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’**  
*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
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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, RIAH 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 – 13.00  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 V Jain

- **14.15 Autism stereotypies?: 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**

- **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**

15.00 onwards **Explore Edinburgh**

19.30 for 20.00 **Reception and dinner at Murrayfield**
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, RJAH Orthopaedic NHS Trust and Birmingham Children’s Hospital

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

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
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<th>Time</th>
<th>Title</th>
<th>Speaker</th>
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<tr>
<td>12.45</td>
<td>Paediatric GM2 Gangliosidosis: the changing characteristics of disease in contemporary U.K. patients</td>
<td>Dr NJC Smith</td>
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<td>13.00</td>
<td>Early Recognition and Treatment of Hereditary Folate Malabsorption Reduces the Risk of Long Term Neurological Sequelae</td>
<td>Dr E Meyer</td>
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<td>13.15</td>
<td>Long-term outcome and prognostic features of dancing eye syndrome – retrospective evaluation of 102 patients</td>
<td>Dr A Brunklaus</td>
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<td>14.15</td>
<td>Session 6: Oral presentations</td>
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<td>14.15</td>
<td>Chair: Dr Martin Smith, Consultant Paediatric Neurologist, Birmingham &amp; Oswestry</td>
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<td>14.15</td>
<td>The Clinical Phenotype of Warburg Micro Syndrome</td>
<td>Dr IA Aligianis</td>
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<td>14.30</td>
<td>MECP2 triplication - A rarely described familial cause of neurological regression in boys</td>
<td>Dr S Tang</td>
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<td>14.45</td>
<td>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</td>
<td>Miss D Crompton</td>
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<td>15.00</td>
<td>Predictors of cervical arterial abnormalities in children with acute arterial ischaemic stroke</td>
<td>Dr V Ganesan</td>
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<td>15.15</td>
<td>Preliminary experience of carotid artery stents in young children</td>
<td>Dr V Ganesan</td>
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<td>15.30</td>
<td>Childhood arterial ischaemic stroke without arteriopathy: a distinct entity?</td>
<td>Dr P Munot</td>
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<td>15.45</td>
<td>Tea and coffee prior to journey home</td>
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