

Cynhadledd Flynyddol CBNB 2010/BPNA Annual Conference 2010
Caeredin/ Edinburgh
Rhaglen/Programme

Wednesday/Dydd Mercher

27 January 2010/27 Ionawr 2010

- Noon Conference Registration and Lunch
- 12.55 – 13.00 Welcome
- 13.00 – 14.30 **Session 1: Oral Presentations**
Chair: Dr Louise Hartley, Consultant Paediatric Neurologist, Cardiff
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| 13.00 | Prevalence of Vitamin D deficiency in 117 boys with Duchenne muscular dystrophy | Dr P Munot |
| 13.15 | Oral bisphosphonates as prophylaxis of steroid induced osteoporosis in Duchenne Muscular Dystrophy | Dr T Willis |
| 13.30 | An autosomal-recessive form of centronuclear myopathy is caused by mutations in the skeletal muscle ryanodine receptor (RYR1) | Dr H Jungbluth |
| 13.45 | Normal Central Motor Conduction Times (CMCT) in children with severe primary and secondary dystonia under assessment for Deep Brain Stimulation (DBS) | Dr V McClelland |
| 14.00 | Development of the Questionnaire for Young People's Participation – an instrument for adolescents with cerebral palsy | Dr C Tuffrey |
| 14.15 | Controlled long term study of intrathecal baclofen in children with severe spastic Cerebral Palsy | Dr RE Morton |
- 14.30 – 15.15 **Keynote lecture: 'The Paediatric Neurologist and Advanced Fetal Care'**
Dr Omar Khwaja, Director, Fetal Neurology Programme, Attending Physician in Neurology, Children's Hospital, Boston, Assistant Professor of Neurology, Harvard Medical School
- 15.15 – 16.15 Poster viewing (tea & coffee will be served)
- 16.15 – 17.00 **Keynote Lecture: 'Inflicted Head Trauma: recognition and investigation: the evidence base'**
Dr Alison Kemp, Reader in Community Child Health, Cardiff University, Director Welsh Child Protection Systematic Review Group
- 17.10 – 19.00 Annual General Meeting of the British Paediatric Neurology Association
- 19.30 for 20.00 Reception and Dinner at The George Hotel, Edinburgh
- After dinner presentations by BPNA Guests :
- Dr David Chkhartishvili, Pediatric Neurologist, Tbilisi, Georgia
 - Dr Keryma Acevedo, Pediatric Neurologist, Assistant Professor of Pediatrics, University of Chile
 - Dr Gabriel Ofovwe, Consultant Paediatrician/Child Neurologist, University of Benin, Nigeria

Thursday/Dydd Iau

28 January 2010/28 Ionawr 2010

08.00 – 09.00 **Personal practice sessions:**

“Movement Disorders” – Led by Dr Huw Morris, Senior Lecturer in Neurology, MRC Centre for Neuropsychiatric Genetics and Genomics, Cardiff University School of Medicine and Dr Manju Kurian, Research Fellow, Birmingham University and Department of Paediatric Neurology, Birmingham Children’s Hospital

“Foetal Counselling” – Led by Dr Johann te Water Naudé, Consultant Paediatric Neurologist, University Hospital of Wales, Cardiff and Dr Omar Khwaja, Director, Fetal Neurology Programme, Attending Physician in Neurology, Children’s Hospital, Boston, Assistant Professor of Neurology, Harvard Medical School

09.00 – 10.30 **Session 2: Oral Poster Presentations**

Chair: Dr Ros Quinlivan, Consultant in Neuromuscular Disorders, RJAH Orthopaedic NHS Trust and Birmingham Children’s Hospital

Intracranial dissection mimicking transient cerebral arteriopathy in Childhood AIS Dr N Dlamini

Cognitive functioning in children with pantothenate kinase-associated neurodegeneration (PKAN) undergoing Deep Brain Stimulation (DBS) Ms H Gimeno

X- Linked hereditary motor sensory neuropathy (type I) presenting with “stroke” like episode Dr G Anand

Optimising outcomes following paediatric Deep Brain Stimulation (DBS) Ms H Gimeno

The steady mortality in bilateral cerebral palsy Prof G Baird

Aquaporins: age related expression in brain swelling. Miss JJ Wright

King-Denborough Syndrome associated with mutations in the skeletal muscle ryanodine receptor (RYR1) gene Dr H Jungbluth

A review of oral therapy in severe dystonic cerebral palsy Dr C Fairhurst

Profound Amnesia, Cognitive Decline and Dyskinesia Associated with Anti-Basal Ganglia Antibodies: Atypical CNS Autoimmune Disease Dr MA Illingworth

Is there a role of Botulinum Toxin A (BTXa) in symptom control of extrapyramidal movement disorder in children? Dr SS Sarkar

10.30 – 11.15 **Keynote lecture: ‘Hereditary Hyperekplexia – a startling synaptopathy’**

Professor Mark Rees, Director of the Institute of Life Science and Director of the Wales Epilepsy Research Network, School of Medicine, Swansea University

11.15 – 11.45 Tea and coffee break

11.45 – 12.30 **Ronnie MacKeith Guest Lecture: ‘Classification in epilepsy: moving forward’**

Professor Anne Berg, Department of Biology, Northern Illinois University

Chair: Professor Helen Cross, The Prince of Wales’s Chair of Childhood Epilepsy, UCL-Institute of Child Health, Great Ormond Street Hospital & NCYPE

12.30 – 13.30 Lunch

13.30 – 15.00 Session 3: Video session with oral presentations

Chair: Dr Cathy White, Consultant Paediatric Neurologist, Swansea

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| 13.30 | Devastating Epileptic Encephalopathy in School-Aged Children (DESC) with Elevated Voltage-Gated K ⁺ Channel Antibodies (VGKC-Ab) | Dr MA Illingworth |
| 13.45 | Anti-NMDA receptor encephalitis- emerging diagnosis in children presenting with neuropsychiatric symptoms and dyskinesia | Dr A Sudarsanam |
| 14.00 | An infantile onset epileptic encephalopathy associated with a homozygous missense variant in the SCN1A gene | Dr V Jain |
| 14.15 | Autism stereotypes?: The widening spectrum of Allan-Herndon-Dudley syndrome | Dr A Gika |
| 14.30 | I am a morning person.....! | Dr GP Ambegaonkar |
- 14.45 – 15.00 British Paediatric Neurology Surveillance Unit – Dr Finbar O’Callaghan
UKCNRC 2009 – Professor John Osborne
UKCNRC 2010 – Professor Colin Kennedy
- 15.15 – 17.00 Special Interest Group meetings (details in the conference handbook)
- 15.30 – 17.00 Genetics for Beginners Course
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| 15.30 | Molecular Techniques: what the paediatric neurologist needs to know
<i>Dr Andrew Fry, Clinical Genetics, University Hospital of Wales, Cardiff</i> | |
| 15.50 | Rett syndrome and related disorders
<i>Professor Angus Clarke, Clinical Genetics, University Hospital of Wales, Cardiff</i> | |
| 16.10 | Towards treatment for Tuberous Sclerosis
<i>Professor Julian Sampson, Clinical Genetics, University Hospital of Wales, Cardiff</i> | |
| 16.30 | Tubulin genes and cortical malformations
<i>Dr Daniela Pilz, Clinical Genetics, University Hospital of Wales, Cardiff</i> | |
| 16.50 | Discussion | |
| 17.00 | Close | |
- 15.00 onwards Explore Edinburgh
- 19.30 for 20.00 Reception and dinner at Murrayfield

Friday/Dydd Gwener

29 January 2010/29 Ionawr 2010

08.00 – 09.00 **Personal practice sessions:**

“The Teenager with Epilepsy” – Led by Dr Frances Gibbon, Consultant Paediatric Neurologist, University Hospital of Wales, Cardiff and Professor Phil Smith, Consultant Neurologist, University Hospital of Wales, Cardiff

“Evaluating the child with leg pains” – Led by Dr R Quinlivan, Consultant in Neuromuscular Disorders, RJA Orthopaedic NHS Trust and Birmingham Children’s Hospital

09.00 – 10.15 **Session 4: Oral Presentations**

Chair: Dr Frances Gibbon, Consultant Paediatric Neurologist, Cardiff

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| 9.00 | The spectrum of clinical disease in hyperekplexia: genotype and phenotype of forty-two cases | Dr RH Thomas |
| 9.15 | The degree of physicochemical difference between amino acids in missense mutations of SCN1A may help predict the epilepsy phenotype | Dr A Brunklaus |
| 9.30 | The use of Buccal Midazolam in the community for the treatment of seizures in children - A clearer picture. | Dr BG McCullagh |
| 9.45 | Migrating partial seizures in infancy- the first UK series of patients, an update from the current BPNSU study | Dr A McTague |
| 10.00 | Prospective study of POLG1 mutations presenting as epilepsy | Prof J Poulton |

10.15 – 11.00 **Keynote lecture: ‘Whole genome approaches to neurological disease’**

Dr Huw Morris, Senior Lecturer in Neurology, MRC Centre for Neuropsychiatric Genetics and Genomics, Cardiff University School of Medicine

11.00 – 11.30 Tea and coffee break

11.30 – 12.00 **Ronnie MacKeith Prize Lecture: ‘Infantile Parkinsonism-Dystonia: a novel neurotransmitter disorder and ‘Dopamine Transportopathy’**

Dr Manju Kurian, Research Fellow in Paediatric Neurology, Birmingham Children’s Hospital & School of Clinical and Experimental Medicine, University of Birmingham

Chair: Professor Helen Cross, The Prince of Wales’s Chair of Childhood Epilepsy, UCL-Institute of Child Health, Great Ormond Street Hospital & NCYPE

12.00 – 13.30 **Session 5: Oral Presentations**

Chair: Dr Frances Gibbon, Consultant Paediatric Neurologist, Cardiff

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| 12.00 | Plasma 3-O-methyl DOPA as a screening biomarker for Aromatic Amino Acid Decarboxylase (AADC) Deficiency and other Neurotransmitter disorders | Dr DE Lumsden |
| 12.15 | Mild phenotypes of Glut 1 deficiency may be common: consider measuring CSF glucose | Dr G Anand |
| 12.30 | The impact of infection in early childhood on intellectual function in adolescence: evidence from children with sickle cell disease. | Mr S Orfanos |

12.45	Paediatric GM2 Gangliosidosis: the changing characteristics of disease in contemporary U.K. patients	Dr NJC Smith
13.00	Early Recognition and Treatment of Hereditary Folate Malabsorption Reduces the Risk of Long Term Neurological Sequelae	Dr E Meyer
13.15	Long-term outcome and prognostic features of dancing eye syndrome – retrospective evaluation of 102 patients	Dr A Brunklaus
13.30 – 14.15	Lunch	
14.15 – 15.45	Session 6: Oral presentations	
	<i>Chair: Dr Martin Smith, Consultant Paediatric Neurologist, Birmingham & Oswestry</i>	
14.15	The Clinical Phenotype of Warburg Micro Syndrome	Dr IA Aligianis
14.30	MECP2 triplication - A rarely described familial cause of neurological regression in boys	Dr S Tang
14.45	Multiplex Ligation-dependent Probe Amplification (MLPA) Analysis is an Effective Tool for the Detection of Novel Intragenic PLA2G6 Mutations in Phospholipase Associated Neurodegeneration (PLAN): Implications for Molecular Diagnosis	Miss D Crompton
15.00	Predictors of cervical arterial abnormalities in children with acute arterial ischaemic stroke	Dr V Ganesan
15.15	Preliminary experience of carotid artery stents in young children	Dr V Ganesan
15.30	Childhood arterial ischaemic stroke without arteriopathy: a distinct entity?	Dr P Munot
15.45	Tea and coffee prior to journey home	