

## 48<sup>th</sup> BPNA Annual Scientific Meeting Virtual 19-21 January 2022 Programme

Wednesday	Wednesday 19 January 2022	
09.45-10.00	Welcome Dr Alasdair Parker, BPNA President & Dr Kathleen Gorman, Dublin Organising Committee	
10.00-11.55	Epilepsy (Session 1)  Chairs: Dr Mary O'Regan (Dublin), Dr Ailsa McLellan (Edinburgh) & Dr Jiayi Leow (Liverpool)	
10.00-10.35	Keynote Lecture: Laureate Prof Ingrid E Scheffer AO, The Uni Austin Health, Royal Children's Hospital, Florey Institute and Research Institute, Melbourne, Australia 'Precision medicine in genetic epilepsies'	-
10.35-10.50	2206 - Ganaxolone significantly reduces major motor seizures associated with <i>CDKL5</i> deficiency disorder: A randomized, double-blind, placebo-controlled Phase 3 study (Marigold Study)	Sam Amin
10.50-11.05	2459 - Efficacy and safety of Fenfluramine for the treatment of seizures associated with Lennox-Gastaut syndrome: A randomized, double-blind, placebo-controlled clinical trial	Sameer Zuberi
11.05-11.20	2383 - The spectrum of gain-of-function <i>SCN1A</i> disorders: novel phenotypes and response to sodium channel blocking therapies	Andreas Brunklaus
11.20-11.55	Keynote Lecture: Prof Mark Cunningham, Ellens Mayston Bates Professor of Neurophysiology of Epilepsy, Discipline of Physiology, School of Medicine, Trinity College Dublin  'Modelling mitochondrial epilepsy in vitro: conceptualisation, mechanisms, and therapeutic implications'	
11.55-13.00	Poster viewing and exhibition	
12.00-13.00	'Paediatric neurology trainees' meeting: Update on paediatric trainees welcome)	neurology training (All

<sup>\*</sup> Eligible for the prize: Best oral presentation by a trainee

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Wed 19 Jan	Biocodex Sponsored Symposia	
12.15-13.00	biocodex Sporisored Symposia	
	'New avenues to advance our understanding and management of rare epileptic disorders'	
	Dr Andreas Brunklaus (Glasgow), Prof Helen Cross (London), Prof Simona Balestrini (Italy)	
13.00-14.55	Epilepsy Surgery (Session 2)	
	Chairs: Mr Kieron Sweeney (Dublin), Prof. Donncha O'Brien (Du Shahwan (Dublin)	ublin) & Prof. Amre
13.00-13.35	Keynote Lecture: Prof Hans Holthausen, Center for Pediatric Neurology, Neurorehabilitation, and Epileptology, Schoen-Clinic, Vogtareuth, Bavaria, Germany 'Mild Focal Cortical Dysplasias – Severe Epilepsies'	
13.35-13.50	2523 - Epidemiology of rolandic epilepsy in Wales: A population study	Seung Gwan Ryoo**
13.50-14.05	2279 - Improving the diagnostic pathway and management of first seizures in infants with Sturge-Weber Syndrome	Finola Mankel**
14.05-14.20	2418 - A quality improvement project to improve medication administration times in paediatric status epilepticus	Michael Dunphy*
14.20-14.55	Keynote Lecture: Mr Martin Tisdall, MD FRCS(SN), Consultant Paediatric Neurosurgeon, Great Ormond Street Hospital, Honorary Associate Professor, UCL Great Ormond Street Institute of Child Health  'The role of minimally invasive procedures in paediatric epilepsy surgery'	
14.55-15.30	Poster viewing and exhibition	
15.30-17.45	Video Challenge (Session 3) Chair: Professor Manju Kurian (UCL-ICH GOSH, London) Expert Panel: Prof Tim Lynch, Prof Mary King (Dublin), Dr Kathleen Gorman (Dublin) & Prof Dan Healy (Dublin)	
15.30-15.45	Introduction	
15.45-16.05	Two acutely presenting toddlers	Vanita Shukla
16.05-16.25	Abnormal tone and movements in a 8-month old infant	Lianne Doherty*
16.25-16.45	A female infant with episodic movements	Claire Hall*
16.45-17.05	Paroxysmal movements in 2 children	Dora Steel*
17.05-17.25	Teenager with intermittent gait disorder	Martin Smith
17.25-17.45	A girl with gait difficulties	Cláudia Monteiro*

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Thursday 20	January 2022	
08.00-08.45	Personal practice session: Juvenile Myasthenia Gravis	
	Dr Declan O'Rourke (Dublin), Dr John McConville (Belfast), Mr (Dublin) & Dr Ronan Leahy (Dublin)	Jonathan McGuinness
09.00-09.45	9.45 GW Pharmaceuticals Sponsored Symposia	
	'Beyond-seizure benefits of early diagnosis and treatment in radevelopmental-and epileptic encephalopathies'	are, childhood-onset
	Dr Sam Amin (Bristol) & Dr Colin Reilly (Lingfield)	
09.00-09.45	Novartis Gene Therapies Sponsored Symposia	
	'ZOLGENSMA®▼ Gene Therapy in Spinal Muscular Atrophy (SN and early clinical practice'	1A) – developing a service
	Prof Laurent Servais (Oxford), Dr Vasantha Gowda (London) & (Manchester)	Dr Imelda Hughes
09.45-10.00	Poster viewing and exhibition	
10.00-11.45	Stroke & Acute neurology (Session 4)	
	Chairs: Dr Vijeya Ganesan (London), Dr Noelle Enright (London (Canada)	) & Dr Samyami Chowdhury
	Panel Discussion: Dr Vijeya Ganesan (Senior Lecturer in Paediatric Neurology, UCL Great Ormond Street Institute of Child Health, London), Dr Helen McCullagh (Consultant in Paediatric Neurology, Leeds Children's Hospital), Dr Adam Rennie (Consultant Neuroradiologist, Great Ormond Street Hospital, London, MB BS, BMSc, FRCR), Mr Greg James (Consultant Paediatric Neurosurgeon and Associate Professor of Neuroscience, Great Ormond Street Hospital and University College London) & Prof Tufail Patankar (Consultant Interventional Neuroradiologist, Leeds General Infirmary)  'Multidisciplinary practical management of cerebrovascular disease in children'	
11.00-11.15	2354 - Epidemiology of cerebrovascular disorders in a tertiary center	Maymunah Khries*
11.15-11.30	2274 - Incidence of paediatric multiple sclerosis and other relapsing demyelination conditions: 10-year follow-up UK surveillance of paediatric Acquired Demyelinating Syndromes (ADS)	Omar Abdel-Mannan*
11.30-11.45	2365 - Tic-like presentations during the COVID-19 pandemic – a paediatric case series	Sarah Buts*
11.45-13.00	Poster viewing and exhibition	
12.00-13.00	Becoming a paediatric neurologist (All welcome - session aimed at medical students, foundation doctors and ST1-5 paediatric trainees in particular)	
12.15-13.00	Roche Sponsored Symposia	
	'The treatment of infants and older children with spinal muscu	lar atrophy'
	Prof Laurent Servais (Oxford) & Dr Anne-Marie Childs (Leeds)	
13.00-15.30	Charity meeting	

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Thu 20 Jan 13.00-14.55	Neuromuscular (Session 5)  Chairs: Dr Kenneth McWilliam (Edinburgh), Dr Declan O'Rourke Murugan (Bristol)	
13.00-13.35	Keynote Lecture: Prof Laurent Servais, MD, PhD, Professor of Paediatric Neuromuscular Diseases, MDUK Oxford Neuromuscular Center, Department of Paediatrics, University of Oxford  'Newborn screening for Spinal Muscular Atrophy and beyond'	
13.35-13.50	2306 - Onasemnogene Abeparvovec for presymptomatic Infants with spinal muscular atrophy and two copies of <i>SMN2</i> : A Phase III Study	Francesco Muntoni
13.50-14.05	2242 - The RESTORE Registry: real-world assessments of interventions and long-term outcomes in patients with Spinal Muscular Atrophy (SMA)	Laurent Servais
14.05-14.20	2431 - Cross-sectional phenotypic and genotypic analysis of a UK cohort of patients with Nemaline myopathies	Luke Perry*
14.20 -14.55	Keynote Lecture: Prof Carsten Bönnemann, MD, habil, Chief, Neuromuscular and Neurogenetic Disorders of Childhood Section, National Institute of Neurological Disorders and Stroke, NIH, Adjunct Professor of Neurology, University of Pennsylvania School of Medicine  'Gene and Mutation-Specific Precision Therapies: From GAN to the COL6 Related Dystrophies and Beyond'	
14.55-15.30	Poster viewing and exhibition	

15.30-17.45	The Presidential Plenary (Session 6)
	Chairs: Dr Alasdair Parker (BPNA President), Prof Manju Kurian (Chair of BPNA Research) & Dr Jonathon Holland (Cambridge)
15.30-16.05	The Ronnie MacKeith Prize: Dr Joseph Symonds, Paediatric Neurology (ST8) GRID
	trainee, Royal Hospital for Children, Glasgow and Honorary Clinical Lecturer, Institute
	of Health and Wellbeing, University of Glasgow
	'Precision epidemiology in early childhood epilepsies'
16.05-16.20	President's International Guest: Prof Samanmali P. Sumanasena, MD, FRCPCH,
	Professor in Paediatric Disabilities, Department of Disability Studies, Faculty of
	Medicine, University of Kelaniya, Honorary Consultant Paediatrician, Ayati National
	Center for Children with Disabilities and North Colombo Teaching Hospital, Ragama, Sri
	Lanka
	'Successful neurological research in low and middle income settings'
16.20-16.50	President's International Guest: Prof Jo Wilmshurst, Head of Paediatric Neurology, Red
	Cross War Memorial Children's Hospital, University of Cape Town, South Africa and
	Director of the African Paediatric Fellowship Program, UCT
	'Collaborations in child neurology across equipped and limited resource settings – what
	can we learn from each other?'
16.50-17.45	The Ronnie MacKeith Guest Lecture: Prof Tally Lerman-Sagie, MD, Head of Pediatric
	Neurology Unit, Co-director of Magen Rare Disease Center, Co-director of Fetal
	Neurology Clinic, Wolfson Medical Center, Holon, Israel
	'Expansion of paediatric neurology into fetal life-the role of the neurologist in prenatal
	counselling'
17.45-18.00	Poster viewing and exhibition
18.00-19.00	Social Quiz

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Friday 21 Jar	nuary 2022	
08.00-08.45	Personal Practice Session: Pitfalls of genetic testing	
	Dr Kathleen Gorman (Dublin), Dr James O'Byrne (Dublin) & Dr Susan Harvey (Dublin)	
09.00-09.45	Biogen Sponsored Symposia	
	'Leveraging multi-disciplinary teams to support multifaceted as case studies in spinal muscular atrophy (SMA)'	nd evolving patient needs –
	Dr Min Ong (Sheffield), Ms Lindsey Pallant (Leeds) & Mrs Julie	Gray (Southampton)
09.00-09.45	Zogenix Sponsored Symposia	
	'Dravet syndrome: Overcoming treatment barriers and improvi	ing care'
	Prof Helen Cross (London), Prof Rima Nabbout (France) & Prof	Sameer Zuberi (Glasgow)
09.45-10.00	Poster viewing and exhibition	
10.00-11.55	Neuroradiology in clinical diagnosis (Session 7)	
	Chairs: Dr Katheen Gorman (Dublin), Prof David Webb (Dublin) &	Dr Lydia Green (Leeds)
10.00-10.35	Keynote lecture: Dr Shekeeb Mohammad, Paediatric Neurologist, Senior Lecturer, Department of Neurology and University of Sydney Clinical School, The Children's Hospital at Westmead, Sydney, Australia	
	'MRI pattern recognition in childhood onset basal ganglia disor	ders'
10.35-10.50	2497 - Mutations in HOPS-complex genes <i>VPS16</i> and <i>VPS41</i> cause early-onset movement disorders	Dora Steel*
10.50-11.05	2415 - A patient-derived midbrain dopaminergic model of Beta-Propeller Protein-Associated Neurodegeneration (BPAN): disease modelling and high throughput screening for targeted therapies	Apostolos Papandreou
11.05-11.20	2339 - Paediatric limbic encephalitis: a multi-centre study	Saraswathy Sabanathan
11.20-11.55	Keynote Lecture: Dr David Lynch, National Hospital for Neurology & Neurosurgery, London  'Approach to diagnosis of inherited white matter diseases in older children and adolescents: an adult neurologist perspective'	
11.55-13.00	Poster viewing and exhibition	
12.15-13.00	Biomarin Sponsored Symposia	
	'Efficient trio whole genome sequencing – Practical Tips for the	? Neurologist'
	Dr Alasdair Parker (Cambridge) & Dr Emma Clement (London)	
13.00-14.55	Neurogenetics (Session 8)	
	Chairs: Prof Mary King (Dublin), Prof Nicholas Allen (Galway) & Di	r Joseph Symonds (Glasgow)
13.00-13.35	Keynote lecture: Prof Sally Ann Lynch, Consultant Clinical Geneticist, Children's Health Ireland Temple Street, Dublin 'Irish Travellers, an indigenous European population with a unique culture'	
13.35-13.50	2493 - Genotype-phenotype associations in a large cohort of <i>SCN1A</i> -related epilepsies	Declan Gallagher**
13.50-14.05	2496 - Mutations encoding dopamine receptors cause complex childhood-onset hyperkinetic disorders	Dora Steel*

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14.05-14.20	2466 - Mid-gestation Cytokine profiles in mothers of children affected by autism spectrum disorder: a case-control Study	Michael Carter*
14.20-14.55	Keynote Lecture: Prof Louise Gallagher, MB MRCPsych PhD FTCD, Chair in Child and Adolescent Psychiatry, Trinity College Dublin	
	'What have we learned from 20 years of neurodevelopmental ownat is next?'	disorders genomics and
14.55-15.30	Poster viewing and exhibition	

Fri 21 Jan 15.30-17.15	Hot Topics (Session 9)	
	Chairs: Prof Bryan Lynch (Dublin), Dr Susan Byrne (Dublin) & Dr Kenneith Yong (Edinburgh)	
15.30-16.05	Keynote Lecture: Prof Paul Gissen, Professor in Paediatric Metabolic Medicine, UCL Great Ormond Street Institute of Child Health	
	'Developing therapies for incurable diseases'	
16.05-16.40	Keynote Lecture: Dr Dasha Nicholls, Clinical Reader in Child Psychiatry, Imperial College London	
	'Long COVID-19, long pandemic and mental health in young people'	
16.40-17.15	Keynote Lecture: Prof Rosalie Ferner, Consultant Neurologist and Lead Clinician for Neurofibromatosis, Guy's and St. Thomas' NHS Foundation Trust, London 'Neurofibromatosis 1 in the 21st Century'	
17.15-17.30	Prize giving and close of meeting	
	Dr Alasdair Parker, BPNA President & Prof Manju Kurian, Chair of BPNA Research	

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