# British Paediatric Neurology Association Annual Conference 16-18 January 2008, Leeds Programme

## Wednesday 16 January 2008

| Noon  | Conference Registration and Lunch  |  |
|---|--|--|
| 13.00 – 13.05   | Welcome  |  |
|   | Dr John Livingston, Consultant Paediatric Neurologist, Leeds   |  |
| 13.05 – 14.35   | Session 1: Oral Presentations  |  |
|   | Chair: Professor Helen Cross, London   |  |
| 13.05   | SCN1A mutation analysis aids early diagnosis of infantile onset epilepsies   | Dr S Zuberi  |
| 13.20   | Inherited SCN1A mutation in siblings with Panayiotopoulos syndrome   | Dr J Livingston  |
| 13.35   | Longitudinal cognitive profile in infant onset epilepsy  | Dr A Whitney   |
| 13.50   | Phenotypic characterization of a large family with benign familial neonatal seizures associated with an exon duplication of the KCNQ2 gene   | Dr R Lunan   |
| 14.05   | Efficacy and tolerability of the ketogenic diet in drug resistant epilepsy: a randomised controlled trial of classical and medium chain triglyceride protocols   | Mrs E Neal   |
| 14.20   | Extreme drug toxicity with Clobazam due to genetic polymorphism masquerading as factitious or induced illness  | Dr R Hughes  |
|   | Keynote lecture: 'Restorative technologies for children with disability: is it all hype?"  |  |
| 14.30 – 15.00   |  | lisability: is it all  |
| 14.30 – 15.00   |  | •  |
| 14.30 – 15.00   | hype?" Professor Bipin Bhakta, Charterhouse Professor in Rehabilitation I  | •  |
| 14.30 – 15.00<br>15.00 – 16.30                            | hype?"  Professor Bipin Bhakta, Charterhouse Professor in Rehabilitation In University of Leeds  | •  |
|   | hype?"  Professor Bipin Bhakta, Charterhouse Professor in Rehabilitation In University of Leeds  Chair: Dr Mike Clarke, Leeds  | •  |
| 15.00 – 16.30   | hype?"  Professor Bipin Bhakta, Charterhouse Professor in Rehabilitation In University of Leeds  Chair: Dr Mike Clarke, Leeds  Poster viewing (tea & coffee will be served)  | •  |
| 15.00 – 16.30   | hype?"  Professor Bipin Bhakta, Charterhouse Professor in Rehabilitation In University of Leeds  Chair: Dr Mike Clarke, Leeds  Poster viewing (tea & coffee will be served)  Presentations by Guests of the BPNA   | Medicine   |
| 15.00 – 16.30<br>16.30 – 17.00                            | hype?"  Professor Bipin Bhakta, Charterhouse Professor in Rehabilitation In University of Leeds  Chair: Dr Mike Clarke, Leeds  Poster viewing (tea & coffee will be served)  Presentations by Guests of the BPNA  Chair: Dr Mike Pike, Oxford  Dr Anannit Visudtibhan, Associate Professor in Pediatrics and Pediatrics  | Medicine iatric Neurology,   |
| 15.00 – 16.30<br>16.30 – 17.00<br>16.30                   | hype?"  Professor Bipin Bhakta, Charterhouse Professor in Rehabilitation In University of Leeds  Chair: Dr Mike Clarke, Leeds  Poster viewing (tea & coffee will be served)  Presentations by Guests of the BPNA  Chair: Dr Mike Pike, Oxford  Dr Anannit Visudtibhan, Associate Professor in Pediatrics and Ped Ramathibodi Hospital, Bangkok, Thailand  Dr Tran Thi Thu Ha, Vice Director of Rehabilitation Department, Na   | Medicine iatric Neurology,   |
| 15.00 – 16.30<br>16.30 – 17.00<br>16.30<br>16.40          | Professor Bipin Bhakta, Charterhouse Professor in Rehabilitation In University of Leeds Chair: Dr Mike Clarke, Leeds Poster viewing (tea & coffee will be served)  Presentations by Guests of the BPNA Chair: Dr Mike Pike, Oxford  Dr Anannit Visudtibhan, Associate Professor in Pediatrics and Ped Ramathibodi Hospital, Bangkok, Thailand  Dr Tran Thi Thu Ha, Vice Director of Rehabilitation Department, Na Pediatrics, Hanoi, Vietnam  Dr Ramesh Kant Adhikari, Professor in Child Health and Dean, Institution Department, Institution Departm | Medicine iatric Neurology,   |
| 15.00 – 16.30<br>16.30 – 17.00<br>16.30<br>16.40<br>16.50 | Professor Bipin Bhakta, Charterhouse Professor in Rehabilitation In University of Leeds Chair: Dr Mike Clarke, Leeds Poster viewing (tea & coffee will be served)  Presentations by Guests of the BPNA Chair: Dr Mike Pike, Oxford  Dr Anannit Visudtibhan, Associate Professor in Pediatrics and Ped Ramathibodi Hospital, Bangkok, Thailand  Dr Tran Thi Thu Ha, Vice Director of Rehabilitation Department, Na Pediatrics, Hanoi, Vietnam  Dr Ramesh Kant Adhikari, Professor in Child Health and Dean, Ins Kathmandu, Nepal  | Medicine iatric Neurology, ational Hospital of titute of Medicine, |

### Thursday 17 January 2008

### 08.00 - 09.00 Personal practice sessions:

- The management of non-organic disorders that present to the neurologist Led by Dr M Eminson & Professor D Cottrell
- The practical management of increased muscle tone
   Led by Dr M Clarke & Dr N Basheer & Ms S Garbutt & Dr A Musson

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

Chair: Dr Neti Gayatri, Leeds

| 09.00 | Measuring tympanic membrane displacement in raised intracranial pressure   | Dr M Lim        |
|-------|--|-----------------|
| 09.0  | Bannayan-Riley-Ruvalcaba syndrome: a common cause of extreme macrocephaly and neurodevelopmental delay                                   | Dr N Lynch      |
| 09.10 | Factors influencing admission to ICU in children presenting with status epilepticus (SE)   | Dr S Tirupathi  |
| 09.1  | A simple screening test for defects in creatine metabolism   | Dr S Hogg       |
| 09.20 | A contributory role for neuroinflammation in Batten disease  | Dr M Lim        |
| 09.2  | Making the diagnosis in leukoencephalopathy - key features in a cohort of children with Pelizaeus Merzbacher disease                     | Dr A Maw        |
| 09.30 | Infantile Parkinsonism- dystonia with raised CSF dopamine metabolites  | Dr J Ng         |
| 09.3  | Acute necrotising encephalopathy (ANE), associated with a missense mutation in the nuclear pore component ran binding protein 2 (RANBP2) | Dr A-M Childs   |
| 09.40 | Posterior reversible leucoencephalopathy syndrome (PRES) as a presentation of Guillain Barre syndrome (GBS) in a child                   | Dr A Desurkar   |
| 09.4  | Novel neuro-renal syndrome due to AQP4 antibody mediated disease   | Dr V Ramesh     |
| 09.50 | Dominantly inherited leucoencephalopathy with recurrent haemorrhagic stroke due to COL4A1 mutation                                       | Dr S Shah       |
| 09.5  | Diffuse hemispheric DNET: a new radiological variant associated with early onset severe epilepsy   | Dr K Sarsfield  |
| 10.00 | Primary diffuse leptomeningeal melanosis mimicking tuberculous meningitis  | Dr V Jain       |
| 10.0  | Double trouble: central core myopathy with RYR1 mutation masks 5q spinal muscular atrophy  | Dr S Chandratre |
| 10.10 | Congenital myasthenia due to COLQ gene mutation responsive to ephedrine therapy  | Dr R Pandey     |
| 10.1  | Establishing the parameters for clinical trials of antisense oligonucleotide therapy in Duchenne muscular dystrophy                      | Dr M Kinali     |

| 10.20           | Long-term use of intermittent low-dosage prednisolone therapy in Duchenne muscular dystrophy, tolerance and effect on functional outcome                | Dr M Kinali          |
|-----------------|---|----------------------|
| 10.25           | Perinatal dyskinesia as a presenting feature in Prader Willi syndrome   | Dr N McSweeney       |
| 10.30 – 11.00   | Keynote lecture: 'Aicardi-Goutières syndrome and other genetic causes of intracranial calcification'  |                      |
|                 | Dr Yanick Crow, Senior Lecturer and Honorary Consultant in Clinical Genetics,<br>Institute of Molecular Medicine, St James's University Hospital, Leeds |                      |
|                 | Chair: Dr Anne-Marie Childs, Leeds  |                      |
| 11.00 – 11.30   | Tea and coffee break  |                      |
| 11.30 – 12.00   | Session 3: Oral Presentations   |                      |
|                 | Chair: Dr Anne-Marie Childs, Leeds  |                      |
| 11.30           | Health related quality of life (HRQL) and its determinants at 1, 6 and 12 months in Traumatic Brain Injury (TBI) children and non-injured controls      | Dr B Hameed          |
| 11.45           | Bringing back the child: development after extreme deprivation  | Dr L-J Brown         |
| 12.00 – 13.00   | - 13.00 Ronnie MacKeith Guest Lecture: 'Sam and Ronnie: attention and personality   |                      |
|                 | Professor George Rousseau, Co-Director, Oxford University Centre<br>Childhood   | e for the History of |
|                 | Chair: Dr Mike Pike, Oxford   |                      |
| 13.00 – 14.00   | Lunch   |                      |
| 14.00 – 15.00   | Session 4: Oral Presentations   |                      |
|                 | Chair: Debbie Murdoch-Eaton, Professor of Medical Education, Leeds  |                      |
| 14.00           | Innovative speech and language therapy for school age children with language impairment: findings from a randomised controlled trial                    | Prof A O'Hare        |
| 14.15           | Botulinum toxin type A as a treatment of pain in cerebral palsy   | Dr C Lundy           |
| 14.30           | Parent and child perception of nitrous oxide use for botulinum toxin injections of upper and lower limb spasticity                                      | Dr K Pysden          |
| 14.45           | Deep brain stimulation improves motor ability and reported quality of life in primary and secondary generalised dystonia                                | Dr T Kerr            |
| 15.00 – 15.10   | British Paediatric Neurology Surveillance Unit  |                      |
|                 | Dr Finbar O'Callaghan, Chair BPSU, Consultant Paediatric Neurologist, Bristol   |                      |
| 15.00 – 17.00   | Special Interest Group meetings (details in the conference handbook)  |                      |
| 19.30 for 20.00 | Reception, dinner and ceilidh   |                      |

## Friday 18 January 2007

| 08.00 – 09.00 | Personal practice sessions:  1. Optimal management of CNS Tumour Led by Dr M Elliott, Mr P Chumas & Dr J Livingston  2. The management of newly diagnosed epilepsy Led by Dr C Ferrie & Dr T Martland |                      |  |
|---------------|---|----------------------|--|
| 09.00 – 10.30 | Session 5: Oral Presentations   |                      |  |
|               | Chair: Dr Philip Jardine, Bristol   |                      |  |
| 09.00         | Spectrum of brain involvement in muscular dystrophies with defective dystroglycan glycosylation   | Prof F Muntoni       |  |
| 09.15         | Dystrophin positive revertant fibres do not increase with age in<br>Duchenne muscular dystrophy   | Dr M Kinali          |  |
| 09.30         | Hereditary Motor Sensory Neuropathy (HMSN) with superadded inflammatory polyneuropathy in two children  | Dr A Desurkar        |  |
| 09.45         | Congenital myasthenic syndromes in childhood: diagnostic pitfalls and management issues   | Dr S Robb            |  |
| 10.00         | A national prospective population-based study of children with mitochondrial disease: clinical presentation and method of diagnosis in 101 cases  | Dr D<br>Krishnakumar |  |
| 10.15         | Significant variation in the prevalence and spectrum of neuromuscular disease among different ethnic groups: an observational study   | Dr A-M Childs        |  |
| 10.30 – 11.00 | Keynote lecture: 'The delivery of paediatric neurosurgery: a time for change'  Mr Paul Chumas, Consultant Paediatric Neurosurgeon, Leeds  |                      |  |
|               |   |                      |  |
|               | Chair: Dr John Livingston, Leeds  |                      |  |
| 11.00 – 11.30 | Tea and coffee break  |                      |  |
| 11.30 – 12.00 | Ronnie MacKeith Prize Lecture: 'IS you is, or IS you ain't (ma' Baby)? And how should we treat infantile spasms anyway?'  |                      |  |
|               | Dr Andrew Lux, Consultant in Paediatric Neurology, Bristol Royal Ho   | spital for Children  |  |
|               | Chair: Dr Penny Fallon, London  |                      |  |
| 12.00 – 13.15 | Session 6: Oral Presentations   |                      |  |
|               | Chair: Dr Nigel Basheer, Leeds  |                      |  |
| 12.00         | Improving the yield of investigations in mitochondrial disorders  | Dr A Parker          |  |
| 12.15         | The use of aciclovir in children with possible viral encephalitis   | Dr R Kneen           |  |
| 12.30         | Cerebrovascular dysplasia in neurofibromatosis Type 1   | Dr A Cairns          |  |
| 12.45         | Use of cerebral Magnetic Resonance Imaging (MRI) and Transcranial Doppler (TCD) to monitor cerebral vascular and perfusion abnormalities in children with sickle cell disease: the Irish experience   | Dr N Lynch           |  |

| 13.00         | Neurological complications associated with isolated liver transplant – a retrospective long term study   | Dr T El-Azzabi  |
|---------------|--|-----------------|
| 13.15 – 14.00 | Lunch  |                 |
| 14.00 – 15.00 | Session 7: Video session with oral presentations Chair: Dr Colin Ferrie, Leeds   |                 |
| 14.00         | Hereditary neuralgic amyotrophy: a differential for recurrent shoulder pain and weakness   | Dr S Chandratre |
| 14.12         | Improved motor function and resumption of speech and oral feeding in a case of non-DYT1 idiopathic torsion dystonia following pallidal deep brain stimulation (DBS). | Dr T Kerr       |
| 14.24         | Movement disorders associated with complex regional pain syndrome  | Dr S Agrawal    |
| 14.36         | Paroxysmal episodes, "re-build up" phenomenon and moyamoya disease   | Dr N Dlamini    |
| 14.48         | Creatine synthesis disorder: atypical presentation of a treatable disorder   | Dr D O'Rourke   |

#### 15.00 - 15.30 BPNA 'Grand Round'

An acute fatal neurological syndrome in a teenage boy is presented by Dr Basheer (Leeds General Infirmary), Dr Carlos de Sousa (Great Ormond Street Hospital, London), and Dr Phil Jardine (Frenchay Hospital, Bristol).

A previously healthy 14-year-old male with an acute fatal brainstem syndrome characterized by rapidly progressive course is presented. An approach to investigation, treatment and differential diagnosis will be discussed in terms of clinical-radiological correlation. Audience participation is invited.

15.30 Tea and coffee prior to journey home