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Ayesha, age 8

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

Research Review 2007

UCL INSTITUTE OF CHILD HEALTH

Great Ormond Street Hospital for Children NHS Trust

Research Review 2007

Contents

Harry, age 5 weeks

The UCL Institute of Child Health, in partnership with Great Ormond Street Hospital, is the largest centre in Europe devoted to clinical and basic research and postgraduate teaching in children's health.

Jennifer Frost, PhD Student, Clinical and Molecular Genetics Unit

Dean's report

"Postgraduate education in a range of child health areas has long been a strength at the Institute, but during 2007 the Institute took a further step into undergraduate education."

For the UCL Institute of Child Health, like all university departments across the UK, 2007 was the year of the Research Assessment Exercise (RAE). The RAE evaluated all aspects of research performance during 2001–2007. The Institute submitted 134 senior research staff to the RAE, including 14 consultants in paediatric medicine at Great Ormond Street Hospital who, despite heavy clinical workloads, had achieved a particularly high level of research output. The Institute formed the largest part of an experimental medicine submission to the RAE, together with the UCL Institute of Ophthalmology and UCL Division of Medicine.

There were many other highlights during 2007. We welcomed Francesco Muntoni as Professor of Paediatric Neurology and head of the Dubowitz Neuromuscular Centre, a unit with an international reputation for research and clinical practice in muscular dystrophies. Professor Muntoni is one of three leaders (together with Professor Mike Hanna at the UCL Institute of Neurology, and our own Professor Martin Koltzenburg) of the newly formed Medical Research Council Centre for Neuromuscular Diseases, which opened in early 2008.

Also new to the Institute were Professor Tessa Crompton, who established a research team investigating the development of the immune system, and Professor Peter Hammond, who joined us from the Eastman Dental Institute to pursue research on the analysis of facial features in genetic disease.

Sadly, Professor Robert Surtees, one of the Institute's longest serving and most gifted academic paediatricians, died during the summer. Professor Surtees was widely known and respected for his expertise in all aspects of paediatric neurology, but with particular reference to neurochemistry and movement disorders. He is greatly missed by all his colleagues both at the Institute and hospital.

Further awards were achieved under the Higher Education Funding Council for England Clinical Senior Lecturer Scheme. In 2006, Dr Shamima Rahman and Dr John Anderson obtained one of these prestigious awards, and this continued in 2007 with awards to Dr Persis Amrolia, Dr Neil Sebire and Dr Russell Viner. We were also delighted that Dr John Achermann was awarded a Wellcome Trust Senior Clinical Research Fellowship.

While continuing to pursue its research interests throughout the paediatric specialties, the Institute has established an academic unit of General and Adolescent Paediatrics. Its leader, Professor Brent Taylor, moved to the Institute during the year, bringing with him the UCL undergraduate medical degree module in Child and Family Health. A long-term strategic goal is to build this new grouping into a significant element of the Institute's overall research portfolio, providing a higher profile than before in the area of general and adolescent paediatrics in the UK and beyond.

Postgraduate education in a range of child health areas has long been a strength at the Institute, but during 2007 the Institute took a further step into undergraduate education with the development of an intercalated BSc in International Health and Development, run by Professor Anthony Costello. This course forms an important part of a new UCL strategic development in Global Health, which brings together a range of UCL departments for which international health is a common research and educational interest.

During 2007, more of the Institute's senior staff were successful in UCL's promotions exercise than ever before. Helen Cross became Professor of Paediatric Neurology for research achievements in childhood epilepsy. Bobby Gaspar became Professor of Paediatrics and Immunology, for his groundbreaking research in gene therapy for immunodeficiency disease. Ruth Gilbert became Professor of Clinical Epidemiology for her research on childhood infection at a population level. Catherine Law became Professor of Public Health and Epidemiology, for her contributions to research-based public health policy for children.

Promoted to Reader during 2007 were: Dr Persis Amrolia for immunology of bone marrow transplantation, Dr Michelle de Haan for memory and the psychology of childhood perception, Dr Jugnoo Rahi for childhood visual impairment and prevention of blindness, Dr Lesley Rees for growth and heart disease in children with kidney failure, Dr Paul Riley for stem cells that can repair heart blood vessels, Dr Neil Sebire for pathology research into childhood and placental disease, Dr Andrew Taylor for heart imaging research, Dr Paul Veys for bone marrow transplantation and Dr Russell Viner for adolescent medicine and obesity. Promoted to Senior Lecturer were Dr Andrew Cook for education in congenital heart disease and Dr Eleanor Main for education and research in physiotherapy for lung disease.

As the year closed, the Institute embarked upon an in-depth re-evaluation of its research and academic strategy, in conjunction with our clinical partner Great Ormond Street Hospital. Having surveyed and evaluated research performance since 2001 in the RAE, it now seems appropriate to look forward over the next five years to determine the priority areas for research and development. As I write this report, we are in the final stages of developing our strategic plan, and I will return to this topic in next year's report.

Andrew Copp
Dean, UCL Institute of Child Health

Hannah, age 20 months

Chief Executive's report

"Together the UCL Institute of Child Health and Great Ormond Street Hospital form the largest paediatric centre in Europe dedicated to clinical and basic research."

As a hospital that values research as highly as the quality of clinical care, the last year has been particularly challenging. We have needed to start adapting to the loss of the annual NHS Culyer research and development block grant of £34 million and to find new alternative funding opportunities. It has been made more difficult because we face a number of other significant funding changes which affect the income we get for treating patients, all of which have had an impact on our financial position. Juggling all of this while applying to become a Foundation Trust and yet protecting the research which benefits our patients, has not been easy and we don't have all the solutions yet.

The good news is that we now understand what the opportunities are much more clearly and therefore should be able to maximise our chances of success in our funding applications. We can also start to see how this new system, which is completely transparent, will more explicitly benefit patients. The great disadvantage of the old block grant system was that we couldn't always be clear how the money supported specific research outcomes and outputs but these are totally transparent under the new system.

The successful award of Specialist Biomedical Research Centre status has been a good start. In partnership with UCL Institute of Child Health we are the only specialist centre dedicated to experimental medical research for children. Our success was based on our track record of being able to demonstrate that the programmes of research included in this bid will translate into real benefits for the children we treat at Great Ormond Street Hospital.

Some pieces of work are the building blocks for future research proposals. We think these will be much more difficult to fund under the new regime and yet losing these building blocks would have a real negative impact.

An example would be the renal and cancer databases that, with consent from children and families, consist of longitudinal data collected on patients for many years. Such data provides invaluable information, for example on the natural history of a particular problem or the long-term effects of different treatment regimes. Fortunately Great Ormond Street Hospital Children's Charity is willing to fund this sort of work as one of the ways it supports our research.

We also need to make sure we have excellent facilities for research. The opening of The Somers Clinical Research Facility later in 2008 will provide this, enabling children to be enrolled in pioneering research. Previously investigators have used any space they could beg, borrow or steal around the hospital when they wanted to do a study, which meant a lot of time was wasted. The Somers Clinical Research Facility will provide robust systems, processes and infrastructure to make research as easy as possible. Its staff will provide support to any investigator wishing to do a study. Many of the studies conducted in this area will also bring in additional funding from the Department of Health and elsewhere. This development is possible through the generosity of both the J N Somers Charitable Will Trust and Friends of the Children of Great Ormond Street.

Together the UCL Institute of Child Health and Great Ormond Street Hospital form the largest paediatric centre in Europe dedicated to clinical and basic research. Maintaining this pre-eminence in a competitive environment is not easy but is extremely important for the children who come to the hospital and to the children who don't.

Jane Collins

Dr Jane Collins
Chief Executive

Emily, age 4

Research and development report

During 2007, many of the recent changes in NHS research and development funding began to impact on the activity at Great Ormond Street Hospital and UCL Institute of Child Health (ICH).

The greatest impact thus far has been the establishment of the Great Ormond Street Hospital /ICH Specialist Biomedical Research Centre (BRC) focused on children's health, which was recently awarded to us in open competition. The award of centre status is recognition of the outstanding research and worldwide scientific and clinical importance of the work done by the joint institution and is the only BRC dedicated to children's research in the UK.

Research within the centre is focused on experimental medicine (proof of concept or first-in-human studies) and includes the following three research themes:
- Molecular basis of childhood diseases
- Gene, stem and cellular therapy
- Novel therapies for childhood diseases

The BRC was formally opened on 22 January, 2008 with the keynote address delivered by Dr John Gallin, director of the US National Institutes of Health Clinical Centre in Bethesda, Maryland. Dr Gallin runs the largest research hospital in the world based on the National Institutes of Health campus outside Washington DC.

As a result of this award we have been able to continue our experimental research programmes as well as implement a number of new initiatives. Academic training is an important part of the functioning of the BRC and we have allocated funds to appoint new PhD students, clinical research fellows, academic clinical lecturers and clinician scientists. We have also been able to allocate funds to pump prime new areas of experimental medicine.

"The award of centre status is recognition of the outstanding research and worldwide scientific and clinical importance of the work done by the joint institution and is the only Biomedical Research Centre dedicated to children's research in the UK."

This award also provides essential funds to staff our new Somers Clinical Research Facility, which will open in 2008. This bespoke, state-of-the-art facility will contain dedicated research space for outpatients and day cases and will be staffed by nurses trained in research methodology and good clinical practice. Modern IT systems will facilitate the conduct of high quality research for children.

This year the Special Trustees of Great Ormond Street Hospital Children's Charity have agreed to provide additional financial support for research across the joint organisation. This funding is allowing us to continue the excellent research undertaken at Great Ormond Street Hospital/ICH.

For the 2007/2008 edition of the Research Review, the focus has changed to highlight the excellent experimental medicine being undertaken as part of our Biomedical Research Centre, as well as translational research.

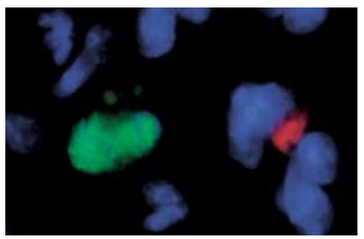
Professor David Goldblatt
Director of Clinical Research and Development

Ms Emma Pendleton
Head of Research and Development Office

Visualising different mouse tissue proteins

In the news

Stephen Cox, Head of Communications, reports on the top stories that hit the headlines in 2007.



Common gene variation protects against cancer

A common variation in the gene B-MYB might have a significant protective effect against a wide range of cancers. The study, led by Dr Arturo Sala was published in *Oncogene*. A particular polymorphism is present in 40 per cent of the Italian population, and the study identified that those carrying it had half the incidence of cancer (as a whole) compared with those in the control group. Dr Sala said: "This would suggest that we have found a key player in the genetic influences in cancer. Although the results are statistically significant, we would certainly want to see the scale of the effect confirmed in a much larger study." The study is of particular interest since the polymorphism is common; only one copy is needed for any effect, and it appears to affect a wide range of cancers. Incidence varies widely between ethnic groups.

A gene for 'stem-cell-ness'?

Researchers have identified a gene which appears key to the ability of blood stem cells to form the other cells in the blood. Translocations involving the MLL gene are the cause of most infant leukaemias, which have very poor survival rates. However, the function of healthy MLL was not previously known. Research by Dr Hugh Brady and colleagues in *Cell Stem Cell* (a new scientific journal) has shown that in mice where the MLL gene in blood stem cells was turned off, the cells lost the ability to make other cells. A working MLL gene seems crucial to making stem cells effective. Dr Hugh Brady said: "We do need to better understand the molecular biology of leukaemia. We think this gives us good insight. We're still a long way off understanding how children born with a damaged MLL gene might be prevented from developing the disease."

CHILDREN with LEUKAEMIA fund the research.

Allergy and eczema: a new explanation

The rising incidence of allergy and allergic diseases in the developed world might be attributable to excessive washing with harsh soaps and abrasive skin care products which strip away a protective layer of skin. Researchers challenge the so-called hygiene hypothesis, which claims that reduced exposure to infectious disease, particularly in childhood, weakens the immune system and allows allergic responses to emerge.

Professors Robin Callard and John Harper's paper published in *Trends in Immunology* brings together two separate lines of work; genetic investigation into defects of the protective skin layer, and direct experiment. Professor Callard explained: "Too much washing with strong soaps, using exfoliants and other such skin care products, and perhaps biological washing powders could be stripping away the skin's outer protective layer, resulting in allergic responses to allergens in susceptible individuals.

"We have shown that children and adults with a rare genetic skin disease who develop atopic dermatitis and allergy also have a weakened skin protective layer. In the lab, we have shown that if the outer protective layer of the skin is stripped away using something as simple as sellotape, allergens and other proteins are able to penetrate the skin and be taken up by specialised cells called Langerhans cells in the epidermis. The Langerhans cells then move from the skin to the local lymph nodes and induce the classic Th2 allergic immune response."

A community programme for obesity works

The MEND programme for tackling child obesity has statistically significant benefits for children, which have been sustained a full 12 months after they started the trial. MEND is the first obesity programme to have a successful Randomised Controlled Trial (RCT). Results for the 107 families following the nine-week intervention include statistically significant improvements in the children's body mass index, waist circumference, fitness, lifestyle and self-esteem.

MEND director Paul Sacher said: "Obviously sustaining a healthy lifestyle is the Holy Grail of health and fitness. The MEND programme is not a diet but rather helps overweight children and their families build a foundation for healthy living – for life."

Professor Alan Lucas, director of the Medical Research Council Childhood Nutrition Research Centre at the UCL Institute of Child Health, who oversaw the effectiveness study said: "Thirty per cent of UK children are now considered to be obese or overweight. It is an immense public health issue in both immediate and long-term health. Obesity costs the nation £7 billion a year. This popular community-based programme has the potential to underpin effective national strategies for obesity treatment and prevention."

200 million children fail to make intellectual and educational potential

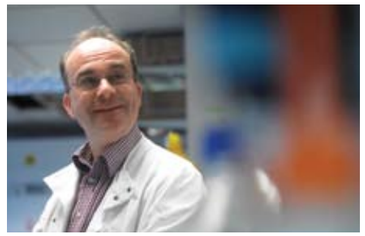
Professor Sally McGregor led a team of international experts in a pioneering series in *The Lancet*. She identified that intervention for children less than five years of age can make a significant difference in their ability to learn, and tackle global poverty. Better education not only lifts individuals out of poverty but is also proven to improve the life chances of their own children, and encourages better control by women over family size. Tackling these issues will bring benefits down the generations.

"Investing in programmes in children under the age of five will be both more effective and far cheaper than leaving it until later," Professor McGregor said. "Bluntly, we can make these children more intelligent and help them benefit more from education. We can show that the benefits of these programmes last into adulthood. These are not high-tech interventions. Research over decades in Jamaica (and other countries) has shown that women with only primary school-level education and a few home made toys can be trained to make a significant difference in the education, intelligence and mental health of disadvantaged children."

Great Ormond Street Hospital provides an almost unique resource of children with a wide range of disease conditions, thereby stimulating and enabling research.

Professor Francesco Muntoni, Head of the Dubowitz Neuromuscular Centre

Kidney chaos



Professor Adrian Woolf
Genes, development and disease

Biography

- Graduated in medicine, 1981
- Honorary consultant in nephrology at Great Ormond Street Hospital, 1994
- At ICH Nephro-Urology Unit since inception, 1997

Funding

- UCL Bogue Fellowship
- Wellcome Trust
- Kids Kidney Research
- Great Ormond Street Hospital Children's Charity
- Kidney Research UK

Collaborators

- University of Florida, USA: Dr R Johnson
- Regeneron Pharmaceuticals, USA: Dr G Yancopoulos and Dr J Rudge
- King's College London, UK: Dr L Gnudi
- Great Ormond Street Hospital/ICH: Professor P J Scambler, Dr J Pitera, Dr D Long
- Harvard Medical School, Boston, USA: Dr H Yuan

In the UK there are more than 30,000 people with severe kidney failure requiring transplant or long-term dialysis, which for many of them originated in childhood. There are some 1,000 children in the UK with severe kidney failure and Great Ormond Street Hospital treats 20 per cent of them.

Using fetal therapies to rescue malformed kidneys before birth and breaking the cycle of kidney damage after birth are two innovative areas of research under investigation by Professor Adrian Woolf's research team in the UCL Institute of Child Health (ICH) Nephro-Urology Unit.

In the UK, fetal ultrasound scans, to check whether major organs have developed normally, are routinely performed around the middle of pregnancy. Using this screening technique, kidney malformations are one of the common abnormal findings. In the most severe cases a baby is born with no kidneys, a condition called renal agenesis. The questions Professor Woolf and his colleagues are trying to answer are: why does agenesis happen; and can anything be done to make the kidneys grow before birth?

Babies born with a rare condition called Fraser syndrome have multiple birth defects, including kidney agenesis. Professor Peter Scambler's team (Molecular Medicine Unit) had previously discovered that a gene which is mutated in Fraser syndrome normally makes a protein that coats the surface of cells, and Dr Jolanta Pitera, a scientist working with Professors Woolf and Scambler, found that this same protein mediates interactions between kidney cells just at the moment the kidneys are forming. She discovered that in a model of Fraser syndrome, the embryonic kidneys begin to form but soon 'commit suicide' and fall apart by a process called apoptosis.

Dr Pitera also found that, by removing the part of the embryo that forms the kidneys and adding back specific molecules called growth factors, the Fraser syndrome kidneys resumed a more normal pattern of growth, at least in a Petri dish. This striking observation raises the question whether growth factors could be replaced in vivo, for example by giving them to a woman carrying a fetus with severely malformed kidneys.

In this way, the need for babies to undergo life-saving dialysis might be at least delayed until later in childhood. One challenge would be to establish whether the large growth factor molecules could move across the placenta into the embryo after being given to a mother, and more work is needed to establish whether this is feasible.

Another project, also involving growth factor therapies, investigates the possibility of intervention to stop the progression of kidney damage in children with abnormal kidneys.

Professor Woolf said once the normal physiology of a kidney has been interrupted for any reason, a cycle of damage can be triggered which leads to worsening function and the eventual need for transplantation or dialysis.

He explained: "The kidney is a very blood vessel-rich organ, with one-fifth of the cardiac output going through both kidneys. During the progression of kidney disease, tiny blood vessels called capillaries wither and die. Consequently the kidney tissue becomes starved of oxygen and this worsens scarring."

Following on from this, his team is investigating whether the progressive loss of kidney blood vessels can be prevented by giving vascular growth factors. Dr David Long, a scientist who completed his PhD at the ICH Nephro-Urology Unit and then studied in the universities of Texas and Florida, supported by a UCL Bogue Fellowship, is undertaking this work.

In the USA, he worked with Dr Rick Johnson, a pioneer of growth factor therapy for kidney disease. Back at ICH, Dr Long, currently a Kidney Research UK Senior Fellow, has been using vascular growth factors in models of kidney failure and the early results show that the cycle of capillary loss can sometimes be broken.

Professor Woolf said: "Similar vascular growth factors are already being used in humans to treat other diseases – for example, to improve blood flow in damaged hearts and limbs. However, like all powerful biological therapies, these factors can have side effects, and it is possible to have 'too much of a good thing'. Much work needs to be done in disease models with regard to the dosing and timing of these treatments."

Sections of kidneys ready to be analysed by microscopy

Decoding disease

Professor Phil Beales
Genes, development and disease

Biography

- Post doctoral studies at Baylor College of Medicine, Houston, Texas, 1999–2000
- Joined UCL Institute of Child Health (ICH), 2000
- Honorary consultant in clinical genetics at Great Ormond Street Hospital and Guys' and St Thomas' NHS Trust, 2000
- Wellcome Trust senior research fellow in clinical science at ICH, 2002
- Awarded chair of Medical and Molecular Genetics at UCL, 2005

Funding

- Wellcome Trust
- Medical Research Council
- Kidney Research
- Newlife (formally BDF)

Collaborators

- Great Ormond Street Hospital/ICH: Professor P Hammond and Professor P J Scambler
- University College London, UK: Dr M Tada, Dr R Mayor, Professor A Forge
- Johns Hopkins University, Baltimore, USA: Dr N Katsanis
- Simon Fraser University, Vancouver, Canada: Dr M Leroux
- Baylor College of Medicine, Houston, Texas, USA: Professor J Lupski
- University of Strasbourg, France: Professor H Dollfus

Understanding the molecular basis of childhood genetic diseases with obesity, especially those caused by dysfunction of cilia – hair-like projections from the cell's surface – is the focus of the work by Professor Phil Beales and his research team. "Scientists use the same shared technology to investigate the aetiology of many of these disorders," Professor Beales said.

New technology has enabled some of these conditions to be re-visited in the form of comparative genomic hybridisation (CGH) to provide new diagnoses that were not possible using traditional tests. Professor Beales explained these molecular forms of karyotyping have a far greater resolution, enabling the identification of small deletions or duplications of the chromosome not visible to the eye. He is using CGH to investigate unusual causes of obesity.

Professor Beales explained: "We are now starting to look at genes that might contribute to obesity, by studying 40 cases of obesity and mental retardation in children under the age of 16. So far, with the Cytogenetics Department, we have found five deletions and, two duplications; two of these recur in two or more patients, paving the way for potentially, identifying obesity-related genes."

Professor Beales is particularly interested in single gene syndromes like Bardet-Biedl Syndrome (BBS). A child born with this rare inherited syndrome has symptoms including retinal degeneration, early-onset obesity, learning difficulties, extra digits, genital malformation, organ reversal and renal dysfunction (including kidney cysts).

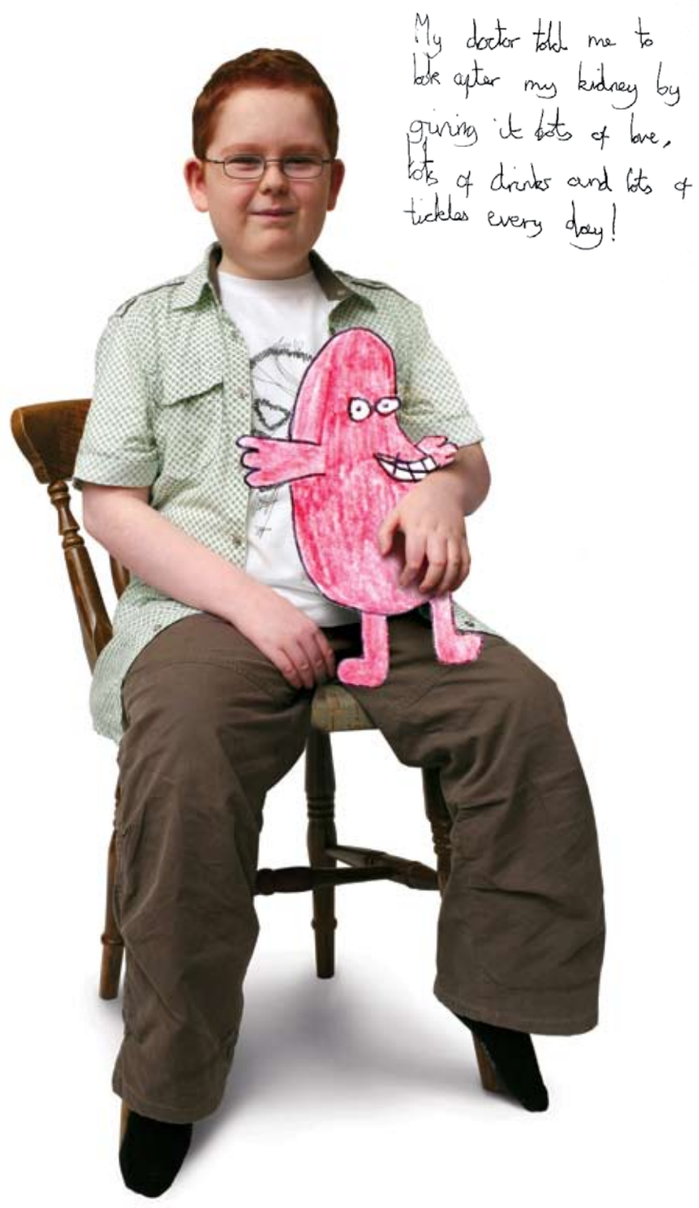
"BBS is caused by mutations in 12 genes, the identification of which has enabled us to develop diagnostic, pre-natal and carrier tests for at-risk families," he said. The syndrome cannot be cured but it is hoped that therapies can be designed with each problem in mind. Professor Beales has been investigating the role played by cilia in BBS. All cells in the body have cilia. They are often adapted to perform special functions and are important for early development.

A specialised form of non-beating cilia are found in the kidneys and the eyes and are responsible for the processes leading to the formation of left-right asymmetry in the developing embryo (defects of this process can cause organ reversal). All of these functions are impaired in BBS patients.

By creating mouse models of BBS, Professor Beales' group has made new discoveries in humans. For example, cilia in the nose, important for our sense of smell, were found to be lacking in BBS mice, which led to olfactory testing of BBS patients. Over half had difficulty with their sense of smell – permitting Professor Beales to implement another clinical aid to diagnosis.

BBS has become a byword for a new group of emerging syndromes, the 'ciliopathies', in which cilia are presumed to not work correctly. Professor Beales' group recently collaborated with Professor Peter Scambler and colleagues to study another disease called Jeune syndrome (JATD). Another rare genetic disorder, JATD patients characteristically have shortened limbs and a poorly developed ribcage, which can severely restrict breathing. By predicting that JATD may also be a ciliopathy, the team identified the first disease-causing gene, which it showed to be important for building a cilium. This revelation now expands the definition of a ciliopathy to include some diseases with abnormal bone and cartilage development.

Kidney cysts are a common feature of BBS, JATD and many other ciliopathies. Professor Beales' group is examining possible existing therapies that may alleviate or even prevent cysts forming and kidney failure. "Although these are still early days, my hope is that soon we can prevent or at least delay the onset of kidney failure in these patients," Professor Beales said. "We are trying to apply what we have learnt from animal models and from one disease to another."



Case study 1
Dan, age 11

Dan has Laurence-Moon-Bardet-Biedl Syndrome (LMBBS) also referred to as Bardet-Biedl Syndrome: a non-curable condition that can be life-limiting. Dan is treated by a multi-disciplinary team at Great Ormond Street Hospital and his mum, Tonia, tells his story.

"At four days old Dan was admitted to special care at our local hospital due to dehydration and weight loss, but four months later he started piling on the pounds and within one month was classed as clinically obese. At six months old he was re-admitted with a viral infection and tests showed he had impaired kidney function. He also presented with obesity, an extra digit and under-developed testes (hypogonadism). After a referral to Great Ormond Street Hospital for further tests, Dan was diagnosed with LMBBS.

One of the main features of this syndrome is loss of vision. Dan was already showing the other main symptoms but we had no idea about the eyes, which came as a bit of a shock. Dan didn't crawl until he was about a year old and didn't walk until he was nearly two. However he is a very bright boy and always has his head in a book.

We have had to maintain a very healthy low-fat, low-sugar diet and have managed to keep his weight under control. He copes brilliantly, demonstrating a willpower most of us can only dream about.

Dan's kidneys started to deteriorate rapidly in early 2006, and he had a pre-emptive live transplant (from me) performed at Great Ormond Street Hospital and his new kidney is functioning well.

Dan's condition is degenerative; his eyes will get worse and his new kidney may not last his lifetime but he is a happy, positive boy and doesn't get down about his disabilities."

Syndromes – a complete picture

Professor Raoul Hennekam
Genes, development and disease

Biography

- Qualified as physician in Utrecht, The Netherlands, 1980
- Trained as a paediatrician and clinical geneticist in Utrecht, 1984
- PhD thesis: Rubinstein-Taybi syndrome, 1990
- Professor of paediatrics and clinical genetics at Amsterdam University, where he still has a small appointment, 2002
- Professor in clinical genetics and dysmorphology at Great Ormond Street Hospital/ICH, 2005
- Research focus: dysmorphology, skeletal dysplasias, causes of birth defects, autism, and the natural history of syndromes

Funding

- Birth Defects Foundation Newlife
- March of Dimes
- Cerebra

Collaborators

- Great Ormond Street Hospital/ICH: Professor P Beales, Dr B Gibbons, Professor P Hammond, Professor J Harper, Professor G Moore, Professor P J Scambler and Dr A Shaw
- Academic Medical Centre, Amsterdam: Dr M Alders and Professor C Van der Horst
- Children's Hospital of Eastern Ontario, Ottawa, Canada: Professor J Allanson
- National Institutes of Health, Maryland, USA: Dr L Biesecker
- University Hospital of Utah, Salt Lake City, USA: Professor J Carey
- University of Birmingham, UK: Professor C Oliver

Building a complete picture of a syndrome for the patient and their family, and using this information to help treat other patients, is the work of Professor Raoul Hennekam. He works with small groups of patients with rare conditions, such as Tricho-rhino pharyngeal (TRP) syndrome and Rubinstein-Taybi Syndrome (RTS).

TRP is a condition characterised by an unusual face, for example a pear-shaped nose, and all kind of bone anomalies, including frequent hip problems. It is caused by a small genetic error on chromosome 8.

RTS is an equally rare condition characterised by a small stature, again remarkable facial characteristics, broad thumbs and big toes, a delayed development and unusual behaviour, including a very poor short-term memory. It is caused by a defect in a gene called CBP. Professor Hennekam said: "Our first goal is to know how people with these syndromes are doing, and to give that information back to the families. The behaviour of children with syndromes is studied in particular, to create a complete picture."

He explained paediatrics and genetics had come a long way in diagnostics, after many years of establishing which patients, and which diseases, belonged together. Now the next step is knowing more about the natural history of all these entities.

The Medical Research Council has set up the Patient Research Cohorts Initiative Call, and Professor Hennekam and his team are applying to be a part of this initiative. It is hoped that by studying groups of patients with the same syndrome over a long time, they will gain a better understanding of their syndromes, improved diagnosis and improved care.

But creating such cohorts of patients has another advantage: it creates a very well defined group, facilitating the start of any new medical interventions. "To intervene you need to know a group of patients very well, know their background," Professor Hennekam explained. "We need to know what would happen if we did not do anything, before we can study what happens if we do something."

Some of the characteristics in rare syndromes like TRP and RTS can also be present in people without the condition, such as hip problems found in TRP patients or short-term memory problems in those with RTS.

"By studying the patients who have these syndromes closely we can do something for people who may not have them, but still have problems such as bad hips," Professor Hennekam said. "So in fact studying groups of patients with rare entities helps us understand medical problems that are frequently encountered in everyday life."

His team has very recently started the clinic, with the help of the patient support groups of the two syndromes, whom they've known for years. Dr Adam Shaw is performing most of the work in the clinic.

"I make use of the unusual collaboration between a world-class children's hospital and an equally world-class research institute. This way we can easily build further bridges between Great Ormond Street Hospital and the UCL Institute of Child Health," Professor Hennekam said. "Our follow-up will give results now but will also be very important in the future, creating a clinical treasure for anyone who wants to study the syndromes. In the end, this will translate into the best possible care for all children with these syndromes, and for others that show some of the complications associated with it."

Pedigree of a family with a genetic condition

Reproductive development

Dr John Achermann
Genes, development and disease

Biography

- Qualified in medicine at Cambridge University, 1990
- General paediatric, paediatric endocrinology and clinical research training in London (MD 1998)
- Fellowship in molecular medicine at Northwestern University, Chicago, USA, 1998–2001
- Wellcome Trust Clinician Scientist at UCL, 2002–2006
- Wellcome Trust Senior Fellow in Clinical Sciences with a clinical and research focus on disorders of adrenal and sex development, steroidogenesis and nuclear receptors, 2006

Funding

- The Wellcome Trust
- Coodenaçao de Aperfeiçoãmento de Pessoal de Nivel Superior, Brazil

Collaborators

- Great Ormond Street Hospital/ICH/UCL: DSD clinical team, Dr D Gerrelli, Dr M Hubank, Dr K Mills and Dr J Pallas
- Birmingham University, UK: Professor W Arlt
- University of Cambridge, UK: Professor I Hughes
- Barts and The Royal London Hospitals, London, UK: Professor A Clark
- National Institute of Medical Research, London, UK: Professor R Lovell-Badge
- Northwestern University, Chicago, USA: Professor J Jameson
- University of Texas Southwestern, Dallas, USA: Professor R Auchus
- University of California, San Francisco, USA: Professor W Miller
- Charité University, Berlin, Germany: Professor A Grüters

Dr Achermann's research results are recorded for the patient, the family and future generations

Understanding human adrenal and reproductive development, and taking that knowledge into the clinic, is the focus of Dr John Achermann's work.

The adrenal glands and gonads have a common embryonic origin, and so many aspects of their development and regulation are shared. The adrenal gland secretes hormones, which control vital functions such as blood pressure and glucose levels. Adrenal complaints are difficult to diagnose and can be life threatening, but can be treated with steroids.

Disorders of sex development can lead to a spectrum of problems such as ambiguous genitalia in neonates, failure to enter puberty at adolescence or infertility in later life. Some of these children have adrenal problems too, so making a prompt and appropriate diagnosis is important.

Dr Achermann is part of the hospital's Disorders of Sex Development (DSD) clinic and works with a multi-disciplinary team including urologists, gynaecologists, biochemists, geneticists and psychologists. The clinic aims to diagnose and plan the management of patients with DSD and is an example of translational research, where science and clinical research work side-by-side.

"Disorders of adrenal or gonad development and function can have a wide range of causes," Dr Achermann said. "Many of these conditions can present with a similar clinical picture, but can require very different approaches to management."

Dr Achermann believes that trying to get an exact diagnosis is important, but in many cases the specific problem is not known, and individual cases may be rare.

"In situations where the gonad doesn't form properly, we probably make a specific diagnosis in about 20 per cent of cases," he explained. "The diagnosis might influence gender assignment, options for fertility and the risk of developing a tumour. Working as a multi-disciplinary team is essential to pool everyone's skills and experience."

Dr Achermann has been studying the role of SF-1, a transcription factor which turns genes on and off, and its associated nuclear receptor, DAX-1, in human adrenal and reproductive development and disease. Initial work focused on patients with adrenal and gonadal problems, however recent findings have shown that milder changes in SF-1 can be found in approximately 15 per cent of individuals who have impaired production of androgen hormones and milder defects in the development of the testes.

He said: "It seems that adrenal function is normal in these cases, but will need careful monitoring as they might develop adrenal failure in the future."

SF-1 may also be a key regulator of many other aspects of adrenal and gonadal development and function. The researchers have been using various approaches to up and down regulate SF-1 in cells. Dr Achermann said: "We are hoping that by regulating it we can identify new target genes and proteins. These new genes could turn out to be important factors responsible for adrenal and reproductive disorders in patients where the cause currently isn't known."

SF-1 variability in a UK population cohort is another area Dr Achermann is investigating. His findings will reveal the impact of milder changes of SF-1 in the whole population. Understanding the biology of these disorders of adrenal and reproductive development and function helps patients manage their condition. Long-term monitoring of these children into adulthood is vital.

Dr Achermann and his team are trying to find causes for the remaining cases of adrenal development and gonad problems where a specific diagnosis currently cannot be reached.

Non-toxic treatment

Dr Paul Veys
Cancer

Biography

- Qualified at St Bartholomew's Hospital Medical School, 1983
- Trained in Haematology/BMT at The Royal London Hospital, The Royal Free Hospital, Great Ormond Street Hospital and SickKids in Toronto, Canada
- Director of Blood and Marrow Transplant Unit at Great Ormond Street Hospital since 1994
- Reader in stem cell transplantation in UCL Institute of Child Health Molecular Immunology Unit, 2007
- Research focus: reduced-intensity conditioning regimens

Funding

- National Commissioning Group
- Leukaemia Research Fund
- Cord Blood Charity
- Great Ormond Street Hospital Children's Charity
- Olivia Hodson Cancer Fund

Collaborators

- Great Ormond Street Hospital/ICH: Dr P Amrolia, Professor B Gaspar, Dr N Goulden, Dr W Qasim and Professor A Thrasher
- Southampton University Medical School, UK: K Orchard, Consultant Haematologist

Reducing the toxicity of Acute Leukaemia (AL) treatment and prompting graft-verses-leukaemia responses is an area of investigation by Bone Marrow Transplant (BMT) Consultant Dr Paul Veys.

Treatment by high dose chemotherapy, radiotherapy and stem cell transplantation is the usual option for children with high-risk AL – this eradicates the leukaemia and suppresses the immune system so healthy cells can replace the diseased bone marrow.

However BMT is very toxic, with a high risk of death either from the treatment itself, or relapse of the leukaemia. Dr Veys is exploring whether treatment 'targeted' to the bone marrow using radio-labelled and non-labelled antibody therapy, in conjunction with stem cell transplantation, may allow an increase of therapy to be delivered to disease-specific sites, while sparing non-diseased tissue.

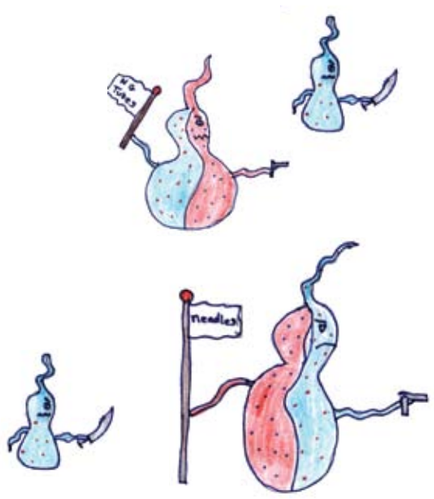
Developing a reduced or minimal intensity transplant for AL is being sought following success with a similar approach in genetic diseases. "Preparation for transplant in AL has historically involved full-intensity regimens. If you cut back the drugs before transplantation in leukaemia, the disease may relapse quickly," Dr Veys said. "So we are trying to maximise the treatment to the bone marrow, where the AL hides, and reduce the side effects of the treatment elsewhere in the body."

In some cases it may be the new graft that cures the leukaemia and not the drugs. "If the cancer is not cured after the first BMT we have been able to achieve a cure with a second reduced-intensity procedure, this time promoting a graft-versus-leukaemia response to cure the leukaemia," he added.

This idea has challenged the concept that the patient's cells need to be completely destroyed by drugs and radiation to achieve cure of the leukaemia.

Having reduced-intensity treatment also means more patients could be treated. Very sick children (with organ damage), who wouldn't have been able to be treated with full-intensity therapy, could be treated with minimal intensity BMT.

Looking to the future, Dr Veys thinks we will continue to use less and less high dose chemotherapy and radiotherapy in BMT in children. "Once we can make the graft cells eradicate the leukaemic cells more specifically then we can reduce the need for high doses of chemotherapy and radiotherapy," he said. "This will allow us to reduce the long-term side effects of BMT such as growth problems, second tumours and infertility."



Case study 2
Alex, age 11

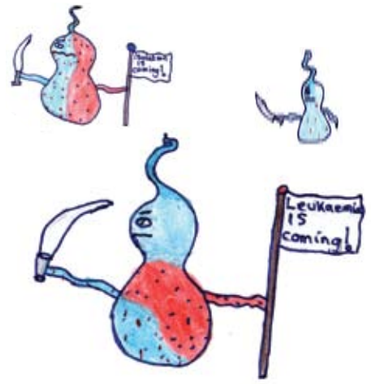
Alex was diagnosed with Juvenile Myeloid Monocytic Leukaemia (JMML) when he was just six years old. Alex's parents, Peter and Angela, tell his story.

"Alex was lethargic, pale and quiet. When doctors told us Alex had leukaemia we felt that the bottom had fallen out of our world. He was taken to Great Ormond Street Hospital and diagnosed with JMML.

Alex was put on the Bone Marrow Transplant (BMT) waiting list and in May 2003, an unrelated donor was found. Alex began chemotherapy in preparation for the BMT and was in hospital for 10 weeks recovering. He lost all his hair because of the chemotherapy and spent most of the time in isolation, which he found hard.

In April 2004 Alex relapsed and needed another BMT. His consultant at the time, Dr Paul Veys, said Alex needed another course of chemotherapy and a second reduced-intensity transplant, followed by experimental treatment with a drug called interferon to increase the 'graft versus leukaemia' effect of the second graft.

Alex's response to the second BMT was fantastic and since then he's been doing really well."



The vaccine approach

Dr John Anderson
Cancer

Biography

- Qualified in biochemistry at Oxford University, 1986
- Qualified in medicine at Newcastle University, 1991
- Trained in paediatrics and developed an interest in oncology
- PhD thesis at the Institute of Cancer Research on sarcoma oncogenic fusion protein transcription factors, 1995–1998
- Joined Great Ormond Street Hospital/ICH, 1998
- Research focus: the interaction between tumours and the immune microenvironment and identification of new approaches to enhance therapy

Funding

- Cancer Research UK
- SPARKS
- Department of Health
- Children's Cancer and Leukaemia Group

Collaborators

- University of London School of Pharmacy, UK: Professor D Thurston
- UCL Department of Haematology, UK: Dr M Pule
- Royal Free Hospital, London, UK: Professor H Stauss
- Institute of Cancer Research, Sutton, UK: paediatric oncology clinicians and scientists

Dr John Anderson and his team in UCL Institute of Child Health (ICH) are using vaccine approaches to treat childhood cancers by using the patient's own immune cells and developing targeted responses to cancer.

The vaccine approach is much less toxic for patients who are routinely given radiotherapy and chemotherapy to treat the cancer. Made in the gene and cell therapy laboratory at Great Ormond Street Hospital, this vaccine uses cells from the patient's own immune system to attack malignant cells.

Clinical trials are part of the research and include a dendritic cell vaccination for osteosarcoma (bone cancer) trial funded by Cancer Research UK (CRUK). Eight patients with relapsed disease have been recruited onto the trial so far and these are patients who have no other treatment options.

Dr Anderson additionally runs several laboratory-based projects in the area of developing cancer immunotherapy. The first is redirecting the immune system to create a response. "This involves understanding the immune environment, redirecting immune response to tumours and combining this with other, more conventional therapeutic approaches," Dr Anderson explained. "We are developing this by increasing our understanding of the basic biology of childhood cancers."

Another project involves a disease-based approach – trying to identify proteins found in most diseases that could be suitable for targeted therapy. It is an experimental approach predicting cell surface markers and confirming their expression. The research involves using human blood and tumour cells to manipulate immune cells in culture and testing their therapeutic applications in models of paediatric cancers. "We are moving towards cloning the T-cell receptors and genes with a view to creating reagents for clinical therapy," Dr Anderson said.

Combining immunotherapy progress with investigating the basic biology of cancer cells and dendritic cells is another project. The work has involved identifying patients who will have an immune response against their own tumour and identifying the natural target for that immune response.

Other work involves studying how chemotherapy and vaccines could work together in the clinical setting to treat high-grade gliomas, which have a poor prognosis. The molecular basis of the immune environment is also being researched. This has involved looking at new inhibitors and small molecule targets, for example, STAT-3. A collaboration has been funded by CRUK with the University of London School of Pharmacy to develop new inhibitors of STAT-3. "The work is a massive collaborative effort – we rely on research nurses for sample collection and trial recruitment, and on all the oncologists and radiologists," Dr Anderson said. "It could not work if we did not have a joint ICH/Great Ormond Street Hospital setting."

Techniques such as the Elispot assay allow quantification of immune responses

UCL Institute of Child Health pursues an integrated, multidisciplinary approach to enhance understanding, diagnosis, therapy and prevention of childhood disease.

Harrison, age 3

Paediatric nutrition and health prediction

Dr Atul Singhal
Biochemistry

Biography

- Qualified at the Royal Free Hospital and trained in paediatrics, 1986
- Appointed head of the Clinical Trials and Nutrition in Cardiovascular Disease Group at the Medical Research Council Childhood Nutrition Centre, 1998
- Reader in paediatric nutrition, 2004
- Honorary Consultant Paediatrician at the Whittington Hospital, 1998
- Honorary consultant paediatrician at Great Ormond Street Hospital, June 2003

Funding

- Medical Research Council (MRC), UK

Collaborators

- Great Ormond Street Hospital/ICH:
  - Vascular Physiology Unit: Professor J Deanfield
  - Centre for Paediatric Epidemiology and Biostatistics: Professor T Cole
- Dr Singhal also works closely with several academics and consultant paediatricians from a national network of collaborating hospitals where much of the clinical research is conducted

Dr Atul Singhal and the research team in the Medical Research Council Childhood Nutrition Research Centre are investigating obesity, cardiovascular disease and the link with childhood nutrition and growth patterns. Dr Singhal said that paediatric nutrition is a key environmental determinant of health, with a major impact on cardiovascular disease (CVD) risk, cognitive functioning and diseases with large health costs, such as obesity.

The centre has four major programmes to address areas of public health importance influenced by nutrition: early origins of cardiovascular disease, cognition and the brain, bone health and body composition, growth and obesity.

Dr Singhal's work focuses on two main areas: the importance of nutrition for weight gain in infancy and long-term cardiovascular health, and obesity in childhood and the risk of later heart disease.

Research has shown that the pattern of infant and childhood growth has a major impact on cardiovascular disease. For example, atherosclerotic cardiovascular disease has a long pre-clinical phase with development of pathological changes in the arteries of children and young adults decades before overt clinical manifestations of the disease.

Nutritional factors in both infancy and childhood have been shown to be important in this process and affect lifetime cardiovascular disease risk. For instance, breast-feeding is associated with benefits for long-term cardiovascular risk factors, possibly as a consequence of a slower pattern of growth in breast-fed compared with formula-fed infants.

An intervention study in small full-term infants showed those randomly assigned to a standard formula for the first nine months had lower blood pressure six to eight years later than infants fed a nutrient-enriched formula which promoted growth.

"The World Health Organisation has accepted that formula-fed babies have been overfed, as the energy content in formula has been overestimated," Dr Singhal said. "There is on-going research to reduce the amount of energy and protein in formula milk to make it more like breast milk."

It is now accepted that slower infant growth is better for long-term health, and particularly the risk of obesity. "Some 30 per cent of obesity in adults could be explained by infancy growth," Dr Singhal said. "Optimising nutrition and growth in infancy will be an important public health-based intervention and could help in the primary prevention of cardiovascular disease, the most important cause of death in the populations of Western countries."

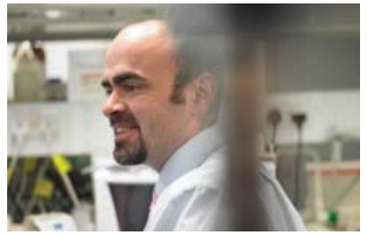
There is evidence that being obese as a child increases the risk of heart disease in later life. Dr Singhal said that the MEND programme, designed by Clinical Research Fellow Paul Sacher and one of the first community-based randomised trials for obesity treatment in the UK, had shown quite a marked improvement. Nationally, more than 28,000 children will use the obesity intervention.

The research group is currently looking at the effects of weight loss on blood vessels to try to understand the effects of childhood obesity on long-term CVD.

"Half the population is overweight and there are three factors governing this: development, environment and genetics. We can't do much about our genes but we can do something about early nutrition," Dr Singhal said. "Changing the mindset of parents to prevent overfeeding in infancy is important. We cannot get rid of the risk altogether but we can educate mothers about the importance of childhood nutrition."

Dr Singhal's case study files

Stem cell potential



Dr Paolo De Coppi
Biochemistry

Biography

- Graduated, completed surgical training and awarded PhD in tissue engineering at University of Padua, 2002
- Research Fellow at Harvard University, Boston, USA, 2000–2002
- Assistant Professor Paediatric Surgery, University of Padus 2002-2005
- Clinical Senior Lecturer and Honorary Consultant at ICH, 2006

Funding

- Telethon Foundation
- Association Français de Microminéralogie, Française Contre Les Myopathies
- The Royal Society, UK – Royal Society Wolfson Laboratory Refurbishment Grants
- Fondazione Citta' della Speranza, Malo, (VI) Italy

Collaborators

- Great Ormond Street Hospital/ICH: Professor A Copp, Dr P Ferretti, Dr N Taphar, Dr A Burns, Dr J Sowden, Dr M Lythgoe, Dr W Qasim, Dr P Riley and Professor B Gaspar
- Laboratoire de thérapie cellulaire et génique Hôpital Necker Enfants Malades, Paris, France: Professor M Cavazzana Calvo
- Wake Forest Institute for Regenerative Medicine, North Carolina, USA: Professor Anthony Atala
- Istituto M Negri, Bergamo, Italy: Professor G Remuzzi
- University of Padua, Italy: Professor N Elvassore

The potential of amniotic stem cells in therapy is being investigated by Great Ormond Street Hospital paediatric surgeon and UCL Institute of Child Health (ICH) researcher Dr Paolo De Coppi. In collaboration with a US research group, Dr De Coppi has investigated the potential of these cells, and found they had the ability to become many different types of cells and to be used to replace damaged tissues.

The group's research paper, *Isolation of amniotic stem cell lines with potential for therapy,* was published as the cover article in *Nature Biotechnology* in January 2007 and prompted much debate.

Dr De Coppi explained it might be possible to correct defects in the womb using these amniotic cells. "In a congenital malformation, such as heart disease, you can get a diagnosis of the condition at 21 weeks gestation," he said. "In theory you can take cells from the fluid that surrounds the fetus, culture them and use them to repair the defect."

Babies with a cardiac condition do not need treatment before birth, because the heart works differently in the womb as the blood does not need to be oxygenated by the lungs.

"However they often need an operation soon after birth," Dr De Coppi said. "If you can create a tissue and implant at birth that would be ideal. We are trying to extract the cells prenatally and make them differentiate into different tissues, then use this tissue to create a material to implant in vivo after birth."

The researchers have collaborated with various groups in order to understand the biological characteristics of these cells. The cells have some characteristics in common with embryonic stem cells, but unlike these cells do not form tumours when transplanted.

These cells also grow very easily, unlike adult stem cells. Dr De Coppi explained: "These cells are ideal for therapy. You can get lots of them because they grow faster and they have better potential than adult stem cells. We want to understand the characteristics of these cells and to know how far we have to induce them in vitro before putting them into a patient.

"For example, do they need to be committed to perform a function before they are inserted? We've been using animal models to investigate."

In a mouse model the cells were introduced and successfully started producing key chemicals in the brain and the liver.

Dr De Coppi has been collaborating with Professor Agostino Pierro and Research Fellow Dr Simon Eaton, and there are four people working on these cells constantly as part of the project.

"We are trying to establish tissue engineering, and we are the first centre outside the USA to be using these cells," Dr De Coppi said. "My appointment here was to develop tissue engineering. We have received a grant from the Royal Society and are opening the new tissue engineering centre in May."

Professor Pierro said the centre would start by trying to engineer oesophagus tissue, as it was quite simple, and would then move on to other tissues and organs. "It's very exciting to start this collaboration with surgery, gastroenterology and chemical engineering," he said.

Dr De Coppi's research is looking at the possibility of using cells taken from the fluid that surrounds the fetus to correct defects

Epilepsy – alternatives to drug treatment

Professor Helen Cross
Neuroscience

Biography

- Qualified at Birmingham University, 1984
- Paediatric training in Birmingham and paediatric neurology training at Great Ormond Street Hospital
- Consultant at Great Ormond Street Hospital since 1996
- Senior Academic in neurosciences since 2000
- Research focus: neuroimaging, the role of early surgical intervention and new treatments in childhood epilepsy.

Funding

- Epilepsy Research UK
- The Foyle Foundation
- SPARKS
- Scientific Hospital Supplies International
- Milk Developing Council
- The Bailey Thomas Charitable Trust Fund
- CURE

Collaborators

- Great Ormond Street Hospital/ICH: Professor B Neville, Dr R Scott, Dr M Lawson, Dr C Clarke, Professor D Gadian, Mr W Harkness, Professor F Vargha Khadem, Dr T Baldeweg, Dr M de Haan
- UCL Institute of Neurology, London, UK: Professor S Sisodiya, Professor L Lemieux
- Free University of Brussels, Erasme Hospital, Belgium: Dr X de Tiege
- Austin Health, Melbourne, Australia: Professor I Scheffer
- Children's Memorial Hospital, Chicago, USA: Professor D Nordli
- Comprehensive Epilepsy Programme, National Institutes of Health, Washington DC, USA: Dr W Gaillard
- University od California, Los Angeles, USA: Professor G Mathern
- Johann Wolfgang Goethe-University, Frankfurt am Maine, Germany: Dr H Laufs
- Pediatric Hospital A. Meyer-University of Firenze, Florence, Italy: Professor R Guerrini

Strawberries, yoghurt and double cream; a breakfast option for patients on a ketogenic diet

The effect of diet, imaging and early intervention in epilepsy patients is being investigated by neurology consultant Professor Helen Cross and her collaborators at UCL Institute of Child Health (Wolfson Centre) and Great Ormond Street Hospital. Professor Cross and her team have been conducting randomised trials and investigations of patients to determine ways of controlling the condition where medication has failed.

About 75 per cent of children with epilepsy respond to drug treatment. "The children that we see are resistant to drug treatment," Professor Cross explained. "We are developing ways to treat these children, and also deal with the associated learning and behavioural problems."

Treatments for these children range from surgery to dietary. New ways of brain imaging are being used as a non-invasive way to find the area of the brain responsible for epilepsy, as seizures may arise from one area. In particular, work is being done to combine EEG and MRI to get a functional MRI.

Combining these imaging techniques means areas of the brain that may be responsible for seizure onset may light up, helping to localise brain abnormality. This type of imaging is a step forward in treating patients with surgery – which involves removing the small section of the brain causing the epilepsy. Professor Cross and her team are identifying more patients who might be suitable for this ongoing trial. Professor Cross has also been exploring whether early intervention could be beneficial with a cohort of children with epilepsy in north London. It is thought that children with drug- resistant epilepsy are not being seen early enough to have an impact on their learning and behaviour in the long term.

"We get the impression that children are coming to us late for consideration for surgery – when they've been having seizures for many years," Professor Cross said. "Our hypothesis is that if children suitable for surgery were recognised early, their developmental outcome may be optimised."

The cohort of children is being seen from diagnosis and the plan is to extend the study up to five and then 10 years. "It's a way of tackling the condition from different angles," Professor Cross said. "We think these children might be better off with early intervention and surgery but it's not proven that this might be the case as we do not know exactly what happens to the range of children who present at this age. The study will explore the natural history of these children so information acquired can be used as the basis for further studies."

One treatment that Professor Cross has been investigating is the ketogenic diet; a diet that is high in fat but low in carbohydrate and protein that mimics the body's response to starvation, replacing glucose with fats as a major energy source. Broken down fat produces ketones, and it is proposed that these may control seizures.

This is not a new treatment, but up until the randomised controlled trial conducted by Professor Cross and her team, there was no evidence that either the classical diet or the more recently developed medium chain triglyceride diet could be effective to control the condition. In the first randomised controlled trial completed by Professor Cross and her team the diet worked as well as any new anti-convulsant drug, and that each of the two ways of giving the diet were equal in their effect.

These results mean parents are able to choose the diet that suits their child's needs. Similar findings were seen after three, six and 12 months on the diet.

Gene therapy for muscular dystrophy

Professor Francesco Muntoni
Neuroscience

Biography

- Qualified at Cagliari University, Italy, 1984
- Completed a two-year MD thesis on the mechanism of ethanol-induced reward studying the electrical activity of dopaminergic neurons in rats, 1984
- Trained in child neurology and psychiatry in Sassari, Italy, 1989
- Postgraduate training period at the Hammersmith Hospital in London, 1993
- Imperial College London and Hammersmith, 2003–2007: Professor of Paediatric Neurology and Honorary Consultant in Paediatric Neurology, Head of Dubowitz Neuromuscular Centre, including the Nationally Commissioned Group Centre for Congenital Muscular Dystrophies and Congenital Myopathies, Great Ormond Street Hospital/ICH, from end of 2007
- Great Ormond Street Hospital/ICH, from end of 2007
- Research focus: genetic causes, translational research including clinical trials in muscular dystrophies

Funding

- Wellcome Trust
- Muscular Dystrophy Campaign
- Department of Health
- Medical Research Council
- Jennifer Trust for Spinal Muscular Atrophy
- Muscular Dystrophy Association, USA
- European Community

Collaborators

- Great Ormond Street Hospital/ICH: Professor A Pierro, Professor G. Moore, Dr J Morgan and Professor A Thrasher.
- Imperial College Healthcare NHS Trust, London, UK: Dr S Brown; Professor D. Wells
- UCL Institute of Neurology, London, UK: Professor M Hanna and Dr M Reilly
- Newcastle University: Professors K Bushby and Volker Straub

Correcting the faulty gene that causes the progressive muscle-weakening condition muscular dystrophy is the area of research interest for Professor Francesco Muntoni. Muscular dystrophy is a life-limiting condition and there are several types: congenital, Duchenne and adult onset.

Professor Muntoni has been investigating the most common form of the condition, known as Duchenne muscular dystrophy. The genetic defect responsible for this condition is linked to the X chromosome and affects one in every 3,500 boys. Boys with this type of the condition have an inability to produce the protein dystrophin, which keeps the muscle intact.

There are a number of approaches that are being used to treat these disorders, including experimental research into gene therapy in collaboration with Professor Adrian Thrasher that aims to use the patient's own stem cells to correct the defective gene that causes the disorder. Dr Jenny Morgan, a member of the neuromuscular centre, is trying to isolate the best cells to use in this work. The advantage of this approach is that it avoids the need for donor cells and the immunosuppressant drugs the patient would have to take for life.

Professor Muntoni and his research team started a proof-of-concept trial in December 2007, funded with a five-year grant from the Department of Health, to create a molecular bridge to 'span' the missing part of the gene – allowing shortened but functional proteins to be formed.

This concept has worked well in models, and similar success is expected in the trial. Patients on the trial were injected with the molecular bridge and a muscle biopsy was taken to see if the protein was present.

Once this intramuscular study is complete the next stage is systemic administration, where the molecular bridge is injected into the bloodstream, rather than each muscle. This will be funded by the Medical Research Council.

This step would remove the need for injections into individual muscles. Professor Muntoni said an exciting piece of research was investigating the repair of the molecular bridge permanently and correcting the gene – stopping the need for injections altogether.

Professor Muntoni said that the advances in treatment and care of patients with muscular dystrophy has improved: over the past 40 years the average life expectancy has doubled to 30 years old.

Another piece of research, undertaken by Dr Susan Brown, and Drs Silvia Torelli and Martin Brockington in the Dubowitz centre is identifying a novel mechanism where a gene can be switched on and off to enable the protein to work. The problem in some variants of muscular dystrophy is not so much the lack of a structural protein in the muscle, but lack of a number of enzymes which put sugar on certain parts of a structural protein called dystroglycan, which enable it to function.

"We have demonstrated in various cellular models that there is a particular pathway where you can increase the number of sugars you can put on the protein dystroglycan," Professor Muntoni said. "We are currently interested in finding drugs that could enhance this particular pathway as this could be beneficial for a number of muscular dystrophies with reduced sugar content in dystroglycan."

Separating muscle cell proteins by size

Repairing the retina

Dr Jane Sowden and Professor Robin Ali
Neuroscience and immunology

Biography: Professor Robin Ali

- Lecturer, Division of Molecular Genetics, UCL Institute of Ophthalmology & Molecular Immunology Unit, UCL Institute of Child Health, 1991, (joint appointment)
- Professor of Human Molecular Genetics, Division of Molecular Genetics, UCL Institute of Ophthalmology & Molecular Immunology Unit, UCL Institute of Child Health, 2003, (joint appointment)
- Appointed Head of the Division of Molecular Therapy, UCL Institute of Ophthalmology, 2004
- Elected to the Academy of Medical Sciences, 2007
- Faculty of UCL/Moorfields Eye Hospital Biomedical Research Centre for Ophthalmology, 2008

Biography: Dr Jane Sowden

- Medical Research Council Career Development Award from UCL Institute of Opthalmology, 1996
- Appointed Lecturer at ICH, 1998
- Appointed Senior Lecturer at ICH, 2002
- Research focus: Developmental biology of the retina with a focus on the analysis of factors regulating retinal stem cells and their potential use for retinal repair

Funding

- Medical Research Council
- Fight for Sight
- Macula Vision Research Foundation
- Ulverscroft Foundation

Collaborators

- Moorfields Eye Hospital/UCL Institute of Ophthalmology, London, UK: Dr R Pearson, Dr R MacLaren
- University of Michigan, USA: Dr A Swaroop
- City University, London, UK: Professor R Douglas

Upright microscope and imaging system used to capture digital images of tissue and cells at high resolution

An exciting breakthrough in restoring vision in blind mice has potentially major implications for patients with untreatable eye conditions. A collaborative research programme between UCL Institute of Child Health (ICH) and UCL Institute of Ophthalmology is testing the idea that damaged photoreceptors in the eye can be repaired by transplanting new cells into the retina of a mouse model. Dr Jane Sowden is a research leader in this programme and, together with Professor Robin Ali, runs the research programme at ICH.

Retinal diseases leading to loss of photoreceptor cells are one of the most common causes of blindness. For example, inherited retinal degenerations affect one in 3,000 people with conditions such as retinitis pigmentosa causing progressive loss of vision, and Leber's congenital amaurosis causing childhood blindness. The retina cannot repair itself once the photoreceptors die, so the idea of transplanting cells to create new photoreceptor cells has always been attractive to researchers.

"There has been a longstanding interest in this idea for several decades and recent research has involved transplanting stem cells that have the potential to become different types of cells," Dr Sowden said. "The outcomes had not been successful in previous attempts, as the transplanted cells failed to move to the correct location in the retina to make photoreceptor cells."

The problem was how to direct the transplanted cells to differentiate and become new photoreceptor cells that make connections with the visual centres in the brain. The research group decided to try transplanting cells at a later developmental stage and were pleased to discover that the transplant worked. "The problem has been selecting the cells at the correct stage of development," Dr Sowden said.

Cells were taken from the peak rod photoreceptor genesis stage of development in the mouse model, when the retina is about to be formed, and were successfully transplanted and integrated into the retina of young mice. "If we take these same stage cells and put them into the adult or degenerating retina it works also. If the cells are partially instructed and then put into an adult they are still able to make photoreceptor cells." This means the mature system is still receptive to new cells, provided the cells are committed to becoming photoreceptors before they are transplanted. "This is important as it suggests it might be possible to repair the damaged retina and restore vision," Dr Sowden said.

A photoreceptor precursor was labelled with a gene encoding a fluorescent protein so the process of making new photoreceptor cells could be detected. Blind mice, which were given the new cells, also responded to light sensitivity tests.

The next step is to use a renewable source of cells in the laboratory to create cells at the correct stage of development for transplantation. "We need to make rod precursors by reproducing the process that normally occurs in development," Dr Sowden said. "The cells will be given transcription and growth factors in an attempt to make these cells identical to the cells created naturally." This process benefits from use of the microarray and FACS (Fluorescence Activated Cell Sorting) technologies at ICH enhanced by new funding for the Great Ormond Street Hospital/ICH Biomedical Research Centre.

Also, many more cells need to be produced for this research programme. Currently, hundreds are being used to improve light sensitivity in mouse models, but quantities need to be in the thousands, so better outcomes are achieved and can be assessed. One potential source of these cells is from the patients themselves – which would avoid rejection of the new cells.

Even though the team's research is a long way from clinical trials, this research opens up the potential for treating blindness in humans.

UCL Institute of Child Health aims to improve the health and wellbeing of children, and the adults they will become, through world-class research, education and public engagement.

Ana, age 4

Gene therapy and the immune system

Professor Bobby Gaspar (left) and Professor Adrian Thrasher (right)
Immunology

Biography: Professor Bobby Gaspar

- Trained in paediatrics at King's College Hospital, London, BSc 1986, MBBS 1989
- PhD in the Molecular Immunology Unit at ICH, 1998
- Professor of Paediatrics and Immunology at ICH, 2007
- Research focus: Different aspects of primary immunodeficiency including understanding the molecular and cellular defects and disease pathogenesis, bone marrow transplantation for severe immunodeficiencies and the development of gene therapy for these conditions

Biography: Professor Adrian J Thrasher

- Consultant Clinical Immunologist at Great Ormond Street Hospital, 1998
- Wellcome Trust Senior Clinical Fellow in the Molecular Immunology Unit at the ICH, 1999
- Professor of Paediatric Immunology at ICH, 2002
- Director of the Centre for Immunodeficiency at Great Ormond Street Hospital/ICH with clinical responsibilities for care of patients with primary immunodeficiency syndromes, 2006

Funding

- Medical Research Council
- Department of Health
- Leukaemia Research Fund
- Wellcome Trust
- Chronic Granulomatous Disease Research Trust
- Primary Immunodeficiency Association
- European Union

Collaborators

- Numerous collaborators within Great Ormond Street Hospital/ICH including clinical and academic staff
- Purine Research Lab, Guy's and St Thomas' Hospital, London, UK: Dr L Fairbanks
- EUFETS AG, Germany
- Numerous European and international collaborations

Creating a safer way to treat children with immunodeficiencies is the work of Professors Bobby Gaspar and Adrian Thrasher. The pair have been investigating the potential of gene therapy for patients with inherited diseases of the immune system: the severest being SCID (Severe Combined Immunodeficiency), when a patient is born with no immune system.

The existing treatment for these patients is a Bone Marrow Transplant (BMT), which is very successful if a full donor match is available. However a BMT can have serious side effects and complications if there is no matched donor and a BMT from a mismatched donor has to be undertaken.

For this reason, they have been working on alternative treatments including gene therapy. "We have been running treatment trials for three different inherited immune deficiencies," Professor Gaspar explained. "We have now treated 19 children on these three different trials, and most have had good clinical outcomes."

The children treated on the trials have a problem or mutation in genes that affects either the development or function of cells in their immune system. Essentially, Professors Gaspar and Thrasher have been trying to develop a form of genetic medicine with which to treat them. "We are going back to basics and trying to repair the problem at a genetic level," Professor Gaspar explained. "We can manufacture a working copy of the gene in our laboratory and this is carried by a disabled virus which then has the ability to enter and insert the gene into the child's own cells."

Gene therapy involves using the child's own cells – and so it can avoid problems like Graft-versus-Host Disease (GvHD), where the transplanted cells begin to attack the patient. In addition, either there is no need for chemotherapy, or milder forms of chemotherapy are used.

Professor Thrasher said it could be a safer procedure, certainly in the short term. In the case of X-linked SCID (X-SCID), a single faulty gene can be responsible for the failure of the immune system in boys, and has been treated at Great Ormond Street Hospital with gene therapy. This involves bone marrow being removed, treated with an engineered retro-virus which carries the corrected gene to these cells, and returned to the child to generate immune cells. In time the child will develop a working immune system.

This therapy has also successfully treated children with other disorders such as ADA-SCID (Adenosine Deaminase) and X-linked Chronic Granulomatous Disease (X-CGD).

The treatment of X-SCID does not require chemotherapy, so a patient spends just one day in hospital and is then treated as an outpatient.

This therapy is an example of how the close relationship between the UCL Institute of Child Health (ICH) and Great Ormond Street Hospital creates an ideal environment to combine science and clinical medicine. Professor Gaspar said: "It has been very rewarding to move the research we have done in our laboratories into treating patients. We are also very lucky to have a big support team of individuals who are involved in the work that we do."

A major component of the work that Professors Gaspar and Thrasher do has helped Great Ormond Street Hospital/ICH achieve Biomedical Research Centre status.

Looking to the future, they want to develop gene therapy to make a safer and more effective way to treat patients with other conditions. "We hope that the work that has been done on these severe immunodeficiencies will now allow gene therapy to be developed for other genetic diseases such as disorders of the blood such as thalassaemia and sickle cell disease, and metabolic disorders."

Case study 3
Bradley, age 2

Bradley was just three months old when he was first admitted to Great Ormond Street Hospital and was diagnosed with ADA-SCID. His parents Karen and Mark tell his story.

"We had been in and out of our local hospital for tests since Bradley was two weeks old. He had started to vomit, had diarrhoea, a persistent cough and developed an ear infection. After several chest X-rays and two sweat tests for Cystic Fibrosis (CF), Bradley was referred to a respiratory consultant who re-tested for CF and took another chest X-ray which showed that his ribs were splayed. The consultant analysed Bradley's blood results and found that his lymphocyte and neutrophil counts were very low. Within four days he was admitted to Great Ormond Street Hospital for further tests and was diagnosed with ADA-SCID.

For the first year of his life we had to make weekly trips to Great Ormond Street Hospital for Bradley to receive PEG-ADA enzyme replacement injections and he was put on prophylactic medication and intravenous immunoglobulin. His inability to produce a healthy immune system meant we had to follow very strict guidelines to keep him safe from infections. We became isolated as a family; our only visitors were health professionals and grandparents.

Bradley received gene therapy in May 2006 and his condition has improved considerably; he is developing a healthy immune system and is now beginning to do all the things normal two year olds do.

We have always received excellent support from Bradley's Consultant, Professor Bobby Gaspar, and the immunology team."

Heart vessel inflammation

Professor John Deanfield
Cardiorespiratory

Biography

- Undergraduate training at Churchill College, Cambridge and The Middlesex Hospital, London
- Subsequently trained at The Hammersmith Hospital, London, 1976–1977, and Great Ormond Street Hospital, 1983–1984
- Consultant Cardiologist at The Heart Hospital, London since 1998 and Great Ormond Street Hospital since 1984
- British Heart Foundation Vandervell Professor of Cardiology, ICH, 2003
- Principal research focus: vascular medicine concentrating on the role of the endothelium in atherosclerosis

Funding

- British Heart Foundation

Collaborators

- UCL Eastman Dental Institute, UK: Professor N Donos and team and Dr A Hingorani
- University of Bristol, UK: Professor G Davey-Smith
- Great Ormond Street Hospital/ICH: Professor N Klein

Professor John Deanfield's group has been leading in the field of the vascular biology of pre-clinical atherosclerosis. "We know that classical risk factors, such as cholesterol, smoking and hypertension are important, but recently unexpected inflammatory influences have been implicated in the development of early arterial disease," he said.

The group has examined the link between both acute and chronic inflammation and changes in the vessel wall.

The Avon Longitudinal Study of Parents and Children (ALSPAC) looked at the impact of acute common childhood infections on the vascular endothelium, which is known to be a key factor in early arterial disease. Professor Deanfield worked with the ALSPAC Group in Bristol; a very large follow-up study of 14,000 children born in 1991.

They used non-invasive ultrasound (flow mediated dilatation of the brachial artery) to quantify arterial ejection in endothelial function and investigated the impact of acute or recent common childhood infections, such as cough or sore throats in 600 10-year-old children. Based on parents' questionnaires, children were classified into three groups: those with an acute illness at the time of study, children who had an illness within one month of study and a third group for children without recent illness.

Endothelial function was significantly lower in the children with acute infections than in the non-invasive controls. Endothelial function was also significantly lower in the children with recent illness, but was less impaired than in the acute illness group. No long-term hygiene or infection history measures predicted endothelial function.

The sub-group of children who had had infection were asked to return a year later for further vascular testing while they were well. In almost all cases, endothelial function had recovered to normal levels. This study showed that minor childhood infections could lead to a change in arterial endothelial function in normal children.

Professor Deanfield and Professor Nigel Klein are currently studying the long-term impact of more severe or chronic inflammatory conditions on arterial structure and function. This may be an important risk factor for the silent development of arterial disease from early life.

In collaboration with Professor Nikos Donos' team at the UCL Eastman Dental Hospital, the research team has been examining a common chronic source of generalised inflammation – gum disease – and its effects on vascular function.

Some 120 young adults with periondontitis but no signs of cardiovascular disease were randomised to two forms of oral treatment. The first received a standard cycle of scaling and polishing, while the second had a more intensive regime of plaque removal and/or dental extraction.

A range of measurements of endothelial function was made before dental treatment and for six months afterwards at regular intervals. The standard treatment produced little resolution of the gum disease and had no effect on the arterial wall; in contrast the intensive periodontal protocol produced an acute systemic inflammatory response for a few days, which was associated with a significant transient impairment in endothelial function.

As gum hygiene improved, endothelial function rose steadily so that by six months, there was significantly better endothelial function than at base line. This is the first evidence from a clinical trial, that the level of a systemic source of chronic inflammation has a direct impact on arterial wall function. This highlights not only the potential importance of good oral care for cardiovascular disease prevention, but also the opportunities to reverse the abnormal vascular biology associated with atherosclerosis by novel treatment approaches.

Professor Deanfield and his team are working with collaborators Professor Donos and Professor Aroon Hingorani at UCL to determine in a new trial whether recovery of endothelial function is associated with a slowing of progression of markers of structural changes in the arterial wall.

"Waiting for clinical events before intervening in adults with arterial disease is not the best strategy," Professor Deanfield said. "We need to invest in our arteries from much earlier if the impact of cardiovascular disease in the next generation is to be reduced."

Chloe, age 5

Heart-valve replacement – sharing innovation

Professor Philipp Bonhoeffer
Cardiorespiratory

Biography

- Trained at University of Milan, Italy, 1983–1990
- Worked at Necker Hospital, Paris, France, 1994–2001
- Great Ormond Street Hospital, 2001
- Research focus: development of non-surgical techniques for treatment of valvar diseases

Funding

- Medtronic

Collaborators

- Great Ormond Street Hospital/ICH: Dr S Schievano, Dr P Lurz, Dr J Nordmeyer, Dr A Taylor and Dr S Khambadkone

Rolling out a worldwide educational programme so hospitals and centres can perform an entirely new heart-valve replacement technique is part of the work being done by Great Ormond Street Hospital Cardio-Respiratory Professor, Philipp Bonhoeffer.

Professor Bonhoeffer pioneered the technique whereby a patient can have a valve implanted in the pulmonary artery without opening the chest. The one-hour procedure uses a catheter inserted through the leg to guide the valve gently through the heart and into the pulmonary artery. This has enormously improved the outlook for children and adults with congenital heart conditions by replacing the need for the major open-heart surgeries.

This procedure is now being rolled out worldwide and Professor Bonhoeffer and his team are demonstrating the technique to clinical institutions. "The last part of translational research is moving new techniques into the clinical environment safely," he said. "The procedure can be dangerous if inappropriately used. One has to look at transferring the knowledge about the potential problems with the procedure and make decisions about choice of institution and expertise of operators in order to minimise the risk to patients when the technique is introduced elsewhere."

Professor Bonhoeffer explains that the operator needs to be experienced in interventional cardiology and have a good understanding about safety. "Operators also need to have a team to support them," he said.

Since September 2006, a monthly educational programme has been running for investigators from all over the world who are now introducing pulmonary implantations in their countries. They spend a whole day talking about potential problems of performing the heart valve procedure. "During the second day they see a procedure and see things in practice," he explained.

Professor Bonhoeffer said in the future he wanted to develop a simulator, which would allow trainees to perform the procedure with hands-on experience. This would reduce the risk of operators performing the technique with too little experience in clinical practice.

Investigators bring their patients' files to be discussed and the suitability of each case is weighed up in an expert environment so no mistakes are made in the patient selection. The next step is then to help plan the procedure and assist with the heart valve implant at their own centres by the formal proctors of the programme.

"They become independent only when the proctor and the cardiologist feel they have reached the level of necessary expertise. However, they will always be supported by the expertise of the product specialists who are employed by the valve manufacturer," Professor Bonhoeffer said.

"With this programme we could treat more than 400 patients, and with our meticulous introduction we have been able to keep the procedure mortality to zero. Worldwide, a new hospital is starting to use the procedure every week," he added.

As a result, the programme roll-out requires a lot of travel on behalf of the proctors – who currently support many of the procedures performed.

"We had some teething problems but now the institutions know what they need to do and are prepared," Professor Bonhoeffer said. "There is more technology to come. Completing our cycle of translational research with the first valve will help us to do this again with much more ease when further treatment options are ready for clinical practice in the near future."

Case study 4
Thomas, age 13

Thomas has been treated at Great Ormond Street Hospital for his congenital heart condition since he was one day old. His parents Mark and Karen tell his story.

"Thomas has pulmonary atresia with ventricular septal defect. A few hours after he was born nurses noticed he was blue, and tests revealed something wrong with his heart. He was transferred to Great Ormond Street Hospital for immediate surgery and had a shunt fitted. After a year a bigger shunt was fitted.

At the age of four Thomas needed open-heart surgery. He had a homograft (cardiac valve) inserted and a hole in his heart repaired, but after a few months the homograft had narrowed and needed to be stretched by a balloon catheter.

When Thomas was 11 years old his Consultant, Dr Phillip Rees, introduced us to Professor Philipp Bonhoeffer who told us Thomas would benefit from a new heart-valve procedure; removing the need for open-heart surgery.

After Thomas had the heart valve replacement procedure we could not believe how pink he was. After just one night in hospital, he was allowed home and the next day he was full of energy.

However in 2007 Thomas started to tire easily. He had further tests and Professor Bonhoeffer and his team decided he needed another valve, which was successfully performed. Thomas visits Great Ormond Street Hospital for regular appointments, and will need open-heart surgery to replace the homograft when he is 15. Thomas is in good health at the moment."

You can watch Thomas' first heart-valve procedure on the hospital's children and families health information website Children First for Health: www.childrenfirst.nhs.uk/teens/gosh_tv/thomas/index.html

Childhood immunisation

Dr Helen Bedford and Dr David Elliman
Population health sciences

Biography: Dr David Elliman

- Qualified at St George's Hospital, 1973
- Joint post at Great Ormond Street Hospital and Islington PCT, 2003
- Honorary Senior Lecturer at ICH, 2000
- Research focus: preventive medicine, particularly immunisation and screening. Collaborated with various members of the Centre for Paediatric Epidemiology and Biostatistics

Biography: Dr Helen Bedford

- Trained and practiced as a nurse and health visitor, 1980
- Joined ICH in 1986 on a large study with Professor Catherine Peckham looking at the determinants of vaccine uptake, and immunisation has remained a major interest
- Conducted research into the long-term outcome of meningitis in infancy, work that formed the basis of PhD, 2000
- Director of the MSc in Community Child Health, 2002
- Teaches on ICH-based courses as well as being involved in immunisation training for health professionals in a number of Primary Care Trusts

Funding

- Economic and Social Research Council – Professor H Joshi for the Millenium Cohort Study
- Department of Health – Professor C Law and Ms A Pearce to conduct immunisation studies

Collaborators

- Great Ormond Street Hospital/ICH: Professor C Law, Ms A Pearce, Professor T Cole, Professor C Peckham and Professor C Dezateux
- International Centre for Child Studies, Institute of Education, Bristol, UK: Professor N Butler (deceased)

Drs Helen Bedford and David Elliman work in the area of childhood immunisation: Dr Elliman as a consultant in community child health and Dr Bedford as a senior lecturer in children's health in the Centre for Paediatric Epidemiology and Biostatistics. Their collaboration is an example of successful working between UCL Institute of Child Health (ICH) and Great Ormond Street Hospital.

Both Dr Elliman and Dr Bedford have been involved in several studies focusing on immunisation. One study set out to explore the immunisation status of inpatients at the hospital. Evidence already existed to show that hospital inpatients, in general, are often under-immunised and they were concerned that this may also apply at Great Ormond Street Hospital.

Together with Dr Suzanne Walton, an audit of patients' records was conducted and it was found that a significant proportion of patients were not up-to-date with their routine vaccines. "Children treated at Great Ormond Street Hospital often have complex medical conditions and this can mean that health visitors, GPs and parents worry about the safety of immunising them," Dr Bedford said. Dr Bedford explained that the hospital had an important role in setting an example and promoting public health, as well as looking after children with complex conditions. "We wanted to raise the profile of immunisation in the hospital and train nurses in immunisation, so that they are able to give vaccines if it is acceptable to parents," she said.

As part of the study, the recording of patients' immunisation status by hospital staff was examined and it was found that in many cases it lacked the necessary detail. "We have now developed a form which means a more detailed history can be recorded and have also been involved in training hospital staff in the current immunisation schedule," Dr Elliman said.

Another aspect of the population immunisation work is based on the Millennium Cohort Study – a cohort of over 18,000 children born in the UK in 2000/01 who are being followed up at intervals.

A number of studies have been carried out by the Millennium Study Child Health Group led by Professor Carol Dezateux. The first study conducted with Dr Lamiya Samad, looked at factors determining whether children received the primary vaccines at two, three and four months. In this study the researchers were also able to look at those children who had set out on the immunisation course but had not completed it.

When the children were three years of age, information was gathered about the uptake of the MMR vaccine. Parents were asked if their child had the combined vaccination, single vaccines or no MMR vaccination at all. "In the first study we found that mothers of children who had no immunisations at all tended to be older and more highly educated. Mothers of children who had an incomplete immunisation course tended to be more disadvantaged and were often young and less well educated."

These findings suggest that different interventions are needed for different groups. Parents who decline vaccinations require evidence-based information and an opportunity to discuss any concerns they may have. For other parents all that might be needed is improved access to immunisation, and taking advantage of any opportunities to have the child immunised that may arise. This is when immunisation in hospital is important.

The aim of gathering such evidence is to inform policy and practice and in this case the results of these studies are very timely: the National Institute for Health and Clinical Excellence (NICE) is about to start to formulate recommendations on best practice in interventions for reducing inequalities in childhood immunisation.

Case study 5
Lydia, age 16 months

Lydia was seven months old when she contracted measles and ended up fighting for her life. She was too young to be immunised and caught the disease because older children around her had not been immunised. Her mum Annette tells her story.

"In mid-August 2007 Lydia developed an excessively running nose and a worsening cough. By the end of August she was still unwell and my husband and I took her to A&E. The doctors prescribed pain relief and advised us to keep her comfortable at home.

Forty-eight hours later florid rashes began to appear on Lydia's face, chest, back and neck. I took her to our GP who immediately suspected measles and sent us back to hospital. The paediatrician confirmed the diagnosis and prescribed more pain relief, as well as antibiotics and eye drops, as Lydia now had conjunctivitis and significant breathing difficulties too.

In the early hours of the following morning, Lydia became so tired her lips turned blue: we raced back to the resuscitation unit. Her heart was racing and it was difficult to stablise her temperature. She was put on a ventilator and transferred to Great Ormond Street Hospital. Lydia remained there for nine days – three of those on a ventilator. She also needed a blood transfusion.

I never, ever suspected Lydia might have measles. I was shocked to see how rapidly she deteriorated.

Children in the UK are not vaccinated until they are aged between 12 and 15 months (before this age the vaccine does not work very well).

I am pleased to say Lydia made a full recovery."

Science-based policymaking

Professor Catherine Law
Population health sciences

Biography

- Trained in paediatrics in London (Northwick Park, Hammersmith, Great Ormond Street and Royal Brompton Hospitals), 1980–1985
- Trained in epidemiology and public health at Johns Hopkins School of Hygiene and Public Health in Baltimore, Maryland, USA and Maryland State Health Department, USA, 1985–1987
- Worked at the Medical Research Council Environmental Epidemiology Unit, University of Southampton, UK, 1987–2003
- Worked with regional and national government since 1992
- Director of the Centre for Policy Research ICH Centre for Paediatric Epidemiology and Biostatistics since 2003
- Chair of the Public Health Interventions Advisory Committee of the National Institute for Health and Clinical Excellence, since 2005
- Research focus: child public health, particularly physical growth, inequalities in health, and the use of research for public policy

Funding

- Department of Health
- INVOLVE

Collaborators

- Great Ormond Street Hospital/ICH: Dr D Elliman, Ms M Collins, Professor C Dezateux and Professor T Cole
- Haringey Teaching Primary Care Trust, London, UK: Dr A Connolly
- University of York, UK: Professor H Graham
- York and Humber Public Health Observatory, UK: Professor B Ferguson
- National Children's Bureau, London, UK: Ms C Shaw and Ms L Brady

Increasing the use of science and scientific method in the field of policymaking in children's health is the overall aim of Professor Catherine Law and her team.

"We do this by collating scientific information and by trying to conduct our research in ways which are useful to policymakers," she explained. "Often these aims can be achieved by phrasing the research question slightly differently or attempting to interpret findings through a policymaker's eyes."

The team recently analysed the relationship between maternal employment and children's health. The Government has pledged to put an end to child poverty and some of the main features of this policy are incentives and schemes to get one or both parents into paid employment. Although it is known that unemployment is bad for adult health, very little work has been done on how maternal employment is related to child health. This is important as the number of women with young children in the workforce is increasing.

The team's analysis of the Millennium Cohort Study showed that maternal employment was associated with higher rates of early childhood obesity, particularly when the mother worked longer hours.

"This caused quite a stir in the national press," said Professor Law, "with some commentators questioning whether this research should have been conducted at all. They felt it victimised women. However, I feel it is important that we try to find out what the unintended consequences of policies are." The team is now carrying out a systematic review of the literature to assess what other research may link maternal employment to child health.

One piece of evidence that is often missing from policy development, and also from research, is the perspectives of children. Yet including children's views can have a positive influence on the development of policies and interventions. Professor Law adds: "Furthermore, we know that children carry their views forward into their adult lives and that this, in turn, influences how they bring up their own children."

Professor Law said it had been a privilege to work with the National Children's Bureau on a pilot project seeking views from young people on public health research and policy. "We set up a young people's reference group on public health and the group members have acted as consultants to research projects about young people, presented their work at the INVOLVE conference and produced a young person's version of the Association of Public Health Observatories Indications report on Child Health." Following the successful pilot, the group and its work will be expanded in the next year.

Policymaking is not just about government – local organisations also have to make decisions and children's perspectives should also inform these decisions. In collaboration with Haringey Teaching Primary Care Trust, Professor Law and her team ran a multi-methods research project to gain children's views on how the environment influenced their diet and physical activity patterns. Children took photos, drew maps and discussed in groups what was important to them. The results are being used locally to plan services.

Professor Law said research on child health needed to fit into a bigger picture. "On the Public Health Interventions Advisory Committee at NICE (National Institute for Health and Clinical Excellence), which I chair, we are trying to use the evidence base for improving population health through the development of cost-effective public health practice. NICE pioneered these methods for clinical practice and I am enjoying the challenge of developing them for public health."

Dr Law's research files

The ICH's multidisciplinary approach is necessitated by the important principle that the child is not merely a small adult.

Dr Marta Fernandez-Fuente, Post-Doctoral Scientist in comparative neuromuscular disease

Awards, honours and prizes 2007

Staff from UCL Institute of Child Health (ICH) and Great Ormond Street Hospital received national and international recognition for their research achievements during 2007.

Dr Noraishah Abdul Aziz was awarded a PhD for the thesis: *Molecular basis of neural fold adhesion and fusion in closure of the spinal neural tube.*

Dr John Achermann was senior author on the abstract that won the Henning Andersen Prize (Clinical) at the 46th Annual Meeting of the European Society for Paediatric Endocrinology, Helsinki. Dr Achermann was also invited to join the Annual Meeting Steering Committee of the US Endocrine Society and awarded the RCPCH/SPARKS Young Investigator of the Year Medal 2007 by the Royal College of Paediatrics and Child Health.

Dr Rachel Agbeko won the World Federation of Paediatric Intensive Care Society's Best Basic Science Award for her paper entitled: *The association of mannose binding lectin deficiency and systemic inflammatory response syndrome is unaffected by other complement and cytokine single nucleotide polymorphisms* at the 5th World Congress on Paediatric Critical Care, Geneva.

Professor Robin Ali was awarded the first Academy of Medical Sciences Foulkes Foundation Medal.

Dr Persis Amrolia was awarded a Higher Education Funding Council for England (HEFCE) clinical lectureship.

Dr Susannah Bailey won the Fairbairn Award for her oral presentation to the British Society for Gene Therapy in Warwick, and was awarded a PhD for the thesis: *Self-inactivating retroviral vectors for gene therapy of X-linked severe combined immunodeficiency.*

Ms Suzanne Bartington was the winner of the oral presentation prize for her presentation: *Factors associated with maternal report of varicella in pre-school aged children in the UK* at the European Medical Student Conference, Berlin.

Ms Kate Bennett was awarded a travel fund by the British Society of Investigative Dermatology to present: *A proteomic approach to identify targets of LEKTI – a protein implicated in the barrier function of the skin* at the annual meeting of the British Society for Investigative Dermatology (BSID).

Dr Michael Blundell was awarded a PhD for the thesis: *Molecular investigations into Wiskott-Aldrich syndrome* and was also a recipient of the John Lipscombe Memorial Travel Award for travel to present at the European Society for Gene and Cell Therapy meeting in Athens.

Dr Mario Cortina Borja was appointed to the Royal Statistical Society Publications Network Committee.

Dr Parjeet Boughan was awarded a PhD for the thesis: *Innate host defence to Helicobacter pylori.*

Dr Daniel Brewer was awarded a PhD for the thesis: *Modelling the p53 gene regulatory network.*

Dr Anastasios Chanalaris was awarded a PhD for the thesis: *Effects of the urocortin family of peptides on cardiac neonatal myocytes. Antiapoptotic and hypertrophic effects.*

Dr Sirinuch Chomtho was awarded a PhD for the thesis: *The influence of early nutrition and growth on body composition in childhood and early adult life.*

Dr Suparna Choudhury was awarded a PhD for the thesis: *The development of social cognition during adolescence.*

Dr Tanzina Chowdhury was awarded a PhD for the thesis: *Investigation of the role of MLL-ENL in leukaemogenesis.*

Professor Tim Cole was elected to become a fellow of the Academy of Medical Sciences.

Professor Helen Cross has been made president elect of the British Paediatric Neurology Association (BPNA). Professor Cross has also been made an Ambassador for Epilepsy by the International League Against Epilepsy (ILAE), awarded during the 27th International Epilepsy Congress held in Singapore.

Dr Phillippa Cumberland received an award from the Dean's Travel Fund towards expenses to attend and make an oral presentation at the annual meeting of the Association for Research in Vision and Ophthalmology in Fort Lauderdale, Florida.

Professor Mehul Dattani was appointed as Chair of the Scientific Evaluation Committee of E-RARE (EU call for research into rare disorders).

Professor Carol Dezateux took part in a media briefing discussing concerns raised by the Connecting For Health NHS IT project on behalf of the Wellcome Trust and the Medical Research Council (MRC). Professor Dezateux was also invited as an expert witness to address the Health Select Committee along with colleagues from The Wellcome Trust and the UK Clinical Research Collaboration, and to give evidence to the Home Affairs Committee at the House of Commons along with colleagues from the Academy of Medical Sciences.

Dr Louisa Dunlevy was awarded a PhD for the thesis: *Investigating the role of the methylation and folate cycles in neural tube closure.*

Dr Patrizia Ferretti was invited to take part in a one-day workshop concerned with Medical Research Council (MRC) future strategy for supporting stem cell research and in particular discuss the need for additional stem cell lines, and was invited by the MRC to join a stem cell expert group to give expert opinions on fellowship applications in the field. Dr Ferretti was also invited to join the *Open Neurology Journal* as an Editorial Board Member and recently received an invitation to join the *Open Tissue Engineering and Regenerative Medicine Journal* in this capacity.

Dr Katy Fidler was awarded a PhD for the thesis: *The role of mannose binding lectin in infection and inflammation.*

Ms Claire Gannon was awarded first prize in the Biomedical Sciences category at the UCL Graduate School poster competition for a poster entitled: *The teashirt3 transcription factor marks a novel renal tract lineage and controls smooth muscle development in the ureter.*

Dr Faith Gibson was awarded the prestigious Fellowship of the Royal College of Nursing. It acknowledged her outstanding contribution to the advancement of the nursing of children and young people in the specialty of paediatric oncology, and her exceptional contribution to nursing research and leadership across international boundaries.

Professor Ruth Gilbert was appointed a member of the Department of Health Advisory Committee on Antimicrobial Resistance and Healthcare-Associated Infections. Professor Gilbert was also appointed to the Paediatric Medicines Expert Advisory Group (PMEAG) of the Medicines and Healthcare Products Regulatory Agency.

Professor David Goldblatt was appointed co-chair of the Immunology and Infectious Disease Funding Committee at the Wellcome Trust (for three years) and to the Wellcome Trust Pathogens, Immunology and Population Health Strategy Committee.

Professor Sheila Haworth was awarded a CBE in the New Year's Honours List. This award acknowledges Professor Haworth's contribution to cardiovascular science and clinical care at ICH and Great Ormond Street Hospital over many years.

Dr Wendy Heywood won the poster prize and the best talk at the annual Special Advances in Fetal Medicine meeting in Bristol.

Dr Steven Howe was a recipient of the John Lipscombe Memorial Travel Award for travel to present at the American Society of Gene Therapy in Baltimore, USA.

Dr Elina Hypponen was appointed as an honorary senior lecturer at Imperial College, Department of Epidemiology and Public Health, and invited as a member for the Medical Research Council (MRC) College of Experts.

Dr Marianne Jacobsen was awarded a PhD for the thesis: *Regulation of endothelial E-selectin molecule expression by neisseria meningitidis.*

Dr Tom Jacques passed the Membership of the Royal College of Pathologists (MRCPath).

Dr Shalini Jadeja was awarded a PhD for the thesis: *Fraser syndrome and mouse blebbed mutants.*

Dr Barbara Jefferis was awarded a PhD for the thesis: *How do childhood cognition and life-course health behaviours affect adult glucose homeostasis?*

Dr Dagan Jenkins was awarded a PhD for the thesis: *Genetics of human renal tract malformations.*

Dr Mazyar Kanani was awarded a PhD for the thesis: *Clinico-morphologic integration in the surgical repair of atrioventricular septal defect with common atrioventricular junction.*

Ms Yukiko Kimura was awarded second prize for her presentation: *Development of a method to measure glutathione synthesis by GC-combustion/high temperature conversion-irms* at the SIMSUG 2007 Conference held in Newcastle.

Professor David Latchman has been appointed as a member of the London Council of the Confederation of British Industry. Professor Latchman was also appointed as Higher Education Observer on the Board of the London Development Agency.

Professor Catherine Law received the Wilfrid Harding Prize, a Faculty of Public Health Award, awarded biennially for outstanding effort in promoting the aims of the Faculty.

Dr Sandy Lee was awarded a PhD for the thesis: *Understanding PTPó function: dimerisation and the search for interacting proteins.*

Dr Lin Lin was awarded the Endocrinology Section Prize at the Medical Research Society meeting.

Dr Stavros Loukogeorgakis was awarded a PhD for the thesis: *Remote ischaemic preconditioning in humans.*

Dr Stephen Marks' book *The Great Ormond Street Colour Handbook of Paediatrics and Child Health,* co-edited with Professor Stephan Strobel, Dr Peter Smith, Dr Magdi El Habbal and Professor Lewis Spitz, was awarded Joint Winner in the category of new edited book (retailing at less than £75) at the Society of Authors and the Royal Society of Medicine Book Awards. This is following being highly commended in the Paediatrics category of the BMA Medical Book Competition.

Dr Liam McCarthy was awarded a PhD for the thesis: *Extracellular matrix biology in normal and abnormal bladder development.*

Dr Merrill McHoney was awarded a PhD for the thesis: *The metabolic and inflammatory response to laparoscopic surgery in infants and children.*

Dr Kathryn McMahon was awarded a PhD for the thesis: *An investigation into the role of MLL in murine haematopoiesis.*

Professor Mortimer Mishkin was awarded a UCL Honorary Doctorate in September 2007 as a tribute to his outstanding contribution to the field of neuroscience and his support of research at UCL.

Dr Halima Moncrieffe has been awarded a Rheumatology Young Researcher Travel Award worth €1,000 for her to present at the 27th European Workshop for Rheumatology Research (EWRR).

Dr Deven Patel was awarded a PhD for the thesis: *Statistical modelling of virological and immunological patterns in HIV-1 infected pregnant women in Europe.*

Dr Jugnoo Rahi was invited to join the Medical Research Council (MRC) College of Experts, to give expert opinions on new applications, as part of the refereeing process; to provide advice to the MRC on high-profile issues such as new schemes and strategic reviews; and to sit on ad hoc panels and committees.

Mr Ori Ron won the Poster Prize for *Outcomes following the 'Clip and Drop' for multifocal necrotizing enterocolitis* at the British Association of Paediatric Surgeons Annual Conference, Edinburgh.

Dr Rod Scott gave the 3rd Butler Lecture on Epilepsy at the University of Alberta, Canada.

Dr Neil Sebire was awarded a HEFCE clinical lectureship.

Dr Rukshana Shroff was awarded first prize for the best poster presentation: *A model of intact human arteries to study the pathophysiology of medial calcification in children with chronic kidney disease – clinical and laboratory correlations* at the International Paediatric Nephrology Association meeting in Budapest. She was also awarded first prize for the best oral presentation: *An in vitro model of intact human arteries to study the pathophysiology of medial calcification in children with chronic kidney disease* at the British Association for Paediatric Nephrology in York.

Dr Jane Sowden was awarded the Santen Prize for Excellence at the Pacific Ocular Regeneration Biology Conference XII in California, USA for her presentation: *Retinal stem cell therapy.*

Professor David Taylor was awarded the gold medal of the Saudi Ophthalmological Society.

Dr Rachel Thomasson was awarded a PhD for the thesis: *Visual cognition after hemispherectomy: a neuropsychological study.*

Dr Jeni Tregay was awarded a PhD for the thesis: *The structure and cognitive underpinnings of rigid and repetitive behaviour: insights from autism spectrum disorder, Prader-Willi syndrome and typical development.*

Dr Joanna Tully was awarded a PhD for the thesis: *Risk and protective factors for the development of meningococcal disease in adolescence: a biophysical investigation.*

Dr Mieke Van Haelst was awarded a PhD for the thesis: *Clinical and molecular genetics of Fraser syndrome.*

Professor Faraneh Vargha-Khadem was invited to deliver the Birch Lecture at the Portland meeting of the International Neuropsychological Society. She also gave plenary lectures at the Center for the Brain Basis of Cognition (CBBC) in Washington, the National Institute of Mental Health in Bethesda, Washington, the EMBL-EBI on Biology and Language and Endel Tulving's Festschrift held in Tallinn, Estonia.

Dr Russell Viner was awarded a HEFCE clinical lectureship.

Dr Lucy Wedderburn became a Fellow of the Royal College of Physicians (FRCP).

Dr Lisa Willats was awarded a PhD for the thesis: *Improved quantification of perfusion in patients with cerebrovascular disease.*

Dr Marcelo Zamparelli was awarded a PhD for the thesis: *The metabolic and thermogenic response to anaesthesia and surgery in neonates.*

Dr Augusto Zani was awarded the prize for Best Research Paper Presented as an Oral Communication for the paper entitled: *Captopril reduces the severity of bowel damage in a neonatal rat model of necrotizing enterocolitis* at the 38th Congress of Italian Society of Paediatric Surgery, Florence, Italy.

Dr Matthias Zilbauer was awarded a PhD for the thesis: *Innate immune defence to Campylobacter jejuni.*

Grants and donations 2007

We are grateful to the organisations listed below for their generosity. The research at UCL Institute of Child Health (ICH) and Great Ormond Street Hospital also depends on the ongoing support of a number of organisations acknowledged at the end of this list.

Actelion Pharmaceuticals
Dr A Vellodi, Professor R A H Surtees and Ms E Davies received funding for the project: *Quantifying neurology in lysosomal storage disorders (LSD).*

Action Medical Research
Dr L Wedderburn for an investigation into the basic pathogenic mechanism of juvenile dermatomyositis (JDM).

Anatomical Society of Great Britain and Ireland
Dr A Burns for an investigation of the embryological origin and development of intrinsic ganglia within the mammalian lung.

Arthritis Research Campaign
Dr P Brogan received a Fellowship award for Dr D Efetheriou for the project: *Inflammation and endothelial injury and repair in childhood central nervous system vasculitis: identifying biomarkers predictive of disease progression.*

Dr P Brogan, Professor P Woo, Professor N Klein, Dr V Shah and Dr J Halcox received funding for the project: *Endothelial injury and repair in vasculitis of the young.*

Dr L Wedderburn, Dr K Nistala and Professor P Woo to investigate the role of TH17 cells in JIA: a new pathogenic mechanism in autoimmune arthritis.

Astella Pharma GmbH
Dr R S Trompeter received additional funding for the study: *Sub-study immunology and sub-study pharmacology.*

Astra Zeneca (UK) Ltd
Dr C Brain and Ms E Davies for an open-label, non-comparative trial to evaluate the safety, efficacy and pharmacokinetics of FASLODEX (fulvestrant) in girls with progressive precocious puberty associated with McCune-Albright syndrome.

Autism Speaks
Professor T Charman for the further development of the Co-operation of Safety in Medicines (COSMIC) as a measure of the generalisation of treatment efficacy in the PACT study.

BDF Newlife
Dr L Chitty, Dr J Waters and Dr A Wade received funding for the project: *Multiplex ligation-dependent probe amplification (MLPA): a reliable and cost efficient 'single-test' alternative for fetal chromosome analysis.*

Dr A C Offiah, Dr L Wilson, Professor R C Hennekam and Professor C Hall to digitise the radiographic collection of Professor Robert Gorlin for incorporation into the second edition of REAMS.

Big Lottery Fund
Professor A Costello to improve maternal, newborn and child health in low-income countries.

Biotechnology and Biological Sciences Research Council (BBSRC)
Professor R Callard received funding for the project: *Applied statistical and mathematical modelling of peripheral T-cell homeostasis.*

Professor P J Scambler received funding for the project: *Generation and analysis of a conditional mutation of the HIRA Gene.*

Biotechnology and Biological Sciences Research Council (BBSRC)/ Organon International
Professor M Koltzenburg received funding for the project: *Keeping the right balance: Ion channels regulating the membrane potential of sensory neurons.*

Bone Cancer Research Trust
Dr J Anderson for the development of clinical protocols for Ewing sarcoma treatment using gene transfer of T-cell receptors.

British Council
Professor A S Woolf and Miss C Gannon to investigate the roles of teashirt (tsh) transcription factors in the developing and injured/regenerating mouse renal system.

British Heart Foundation
Dr A Cook received funding for the project: *New horizons in cardiac morphology, national education, research and E-Learning Centre at the Institute of Child Health, UCL.*

Professor J Deanfield received funding for the project: *Genetic and environmental determinants of arterial function in childhood: insight into causal pathways from the Avon Longitudinal Study of Parents and Children (ALSPAC).*

Dr L Rees and Professor J Deanfield received funding for the project: *An in-vitro model of intact human arteries to study the mechanisms of vascular calcification in chronic kidney disease: clinical and laboratory correlation.*

Dr P Riley received a studentship for Mr A Rossdeutch to investigate the role of thymosin beta4 during coronary vessel development and neovascularisation.

Professor P J Scambler to study the role of CHARGE syndrome gene CHD7 in cardiovascular morphogenesis and its interaction with the DiGeorge Syndrome gene Tbx1.

British Heart Foundation via St George's Hospital Medical School
Professor J Deanfield received funding for the project: *Early markers of vascular disease in British children of South Asian, African-Carribean and white European origin.*

British Heart Foundation via University of Oxford
Professor A Lucas and Mrs E Sutton received additional funding for the project: *Endothelial phenotype determined by genetic variation and environment in early life: impact on cardiovascular disease development in adulthood.*

Cancer Research UK
Dr R Dommett, Dr J Chisholm, Dr M Bajaj-Elliott and Professor N Klein to investigate the role of innate immunity in febrile neutropenic patients.

Cathal Hayes Research Foundation
Dr L Wedderburn received funding for the project: *The JDM National Registry and Repository of UK and Ireland.*

Charles Hawkin Fund for Handicapped Children
Professor T Charman received funding for the project: *Does the use of screening instruments for autism improve the accuracy of referrals to specialist paediatric services?*

Child Growth Foundation
Professor M T Dattani, Dr A P Salt, Dr N J Dale and Professor P C Hindmarsh for an investigation into the role of growth hormone on higher functioning in children.

Child Health Research Appeal Trust (CHRAT)
Dr S Burns received funding for a studentship to investigate dendritic cell function in severe combined immunodeficiency.

Dr C Clark received funding for a studentship to map white matter tracts in children using MRI: applications in neurosurgical planning.

Dr P Ferretti received funding for a studentship to investigate the role of neural stem cells, neurogenesis and doublecortin in maintaining regenerative capability in the developing spinal cord.

Dr D Osrin received funding for a studentship for a randomised controlled trial of the effect of community groups on maternal and infant nutrition.

Professor P J Scambler received funding for a studentship to examine Tbx1 and its role in stem cells/progenitor cells of the heart.

Dr P Stanier received funding for a studentship to investigate the role of the T-box transcription factor TBX22 in craniofacial development.

Children's Hyperinsulinism Fund
Dr K H Hussain to investigate the genetics of hypoglycaemia.

Children's Research Fund
Professor A Pierro to investigate the effect of glutamine on liver metabolism during sepsis in infants and children.

Professor A Pierro received funding for the project: *Therapeutic controlled hypothermia in the treatment of neonates with severe necrotising enterocolitis.*

CLIC Sargent
Dr F Gibson received funding for the project: *Cancer in young people: a narrative study to explore their experiences during the diagnostic phase.*

Coeliac UK via University of Southampton
Professor M Londei and Dr K Lindley received funding for the project: *Unravelling innate adaptive immune responses to gluten in coeliac disease: implications for new therapies.*

CURE via Children's Memorial Hospital, Chicago
Professor J H Cross for a registry of patients with regard to their response to medication (children with epilepsy presenting under two years of age).

Cystinosis Foundation Ireland
Dr W Van't Hoff and Ms S Collin for a national registry of cystinosis patients.

Cystinosis Research Network, Inc.
Dr K Nischal, Dr R K Sekhri, Dr W Van't Hoff, Professor T J Cole and Mr N Geddes for the development of a cysteamine in situ gelling system for the topical treatment of corneal crystals in cystinosis.

Deafness Research
Dr M A K Bitner-Glindzicz, Professor L Luxon, Dr M Cohen, Dr D A Thompson and Miss I M Russell-Eggitt received further funding for the project: *The National Collaborative Usher Study.*

Department of Health
Professor C Dezateux received additional funding for a UK collaborative study of newborn screening for medium chain acyl CoA dehydrogenase deficiency (UKCSNS – MCADD).

Professor C M Law to bridge the gap between research and policy for public health and for children's health.

Professor B Gaspar, Dr W Qasim, Professor A Thrasher and Dr P Veys received funding for a phase I/II clinical trial of T-cell suicide gene therapy following allogenetic haematopoietic stem cell transplantation.

Department of Health via Health Protection Agency (HPA)
Professor D Goldblatt received additional funding for the project: *The National Vaccine Evaluation Consortium Work Programme 2005–2010.*

Department of Health via York University
Professor C M Law for the Public Health Research Consortium Management Group.

Department for Innovation, Universities and Skills
Dr B Gao received a UK–China Fellowship for Excellence.

Dr T Hesketh to study the impact of high sex ratios in urban and rural China.

Diabetes UK via International Insulin Foundation
Mr D Beran to improve diabetes care in Mozambique.

Economic and Social Research Council (ESRC)
Dr T Baldeweg received a Fellowship award for Dr L F Halliday for the project: *Are auditory processing deficits linked to literacy problems? A comparison of specific reading disability and mild to moderate hearing loss.*

Professor P Hobson received a Fellowship award for Mr D Williams for the project: *Novel approaches to the study of self-awareness among persons with autism, with special reference to inner speech.*

Economic and Social Research Council (ESRC) via Institute for Fiscal Studies
Dr D Osrin and Professor A Tomkins for human development and poverty reduction in developing countries.

Epilepsy Research Foundation
Dr R C Scott, Dr M Lythgoe and Professor D G Gadian to investigate the role of inflammation in brain injury and epileptogenesis following status epilepticus.

EU Commission Marie Curie Fellowship
Dr G Bouma and Professor A Thrasher received funding for the project: *Trafficking of DC in Wiskott Aldrich syndrome (WASpTrafficDC).*

European Commission (EC)
Professor D Skuse received funding for the project: *Towards the genetic basis of co-operation.*

Dr A W Stoker received funding for the project: *Protein tyrosine phosphatases: structure, regulation and biological function.*

European Society for Immunodeficiencies (ESID)
Dr G Davies, Professor B Gaspar, Dr A M Jones, Dr C M Cale and Professor A Thrasher for a UK database for severe primary immunodeficiency disorders in children to include internet transfer of data to the European Society for Immunodeficiencies (ESID) patient database.

European Society for Paediatric Endocrinology
Professor M Dattani to investigate the role of SOX2 and SOX3 in disorders of eye, forebrain and pituitary development in humans.

European Union (EU)
Dr P J Amrolia, Professor A Thrasher, Dr N Goulden and Dr P Veys received funding for the project: *Chimaeric T-cells for the treatment of paediatric leukaemia/lymphoma.*

Professor D Skuse received a programme grant for the project: *Integrating co-operation research across Europe.*

Fight For Sight
Dr J Sowden for the development of retinal stem cell therapy: Isolation of rod precursors for photoreceptor regeneration.

Fondation Genevoise de Bienfaisance Valeria Rossi di Montelera
Professor A Pierro, Dr S J Eaton, Dr A Burns, Dr N Greene, Dr N Thapar, Dr K Mills and Dr L Cordischi received funding for the project: *Amniotic fluid and intestinal dysfunction in fetal gastroschisis.*

Professor A Pierro, Dr S J Eaton, Dr J Brierley and Dr C Barbara received funding for the project: *Energy expenditure and capillary leak in children receiving surgery or intensive care.*

Genex Biosystems
Dr S Hart to research tumour-targeted nanoparticles for delivery of molecular therapies.

Genzyme
Dr E Pope-Davies received funding for the purchase of equipment for the project: *Develop a severity scoring tool for neuronopathic Gaucher disease.*

Great Ormond Street Hospital/ICH Science Development Initiative

Dr A J Burns and Dr N Thapar for an investigation of the role of RET as a dependence receptor during enteric nervous system development.

Dr S Keating for a molecular analysis of the immune response to vaccines and infectious diseases: optimisation of antigen-specific B cells.

Dr M Bajaj-Elliott and Dr K J Lindley received funding for the project: *Dysregulation of IL-23/IL-17 axis and FoxP3+ T-cell regulatory function by microbial-innate activation pathways contributes to enhanced autoimmunity in ulcerative colitis.*

Dr H Baxendale, Dr K Gustafsson and Dr S Hart to study DNA vaccine encoding targets for natural antibody-mediated immune complex formation.

Dr D Bockenhauer, Dr W Van't Hoff and Dr K L Price for an investigation of glomerular defects in the hyperoxaluric knock-out mouse.

Dr L S Chitty, Dr W Heywood and Dr K Mills received funding for the project: *Biomarkers in maternal plasma – potential for non-invasive prenatal diagnosis of trisomy 21.*

Dr G Davies, Professor A J Thrasher, Dr K Parsley, Dr K Gilmour and Ms L D Henderson to study cultured thymic epithelial transplantation for complete DiGeorge syndrome.

Dr W L Di, Dr W Qasim, Professor A J Thrasher, Professor J Harper and Professor R E Callard received funding for the project: *Lentiviral gene therapy for Netherton syndrome.*

Dr K C Gilmour, Dr C M Cale, Professor B Gaspar, Dr R Wheeler, Dr S O Burns and Dr A Vellodi for the development of diagnostic tests for primary immune deficiency.

Dr T Jacques, Dr L Harding, Professor J H Cross and Dr W Harkness for an investigation of the pathological mechanisms in childhood epilepsy.

Dr J C K Wells, Dr R M Viner, Dr D Haroun and Professor T J Cole received funding for the project: *3D Body scanning and cardiovascular risk factors in multi-ethnic adolescents.*

Great Ormond Street Hospital Children's Charity

Dr S Amitay and Professor F Vargha-Khadem for the purchase of diagnostic audiometer, calibration equipment and software for the investigation of non-verbal auditory long-term recognition memory in normal adults and adults with developmental amnesia.

Dr M Bajaj-Elliott and Dr K J Lindley for the purchase of an Ussing Chamber to study how gut from healthy children interacts with bacteria compared with those that become susceptible to diarrhoea and inflammation.

Dr T Baldeweg for the purchase of brain functioning equipment upgrade for functional Magnetic Resonance Imaging (fMRI).

Dr H Baxendale for the purchase of an ELISA Plate Reader and an ELISA Plate Washer.

Dr H Brady received studentship funding for the project: *Molecular pathways in infant and childhood leukaemia.*

Dr H Brady and Professor M Monk received funding for the project: *Aberrant imprinted gene expression as a marker for acute childhood leukaemia.*

Professor D Goldblatt, Professor A Thrasher and Ms K T Lant received funding for the project: *Centralised chronic granulomatous disorder (CGD) clinical nurse specialist funding.*

Professor C Kinnon for a Great Ormond Street Hospital/ICH cell analysis and sorting facility, from Miss A E Clarke.

Professor C Kinnon and Dr H Brady for a FACS Canto 2 Analyser in the Great Ormond Street Hospital/ICH cell analysis and sorting facility, from Rays of Sunshine and Investec.

Professor C Kinnon and Dr H Brady for a Great Ormond Street Hospital/ICH cell analysis and sorting facility, from D B Hussey.

Professor C Kinnon and Dr H Brady for a Great Ormond Street Hospital/ICH cell analysis and sorting facility, from the Jane Hodge Foundation.

Dr M Fewtrell for the purchase of a Peripheral Quantitative Computed Tomography (pQCT) machine.

Dr N Greene, Professor A Copp, Dr K Mills, Dr L Chitty, Professor P Clayton, Professor J Harper, Dr P Ferretti, Professor M Koltzenburg and Dr A Stoker for the purchase of 2D Protein Gel Analysis Software for analysis of the proteins contained in a tissue sample, from a bequest in a will made by Cynthia Rose Gwilliam in memory of her late son, Mark Gwilliam.

Dr K Hussain and Professor P Beales received a legacy for homozygosity mapping to identify potential candidate genes leading to abnormalities in pancreatic development and diabetes mellitus.

Dr D Long and Dr P Winyard for the purchase of a PCR-G Storm GS4 Thermal Cyler Base Unit, Benchmark Plus Spectrophotometer System.

Dr S Lum for the purchase of four Masimo oxygen saturation monitors with 40 free sensors, six Fleisch flowmeters for respiratory physiology: diffusing capacity equipment.

Dr E Main for the purchase of a Novel pliance®-x-32/E system.

Dr M Munoz-Lopez, Professor F Vargha-Khadem, Dr B N Harding, Dr T Jacques and Professor D Gadian received equipment funding to purchase an eye tracking system suitable for fMRI experiments.

Dr C Owens towards the cost of a Dual Source Multi-Detector CT (MDCT) Scanner.

Dr D Roebuck for the purchase of a LightLab M2 Optical Coherence Tomography and four LightLab Imagewire Probes.

Dr A Sala and Dr J Anderson received a legacy for: *Research into cancer and arthritis in children of Great Ormond Street Hospital.*

Mrs H Shannon and Dr E Main for the purchase of a Trucorp 'Truman' Manikin for Cardiopulmonary Management System.

Dr A Stoker, Dr P Ferretti, Dr J Sowden, Dr S Burns, Dr A Burns, Dr T Jacques, Dr N Thapar, Dr P Winyard, Professor P J Scambler for the purchase of Linear Encoded Microscope Stage with Joystick and Stage Inserts, Exfo Light Source, Firewire black and white cooled CCD Camera, Volicity Acquisition software, Mac Pro computer and monitor.

Dr P Veys, Dr N Goulden, Dr P J Amrolia, Dr K Rao and Dr L Biassoni received a legacy for: *Targeted radiotherapy in conjunction with stem cell transplantation for refractory acute myeloid leukaemia in children.*

Guide Dogs for the Blind Association

Dr J Rahi, Mr D S I Taylor, Professor A Moore, Mrs P Cumberland, Dr A P Salt, Dr N J Dale, Professor G Hundt (Warwick) and Professor P T Khaw (Moorfields/Institute of Ophthalmology) for an investigation of the quality of life of visually impaired children – investigation and development of a novel vision-related quality of life instrument.

Guy's and St Thomas' via Aspreva Pharmaceutical

Professor J Deanfield and Ms A E Donald for a prospective, randomised, double-blind, placebo-controlled trial evaluating the effects of mycophenolate mofetil (MMF) on 'surrogate markers' for atherosclerosis in female patients with systemic lupus erythematosus.

Health Protection Agency (HPA)

Professor C Dezateux received funding for the project: *Measuring environmental exposures in childhood using shed milk teeth (contribution of environmental lead exposure in early childhood in inequalities in child health and cognitive development in the UK Millennium Cohort Study).*

Professor D Goldblatt for a co-ordinated programme of research designed to inform key policy decisions relevant to the future use of vaccines in the UK.

Health Technology Assessment (HTA) via Royal Liverpool Children's NHS Trust

Dr A Sutcliffe and Dr P J Santosh received funding for the project: *The use of melatonin in children with neuro-developmental disorders and impaired sleep; a randomised, double-blind, placebo-controlled, parallel study.*

Health Technology Assessment (HTA) via University of Exeter

Dr A Vellodi for a proposal to conduct a long-term cohort study of people with lysosomal storage disorders.

Hestia Foundation and the Ruth Lilly Philanthropic Foundation

Professor A Tomkins and Dr S Filteau for the support of community groups for counselling of HIV infected mothers in the Infant Feeding and HIV Project in Lusaka, Zambia.

Horst Bickel-Stiftung

Professor P T Clayton, Professor J H Cross, Professor R A H Surtees, Dr P B Mills and Dr P Lister received funding for the project: *Pyridoxal phosphate (PLP) and epilepsy.*

Hyperinsulinism Parent Support Group

Dr K H Hussain and Ms C Gilbert to investigate the genetics of hypoglycaemia.

ID Greening Budget

Professor N Klein, Dr A Prendergast and Ms M Clapson to investigate the role of the cellular immune system in control of HIV in young people.

Institute of Biomedical Science

Dr T Jacques and Ms K Miller to investigate a role for glial signalling in the regulation of neural stem cell proliferation.

Institute of Education

Professor C Dezateux received funding for the project: *Measuring fat-free mass and physical activity patterns in the UK Millennium Cohort.*

International Insulin Foundation via Diabetes UK

Mr D Beran to improve diabetes care in Mozambique.

Inveresk/The Antiretroviral Pregnancy Registry

Professor M L Newell, Dr C Thorne and Professor C Peckham for a European collaborative study on children born to HIV infected mothers (ECS-HIV).

Ipsen Ltd

Professor M T Dattani received funding for the project: *International Co-operative Growth Study (INCGS).*

ISIS Pharmaceuticals

Professor J Deanfield, Dr P J Lee, Miss L Redmond and Dr J Halcox for a randomised, double-blind, placebo-controlled study to assess the safety and efficacy of isis 301012 as add-on therapy in homozygous familial hypercholesterolaemia subjects.

Johns Hopkins University
Professor D Goldblatt received funding for the project: *Immunity to prevent pneumococcal transmission: correlates of protection and herd immunity.*

Kelloggs Co.
Miss J Lanigan, Dr A Singhal and Dr M Lawson received funding for the study: *Dietary long chain n-3 fatty acids: development of a dietary assessment tool.*

Kids Company via Trustee Man Group PLC Charitable Trust
Professor F Vargha-Khadem, Professor D G Gadian, Professor R A H Surtees, Dr K H Hussain, Dr P J Santosh, Dr W K Chong and Dr Q Mok received funding for the project: *Neural and physiological markers of antisocial behaviour in adolescents.*

Kids Kidney Research
Dr L Rees, Dr D K Hothi and Dr J Marek received funding for the study: *Myocardial stunning during paediatric dialysis and the effects of cooling the dialysate haemodialysis.*

Professor A Woolf, Professor P J Scambler and Dr J Pitera to examine roles for Tbx1 in normal and abnormal renal tract morphogenesis.

Leukaemia and Lymphoma Society
Dr A Sala received funding for the workshop: *MYB proteins in death, differentiation and disease.*

Leukaemia Research Fund
Professor T Crompton to investigate the function of the GLi family of transcription factors in T-cell development.

Professor C Kinnon to investigate the effect of bone morphogenetic protein 4 on haematopoietic differentiation of murine embryonic stem cells.

Dr W Qasim and Professor B Gaspar for strategies for the control and treatment of viral infections following bone marrow transplantation using interferon gamma secretion assays.

Dr W Qasim, Professor A Thrasher, Professor B Gaspar, Professor C Kinnon and Dr P Veys for the development of cell therapies to improve haematopoietic cell transplantation.

Dr O Williams and Dr H Brady to generate an in vivo model for TEL-AML1-induced acute lymphoblastic leukaemia using disease-associated secondary mutations.

Macula Vision Research Foundation
Dr J Sowden received funding for the project: *Towards retinal stem cell therapy: transplanting post-mitotic cone and rod photoreceptor precursors for retinal repair.*

Mason Medical Research Foundation
Dr G Davies, Professor A Thrasher, Dr K Parsley, Dr K Gilmour, Ms L D Henderson to study cultured thymic ephithelial transplantation for complete DiGeorge syndrome.

Medical Research Council MRC
Dr P J Amrolia, Dr P Veys and Dr S Samarasinghe received funding for the project: *Selective depletion of donor allo-reactive t-cells to improve anti-viral and anti-leukaemic responses post haplo-identical stem cell transplant.*

Dr J Cohen for a detailed evaluation of the immunogenicity and protective efficacy of a peptide mimic of pneumococcal capsular polysaccharide Type 6B – a novel vaccine candidate antigen.

Dr M Fewtrell and Professor A Lucas received funding for the project: *Early growth and later bone health: a project based on two randomised intervention trials.*

Professor B Gaspar and Professor A Thrasher for the development of an enhanced lentiviral vector for gene therapy of ADA-SCID.

Dr N Greene and Professor A Copp received funding for the project: *Inositol-preventable neural tube defects: understanding the molecular causes and mechanisms of prevention.*

Dr E Hyppönen and Professor C Power received funding for the project: *Vitamin D and health: genome–wide analysis and insights from mendelian randomisation.*

Dr L Kerecuk, Professor A Woolf and Professor P J Scambler to explore the expression and potential roles of Fras1 and Frem2 in models of kidney diseases affecting the collecting duct lineage.

Dr L Li received funding for the project: *Statistical approaches for life course studies: developmental trajectories and adult health.*

Dr A Singhal and Professor A Lucas received funding for the project: *Nutritional interventions in childhood and cardiovascular disease risk.*

Medical Research Council Capacity Building Area Studentship
Dr M Cortina-Borja and Dr A M Wade received funding for the project: *Models for discrete epidemiological and clinical data.*

Medical Research Council via Newcastle University
Professor A Woolf received funding for the project: *SNP based sib-pair linked study to identify loci contributing to vesicoureteric reflux.*

Medical Research Council via University of Nottingham
Professor J Stocks, Dr S-Y Lum, Dr J Kirkby, Dr S Sonnappa, Dr C Bastardo, Dr C Oliver and Mr L Welsh received further funding for the project: *Extremely preterm infants – population-based studies of survival and health status of infants born at less than 26 completed weeks of gestation age.*

Medtronic
Professor P Bonhoeffer received funding for the study: *Right ventricular outflow tract reconstruction from magnetic resonance data.*

MEND Central
Mr P Sacher for a clinical trial of the MEND programme to improve health outcomes in obese children.

Meningitis Trust
Dr R Viner, Dr D Christie, Dr H Bedford, Dr R Booy and Dr H El-Bashir for the project: *Meningococcal outcomes in children and adolescent (MOCA) study.*

Meningitis UK
Dr R Viner, Dr H El-Bashir and Dr H Rashid for a literature review on outcomes of meningitis.

Merck
Professor J Deanfield for a worldwide, double-blind, randomised, placebo-controlled study of MK-0524A 2g co-administered with an intensive LDL-C lowering therapy compared with intensive LDL-C lowering therapy alone on carotid artery intima-media thickness (cIMT) in patients with HeFH.

Dr W Van't Hoff for a randomised double, parallel, placebo or amiodipine controlled study of the effects of losartan on proteinuria in paediatric patients with or without hypertension.

Milena Carvajal – Prokartagener Foundation
Dr H Mitchison for an NP-based genome-wide linkage scan to identify new genes causing Primary Ciliary Dyskinesia (PCD).

Muscular Dystrophy Campaign
Professor F Muntoni received a combined Dubowitz and IoN Neuromuscular Centres grant.

Myositis Support Group
Dr L Wedderburn, Professor P Woo and Dr C Pilkington received funding for the project: *The JDM National Registry and Repository of UK and Ireland.*

National Institute of Mental Health (NIMH)
Professor F Vargha-Khadem and Professor M Mishkin (NIHM) to investigate interactions between cognitive-based versus habit-based memory for speech and language – comparisons of the hippocampal versus neostriatal/prefrontal systems.

North Bristol NHS Fund
Dr S L Hart received funding for the project: *Novel therapies for glioblastoma using receptor-targeted nanoparticles.*

Novartis
Professor D Goldblatt to measure pneumococcal antibodies.

Professor P Woo for a multi-centre open label, repeated dose range-finding study to evaluate the safety, tolerability, immunogenicity, pharmacokinetics and efficacy of an anti-iL-beta monoclonal antibody (ACZ885) given subcutaneously in paediatric subjects with sJIA.

The Nuffield Foundation
Dr P Brogan and Miss L Clarke received funding for undergraduate student Ms A Standing for the project: *Endothelial injury and repair in vasculitis of the young.*

Olivia Hodson Cancer Fund
Dr H Brady received funding for the project: *Elucidating the biochemical mechanisms underlying senescence-based chemotherapy for cancer.*

Dr F Gibson, Ms A Barry, Ms J Bayliss, Mrs A Conley and Mrs V Wigg received funding for the project: *What it's like when you find eating difficult: children and parents' experience of food intake.*

Options Consultancy Services
Professor A Costello and Dr J Borghi to evaluate the cost-sharing scheme for delivery care in Nepal.

Orphan Europe
Dr W Van't Hoff for a national registry of cystinosis patients.

Paediatric Rheumatology Discretionary Fund
Dr L Wedderburn received funding for the project: *The JDM National Registry and Repository of UK and Ireland.*

Pfizer Consumer Health Products
Professor L Franck received funding for the study: *Translating the tears: what words do young children use to tell their parents they have pain or fever?*

Research into Childhood Cancer
Dr A Michalski received a donation from Mr and Mrs Sacarello.

Rho Inc.
Dr R Liesner and Ms K Khair received funding for the study: *Haemophilia Inhibitor Genetics Study (HIGS).*

Royal College of Surgeons of England
Dr C Clark, Dr W Harkness, Dr R Hayward and Dr W Chong for the development of Magnetic Resonance Tractography for Paediatric Neurosurgical Operating Planning.

Mr N Alexander received a Fellowship Award for the project: *Regulation of monocyte MHC Class II– understanding post-operative immunoparalysis.*

Royal Society Wolfson Foundation Laboratory Refurbishment Grants Scheme
Professor A Pierro, Dr N Thapar, Dr P De Coppi, Dr S J Eaton and Dr A Burns received funding for the project: *Gut tissue engineering.*

Samantha Dickinson Research Trust via Institute of Neurology (IoN)
Dr T Jacques received funding for the project: *Wnt signalling in neural stem cell differentiation and tumourigenesis of the CNS.*

Sanofi Pasteur
Professor D Goldblatt for the testing of human serum samples for antibody titres against Streptococcus pneumoniae serotypes.

Professor D Goldblatt received funding for the project: *Contract serology.*

Save the Children US
Professor A Costello, Professor M L Newell and Dr D Osrin to improve essential maternal and newborn care in poor rural communities in Malawi.

School of Medical Sciences, Bristol
Professor D Goldblatt received funding for the project: *Contract serology.*

Shire Human Genetics Therapies AB
Dr A Vellodi and Dr M Clearly for an international, multi-centre, long-term observational study of patients with Hunter syndrome (Mucopolysaccharidosis II).

Simons Foundation via Spring Harbour Laboratories
Professor D Skuse received funding for the project: *Neurocognitive and genetic investigations of families with an autistic child: quantifying the broader phenotype.*

Society for Paediatric Radiology
Dr O Olson, Dr C Owens and Dr I A Mendichovszky received funding for the project: *T1 mapping of childhood solid tumours.*

Sport Aiding Medical Research for Kids (SPARKS)
Dr M Bitner-Glindzicz, Dr S Rahman and Professor M Pembrey received funding for the project: *Preventable hearing loss: what is the prevalence and penetrance of the mitochondrial m.1555A>G mutation associated with aminoglycoside-induced hearing loss?*

Professor G E Moore, Dr L M Chitty and Mr D Peebles received funding for the project: *Can imprinted genes act as diagnostic markers for intrauterine growth restriction during pregnancy?*

Professor A Pierro, Dr S J Eaton and Professor N Klein received funding for the project: *Microbial invasion during parenteral nutrition in surgical infants receiving glutamine.*

Dr A Sala received funding for the project: *Green tea catechins and red pepper capsaicin: safe, natural compounds as a novel treatment approach for neuroblastoma.*

Dr L Wedderburn for the characterisation of the biological mechanisms of and psychological responses to success or failure of drug treatment in childhood arthritis.

Dr L Wedderburn, Ms P Livermore, Dr M Fife and Professor P Woo received an award extension for the project: *Characterisation of the biological mechanisms of and psychological responses to success or failure of drug treatment in childhood arthritis.*

Teenage Cancer Trust
Dr F Gibson to evaluate the advanced symptom management system (ASyMS) to monitor and manage chemotherapy-related toxicity: *The AsyMS Study.*

Tibotec Pharmaceuticals
Dr V Novelli for a phase I, open-label trial to investigate pharmacokinetics, safety and tolerability of TMC125 at steady-state in treatment-experienced HIV-1 infected children.

UCB Pharma
Professor J H Cross, Dr C Eltze, Dr M de Haan and Dr R C Scott received an unrestricted educational grant for the project: *Epilepsy in infancy: spectrum of aetiologies, natural history and outcome predictors.*

UCL Business
Dr H Baxendale to establish a proof of concept for using human natural IgM as a vaccine adjuvant in viral and bacterial vaccines and a therapeutic in pneumococcal and influenza infection.

UK Clinical Research Collaboration
Professor C Dezateux and Professor C Peckham received funding for the project: *NHS electronic records and their linkage for epidemiological research: a simulation for UK Clinical Research Collaboration and Connecting For Health.*

University of London Central Research Fund
Professor C Dezateux and Professor P J Scambler received an equipment grant for the project: *Realising the research potential of the national newborn bloodspot bank to advance science and health.*

Professor C Kinnon and Mr A Abeyewickreme to investigate the effect of bone morphogenetic protein 4 on haematopoietic differentiation of murine embryonic stem cells.

Wellcome Trust
Professor P Beales received a renewal of his fellowship to define the role of primary cilia in development and disease.

Professor A Copp received funding for the project: *Consolidation of the MRC/Wellcome Trust human developmental biology resource.*

Dr K H Hussain received funding for the project: *Defining genotype/phenotype relationships in known and novel causes of hyperinsulinaemic hypoglycaemia.*

Dr L Wedderburn and Mr P Knopp received funding to investigate satellite cell biology in juvenile dermatomyositis.

Professor A D Phillips and Dr S Schuller to investigate the interaction of enterohaemorrhagic Escherichia coli with human intestinal epithelium under polarised intestinal simulated conditions.

Professor G E Moore and Dr D Monk received funding for a vacation scholarship for Mr J Chong for the project: *Discovering new human imprinted genes in the placenta.*

Dr D Osrin was awarded a Career Development Fellowship for the project: *Cluster randomised controlled trial of the effect of community mobilisation on neonatal survival in Mumbai slums.*

Dr R C Scott and Professor D G Gadian to examine the consequences and outcomes of convulsive status epilepticus in childhood.

Dr C Thorne received funding for the project: *Pregnancy, antiretroviral therapy and HIV disease progression in women.*

Professor B Gaspar received a Fellowship award for Dr C Booth for the project: *Regulated SAP gene transfer for correction of X-linked lymphoproliferative disease.*

World Health Organization (WHO)
Professor D Goldblatt for assay standardisation and development for pneumococcal vaccine.

Dr A Seal received an award for the performance of work for WHO.

The ICH also continues to receive grants from the following organisations

Abbott Laboratories
Academy of Medical Sciences
Addenbrooke's NHS Trust
Alcon Laboratories Ltd UK
Alzeimers Research Trust
Ambu A/S
Association for International Cancer Research
Association of Anaesthetists of GB and Ireland
Association of Paediatric Anaesthetists
Asthma UK
Avent Ltd
Avidex Ltd
Baily Thomas Charitable Trust
Barts and The London Charitable Foundation
Baxter Healthcare Corporation
Bill Gates Foundation
Bioenvision
Biomarin Pharmaceutical Inc
Biophage Limited
Bio Products Laboratory
Birdseye Walls Ltd
Bliss – The Premature Baby Charity
British Educational Communications and Technology AG
British Eye Research Foundation
British Lung Foundation
British Medical Association
British Neuropathalogical Society
BUPA Foundation Medical Research Charity
Centocor Inc
Cerebra
Channel Four Television Corporation
Charite-Universitatsmedizin Berlin
CHILDREN with LEUKAEMIA
Children's Trust
Chiron Srl
Chronic Granulomatous Disease Trust
Cleft Lip and Palate Association
Colt Foundation
CORDA
Cord Blood Charity
Coronary Artery Disease Research Association
CP Charitable Trust
Crohn's in Childhood Research Appeal
CSL Behring AG
Cyberonics Inc
Cystic Fibrosis Research Trust
Department for Education and Skills
Department for International Development
Dona Estefania Hospital
Down's Syndrome Association
Dystrophic Epidermolysis Bullosa Research Association
Edward Jenner Institute
Eli Lilly & Co
Elimination of Leukaemia Fund
Engineering and Physical Sciences Research Council
Enid Linder Foundation
European Society for Paediatric Endocrinology
Fondation Milena Carvajal

Foundation for the Study of Infant Deaths
General Charitable Trust of ICH
GlaxoSmithKline
Health Foundation
Help the Aged
H.J. Heinz Company Limited
Hospital for Sick Children
HSA Charitable Trust
Human Early Learning Partnership
ICN Pharmaceuticals Ltd
Imperial College of Science, Technology and Medicine
International Association for the Study of Pain
International Centre for Child Studies
Janssen-Cilag Ltd
Kidney Research Aid Foundation
Macular Disease Society
March of Dimes Birth Defects Foundation
Marie Curie Cancer Care
Mary Kitzinger Trust
Medical Research Council of Canada
Merck, Sharp and Dohme
Moulton Charitable Trust
Muscular Dystrophy Association
Nancy Lurie Marks Family Foundation
National Academy of Education
National Alliance for Autism Research
National Eczema Society
National Institutes of Health
National Kidney Research Fund
Neuroblastoma Society
NHS Innovations London
North Central London Innovation Hub
Nutricia Ltd
Oxford Glycosciences (Vic) Limited
Paediatric Rheumatology Discretionary Fund
Parkinson's Disease Society
Pathalogical Society of Great Britain and Ireland
Pearsalls Ltd
Pharmaxis Ltd
Pharm Research Associates (UK) Ltd
PHLS Communicable Disease Surveillance Centre
Physiotherapy Research Foundation
Quintiles (UK) Ltd
Rank Bequest
Roche Products Limited
Royal College of Paediatrics
Royal College of Radiologists
Royal College of Surgeons of Edinburgh
Royal Society
Sainsbury's Supermarkets Ltd
Sankyo Pharma Development
Sanofi-Aventis
Save the Children
Search
Sense (The National Deafblind and Rubella Association)
SHS International Limited
Siemens Plc
Sir Halley Stewart Trust
Sir Siegmund Warburg's Voluntary Settlement
Smiths Medical
Society for Mucopolysaccharide Diseases
Society for Paediatric Radiology
Spencer Dayman Meningitis Laboratories
Stanford University
Stroke Association
Tanita UK Limited
Tavistock and Portman NHS Trust
Transkaryotic Therapies Inc
UBS AG
United Kingdom Children's Cancer Study Group
University of Iowa
United Nations Children's Fund
University of Southampton
US Agency for International Development
Vitol Charity Fund
Volkswagen Stiftung
Walter Swindon Charitable Trust
Wellbeing (The Health Charity for Women and Babies)
Wellchild
Welton Foundation
Wyeth Laboratories
Wyeth-Lederle Vaccines

Senior academic staff 2007

Biochemical and nutritional sciences theme

Theme Leader:
Professor Alan Lucas

Nutrition unit

MRC Professor of Paediatric Nutrition and Head of Unit
Professor Alan Lucas MA MB BChir FRCP MD FRCPCH FmedSci
Professor of Biochemistry
Professor David Muller BSc PhD
Emeritus Professor
Professor Brian Wharton MD MBA DSc FRCP(L)(E)(G) FRCPCH DCH
Reader in Childhood Nutrition
Dr Atul Singhal MB BS DCH MRCP MD
Reader in Paediatric Nutrition
Dr Jonathan Wells MA MPhil PhD
Reader in Childhood Nutrition
Dr Mary Fewtrell MD BMBCh FRCPCH MRCP DCH MA
Senior Lecturers
Dr Virgilio Carnielli PhD MD (left February 2007)
Dr Margaret Lawson MSc PhD SRD

Surgery unit

Nuffield Professor of Paediatric Surgery and Head of Unit
Professor Agostino Pierro MD FRCS (Eng) FRCS (Ed)
Senior Lecturer
Dr Simon Eaton BSc PhD
Honorary Senior Lecturers
Mr David Albert FRCS
Mr Peter Ayliffe FRCS (Eng) FRCS (Maxfac) FDS RCS (Eng)
Mr Martin Bailey BSc FRCS
Mrs Mary Calvert BDS FDSRCS (Ed) MOrth MSc
Mr Joe Curry MBBS FRCS (Eng) FRCS (Paed Surg)
Mr David Drake MB BChir FRCS FRCPCH
Mr Robert Evans BSc BDS MScD FDSRCS (Eng) DOrth MOrth RCS (Ed)
Mr Ben Hartley BSc MB BS FRCS (ORL-HNS)
Mr Robert Hill RCS
Dr Susan Hill BM MRCP DCH
Mr Barry Jones MS FRCS
Mr David Jones FRCS FRCS Ed (Orth)
Mr Loshan Kangesu BSc MBBS FRCS MS FRCS (Plast)
Mr Edward Kiely FRCSI FRCS
Dr Michael Mars PhD BDS FDS DOrth
Mr Fergal Monsell MSc FRCS FRCS (Orth)
Mr Hilali Noorden MA FRCS (Eng) FRCS (Orth)
Dr Neil Shah MB BS PhD
Mr Paul Smith FRCS
Dr Virpi Smith FIBMS PhD
Mr Brian Sommerlad FRCS
Mr Stuart Tucker FRCS

Cancer theme

Molecular haematology and cancer biology unit

Head of Unit
Vacant
Reader in Molecular Haematology and Cancer Biology
Dr Hugh Brady
Emeritus Professor of Haematology and Oncology
Professor Judith Chessells MD FRCP FRCPath
Visiting Professor of Molecular Haematology
Professor Paul Brickell BA MA PhD
Visiting Professor of Haematology and Oncology
Professor Ian Hann MD FRCP FRCPath

Developmental neurosciences and behavioural paediatrics unit

Reader in Molecular Neurobiology and Deputy Head of Unit
Dr Jonathan Ham BSc PhD
Reader In Paediatric and Developmental Pathology
Dr Neil Sebire BSc MB BS DM FRCPath
Senior Lecturers
Dr John Anderson BA MB BS MRCP PhD
Dr Michael Hubank BA PhD
Dr Arturo Sala PhD
Honorary Senior Lecturers
Dr Peppy Brock MD PhD
Dr Julia Chisholm MRCPCH MA PhD
Dr Ann Goldman MB BCh FRCP
Dr Gill Levitt BSc MRCP DCH
Dr Raina Liesner BA MB BChir MRCP
Dr Antony Michalski MB ChB MRCP
Dr David Webb MD FRCP FRCPath MRCPCH
Lecturer
Dr Owen Williams BSc PhD
Honorary Lecturer
Dr Alison Leiper MB BS MRCP

Cardiorespiratory sciences theme

Theme Leader:
Professor Michael (Monty) Mythen (until December 2007)
Professor John Deanfield (from December 2007)

Cardiac unit

The British Heart Foundation Vandervell Professor of Congenital Heart Disease and Head of Unit
Professor John Deanfield MD BChir FRCP

Professor of Cardiothoracic Surgery
Professor Marc de Leval MD FRCS (retired 2006, but still holds honorary contract)
The British Heart Foundation Joseph Levy Professor of Paediatric Cardiac Morphology
Professor Robert Anderson BSc MD FRCPath
Professor of Cardiology
Professor Philipp Bonhoeffer MD FSCAI
Professor of Cardiothoracic Surgery
Professor Martin Elliott MD FRCS
Professor of Cardiology
Professor William McKenna BA MD DSc FRCP FESC FACC
Reader in Cardiovascular Imaging
Dr Andrew Taylor BA (Hons) MD MRCP(UK) FRCR
Senior Lecturers
Dr Andrew Cook PhD (British Heart Foundation lecturer)
Dr Perry Elliott MBBS MD MRCP
Dr Julian Halcox MA MB BChir MD MRCP (Al Maktoum British Heart Foundation Senior Lecturer in Cardiology) (left October 2007)
Ms Catharina van Doorn MD FRCS (C/Th)
Honorary Senior Lecturers
Dr Kate Brown BChir MRCP (joint with Portex)
Dr Michael Burch MB ChB MD FRCP FRCPCH
Dr Allan Goldman MB BChB MRCP MSc
Dr Nick Piggott MB BS MRCPI MRCPCH
Dr Philip Rees MB BChir DRCOG FRCP
Dr Margrid Schindler MD MB BS FFICI-ANZCA (joint with Portex)
Dr Ian Sullivan MB BChir FRACP
Mr Victor Tsang MB BS MS MSc FRCS FRCS (Ed)
Dr Robert Yates BSc(Med) MB Bch

Portex anaesthesia, intensive therapy and respiratory medicine unit

Smiths Medical Professor of Anaesthesia and Critical Care and Head of Unit
Professor Michael (Monty) Mythen FRCA
Professor of Respiratory Physiology
Professor Janet Stocks PhD
Honorary Reader in Cardiovascular Genetics
Dr Hugh Montgomery BSc MB BS MRCP MD
Senior Lecturers
Dr Eleanor Main BA PhD
Dr Mark Peters MB BCh MRCP
Dr Suellen Walker MB BS MM(PM) MSc FANZA FFPMANZCA
Honorary Senior Lecturers
Dr Paul Aurora BSc MB BS MRCP MSc
Dr Robert Bingham MB BS FRCA
Dr Ann Black MB BS DRCOG FRCA
Dr David de Beer BSc MB ChB DCH FRCA
Dr Robert Dinwiddie MB ChB FRCP FRCPCH DCH
Dr Hilary Glaisyer MB BS MRCP FRCA
Dr Louise Harding MB BS FRCS
Dr Jane Herod BSc MB BS FRCA
Dr Richard Howard BSc MB ChB FRCA
Dr Elizabeth Jackson BSc MB BS MRCP FRCA
Dr Ian James MB ChB FRCA
Dr Adrian Lloyd-Thomas MB BS FRCA
Dr Angus McEwan MB ChB FRCA
Dr Quen Mok MB BS MRCP MRCPI DCH
Dr Andy Petros MB BS MSc FRCP FRCPCH
Dr Christine Pierce MD BSc BBS MRCP
Dr Nick Pigott MB BS MRCPI MRCPCH
Dr Michael Sury MB BS DA FRCA
Dr Mark Thomas BSc MBBChir FRCA
Dr Isabeau Walker BSc MBBChir FRCA
Dr Colin Wallis MB ChB FCP (Paed) MD DCH FRCP
Dr Glyn Williams MBBS FRCA MD

Centre for nursing and allied health professions research

Chair of Children's Nursing Research and Head of Unit
Professor Linda Franck PhD RN RGN RSCN FRCPCH FAAN
Senior Lecturer
Dr Faith Gibson MSc (Cancer Nursing) RSCN RGN CertEd RNT PhD
Honorary Senior Lecturer
Dr Debbie Sell SRSLT FRCSLT PhD

General and adolescent paediatrics theme

General and adolescent paediatrics unit

Professor of Child Health and Head of Unit
Professor Brent Taylor PhD MB ChB FRCP FRACP
Professor of Paediatrics
Professor R Mark Gardiner MBBCh MD FRCPCH FMedSci
Honorary Professor of Paediatric Medicines Research
Professor Ian Wong BSc MSc PhD MRPharmS ILTM (HE)
Reader in Molecular Cell Biology
Dr Sara Mole PhD
Reader in Adolescent Health
Dr Russell Viner FRCP FRCPCH FRACP PhD MBBS
Senior Lecturers
Dr Eddie Chung MBChB MRCP
Dr Hannah Mitchison PhD
Dr Alastair Sutcliffe MD MRCP MRCPCH
Lecturers
Dr Indrani Banerjee PhD
Dr Jill Ellis PhD
Dr Kate Everett PhD
Dr Christina Georgoula MRCPCH
Dr Camilla Salvestrini MD

Genes, development and disease theme

Theme Leader:
Professor Peter Scambler

Clinical and Molecular Genetics Unit

Professor of Clinical and Molecular Genetics and Head of Unit
Professor Gudrun Moore BA PhD
Professor of Paediatric Metabolic Disease and Hepatology
Professor Peter Clayton MD FRCP FRCPCH
Professor of Clinical Genetics and Dysmorphology
Professor Raoul Hennekam MD PhD
Professor of Paediatric Endocrinology
Professor Peter Hindmarsh BSc MB MD BS FRCP (joint with UCL Medicine)
Professor of Paediatric Genetics
Professor Marcus Pembrey BSc MB BS MD FRCP FRCPCH FMedSci
Professor of Child Health and Growth
Professor Michael Preece MD MSc FRCP FRCPCH
Professor of Paediatric Endocrinology
Professor Mehul Dattani MD FRCP
Professor of Biochemistry and Head of Unit
Professor Bryan Winchester MA PhD
Emeritus Professor of Molecular Genetics
Professor Susan Malcolm PhD FRCPath
Emeritus Professor of Molecular Embryology
Professor Marilyn Monk
Honorary Professor of Neonatal Paediatrics
Professor John Wyatt BSc MB BS DCH FRCP (joint with UCL Paediatrics and Child Health)
Reader and Honorary Consultant in Clinical Genetics
Dr Maria Bitner-Glindzicz BSc MB BS PhD FRCP
Reader in Genetics and Fetal Medicine
Dr Lyn Chitty BSc PhD MB BS MRCOG
Senior Lecturer and Wellcome Trust Senior Fellow in Clinical Science
Dr John Achermann MA MD MRCP MRCPCH (joint with UCL Medicine)
Senior Lecturer
Dr Elizabeth Carrey PhD
Honorary Senior Lecturers
Dr Angela Barnicoat BSc MD DRCOG FRCP
Dr Caroline Brain MB MD FRCP FRCPCH
Dr Maureen Cleary MD MRCP MB ChB
Dr Ying Foo PhD
Dr Barbara Gibbons BSc FRCPath
Dr Stephanie Grunewald MD
Dr Khalid Hussain MB ChB MSc MRCP MRCPCH
Dr Richard Jones BSc MBChB DPhil MRCPath
Dr Melissa Lees MRCP MSc MD FRACP
Dr Alison Male BSc MBBS MRCP
Mrs Gail Norbury MA MSc MRCPath
Dr Elisabeth Rosser FRCP BSc MB BS
Dr Richard Stanhope BSc FRCP FRCPCH (retired November 2007)
Dr Ashok Vellodi FRCP FRCPCH
Dr Louise Wilson BSc MB ChB FRCP
Lecturers
Dr Shamima Rahman MA MRCP MRCPCH PhD
Dr Owen Williams PhD (joint with Molecular Haematology and Cancer Biology)

Molecular medicine unit

Professor of Molecular Medicine and Head of Unit
Professor Peter Scambler BSc MB ChB FRCPath FMedSci
Professor of Medical and Molecular Genetics and Wellcome Trust Senior Research Fellow and Honorary Consultant in Clinical Genetics
Professor Philip Beales BSc MD MRCP
Reader in Molecular Cardiology
Dr Paul Riley BSc PhD

Medical molecular biology unit

Professor of Human Genetics and Head of Unit
Professor David Latchman MA PhD DSc FRCPath FRSA
Reader in Molecular and Cellular Biology
Dr Anastasis Stephanou BSc PhD
Honorary Senior Lecturer
Dr Richard Knight MD PhD
Lecturer
Dr Vishwanie Budhram-Mahadeo BSc PhD

Nephro-urology unit

Professor of Nephrology and Head of Unit
Professor Adrian Woolf MA MD FRCPCH
Emeritus Professors of Paediatric Nephrology
Professor Martin Barratt FRCP CBE
Professor Michael Dillon FRCP FRCPCH
Reader in Paediatric Nephrology
Dr Lesley Rees MD FRCP FRCPCH
Honorary Readers in Paediatric Nephrology
Dr Richard Trompeter FRCP FRCPCH
Dr William Van't Hoff BSc MD FRCP FRCPCH
Senior Lecturers
Mr Philip Ransley MA MB BChir FRCS (retired September 2007)
Dr Paul Winyard BM BCh MA PhD FRCPCH
Honorary Senior Lecturers
Dr Detlef Böckenhauer MD PhD
Mr Peter Cuckow FRCS (joint with The Middlesex Hospital)
Mr Patrick Duffy MB FRCS
Mr Geoff Koffman MBChB FRCS (joint with Guy's and St Thomas' NHS Foundation Trust)
Miss Rozanne Lord MB BS FRCS (joint with Royal Free Hampstead NHS Trust)
Dr Stephen Marks MBChB MSc MRCP(UK) DCH FRCPCH
Mr Imran Mushtaq MD FRCS
Mr John Taylor MD FRCS (joint with Guy's and St Thomas' NHS Foundation Trust)
Dr Kjell Tullus MD PhD FRCPCH
Honorary Lecturers
Ms Eileen Brennan RGN RSCN ENB 147 DMS MSc
Mr Divyesh Desai MB MChir
Dr Sarah Ledermann MRCP

Infection and immunity theme

Theme Leader:
Professor Christine Kinnon

Gastroenterology and autoimmunity unit

Professor of Autoimmunity and Head of Unit
Professor Marco Londei MD PhD
Emeritus Professor of Paediatric Gastroenterology and Nutrition
Professor Peter Milla MSc MB BS FRCP
Reader in Paediatric Gastroenterology
Dr Keith Lindley BSc PhD MRCP(UK) MRCPCH

Immunobiology unit

Professor of Vaccinology and Immunology, Director of Clinical R&D and Head of Unit
Professor David Goldblatt MB ChB PhD FRCP FRCPCH
Professor of Immunology
Professor Robin Callard BSc MSc PhD DipMath

Professor of Experimental Immunology
Professor Tessa Crompton PhD
Professor of Paediatric Dermatology
Professor John Harper MD FRCP FRCPCH
Emeritus Professor of Molecular Immunology
Professor Malcolm Turner DSc (Med) PhD FRSC FRCPath
Honorary Senior Lecturer
Dr David Atherton MA MB BChir FRCP
Lecturer
Dr Wei-Li Di MB BS PhD

Infectious diseases and microbiology unit

Professor of Infectious Disease and Immunology and Head of Unit
Professor Nigel Klein BSc MB BS MRCP PhD FRCPCH
Honorary Professors
Professor Diana Gibb MBChB (Hons) MRCP MD MSc Dip Obs FRCPCH
Professor Alan Phillips PhD FRCPCH
Senior Lecturer
Dr M Bajaj-Elliott BSc PhD
Honorary Senior Lecturers
Dr Paul Brogan BSc (Hons) MBChB (Hons) MRCPCH MSc PhD (joint with Rheumatology Unit)
Dr David Cubitt MSc PhD
Dr John Hartley BSc MB BS MSc DTM&H MRCP FRCPath
Dr Marian Malone MB BCh BAO FRCPath
Dr Karyn Moshal MBChB MRCP MRCPCH DTM&H
Dr Vas Novelli FRACP FRCP FRCPCH
Dr Delane Shingadia MBBS MPh MRCP FRCPCH
Dr James Soothill MD MB BS FRCPath
Clinician Scientist
Dr Helen Baxendale BSc MB BS PhD MRCP MRCPCH
Honorary Clinical Senior Lecturer
Dr Garth Dixon BSc MB ChB PhD MRCP MRCPath

Molecular immunology unit

Professor of Molecular Immunology and Head of Unit
Professor Christine Kinnon BSC PhD
Professor of Paediatric Immunology
Professor Bobby Gaspar BSc MB BS MRCP
Professor of Paediatrics and Immunology and Wellcome Trust Senior Fellow
Professor Adrian Thrasher MB BS PhD FRCP FMedSci
Professor of Human Molecular Genetics
Professor Robin Ali BSc PhD (joint with Institute of Ophthalmology)
Reader in Molecular Biology
Dr Kenth Gustafsson PhD
Reader in Transplantation Immunology
Dr Persis Amrolia BSC MBBS MRCP MRCPath PhD
Reader in Molecular Genetics
Dr Stephen Hart BSc MSc PhD
Reader in Stem Cell Transplantation
Dr Paul Veys MBBS FRCP FRCPath FRCPCH
Senior Lecturer
Dr Waseem Qasim BMedSci MBBS MRCP MRCPCH PhD
Honorary Senior Lecturers
Dr Cathy Cale BSc MB ChB PhD MRCP MRCPCH MRCPath
Dr Graham Davies MA FRCP FRCPCH
Dr Alison Jones MRCP PhD
Lecturers/Clinician Scientists
Dr Siobhan Burns MB BCh MRCPI PhD
Dr Nicola Philpott BSc PhD (left November 2007)

Rheumatology unit

Professor of Paediatric Rheumatology and Head of Unit
Professor Patricia Woo CBE MB BS BSc PhD FRCP FRCPCH FMedSci (Head of Unit until December 2007)
Reader in Paediatric Rheumatology and Head of Unit (Head of Unit from December 2007)
Dr Lucy Wedderburn BA PhD MB BS FRCP MRCPCH
Honorary Senior Lecturers
Dr Paul Brogan BSc (Hons) MBChB (Hons) MRCPCH MSc PhD (joint with Infectious Diseases and Microbiology)
Dr Clarissa Pilkington BSc MBBS MRCPCH
Dr Nathan Hasson MBChB FRCPCH
Lecturers
Dr Bin Gao MMed PhD

Neurosciences and mental health theme

Theme Leader:
Professor Martin Koltzenburg (until September 2007)
Professor Francesco Muntoni (from December 2007)

Audiological medicine unit

Professor of Audiological Medicine and Head of Unit
Professor Linda Luxon BSc MB BS FRCP
Honorary Senior Lecturers
Dr Ewa Raglan Med Dip (Hons) LRCP MRCS FRCS
Dr Doris-Eva Bamiou ENTspec MSc (Distinction) PhD

Behavioural and brain sciences unit

Professor of Behavioural and Brain Sciences and Head of Unit
Professor David Skuse MD FRCP FRCPsych FRCPCH
Professor of Developmental Psychopathology
Professor Peter Hobson MB BChir PhD CPsychol FRCPsych
Professor of Neurodevelopment Disorders
Professor Tony Charman MA MSc PhD CClinPsy
Honorary Senior Lecturers
Dr Danya Glaser MB BS DCH FRCPsych
Dr Jon Goldin MB BS
Dr Jill Hodges BA MSc PhD
Dr Dasha Nicholls MB BS MRCPsych
Lecturer
Dr Kate Lawrence MA PhD (left August 2007)

Developmental biology unit

Reader in Developmental Biology and Head of Unit
Dr Patrizia Ferretti PhD
Reader in Craniofacial Developmental Biology and Genetics
Dr Phil Stanier PhD (Joint with Neural Development Unit)
Senior Lecturer
Dr Jane Sowden BA PhD
Honorary Senior Lecturer
Dr Agnés Bloch-Zupan BChD MBiolMedSc Specialist Certificate PhD

Developmental cognitive neuroscience unit

Professor of Developmental Cognitive Neuroscience and Head of Unit
Professor Faraneh Vargha-Khadem MA PhD
Visiting Professor
Professor Mortimer Mishkin MA PhD
Reader in Developmental Cognitive Neuroscience
Dr Torsten Baldeweg MD
Reader in Developmental Cognitive Neuroscience
Dr Michelle de Haan PhD
Honorary Lecturers
Dr Luc Berthouze BSc Msc PhD (joined September 2007)
Dr Margaret Mayston BSc Msc PhD (joined September 2007)
Dr Peter Rankin BSc Msc DClinPsy (joined October 2007)
Lecturers
Dr Sygal Amitay BSc MSc PhD (left August 2007)
Dr Federique Liegeois BSc MSc PhD

Dubowitz neuromuscular centre

Professor of Paediatric Neurology and Head of Unit
Professor Francesco Muntoni MD FMedSci

Neural development unit

GlaxoWellcome Professor of Developmental Neurobiology, Head of Unit and Dean
Professor Andrew Copp MBBS DPhil FRCPath FMedSci
Reader in Craniofacial Developmental Biology and Genetics
Dr Phil Stanier PhD (Joint with Developmental Biology Unit)
Reader in Development Neurobiology
Dr Andrew Stoker PhD
Wellcome Trust Senior Fellow
Dr Juan Pedro Martinez-Barbera BA PhD
Senior Lecturer
Dr Alan Burns BSc PhD
Lecturer
Dr Nick Greene BA PhD
Clinician Scientists
Dr Thomas Jacques BA MA MB BChir PhD MRCP
Dr Nikhil Thapar BSc BM MRCP(UK) MRCPCH(UK) PhD

Neural plasticity unit

Professor of Clinical Neurophysiology and Head of Unit
Professor Martin Koltzenburg MD FRCP
Senior Lecturer
Dr Antoni Matilla BSc MB DSc (left February 2007)
Lecturer
Dr Martin Payne Smith BSc (Hons) PhD (left August 2007)

Neurosciences unit

Professor of Paediatric Neurology and Head of Unit (until August 2007)
Professor Robert Surtees MA BM BCh PhD FRCP
Professor of Paediatric Neurology and Head of Unit (from October 2007)
Professor Helen Cross MB ChB MRCP
Prince of Wales' Professor of Childhood Epilepsy
Professor Brian Neville FRCP FRCPCH
Professor of Paediatric Neurosurgery
Professor Richard Hayward FRCS
Professor in Paediatric Neurosciences and International Child Health
Professor Charles Newton MB ChB MD MRCP
Professor of Paediatric Neurology
Professor Fenella Kirkham MA MB BCh MRCP FRCP
Honorary Professor
Professor Sheena Reilly BappSci PhD
Visiting Professor
Professor David Taylor MD FRCP FRCPsych (Hons) FRCPCH
Senior Lecturers
Dr Vijeya Ganesan MB ChB MD MRCP MRCPCH
Dr Rod Scott MB ChB PhD MRCP MRCPCH
Honorary Senior Lecturers
Dr Sarah Aylett MB BS MRCP FRCPCH
Dr Martin Bax MB BCh DM MA FRCP (Hons) FRCPCH
Dr Sarah Benton MB ChB FRCP FRCPCH
Dr Stewart Boyd MD FRCPCH
Dr Lucinda Carr MD MBChB DCH FRCPCH
Dr Hilary Cass BSc FRCP FRCPCH MILT
Dr Carlos de Sousa MB BS BSc MD FRCP FRCPCH
Dr Catherine DeVile MA MB BS MD MRCP MPCPCH
Dr Jane Evans MD
Miss Silvia Gatscher Dr med univ
Dr Brian Harding MA DPhil BM BCh FRCPath
Mr William Harkness FRCS
Dr Isabel Heyman BSc MB BS MRCPsych PhD
Dr Helen McConachie MA MPhil PhD
Dr Paola Nicolaides MB ChB FRCPCH
Dr Matthew Pitt MD MRCP
Dr Alison Salt MSc DCH FRCAP FRCPCH
Mr David Scrutton MSc
Dr Patricia Sonksen MD FRCP DObst MRCOG
Mr Dominic Thompson MB BS BSc FRCS (SN)
Dr Steve White MA DPhil MB BChir MRCPsych FRCP
Lecturer
Dr Imogen Newsom-Davis BA DClinPsych

Visual sciences unit (until June 2007)

Professor of Paediatric Ophthalmology and Head of Unit
Professor David Taylor FRCOphth FRCPCH DSc (Med) (retired June 2007)
Honorary Professors
Professor Richard Abadi PhD
Professor Tony Moore FRCOphth
Honorary Reader
Miss Isabelle Russell-Eggitt MA FRCS FRCOphth
Principle Research Fellow
Dr Richard Clement PhD BSc
Honorary Senior Lecturers
Mr Kanwal Nischal FRCOpth
Dr Dorothy Thompson BSc PhD MBCO
Honorary Lecturer
Dr Alki Liasis PhD CPSM

Ulverscroft research group (from June 2007)

Reader in Ophthalmic Epidemiology and Director of the Ulverscroft Visual Research Group
Dr Jugnoo Rahi MSc PhD FRCOphth

Honorary Professors
Professor Richard Abadi PhD
Professor Tony Moore FRCOphth
Honorary Reader
Miss Isabelle Russell-Eggitt MA FRCS FRCOphth
Senior Lecturer
Dr Jane Sowden BA PhD
Principle Research Fellow
Dr Richard Clement PhD BSc
Honorary Senior Lecturers
Mr Kanwal Nischal FRCOpth
Dr Dorothy Thompson BSc PhD MBCO
Honorary Lecturer
Dr Alki Liasis PhD CPSM

Radiology and physics unit

Rank Professor of Biophysics and Head of Unit
Professor David Gadian DPhil FMedSci
(Chair of Biophysics)
Professor of Paediatric Radiology
Professor Christine Hall DMRD FRCR MD
(retired 2006 but finished working March 2007)
Professor of Paediatric Imaging
Professor Isky Gordon FRCR FRCP FRCPCH
Honorary Professor of Medical Physics
Professor Andrew Todd-Pokropek PhD (joint with Department of Medical Physics & Bioengineering, UCL)
Honorary Senior Lecturers
Dr Alex Barnacle BA MRCP FRCR
Dr Lorenzo Biassoni MD FEBNM
Dr Tim Cox FRCR
Dr Rose de Bruyn DMRD FRCR
Dr Marian Easty MBBS MRCP FRCR
Dr Melanie Hiorns MB BS MRCP FRCR
Dr Wui Khean 'Kling' Chong MD MRCP FRCR
Dr Keiran McHugh DCH FRCR FRCPI
Dr Amaka Offiah MRCP FRCR PhD
Dr Catherine Owens MRCP FRCR
Dr Oystein Ølsen cand.med (Norway) PhD
Dr Derek Roebuck BMedSc FRCR FRANZCR FHKAM (Radiology)
Dr Dawn Saunders MB MD FRCR
Senior Lecturers
Dr Christopher Clark PhD
Dr Mark Lythgoe PhD
Lecturers
Dr Martin King PhD
Honorary Lecturer
Dr Edward Proctor MD FRCS (retired in September 2007)

Population health sciences theme

Theme Leader:
Professor Carol Dezateux

Centre for international health and development

Professor of International Child Health and Head of Unit
Professor Anthony Costello MA MB BChir FRCP FRCPCH
Professor of Child Health and Nutrition
Professor Sally Grantham-McGregor MB BS MD DPH FRCP
Professor of Disability Studies
Professor Sheila Wirz MEdFCST PhD
Professor of International Child Health
Professor Andrew Tomkins MB BS FRCP FRCPCH FFPHM FMedSci
Senior Lecturer
Dr Therese Hesketh MFPHM MRCPCH PhD MPH DTM&H DCH
Honorary Senior Lecturers
Dr Richard Lansdown MA PhD DipPsych FBPsS Cpsychol
Dr Felicity Savage MS BM BCh FRCP
Lecturers
Dr Sarah Barnett PhD
Dr Julie Carter PhD (left October 2007)
Dr Zelee Hill PhD
Dr Audrey Prost PhD
Dr Andrew Seal PhD

Paediatric epidemiology and biostatistics unit

Professor of Epidemiology and Head of Unit
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Professor of Medical Statistics
Professor Timothy Cole BA BPhil MA PhD ScD HonFRCPCH FMedSci
Professor of Clinical Epidemiology
Professor Ruth Gilbert MSc MD MRCP
Professor of Public Health and Epidemiology
Professor Catherine Law OBE MD FRCP FRCPCH FFPH
Professor of Paediatric Epidemiology
Professor Marie-Louise Newell MSc PhD MFPHM
Professor of Epidemiology and Public Health
Professor Christine Power BA MSc PhD MFPHM
Professor of Epidemiology
Professor Catherine Peckham CBE MD FFPHM FRCP FRCPCH FRCPath FRCOG FMedSci
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Professor Clyde Herztman MD BSc MSc FRCPC
Professor Heather Joshi MA Mlitt
Professor Orly Manor BSc MSc PhD
Professor Brent Taylor MB ChB PhD FRACP FRCPCH
Reader in Ophthalmic Epidemiology and Director of the Ulverscroft Visual Research Group
Dr Jugnoo Rahi MSc PhD FRCOphth
Senior Lecturers
Dr Helen Bedford BSc MSc PhD FFPH FRCPCH
Dr Mario Cortina Borja BSc MSc PhD
Dr Patricia Tookey BA MSc PhD MFPHM
Dr Angela Wade BSc CStat MSc PhD ILTM
Honorary Senior Lecturers
Dr Catherine Bull MA MB BChir MRCP DCH
Dr David Elliman FRCPCH MFPHM
Dr Carlo Giaquinto MD
Dr Elizabeth Miller BSc MB BS MFPH FRCPath
Dr Angus Nicoll CBE MSc FRCP FFPHM FRCPCH
Dr Sandy Oliver BA PhD
Dr Nigel Rollins MB BCH MRCP DCH MD FRCPch
Dr Christopher Wren MB BCh FRCP
Lecturers
Dr Elina Hypponen MSc MPH PhD
Dr Tessa Parsons BSc PhD (until November 2007)
Dr Claire Thorne BA MSc PhD

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Professor Brent Taylor PhD MB ChB FRCP FRACP
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Professor Christine Kinnon BSC PhD
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Kianna, age 3