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	
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'**	
	Professor Alison Kemp, Professor of Child Health, Cardiff University, Director Welsh Child Protection Systematic Review Group	
17.10 – 19.00	Annual General Meeting of the British Paediatric Neurology Association	
19.15 for 19.45	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

Thursday/Dydd Iau

28 January 2010/28 Ionawr 2010

08.00 – 09.00 **Personal practice sessions:**

MAIN AUDITORIUM: *"**Movement Disorders: 'From Clinical Semiology to Genetic Aetiology'**"* – 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

NEW LIBRARY: *"**Paediatric Neurology and Fetal Medicine**"* – 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

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 R Jain
14.15	Autism sterotypies?: 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

15.00 onwards Explore Edinburgh

15.15 – 17.30 Special Interest Group meetings (details in the conference handbook)

15.30 – 17.00 **Update on Genetics**

15.30 Molecular Techniques: what the paediatric neurologist needs to know
Dr Andrew Fry, Clinical Genetics, University Hospital of Wales, Cardiff

15.50 Rett syndrome and related disorders
Professor Angus Clarke, Clinical Genetics, University Hospital of Wales, Cardiff

16.10 Towards treatment for Tuberous Sclerosis
Professor Julian Sampson, Clinical Genetics, University Hospital of Wales, Cardiff

16.30 Tubulin genes and cortical malformations
Dr Daniela Pilz, Clinical Genetics, University Hospital of Wales, Cardiff

16.50 Discussion

17.00 Close

19.30 for 20.00 Reception and dinner at Murrayfield

Coaches will depart for Murrayfield at:

19.00 from The George Hotel

19.00 from Jury's Inn

19.30 from The George Hotel *for people attending late meetings*

Coaches will return from Murrayfield at:

22.00 (or when dinner has finished, if later) to drop off at The George and Jury's Inn

23.00 to drop off at both The George Hotel and Jury's Inn

24.00 to drop off at both The George Hotel and Jury's Inn

Friday/Dydd Gwener

29 January 2010/29 Ionawr 2010

08.00 – 09.00 **Personal practice sessions:**

*MAIN AUDITORIUM: "**Evaluating the child with leg pains**"* – Led by Dr Ros Quinlivan, Consultant in Neuromuscular Disorders, RJAH Orthopaedic NHS Trust and Birmingham Children's Hospital

*NEW LIBRARY: "**Teenagers and epilepsy: What's occurrin'?"*** – Led by Dr Frances Gibbon, Consultant Paediatric Neurologist, University Hospital of Wales, Cardiff and Professor Phil Smith, Consultant Neurologist, University Hospital of Wales, Cardiff

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

Chair: Dr Frances Gibbon, Consultant Paediatric Neurologist, Cardiff

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

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**

Chair: Dr Martin Smith, Consultant Paediatric Neurologist, Birmingham & Oswestry

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 Phospholiase 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