Andreas Roposch’s work looks at how to improve decision-making across the whole care pathway – from the community to the hospital – to improve outcomes for patients.

“I’m interested in how clinicians reason, formulate judgements and make decisions,” says Professor Andreas Roposch, Consultant in Paediatric Orthopaedic Surgery at Great Ormond Street Hospital (GOSH) and Honorary Reader at the UCL Institute of Child Health (ICH). “In particular, I am interested in how we make diagnoses, and the implication this has for patients and the NHS.”

Much of Professor Roposch’s research focuses around hip dysplasia – a condition where the joint is malformed. If the condition is diagnosed early, it can be treated relatively easily in most cases, using a special harness to keep the hips aligned and stable.

“There is, however, a downside with early diagnosis,” explains Professor Roposch. “Doctors very often diagnose hip dysplasia when it isn’t actually there. We call this an overdiagnosis. It can occur for several reasons, one being a doctor’s reasoning and judgement. If a baby is given the disease label when their hips are actually normal, they will experience some form of harm or burden. Their family will have to travel to hospital, the child will have to have tests and they might be given treatments that they might not need at all. All of that is costly for families and for the NHS.

“The research I’ve been doing over the past decade is about making sure that diagnosis of hip dysplasia is more precise and evidence-based. We wanted to make sure that we can identify those children with hip dysplasia that could benefit from early treatment while also identifying those that could safely be discharged from medical care without risk to their health. Previous research in this area was confounded by variable

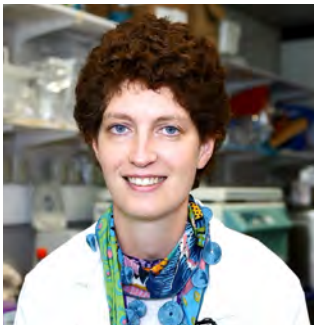
disease definitions and there was a need to return to basic principles to sort through the many criteria and definitions related to hip dysplasia. We looked at 34 countries and found a wide variation in how expert surgeons diagnose dysplasia.

“By working closely with 300 clinical experts, we have now created a diagnostic index, which enables clinicians to diagnose hip dysplasia more consistently. We now hope to use this index to tackle the problem of overdiagnosis in primary care. We are currently preparing a clinical trial of GP practices in England, which will investigate whether using our diagnostic tool is cost-effective and leads to better care for infants.”

Alongside this work, Professor Roposch is conducting genetic research to look for markers that could help further refine diagnosis for children with hip dysplasia. “We see quite a lot of variability in children with hip dysplasia in that some respond well to treatment and others don’t – hip dysplasia doesn’t seem to be a single disease. Using genetic techniques, we want to identify the different types of disease, so that we can pinpoint more accurately which patients are most at risk and which are unlikely to respond well to treatments.”

The work of Professor Roposch and his team is vital to creating more effective ways to define groups of patients. He adds: “Sometimes, it is important to step back and consider what we call a disease. Labelling a clinical phenomenon as a disease always leads to some clinical response. This response may cause burden to patients and the NHS or society. We need to make sure we are doing what is best for the patients and their family while taking into account the costs and societal consequences as well.”





**Dr Karin Straathof**

“My interest in immunotherapy goes way back to the very beginning of my research career. In medical school I did an intercalated BSc biomedical science degree and my main project was immunotherapy. I was involved in identifying which bit of HPV (human papilloma virus) was recognised by T-cells, which played a small part in what was eventually used to design the vaccine that has been implemented to protect against cervical carcinoma. That was a real demonstration of how immunotherapy can be used to prevent and treat cancer.

“I was given a prize for that project and I decided to go to Baylor College of Medicine, which is linked to Texas Children’s Hospital. That was where I started working on immunotherapy before coming to GOSH to join an excellent team of researchers to continue this work. What inspires me to continue doing the research is, without a doubt, the patients.”

- Dr Straathof’s research outlined in this report was supported by grants from:**
- Great Ormond Street Hospital Children’s Charity
  - National Institute for Health Research Biomedical Research Centre at Great Ormond Street Hospital for Children NHS Foundation Trust and University College London
  - Wellcome Trust
  - Cancer Research UK

## Harnessing the immune system to fight childhood cancers

Dr Karin Straathof is investigating how to teach a child’s immune system to recognise and destroy neuroblastoma cells, a type of cancer that is very difficult to treat.

Our immune system is an extremely sophisticated defence network, armed and ready to fight off the viruses and bacteria that we encounter every day to keep us healthy. In recent years, scientists have begun to recognise that the extraordinary power of the immune system can also be turned on cancer to attack and destroy tumours. This approach is exciting cancer scientists across the world and could soon be used to help children with neuroblastoma, as Dr Karin Straathof, National Institute for Health Research Academic Lecturer in Paediatric Oncology at Great Ormond Street Hospital (GOSH), explains.

“This type of therapy is called immunotherapy, and is starting to work really well for ‘liquid cancers’ such as leukaemia, where children and adults whose cancer was not responding to chemotherapy have been cured. What we’re trying to do is make the same type of treatment work for solid tumours, such as neuroblastoma.”

Neuroblastoma is a common childhood cancer, and is currently very difficult to treat, at least for a significant number of patients. The tumour usually develops as a lump in the tummy, but then spreads to other parts of the body. Unfortunately, nearly half of the patients with neuroblastoma have an aggressive form of this disease.

“At the moment, the best treatment available for these children is a combination of chemotherapy, surgery and radiotherapy,” explains Dr Straathof. “This very intensive treatment often has nasty side effects. You have the well-known immediate side effects, such as hair loss, nausea, tummy pain and diarrhoea, but these treatments can also have delayed effects: chemo- and radiotherapy can damage the heart and the kidneys and can cause hearing loss and infertility.”

Immunotherapy would reduce those side effects, as it is better targeted towards the tumour making the treatment less toxic. “Our immunotherapy technique involves taking special immune cells from the patient – called T-cells – and modifying them using gene therapy so that these cells can recognise and destroy neuroblastoma cells, while leaving healthy cells unharmed,” says Dr Straathof. “These modified T-cells are then tested in the lab before they are given back to the patient.

“The important thing here is that the treatment is not a drug, nor an antibody. It’s a living cell, which means it will divide and multiply and ultimately amplify the tumour-killing response. The hope is that these immune cells are able to destroy all tumour cells as well as stopping the cancer from returning.”

Neuroblastoma may be particularly amenable to treatment by immunotherapy as the tumour is coated with a molecule called GD2, which makes it easier for the body to distinguish it from the normal tissue. To exploit this, Dr Straathof and her team have been developing and optimising the immunotherapy technique for years to discover the best way of engineering immune cells to recognise GD2-coated neuroblastoma cells.

“Excitingly, we are now ready to start a clinical trial where we use this type of immunotherapy later this year,” says Dr Straathof. “My overriding ambition is to improve the outcome for childhood cancers, starting with neuroblastoma, but then also applying this strategy to other tumours. I want to increase the number of children with a cure, as well as reduce the number of side effects along the way, so that survivors can lead normal lives and achieve just like their peers.”







**Professor Faith Gibson**

“In essence, what we’re trying to do in ORCHID is to undertake research that improves patient and family experience while they are in hospital,” says Professor Faith Gibson, Clinical Professor of Children’s and Young People’s Cancer Care at GOSH and London South Bank University.

“If we want to be the best hospital in the world, then we need to have a broader understanding of child health. Clearly, research that improves diagnosis and treatment and increases survival is important, but that has to be complemented by research aimed at improving patient experience.

“The kinds of questions we’re interested in are: how can we make sure that patients’ time in the hospital is as good an experience as it can be? How do we know that the care we are providing is responsive, effective, and appropriate? How can we better prepare children and young people for life after hospital?

“Other areas include helping patients explain their condition to their friends at school, finding out what it’s like living with a chronic illness or disability, and what interventions we can introduce to help support families.”

“In the past, we used to try and glean this kind of information from parents or other carers. But if we want to try and deliver true patient and more specifically person-centred care at GOSH, we need to gather the perspectives from those receiving our care. And that’s what ORCHID is really about.”

**The Centre for Outcomes and Experience Research in Children’s Health, Illness and Disability**

Professor Faith Gibson leads a team of researchers dedicated to improving children’s experience of Great Ormond Street Hospital (GOSH) – the Centre for Outcomes and Experience Research in Children’s Health, Illness and Disability (ORCHID).

ORCHID’s core team is made up of Professor Gibson, who leads the group, Dr Kate Oulton, Dr Debbie Sell and Dr Jo Wray. The researchers come from a range of different professional backgrounds, which is a huge strength for the group. Professor Gibson trained as a children’s cancer nurse, and Dr Oulton as a children’s nurse with a specific interest in children with complex needs. Dr Sell is a Speech and Language Therapist and Dr Wray is a Psychologist.

“We’re very varied,” says Professor Gibson. “But what we have in common is a shared focus on trying to understand and improve the patient and family experience for everyone coming through the doors of the hospital. We work across the multi-professional teams and collaborate closely with those working in the Somers Clinical Research Facility and other clinical units, as well as consultant nurses working in the Trust.”

In the past year, ORCHID have achieved a huge amount. The group have completed a number of studies, including a small project exploring children’s understanding of being involved in a research study. They have also worked with GO Create! – the hospital’s arts programme – and Medical Illustration to develop an app called BloodQuest, which can be used as a distraction and preparation tool for children having their blood taken.

In the future, Professor Gibson would like to see research woven into the fabric of the hospital so that children can look forward to better care. Professor Gibson adds: “We have supported a number of nurses and Allied Health Professionals (AHPs) to secure

National Institute for Health Research (NIHR) funding to complete their master’s degrees in Clinical Research,” says Professor Gibson. “With funding from the NIHR Biomedical Research Centre, we have also offered 13 internships to nurses, AHPs and clinical scientists across the Trust and ICH to support them and help them apply for doctoral and post-doctoral NIHR fellowships. To complement this activity we held a study day, planned an academic weekend and have established an academic inquiring minds group. All of this goes some way to positively influencing the Trust culture and perceptions of research.”

“We recently led on a Trust-wide staff survey exploring the barriers and facilitators to GOSH becoming a true research hospital. We’d like to get to a stage where research is embedded into everything that we do as nurses and AHPs. That doesn’t mean that everyone has to have a PhD. It means that everyone understands that research can make things better. It means putting the patient at the centre of everything we do.”

Nine-year-old Daisy on Elephant Ward.



Grants and donations

Great Ormond Street Hospital (GOSH) and the UCL Institute of Child Health (ICH) continue to receive grants and donations towards research from the following individuals and organisations.

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University of Tartu

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Vasculitis Foundation  
Vasculitis UK  
The 3VB Charitable Trust  
Vitaflo Ltd  
The Waterloo Foundation  
Wellbeing of Women  
WellChild  
Wellcome Trust  
The Welton Foundation  
The Wenner-Gren Foundation  
World Health Organization  
Worldwide Cancer Research

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# UCL Institute of Child Health’s academic programmes

## Developmental biology and cancer

**Head of Programme**  
Professor Andrew Copp

**Deputy Head**  
Professor John Anderson

The aim of the developmental biology and cancer programme is to study the key processes necessary for normal development and tissue homeostasis that are altered in childhood diseases.

**Cancer** – to understand the molecular basis of childhood cancers and to develop more effective therapies for children with cancer, by combining basic research in cell and molecular biology with translational cancer research and clinical trials.

**Developmental biology of birth defects** – to use techniques of genetics and developmental biology to improve our understanding of, and develop novel treatments and preventive strategies for, clinically important birth defects.

**Stem cells and regenerative medicine** – to promote and expand translational stem cell research and harness the great potential of regenerative medicine for childhood disease.

## Developmental neuroscience

**Head of Programme**  
Professor Francesco Muntoni

**Deputy Head**  
Professor Helen Cross

The focus of the developmental neuroscience programme is to minimise the impact of disorders affecting the developing central and peripheral nervous system.

**Anatomical, genetic and functional aspects of neurodevelopment** – to understand normal and pathological neurodevelopment using innovative technologies.

**Improving neurodevelopmental outcomes** – to promote early diagnosis and improve the treatment of children with developmental or acquired disorders of the brain through robust clinical trials.

**Shape national guidelines and policy** – to contribute to the development of national guidelines and policies, and set national/international standards of clinical care for children with neurodevelopmental disorders and diseases affecting the function of the nervous system.

## Genetics and genomic medicine

**Head of Programme**  
Professor Phil Beales

**Deputy Head**  
Professor Maria Bitner-Glindzicz

The aim of the genetics and genomic medicine programme is to use genetics, imaging and biological indicators to understand predisposition to disease and what constitutes health during childhood and throughout the life course.

**Gene and protein function** – to develop tools, skills and resources to investigate gene function and to inform development of new therapies using state-of-the-art technologies.

**Translational ‘omics’** – to create, integrate and maintain data and informatics platforms to support genomic, proteomic and other ‘-omic’ research and its healthcare applications.

**Personalised medicine** – to ensure discoveries of disease mechanisms and patients’ genomes are used to best effect to improve patients’ lives. This will include better diagnostics, identification of biomarkers and targeting of therapies.

## Infection, immunity, inflammation and physiological medicine

**Head of Programme**  
Professor Adrian Thrasher

**Deputy Head**  
Professor Lucy Wedderburn

The aim of the infection, immunity and physiological medicine programme is to deliver world-class interdisciplinary research for children with infectious, immunological and inflammatory disease, children with life-threatening respiratory disease, children in pain and critically ill children on intensive care.

**Molecular basis of immunological and inflammatory disease** – to apply high-throughput genetics to understand the molecular basis of these immunological and inflammatory diseases.

**Basic immunological mechanisms** – to investigate immunological mechanisms and function in model systems and in children.

**Pathogen action** – to understand molecular mechanisms of pathogen action through study of microbial genetics and host response to challenge.

**Disease prevention** – to develop effective methods to enhance disease diagnosis in early childhood and use effective vaccination strategies.

**Development of a world-leading translational research programme** – to undertake clinical trials in children with immunological and inflammatory diseases using novel therapies.

**Respiratory, critical care and pain** – to improve the understanding of early lung disease and the pathophysiology of children with life-threatening respiratory disease, critically ill children on intensive care and children in pain, and to develop novel therapeutic and management strategies to improve their outcome.

## Population, policy and practice

**Head of Programme**  
Professor Catherine Law  
(until Oct 2015)

Professor Helen Roberts  
(interim Head between Oct 2015 – Jan 2016)

Professor Monica Lakhanpaul  
(Head from Jan 2016)

**Deputy Head**  
Professor Russell Viner

The aim of the population, policy and practice programme is to conduct research that promotes the health of children and the adults they will become.

**Research across the life course** – identifying risk factors for disease, testing hypotheses on causal mechanisms and informing public health policy.

**Burden of disease** – to investigate common, chronic and complex conditions of children and young people, using population-based and clinical approaches.

**Promote children and young people’s health** – to define and measure what matters for children, young people and their families and use this information to promote children and young people’s health.

**Children in their contexts** – to carry out research on health services, the environment and other systems and contexts to prevent, diagnose and treat disease and to promote health and wellbeing.

## Cross-cutting theme for rare diseases

**Theme Lead**  
Professor Bobby Gaspar

The cross-cutting theme for rare diseases will harness the excellence in these fields within the programmes above and seek to:

**Improve the lives of children** with rare diseases through basic and translational research.

**Create greater visibility and emphasis on rare diseases** by enhancing research activity, facilitating joint working between the ICH and GOSH and engaging with external partners.

**Offer tangible benefits for children with rare diseases** either at GOSH or nationally and internationally using the latest technologies.



# Administration

## Great Ormond Street Hospital Board of Directors

Chairman of the Trust Board and Members' Council

### Baroness Tessa Blackstone

Non-Executive Directors

Yvonne Brown  
David Lomas  
Mary MacLeod  
Akhter Mateen (from April 2015)  
John Ripley (until March 2015)  
Professor Rosalind Smyth  
Charles Tilley

Trust Board Executive Directors

### Chief Executive

Dr Peter Steer (from January 2015)  
Julian Nettel (until December 2014)

### Chief Finance Officer

Claire Newton

### Chief Nurse

Juliette Greenwood (from May 2015)  
Elizabeth Morgan (until March 2015)

### Medical Director

Dr Vinod Dawakar (from July 2015)  
Dr Catherine Cale and Professor Martin Elliott (Co-medical Directors until June 2015)

### Director of Human Resources and Organisational Development

Ali Mohammed

### Chief Operating Officer

Rachel Williams (Dena Marshall interim maternity leave cover from May 2015)

## UCL Institute of Child Health Leadership

Senior Management Team

### Director

Professor Rosalind Smyth

### Deputy Director (Education)

Professor Christine Kinnon

### Deputy Director (NHS engagement)

Professor David Goldblatt

### Deputy Director

(Strategy and Partnerships)

Professor Catherine Law

### Institute Manager

Wendy Knowles

Academic programme management

### Head of Developmental Biology and Cancer Programme

Professor Andrew Copp

### Head of Developmental Neurosciences Programme

Professor Francesco Muntoni

### Head of Infection, Immunity, Inflammation and Physiological Medicine Programme

Professor Adrian Thrasher

### Head of Population, Policy and Practice Programme

Professor Catherine Law

### Head of Genetics and Genomic Medicine Programme

Professor Philip Beales

### Theme Lead for Rare Disease

Professor Bobby Gaspar

## Trustees for Great Ormond Street Hospital Children's Charity

### Chair

Alan Hodson  
(Chair until 31 March 2015)  
John Connolly  
(Chair from 1 April 2015)

### Trustees

Gabrielle Abbott (until 31 March 2015)  
Susan Burns (until 30 September 2015)  
John Connolly  
Dr Diana Dunstan  
Hugo Llewelyn (until 30 September 2015)  
Christopher Spratling (until 26 June 2015)

### New Trustees

David Claydon (from 1 April 2015)  
Margaret Ewing (from 1 April 2015)  
Kaela Fenn-Smith (from 1 April 2015)  
Frances Murphy (from 1 April 2015)  
Steven Sharp (from 1 November 2015)  
Mark Sartori (from 11 November 2015)

### Associate Trustees

Simon Brewer  
Ruary Neill  
Simon Stormer  
Humphrey van der Klugt  
Michael Weston (until 14 October 2014)  
Dr Edward Wozniak

## Trustees for The Child Health Research Charitable Incorporated Organisation

### Chair

Mr Andrew Fane

### Trustees

Mr Tim Clarke  
Professor Andrew Copp  
Professor David Goldblatt  
Professor Graham Hart  
Ms Sara Payne  
Mr Andrew Ross  
Professor Rosalind Smyth  
Dr Peter Steer

Right: Five-year-old Shiloh on Badger Ward.