British Paediatric Neurology Association
Annual Conference 28-30 January 2009
National Motorcycle Museum, Birmingham
Programme

Wednesday 28 January 2009

Noon  Conference Registration and Lunch

12.55 – 13.00  Welcome

13.00 – 14.30  **Session 1: Oral Presentations**
Chair:  Professor Stefano Seri, Birmingham

- Acute spinal cord lesions in children  B Harms
- Is it possible to diagnose mitochondrial disease on the basis of the clinical presentation?  C Verity
- Aicardi-Goutières syndrome (AGS) and other congenital infection-like phenotypes: preliminary data and emerging patterns  J Livingston
- Recovery of consciousness following acute symptomatic seizures due to central nervous system infections in children  E King
- Appearances of the cerebellum on diffusion weighted imaging following preterm birth  A Hart
- The multiple phenotypes of arthrogryposis multiplex congenita (AMC) with special reference to the neurogenic variant  G Ambegaonkar

14.30 – 15.00  **Keynote lecture: ‘Update on leucoencephalopathies’**
*Professor van der Knaap, Holland*
Chair:  Dr Raj Gupta, Consultant Paediatric Neurologist, Birmingham

15.00 – 16.30  Poster viewing (tea & coffee will be served)

16.30 – 17.00  **Presentations by Guests of the BPNA**
Introduced by Professor Helen Cross, London, BPNA President

- Dr Inga Talvik, Senior Consultant in Pediatric Neurology
  Children’s Clinic of Tartu University Hospital, Estonia

- Professor Dana Craiu, Professor of Paediatric Neurology
  Alexandru Obregia Hospital, Bucharest, Romania

- Professor Nuri Shembesh, Consultant & Head of Neurology
  Al-fateh Children’s Hospital, Beghazi, Libya

17.00 – 17.10  Eulogy for Dr Sarah Benton by Dr Carlos de Sousa

17.10 – 19.00  Annual General Meeting of the British Paediatric Neurology Association
*For members of the Association*

- 19.00  Reception in the National Motorcycle Museum
- 20.00  Piano recital in memory of the late Dr Stuart Green
- 20.30  Dinner in the Britannia Suite
Thursday 29 January 2009

08.00 – 09.00  **Personal practice sessions:**

1. “White matter disorders – it’s not all black and white”
   Chair: Dr E Wassmer, Panel: Prof M Van der Knaap, Dr L MacPherson

2. “Dystonia / movement disorders – video based discussions”
   Chair: Dr M Smith, Panel: Prof E Fernandez-Alvarez, Dr H Pal, Dr J-P Lin

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

Chair: Dr Tracey Willis, Research Fellow, Newcastle Upon Tyne

- Electro-clinical phenotype of males with MECP2 duplication: J Kisler
- Mortality from childhood stroke in England and Wales 1921-2000: A Mallick
- Continuous intrathecal baclofen in non ambulant children with cerebral palsy: R Morton
- Evaluation of the efficacy and acceptability of cardiac biomarker N-terminal pro-brain natriuretic peptide as a method of early identification of cardiac impairment in children with Duchenne muscular dystrophy: K Lewis
- Familial hemiplegic migraine with a novel mutation in the ATP1A2 gene associated with febrile seizures and recurrent unprovoked encephalopathy: V Jain
- Adenylosuccinate lyase deficiency in the United Kingdom – an under recognised cause of developmental delay, epilepsy and delayed myelination?: C Lundy
- Have guidelines improved the use of lumbar punctures for children with suspected central nervous system Infections?: T Bugembe
- Neurological features and brain magnetic resonance imaging findings in patients harbouring mutations in the SLC16A2 gene: A Gika
- Epilepsy and mental retardation limited to females (EFMR): paediatric presentation of the first UK family: M Illingworth
- A novel syndrome of severe mental retardation, spasticity and tapetoretinal degeneration (MRST) is linked to chromosome 15q.25.1: M Kurian
- Response to deep brain stimulation (DBS) in primary and secondary childhood dystonias improves with the volume of globus pallidus internus (GPI) stimulated as determined by the number of activated contacts: D Lumsden
- Partially dopa responsive dystonia in a compound heterozygote for GTP- cyclohydrolase 1 gene mutations: K McWilliam
- Defining normal values for two simple test of strength in young children: J Ng
- Psychological status at 1, 6 and 12 months in children with severe/moderate and mild traumatic brain injury (TBI) and non-injured controls; relationship to injury severity, health related quality of life (HRQL), socio-economic status and family burden: R Lakschmi
- Childhood absence seizures and GLUT 1 deficiency: D Webb
Effect of attributional style and maternal mental health on quality of life of children treated for posterior fossa tumours: interim results  
C Kennedy

A diagnostic algorithm for the diagnosis of neuronal ceroid lipofuscinosis in children  
R Williams

Dihydrolipoamide acetyltransferase deficiency in cases of atypical pantothenate kinase associated neurodegeneration  
C McWilliam

10.30 – 11.00  
Keynote lecture: ‘Dopamine defects in children’
Professor Emilio Fernandez-Alvarez, Professor Titular Universidad de Barcelona, Hospital San Juan de Dios, Barcelona, Spain
Chair: Dr Sunny Philip, Consultant Paediatric Neurologist, Birmingham

11.00 – 11.30  
Tea and coffee break

11.30 – 12.15  
Keynote lecture: ‘From pathology to therapy in muscular dystrophies’
Professor Caroline Sewry, RJAH Orthopaedic Hospital, Oswestry & Professor Francesco Muntoni, Institute of Child Health, London
Chair: Dr Ros Quinlivan, Consultant, RJAH Orthopaedic NHS Trust

12.15 – 13.00  
Ronnie MacKeith Guest Lecture: ‘Epilepsy genetics: new twists in the tale’
Professor Ingrid Scheffer, University of Melbourne
Introduced by Professor Helen Cross, London, BPNA President

13.00 – 14.00  
Lunch

14.00 – 15.00  
Session 3: Oral Presentations
Chair: Dr Raj Gupta, Consultant Paediatric Neurologist, Birmingham

Non-accidental head injury: a disorder of deprivation?  
P Jones

Radiological and clinical findings that aid in the diagnosis of traumatic head injury in infancy  
V Kelly

Use of hierarchical linear modelling of verbal and non-verbal memory function to investigate the effects of injury severity, age and gender on levels of outcome and rate of change in children with traumatic brain injury (TBI) and non-injured controls  
H Miller

Behavioural problems, adaptive behaviour, cognitive status, emotional status, health status, and HRQL in the first 12 months after diagnosis in children with brain tumours; a prospective longitudinal study  
A Penn

15.00 – 15.15  
British Paediatric Neurology Surveillance Unit
Dr Finbar O’Callaghan, Consultant Paediatric Neurologist, Bristol

UK Children’s Neurological Research Campaign
Professor John Osborne, Bath

15.15 – 17.00  
Special Interest Group meetings (details in the conference handbook)

15.30 – 17.00  
Metabolic Course for those who have pre registered

19.30 for 20.00  
‘A burst of spice – a night of Bollywood Dreams!’  
Centennial Centre, Birmingham
Friday 30 January 2009

08.00 – 09.00  **Personal practice sessions:**

1. “Management of spasticity in a diplegic child”  
   Chair: Dr M Smith, Panel: Mr A Roberts, Dr R Morton

2. “Multiple sclerosis – early intervention”  
   Chair: Dr E Wassmer, Panel: Dr C Hemingway, Dr M Lim

3. “Ketogenic diet in epilepsy – from fasting to Atkins”  
   Chair: Dr S Philip, Panel: Prof H Cross, Dr E Neal, Ms G Magrath

09.00 – 10.30  **Session 4: Oral Presentations**

Chair: Dr Raj Gupta, Consultant Paediatric Neurologist, Birmingham

Seizures in comatose children in the paediatric intensive care unit (PICU)  
A Shahwan

Adherence to anti-epileptic medication in children with epilepsy from a Scottish population cohort  
J Shetty

Parent’s views about drug trials in children with refractory convulsive status epilepticus  
S Agrawal

Molecular genetic investigation of autosomal recessive cryptogenic infantile spasms (ARCIS): evidence of clinical and genetic locus heterogeneity  
M Kurian

Multiplex ligation probe amplification detects SCN1A gene alterations in >10% of mutation-negative Dravet Syndrome / core SMEI patients  
A Brunklaus

The clinical spectrum of gelastic seizures in children  
M Chitre

10.30 – 11.00  **Keynote lecture: ‘Discovering genes for recessively inherited disorders’**

*Professor Eamonn Maher, Medical and Molecular Genetics, University of Birmingham*

Chair: Dr Sunny Philip, Consultant Paediatric Neurologist, Birmingham

11.00 – 11.30  **Tea and coffee break**

11.30 – 12.00  **Ronnie MacKeith Prize Lecture: ‘Identifying clinical phenotypes in the search for autism susceptibility genes’**

*Dr Jeremy Parr, Great Ormond Street Hospital, London & The University of Oxford*

Introduced by Professor Helen Cross, London, BPNA President
12.00 – 13.15 **Session 5: Oral Presentations**
Chair: Dr Martin Smith, Consultant Paediatric Neurologist, Birmingham

Automated visual field assessment in young children using eye tracking – preliminary data from 9 children  
M MacRae

Evaluation of the diagnostic screening service for PLA2G6 mutations in phospholipase associated neurodegeneration (PLAN)  
M Kurian

Trends in BFMDRS scores in patients receiving pallidal deep brain stimulation  
D Lumsden

Disease progress modelling improves trial efficiency: work smarter not harder  
R Forsyth

Clinical and radiological features of vein of galen malformation  
N McSweeney

13.15 – 14.00 Lunch

14.00 – 15.30 **Session 6: Video session with Oral presentations**
Chair: Dr Helen Roper, Birmingham

Non-epileptic status epilepticus  
S Haden

Immune mediated encephalopathy with chorea, seizures and developmental regression- case report and literature review  
S Agrawal

Video capture of rapid recovery from status dystonicus in an 11-year old girl with PANK2 disease after switching on Kinetra® DBS stimulator  
J-P Lin

Epileptic apnoea as the only presentation of temporal lobe epilepsy in a child with chromosomal anomaly  
A Desurkar

Evidence of defective neuromuscular transmission (NMT) in a patient with aromatic L-amino acid decarboxylase (AADC) deficiency  
A Gika

Acute life-threatening episodes (ALTE) secondary to a dystonic movement disorder in Rett syndrome (RS): treatment with Trihexyphenidyl (THP)  
A Gika

Vigabatrin induced movement disorder in West syndrome  
A Nussbaumer

A novel SCN1A mutation with unusual seizure semiology  
V Jain

15.30 Closing – Professor Helen Cross, BPNA President

15.35 – 16.00 Tea and coffee prior to journey home