UCL
GREAT ORMOND STREET INSTITUTE OF CHILD HEALTH

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NHS Foundation Trust

THE NATIONAL CENTRE FOR YOUNG PEOPLE WITH EPILEPSY CHARITABLE TRUST

Paediatric Epilepsy Research and Impact Report

2024

Young Epilepsy

Contents

Introduction

I am delighted to present our Annual Research Report for the period July 2023 to June 2024, showcasing the achievements of the paediatric epilepsy research unit across Young Epilepsy, UCL GOS – Institute of Child Health and Great Ormond Street Hospital for Children.

2024 has been a fantastic year for our research programme, marked by significant achievements and exciting new milestones. During this period, we conducted 46 active projects, launched 8 new studies and successfully completed 3. Highlights among the new projects include investigations into the role of DNA methylation in epilepsy, the development of zebrafish models to study genetic disorders linked to vitamin B6 metabolism and a pioneering initiative combining multi-scale imaging and genomics to explore neurodevelopmental disorders and epilepsy. Additionally, we initiated a project which examines physical activity in primary school-aged children with epilepsy, launched a pilot of Acceptance Commitment Therapy (ACT) for children and young people with epilepsy and began a natural history study of SCN1A related epilepsies in the UK. These projects span a wide array of topics, demonstrating our commitment to advancing knowledge and improving outcomes across all aspects of childhood epilepsy.

The impact of our efforts is evident in this year's publication record. Between July 2023 and June 2024, we produced 86 peer-reviewed research articles, 27 reviews and expert commentaries, and a book chapter. Five of these publications ranked among the top 5% of all research papers published globally, underscoring their significance and quality. Notable publications include the findings of the Mental Health in Childhood Epilepsy (MICE) project, a multi-centre randomised controlled trial evaluating psychological therapy for children and young people with epilepsy; the Gene-STEPS project, which demonstrated the diagnostic utility of rapid genome sequencing for infantile epilepsy and innovative research introducing advanced scanning techniques for patients with epilepsy.

Another standout event from the year was the 14th Paediatric Epilepsy Research Retreat in January 2024, moderated by epileptologist and child neurologist, Professor Stéphane Auvin from Robert Debré University Hospital and Université de Paris. The retreat brings early-career and established researchers together to share insights and foster collaboration, with attendees particularly valuing the opportunity to network and exchange ideas within this multidisciplinary forum.

We also hosted several other impactful events, including a Joint Research Event with the Epilepsy Research Institute UK, attended by over 150 participants. This event featured inspiring talks on topics such as fostering a sense of community within epilepsy and supporting the mental health of children and young people. As a proud founding member of the Epilepsy Research Institute, we were also excited to learn about their recent advancements. Events such as this demonstrate the vital role of collaboration between charities, beneficiaries and supporters in driving meaningful change and improving the lives of those affected by epilepsy.

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 collaborative research programme exists to establish successively better outcomes by driving early diagnosis and intervention in every aspect of childhood epilepsy. I do hope you enjoy reading this report and feel inspired by the progress made in improving the lives of children and young people with epilepsy.

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' Chair of Childhood Epilepsy is jointly grant funded by GOSH charity and Young 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 hosts 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.

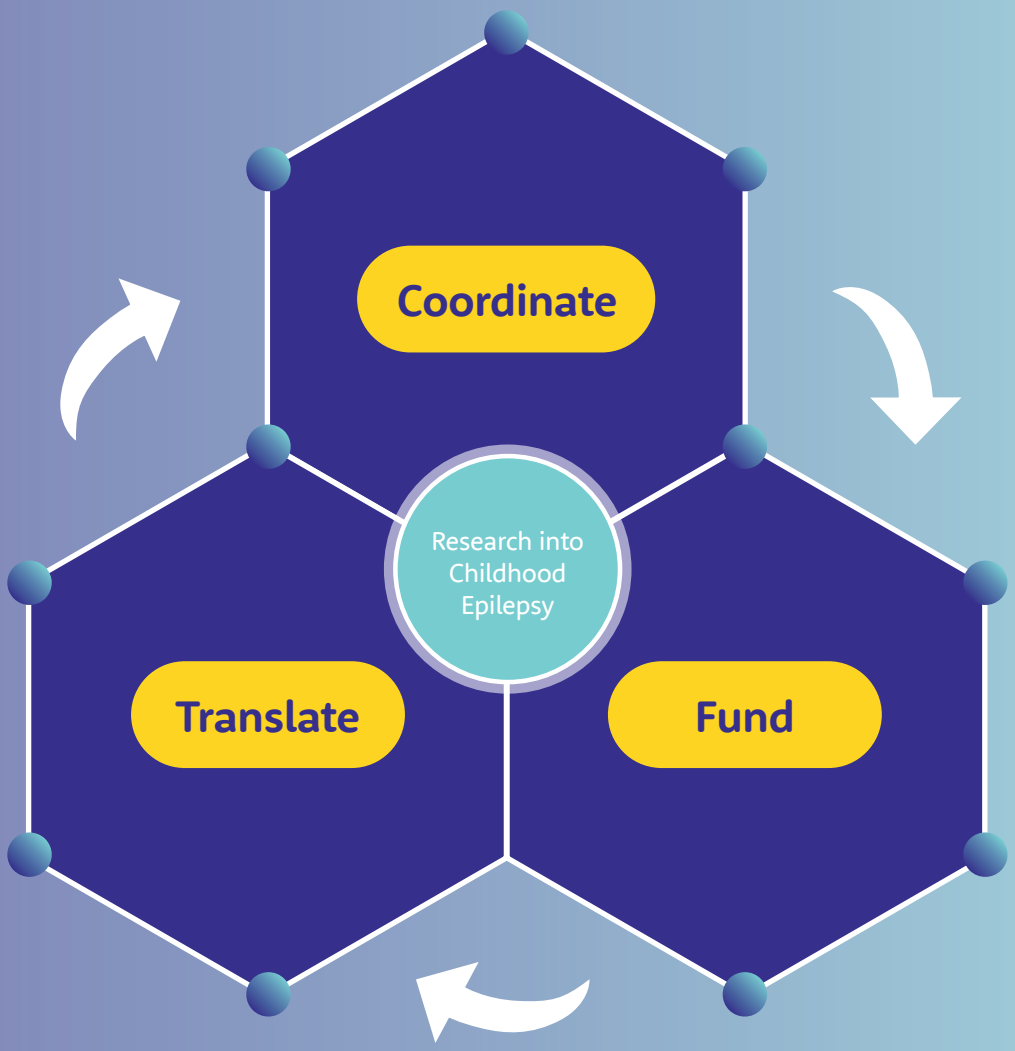
The inspirational mission of the UCL Great Ormond Street Institute of Child Health is to "improve the health and well-being of children, and the adults they will become, through world-class research, education and public engagement".

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...

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



Animal Welfare Policy

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.

Research Strategy & Goals

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

27% projects currently contribute to this goal

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

GOAL 02

Gain a better understanding of how epilepsy affects development and behaviour

19% projects currently contribute to this goal

Epilepsy is associated with a myriad of 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

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

28% projects currently contribute to this goal

The longer an individual 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

Research Strategy & Goals

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

5% projects currently contribute to this goal

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

GOAL 05

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

19% projects currently contribute to this goal

Childhood epilepsy can affect the whole family and treatment must involve multiple disciplines and agencies. Support for families must be evidenced and 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

GOAL 06

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

2% projects currently contribute to this goal

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

Research Projects

Young Epilepsy

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 2023 to June 2024.

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

Key Projects

Optically Pumped Magnetometer Magnetoencephalography (OPM-MEG) at Young Epilepsy

2024 has been a big year for the Diagnostic Suite, with the wearable optically pumped magnetometer magnetoencephalography (OPM-MEG) study entitled *Determining the utility of OPM-MEG in a clinical context commencing recruitment.*

The study aims to show how well the OPM-MEG can map epileptic foci in children with epilepsy. It has two main arms:

- ✓ **Mapping epileptic activity in paediatric presurgical candidates with focal epilepsy**
- ✓ **Collecting a bank of OPM-MEG data in children and young people presenting for diagnostic electroencephalography (EEG)**

Data from the OPM-MEG scan will be directly compared to previous clinical EEG and other neuroimaging data, as well as concurrent research EEG for a subset of participants. The study leverages the depth of expertise across the diagnostic suite, spanning clinical EEG, research and clinical MEG and cognitive neuroscience.

Collecting concurrent EEG in a subset of these participants allows for the closest comparison to current clinical standards (EEG/cryogenic MEG) and helps to inform how this data can be interpreted for clinical adoption in the future. During the scan, participants can relax and watch TV or engage in tasks designed to map eloquent cortices, including motor, visual, and auditory functions. By examining brain activity during these behavioural tasks, the scan not only captures epileptic activity but also identifies critical functions, such as language and movement, that must be preserved if surgery is considered. This approach enhances the likelihood of improved post-surgical outcomes.

While OPM-MEG technology is still at the research stage, this work will offer ground-breaking opportunities to advance medical sciences across epilepsy and other childhood neurological disorders. The technology offers increased accessibility and compliance for children with epilepsy, particularly relative to cryogenic MEG, by providing a flexible fit with sensors closer to the head. Once clinically approved, OPM-MEG will enable earlier assessments for epilepsy surgery, improving seizure control through more accurate pre-surgical evaluations. Successful surgeries could also lead to developmental and cognitive gains, delivering better outcomes for patients, families and society.

Our collaborations have expanded to include several Children's Epilepsy Surgery Service (CESS) centres, such as Bristol Children's Hospital and King's College Hospital, alongside our established partnership with Great Ormond Street Hospital. We are also working with regional NHS trusts, including Maidstone and Tunbridge Wells, Surrey and Sussex Hospitals, and University Hospitals Sussex. These collaborations ensure a large, diverse recruitment pool while strengthening the science through input from clinicians and researchers dedicated to improving outcomes for children with epilepsy. We welcome partnerships and collaborations to increase recruitment and are open to building new studies and grants focussed on neurological conditions impacting children. Initial findings from the first participants were shared at the MEG UKI conference in Birmingham this October. This international gathering of MEG specialists brings together physicists, engineers, and neuroscientists to advance the measurement of magnetic signals from the human brain. This community is highly productive and generates hundreds of high-profile publications annually across the field, and the meeting provides a kickstart to new collaborations and ideas. We will also be presenting data at our annual research retreat and in upcoming meetings next year as our recruitment continues.

If you're interested in exploring how OPM-MEG can contribute to functional neuroimaging in your area of expertise, please get in touch as we would be delighted to discuss how our interests and expertise may align.

research@youngepilepsy.org.uk

Medicinal Cannabis in Refractory Epilepsies

After several years in development, approval was given for funding through the NIHR for two clinical trials assessing efficacy and safety of cannabinoid products in the treatment of epilepsy.

For full details see:

https://www.fundingawards.nihr.ac.uk/award/NIHR131309

Chief Investigators:

Professor Finbar O'Callaghan & Professor Helen Cross

Co-Investigators:

Dr Anita Devlin, Dr Hakim-Moulay, Dr Sallie Baxendale, Ms Felicia ikeji, Ms Gemma Jones, Professor Anthony Marson, Professor Dyfrig Hughes, Professor Martin Kirkpatrick, Professor Nick Freemantle, Professor Sanjay Sisodiya

Background:

Although anti-seizure medications (ASMs) successfully control epilepsy in around 70% of patients, approximately 30% will have seizures that are resistant to current treatments.

Historically there has been much interest in the use of cannabinoids for the treatment of the epilepsies. Recently there has been public and media interest in their use particularly in children with drug resistant seizures and learning impairment. The Cannabis plant produces in excess of 140 phyto-cannabinoids but almost all clinical research to date has focused on two: cannabidiol (CBD) and tetrahydrocannabinol (THC). There have been randomised controlled trials (RCTs) investigating the use of a pure CBD product (Epidyolex ®) in three rare conditions associated with difficult epilepsies: Dravet Syndrome, Lennox Gastaut Syndrome, and tuberous sclerosis complex. These trials have suggested benefit but there have been no RCTs in the broader group of children or adults with intractable epilepsy. Moreover, there have been no RCTs to determine the efficacy of products containing both CBD and THC or studies addressing the possibility of any cognitive or behavioural effect of chronic exposure to THC.

Aims and objectives:

The primary question being addressed by this study is whether CBD, and CBD with THC, reduce convulsive seizure frequency in a range of epilepsies more effectively than placebo, in the absence of any effect on neuropsychological function. Specifically, the study will:

- Investigate whether CBD and CBD+THC are effective in reducing seizures in a broad range of epilepsies and across a broad range of seizure types
- Investigate whether CBD and CBD+THC have any impact on cognition, quality of life, levels of behaviour and anxiety, sleep quality and parental stress
- Estimate the cost-effectiveness of CBD and CBD+THC from the perspective of the National Health Service and Personal Social Services

Methods:

Patients at multiple NHS sites across the UK will be randomly assigned either CBD, a combination of CBD and THC, or a matched placebo. The first trial will investigate cannabis-based medicines for adults and children with epilepsies that start in the first five years of life. The second trial will look at patients with genetic generalised epilepsies (GGE) that have not responded to standard treatment. Patients with GGE usually do not have learning disabilities associated with their epilepsy so this trial will allow researchers to look at the effects of cannabis-based medicines on learning and memory.

After a baseline period, patients will be randomised to one of three treatment arms and reviewed for 24 weeks. Frequency of seizures will be assessed over a baseline period of 8 weeks and again between 16 and 24 weeks of treatment. Cognitive and/or behavioural assessments will be conducted at baseline and 24 weeks. A total of 240 patients are required in each study, with a total of 480 patients recruited across the UK over 18 months. Recruitment of patients for the trials is expected to begin in 15 months, once regulatory and safety approvals have been granted.

What this means:

These studies will answer specific questions as to whether CBD and CBD with THC are safe, effective and cost effective in the treatment of seizures in a range of epilepsies. Data from the trial could, in future be used to enable products to be licensed by the UK medicines regulatory, the MHRA and then commissioned by the NHS, if shown to be clinically and cost effective.

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 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)

02 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

03 Is there an episignature in the rare epilepsies?

Project Aim: To understand the role of DNA methylation in rare epilepsies.
Investigators: Amy McTague, Manju Kurian

04 Understanding the role of vitamin B6 dyshomeostasis in epilepsy disorders

Project Aim: Establish and characterise zebrafish models of different genetic disorders of vitamin B6 metabolism associated with early onset epilepsy.
Investigators: Karin Tuschl, Philippa Mills, Richard Rosch, Isaac Bianco, Stephen Wilson

05 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

06 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

07 Transforming neurodevelopmental disorders using multi scale imaging and genomics

Project Aims:

- Develop computational tools to identify individual subject-level imaging abnormalities in neurodevelopmental disorders.
- Create a multiscale genetic, cellular and imaging framework for understanding the common and diverging neurobiological causes of epilepsy and ASD.
- Test the potential of these tools for linking genetics, imaging and phenotypes with known mutations in genes associated with epilepsy and autism.

Investigators: Konrad Adler-Wagstyl, Sophie Adler-Wagstyl, Helen Cross, Finbar O'Callaghan, Amy McTague, Andreas Brunklaus, Armin Raznahan, Juan Eugenio Iglesias.

08 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

09 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

10 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

11 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

12 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

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, Lara Carr, and Helen Cross

18 SCN1A Horizon's: A natural history study of SCN1A related epilepsies in the UK

Project Aim: The SCN1A Horizons natural history study will establish a national UK platform for long term data collection on assessment and therapy of up to 400 child and adult patients with a genetically confirmed SCN1A variant. This study will increase our understanding of Dravet syndrome and SCN1A-related epilepsies by allowing us to learn more about the seizures, learning abilities and behavioural difficulties that children and adults with an SCN1A-related epilepsy live with.
Investigators: Andreas Brunklaus, Helen Cross, Amy McTague, Michael Absoud

Current projects

Workstream 2: Outstanding Treatment

19 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, Shan Tang, Dr Simon Richardson, Serena Counsell, Alex Hammers, Jonathan O'Muircheartagh

20 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

21 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, David Carmichael, Torsten Baldeweg

22 Comprehensive neuroimaging characterization of neurodegeneration and brain plasticity in children with Rasmussen Syndrome

Project aim: The primary objective of the project is to identify predictors of successful cognitive recovery after surgical treatment.
Investigators: Torsten Baldeweg, Suresh Pujar, Patricia Sanfilippo, Marios Kaliakatsos

23 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

24 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

25 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, Caroline Scott, Lara Carr, Dominic Sims, Elena Boto, Matt Brookes, Helen Cross

26 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

27 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

28 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

29 Improved diagnosis and monitoring of treatment for patients with epilepsy caused by mutations in ALDH7A1

Project Aim: To work out the most reliable compound for detection of ALDH7A1-deficiency which could be used for newborn screening.
Investigators: Philippa Mills, Emma Footitt, Helen Aitkenhead, Peter Clayton, Alistair Horman, Youssef Khalil

30 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

31 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

32 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

33 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, Rory Piper

34 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]

35 Management of seizures in patients with primary mitochondrial diseases: consensus statement from the InterERNs Mitochondrial Working Group

Project Aim: We aim to develop guidelines and consensus recommendations on safe medication use and seizure management in mitochondrial epilepsy using Delphi methodology.
Investigators: Michelangelo Mancuso, Maria T Papadopoulou; Yi Shiau Ng; Anna Ardissone; Marcello Bellusci; Enrico Bertini; Lidia Di Vito; Teresinha Evangelista; Carmen Fons; Omar Hikmat; Rita Horvath; Thomas Klopstock; Cornelia Kornblum; Costanza Lamperti; Laura Licchetta; Maria Judit Molnar; Kristin N Varhaug; Mar O'Callaghan; Ronit M Pressler; Manuel Schiff; Serenella Servidei; Nora Szabo; Gráinne S Gorman; Helen J Cross; Shamima Rahman

Current projects

Workstream 3: Outstanding Support

36 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

37 Epilepsy Pathway Innovation in Africa (EPInA)

Project 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

38 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.

39 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.

40 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

41 Physical Activity in Childhood Epilepsy (PACE) Prime

Project Aim:

- To compare levels of physical activity in primary 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, Natalie Pearson, Lauren Sherar, Monica Lakhanpaul, Kerry Robinson, Lara Carr and Helen Cross

42 Acceptance & Commitment Therapy (ACT) in Children and Young People with epilepsy

Project Aims: To develop, deliver and evaluate pilot ACT intervention groups to improve mental health support for young people with epilepsy living in the South of England.
Investigators: Natasha Hughes, Emily Rhidian, Lara Carr, Ingram Wright, Alexander Marsh

43 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

44 Functional effects of SCN1A mutations – New insights from biophysics and computational modellinga

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

45 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

46 Multiscale modelling of epileptic networks from SEEG recordings

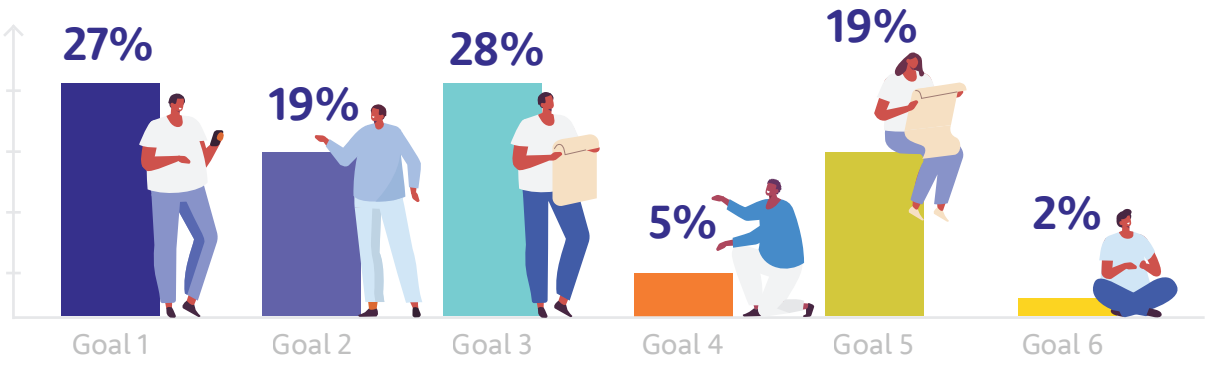
Project Aim: Develop analysis techniques that allow us to understand how changes in brain networks in patients with drug-resistant epilepsy undergoing epilepsy surgery result in the patients' epilepsy.
Investigators: Richard Rosch, Ulrich Stoof, James Wilsenach, Aswin Chari, Martin Tisdall, Gerald Cooray,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

Impact of our research

Young Epilepsy

Current and Past Impact

Between July 2023 and June 2024 the programme portfolio consisted of

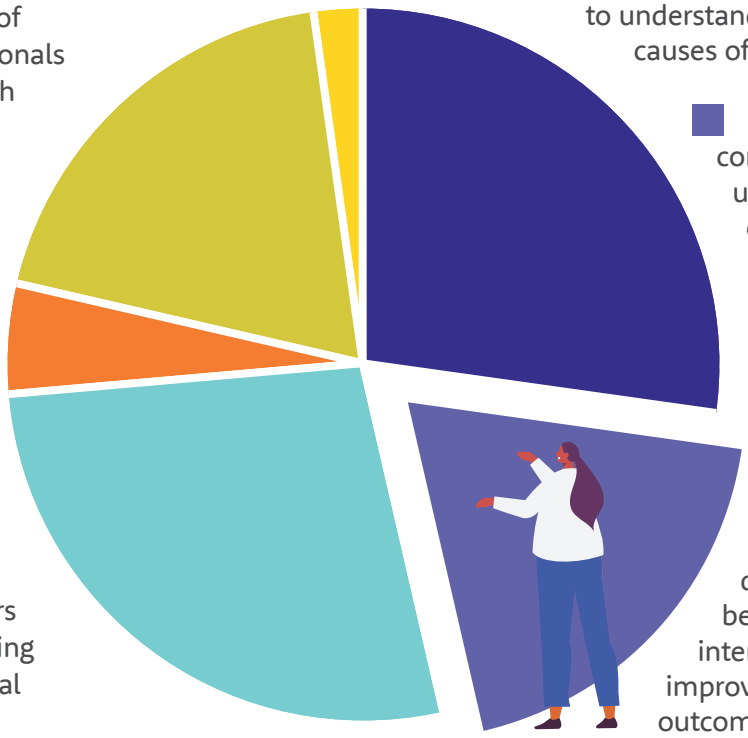


contributing to each goal respectively

2% of projects contributed to developing a network of multidisciplinary professionals to strengthen our research and shape the education of future practitioners

19% of projects contributed to making life better for children and families and making support systems more effective

5% of projects contributed to understanding the barriers to learning and determining the benefits of educational interventions



27% of projects contributed to understanding the medical causes of epilepsy

19% of projects contributed to understanding how epilepsy affects development and behaviour

28% of projects contributed to improving diagnosis and treatment to determine the benefits of early intervention in improving long-term outcomes

We are currently involved in the development of:

2 Genetic Therapy Treatments

1 Antisense Oligonucleotide's for Dravet Syndrome (ASO; Stoke therapeutics)

Admiral initial safety and tolerabilty and Longwing continuation

1 ASO for Angelmans, Ultragenyx_GTX

86 published
peer-reviewed items of primary research

27 reviews and
1 further chapter
in books

Specialist PPI network for childhood epilepsy with over **195 members.**

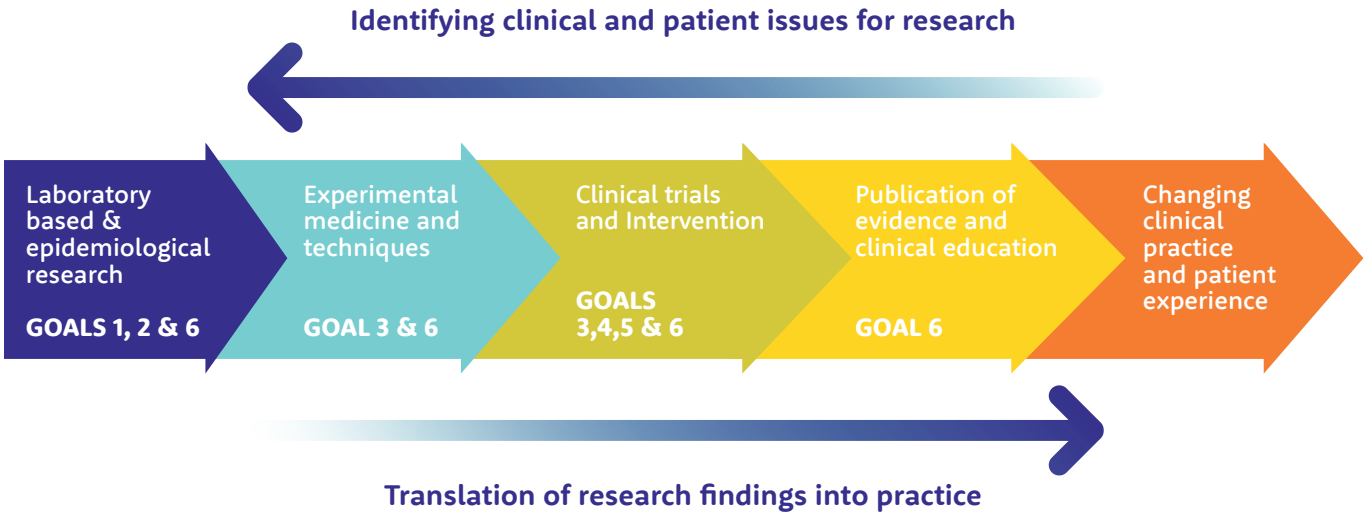
Nearly **800 attendees** across 3 events. The Annual Research Retreat had almost **100 attendees** and our Joint Event with the Epilepsy Research Institute had **150**.

Our webinar attracted almost **500 registered attendees!**

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.



There was a total of 46 active research projects in the 2023/24 year (Figure 1), a slight increase in the number of total projects from last year. This coupled with the addition of new initiatives and successful completion of others underscores our commitment to advancing knowledge and driving meaningful impact. We track the contribution of active projects to the three workstreams and six strategic goals (Figure 2). Historically our strengths lie in Workstream 1 and 2: Understanding Childhood Epilepsies and Outstanding Treatments (Figure 3). At the same time, we are actively expanding our focus on Workstream 3: Outstanding Support, encouraging educational, psychosocial, and service-based research to enhance support for individuals and families living with epilepsy.

Figure 1: Number of active research projects per year

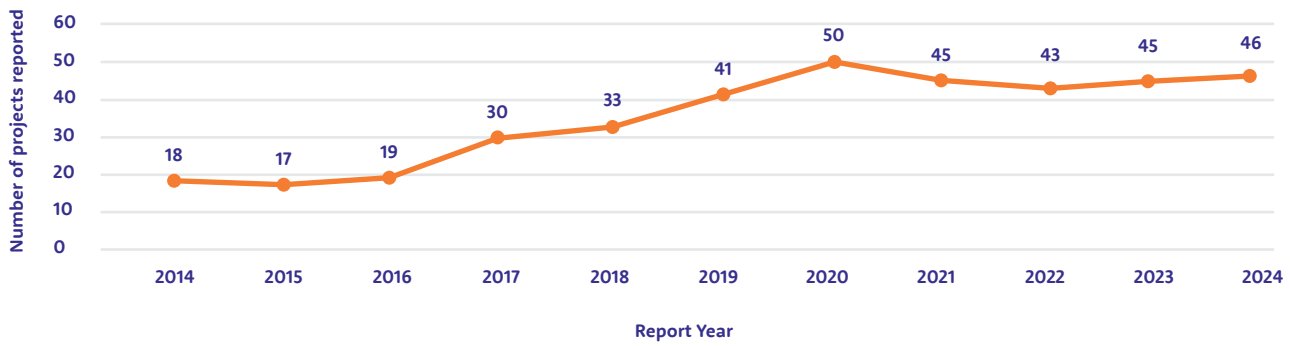
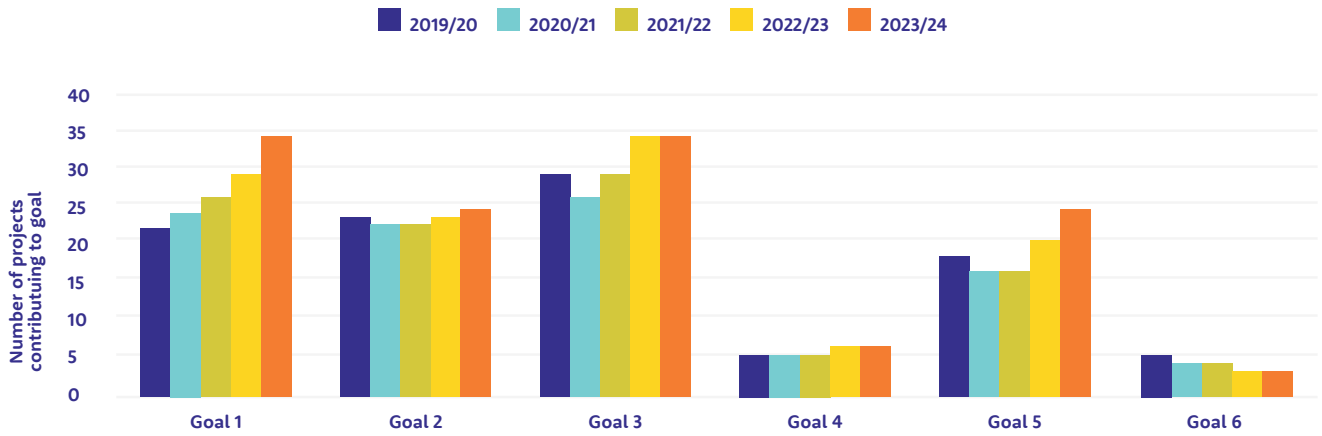


Figure 2: Number of active projects contributing to each goal

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



The strength of the evidence we publish

Over the past 10 years, our research programme has grown from having just four Principal Investigators (the leaders of research units and laboratories, often Professors), to having 32 Principal Investigators supervising 32 PhD students and working alongside an additional 30 international collaborating researchers (Figure 4).

2024 has marked a significant increase in the publication of original research (Figure 5), signalling a strong recovery in research output to pre-COVID-19 levels. While there has been a slight decline in published reviews and expert opinion pieces, the overall number of publications has risen. This upward trend, coupled with the increasing number of active projects, suggests that our research output will continue to expand in the years ahead.

Figure 4: Annual growth of the Research Unit Network

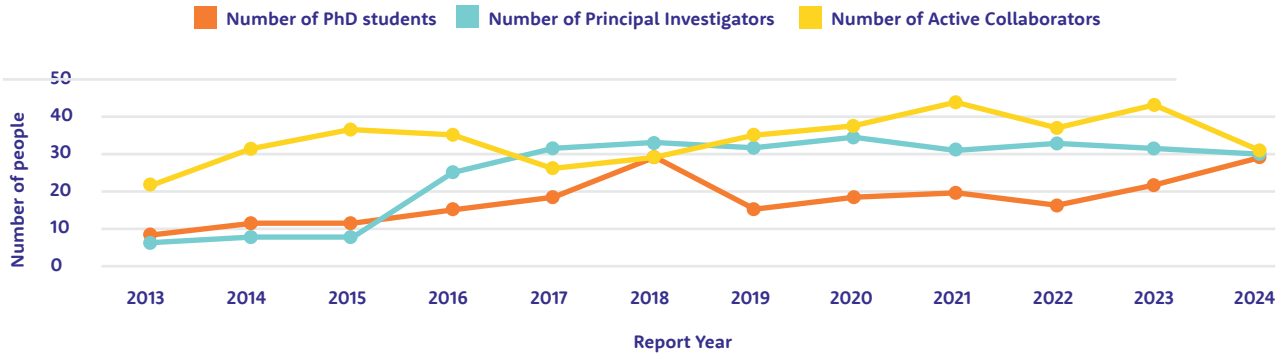
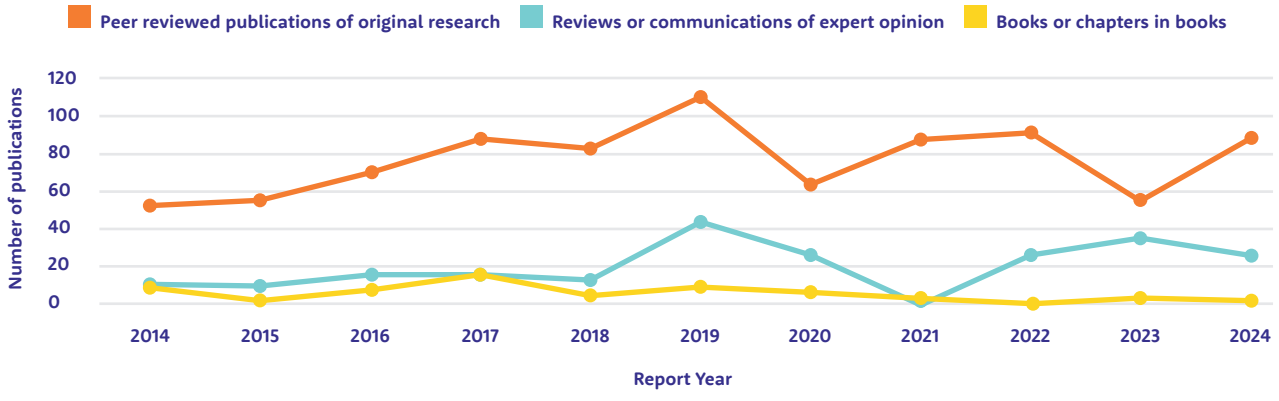
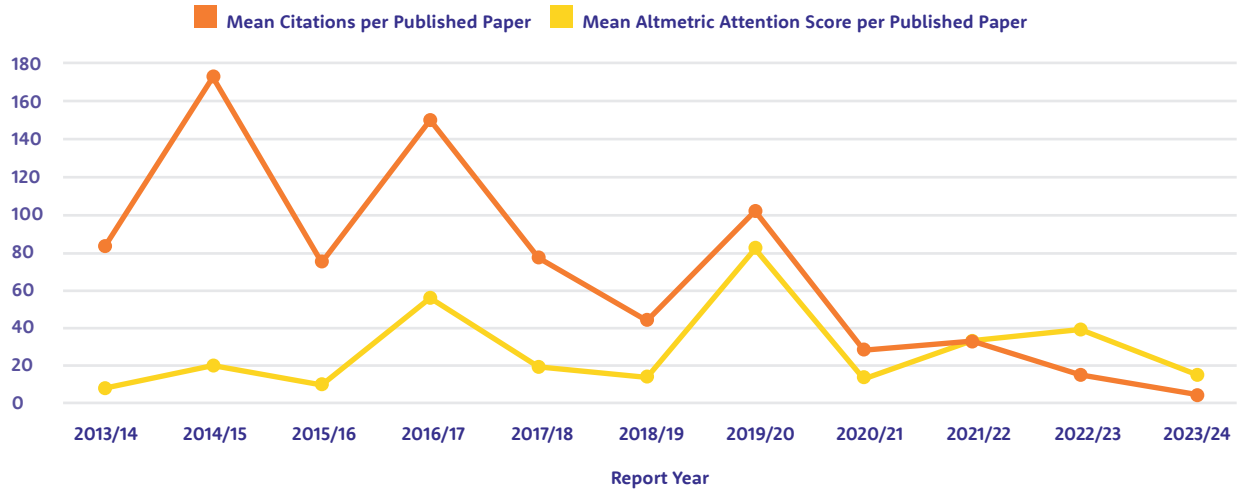


Figure 5: Number of research publications produced per year



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



This year's publications have been cited less frequently than those from previous years, which is expected given that citations typically accumulate over time. Additionally, there has been a decrease in the average Altmetric score compared to 2022/23, suggesting that recent publications may have attracted less immediate attention (Figure 6). Notably, last year's highest-scoring papers focused on genetics and COVID-19—topics with broad, global relevance. In contrast, research specific to epilepsy often requires more time to garner attention and citations.

Despite this year's slight decline, we achieved strong impact, with five papers receiving an Altmetric score of over 100, placing them in the top 5% of all research outputs tracked by ReadCube. This is comparable to last year, which saw six such high-scoring publications.

In 2024, for the 5th year, Professor Helen Cross was in Clarivate's annual 'Highly Cited Researchers List' which recognises authors of the most influential research papers around the world. One of 74 at UCL overall.

Topics of the 2023/24 high impact papers cover:

- A multi-centre randomised controlled trial reporting on the effectiveness of a psychological therapy for children and young people with epilepsy (Mental Health Intervention in Childhood Epilepsy; MICE).
- A pilot and feasibility study investigating transcranial electrical stimulation during functional magnetic resonance imaging in patients with genetic generalised epilepsy.
- Evaluation of the feasibility, diagnostic yield, and clinical utility of rapid genome sequencing in infantile epilepsy (Gene-STEPS).
- The development of a system to concurrently measure spinal and brain signals with optically pumped magnetometers.
- An evaluation of the pharmacokinetics, safety, and tolerability of brivaracetam in neonates with repeated electroencephalographic seizures not controlled with previous antiseizure medications.

Importance of PPI

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. The E-CURe (Epilepsy Carers Uniting with Researchers) network connects parents and caregivers of children with epilepsy and dedicated researchers, ensuring their voices can shape the future of epilepsy reserach. We currently have 195 members but would welcome more. Please see page 48 for details on how to get involved.

I have been involved with Young Epilepsy for the last 15 years and it is great to see how it has evolved into such a vital organisation with national reach. Whether working with young people themselves or with their families, Young Epilepsy offers a variety of much needed support services and now, through the E-Cure Network, the opportunity to collaborate on pioneering research projects. Since being part of E-Cure, I have been impressed by the huge range of studies young people and their families can take part in and the many opportunities to shape research from the earliest design phases, to ensure outcomes are meaningful and impactful.

Emma Dalrymple
Parent of a young person with epilepsy

Being involved in the coproduction workshops has been very insightful. Listening and discussing with other carers going through similar situations, has made me realise that there is an absence of adequate mental and additional support for both children and parents during this overwhelming time. In the three years since my daughter's diagnosis there has been very little in terms of help to assist us in understanding epilepsy, other than a few brochures posted to us at the start, and that needs to change in future.

This workshop gives the parents a voice, as they know more than anyone what help is missing and what is needed to get them through this difficult time. Hopefully the suggestions in the meetings are listened to and possibly implemented in the future.

My daughter was diagnosed with epilepsy when she was 13 years old. I've been desperate to find out more about epilepsy ever since and jumped at the chance to get involved in Young Epilepsy's Parenting Plus co-production group. Around six of us meet in this online group to share our thoughts about what support we think other carers would find useful. I find it's a constant battle to navigate each particular challenge my daughter faces, so it's great to work on something that will give parents and carers more help and support.

Emma Campbell
Parent of a young person with epilepsy

It is hugely important to me to be able to contribute to the coproduction workshops and share experiences of parenting a child with epilepsy as it seems to be the only way to have an impact and make change that will benefit others and make the journey less challenging.

Working with the other parents is really interesting as no child has identical seizures/co-morbidities, but there are so many similarities and connections that are very different to parenting children with other medical conditions.

It is a relief to speak with people who understand the condition and we and the researchers all have the same goal which is to make things better than they currently are.

It is important for me to share my daughter's voice as she is often not in a position to share herself due to a variety of reasons. Whilst I can only try to imagine what it feels like to live with epilepsy, no one really knows apart from the person who has the condition, so their voice is the most crucial.

We also work very closely with our Youth Voice Network to ensure that the voices of children and young people are represented in our research. The Youth Voice Network is a group of over 100 young people with epilepsy between the ages of 13-25, who help to ensure that young people are at the centre of everything we do at Young Epilepsy. To find out more and get involved:

www.youngepilepsy.org.uk/get-involved/give-time/be-member-youth-voice-network

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.

Young Epilepsy Paediatric Epilepsy Research Retreat 2024

The Young Epilepsy Research Retreat, led by The Prince of Wales's Chair of Childhood Epilepsy, is an annual event that brings together researchers and collaborators from across our research unit. This unique gathering provides a platform for sharing ongoing and completed projects, fostering discussions, and exploring future research directions.

In 2024, we proudly hosted our 14th Research Retreat, welcoming nearly 100 attendees over two days. The programme featured 28 exceptional presentations spanning topics across our research portfolio, including highlights such as the Turning Six study and the Mental Health in Childhood Epilepsy project. Each presentation was followed by engaging discussions, offering presenters valuable feedback from peers and principal investigators representing diverse fields of expertise.

The event was chaired by Professor Helen Cross, with Professor Stéphane Auvin, a renowned epileptologist and child neurologist from Robert Debré University Hospital and Université de Paris, serving as our distinguished moderator.

The retreat was a resounding success, with attendees particularly appreciating the opportunity to network and exchange ideas within this multidisciplinary forum. The feedback received underscores the event's value in fostering collaboration and advancing our collective research impact.

Research Events

Joint Research Event - Epilepsy Research Institute

In May 2024, we partnered with the Epilepsy Research Institute to co-host a Joint Research Event at the renowned Francis Crick Institute. The event brought together over 150 attendees, who heard a series of inspiring talks on topics such as fostering community within epilepsy and supporting the mental health of children and young people.

As a proud founding member of the Epilepsy Research Institute, we were also excited to learn about their groundbreaking advancements, which hold the potential to significantly accelerate progress in epilepsy research.

The event underscored the vital role of collaboration between charities, beneficiaries, and supporters in driving meaningful change and improving the lives of those affected by epilepsy

Ketogenic Diet in Infants with Epilepsy (KIWE) Webinar

The Ketogenic Diet in Infants with Epilepsy (KIWE) Webinar, which took place on the 17th April was a huge success with just under 500 attendees.

The webinar began with Professor Helen Cross presenting the history of the ketogenic diet in paediatric epilepsy, followed by Dr Natasha Schoeler summarising the KIWE project's findings. Rai Royal, a parent of an infant with epilepsy, shared their firsthand experience of participating in the KIWE trial and finally we heard from Julie Fountain, the CEO of Matthews Friends, who reflected on the implications of the KIWE trial and potential future directions. The session concluded with an engaging Q&A, allowing attendees to interact directly with the panel and deepen their understanding of the topics discussed

www.youtube.com/watch?v=VbAnpt3W-go&feature=youtu.be

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.

Action Medical Research
Angelini Pharma
Anna Mueller Grocholski Foundation
Autistica
Brain Tumour Charity
BRC Cambridge
Cancer Research UK
Child Health Research Charity
Children with Cancer UK
Epilepsy Research Institute
Evelyn Trust
George E Neville Foundation
GOSH NIHR BRC
Great Ormond Street Children's Charity
Horizon Medical
Human Brain Project
Innovate UK
Jazz Pharma
Medical Research Council Clinician Scientist Fellowship
National Institute of Health Research (HTA)
Nevilles PLC
Oakgrove Foundation
PI EANS
Rosetree's Trust
SBRI Healthcare
The Oakdale Trust
Wellcome Trust

We remain ever grateful for the generosity and dedication of the organisations and individuals who support out work. Thank you! To find out how you can get involved in our vital work, visit:

www.youngepilepsy.org.uk/get-involved


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 Institute 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 executive committee
Chair – International Neurological Ketogenic Society

Dr Patricia Atkinson
Consultant Community Paediatrician

Sussex Community NHS Foundation Trust

Dr Sarah Aylett
Consultant Paediatric Neurologist

Great Ormond Street Hospital for Children

Additional Roles:
Caldicott Guardian Postgraduate Teaching - ICH-UCL

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

Department of Imaging Neuroscience, UCL

Dr Stewart Boyd
Consultant Clinical Neurophysiologist

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

Dr David Carmichael
Professor of MRI

School of Biomedical Engineering and Imaging Sciences, King's College London

Additional Roles:
Honorary Reader in Neuroimaging and Biophysics, Reader in Magnetic Resonance Physics
UCL GOS - Institute of Child Health and Wellcome/EPSRC Centre for Medical Engineering, Kings College London

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

UCL GOS - Institute of Child Health

Dr Maria Clark
Consultant Paediatric Neurologist

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

Dr Felice D'Arco
Consultant Paediatric Neuroradiologist

Great Ormond Street Hospital for Children

Additional Roles:
Chair - GOSH MRI Safety Group
Honorary Senior Lecturer - UCL GOS - ICH and UCL Institute of Neurology
Lecturer - European Course of Paediatric Neuroradiology
Member - European Network for Brain Malformations
Member of the Editorial Board - Journal of the European Society of Neuroradiology

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
Course Speaker, MSc in Cognitive Neuroscience, Translational Research Module - University College London
Deputy Director, MSc in Clinical & Applied Paediatric Neuropsychology - UCL GOS - ICH
Director, MSc in Infancy and Early Childhood Development - UCL GOS - ICH
Membership of Steering Committees - Centre for Developmental Cognitive Neuroscience UCL
Membership of Steering Committees - Centre for Developmental Cognitive Neuroscience UCL

Dr Krishna Das
Consultant Paediatric Neurologist

Young Epilepsy and Great Ormond Street Hospital for Children

Professor Michelle De Haan
Professor in Infant and Child Development

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

Additional Roles:
Affiliated Scientist - British Autism Study of Infant Siblings Network

Course Speaker, MSc in Cognitive Neuroscience, Translational Research Module - University College London

Deputy Director, MSc in Clinical & Applied Paediatric Neuropsychology - UCL GOS - ICH

Director, MSc in Infancy and Early Childhood Development - UCL GOS - ICH

Membership of Steering Committees - Centre for Developmental Cognitive Neuroscience UCL

Dr Christin Eltze
Consultant Paediatric Neurologist

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

Professor Isobel Heyman
Consultant Child and Adolescent Psychiatrist

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

Professor Tom Jacques
Professor of Paediatric Neuropathology

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

Additional Roles:
Editor in Chief - Journal of Neuropathology and Applied Neurobiology

Lead - Paediatric Tumour Genomics England Clinical Interpretation Partnership (GeCIP)

Pathology representative on the Central Nervous System subgroup - National Cancer Research Institute (NCRI) Children's Cancer and Leukaemia Clinical Studies Group

Dr Marios Kaliakatsos
Paediatric Neurologist

Great Ormond Street Hospital for Children

Dr Amy McTague
Principal Research Fellow and Honorary Consultant Paediatric Neurologist

UCL GOS - Institute of Child Health

Additional Roles:
Member of work package 2, Laboratory Diagnostics - EpiCARE and Genetic Research work package

Scientific Adviser - KCNT1 Epilepsy Foundation

Member of Epilepsy Research Institute UK Scientific Advisory Committee and member of Advanced Therapeutics Task Force

Professor Philippa Mills
Professor of Inherited Paediatric Metabolic Disease

UCL GOS - Institute of Child Health

Additional Roles:
Course Contributor - UCL GOS - ICH

Treasurer - Society of the Study of Inborn Errors of Metabolism

Dr Friederike Moeller
Consultant Clinical Neurophysiologist

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

Professor Finbar O'Callaghan
Professor of Paediatric Neuroscience, Head of UCL GOS - ICH Clinical Neurosciences Section

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

Additional Roles:
President - British Paediatric Neurology Association (BPNA)

Secretary and Board Member- European Paediatric Neurology Society (EPNS)

Dr Ronit Pressler
Consultant Clinical Neurophysiologist, Great Ormond Street Hospital for Children

Cambridge University Hospital and UCLH

Associate Professor in Clinical Neuroscience

UCL GOS - Institute of Child Health

Additional Roles:
On British Society for Clinical Neurophysiology (BSCN council), current position: president-elect

Council member of the ILAE UK Branch

Course Director, EEG in the First Year of Life teaching course – ILAE

Associated Editor for Epilepsia Open

Member of the Editorial Board of European Journal of Paediatric Neurology and Neurophysiologie Clinique

Member of the ILAE Task Forces: Neonatal seizures and Acute Symptomatic Seizures

Web based teaching: e-brain and VIREPA (paediatric EEG)

Professor Shamima Rahman
Professor of Paediatric Metabolic Medicine

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

Additional Roles:
Adviser to Statutory Approvals Committee - Human Fertilisation and Embryology Authority

Contributor, MSc courses - UCL GOS ICH, UCL Division of Biosciences, UCL Institute of Cardiovascular Science, UCL QS Institute of Neurology and Genomics England

Editor-in-Chief - Journal of Inherited Metabolic Disease

Council Member, Society for the Study of Inborn Errors of Metabolism (SSIEM)

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Pathways and mechanisms affected in individuals with vitamin B6-responsive epilepsy

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

Dominic Burrows
Brain-wide abnormal dynamics during epileptic seizures at single cell resolution

Barbora Cerna
Epilepsy in 3D: investigation of a novel RNA-based therapy in cerebral organoids

Dimitrios Champsas
Improving the understanding of FIRES – febrile infection-related epilepsy syndrome

Zachary Cohen
Non-Invasive Acute Neuromodulation and Measurement of Epileptogenicity

Rosie Coleman
Functional and structural plasticity after epilepsy surgery

Georgia Doumou
Mapping human brain development at new spatial resolutions using Artificial Intelligence and 7T Magnetic Resonance Imaging: Application on Paediatric Epilepsy

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 medicated gene therapy for pyruvate dehydrogenase deficiency

Nandaki Keshavan
Gene Therapy for Deoxyguanosine Kinase Deficiency

Jane Kung
Epilepsy in infancy – relating phenotype to genotype

Mei-Ju Lai *Investigating cellular identity in childhood epilepsy*

Jade Lau
Zebrafish models of vita- min B6-dependent epilepsies

Jyoti Mangal
Developing Ultra-High Resolution Quantitative MRI at 7T for Clinical Applications

Rebecca Meagher
Optimising EEG hardware for use at 7T for effective deployment in simultaneous EEG-fMRI for better epilepsy surgery localisation

Jamie Norris
Variability in epilpetiform activity and the brain's response to stimulation

Jack O'Brien
Cairney Novel models of autoimmune epilepsies Rory Piper Network-guided epilepsy surgery for children

Vaisakh Pulappatta Azhakapath
Timings and origin of Hypoxic Ischaemic Encephalopathy in the low-and-middle income countries

Mathilde Ripart
Automated detection of MRI pathology in epilepsy Flavia Matos Santo The mosaic brain: a new diagnostic approach in focal epilepsies

Oliver Sherwood
Steering epilepsy networks in real time: a multimodal approach

Izabella Smolicz
The biology of paediatric central nervous system tumours at post-mortem

Ulrich Stoof
Multiscale modelling of epileptic networks from SEEG recordings

Aitkaterini Vezyroglou
Deep phenotyping of alternating hemiplegia in childhood

Ella Whittle (SGUL)
Elucidating the genetic background of rare neurological diseases: with a focus on paediatric neurological disorders

Lottie Wood
Comprehensive Neuroimaging characterization of neurodegeneration and brain plasticity in children with Rasmussen Syndrome

Yutong Yao
Modelling epileptogenic networks in concurrent iEEG-fMRI

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Young
Epilepsy

Research Publications

Primary Research

1. Arnhem, M. M. L. van, Munckhof, B. van den, Arzimanoglou, A., Perucca, E., Metsähonkala, L., Rubboli, G., Khinchi, M. S., Saint-Martin, A. de, Klotz, K. A., Jacobs, J., Cross, J. H., Morales, I. G., Otte, W. M., Teeseling, H. C. van, Leijten, F. S. S., Braun, K. P. J., Jansen, F. E., group, R. E. study, Jansen, A., … Ramantani, G. (2024). **Corticosteroids versus clobazam for treatment of children with epileptic encephalopathy with spike-wave activation in sleep (RESCUE ESES): a multicentre randomised controlled trial**. *The Lancet Neurology, 23*(2), 147–156. https://doi.org/10.1016/s1474-4422(23)00409-x

2. Bennett, S. D., Cross, J. H., Chowdhury, K., Ford, T., Heyman, I., Coughtrey, A. E., Dalrymple, E., Byford, S., Chorpita, B., Fonagy, P., Moss-Morris, R., Reilly, C., Smith, J. A., Stephenson, T., Varadkar, S., Blackstone, J., Quartly, H., Hughes, T., Lewins, A., … Shafran, R. (2024). **Clinical effectiveness of the psychological therapy Mental Health Intervention for Children with Epilepsy in addition to usual care compared with assessment-enhanced usual care alone: a multicentre, randomised controlled clinical trial in the UK**. *The Lancet, 403*(10433), 1254–1266. https://doi.org/10.1016/s0140-6736(23)02791-5

3. Bennett, S. D., Rojas, N., Catanzano, M., Roach, A., Ching, B. C., Coughtrey, A. E., Heyman, I., Liang, H., Team, L. P. & Shafran, R. (2023). **Feasibility, acceptability and preliminary effectiveness of a mental health drop-in centre for the siblings of young people attending a paediatric hospital**. *Journal of Child Health Care*, 13674935231206896. https://doi.org/10.1177/13674935231206895

4. Bjurulf, B., Reilly, C. & Hallböök, T. (2024). **Caregiver reported behavior, sleep and quality of life in children with Dravet syndrome: A population-based study**. *Epilepsy & Behavior, 150*, 109560. https://doi.org/10.1016/j.yebeh.2023.109560

5. Bonthrone, A. F., Green, D., Morgan, A. T., Mankad, K., Clark, C. A. & Liégeois, F. J. (2024). **Attention and motor profiles in children with developmental coordination disorder: A neuropsychological and neuroimaging investigation**. *Developmental Medicine & Child Neurology, 66*(3), 362–378. https://doi.org/10.1111/dmcn.15745

6. Burgod, C., Mazlan, M., Pant, S., Krishnan, V., Garegrat, R., Montaldo, P., Muraleedharan, P., Bandiya, P., Kamalaratnam, C. N., Chandramohan, R., Manerkar, S., Jahan, I., Moni, S. C., Shahidullah, M., Rodrigo, R., Sumanasena, S., Sujatha, R., Sathyanathan, B. P., Joshi, A. R., … Thayyil, S. (2024). **Duration of birth depression and neurodevelopmental outcomes after whole-body hypothermia for hypoxic ischemic encephalopathy in India, Sri Lanka and Bangladesh – an exploratory analysis of the HELIX trial**. *The Lancet Regional Health - Southeast Asia*, 20, 100284. https://doi.org/10.1016/j.lansea.2023.100284

7. Carmichael, D. W., Vulliemoz, S., Murta, T., Chaudhary, U., Perani, S., Rodionov, R., Rosa, M. J., Friston, K. J. & Lemieux, L. (2024). **Measurement of the Mapping between Intracranial EEG and fMRI Recordings in the Human Brain**. *Bioengineering, 11*(3), 224. https://doi.org/10.3390/bioengineering11030224

8. Carroll, J. H., Parkin, T., Cross, J. H., Hickson, M., Williams, E., Aldridge, V. & Collinson, A. (2024). **Drug-resistant epilepsy and ketogenic diet therapy – a qualitative study of families' experiences**. *Seizure: European Journal of Epilepsy, 118*, 137–147. https://doi.org/10.1016/j.seizure.2024.04.024

9. Casella, C., Vecchiato, K., Cromb, D., Guo, Y., Winkler, A. M., Hughes, E., Dillon, L., Green, E., Colford, K., Egloff, A., Siddiqui, A., Price, A., Grande, L. C., Wood, T. C., Malik, S., Teixeira, R. P. A. G., Carmichael, D. W. & O'Muircheartaigh, J. (2024). **Widespread, depth-dependent cortical microstructure alterations in pediatric focal epilepsy**. *Epilepsia, 65*(3), 739–752. https://doi.org/10.1111/epi.17861

10. Casey, T., Thachuthara, A. J., Fogarty, L., Livingstone, V., Haan, M. D., Marlow, N., Kiely, M. E. & Murray, D. M. (2023). **Validation of a touchscreen assessment tool to screen for cognitive delay at 24 months**. *Developmental Medicine & Child Neurology, 65*(9), 1206–1214. https://doi.org/10.1111/dmcn.15555

11. Chalia, M., Singh, D., Boyd, S. G., Hannam, S., Hoskote, A. & Pressler, R. (2024). **Neonatal seizures during extra corporeal membrane oxygenation support**. *European Journal of Pediatrics, 183*(6), 2605–2614. https://doi.org/10.1007/s00431-024-05510-w

12. Chari, A., Adler, S., Wagstyl, K., Seunarine, K., Tahir, M. Z., Moeller, F., Thornton, R., Boyd, S., Das, K., Cooray, G., Smith, S., D'Arco, F., Baldeweg, T., Eltze, C., Cross, J. H. & Tisdall, M. M. (2024). **Lesion detection in epilepsy surgery: Lessons from a prospective evaluation of a machine learning algorithm**. *Developmental Medicine & Child Neurology, 66*(2), 216–225. https://doi.org/10.1111/dmcn.15727

13. Cipriani, V., Vestito, L., Magavern, E. F., Jacobsen, J. O., Arno, G., Behr, E. R., Benson, K. A., Bertoli, M., Bockenhauer, D., Bowl, M. R., Burley, K., Chan, L. F., Chinnery, P., Conlon, P., Costa, M., Davidson, A. E., Dawson, S. J., Elhassan, E., Flanagan, S. E., … Smedley, D. (2023). **Rare disease gene association discovery from burden analysis of the 100,000 Genomes Project data**. *medRxiv*, 2023.12.20.23300294. https://doi.org/10.1101/2023.12.20.23300294

14. Clayton, L. M., Azadi, B., Eldred, C., Wilson, G., Robinson, R. & Sisodiya, S. M. (2024). **Feeding Difficulties and Gastrostomy in Dravet Syndrome**. *Neurology: Clinical Practice, 14*(3), e200288. https://doi.org/10.1212/cpj.0000000000200288

15. Cohen, Z., Steinbrenner, M., Piper, R. J., Tangwiriyasakul, C., Richardson, M. P., Sharp, D. J., Violante, I. R. & Carmichael, D. W. (2024). **Transcranial electrical stimulation during functional magnetic resonance imaging in patients with genetic generalized epilepsy: a pilot and feasibility study**. *Frontiers in Neuroscience, 18*, 1354523. https://doi.org/10.3389/fnins.2024.1354523

16. Coughtrey, A. E., Bennett, S. D., Stanick, C., Chorpita, B., Dalrymple, E., Fonagy, P., Cross, J. H., Ford, T., Heyman, I., Moss-Morris, R., Team, M. S. & Shafran, R. (2024). **Training and supervision of physical health professionals to implement mental health care in paediatric epilepsy clinics**. *Epilepsy & Behavior, 157*, 109905. https://doi.org/10.1016/j.yebeh.2024.109905

17. Cross, J. H., Schoeler, N. E., Marston, L., Freemantle, N. & investigators, K. (2024). **Implications of the KIWE trial for low-income and lower-middle-income countries – Authors' reply**. *The Lancet Neurology, 23*(4), 332–333. https://doi.org/10.1016/s1474-4422(24)00080-2

18. Dalen, T. van, Kirkham, J. F., Chari, A., D'Arco, F., Moeller, F., Eltze, C., Cross, J. H., Tisdall, M. M. & Thornton, R. C. (2024). **Characterizing Frontal Lobe Seizure Semiology in Children**. *Annals of Neurology, 95*(6), 1138–1148. https://doi.org/10.1002/ana.26922

19. Louw, E. van der, Trimmel-Schwahofer, P., Devlin, A., Armeno, M., Thompson, L., Cross, J. H., Auvin, S. & Dressler, A. (2024). **Human milk and breastfeeding during ketogenic diet therapy in infants with epilepsy: Clinical practice guideline**. *Developmental Medicine & Child Neurology, 66*(10), 1276–1288. https://doi.org/10.1111/dmcn.1592

20. Defelippe, V. M., Thiel, G. J. M. W. van, Otte, W. M., Schutgens, R. E. G., Stunnenberg, B., Cross, H. J., O'Callaghan, F., Giorgis, V. D., Jansen, F. E., Perucca, E., Brilstra, E. H. & Braun, K. P. J. (2023). **Toward responsible clinical n-of-1 strategies for rare diseases**. *Drug Discovery Today, 28*(10), 103688. https://doi.org/10.1016/j.drudis.2023.103688

21. Dellenmark-Blom, M., Ax, S. Ö., Lilja, H. E., Reilly, C., Svensson, J. F., Kassa, A.-M., Jönsson, L., Abrahamsson, K., Gatzinsky, V., Omling, E., Tollne, A., Stenström, P. & Öst, E. (2023). **Prevalence of Mental Health Problems, Associated Factors, and Health-Related Quality of Life in Children with Long-Gap Esophageal Atresia in Sweden**. *Journal of Pediatric Surgery, 58*(9), 1646–1655. https://doi.org/10.1016/j.jpedsurg.2022.12.004

22. D'Gama, A. M., Mulhern, S., Sheidley, B. R., Boodhoo, F., Buts, S., Chandler, N. J., Cobb, J., Curtis, M., Higginbotham, E. J., Holland, J., Khan, T., Koh, J., Liang, N. S. Y., McRae, L., Nesbitt, S. E., Oby, B. T., Paternoster, B., Patton, A., Rose, G., … McTague, A. (2023). **Evaluation of the feasibility, diagnostic yield, and clinical utility of rapid genome sequencing in infantile epilepsy (Gene-STEPS): an international, multicentre, pilot cohort study**. *The Lancet Neurology, 22*(9), 812–825. https://doi.org/10.1016/s1474-4422(23)00246-6

23. Diodato, D., Schiff, M., Cohen, B. H., Bertini, E., Rahman, S., Bertini, E., Cohen, B. H., Rahman, S., Schiff, M., Ardissone, A., Bertini, E., Christodoulou, J., Cohen, B. H., Falk, M., Goldstein, A., Haas, R., Horvath, R., McFarland, R., Murayama, K., … Yeske, P. (2023). **258th ENMC international workshop Leigh syndrome spectrum: genetic causes, natural history and preparing for clinical trials 25-27 March 2022, Hoofddorp, Amsterdam, The Netherlands**. *Neuromuscular Disorders, 33*(8), 700–709. https://doi.org/10.1016/j.nmd.2023.06.002

24. Dubenko, A., Morelli, R., Cross, J. H., Hall, J., Kharytonov, V., Michaelis, R., Wiebe, S. & Ukraine, the I. E. T. F. for. (2024). **Mental health and quality of life of individuals with epilepsy during the war in Ukraine**. *Epilepsia, 65*(9), 2718–2727. https://doi.org/10.1111/epi.18052

25. El-Dib, M., Abend, N. S., Austin, T., Boylan, G., Chock, V., Cilio, M. R., Greisen, G., Hellström-Westas, L., Lemmers, P., Pellicer, A., Pressler, R. M., Sansevere, A., Tsuchida, T., Vanhatalo, S., Wusthoff, C. J., Bonifacio, S., Wintermark, P., Aly, H., Chang, T., … McCaul, M. C. (2022). **Neuromonitoring in neonatal critical care part I: neonatal encephalopathy and neonates with possible seizures**. *Pediatric Research, 94*(1), 64–73. https://doi.org/10.1038/s41390-022-02393-1

26. Elkhateeb, N., Issa, M. Y., Elbendary, H. M., Elnaggar, W., Ramadan, A., Rafat, K., Kamel, M., Abdel-Ghafar, S. F., Amer, F., Hassaan, H. M., Trunzo, R., Pereira, C., Abdel-Hamid, M. S., D'Arco, F., Bauer, P., Bertoli-Avella, A. M., Girgis, M., Gleeson, J. G., Zaki, M. S. & Selim, L. (2024). **The clinical and genetic landscape of developmental and epileptic encephalopathies in Egyptian children**. *Clinical Genetics, 105*(5), 510–522. https://doi.org/10.1111/cge.14481

27. Eriksson, M. H., Ripart, M., Piper, R. J., Moeller, F., Das, K. B., Eltze, C., Cooray, G., Booth, J., Whitaker, K. J., Chari, A., Sanfilippo, P. M., Caballero, A. P., Menzies, L., McTague, A., Tisdall, M. M., Cross, J. H., Baldeweg, T., Adler, S. & Wagstyl, K. (2023). **Predicting seizure outcome after epilepsy surgery: Do we need more complex models, larger samples, or better data?** *Epilepsia, 64*(8), 2014–2026. https://doi.org/10.1111/epi.17637

28. Eriksson, M. H., Whitaker, K. J., Booth, J., Piper, R. J., Chari, A., Sanfilippo, P. M., Caballero, A. P., Menzies, L., McTague, A., Adler, S., Wagstyl, K., Tisdall, M. M., Cross, J. H. & Baldeweg, T. (2023). **Pediatric epilepsy surgery from 2000 to 2018: Changes in referral and surgical volumes, patient characteristics, genetic testing, and postsurgical outcomes**. *Epilepsia, 64*(9), 2260–2273. https://doi.org/10.1111/epi.17670

29. Fasaludeen, A., McTague, A., Jose, M., Banerjee, M., Sundaram, S., Madhusoodanan, U. K., Radhakrishnan, A. & Menon, R. N. (2024). **Genetic variant interpretation for the neurologist – A pragmatic approach in the next-generation sequencing era in childhood epilepsy**. *Epilepsy Research, 201*, 107341. https://doi.org/10.1016/j.eplepsyres.2024.107341

30. Fleming, J. E., Benjaber, M., Toth, R., Zamora, M., Landin, K., Kavoosi, A., Ottoway, J., Gillbe, T., Piper, R. J., Noone, T., Campbell, H., Gillbe, I., Kaliakatsos, M., Tisdall, M., Valentín, A. & Denison, T. (2023). **An embedded intracranial seizure monitor for objective outcome measurements and rhythm identification**. *2023 45th Annual International Conference of the IEEE Engineering in Medicine & Biology Society (EMBC)*, 00, 1–6. https://doi.org/10.1109/embc40787.2023.10340850

31. Gifford, A., Griffiths, M. J., Rodie, P., Wilmshurst, J., Ball, J., Dunkley, C., McLellan, A., O'Callaghan, F. & Kirkpatrick, M. (2024). **Reducing epilepsy diagnostic and treatment gaps: Standardized paediatric epilepsy training courses for health care professionals**. *Developmental Medicine & Child Neurology, 66*(8), 1045–1052. https://doi.org/10.1111/dmcn.15864

32. Gill, S. K., Rose, H. E. L., Wilson, M., Gutierrez, D. R., Worthington, L., Davies, N. P., MacPherson, L., Hargrave, D. R., Saunders, D. E., Clark, C. A., Payne, G. S., Leach, M. O., Howe, F. A., Auer, D. P., Jaspan, T., Morgan, P. S., Grundy, R. G., Avula, S., Pizer, B., … Peet, A. C. (2024). **Characterisation of paediatric brain tumours by their MRS metabolite profiles**. *NMR in Biomedicine, 37*(5), e5101. https://doi.org/10.1002/nbm.5101

33. Goselink, R. J., Eklund, A., Olsson, I., Hallböök, T., Malmgren, K. & Reilly, C. (2024). **Transitioning from paediatric to adult care in epilepsy: A qualitative study of adolescent experiences**. *Seizure: European Journal of Epilepsy, 119*, 92–97. https://doi.org/10.1016/j.seizure.2024.05.011

34. Gurung, S., Karamched, S., Perocheau, D., Seunarine, K. K., Baldwin, T., Alrashidi, H., Touramanidou, L., Duff, C., Elkhateeb, N., Stepien, K. M., Sharma, R., Morris, A., Hartley, T., Crowther, L., Grunewald, S., Cleary, M., Mundy, H., Chakrapani, A., Batzios, S., … Baruteau, J. (2024). **The incidence of movement disorder increases with age and contrasts with subtle and limited neuroimaging abnormalities in argininosuccinic aciduria**. *Journal of Inherited Metabolic Disease, 47*(6), 1213–1227. https://doi.org/10.1002/jimd.12691

35. Hikmat, O., Naess, K., Engvall, M., Klingenberg, C., Rasmussen, M., Brodtkorb, E., Ostergaard, E., Coo, I. de, Pias-Peleteiro, L., Isohanni, P., Uusimaa, J., Majamaa, K., Kärppä, M., Ortigoza-Escobar, J. D., Tangeraas, T., Berland, S., Harrison, E., Biggs, H., Horvath, R., … Bindoff, L. A. (2024). **Status epilepticus in POLG disease: a large multinational study**. *Journal of Neurology, 271*(8), 5156–5164. https://doi.org/10.1007/s00415-024-12463-5

36. Hill, M., Iro, M., Sadarangani, M., Absoud, M., Cantrell, L., Chong, K., Clark, C., Easton, A., Gray, V., Kneen, R., Lim, M., Liu, X., Pike, M., Solomon, T., Vincent, A., Willis, L., Yu, L.-M., Pollard, A. J., Pollard, A. J., … Collinson, A. (2023). **Intravenous immunoglobulin treatment in childhood encephalitis (IgNiTE): a randomised controlled trial**. *BMJ Open, 13*(11), e072134. https://doi.org/10.1136/bmjopen-2023-072134

37. Hutton, D., Kameda-Smith, M., Afshari, F. T., Elawadly, A., Hogg, F., Mehta, S., Samarasekara, J., Aquilina, K., Jeelani, N. ul O., Tahir, M. Z., Thompson, D., Tisdall, M. M., Silva, A. H. D., Hatcher, J. & James, G. (2023). **Intracranial invasive group A streptococcus: a neurosurgical emergency in children**. *Journal of Neurosurgery: Pediatrics, 32*(4), 1–10. https://doi.org/10.3171/2023.5.peds23109

38. Idowu, J., Pearson, N., Meades, C., Cross, J. H., Muggeridge, A., Lakhanpaul, M., Robinson, K., Sherar, L. B. & Reilly, C. (2024). **Accelerometer and Survey Assessed Physical Activity in Children With Epilepsy: A Case-Controlled Study**. *Pediatric Exercise Science*, 1–9. https://doi.org/10.1123/pes.2023-0065

39. Iro, M. A., Sadarangani, M., Absoud, M., Cantrell, L., Chong, W. K., Clark, C., … & IgNiTE Study Team. (2024). **Intravenous immunoglobulin treatment for encephalitis in children aged 6 months to 16 years: the IgNiTE RCT**. *Efficacy and Mechanism Evaluation, 11*(6).

40. Kameda-Smith, M. M., Mendoza, M., Brown, L.-A., Hartley, J., Aquilina, K., James, G., Jeelani, N. O., Silva, A. H. D., Thompson, D., Tisdall, M., Tahir, M. Z. & Hatcher, J. (2023). **Comparison of endoscopic sinus sampling versus intracranial sampling for microbiological diagnosis of intracranial infection in children: a case series and literature review**. *Child's Nervous System, 39*(12), 3561–3570. https://doi.org/10.1007/s00381-023-06038-4

41. Kariuki, S. M., Wagner, R. G., Gunny, R., D'Arco, F., Kombe, M., Ngugi, A. K., White, S., Odhiambo, R., Cross, J. H., Sander, J. W. & Newton, C. R. J. C. (2024). **Magnetic resonance imaging findings in Kenyans and South Africans with active convulsive epilepsy: An observational study**. *Epilepsia, 65*(1), 165–176. https://doi.org/10.1111/epi.17829

42. King, T., Hui, G. C., Muschialli, L., Shafran, R., Ritchie, B., Hargreaves, D. S., Heyman, I., Griffiths, H. & Bennett, S. (2024). **Mental health interventions for children and young people with long-term health conditions in Children and Young People's Mental Health Services in England**. *Clinical Child Psychology and Psychiatry, 29*(3), 799–819. https://doi.org/10.1177/13591045231216134

43. Knöpfel, N., Zecchin, D., Richardson, H., Polubothu, S., Barberan-Martin, S., Cullup, T., Gholam, K., Heales, S., Krywawych, S., López-Balboa, P., Muwanga-Nanyonjo, N., Ogunbiyi, O., Puvirajasinghe, C., Solman, L., Swarbrick, K., Syed, S. B., Tahir, Z., Tisdall, M. M., Allgrove, J., … Kinsler, V. A. (2024). **GNAQ/GNA11 Mosaicism Is Associated with Abnormal Serum Calcium Indices and Microvascular Neurocalcification**. *Journal of Investigative Dermatology, 144*(4), 820-832.e9. https://doi.org/10.1016/j.jid.2023.09.008

44. Krishnan, V., Ujjanappa, V., Vegda, H., Annayappa, M. K., Wali, P., Fattepur, S., Chandriah, S., Devadas, S., Kariappa, M., Gireeshan, V. K., Thamunni, A. V., Montaldo, P., Burgod, C., Garegrat, R., Muraleedharan, P., Pant, S., Newton, C. R., Cross, J. H., Bassett, P., … Pressler, R. M. (2024). **Sequential levetiracetam and phenytoin in electroencephalographic neonatal seizures unresponsive to phenobarbital: a multicenter prospective observational study in India**. *The Lancet Regional Health - Southeast Asia, 25*, 100371. https://doi.org/10.1016/j.lansea.2024.100371

45. Kristensen, E., Mathisen, L., Berland, S., Klingenberg, C., Brodtkorb, E., Rasmussen, M., Tangeraas, T., Bliksrud, Y. T., Rahman, S., Bindoff, L. A. & Hikmat, O. (2024). **Epidemiology and natural history of POLG disease in Norway: a nationwide cohort study**. *Annals of Clinical and Translational Neurology, 11*(7), 1819–1830. https://doi.org/10.1002/acn3.52088

46. Lara, M. K., Brabec, J. L., Hernan, A. E., Scott, R. C., Tyler, A. L. & Mahoney, J. M. (2024). **Network-based analysis predicts interacting genetic modifiers from a meta-mapping study of spike–wave discharge in mice**. *Genes, Brain and Behavior, 23*(2), e12879. https://doi.org/10.1111/gbb.12879

47. Lee, K. S., Seunarine, K. K., Barnes, N., Tahir, M. Z., Varadkar, S. M. & Tisdall, M. M. (2023). **Accuracy of robot-assisted stereotactic MRI–guided laser ablation in children with epilepsy**. *Journal of Neurosurgery: Pediatrics, 32*(2), 1–9. https://doi.org/10.3171/2023.4.peds2318

48. Lin, H., Figini, M., D'Arco, F., Ogbole, G., Tanno, R., Blumberg, S. B., Ronan, L., Brown, B. J., Carmichael, D. W., Lagunju, I., Cross, J. H., Fernandez-Reyes, D. & Alexander, D. C. (2023). **Low-field magnetic resonance image enhancement via stochastic image quality transfer**. *Medical Image Analysis, 87*, 102807. https://doi.org/10.1016/j.media.2023.102807

49. Macrae, E., Milosavljevic, B., Katus, L., Mason, L., Amadó, M. P., Rozhko, M., Haan, M. de, Elwell, C. E., Moore, S. E., Lloyd-Fox, S. & Team, T. B. P. (2024). **Cognitive control in infancy: Attentional predictors using a tablet-based measure**. *Infancy, 29*(4), 631–655. https://doi.org/10.1111/infa.12599

50. Mancuso, M., Lopriore, P., Lamperti, C., Klopstock, T., Rahman, S., Licchetta, L., Kornblum, C., Wortmann, S. B., Dollfus, H., Papadopoulou, M. T., Arzimanoglou, A., Scarpa, M., Graessner, H. & Evangelista, T. (2024). **Current management of primary mitochondrial disorders in EU countries: the European Reference Networks survey**. *Journal of Neurology, 271*(2), 835–840. https://doi.org/10.1007/s00415-023-12017-1

51. Mancuso, M., Papadopoulou, M. T., Ng, Y. S., Ardissone, A., Bellusci, M., Bertini, E., Vito, L. D., Evangelista, T., Fons, C., Hikmat, O., Horvath, R., Klopstock, T., Kornblum, C., Lamperti, C., Licchetta, L., Molnar, M. J., Varhaug, K. N., O'Callaghan, M., Pressler, R. M., … Rahman, S. (2024). **Management of seizures in patients with primary mitochondrial diseases: consensus statement from the InterERNs Mitochondrial Working Group**. *European Journal of Neurology, 31*(7), e16275. https://doi.org/10.1111/ene.16275

52. Mankel, F. L., Papandreou, A., Mankad, K., Richardson, H. & Aylett, S. E. (2024). **Diagnostic pathway and management of first seizures in infants with Sturge–Weber syndrome**. *Developmental Medicine & Child Neurology*. https://doi.org/10.1111/dmcn.15983

53. Mardell, L. C., Spedden, M. E., O'Neill, G. C., Tierney, T. M., Timms, R. C., Zich, C., Barnes, G. R. & Bestmann, S. (2024). **Concurrent spinal and brain imaging with optically pumped magnetometers**. *Journal of Neuroscience Methods, 406*, 110131. https://doi.org/10.1016/j.jneumeth.2024.110131

54. McCormick, E. M., Keller, K., Taylor, J. P., Coffey, A. J., Shen, L., Krotoski, D., Harding, B., Panel, N. C. U. M. D. G. C. E., Gai, X., Falk, M. J., Zolkipli-Cunningham, Z. & Rahman, S. (2023). **Expert Panel Curation of 113 Primary Mitochondrial Disease Genes for the Leigh Syndrome Spectrum**. *Annals of Neurology, 94*(4), 696–712. https://doi.org/10.1002/ana.26716

55. Meinhardt, A., Reilly, L., Kaliakatsos, M., Abdel-Aziz, K., Alsharidah, S., Bodi, I., Booth, C., Chetty, K., Evans, J., Ferreras-Antolín, L., Froude, S., Galtrey, C. M., Guruprasad, S., Hadden, R. D., Hassell, J., Hyöty, H., Kreins, A. Y., Laiho, J. E., Lowe, D. M., … Kadambari, S. (2024). **Novel antivirals for severe enterovirus infection in immunocompromised hosts; A case series**. *Journal of Infection, 88*(5), 106142. https://doi.org/10.1016/j.jinf.2024.106142

56. Mellor, S., Tierney, T. M., Seymour, R. A., Timms, R. C., O'Neill, G. C., Alexander, N., Spedden, M. E., Payne, H. & Barnes, G. R. (2023). **Real-time, model-based magnetic field correction for moving, wearable MEG**. *NeuroImage, 278*, 120252. https://doi.org/10.1016/j.neuroimage.2023.120252

57. Mellor, S., Timms, R. C., O'Neill, G. C., Tierney, T. M., Spedden, M. E., Consortium, T. M. P., Spitzer, H., Ripart, M., Whitaker, K., Napolitano, A., Palma, L. D., Benedictis, A. D., Foldes, S., Zhang, K., Hu, W., Mo, J., Likeman, M., Davies, S., Güttler, C., … Barnes, G. R. (2024). **Combining OPM and lesion mapping data for epilepsy surgery planning: a simulation study**. *Scientific Reports, 14*(1), 2882. https://doi.org/10.1038/s41598-024-51857-3

58. Mitchell, J. W., Sossi, F., Miller, I., Jaber, P. B., Das-Gupta, Z., Fialho, L. S., Amos, A., Austin, J. K., Badzik, S., Baker, G., Zeev, B. B., Bolton, J., Chaplin, J. E., Cross, J. H., Chan, D., Gericke, C. A., Husain, A. M., Lally, L., Mbugua, S., … Berg, A. T. (2024). **Development of an International Standard Set of Outcomes and Measurement Methods for Routine Practice for Infants, Children, and Adolescents with Epilepsy: The International Consortium for Health Outcomes Measurement Consensus Recommendations**. *Epilepsia, 65*(7), 1938–1961. https://doi.org/10.1111/epi.17976

59. Mitchell, J. W., Sossi, F., Miller, I., Jaber, P. B., Das-Gupta, Z., Fialho, L. S., Amos, A., Austin, J. K., Badzik, S., Baker, G., Zeev, B. B., Bolton, J., Chaplin, J. E., Cross, J. H., Chan, D., Gericke, C. A., Husain, A. M., Lally, L., Mbugua, S., … Berg, A. T. (2024). **Development of an International Standard Set of Outcomes and Measurement Methods for Routine Practice for Adults with Epilepsy: The International Consortium for Health Outcomes Measurement Consensus Recommendations**. *Epilepsia, 65*(7), 1916–1937. https://doi.org/10.1111/epi.17971

60. Montaldo, P., Cirillo, M., Burgod, C., Caredda, E., Ascione, S., Carpentieri, M., Puzone, S., D'Amico, A., Garegrat, R., Lanza, M., Morales, M. M., Atreja, G., Shivamurthappa, V., Kariholu, U., Aladangady, N., Fleming, P., Mathews, A., Palanisami, B., Windrow, J., … Moschella, S. (2024). **Whole-Body Hypothermia vs Targeted Normothermia for Neonates With Mild Encephalopathy**. *JAMA Network Open, 7*(5), e249119. https://doi.org/10.1001/jamanetworkopen.2024.9119

61. Nabbout, R., Arzimanoglou, A., Auvin, S., Berquin, P., Desurkar, A., Fuller, S., D. & Cross., … J. Helen. (2023). **Retrospective chart review study of use of cannabidiol (CBD) independent of concomitant clobazam use in patients with Lennox-Gastaut syndrome or Dravet syndrome**. *Seizure*. https://doi.org/10.1016/j.seizure.2023.05.003

62. Panagi, L., Newlove-Delgado, T., White, S. R., Bennett, S., Heyman, I., Shafran, R. & Ford, T. (2024). **Trends in comorbid physical and mental health conditions in children from 1999 to 2017 in England**. *European Child & Adolescent Psychiatry, 33*(1), 33–38. https://doi.org/10.1007/s00787-022-02112-5

63. Panagi, L., White, S. R., Dai, X., Bennett, S., Shafran, R. & Ford, T. (2024). **Risk of new onset and persistent psychopathology in children with long-term physical health conditions: a population-based cohort study**. *European Child & Adolescent Psychiatry, 33*(2), 439–449. https://doi.org/10.1007/s00787-023-02170-3

64. Panagi, L., White, S. R., Howdle, C., Bennett, S., Heyman, I., Shafran, R. & Ford, T. (2024). **Predictors of contact with services for mental health problems among children with comorbid long-term physical health conditions: a follow-up study**. *European Child & Adolescent Psychiatry, 33*(1), 21–31. https://doi.org/10.1007/s00787-022-02105-4

65. Parikh, N., Faulkner, A., Hadji-Michael, M., Heyman, I., Murphy, T. & McAllister, E. (2024). **Group-based parent training programme for children with neurological conditions: a feasibility study**. *Archives of Disease in Childhood, 109*(2), 138–143. https://doi.org/10.1136/archdischild-2023-326174

66. Pavel, A. M., Rennie, J. M., Vries, L. S. de, Mathieson, S. R., Livingstone, V., Finder, M., Foran, A., Shah, D. K., Pressler, R. M., Weeke, L. C., Dempsey, E. M., Murray, D. M., Boylan, G. B., Consortium, Ans., Pavlidis, E., Kharoshankaya, L., Marnane, L., Lightbody, G., O'Leary, J., … Huffelen, A. C. van. (2024). **Temporal evolution of electrographic seizures in newborn infants with hypoxic-ischaemic encephalopathy requiring therapeutic hypothermia: a secondary analysis of the ANSeR studies**. *The Lancet Child & Adolescent Health, 8*(3), 214–224. https://doi.org/10.1016/s2352-4642(23)00296-1

67. Piper, R. J., Dasgupta, D., Eriksson, M. H., Ripart, M., Moosa, A., Chari, A., Seunarine, K. K., Clark, C. A., Duncan, J. S., Carmichael, D. W., Tisdall, M. M. & Baldeweg, T. (2023). **Extent of piriform cortex resection in children with temporal lobe epilepsy**. *Annals of Clinical and Translational Neurology, 10*(9), 1613–1622. https://doi.org/10.1002/acn3.51852

68. Pressler, R., Boylan, G., Dempsey, E., Klotz, K. A., Krauwinkel, W., Will, E., Morita, D., Floricel, F., Elshoff, J. & Anker, J. van den. (2024). **Pharmacokinetics and safety of brivaracetam in neonates with repeated electroencephalographic seizures: A multicenter, open-label, single-arm study**. *Epilepsia Open, 9*(2), 522–533. https://doi.org/10.1002/epi4.12875

69. Rahimian, E., D'Arco, F., Sudhakar, S., Tahsini, Majid. R., Azin, N., Morovvati, M., Karimzadeh, P. & Farahvash, M. A. (2024). **The full spectrum of MRI findings in 18 patients with Canavan disease: new insights into the areas of selective susceptibility**. *Neuroradiology, 66*(10), 1829–1835. https://doi.org/10.1007/s00234-024-03388-x

70. Ramantani, G., Cserpan, D., Tisdall, M., Otte, W. M., Dorfmüller, G., Cross, J. H., Schooneveld, M. van, Eijsden, P. van, Nees, F., Reuner, G., Krayenbühl, N., Zentner, J., Bulteau, C. & Braun, K. P. J. (2024). **Determinants of Functional Outcome after Pediatric Hemispherotomy**. *Annals of Neurology, 95*(2), 377–387. https://doi.org/10.1002/ana.26830

71. Reilly, C., Bjurulf, B. & Hallböök, T. (2024). **Autism and attention-deficit/hyperactivity disorder in children with Dravet syndrome: A population-based study**. *Developmental Medicine & Child Neurology, 66*(11), 1457–1465. https://doi.org/10.1111/dmcn.15937

72. Roach, A., Bennett, S., Heyman, I., Coughtrey, A., Stokes, I., Ndoci, X., Balakrishnan, S., Astle, N., Drinkwater, J., Evans, R., Frederick, U., Groszmann, M., Jones, S., McDonnell, K., Mobley, A., Murray, A., O'Sullivan, H., Ormrod, S., Prendegast, T., … Shafran, R. (2024). **Clinical effectiveness of drop-in mental health services at paediatric hospitals: A non-randomised multi-site study for children and young people and their families – study protocol**. *PLOS ONE, 19*(5), e0302878. https://doi.org/10.1371/journal.pone.0302878

73. Roth, J., Bergman, L., Weil, A. G., Brunette-Clement, T., Weiner, H. L., Treiber, J. M., Shofty, B., Cukiert, A., Cukiert, C. M., Tripathi, M., Chandra, P. S., Bollo, R. J., Machado, H. R., Santos, M. V., Gaillard, W. D., Oluigbo, C. O., Ibrahim, G. M., Jallo, G. I., Shimony, N., … Uliel-Sibony, S. (2023). **Added value of corpus callosotomy following vagus nerve stimulation in children with Lennox–Gastaut syndrome: A multicenter, multinational study**. *Epilepsia, 64*(12), 3205–3212. https://doi.org/10.1111/epi.17796

74. Salim, O., Chari, A., Zvi, I. B., Batchelor, R., Jones, M., Baldeweg, T., Cross, J. H. & Tisdall, M. (2024). **Patient, parent and carer perspectives surrounding expedited paediatric epilepsy surgery**. *Epilepsy Research, 200*, 107309. https://doi.org/10.1016/j.eplepsyres.2024.107309

75. Salim, O., Chari, A., Zvi, I. B., Batchelor, R., Baldeweg, T., Cross, J. H. & Tisdall, M. (2023). **Clinician views regarding early surgery for paediatric epilepsy**. *Seizure: European Journal of Epilepsy, 113*, 80–85. https://doi.org/10.1016/j.seizure.2023.11.011

76. Sanchis-Juan, A., Megy, K., Stephens, J., Ricaurte, C. A., Dewhurst, E., Low, K., French, C. E., Grozeva, D., Stirrups, K., Erwood, M., McTague, A., Penkett, C. J., Shamardina, O., Tuna, S., Daugherty, L. C., Gleadall, N., Duarte, S. T., Hedrera-Fernández, A., Vogt, J., … Carss, K. J. (2023). **Genome sequencing and comprehensive rare-variant analysis of 465 families with neurodevelopmental disorders**. *The American Journal of Human Genetics, 110*(8), 1343–1355. https://doi.org/10.1016/j.ajhg.2023.07.007

77. Sanders, M. W., Wolf, I. V. der, Jansen, F. E., Aronica, E., Helmstaedter, C., Racz, A., Surges, R., Grote, A., Becker, A. J., Rheims, S., Catenoix, H., Duncan, J. S., Tisi, J. D., Jacques, T. S., Cross, J. H., Kalviainen, R., Rauramaa, T., Chassoux, F., Devaux, B. C., … (EEBB), as the E. E. B. B. C. (2024). **Outcome of Epilepsy Surgery in MRI-Negative Patients Without Histopathologic Abnormalities in the Resected Tissue**. *Neurology, 102*(4), e208007. https://doi.org/10.1212/wnl.0000000000208007

78. Schoeler, N. E., Marston, L., Lyons, L., Halsall, S., Jain, R., Titre-Johnson, S., Balogun, M., Heales, S. J. R., Eaton, S., Orford, M., Neal, E., Reilly, C., Eltze, C., Stephen, E., Mallick, A. A., O'Callaghan, F., Agrawal, S., Parker, A., Kirkpatrick, M., … group, K. study. (2023). **Classic ketogenic diet versus further antiseizure medicine in infants with drug-resistant epilepsy (KIWE): a UK, multicentre, open-label, randomised clinical trial**. *The Lancet Neurology, 22*(12), 1113–1124. https://doi.org/10.1016/s1474-4422(23)00370-8

79. Jansson, J. S., Bjurulf, B., Blom, M. D., Hallböök, T. & Reilly, C. (2024). **Caregiver perceptions of the impact of Dravet syndrome on the family, current supports and hopes and fears for the future: A qualitative study**. *Epilepsy & Behavior, 156*, 109790. https://doi.org/10.1016/j.yebeh.2024.109790

80. Jansson, J. S., Bjurulf, B., Blom, M. D., Hallböök, T. & Reilly, C. (2024). **Diagnosis, epilepsy treatment and supports for neurodevelopment in children with Dravet Syndrome: Caregiver reported experiences and needs**. *Epilepsy & Behavior, 151*, 109603. https://doi.org/10.1016/j.yebeh.2023.109603

81. Spedden, M. E., O'Neill, G. C., Tierney, T. M., West, T. O., Schmidt, M., Mellor, S., Farmer, S. F., Bestmann, S. & Barnes, G. R. (2024). T**owards non-invasive imaging through spinal-cord generated magnetic fields**. *Frontiers in Medical Technology, 6*, 1470970. https://doi.org/10.3389/fmedt.2024.1470970

82. Sullivan, J., Lagae, L., Cross, J. H., Devinsky, O., Guerrini, R., Knupp, K. G., Laux, L., Nikanorova, M., Polster, T., Talwar, D., Ceulemans, B., Nabbout, R., Farfel, G. M., Galer, B. S., Gammaitoni, A. R., Lock, M., Agarwal, A., Scheffer, I. E. & Group, the Fa. D. S. (2023). **Fenfluramine in the treatment of Dravet syndrome: Results of a third randomized, placebo-controlled clinical trial**. *Epilepsia, 64*(10), 2653–2666. https://doi.org/10.1111/epi.17737

83. Thöny, B., Ng, J., Kurian, M. A., Mills, P. & Martinez, A. (2024). **Mouse models for inherited monoamine neurotransmitter disorders**. *Journal of Inherited Metabolic Disease, 47*(3), 533–550. https://doi.org/10.1002/jimd.12710

84. Tierney, T. M., Seedat, Z., Pier, K. S., Mellor, S. & Barnes, G. R. (2024). **Adaptive multipole models of optically pumped magnetometer data**. *Human Brain Mapping, 45*(4), e26596. https://doi.org/10.1002/hbm.26596

85. Ventura, S., Mathieson, S. R., O'Sullivan, M. P., O'Toole, J. M., Livingstone, V., Pressler, R. M., Dempsey, E. M., Murray, D. M. & Boylan, G. B. (2023). **Parent-led massage and sleep EEG for term-born infants: A randomized controlled parallel-group study**. *Developmental Medicine & Child Neurology, 65*(10), 1395–1407. https://doi.org/10.1111/dmcn.15565

86. Vermeulen, A., D'Arco, F., Groote, L. D. & Cocker, L. J. L. D. (2024). **Signal Abnormalities in Noonan Syndrome are Probably Consistent with Focal Areas of Signal Intensity (FASI)**. *Clinical Neuroradiology, 34*(2), 495–497. https://doi.org/10.1007/s00062-023-01356-1

Reviews or communications of expert opinion

1. Abiramalatha, T., Thanigainathan, S., Ramaswamy, V. V., Pressler, R., Brigo, F. & Hartmann, H. (2023). **Anti-seizure medications for neonates with seizures**. *Cochrane Database of Systematic Reviews, 2023*(10), CD014967. https://doi.org/10.1002/14651858.cd014967.pub2

2. AlMail, A., Jamjoom, A., Pan, A., Feng, M. Y., Chau, V., D'Gama, A. M., Howell, K., Liang, N. S. Y., McTague, A., Poduri, A., Wiltrout, K., Committee, Ipch. E., Bassett, A. S., Christodoulou, J., Dupuis, L., Gill, P., Levy, T., Siper, P., Stark, Z., … Costain, G. (2024). **Consensus reporting guidelines to address gaps in descriptions of ultra-rare genetic conditions**. *Npj Genomic Medicine, 9*(1), 27. https://doi.org/10.1038/s41525-024-00408-w

3. Amin, S. & O'Callaghan, F. (2024). **Response to updated clinical recommendations for the management of tuberous sclerosis complex associated epilepsy**. European Journal of Paediatric Neurology, 49, 129–130. https://doi.org/10.1016/j.ejpn.2024.01.005

4. Ball, M., Thorburn, D. R., & Rahman, S. (1993). **Mitochondrial DNA-Associated Leigh Syndrome Spectrum. GeneReviews®[Internet]**. University of Washington, Seattle.

5. Champsas, D., Zhang, X., Rosch, R., Ioannidou, E., Gilmour, K., Cooray, G., Woodhall, G., Pujar, S., Kaliakatsos, M. & Wright, S. K. (2024). **NORSE/FIRES: how can we advance our understanding of this devastating condition?** *Frontiers in Neurology, 15*, 1426051. https://doi.org/10.3389/fneur.2024.1426051

6. Cross, J. H., Benítez, A., Roth, J., Andrews, J. S., Shah, D., Butcher, E., Jones, A. & Sullivan, J. (2024). **A comprehensive systematic literature review of the burden of illness of Lennox–Gastaut syndrome on patients, caregivers, and society**. *Epilepsia, 65*(5), 1224–1239. https://doi.org/10.1111/epi.17932

7. Cross, J. H. (2024). **Towards equality in global epilepsy care and other neurological disorders**. *Developmental Medicine and Child Neurology, 66*(2), 140-141.

8. Feng, X., Piper, R. J., Prentice, F., Clayden, J. D. & Baldeweg, T. (2024). **Functional brain connectivity in children with focal epilepsy: A systematic review of functional MRI studies**. *Seizure: European Journal of Epilepsy, 117*, 164–173. https://doi.org/10.1016/j.seizure.2024.02.021

9. Gil-Nagel, A., Cross, J. H., Devinsky, O., Ceulemans, B., Lagae, L., Knupp, K., Schoonjans, A., Ryvlin, P., Thiele, E. A., Polega, S., Lothe, A. & Nabbout, R. (2024). **Comprehensive scoping review of fenfluramine's role in managing generalized tonic–clonic seizures in developmental and epileptic encephalopathies**. *Epilepsia, 65*(8), 2186–2199. https://doi.org/10.1111/epi.18020

10. Keshavan, N., Minczuk, M., Viscomi, C. & Rahman, S. (2024). **Gene therapy for mitochondrial disorders**. *Journal of Inherited Metabolic Disease, 47*(1), 145–175. https://doi.org/10.1002/jimd.12699

11. Khamis, S., Brown, A., Kaliakatsos, M., Eyre, M. & Lim, M. (2023). **Acute necrotizing encephalopathy of childhood: Prevention is better than cure especially if the cure remains elusive**. *Developmental Medicine & Child Neurology, 65*(9), 1139–1140. https://doi.org/10.1111/dmcn.15604

12. Messina, M., Vaz, F. M. & Rahman, S. (2024). **Mitochondrial membrane synthesis, remodelling and cellular trafficking**. *Journal of Inherited Metabolic Disease*. https://doi.org/10.1002/jimd.12766

13. Penner, J., Hassell, J., Brown, J. R., Mankad, K., Storey, N., Atkinson, L., Ranganathan, N., Lennon, A., Lee, J. C. D., Champsas, D., Kopec, A., Shah, D., Venturini, C., Dixon, G., De, S., Hatcher, J., Harris, K., Aquilina, K., Kusters, M. A., … Morfopoulou, S. (2023). **Translating metagenomics into clinical practice for complex paediatric neurological presentations**. *Journal of Infection, 87*(5), 451–458. https://doi.org/10.1016/j.jinf.2023.08.002

14. Perucca, E., French, J. A., Aljandeel, G., Balestrini, S., Braga, P., Burneo, J. G., Felli, A. C., Cross, J. H., Galanopoulou, A. S., Jain, S., Jiang, Y., Kälviäinen, R., Lim, S. H., Meador, K. J., Mogal, Z., Nabbout, R., Sofia, F., Somerville, E., Sperling, M. R., … Kapur, J. (2024). **Which terms should be used to describe medications used in the treatment of seizure disorders? An ILAE position paper**. *Epilepsia, 65*(3), 533–541. https://doi.org/10.1111/epi.17877

15. Piper, R. J., Ibrahim, G. M. & Tisdall, M. M. (2024). **Deep Brain Stimulation for Children with Generalized Epilepsy**. *Neurosurgery Clinics of North America, 35*(1), 17–25. https://doi.org/10.1016/j.nec.2023.09.002

16. Pressler, R. M., Abend, N. S., Auvin, S., Boylan, G., Brigo, F., Cilio, M. R., Vries, L. S. D., Elia, M., Espeche, A., Hahn, C. D., Inder, T., Jette, N., Kakooza-Mwesige, A., Mader, S., Mizrahi, E. M., Moshé, S. L., Nagarajan, L., Noyman, I., Nunes, M. L., … Hartmann, H. (2023). **Treatment of seizures in the neonate: Guidelines and consensus-based recommendations—Special report from the ILAE Task Force on Neonatal Seizures**. *Epilepsia, 64*(10), 2550–2570. https://doi.org/10.1111/epi.17745

17. Pujar, S. & Cross, J. H. (2024). **Diagnosis of Lennox-Gastaut syndrome and strategies for early recognition**. *Expert Review of Neurotherapeutics, 24*(4), 383–389. https://doi.org/10.1080/14737175.2024.2323568

18. Rahman, S., Bick, D., & Scott, R. H. (2024). **Whole genome sequencing to screen 100 000 newborns for treatable genetic disorders**. *Journal of Inherited Metabolic Disease, 47*(1), 7-8.

19. Rahman, S., Patterson, M., Peters, V., Morava, E., Zschocke, J. & Baumgartner, M. (2023). **Guidelines in the JIMD: Evidence-based practice for inherited metabolic disease**. *Journal of Inherited Metabolic Disease, 46*(4), 537–538. https://doi.org/10.1002/jimd.12620

20. Ramantani, G., Cserpan, D., Zentner, J., Tisdall, M. M. & Braun, K. P. (2024). **Reader Response: Comparison of Hemispheric Surgery Techniques for Pediatric Drug-Resistant Epilepsy: An Individual Patient Data Meta-Analysis**. *Neurology, 103*(2), e209329. https://doi.org/10.1212/wnl.0000000000209329

21. Reilly, C., Bjurulf, B. & Hallböök, T. (2024). **Do all individuals with Dravet syndrome have intellectual disability?** *Epilepsia, 65*(6), 1801–1802. https://doi.org/10.1111/epi.17847

22. Reilly, C., Jette, N., Johnson, E. C., Kariuki, S. M., Meredith, F., Wirrell, E., Mula, M., Smith, M. L., Walsh, S., Fong, C. Y., Wilmshurst, J. M., Kerr, M., Valente, K. & Auvin, S. (2023). **Scoping review and expert-based consensus recommendations for assessment and management of psychogenic non-epileptic (functional) seizures (PNES) in children: A report from the Pediatric Psychiatric Issues Task Force of the International League Against Epilepsy**. *Epilepsia, 64*(12), 3160–3195. https://doi.org/10.1111/epi.17768

23. Sullivan, J., Benítez, A., Roth, J., Andrews, J. S., Shah, D., Butcher, E., Jones, A. & Cross, J. H. (2024). **A systematic literature review on the global epidemiology of Dravet syndrome and Lennox–Gastaut syndrome: Prevalence, incidence, diagnosis, and mortality**. *Epilepsia, 65*(5), 1240–1263. https://doi.org/10.1111/epi.17866

24. Varadkar, S. M. & Gadgil, P. (2024). **Closing the mental health gap to support good life chances for children and young people**. *The Lancet Global Health, 12*(1), e10–e11. https://doi.org/10.1016/s2214-109x(23)00519-3

25. Winter, S., Walsh, D., Grisold, W., Jordan, J., Singhi, P., Cross, J. H., Sofia, … & F. (2023). **Uniting for global brain health: Where advocacy meets awareness**. https://doi.org/10.1016/j.yebeh.2023.109295

26. Zarakoviti, E., Shafran, R., Skuse, D., McTague, A., Batura, N., Palmer, T., Dalrymple, E., Bennett, S. D. & Reilly, C. (2022). **Factor associated with the occurrence of epilepsy in autism: a systematic review**. *Journal of Autism and Developmental Disorders, 53*(10), 3873–3890. https://doi.org/10.1007/s10803-022-05672-2

27. Ziobro, J., Pilon, B., Wusthoff, C. J., Benedetti, G. M., Massey, S. L., Yozawitz, E., Numis, A. L., Pressler, R. & Shellhaas, R. A. (2024). **Neonatal Seizures: New Evidence, Classification, and Guidelines**. *Epilepsy Currents*, 15357597241253382. https://doi.org/10.1177/15357597241253382

Books or Chapters in Books

1. McTague, A., Scheffer, I. E., Kullmann, D. M. & Sisodiya, S. (2024). **Chapter 11 Epilepsies**. *Handbook of Clinical Neurology, 203*, 157–184. https://doi.org/10.1016/b978-0-323-90820-7.00016-1

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

We are 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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