



**114th Meeting of the British Neuropathological Society
Institute of Child Health, London**

**Symposium: "Advances in Motor Neuron Diseases"
13th March 2013**

Chair: Professor Paul Ince
University of Sheffield

13:15	Registration
14:00	Welcome - Paul Ince (University of Sheffield, UK)
14:15	Genetic characterisation of the motor neuron disease/frontotemporal dementia spectrum Vivianna van Deerlin (University of Pennsylvania, USA)
15:00	Molecular and clinical pathology of genetic variants in the motor neuron disease/ frontotemporal dementia spectrum Manuela Neumann (DZNE/University of Tübingen, Germany)
15:45	Tea break
16:15	Motor neuron differentiation in stem cell research on motor neuron disease Brian Kaspar (Ohio State University, USA)
17:00	Cellular and molecular mechanisms of disease pathogenesis in spinal muscular atrophy Thomas H. Gillingwater (University of Edinburgh, UK)
17:45	Alfred Meyer's Memorial Lecture Pamela Shaw (University of Sheffield, UK) Prospects for therapy development and CNS delivery in motor neuron diseases
18:30	RECEPTION

THURSDAY 14th MARCH 2013

08:00	Registration
08:50	Opening of meeting – Professor Seth Love
09:00 – 10:30	FIRST SCIENTIFIC SESSION – Neuropathology of ageing and dementia Chair: Professor Paul Ince
9:00	The need for precise subgrouping of tauopathies in the elderly 001 <i>G.G. Kovacs, I. Milenkovic</i>
9:15	Selective neuronal changes in prefrontal cortex of post-stroke dementia and ageing- related dementias 002 <i>V. Foster, A.E. Oakley, J. Slade, A. Khundakar, R.N. Kalaria</i>
9:30	Apolipoprotein E ε4 (apoE4) may impair perivascular elimination of amyloid-β (Aβ) 003 <i>C.A. Hawkes, J. Zekonyte, K. Howard, J.A.R. Nicoll, R.O. Weller, R.O. Carare</i>
9:45	The pattern of failure of perivascular elimination of amyloid-β in Alzheimer's disease 004 <i>N.I. Jayakody, M. Gatherer, B. Yuen, J. Attems, R.N. Kalaria C. Smith, J.A.R. Nicoll, R.O. Weller, R. O. Carare</i>
10:00	Diagnostic approach to argyrophilic grain disease (AGD): an UK BDR trial 005 <i>I. Bodi, C. Troakes, S. Maekawa, J. Attems, M.M. Esiri, T. Hortobagyi, A. King, S. Love, D. Mann, J.W. Neal, S. Al-Sarraj</i>

10:15	The role of brain biopsy in the diagnosis of neurodegenerative diseases <i>S. Al-Sarraj</i> , S. Maekawa, I. Bodi, C. Troakes, K. Ashkan, A. King	006
10:30 – 11:00	Coffee break	
11:00 – 12:30	SECOND SCIENTIFIC SESSION Chairs: Professor Tamas Revesz and Dr Johannes Attems– Lewy body diseases and movement disorders	
11:00	LRRK2 regulates autophagy <i>C. Manzoni</i> , A. Mamais, S. Dihanich, S. Tooze, R. Bandopadhyay, P.A. Lewis	007
11:15	Accumulation of alpha synuclein in the bowel in preclinical Parkinson's disease <i>D.A. Hilton</i> , M. Stephens, J. Zajicek, E. Broughton, H. Hagan, P. Edwards, C. Carroll	008
11:30	Exploring the pathogenesis of reduced occipital blood flow in dementia with Lewy bodies <i>H.A. Moulding</i> , J.S. Miners, T. Thomas, J.C. Palmer, S. Love	009
11:45	Parkin disease – a clinicopathological entity? <i>K.M. Doherty</i> , L. Silveira-Moriyama, L. Parkkinen, D.G. Healy, M. Farrell, N.E. Mencacci, Z. Ahmed, F.M. Brett, J. Hardy, N. Quinn, T.J. Counihan, T. Lynch, Z.V. Fox, T. Revesz, A.J. Lees, J. L. Holton	010
12:00	Aβ may promote the formation of Lewy bodies by increasing phosphorylation of α-synuclein at Ser129 <i>M.L.Swirski</i> , J.S. Miners, R. de Silva, S. Love	011
12:15	A choreiform movement disorder due to progressive supranuclear palsy <i>K.S.J. Allinson</i> , W. Scotton, S. Chaal, J. Rowe, D.G. O'Donovan	012
12:30	LUNCH	
13:00	POSTER DISCUSSION	
14:00	THE CAVANAGH PRIZE LECTURE Chair: Professor Seth Love Forget me not: investigating the pathophysiology and treatment of Alzheimer's disease <i>C. Hawkes</i>	
14:45	THIRD SCIENTIFIC SESSION – Paediatric and developmental pathology Chairs: Professor Brian Harding and Dr Thomas Jacques	
14:45	Phenotype-genotype correlation in spinal muscular atrophy: beyond the motor neuron <i>B. Harding</i> , W. Chung, U. Monani, R. Finkel	013
15:00	mTOR-dependent abnormalities in autophagy characterize human malformations of cortical development: evidence from focal cortical dysplasia and tuberous sclerosis <i>S. Yasin</i> , A. Ali, M. Tata, S.R. Picker, G. Anderson, S.L. Nicholson, J.H. Cross, W. Harkness, S.M.L. Paine, T.S. Jacques	014
15:15	The effect of germinal matrix haemorrhage on postnatal oligo/neurogenesis <i>W.J. Dawes</i> , S. Marino	015
15:30	Neuropathological substrates and cellular targets of human CMV in foetal brains <i>N. Teissier</i> , C. Fallet-Bianco, A.L. Delezoide, P. Marcocelles, A. Laquerrière, S. Khung-Savatovsky, Z. Csaba, T. Van Den Abbeele, P. Gressens, H. Adle-Biassette	016
15:45	Familial holoprosencephaly diencephalic hamartoma in Irish siblings <i>C. Keohane</i> , N. Bermingham, A. Greene, S. Ennis, J. Casey, P. McGettigan, T.J. Browne, O. O'Brien, E. Dempsey	017
16:00	Coffee break	
16:15	Business Meeting	
17:15 – 18:15	Professional Affairs Meeting	
19:30	Society Dinner at the Painters' Hall	
http://www.paintershall.co.uk/the-rooms/		

FRIDAY 15th MARCH 2013

9:00 – 10:30	FOURTH SCIENTIFIC SESSION - Neuroinflammation Chairs: Professor Margaret Esiri and Professor Kenneth Smith	
9:00	Tissue and cellular vulnerabilities to hypoxia: insight to the formation of demyelinating lesions in multiple sclerosis? <i>R.A. Desai</i> , K.J. Smith	018
9:15	A role for TNF signalling in cortical grey matter pathology in multiple sclerosis <i>R. Reynolds</i> , R. Magliozzi, C. Gardner, P. Durrenberger, F. Roncaroli, O. Howell	019
9:30	SIRT3 expression is reduced in non-lesional grey matter in multiple sclerosis <i>C.M. Rice</i> , E. Gray, K. Hares, K. Kemp, M. Sun, M. Ginty, S. Barizien, A. Wilkins, N.J. Scolding	020
9:45	Intracortical inflammation and Epstein-Barr virus infection in secondary progressive multiple sclerosis <i>R. Magliozzi</i> , B. Serafini, B. Rosicarelli, G. Chiappetta, C. Veroni, R. Reynolds, F. Aloisi	021
10:00	Efficacy of angiogenesis inhibitors in experimental autoimmune encephalomyelitis <i>C.J. MacMillan</i> , C.D. Doucette, D.W. Hoskin, A.S. Easton	022
10:15	Grey matter pathology in the cerebellum in multiple sclerosis is linked to inflammation in the subarachnoid space <i>O. Howell</i> , E.K. Schulz-Trieglaff, D. Carassiti, S.M. Gentleman, R. Nicholas, F. Roncaroli, R. Reynolds	023
10:30 – 11:00	Coffee break	
11:00 – 12:00	FIFTH SCIENTIFIC SESSION – Tumours of the central nervous system Chairs: Professor Silvia Marino and Professor Geoff Pilkington	
11:00	A national feasibility study of real-time central pathology review (CPR) and molecular diagnostics for medulloblastoma <i>S. Clifford</i> , S. Crosier, S. Nicholson, K. Robson, T.S. Jacques, S. Wharton, N. Bown, A. Michalsk, B. Pize	024
11:15	Structural variants and mutations in subsets of paediatric low-grade gliomas and glioneuronal tumours <i>D. Ellison</i> , G. Wu, B Tang, W. Orisme, C. PUNCHIHewa, R. Tatevossian, S Baker, J. Zhang	025
11:30	Bmi1 controls medulloblastoma invasion through inhibition of BMP-mediated cell adhesion <i>A. Merve</i> , S. Marino	026
11:45	Comparison of quantitative MGMT immunohistochemistry with a novel double-labelling protocol <i>E. Burke</i> , M. Grobler, K. Elderfield, F. Bond, M. Crocker, R. Taylor, L.R. Bridges	027
12:00	Diagnostic Slide Session (incorporating EQA)	
13:00	LUNCH	
13:30	POSTER DISCUSSION	
14:30 – 16:15	SIXTH SCIENTIFIC SESSION – Miscellaneous Chair: Professor Seth Love	
14:30	Brain microvascular accumulation and distribution of GOM and the NOTCH3 ectodomain in CADASIL Y. Yamamoto, L.J.L. Craggs, A. Watanabe, J. Attems, R.W.C. Low, A.E. Oakley, <i>R.N. Kalaria</i>	028
14:45	Detection of protease sensitive PrPSc in the cerebral cortex of fatal familial insomnia patients <i>A.H. Peden</i> , G. Fairfoul, J.W. Ironside, M.W. Head	029
15:00	Filamentous white matter prion protein deposition is a distinctive feature of multiple inherited prion diseases <i>L. Reiniger</i> , I. Mirabile, A. Lukic, J. Linehan, M. Groves, P. Rudge, J. Collinge, S. Mead, S. Brandner	030

15:15	Uncovering early markers of Parkinson's disease pathological progression using proteomics <i>S.N. Pressey, C. E. Murray, W. Heywood, S.M. Gentleman, K. Mills, J.L. Holton, S. Gandhi, T. Revesz</i>	O31
15:30	Degradation of α-synuclein in dementia with Lewy bodies <i>R. Renfrew, J.S. Miners, S. Love</i>	O32
15:45	Glycogen metabolism pathway defects produce gastrointestinal myopathy with polyglucosan bodies: an explanation for the neuropathology and symptoms of gastrointestinal neuromuscular disorder <i>J.E. Martin, Y. Duraisamy, M. West, D.A.T.M. Chowdhury, M. Rahman, P. Rowberry, J. Broad, B. Minassian, S. DiMauro, H.O. Akman, C.H. Knowles, D.B.E. Silk</i>	O33
16.00	Mast cell upregulation in inclusion body myositis: a role for neurogenic inflammation? <i>S. Brady, J. Mack, L. Bindoff, D. Hilton-Jones, H. Miletic, W. Squier</i>	O34
16:15	Announcement of the poster prize winners and close of the meeting	

POSTERS

	A C9orf72 expansion mutation identified in a control case without associated pathology <i>C. Troakes, A. King, B.N. Smith, Y. Lee, S. Maekawa, I. Bodi, C.E. Shaw, S. Al-Sarraj</i>	P01
	IDH mutation analysis in gliomas as a diagnostic and prognostic biomarker <i>K.M. Kurian, H.R. Haynes, C. Crosby, K. Hopkins, M. Williams</i>	P02
	Diagnosis of primary cerebral lymphomas - possible role of PCR prior to rebiopsy <i>A. Shaw, V. Iyer, N. Rooney, R. Wragg, P. Waits, E. Roberts, H. Haynes, K.M. Kurian</i>	P03
	Combined tau, TDP-43 and p62 pathology in a case of FTL associated with both a C9ORF72 repeat expansion and a usually "benign" p.Ala239Thr MAPT (tau) variant <i>A. King, S. Al-Sarraj, C. Troakes, B. Smith, S. Maekaw, M. Iovino, M.G. Spillantini, C. Shaw</i>	P04
	Kallmann syndrome-associated protein anosmin-1 contributes to brain tumour malignancy <i>C.T. Choy, H. Kim, J. Lee, D.M. Williams, D. Palethorpe, K. Laing, L.R. Bridges, F. Howe, S.H. Kim</i>	P05
	Endothelial expression of the tight junction protein Claudin-5 in sporadic cerebral small vessel disease and aged controls <i>C.H.L. Khoong, A.H. Hainsworth, L.R. Bridges</i>	P06
	Clinical and pathological features of frontotemporal dementia linked to C9ORF72 homozygosity <i>T. Lashley, J.D. Rohrer, C.J. Mahoney, J.M. Polke, N.S. Ryan, A. Isaacs, S. Mead, J.D. Warren, T. Revesz</i>	P07
	The Ki-67 labelling index with maximally selected cut-offs is better at predicting recurrence than the WHO grading system of meningiomas <i>R. Laxton, L. Doey, A. Murphy-Stout, E. Davis, R. Bhangoo, R. Gullan, K. Ashkan, S. Al-Sarraj</i>	P08
	The roles of D-serine and D-amino acid oxidase in ALS pathogenesis <i>P. Paul, T. Murphy, Z. Oseni, S. Sivalokanathan, J. de Belleruche</i>	P09
	The balloon cell vascular niche: anatomical evidence of a stem cell-blood vessel relationship in epilepsy-associated malformations of cortical development <i>M.A. Hollingworth, S.M.L. Paine, K. Latak, A. Ganapathi, K. Miller, F. Becherini, J.H. Cross, W. Harkness, B.N. Harding, T.S. Jacques</i>	P10
	APOJ is a reliable marker for traumatic brain injury <i>S. Al-Sarraj, F. Noor, C. Troakes, A. King, G. Ruttly, N. Cary</i>	P11
	Histopathology and clinical characteristics of patients undergoing surgery for refractory epilepsy <i>A. Varghese, P. Chumas, J. Goodden, P. Goulding, A. Ismail</i>	P12
	Establishment and characterization of a novel tumor stem cell-based glioblastoma invasion model <i>S.S. Jensen, M. Meyer, B. Halle, A.M. Rosager, B.W. Kristensen</i>	P13
	Cannabinoid receptor expression in Parkinson's disease <i>D.A. Hilton, M.L. Zeissler, H. Zhang, J. Zajicek, J. Hicks, J. Eastwood, C. Carroll</i>	P14
	Activation of multiple signalling pathways in meningiomas is unrelated to NF2 gene status <i>D.A. Hilton, A. Shivane, L. Turner, S. Ammoun, O. Hanemann</i>	P15
	β-catenin in sporadic inclusion body myositis <i>C.Y. Shim, C. Marini Bettolo, P. Singh, W. Rakowicz, R.J. Lane, F. Roncaroli</i>	P16
	Cognitive impairment in multiple system atrophy: a clinico-pathological study <i>Y.T. Asi*, H. Ling*, Z. Ahmed, A.J. Lees, T. Revesz, J.L. Holton (*Shared first authors)</i>	P17

Oxidative stress and DNA damage in cerebral white matter lesions of the ageing human brain	P18
<i>S.A. Al-Mashhadi, J.E. Simpson, P.R. Heath, M.J. Dickman, P.G. Ince, S.B. Wharton</i>	
Relationship of Apolipoprotein E to Parkinson's disease-related alpha-synuclein pathology	P19
<i>L. Parkkinen, C. Joachim, M.M. Esiri, O. Ansorge</i>	
A previously unrecognised fatal bovine tauopathy is present in aged cattle throughout the UK	P20
<i>M. Jeffrey, P. Piccardo, G. McGovern</i>	
Association of MAPT (tau) haplotype with the pathology of Parkinson's disease	P21
<i>L. Parkkinen, C. Joachim, M.M. Esiri, O. Ansorge</i>	
GM2 -gangliosidosis in British Jacob sheep	P22
<i>M.E. Wessels, J.P. Holmes, M. Jeffrey, M. Jackson, S.F.E. Scholes</i>	
Organising intradural haemorrhage in a 'Sudden Unexpected Infant Death' baby	P23
<i>C. Keohane, M.A. Bolster, N. Bermingham</i>	
Synucleinopathy with a G51D a-synuclein mutation: a neuropathological and genetic study	P24
<i>A.P. Kiely, Y.T. Asi, E. Kara, P. Limousin, H. Ling, P. Lewis, C. Proukakis, N. Quinn, A.J. Lees, J. Hardy, T. Reves, H. Houlden*, J.L. Holton* (* Shared senior authors)</i>	
Cerebellar and brainstem glial pathology in differentiation of tremor dominant and akinetic-rigid dominant Parkinson's disease: a multivariate approach	P25
<i>M. Kokkinou, R.K.B. Pearce, S.M. Gentleman</i>	
Axonal degeneration secondary to cortical neurodegeneration; a potential cause for white matter lesions?	P26
<i>K.E. McAleese, M. Firbank, S. McParland and J. Attems</i>	
Quantification of pathology in mixed Alzheimer's disease and Lewy body dementia	P27
<i>L. Walker, A.J. Thomas, C.M. Morris, J. Attems</i>	
Tissue microarray in the quantification of hyper-phosphorylated tau, amyloid-beta and alpha-synuclein	P28
<i>S. McParland, K. McAleese, L. Walker, M. Johnson, E. Fielder, I. Knowles and J. Attems</i>	
Patterns of hippocampal sclerosis in children	P29
<i>S.M.L. Paine, D.N.R. Bell, V. Melchionda, J. Tan, C. Weereratne, S. Harrison, F. Vargha-Khadem, W. Harkness, J.H. Cross, B.N. Harding, L. Pizarro, T.S. Jacques</i>	
Sparganosis in East Anglia	P30
<i>K.S.J. Allinson, H.P. Mok, N.M. Antoun, M. Berriman, N. Holroyd, H.M. Bennett, I.J. Tsai, M.R. Manford, S.J. Price, T.S. Santarius, E. Gkrania-Klotsas, A.J. Carmichael, P.L. Chiodini, S.B. Lucas, A.F. Dean</i>	
Post-mortem findings in embryonal tumour with abundant neuropil and true rosettes (ETANTR): implications for biopsy diagnosis	P31
<i>S.M.L. Paine, H. Kempinski, W. Harkness, A. Mazumder, K. Mankad, F. Craig, D. Hargrave, T.S. Jacques</i>	
A highly unusual congenital developmental neuroepithelial cyst of the orbit	P32
<i>S.M.L. Paine, M. Shanmuganathan, N.U.O. Jeelani, T.S. Jacques</i>	
Elevated VEGF in the cerebral cortex in Alzheimer's disease is not attributable to arteriosclerotic small vessel disease or cerebral amyloid angiopathy	P33
<i>T.L. Thomas, P.G. Kehoe, S. Love</i>	
Rhabdoid meningioma in a young child: post-mortem findings of a widely disseminated and aggressive tumour	P34
<i>S.M.L. Paine, J. Ternier, D.N.P. Thompson, A. Mazumder, K. Manka, F. Craig, D. Hargrave, T.S. Jacques</i>	
The aggregate proteins of FTL-D-FUS are re-localised to stress granules upon oxidative stress	P35
<i>J. Brelstaff, A. Mamais, T. Lashley, J.L. Holton, T. Revesz, R. Bandopadhyay</i>	
Bmi1 controls proliferation and maintenance of satellite cells and plays an important role in muscle regeneration	P36
<i>V. Di Foggia, X. Zhang, L.G. Robson, S. Marino</i>	
Central nervous system recurrence of acute myeloid leukaemia after allogeneic haematopoietic stem cell transplantation	P37
<i>O.E. Curran, S. Al-Sarraj S, S. Pomplun</i>	
A case of progressive encephalopathy with anti-glutamic acid decarboxylase antibodies and subacute cerebellar degeneration	P38
<i>O.E. Curran, N. Munro, N. Moran, I. Bodi</i>	
A study of gliogenesis in the vicinity of depth electrode tracts	P39
<i>J. Goc, J. Liu, S. Sisodiya, M. Thom</i>	
A comparative study of the dentate gyrus in hippocampal sclerosis in epilepsy and dementia	P40
<i>R. Bandopadhyay, J. Liu, S.M. Sisodiya, M. Thom</i>	

Clinical value of the cancer stem cell-related marker Musashi-1 in gliomas	P41
<i>R.D. Dahlrot, S. Hansen, H.D. Schröder, J. Hjelmberg, B.W. Kristensen</i>	
Using the Anterior Olfactory Nucleus to study Lewy pathology in olfactory structures	P42
<i>S. Rajan, R. Bandopadhyay, A. Kingsbury, H. Ayling, W. Sterlacci, W. Poewe, H. Maier, M.-T. H. Ezquerro, A.J. Lees, T. Revesz, L. Silveira-Moriyama</i>	
Magnetic resonance spectroscopy for 2-hydroxyglutarate identifies IDH mutant gliomas	P43
<i>S.J. Larkin, J. Near, M. Kudari, A. Khrapitchev, N. Sibson, P. Jezzard, I. Pfeffer, K. Al-Qahtani, C. Schofield, J. McCullagh, O. Ansorge</i>	
A pathological basis for the association of parkinsonism and amyotrophic lateral sclerosis	P44
<i>J.R. Highley, J. Cooper-Knock, A. Frolov, G. Charlesworth, J. Kirby, S.B. Wharton, P.G. Ince, P.J. Shaw, N. Wood, O. Bandman</i>	
Epstein-Barr virus-associated smooth muscle tumour presenting as a multifocal brain tumour	P45
<i>Z. Hanzely, A. Kulla, W. Stewart</i>	
Spontaneous malignant transformation of vestibular schwannoma	P46
<i>S. Nagaraju, G. Kulkarni</i>	
Somatic variation of the ataxin-2 CAG repeat in motor neuron disease	P47
<i>S. East, B. Tseu, A. Clift, K. Talbot, O. Ansorge</i>	
Mild malformations of cortical development (type II) in temporal lobe epilepsy	P48
<i>H. Brooke-Ball, M. Tachrount, S. Sisodiya, M. Thom</i>	
Neuropathological changes in Alpers' syndrome	P49
<i>H. Bogle, N.Z. Lax, E Jaros, R McFarland, R.W. Taylor, D.M. Turnbull</i>	
Cerebral Amyloidoma: a case report	P50
<i>A. Shivane, D.A. Hilton, P. Whitefield</i>	