UCL
GREAT ORMOND STREET INSTITUTE OF CHILD HEALTH

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Great Ormond Street Hospital for Children
NHS Foundation Trust

THE NATIONAL CENTRE FOR YOUNG PEOPLE WITH EPILEPSY CHARITABLE TRUST

Paediatric Epilepsy Research and Impact Report

2023

Young Epilepsy

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Introduction

I am delighted to present our annual research report for the period July 2022 to June 2023 for the paediatric epilepsy research unit across Young Epilepsy, UCL GOS - Institute of Child Health and Great Ormond Street Hospital for Children.

Over the past year the research programme has continued to grow conducting 45 active projects, with 5 new projects initiated. These projects focused on a vast array of topics, including a randomised controlled trial of hypothermia as a treatment for newborns with Hypoxic Ischaemic Encephalopathy (HIE), a retrospective investigation of how strongly seizure generating parts of the brain are connected to other healthy parts of the brain and the development of guidelines for the diagnosis, treatment and management of PNPO deficiency. Excitingly, we have also completed 7 projects including an investigation of physical activity in childhood epilepsy, the construction of the new Epilepsy Diagnostic Suite centered around the installation and evaluation of the OPM-MEG technology and a randomised controlled trial of the ketogenic diet for treating drug resistant epilepsy in infants. Once again, the diversity of our research portfolio serves to demonstrate our commitment to encouraging work in all areas of epilepsy research. Whilst we continue to conduct research into understanding and treating epilepsy, we recognise the importance of providing outstanding support through educational, psychosocial, and service-based research. For those of you who are interested, there is a link on page seven where you can find out more detail on any of the 45 research projects summarised in this report.

Reflective of the ongoing work conducted as part of the research programme, this year has seen the publication of 57 peer-reviewed items of primary research, as well as 36 reviews and commentaries of expert opinion and 2 chapters in books. The impact of these publications has been particularly impressive with 6 publications in the top 5% of all research papers published.

In January 2023 we hosted the 13th international Paediatric Epilepsy Research Retreat, moderated by Professor Jo Wilmhurst. The Retreat is a one-of-a-kind event where early career and seasoned researchers meet to constructively share their research and forge new collaborations. For the first time in two years, we were able to return to an in-person format and the event was a huge success with 128 attendees over the two days. The schedule was packed with a range of high-quality presentations, and all attendees agreed it was a fantastic opportunity to celebrate the work conducted as part of the unit. We also hosted a range of other events over the year to further publicise our ongoing research, including a Joint Research Event with Epilepsy Research UK in May 2023. It was here that the Epilepsy Research Institute was announced. The Epilepsy Research Institute will be pivotal in developing the UK epilepsy research ecosystem and Young Epilepsy are thrilled to be a founding member.

Young Epilepsy's vision is to create a society where children and young people with epilepsy are enabled to thrive and fulfil their potential. A society in which their voices are respected, and their ambitions realised. Our research programme exists to establish successively better outcomes by driving early diagnosis and intervention in every aspect of childhood epilepsy, and I do hope you will enjoy reading this report.

Professor Helen Cross OBE
The Prince of Wales's Chair of Childhood Epilepsy

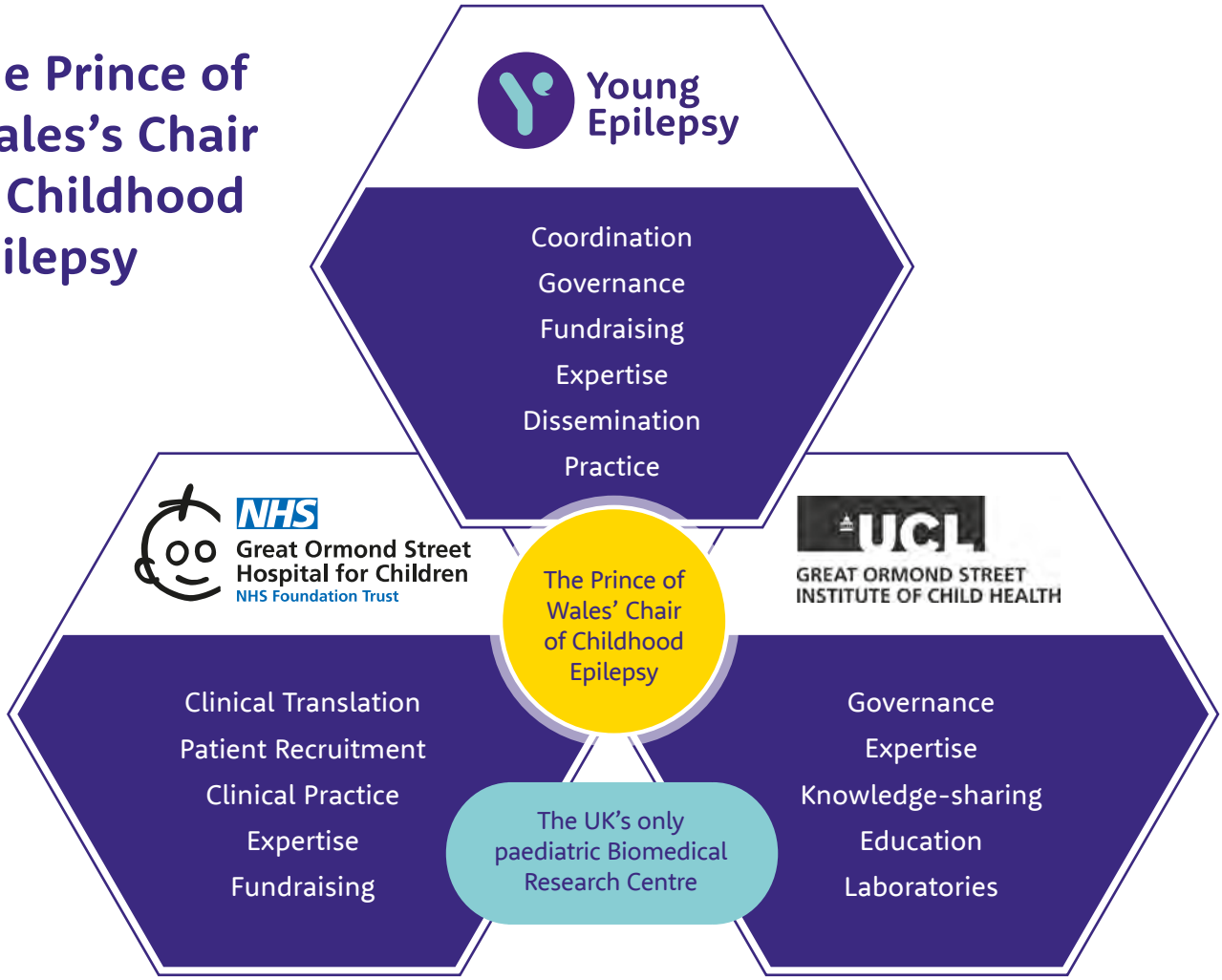
Who we are

Led by the Prince of Wales's Chair of Childhood Epilepsy, Professor Helen Cross, our research programme is a collaborative scheme between Young Epilepsy, Great Ormond Street Hospital and UCL GOS - Institute of Child Health.

Collaboration and integrated working across the partner organisations puts us in a unique position to incorporate data which spans:

- The entire range of complexity and comorbidity in epilepsy
- All stages of diagnosis and care
- The full age range, from neonates to young adults
- Multidisciplinary expertise to improve holistic understanding of epilepsy and service design.

The Prince of Wales's Chair of Childhood Epilepsy



Young Epilepsy exists to create a society where children and young people with epilepsy are enabled to thrive and fulfil their potential. A society in which their voices are respected and their ambitions realised.

Under our three key offers; health and research, voice and support and St Piers special education, we aim to:

- Coordinate research that improves diagnosis and treatments, and deliver cutting-edge health services.
- Campaign for children's rights, supporting them in school and college, and providing innovative tools, information, and practical help for living day-to-day life.
- Provide an innovative and creative environment for children and young people with epilepsy, autism, and severe learning difficulties.

Great Ormond Street Hospital for Children (GOSH) is an international centre of excellence in child healthcare, at the forefront of paediatric training in the UK. Together with UCL GOS - Institute of Child Health, GOSH forms the UK's only Biomedical Research Centre specialising in paediatrics. Most of the children we care for are referred from other hospitals throughout the UK and overseas. There are 63 different clinical specialties at GOSH; the UK's widest range of specialist health services for children on one site. 60% of the UK's epilepsy surgeries are carried out at GOSH.

University College London Great Ormond Street-Institute of Child Health (ICH) together with its clinical partner Great Ormond Street Hospital for Children (GOSH), forms the largest concentration of children's health research in Europe.

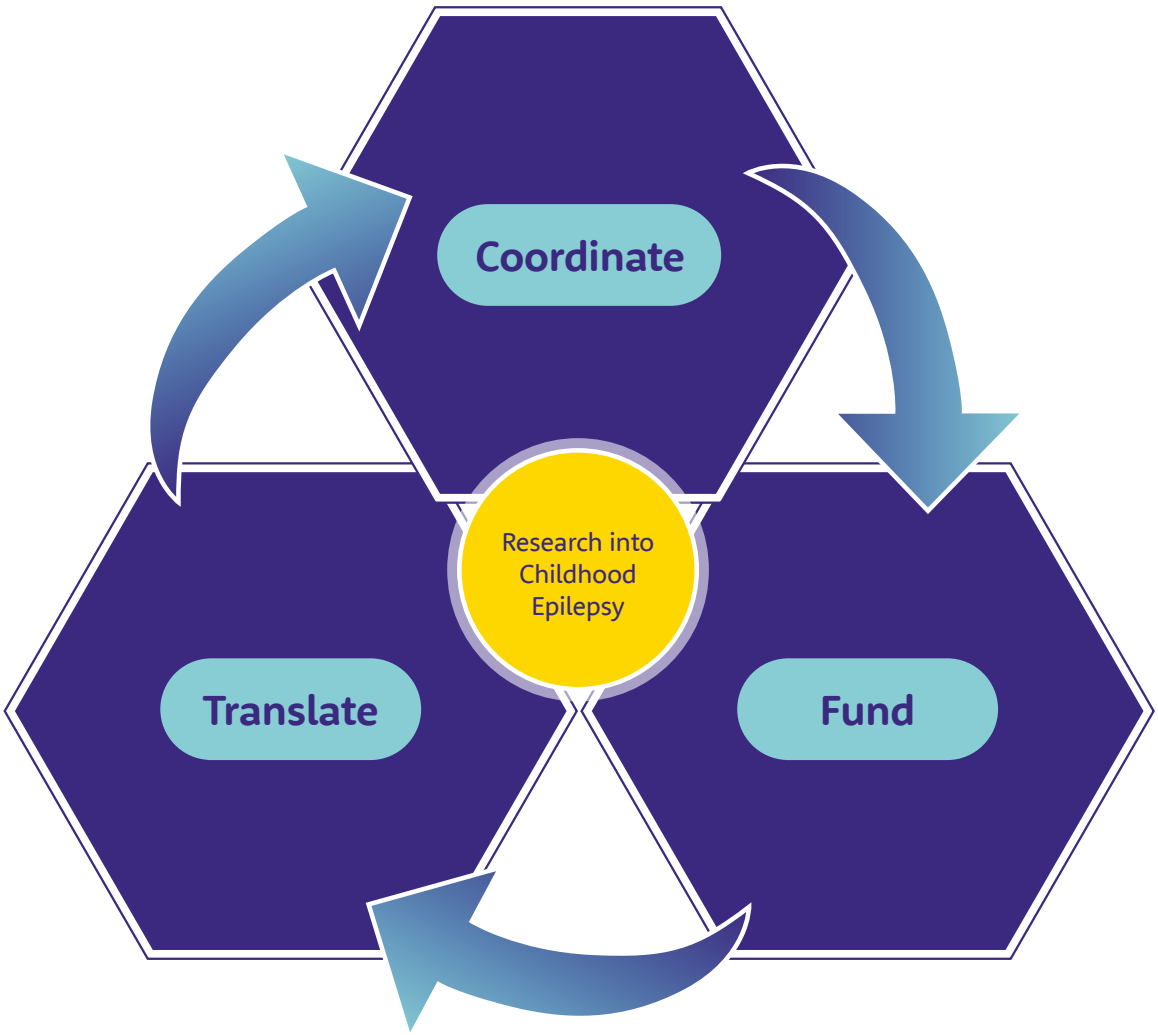
The inspirational mission of the UCL Great Ormond Street Institute of Child Health is to "improve the health and well-being of children, and the adults they will become, through world-class research, education and public engagement".

The academic strategy of GOS ICH is focused on five scientific research and teaching departments:

- Developmental Neurosciences
- Developmental Biology and Cancer
- Genetics and Genomic Medicine
- Infection, Inflammation and Immunology
- Population Policy and Practice

What We Do Research Strategy

Our research programme exists to ensure the best outcome for every child by optimising diagnosis, treatment, and support for all aspects of childhood epilepsy.



* The welfare of animals used in research is very important to Young Epilepsy, GOSH and ICH. Researchers would prefer not to use animals at all so we follow the guidance of the Association of Medical Research Charities. These principles are called the 3Rs:

- **Replace** the use of animals with alternative techniques or avoid the use of animals altogether.
- **Refine** the way experiments are carried out, to make sure animals suffer as little as possible. This includes better housing and improvements to procedures which minimise pain and suffering and/or improve animal welfare.
- **Reduce** the number of animals used to a minimum by seeking ways to find out information from fewer animals or more information from the same number of animals.

Workstream 1: Understanding Childhood Epilepsies

Around half of people diagnosed with epilepsy never learn the cause of it. This is concerning from both the personal and clinician perspective. The more we know about what causes epilepsy and how else the underlying cause is affecting the individual patient, the better clinicians can manage and treat, and the better the patient can understand themselves.

GOAL 01

Gain a better understanding of the medical causes of epilepsy

The majority of epilepsy treatment is symptomatic. The more we know about the underlying causes of the epilepsies, the more chance there is of developing curative, targeted treatments.

- ✓ Cohort studies to evaluate prevalence, natural history and outcome of comorbidities
- ✓ Studies to determine the molecular or genetic basis to the epilepsies
- ✓ Collaborative outcome studies
- ✓ Enhanced structural studies using neuroimaging to increase detection of structural correlates
- ✓ Pathological examination of tissue from surgical specimens to enhance our understating of structural correlates and related epileptogenesis

25% projects currently contribute to this goal

GOAL 02

Gain a better understanding of how epilepsy affects development and behaviour

Epilepsy is associated with myriad comorbidities. Evidence suggests that the effects of these comorbidities have a greater impact than seizures over the course of someone's life. This work will help us to understand how to treat epilepsy holistically.

- ✓ Cohort epidemiological studies to determine incidence, prevalence and outcome
- ✓ Population and family studies to gain further insights into new treatments
- ✓ Correlative studies in neurophysiology to enhance detection of origin
- ✓ Experimental animal model studies* to examine the effects of epileptiform discharges on development
- ✓ Correlative neurophysiology and neuropsychology studies

20% projects currently contribute to this goal

Workstream 2: Outstanding Treatment

Epilepsy treatments have not changed very much over time and the process of finding the right combination of treatments for each patient takes a long time. This is very hard on patients – especially if they are young. Continued advancement of imaging, surgery, dietetics, genomics and targeted treatment, and new medicines is crucial in the quest to effectively treat, and one day perhaps cure, every epilepsy.

GOAL 03

Improving diagnosis and treatment to determine the benefits of early interventions in improving long-term outcomes

The longer one has epilepsy, the longer its underlying cause is able to threaten or cause damage. Effective diagnostic processes, optimal treatments and early intervention are vital in slowing or halting any damage.

- ✓ Short and long-term evaluation of outcome following early epilepsy surgery
- ✓ Evaluation of new medical treatments
- ✓ Evaluation of educational intervention
- ✓ Novel diagnostic and imaging methods

30% projects currently contribute to this goal

Workstream 3: Outstanding Support

This workstream is set to tackle the wider challenges associated with growing up with epilepsy and in treating childhood epilepsies. It is important to know what epilepsy is and how to treat it but if the systems and supports are not in place to act on this knowledge then patients cannot benefit.

GOAL 04

Gain a better understanding of barriers to learning and determine the benefits of educational interventions.

We know that epilepsy can affect the way people learn and therefore may significantly affect someone's academic achievement if not properly understood. We want to know exactly what the challenges are and how best to support children with epilepsy in education.

- Evaluation of measures of progress in children with severe impairments
- Evaluation and development of targeted educational interventions across all educational settings
- Evaluating and enhancing the understanding of professionals working with children with epilepsy

5% projects currently contribute to this goal

GOAL 05

Make life better for children and families and make support systems more effective

Childhood epilepsy can affect the whole family and treatment must involve multiple disciplines and agencies. Support for families must be evidenced, treatment pathways must be made more efficient and the family voice should be reflected in research. Evidencing these needs allows service providers to plan more effective services.

- Patient and public inclusion and representation in research design and management
- Interventional behaviour programmes
- Rehabilitation and follow-up studies
- Assessment of service provision
- Evaluation of the impact of epilepsy on family life
- Evaluation of the economic costs involved in epilepsy care

17% projects currently contribute to this goal

GOAL 06

Develop a network of multidisciplinary professionals to strengthen our research and shape the education of future practitioners

To ensure the continuation of excellent research in paediatric epilepsy by nurturing future talent and continually improving knowledge

- Development of training fellowships
- Projects working towards higher degrees with encouragement for independent working thereafter
- Joint working between ICH, GOSH and Young Epilepsy
- Enhancing research and interoperability across all areas of expertise
- Providing specialist education events and networking opportunities

3% projects currently contribute to this goal

Summary of Research Projects

This section provides a brief overview of two key projects from this year's work. This is followed by a summary list of the current and competed projects during July 2022 to June 2023.

The projects are presented under the workstream that they contribute to most.

To find out more details about each of these projects please visit:

www.youngepilepsy.org.uk/what-we-do/health-research/research

Young Epilepsy

Key Projects

The Mental Health Intervention for Children with Epilepsy (MICE) Project

Chief Investigators:

Professor Roz Shafran and Professor Helen Cross.

Co-Investigators:

Dr. Sophie Bennett, Dr. Anna Coughtrey, Professor Sarah Byford, Professor Bruce Chorpita, Professor Caroline Dore (replaced by Dr. Hakim-Moulay Dehbi), Emma Dalrymple, Professor Peter Fonagy, Professor Tamsin Ford, Professor Isobel Heyman, Professor Rona Moss-Morris, Dr. Colin Reilly, Professor Jonathan Smith, Professor Terence Stephenson, Dr. Sophia Varadkar.

Comprehensive Clinical Trials Unit (CCTU) MICE Team:

James Blackstone, Kashfia Chowdhury, Harriet Quartly

Background:

At least half of young people with epilepsy also have mental health problems such as depression, anxiety and behaviour difficulties. Many young people have more than one of these problems. These difficulties have a very significant negative impact on the quality of life of the young people with epilepsy and their families and often have a greater impact than the epileptic seizures. Existing epilepsy services are separate from mental health services and mental health problems in young people with epilepsy may not be identified or if identified not treated as well as they could be.

Method:

The MICE team worked with health professionals, parents, children and young people to modify the Modular Approach to Therapy for Children ('MATCH-ADTC') so the treatment meets the special mental health needs of young people with epilepsy. This was done by developing an extra module specifically to help children and young people with anxiety, depression or behaviour problems in the context of epilepsy. The researchers recruited young people, aged 3-18 years, with epilepsy across sites in England, to complete the Strengths and Difficulties Questionnaire (SDQ) within the young person's epilepsy service. Those who scored above the clinical threshold using an algorithm developed in our Programme Development Grant (PDG) were invited to complete the Development and Wellbeing Assessment ('DAWBA') online to establish if they meet DSM-V diagnostic criteria for a common mental health disorder.

MICE is an NIHR funded research study that is testing the efficacy of an evidence-based psychological treatment for young people with epilepsy and mental health problems. It began in October 2017. The aim of the project is to improve the treatment of mental health problems in young people with epilepsy.

Those that meet diagnostic threshold were invited to participate in the Randomised Controlled Trial (RCT) until a sample of 334 had been recruited.

Half of the 334 children were randomly assigned to receive the modified version of MATCH-ADTC in addition to their usual care. The other half were randomly assigned to receive an enhanced version of their usual care. Therapists and their clinical supervisors were trained across sites to deliver MATCH.

The purpose of the RCT was to evaluate the clinical and cost-effectiveness of adding MATCH (with the additional epilepsy-specific module) to standard care for mental health disorders. The primary outcome measure is the SDQ, independently assessed six months post-randomisation. All analyses were conducted on an intention-to treat basis and blind to treatment assignment. Measures were repeated one year post-randomisation.

While receiving the MATCH intervention, 24 participants were invited to take part in two in-depth interviews, one before their therapy started and one six months later. The experiences of young participants and/or their carers, with regards to how their epilepsy and psychological and emotional wellbeing evolve over time is being explored. The aim is to complement the quantitative measures to evaluate the intervention's effectiveness in terms of outcomes and processes. The interviews are being analysed longitudinally using Interpretative Phenomenological Analysis (IPA), a method widely used to understand individual experience in health psychology.

The public and patient involvement (PPI) research advisory group (RAG) and epilepsy charities have ensured that the research has focussed on issues that matter most to them. Epilepsy Research UK (ERUK) have published a blog based on the PPI RAG members' experiences of being involved in research and most importantly what they wish they would have known about caring for a child with epilepsy from the start.

Analysis:

The two groups were compared 6 and 12 months after the start of treatment to see if there are differences in terms of the mental and physical health of children and young people. We are also trying to work out if the new treatment offers value for money, as well as talking to the young people and families to understand their experience of the treatment and how we might improve it for future patients.

Results:

Recruitment for the MICE study was concluded in February 2022. Overall, 334 participants were included in the trial. The data analysis for the mental health measures at 6 and 12 months after the start of treatment show that the MICE group had better mental health than the control group at both time-points. Overall, the results of the trial to date demonstrate that mental health comorbidities can be effectively and safely treated by a variety of clinicians, utilising an integrated intervention across ages and in the context of intellectual disability and autism spectrum disorder.

Ketogenic Diet in Infants with Epilepsy (KIWE) Project

For full article, published in Lancet Neurology see:

www.sciencedirect.com/science/article/pii/S1474442223003708

Chief Investigator:

Professor Helen Cross.

Co-Investigators:

Dr Natasha E Schoeler, Prof Louise Marston, Dr Laura Lyons, Dr Sally Halsall, Ruchika Jain, Siobhan Titre-Johnson, Maryam Balogun, Prof Simon J R Heales, Dr Simon Eaton, Dr Michael Orford, Dr Elizabeth Neal, Dr Christin Eltze, Dr Elma Stephen, Dr Andrew A Mallick, Prof Finbar O'Callaghan, Dr Shakti Agrawal, Dr Alasdair Parker, Prof Martin Kirkpatrick, Prof Andreas Brunklaus, Dr Ailsa McLellan, Dr Helen McCullagh, Dr Rajib Samanta, Dr Rachel Kneen, Dr Hui Jeen Tan, Dr Anita Devlin, Dr Manish Prasad, Dr Rohini Rattihalli, Dr Helen Basu, Dr Archana Desurkar, Dr Ruth Williams, Dr Penny Fallon, Prof Irwin Nazareth, Prof Nick Freemantle.

Background:

Infants who develop epilepsy in the first two years of life are most at risk of continuing seizures and poor neurodevelopmental outcomes. Early seizure control has been associated with more favourable neurodevelopmental outcome, but many early-onset epilepsies are associated with poor seizure control. A high-fat, low-carbohydrate diet called the 'ketogenic diet' is a non-pharmacological treatment option for individuals with drug-resistant epilepsy. The diet has been shown to reduce seizures in children older than 2 years of age and adults who continue to have seizures despite taking seizure medication. However, the evidence for the use of ketogenic diets in infants with epilepsy is scarce.

Method:

This multicentre randomised controlled trial aimed to establish whether the ketogenic diet can reduce seizure frequency, compared with further antiseizure medications, in infants (aged 1-24 months) with drug-resistant epilepsy.

Between 1st January 2015 & September 30th 2021, 136 infants were recruited onto the study and were randomly assigned to receive either a classic ketogenic diet or a further antiseizure medication for 8 weeks. 78 infants started a ketogenic diet and 58 started taking another medicine. Before receiving treatment, participants were observed for 1-2 weeks. During this time, seizure frequency was recorded.

For those assigned to the ketogenic diet, food plans were calculated on an individual basis by a paediatric dietitian. Caregivers of infants assigned to the ketogenic diet also underwent training before starting the diet. For those assigned to a further antiseizure medication, the infant's clinician prescribed the most appropriate drug. Infants then stayed on their allocated treatment for 8 weeks, during which time caregivers kept daily seizure diaries. Infants were followed up at 4 and 8 weeks, as well as 6, 9 and 12 months.

KIWE
Ketogenic diet in infants with epilepsy

Findings:

The primary outcome was the number of seizures recorded during weeks 6-8 of the treatment, with adjustment for the number of seizures before the treatment began. Overall, the number of seizures that the infants had was similar in both treatment groups. Specific components of quality of life such as growth, temperament and mood were more favourable at 8 weeks in the ketogenic diet group than the medicine group. Communication and socialisation were also higher in the ketogenic diet group at 12-month follow-up, but these differences were not statistically significant. A similar number of infants in both groups either had to go hospital or stay in hospital for longer due to illness; these stays were often due to seizures. Overall, infants in both groups had a similar rate of side effects.

Conclusion:

In summary, we have no evidence to say that the ketogenic diet was any more effective in infants with epilepsy, compared to further antiseizure medication. Both treatments were well-tolerated and appeared safe to use in this age group. Doctors could, therefore, consider starting a ketogenic diet in babies who continue to have seizures even if they have tried antiseizure medicines.

Current projects

To find out more details about each of these projects please visit:
www.youngepilepsy.org.uk/what-we-do/health-research/research

Workstream 1: Understanding Childhood Epilepsies

01 Functional effects of SCN1A mutations – New insights from biophysics and computational modelling

Project Aim: Linking functional properties of SCN1A miss-sense mutations with their resultant phenotypes.
Investigators: Richard Rosch, Elaine Hughes, Kathleen Gorman, Colin Peters, Peter Ruben

02 Gene-STEPS: Shortening Time of Evaluation in Paediatric epilepsy Services: a multi-centre prospective evaluation of the impact of early genetic diagnosis on patient outcomes

Project Aim: To implement rapid trio WGS for all children, utilise electronic healthcare records and research databases to unite phenotypic and genomic data and assess the impact of early genetic diagnosis on epilepsy, developmental, and health economic outcomes through formal longitudinal assessments of all children enrolled.
Investigators: Amy McTague, Helen Cross, Lyn Chitty, Neil Sebire
With: Annapurna Poduri (Boston Childrens), Katherine Howell, Ingrid Scheffer (Royal Childrens Hospital Melbourne), Gregory Costain, Vann Chau (The Hospital for Sick Children Toronto)

03 Shining a light on the genetic basis of Sunflower syndrome

Project Aim: Investigate the genetic basis of this rare photosensitive epilepsy.
Investigators: Amy McTague, Manju Kurian

04 Multicentre Epilepsy Lesion Detection (MELD) Project

Project Aim: Create open-access, robust and generalisable tools for understanding and detecting focal cortical dysplasias (FCDs) that can assist the pre-surgical evaluation of patients with drug resistant epilepsy.
Investigators: Sophie Adler-Wagstyl, Mathilde Ripart, Hannah Spitzer, MELD consortium, Helen Cross, Torsten Baldeweg, Konrad Adler-Wagstyl

05 MELD Focal Epilepsies Project

Project Aim: To improve epilepsy surgery outcomes by developing Artificial Intelligence (AI) algorithms to automatically find subtle abnormalities on patients' MRI scans and help neurosurgeons to plan operations that will completely remove them.
Investigators: Sophie Adler-Wagstyl, Konrad Adler-Wagstyl, Torsten Baldeweg, John Duncan, Juan Eugenio Iglesias, Helen Cross

06 Modelling childhood genetic epilepsies in zebrafish larvae

Project Aim: Identifying whole-brain network dysfunction at single neuron resolution in larval zebrafish models of genetic epilepsies
Investigators: Richard Rosch, Dominic Burrows, Jade Lau, Martin Meyer

07 The neuropathology of focal epilepsy in children

Project Aim: To understand the biology underlying the diseases that cause focal epilepsy.
Investigators: Tom Jacques, Helen Cross, Martin Tisdall, Darren Hargrave

08 Memory profile and reorganisation after epilepsy surgery in children with intractable Temporal Lobe Epilepsy (TLE)

Project Aim: To characterise the memory profile of children and young people and depict functional and structural reorganisation of memory networks in temporal lobe epilepsy before and after surgery, using functional magnetic resonance imaging (fMRI) and diffusion tensor imaging (DTI) magnetic resonance.
Investigators: Filipa Bastos, Faraneh Vargha-Khadem, Helen Cross, Jonathan Clayden, Sarah Buck

09 The genetics of early onset epileptic encephalopathy

Project Aim: The project aims to identify novel early onset epileptic encephalopathy genes which will contribute to the understanding of the disease mechanisms involved in such epilepsies.
Investigators: Amy McTague, Helen Cross, Dimitri Kullmann, Rod Scott, Manju Kurian

10 A natural history of Pyruvate Dehydrogenase Complex deficiency

Project Aim: To describe the natural history of Pyruvate Dehydrogenase Complex (PDC) deficiency from childhood to adulthood, including the spectrum of molecular diagnoses in affected patients.
Investigators: Nandaki Keshavan, Shamima Rahman

11 Novel network analysis of intracranial stereoelectroencephalography (SEEG)

Project Aim: To characterise interictal abnormalities in single unit neural dynamics and to establish whether the regions that display abnormal dynamics are consistent with the epileptogenic zone
Investigators: Rod Scott, Martin Tisdall, Aswin Chari, Rachel Thornton

12 Multiscale modelling of epileptic networks from SEEG recordings

Project aim: Develop analysis techniques that allow us to understand how changes in brain networks in patients with drug-resistant epilepsy undergoing epilepsy surgery result in the patients' epilepsy.
Investigators: Richard Rosch, Ulrich Stoof, James Wilsenach, Aswin Chari, Martin Tisdall, Gerald Cooray, Karl Friston

13 Landau-Kleffner syndrome: Patterns in the recovery phase

Project Aim: A retrospective case note review examining cognitive and language trajectories across different phases of Landau-Kleffner syndrome (LKS).
Investigators: Maria Clark, Gemma Wilson

14 EAGLET: EEG vs aEEG to improve the diagnosis of neonatal seizures and Epilepsy - a Randomised Trial

Project Aim: EAGLET is a prospective multicentre randomised controlled trial to evaluate whether the combination of cEEG with aEEG is superior to aEEG in the real time evaluation and diagnosis of neonatal seizures and in reducing time to treatment.
CIs: Ronit Pressler and David Rowitch
Co-investigators: Topun Austin, Paul Clarke, Claudia Chetcuti-Ganado

15 The Meerkat Project

Project Aim: The Meerkat project aims to develop non-contact monitoring for neonates in intensive care. A collaboration between the Departments of Engineering and Paediatrics at the University of Cambridge, as well as universities in the UK and Europe, the project will leverage expertise in image processing and machine learning to improve neonatal care.
CI: Kathy Beardsall
Co-investigators: Alex Grafton, Peter Marschik, Ronit Pressler, Oliver Bonner

16 Epilepsy in Infancy: relating phenotype to genotype (EPIPEG)

Project Aim: To identify and follow-up a cohort of children with new onset of epilepsy under 12 months of age to enable definition of neurobehavioral phenotypes; identify risk factors for neurodevelopment and later intellectual disability.
Investigators: Helen Cross, Manju Kurian, Rod Scott, Christin Eltze, Finbar O'Callaghan, Michelle De Haan, Elaine Hughes, Jane Kung, Manuela Pisch, Katy Barwick, Aikaterini Vezyroglou

17 Turning6 - A Clinical and Neurodevelopmental follow up of EpiPEG participants at 60 months

Project Aims:

- ✓ Characterise the neurodevelopmental (cognition, behaviour, sleep) status of children who had epilepsy in the first year of life
- ✓ Examine the association between initial neurodevelopmental and clinical assessment results and performance at follow-up
- ✓ Examine factors including epilepsy factors and neurodevelopmental status associated with current performance and changes in performance between initial assessment and follow-up

Investigators: Colin Reilly, Finbar O'Callaghan, Manuela Pisch, Abigail Wooldridge, Sasha Coates, Colette Meades, Bhavna Sidphara, Amy Muggeridge, Lara Carr, and Helen Cross

Current projects

Workstream 2: Outstanding Treatment

18 Realising the potential of 7T MRI for paediatric imaging

Project Aim: To enable the first 7 Tesla (7T) magnetic resonance imaging (MRI) of paediatric patients with epilepsy being evaluated for surgery at GOSH and Kings College London Hospital (KCLH).
Investigators: David Carmichael, Helen Cross, Martina Callaghan, Shaihan Malik, Thomas Booth, Sila Dokumaci, Fred Dick, Dr Simon Richardson, Serena Counsell, Alex Hammers, Jonathan O'Muircheartagh

19 The fast without the spurious: developing a system for robust and rapid simultaneous EEG-fMRI measurements

Project aim: To develop more advanced EEG-fMRI scans that may better detect brain areas active at the start of seizures. To do this we are trying new motion-correction technology that tells the scanner where the head is using camera and a marker attached to a dental retainer and updates the scanner accordingly.
Investigators: Amy McDowell, Danilo Maziero, David Carmichael, Helen Cross, Kelly St Pier, Nikolaus Weiskopf, Mirja Steinbrenner

20 The 7T Temporal Lobe Epilepsy Study

Project aim: The 7-TLE study is a prospective neuroimaging study that is using super-high-field (7-Tesla) MRI to investigate the network abnormalities in children and adults with temporal lobe epilepsy.
Investigators: Rory Piper, Shan-Shan Tang, Alexander Hammers, Atta Siddiqui, John Duncan, Martin Tisdall and David Carmichael, Torsten Baldeweg

21 Dynamic variability in the epileptic brain

Short Project aim: Investigate how epileptic brain activity changes over time at multiple scales (seconds, minutes, days), in order to understand how our diagnosis and interventions can be targeted appropriately.
Investigators: Richard Rosch, Jamie Norris, Stuart Smith, Martin Tisdall, Gerald Cooray, Karl Fristo

22 The CADET Trial: The Children's Adaptive Deep brain stimulation for Epilepsy Trial

Project Aim: To determine the safety and feasibility of a novel non CE licensed DBS device for children with Lennox Gastaut Syndrome.
Investigators: Martin Tisdall, Helen Cross, Tim Denison, Harutomo Hasegawa, Elaine Hughes, Marios Kaliakatsos, Kei Landin, Rory Piper, Richard Selway, Antonio Valentin

23 Determining the utility of OPM-MEG in a clinical context

Project Aim: This project aims to fast-track regulatory approval of a new OPM-MEG system, making it the first, and only OPM-MEG system in the world to be approved for human use.
Investigators: Christine Embury, Zelekha Seedat, Kelly St Pier, Lara Carr, Eliot Dawson, Freya Jackson, Dominic Sims, Rosemarie Pardington, Elena Boto, Matt Brookes.

24 Modelling neuronal dysfunction in early onset epilepsies; a patient-centric approach

Project Aims:
- To create and characterise a patient-derived induced pluripotent stem cell (iPSC) organoid model Epilepsy of Infancy with Migrating Focal Seizures (EIMFS).
- To investigate the neuronal phenotype of EIMFS at a cellular and network level.
- To investigate the impact of novel therapies.

Investigators: Amy McTague, Dimitri Kullmann, Gabriele Lignani, Jenny Lange, Manju Kurian

25 Is pyridox(am)ine 5'-phosphate oxidase deficiency, an eminently treatable cause of epilepsy, under-recognised in children?

Project Aim: To improve diagnosis and treatment of children with pyridox(am)ine 5'-phosphate oxidase (PNPO) deficiency by using a novel rapid screening dry blood spot assay.
Investigators: Peter Clayton, Philippa Mills, Helen Cross, Ronit Pressler

26 The Diagnosis and Management of Pyridoxamine 5'-Phosphate Oxidase Deficiency

Project Aim: To create guidelines for the diagnosis, treatment and follow up of Pyridoxamine 5'-Phosphate Oxidase Deficiency which will facilitate clinical decision making and improve the care for patients with PNPO-deficiency in a standardised manner.
Investigators: Philippa Mills and Emma Footitt

27 The "Pair Test": an App to diagnose learning and memory impairments in children with Temporal Lobe Epilepsy

Project Aim: To provide better informed diagnosis of memory impairments in children with epilepsy and predict outcome after surgery in the temporal lobe, using the Pair Test.
Investigators: Sarah Buck, Torsten Baldeweg, Filipa Bastos, Faraneh Vargha-Khadem

28 Optimisation and bioperformance of a novel formulation of pyridoxal 5'-phosphate for treatment of pyridox(am)ine 5'-phosphate oxidase deficiency induced epilepsy in children

Project aim: To test the performance in the lab and in vivo of an improved pyridoxal 5'-phosphate (PLP) option for children with pyridox(am)ine 5'-phosphate oxidase deficiency induced epilepsy.
Investigators: Catherine Tuleu, Peter Clayton, Philippa Mills, Emma Footitt, Ahad Rahim, Simon Heales

29 Cooling in Mild Encephalopathy Trial (COMET)

Project Aim: The goal of this randomised control trial is to evaluate the safety, efficacy, and cost-effectiveness of whole-body hypothermia as a therapy for babies with mild HIE.
Investigators: Prof Sudhin Thayyil, Seetha Shankaran, Dr Ronit Pressler, Prof Andrew Shannon, Dr Kerry Woolfall, Prof Samantha Johnson, Prof Patricia Grant, Dr Farah Alobeidi, Prof Stavros Petrou, Mrs Sarah Land, Mrs Mariam Mahmoud, Ms Stuti Pant, Mr Paul Basset, Mr Tony Brady, Prof Victoria Cornelius, Dr Aung Soe, Dr Eleri Adams, Prof Jon Dorling, Dr Ella Chakkarapani, Dr Balamurugan Palanisami, Dr Paolo Montaldo

30 Functional brain connectomics: implications for post-surgical outcomes in children with focal epilepsy

Project Aim: : In this project we will estimate how strongly seizure generating parts of the brain (the surgical target zones) are connected to other, healthy parts of the brain.
Investigators: Xiyu Feng, Jon Clayden, Torsten Baldeweg

31 Reconstruction and Computational Modelling for Inherited Metabolic Diseases [Recon4IMD]

Project Aims: : Using personalised computational modelling to

- Accelerate the diagnosis of patients at risk of an inherited metabolic disorder [IMD].
- Refine the diagnosis of patients at risk of an IMD.
- Stratify IMD patients by clinically actionable compensatory and aggravating metabolic mechanisms that associate with phenotypic severity.

Investigators: Professor Shamima Rahman [UCL is one of 12 participating organisations in this Horizon Medicine grant being coordinated by Professor Ronan Fleming at the University of Galway]

Current projects

Workstream 3: Outstanding Support

32 Epilepsy Carers Uniting with Researchers (E-Cure) PPI network

Project Aim: Strengthen the voice of children and young people with epilepsy in our research by establishing the UKs first network of parents, carers and young people who volunteer to shape childhood epilepsy research.
Investigators: Lara Carr, Samantha Chan, Amy McTague, Helen Cross

33 Epilepsy Pathway Innovation in Africa (EPInA)

Project Aims: : We have four main aims

- Societal change: ensure an enduring, positive change by improving public awareness and reducing the stigma experienced by people with epilepsy in sub-Saharan Africa.
- Diagnose: To improve the rate of accurate diagnosis of epilepsy by primary health care workers with app-based technologies.
- Treatment: increase the adherence to medication using text messaging
- Prevent: reduce the incidence of infection and peri-natal injury in an endemic region in Tanzania and the subsequent risk of epilepsy.

Investigators: Charles Newton, Arjune Sen, Helen Cross, Josemir Sander, Albert Akpalu, Patrick Adjei, Symon Kariuki, Damazo Kadengye, Gershim Asiki, Thomas Kwasa, Bruno Mmbando, Dan Bhwana, Tarun Dua, William Matuja, Sloan Mahone, David McDaid, Richard Walker

34 European Reference Network on rare and complex epilepsies (EpiCARE)

Project Aims:

- To improve accessibility of detailed diagnostics to individuals of all ages with rare and complex epilepsies across Europe, including clinical evaluation and investigation.
- To develop treatment protocols and monitor standardised outcomes of rare and complex epilepsies.
- To improve awareness and accessibility to protocols for physicians and individuals with rare and complex epilepsies across Europe for treatment.
- To enhance educational activities and training opportunities across Europe by interchange across the network.
- To enhance opportunities for registries, and collaborative research for the benefit of individuals with rare and complex epilepsies across Europe

35 Prevention of Epilepsy by reducing Neonatal Encephalopathy (PREVENT) study

Project Aim: To examine a care bundle approach to improve the maternal care around delivery to reduce number of babies sustaining serious birth related brain injury and epilepsy.
Investigators: Sudhin Thayyil, Helen Cross, Ronit Pressler, and many more.

36 Assessment of profound intellectual disability in complex epilepsy

Project Aim: To develop a robust assessment tool for children with complex epilepsy.
Investigators: Maria Clark, Gemma Wilson, Steve Rose, Karen Ray

37 Epilepsy in Schools: Developing web-based training for educational staff who support children with epilepsy in mainstream schools

Project Aims: The overall aim of this project is to develop, pilot and assess the feasibility of web-based interventions for staff currently supporting children with epilepsy.
The specific aims of this project are to:

- ✓ Co-develop web-based training for teachers and other educational staff who support children with epilepsy in mainstream schools.
- ✓ Conduct a pilot study of the developed web-training focusing on the knowledge and attitudes of educational staff in mainstream schools before and after the training.

Investigators: Collette Meades, Joan Idowu, Bhavna Sidhpara, Lara Carr, Helen Cross, Colin Reilly

Completed projects

Workstream 1: Understanding Childhood Epilepsies

38 Development in Hypothalamic Hamartoma

Project Aim: To review the developmental profiles of children with hypothalamic hamartoma in relation to their medical presentation and treatment.
Investigators: Hanna Richardson, Leah Bull, Varsha Siyani

39 Non-invasive modulation of brain network dynamics to suppress epileptic activity and improve cognition (EPICONN TM)

Project Aim: A pilot study to measure a reduction in epileptiform activity associated with transcranial electrical stimulation (TES).
Investigators: David Carmichael, Frederike Moeller, David Sharp, Helen Cross, Mirja Steinbrenner, Martin Tisdall, Mark Richardson, Ines Violante, Rory Piper

Workstream 2 – Outstanding Treatments

40 MELD (Multi-centre Epilepsy Lesion Detection) as an Adjunct for SEEG Trajectories (MAST) trial

Project Aim: Assess the utility of a novel machine learning algorithm in helping to plan electrode trajectories in children undergoing stereoelectroencephalography (SEEG).
Investigators: Aswin Chari, Sophie Adler-Wagstyl, Konrad Wagstyl, Zubair Tahir, Martin Tisdall

41 Wearable magnetoencephalography (MEG) at Young Epilepsy

Project Aim: To develop a new Epilepsy Diagnostic Suite at Young Epilepsy centered around the installation and evaluation of the OPM-MEG technology.
Investigators: Gareth Barnes, Richard Bowtell, Matthew Brookes, Helen Cross, Tim Tierney, Torsten Baldeweg, Rosemarie Pardington, Kelly St Pier, Zelekha Seedat, Konrad Wagstyl, Umesh Vivekananda, David Woolger

42 Development of a lifespan compliant magnetoencephalography system

Project Aim: Build an OP-MEG system for children aged 0-15years, that will offer direct clinical applicability, increased practicality, better data, and lower cost compared to current systems.
Investigators: Matthew Brookes, Richard Bowtell, Gareth Barnes, Helen Cross, Zelekha Seedat, Rosemarie Pardington

43 Ketogenic diet in Infants With Epilepsy (KIWE)

Project Aim: This is a randomised controlled trial to determine the effectiveness on seizure control of the ketogenic diet compared to alternative further antiepileptic drug treatment. Patients are children with epilepsy aged 1 month to 2 years who have failed to respond to two or more pharmacological treatments.
Investigators: Helen Cross, Laura Lyons, Sally Halsall, Natasha Schoeler, Maryam Balogun, Christin Eltze, Simon Heales, Helen McCullagh, Rachel Kneen, Tim Martland, Jeen Tan, Andrew Mallick, Andrew Lux, Alasdair Parker, Helen McCullagh, Archana Desurkar, Penny Fallon, Helen Basu, Anita Devlin, Rajib Samanta, Shakti Agrawal, Manish Prasad, Rohini Rattihalli, Elma Stephen, Andreas Brunklaus, Martin Kirkpatrick, Ailsa McLellan, Nick Freemantle, Louise Marston, Irwin Nazareth

Workstream 3 – Outstanding Support

44 Physical Activity in Childhood Epilepsy (PACE)

Project Aims:

- ✓ To compare levels of physical activity in secondary school-aged children with 'active' epilepsy, and matched healthy controls, using both survey methods and activity trackers.
- ✓ To better understand factors which may be associated with physical activity, including structured exercise/sports participation, in children with epilepsy.
- ✓ Identify the barriers to engagement in physical activity for young people with epilepsy.
- ✓ Explore the feasibility of implementing an intervention to improve levels of physical activity in children with epilepsy.

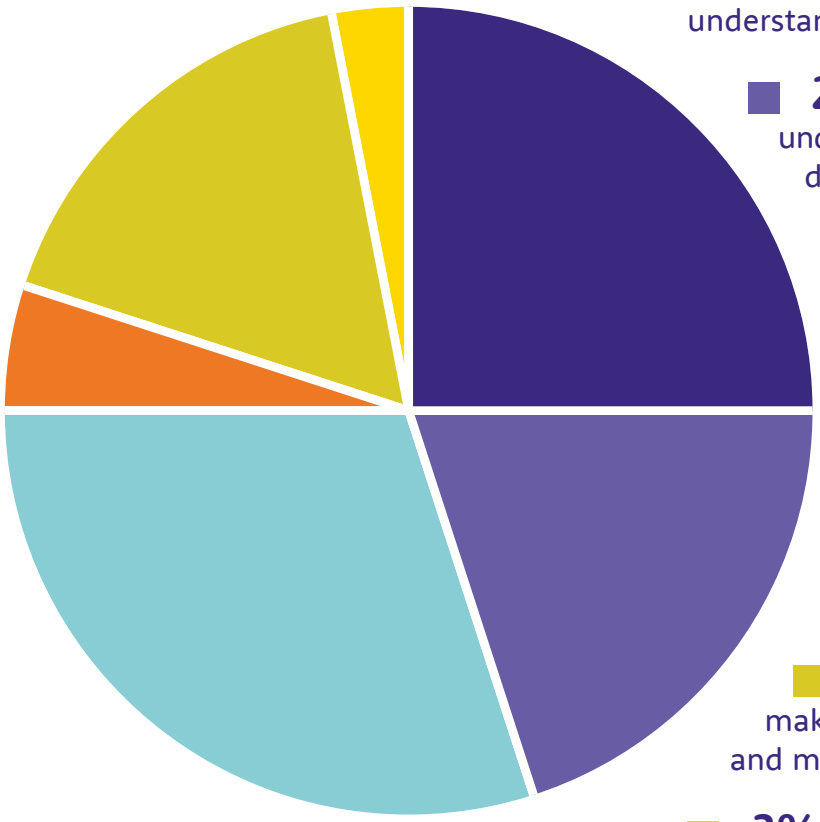
Investigators: Colin Reilly, Joan Idowu, Natalie Pearson, Colette Meades, Helen Cross, Lauren Sherar, Monica Lakhanpaul, Kerry Robinson, Amy Muggeridge, and Helen Cross.

45 Mental Health in Children with Epilepsy (MICE)

Project Aim: Establish the feasibility of routine screening and brief telephone intervention for mental health disorders in paediatric neurology clinics so children and young people with difficulties can access the support they need
Investigators: Roz Shafran, Helen Cross, Sophie Bennett, Sarah Byford, Bruce Chorpita, Anna Coughtrey, Emma Dalrymple, Caroline Dore, Peter Fonagy, Tamsin Ford, Isobel Heyman, Rona Moss Morris, Colin Reilly, Jonathan A Smith, Terence Stephenson, Sophia Varadka

Current & Past Impact

Between **July 2022** and **June 2023** the programme portfolio consisted of **25%, 20%, 30%, 5%, 17%** and **3%** contribute to each goal respectively



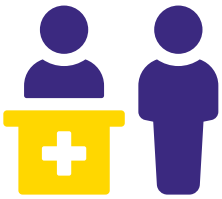
- **25%** of projects contributed to understanding the medical causes of epilepsy
- **20%** of projects contributed to understanding how epilepsy affects development and behaviour
- **30%** of projects contributed to improving diagnosis and treatment to determine the benefits of early intervention in improving long-term outcomes
- **5%** of projects contributed to understanding the barriers to learning and determining the benefits of educational interventions
- **17%** of projects contributed to making life better for children and families and making support systems more effective
- **3%** of projects contributed to developing a network of multidisciplinary professionals to strengthen our research and shape the education of future practitioners

Year on year most of our projects address:

Workstream 1
Understanding Childhood Epilepsies

Workstream 2
Outstanding Treatments

But we are growing our work in:



Workstream 3
Outstanding Services

57 published
peer-reviewed items of primary research

36 reviews and a further **2 Chapters** in Books

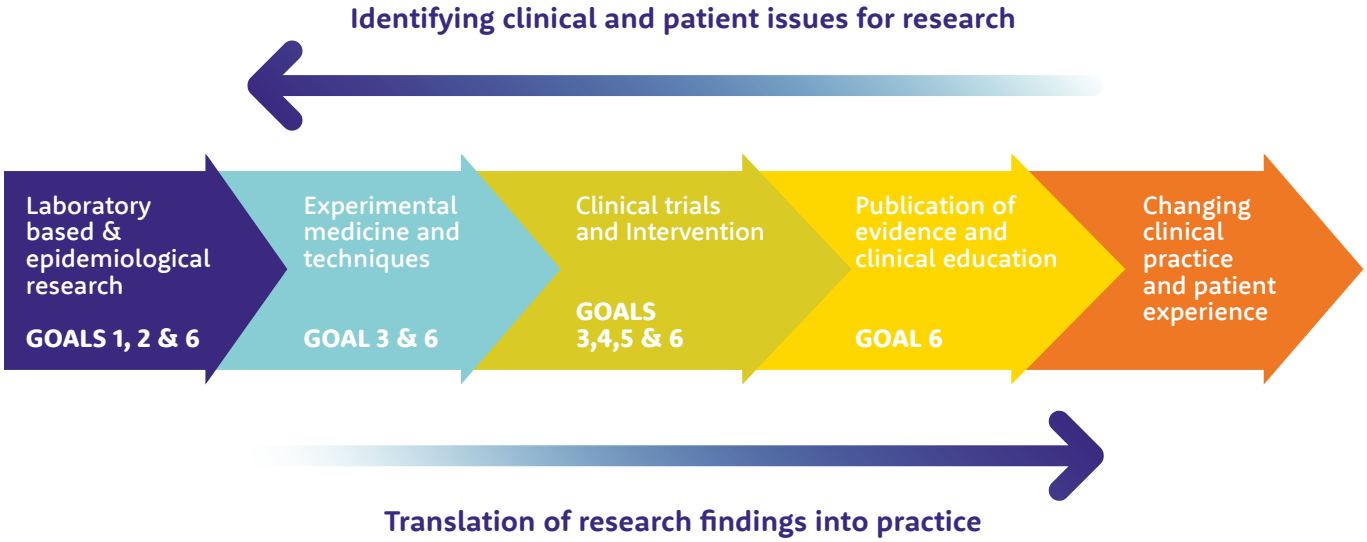
Specialist PPI network for childhood epilepsy with over **140 members**

The largest number of events to date. The Research Retreat was hosted in person for the first time in 2 years with **128 attendees**. We also hosted **2 additional in person events** and **2 webinars**

Meeting our research goals

Our research originates from the identification of clinical problems and feedback from patients. Ideas are then developed into project plans for which funding is sought and an expert team assembled.

The end result is to publish results as original research which has stood up to the review and critique of independent experts – a process known as peer review. This ensures robust evidence on which we can implement changes and/or conduct further research.



The number of research projects within the partnership as increased slightly from last year (Figure 1), with 45 active projects within the current year. In Figure 2 we have traced the contribution of active projects to the three workstreams and six goals. We are historically strongest in addressing Workstream 1 and 2 – Understanding Childhood Epilepsies and Outstanding Treatments (Figure 3). Given our clinical origins this is unsurprising, but we continue to encourage research under Workstream 3, Outstanding Support, through educational, psychosocial, and service-based research.

Figure 1: Number of active research projects per year

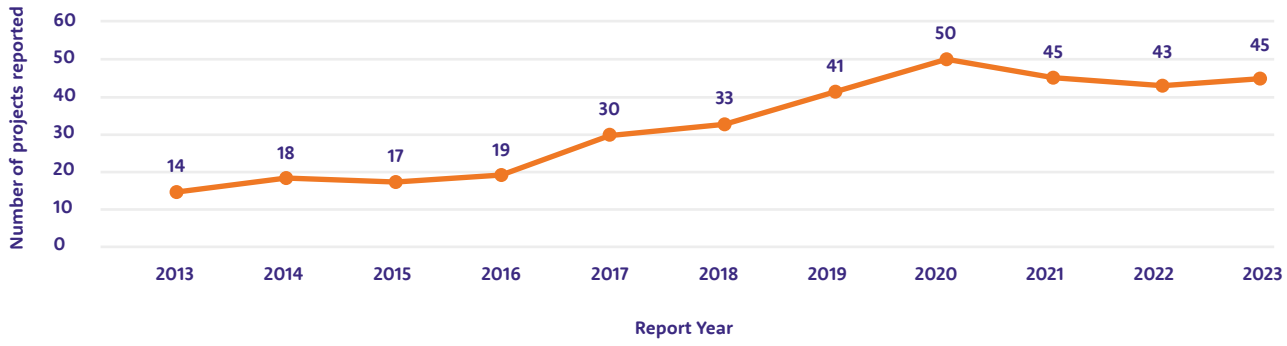
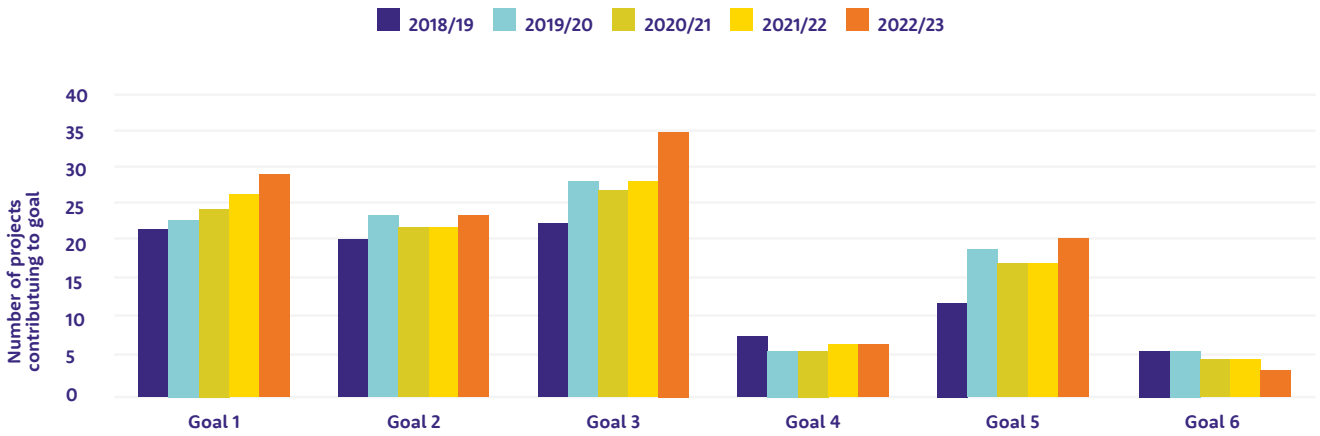


Figure 2: Number of active projects contributing to each goal

Many projects contribute to more than one workstream and/or goal



The strength of the evidence we publish

Figure 4 illustrates how our research programme continues to grow. We now have 31 Principal Investigators, supervising 21 PhD students and working alongside an additional 43 international collaborating researchers.

2023 has seen a rise in both the impact of our publications (Figure 6) and the reviews or communication providing expert opinion (Figure 5). The reduction in the number of publications of original research (Figure 5) is likely reflective of the ongoing impact of COVID-19 with projects that would have been published this year delayed due to imposed restrictions throughout 2020 and 2021. The growth in the number of active projects demonstrates we are likely to see an increase in the number of publications of original research in years to come.

Figure 4: Annual growth of the Research Unit Network

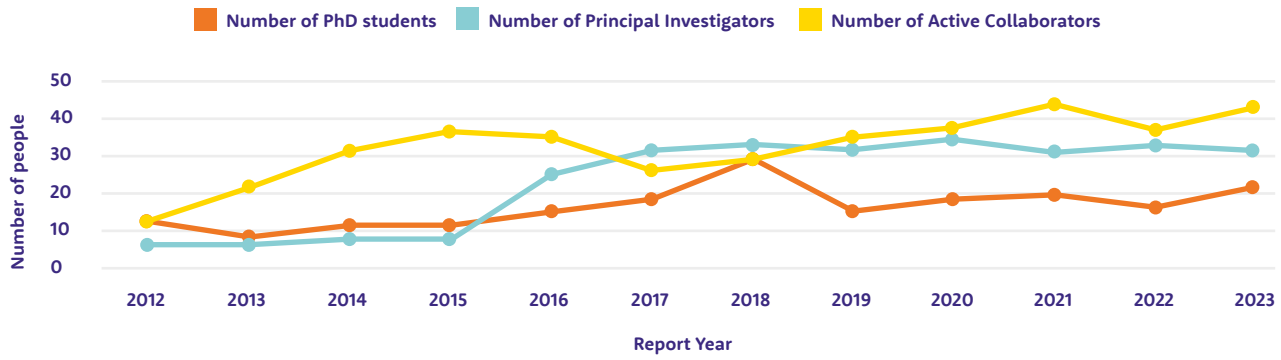
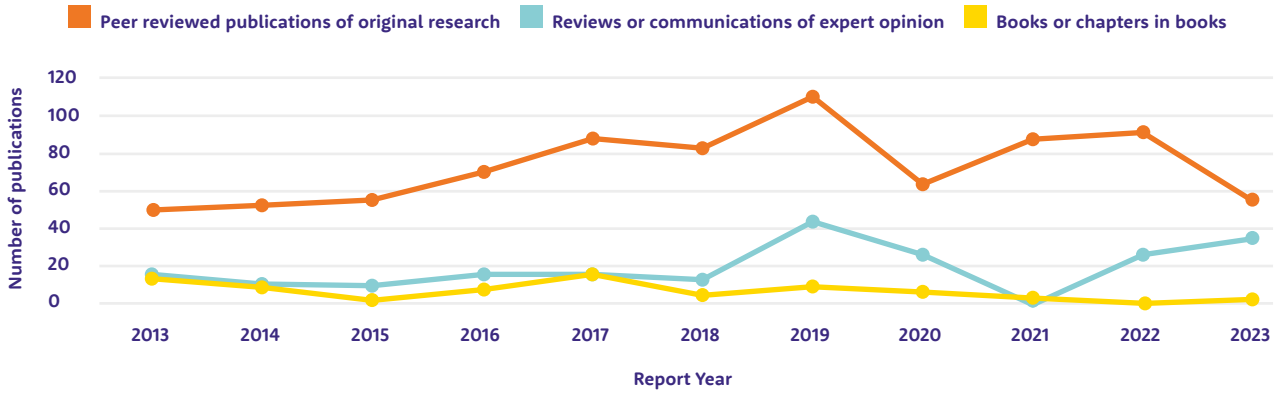


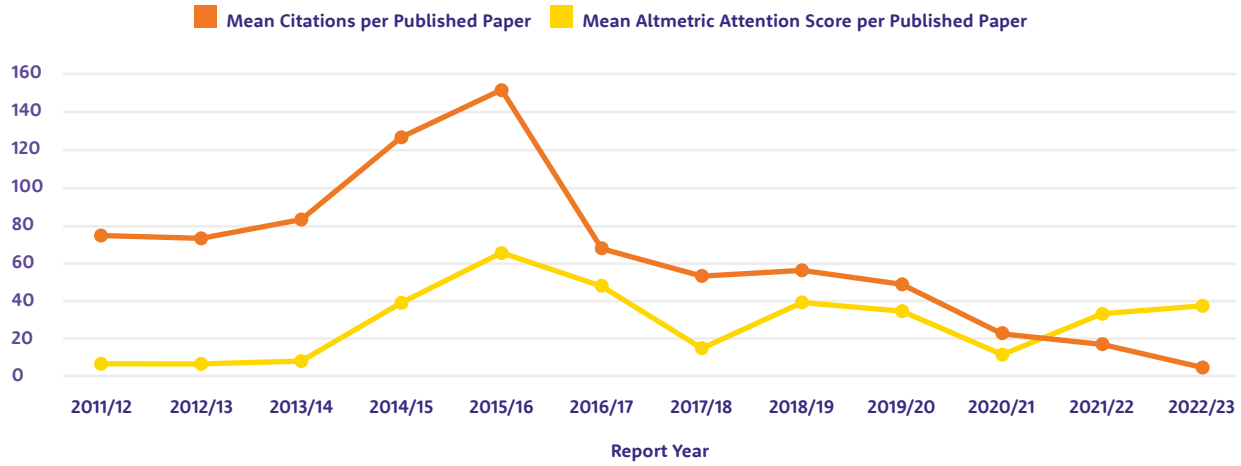
Figure 5: Number of research publications produced per year



We track the progress and influence of these research publications over time using two metrics – citations and an altmetric attention score (Figure 6). A citation is counted when an individual research paper is referred to in a later research publication as a source of evidence.

The altmetric attention score that we use is produced by an independent bibliographic data organisation, Dimensions.ai, and is calculated based on the public attention that an individual publication has received across news articles, social platforms, and policy documents.

Figure 6: Impact of research publications



These data illustrate that our publications this year have made more of an immediate impact than last. This year's publications have been cited less which is to be expected as the longer a publication is available, the more citations you can assume it will accrue. There is, however, a clear increase in the mean altmetric attention score, from 31.11 in 2021/22, to 36.96 in 2022/23 (Figure 6). This year, there are 6 papers with an altmetric score of > 100, meaning that we had 6 publications in the top 5% of all the research papers tracked by ReadCube, compared to 5 last year. Once again, one paper received a score of > 1000. We can perhaps attribute these high altmetric scores to our publications being of current global interest, in areas such as genomics or COVID-19. In 2023, for the fourth year running, Professor Helen Cross was in Clarivate's annual 'Highly Cited Researchers List' which recognises authors of the most influential research papers around the world.

Topics of 2022/23 highest impact papers cover:

- Developing a machine-learning algorithm for automated detection of focal cortical dysplasia.
- The application of a multi-layered diagnostic scheme to better understand and treat focal cortical dysplasia
- Recommendations that epilepsy surgery is offered as soon as drug resistance is ascertained.
- The importance of using specialist multidisciplinary analysis of whole genome sequencing to enhance the diagnostic equity of complex disorders.
- Novel epilepsy phenotypes and the efficacy of potential therapies.
- The incidence of new seizures or epilepsy diagnoses in the 6 months after a COVID-19 diagnosis.

Importance of PPI

How do we ensure research is developed to reflect the real needs of clinicians and patients?

Our Research collaboration with GOSH and ICH ensures that the focus of our research is developed from clinical needs. As part of the this, we consider the views of all individuals who are affected by research. This not only includes the researchers and consultants, but also nurses, support workers, caregivers, parents and crucially the young people themselves. The practice of involving patients in research is called Patient and Public Involvement (PPI) and it is critical in the development of practical, relevant research. Young Epilepsy's E-CURe (Epilepsy Carers Uniting with Researchers) network exists to strengthen the voice of children and young people with epilepsy in our research. We currently have over 140 members but would welcome more. Please see page 48 for details on how to get involved.

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Patient and public involvement in research is essential to ensure research outcomes make meaningful differences in patients' lives. Young Epilepsy ensures that research is holistic to represent all of a patients needs including; school, behaviour, relationships, and treatments for seizures and co-morbidities. They ensure the patient and carers voices are heard through their E-CURe network which unites researchers with carers to understand how their projects may best benefit children with epilepsy. This work is essential as research priorities may be different to the priorities of young people and their carers'. For example, a researcher may want to develop a new type of anti-seizure medication, however, on discussion with patients and families it may be found that a similar anti-seizure medication works but has intolerable side effects and therefore, researching how to modify the toxicity profile of the drug may be more beneficial than a new drug. Equally we know that 30% of patients have intractable seizures. We need to work together as a community to understand where the similarities and differences are within the specific epilepsies associated with intractable seizures and then to path a therapeutic research strategy forwards to help as many patients as possible. This needs to be in the context of understanding meaningful seizure reduction for an individual person.

There are exciting research developments currently underway. Precision medicines and gene therapies are up and coming therapeutic options for some types of epilepsy and we need to ensure that these treatments are funded and there is a mechanism of equitable delivery within the NHS. We also need to ensure there are equitable diagnostic services across the country. Many patients are living with idiopathic epilepsy. We need to ensure that every patient is able to access the multitude of specialist

tests to gain a diagnosis, and if afterwards a diagnosis is not reached then we hope continued research into this population will reveal the answers to the underlying cause.

I have seen first-hand how essential Young Epilepsy values the views of the community particularly the young reps, who are aged 16-25 and are living with epilepsy. The young reps ensure that young people are at the centre of everything Young Epilepsy does. At Young Epilepsy's joint research event with Epilepsy Research UK the young reps introduced each speaker and one bravely and eloquently described the impact of epilepsy on his mental health. They were all incredibly impactful speakers and ensured that the audiences focus was on the patient at every step of the meeting. I was invited to discuss the impact of a rare epilepsy diagnosis from a parent's perspective. Again, this highlights Young Epilepsy's focus on what is important to the community. I was able to describe the emotions and thoughts behind receiving this diagnosis for my daughter but also, the huge amount of work involved to help understand her condition and develop a therapeutic for her and others with rare epilepsy. I encouraged our research community to work together and to work with patient advocacy groups to accelerate the path to improved knowledge, care and treatments.

This joined up approach is being supported by the UK Rare Epilepsies Together (UKRET) organisation which Young Epilepsy is a member. Many of the rare epilepsies priorities and paths towards improved treatments are the same; by joining together we can share knowledge, undertake group projects and achieve more. Young Epilepsy's support to UKRET is vital due to their wealth of experience from working with young people with epilepsy and researchers for many years which provide guidance and oversight which will help UKRET achieve its goals.

As a parent and part of the Young Epilepsy community I am thankful for Young Epilepsy for all that they do to provide resources, care and research. I thank them for listening to people with epilepsy and their carers to ensure that their needs are prioritised.

Dr Stephanie Prince
Parent of a young person with epilepsy, member of the E-CURe Network & board member of Coalition to Cure CHD2

Top 10 Epilepsy Research Priorities

As we have previously reported, Young Epilepsy were honoured to be part of the UK Epilepsy Priority Setting Partnership (PSP) in partnership with Epilepsy Research UK.

The UK PSP were tasked with investigating the health priorities of people with Epilepsy. A survey, completed by 2,014 individuals, identified approximately 5,418 research priorities. From these 110 research questions were drafted, of which 57 were moved forward for prioritisation. 25 of these were shortlisted for discussion at the UK Epilepsy PSP workshop, with the aim of selecting the top 10 priorities for Epilepsy research. The selected Top 10 Epilepsy Research Priorities were:

01 What are the causes and contributing factors of epilepsy-related deaths, including Sudden Unexpected Death in Epilepsy (SUDEP), and how can these deaths be prevented?

02 What underlying mechanisms cause epilepsy in children and adults?

03 What impact do epilepsy, seizures and anti-seizure medication (ASMs) have on brain health – including, cognition, memory, learning, behaviour, and mental health?

04 How does epilepsy and epilepsy treatment impact neurodevelopment, and can this be managed or prevented?

05 How can targeted, personalised medicine, such as gene therapy, be used to treat and/or prevent epilepsy?

06 How can tools, devices and biological markers be used to accurately predict and prevent seizures and the onset of epilepsy?

07 How do hormonal changes in women throughout the lifespan (e.g., puberty, pregnancy, menopause) impact epilepsy, and how can this impact be addressed?

08 How can quality of life be improved for people with epilepsy, their family and carers, including those bereaved by epilepsy?

09 What causes drug-resistant (refractory) epilepsy, and how can it be best treated?

10 How can big data analysis, through artificial intelligence (AI) and machine learning, aid the diagnosis and management of epilepsy?

Creating clearly defined research priorities with input from the entire epilepsy community, allows future research to concentrate on the research areas that matter most.

With this in mind, we have mapped our current studies by their project number onto each of the ten priorities (a project can address more than one priority). At present we are addressing all but two of the priorities, and the majority of our work is focussed on priorities 2, 3, 4, 6. This is unsurprising given that our work is focussed on paediatrics and, in particular, understanding and treating childhood epilepsies.

Retreat 2023

Young Epilepsy Paediatric Epilepsy Research Retreat 2023

The Young Epilepsy Research Retreat, hosted by The Prince of Wales's Chair of Childhood Epilepsy, is an annual gathering of researchers and collaborators across our research unit. This meeting gives researchers the opportunity to share ongoing and/or completed projects, as well as discuss and explore future directions of research. The uniqueness of the event enables researchers, to engage with a rich and diverse network of colleagues from a wide range of backgrounds and thereby benefit from each other's experience and expertise.

Last year, after two years of holding it virtually, we were able to return to its in-person format which was a huge success with 128 attendees over the two days. The schedule was packed with 29 high-quality presentations covering various topics from within our research portfolio. There was an excellent array of presentations and the discussions at the end of each presentation gave investigators the opportunity to receive comments and feedback from fellow researchers and principal investigators representing many different fields.

Our Moderator

Professor Jo Wilmshurst

2023 marked our 13th Retreat and Professor Cross presided over the event for the two days. Alongside Professor Cross we were delighted to welcome as our Moderator, Professor Jo Wilmshurst. Professor Jo Wilmshurst is Head of Paediatric Neurology at the Red Cross War Memorial Children's Hospital, University of Cape Town, in South Africa. She is past-President of the International Child Neurology Association (2018-2022). She is also a member of various Commissions and Task Forces for the International League Against Epilepsy, as well as Chair of the African Commission (2021-2025).

"

I was so impressed by the diversity of research, the cross-discipline activity, the progression in work where one project was clearly evolving to the next stage and most excitingly for me the clinical translation of truly high-level research that would have impact on clinical care but also demonstrated potential reach to the resource limited regions where I practice.

Professor Wilmshurst

Research Events

Rare Epilepsies Event

On the 9th March, Young Epilepsy organised and hosted the 'Better Understanding the Rare Epilepsies' conference at its Lingfield campus, with 53 attendees.

The event was born from discussions between Young Epilepsy and other members of the UK Rare Epilepsisis Together (UKRET) network. Formed in 2021, the UKRET consists of over 25 UK-based patient associations, aiming to work together to effect change for those impacted by rare and complex epilepsies in the UK. The day featured a keynote address from Professor Helen Cross, as well as an update from Dr Amy McTague on her work on Sunflower Syndrome. Allison Watson also introduced the UKRET network, laying out its aims, current activities, and future plans. Following this, we were delighted to welcome two parents and a young person who spoke to their lived experience of the rare epilepsies. As well as the presentations, we had seven other charity exhibitors at the event, all of which are part of the UKRET network.

Overall, we have received some outstanding feedback, with delegates commenting on how informative and valuable they found the event. With this in mind, we plan to instate this as a bi-annual meeting.

Joint Research Event – Epilepsy Research UK

The event, which took place on May 16th, to further share the impact and importance of research into the epilepsies, was a huge success with over 150 attendees.

We have received some excellent feedback from delegates, who have identified the involvement of those with lived experience of epilepsy (i.e., the young reps and the mother of a child with epilepsy) as one of their key standout moments. The announcement of the Epilepsy Research Institute, of which Young Epilepsy will be a founding partner, was also very well received both at the event and on wider channels, such as social media. Every delegate that provided feedback on the event, stated they would be interested in attending something similar in the future. Given such positive responses, we have already begun planning for next year!

Epilepsy in Schools Webinar

On the 14th November, Young Epilepsy were delighted to host the Epilepsy in School webinar, for which we had nearly 800 registered attendees, around 300 of whom worked in educational settings.

The Epilepsy in School Webinar was born from the 'What I Need in School' research project, which crucially asked young people, their parents or caregivers, and teachers what they feel children with epilepsy actually need in school. In keeping with this, as well as hearing about the projects' findings, other speakers included a young person with epilepsy, the parent of a child with epilepsy, and the Assistant Head of St Pier's School. We also hosted speakers from Policy and Advocacy, Youth Development and Support, Health Services, and researchers from the Mental Health in Epilepsy (MICE) project. Overall, the event brought together various essential insights, informing attendees on how we can better support children with epilepsy in school and how Young Epilepsy can assist on the journey to do so. If you want to catch up on the webinar you can watch the recording here:

Physical Activity in Childhood Epilepsy Webinar

The Physical Activity in Childhood Epilepsy (PACE) Webinar, which took place on the 3rd October was a huge success with just over 250 registered attendees.

The webinar began with two presentations from researchers on the PACE project. Following this we heard from individuals with lived experience of epilepsy. The first of these was Jay Bothroyd, a former professional footballer who spoke about his experience of epilepsy in professional sport. We then heard from two members for the Youth Voice Network, Renell and Beth, who spoke about their experiences of physical activity and epilepsy. The webinar concluded with a Q/A session, where useful and informative discussions were held amongst the speakers. Again, if you want to catch up on the webinar you can watch it here:

Research Funding

Central to the research programme is the ability to apply for and manage research grants and other charitable donations.

Our collaborative funding strategy has enabled us to build the world's largest paediatric epilepsy research unit and network of multidisciplinary practitioners.

Alongside academic grants raised by the researchers and their academic institutions, we rely on the additional multidisciplinary fundraising by Young Epilepsy, which allow us to redirect funds where the need is greatest within a project. This flexibility is vital and provides stability during challenges, such as delays due to unforeseen circumstances.

The future of this programme rests on the ability to maintain and build the current infrastructure which allows us to maintain a base of operations to lead, coordinate and provide governance.

We remain ever grateful for the generosity and dedication of the organisations and individuals who support out work.

Action Medical Research
Anna Mueller Grocholski Foundation
Autistica
Brain Tumour Charity
BRC Cambridge
Cancer Research UK
Child Health Research Trust
Children with Cancer UK
D'Oyly Carte Charitable Trust
Desitin
Epilepsy Research UK
Ethypharm
Evelyn Trust
George E Neville Foundation
GOSH NIHR BRC
GOSH-CC
Great Ormond Street Children's Charity
Horizon Medical
The Hospital Saturday Fund
Humnan Brain Project
Innovate UK
James Lewis Foundation
Jazz Pharma
Medical Research Council Clinician Scientist Fellowship
National Institute of Health Research (HTA)
Nevilles PLC
Neuraxpharm
Nutricia
The Oakdale Trust
Oakgrove Foundation
Rosetree's Trust
Persyst
PI EANS
Proveca
PTC Therapeutics
Takeda
UCB Pharma
Waterloo Foundation
Wyfold Charity Trust
Young Epilepsy

Researchers

The research team contribute to a wide spectrum of activities from basic science to patient care. The team consists of a multidisciplinary range of experts working across Young Epilepsy, UCL GOS - Institute of Child Health and Great Ormond Street Hospital for Children.

Principal Investigators

Professor Helen Cross OBE The Prince of Wales's Chair of Childhood Epilepsy and Director UCL GOS - ICH

Young Epilepsy; UCL GOS - Institute of Child Health and Great Ormond Street Hospital for Children

Additional Roles:

Elected President (2021-2025) International League Against Epilepsy (ILAE)

Chair Research Council - European Reference Network for Rare and Complex Epilepsies (EpiCARE)

President - Epilepsy Research UK

Clinical Advisor - Children's Epilepsy Surgery Service (CESS)

Clinical Advisor - Epilepsy Action

Chair of Medical Board - Hope for Hypothalamic Hamartoma

Chair of Medical Board - Matthew's Friends

Chair of the Medical Board - Dravet UK

Associate editor Brain Communications

Editorial Board Epilepsy Research,

Chair, C4C neuroscience expert group

Professor Torsten Baldeweg Professor of Developmental Cognitive Neuroscience, Head of UCL GOS - ICH Developmental Neurosciences Programme

UCL GOS - Institute of Child Health

Additional Roles:

Theme Lead – Capacity Building, Epilepsy Research Institute UK

Chairman of Exam Board, MSc Paediatric Neuropsychology - University College London

Module organiser and lecturer, MSc Paediatric Neuropsychology - University College London

Professor Gareth Barnes Head of Magnetoencephalography

Wellcome Centre for Neuroimaging

Professor Chris Clark Professor of Imaging and Biophysics, Head of UCL GOS – ICH Developmental Imaging and Biophysics Section

UCL GOS - Institute of Child Health

Professor Michelle De Haan Professor in Infant and Child Development

UUCL GOS - Institute of Child Health and Great Ormond Street Hospital for Children

Additional Roles:

Affiliated Scientist - British Autism Study of Infant Siblings Network

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Editor in Chief - Journal of Neuropathology and Applied Neurobiology

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Victoria Bryant *Sudden Unexpected Death in Childhood; characteristics, autopsy findings and investigation*

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Rosie Coleman *Functional and structural plasticity after epilepsy surgery*

Maria Eriksson *Cognitive outcomes after neurosurgical treatment for focal epilepsy: developing a neuroanatomical predictive model for clinical decision making*

Amy Fairchild *Characterisation of high-risk paediatric brain tumours and their aberrant gene networks*

Xiyu Feng *Functional brain connectomics: implications for post-surgical outcomes in children with focal epilepsy*

Robert Flynn *Timings and origin of Hypoxic Ischaemic Encephalopathy in the low- and middle-income countries*

Anna Keegan (IfWH) *AAV9 mediated gene therapy for pyruvate dehydrogenase deficiency*

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Jane Kung *Epilepsy in infancy – relating phenotype to genotype*

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Ulrich Stoof *Multiscale modelling of epileptic networks from SEEG recordings*

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Research Publications

Primary Research

1. Aikaterini Vezyroglou, Akilapa, R., Barwick, K., Saskia Koene, Brownstein, C. A., Holder-Espinasse, M., … Carr, L. (2022). **The Phenotypic Continuum of ATP1A3-Related Disorders**. *Neurology, 99*(14), e1511–e1526. https://doi.org/10.1212/wnl.0000000000200927

2. Anca-Mihaela Vasilica, Winsor, A., Chari, A., Scott, R. C., Torsten Baldeweg, & Tisdall, M. (2023). **The influence of disease course and surgery on quality of life in children with focal cortical dysplasia and long-term epilepsy-associated tumours: A systematic review and meta-analysis**. *Epilepsy Research, 192*, 107132–107132. https://doi.org/10.1016/j.eplepsyres.2023.107132

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5. Bishop, K. I., Isquith, P. K., Gioia, G. A., Knupp, K. G., Scheffer, I. E., Nabbout, R., … & Gammaitoni, A. R. (2023). **Fenfluramine treatment is associated with improvement in everyday executive function in preschool-aged children (< 5 years) with Dravet syndrome.** *Epilepsy & Behavior, 138*, 108994. https://doi.org/10.1016/j.yebeh.2022.108994

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8. Burrows, D. R. W., Diana, G., Pimpel, B., Moeller, F., Richardson, M. P., Bassett, D. S., Meyer, M. P., & Rosch, R. E. (2023). **Microscale Neuronal Activity Collectively Drives Chaotic and Inflexible Dynamics at the Macroscale in Seizures**. *Journal of Neuroscience, 43*(18), 3259–3283. https://doi.org/10.1523/JNEUROSCI.0171-22.2023

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10. Carroll, J. H., Cross, J. H., Hickson, M., Williams, E., Aldridge, V., & Collinson, A. (2023). **A core outcome set for childhood epilepsy treated with ketogenic diet therapy (CORE-KDT study): International parent and health professional consensus. Epilepsia**. https://doi.org/10.1111/epi.17513

11. Carroll, J. H., Martin-McGill, K. J., Cross, J. H., Hickson, M., Williams, E., Aldridge, V., & Collinson, A. (2022). **Core outcome set development for childhood epilepsy treated with ketogenic diet therapy: Results of a scoping review and parent interviews**. *Seizure, 99*, 54–67. https://doi.org/10.1016/j.seizure.2022.05.009

12. Castro-Villablanca, F., Moeller, F., Suresh Pujar, D'Arco, F., Scott, R. C., Muhammad Zubair Tahir, Tisdall, M., J. Helen Cross, & Eltze, C. (2022). **Seizure outcome determinants in children after surgery for single unilateral lesions on magnetic resonance imaging: Role of preoperative ictal and interictal electroencephalography**. *Epilepsia, 63*(12), 3168–3179. https://doi.org/10.1111/epi.17425

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16. Dugan, P., Carroll, E., Thorpe, J., Jette, N., Agarwal, P., Ashby, S., … & Agarwal, P. (2022). **Impact of the COVID-19 pandemic on people with epilepsy: Findings from the US arm of the COV-E study**. *Epilepsia Open, 7*(4), 645-656. https://doi.org/10.1002/epi4.12637

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18. Eriksson, M. H., Ripart, M., Piper, R. J., Moeller, F., Das, K. B., Eltze, C., Cooray, G., Booth, J., Whitaker, K. J., Chari, A., Patricia Martin Sanfilippo, Ana Perez Caballero, Menzies, L., McTague, A., Tisdall, M. M., J. Helen Cross, Torsten Baldeweg, Adler, S., & Konrad Wagstyl. (2023). **Predicting seizure outcome after epilepsy surgery: do we need more complex models, larger samples, or better data?** *Epilepsia*. https://doi.org/10.1111/epi.17637

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21. Hillebrand, A., Holmes, N., Ndedi Sijsma, O'Neill, G., Tierney, T. M., Niels Liberton, Stam, A. H., Nicole van Klink, Stam, C. J., Bowtell, R., Brookes, M. J., & Barnes, G. R. (2023). **Non-invasive measurements of ictal and interictal epileptiform activity using optically pumped magnetometers**. *Scientific Reports, 13*(1). https://doi.org/10.1038/s41598-023-31111-y

22. Mensah, J., Johnson, K., Reilly, C. A., Wilcox, K. S., Rower, J. E., & Metcalf, C. S. (2022). **Evaluating the efficacy of prototype antiseizure drugs using a preclinical pharmacokinetic approach**. *Epilepsia, 63*(11), 2937–2948. https://doi.org/10.1111/epi.17402

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24. Johnson, E. C., Atkinson, P., Muggeridge, A., Cross, J. H.**, & Reilly, C. (2023). Knowledge about, and attitudes towards epilepsy among school staff: A UK-based survey**. *Epilepsy Research, 107116*. https://doi.org/10.1016/j.eplepsyres.2023.107116

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27. Keng Siang Lee, Seunarine, K. K., Barnes, N. P., Muhammad Zubair Tahir, Varadkar, S., & Tisdall, M. (2023). **Accuracy of robot-assisted stereotactic MRI–guided laser ablation in children with epilepsy**. *Journal of Neurosurgery*, 1–9. https://doi.org/10.3171/2023.4.peds2318

28. Krantz, M., Malm, E., Darin, N., Sofou, K., Savvidou, A., Reilly, C., & Boström, P. (2022). **Parental experiences of having a child with CLN3 disease (juvenile Batten disease) and how these experiences relate to family resilience. Child: Care, Health and Development**. https://doi.org/10.1111/cch.12993

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32. McTague, A., Brunklaus, A., Barcia, G., Varadkar, S., Zuberi, S. M., Chatron, N., Parrini, E., Mei, D., Nabbout, R., & Gaëtan Lesca. (2022). **Defining causal variants in rare epilepsies: an essential team effort between biomedical scientists, geneticists and epileptologists**. *European Journal of Medical Genetics, 65*(7), 104531–104531. https://doi.org/10.1016/j.ejmg.2022.104531

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Young
Epilepsy

Reviews or communications of expert opinion

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Books or Chapters in Books

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Epilepsy in childhood:
Carers
Uniting with
REsearchers
...join the conversation"

We Need You

to help us strengthen the voice of children and young people with epilepsy and their families in research.

Following the launch of the first network of young people with epilepsy and their parents, whose sole purpose is to consult on the development of research projects across our partnership, we are keen to continue to grow. If you would like your experiences to ensure epilepsy research is answering the right questions in the right way, please get in contact and sign up to the E-CURe network by scanning the QR code below.

Glossary

Animal models
A non-human species used in medical research because it can mimic aspects of a disease found in humans

Assays
An investigative procedure in laboratory medicine for measuring the presence, amount, or functional activity of a target entity

Biophysical
Methods used in physics to study biological phenomena

Calcium imaging
A technique to optically measure the calcium levels in a cell or tissue

Chronic
Long term

Co-morbidities
Medical conditions that are simultaneously present in a patient

Computational modelling
A mathematical model to study the behaviour of a complex system by computer simulation

Copy number variants
When the number of copies of a particular gene varies between individuals

Cortical
Relating to the outer layer of the uppermost part of the brain

Cox regression
A statistical test

Cryogenic
The production of, and behaviour of, materials at very low temperature

Dietetics
Branch of knowledge concerned with the diet and its effects on health

Electroencephalography (EEG)
A test that detects electrical activity in your brain using small electrodes attached to your scalp. Your brain cells communicate via electrical impulses and activity shows up as wavy lines on an EEG recording

Epidemiological
The branch of medicine which deals with the incidence, distribution, and control of diseases

Epilepsy-dyskinesia
Disorders characterised by recurrent episodes of abnormal movements, co-occurring with epilepsy or other episodic neurological symptoms

Epileptiform discharges
Seen on an EEG, meaning spikes, polyspikes, sharp waves, or spike and slow-wave complexes without observed clinical seizures

Epileptogenesis
The gradual process by which a normal brain develops epilepsy or, the area of epileptogenesis is the area of the brain which causes a patient's epilepsy

Functional validation (of disease-causing genes)
The process of determining whether a particular genetic mutation is causing a disease

Genomics
The study of whole genomes of organisms, and incorporates elements from genetics

Genotype
An organism's set of heritable genes that can be passed down from parents to offspring

Health economics
The study and understanding of how society allocates resources to healthcare and the resource needs of specific healthcare issues

Hemiparesis
Weakness of one entire side of the body

Immunofluorescence
A method in biology that relies on the use of antibodies chemically labelled with fluorescent dyes to visualise molecules under a light microscope

Intractable
Untreatable, hard to manage

Language lateralisation
The phenomenon in which one hemisphere (typically the left) shows greater involvement in language functions than the other

Lesion
A region in an organ or tissue that is abnormal from injury or disease

Magnetoencephalography (MEG)
Functional neuroimaging technique for mapping brain activity by recording magnetic fields produced by electrical currents occurring naturally in the brain

Memory lateralisation
The phenomenon in which one hemisphere (typically the left) shows greater involvement in memory functions than the other

Miss-sense mutation
A point mutation in a gene in which a single nucleotide change results in a codon that codes for a different amino acid

Multi-omic
Or *integrative omics,* is a biological analysis approach in which the data sets are multiple "omes", such as the genome, proteome, transcriptome, epigenome, metabolome, and microbiome

Myoclonia
A form of epileptic seizure manifesting with jerks of the muscles

Natural history
The progression of a disease process in an individual over time, in the absence of treatment

Optically pumped magnetometers (OPM)-MEG
A new type of MEG instrumentation, promising several advantages compared with conventional scanners: higher signal sensitivity, better spatial resolution, more uniform coverage, lifespan compliance, free movement of participants during scanning, and lower system complexity.

Pancytopenia
A condition that occurs when a person has low counts for all three types of blood cells: red blood cells, white blood cells, and platelets

Pathophysiological mechanisms
The cause of a disease associated injury

Phenotype
An individual's observable traits, such as height, eye colour, and blood type. The genetic contribution to the phenotype is called the genotype

PPI
Patient and public involvement

Practice paper
Evaluative summaries of scientific and evidence-based information that address practice topics. Practice papers are often done in emerging areas that might not have sound scientific data yet

Putative variants
A segment of DNA that is believed to be a gene. Putative genes can share sequence similarities to already characterised genes and thus can be inferred to share a similar function, yet the exact function of putative genes remains unknown

Sanger sequencing
A method for determining the nucleotide sequence of DNA

Status epilepticus
A single seizure lasting more than five minutes or two or more seizures within a five-minute period without the person returning to normal between them

Structural correlates
Structural anomalies which correlate to symptoms

Targeted treatment
Treatments which target specific symptoms and potential causes of disease. These treatments are disease modifying

Therapeutic radiofrequency thermocoagulation
A technique of controlled thermal ablation of tissues

Trio whole genome sequencing (WGS)
Whole exome sequencing is a comprehensive method for analysing entire genomes. Trio whole exome sequencing refers to the sequencing of the entire genome of a patient and their biological parents

Western blotting
A widely used analytical technique in molecular biology and immunogenetics to detect specific proteins in a sample of tissue extract

Young Epilepsy, the children and young people's epilepsy charity

We exist to create a society where children and young people with epilepsy are enabled to thrive and fulfil their potential. A society in which their voices are respected and their ambitions realised.

together we create possible.

For more information on our research, or to get involved please contact:

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