



120th Meeting of the British Neuropathological Society
Mary Ward House Conference and Exhibition Centre, 5-7 Tavistock Place, London WC1H 9SN

WEDNESDAY 6th March, 2019

Developmental Neuropathology Symposium

Chair: Professor Tom Jacques, UCL Great Ormond Street Institute of Child Health, UK

12:30 Registration

13:25 **Welcome and Introduction** - Tom Jacques

13:30 **Interneurons in Cerebral Cortical Developmental Disorders**

Jeffrey Golden, Brigham and Women's Hospital and Harvard Medical School Boston, USA

14:15 **Leukodystrophies**

Marianna Bugiani, VU Medical Center, Amsterdam, The Netherlands

15:00 Tea Break

15:30 **Neurodegeneration with Brain Iron Accumulation (NBIA): Genetics, Neuropathology and Novel Treatments**

Manju Kurian, UCL Great Ormond Street Institute of Child Health, UK

16:15 **Preterm brain injury: mechanistic insights from magnetic resonance imaging**

James Boardman, University of Edinburgh, UK

17:00 Short Break

17:15 **Alfred Meyer Memorial Lecture - Neural Tube Defects**

Andrew Copp, UCL Great Ormond Street Institute of Child Health, UK

18:15 Reception

THURSDAY 7th March, 2019

08:00 - Registration

08:50 - Opening of meeting – Professor Colin Smith, BNS President

09:00 - 10:30 FIRST SCIENTIFIC SESSION – cerebrovascular disease

Chairs: Stephen Wharton, Roxana Carare

9:00 A. Willetts, J. Booker, M. Gatherer, N. Albargothy, A. Verma, R. Weller, R. Carare, C. Hawkes
Pathways for flow of solutes along walls of cerebral arteries are maintained even in the presence of Cerebral Amyloid Angiopathy O01

9:15 L. Bridges, L. Kenyon, M. Esiri, D. Bennett, A. Hainsworth
Mural cell depletion and senescence in cerebral small vessel disease: a neuropathology study in older people (aged 80+) O02

9. 30 M. Goldfinger, B. Tilley, M. Sastre, S. Gentleman
A tale of two tauopathies: a comparison of vasculature changes in ARTAG and chronic traumatic encephalopathy O03

9:45 M. Frost, A. Sealy, M. Gatherer, C. Smith, J. Attems, R. Weller, R. Carare
Loss of adrenergic receptors on cerebral blood vessels may contribute to the accumulation of Amyloid- β in Cerebral Amyloid Angiopathy O04

10:00 H. Tayler, R. MacLachlan, I. Schulz, O. Skrobot, S. Miners, S. Love
Post-mortem biochemical markers of vascular dysfunction: associations with midlife hypertension, VCING, endothelin- 1 and dementia O05

10:15 D. Hansen, T. Lashley, J. Holton, T. Warner
Cerebral amyloid angiopathy in Lewy body dementias O06

10:30 – 11:00 Coffee break

11:00 - 12:15 SECOND SCIENTIFIC SESSION – Alzheimer's disease

Chairs: Delphine Boche, Johannes Attems

11:00 C. Toomey, S. Foti, W. Heywood, K. Mills, T. Lashley
Proteomic differences between Alzheimer's disease and Frontotemporal dementia human post-mortem brains O07

11:15 H. Mistry, J. Simpson, A. Chambers, C. Garwood, P. Ince, P. Heath, S. Wharton
Cholesterol dysregulation in the neurone-astrocyte unit as an early contributor to neuronal dysfunction in Alzheimer's Disease O08

11:30 Z. Jaunmuktane, A. Quaegebeur, R. Taipa, S. Mead, S. Brandner
Human transmission of amyloid- β cerebral amyloid angiopathy O09

11:45 K. McAleese, S. Graham, M. Dey, L. Walker, D. Erskine, M. Johnson, A. Thomas, McKeith, C. DeCarli, J. Attems
Fibrinogen leakage in the white matter of Alzheimer's disease and normal aged

	brains: Implications for fibrinogen as a biomarker	O10
12:00	<u>R. Waller</u> , R. Narramore, J. Simpson, F. Matthews, C. Brayne, P. Ince, R. Kalaria, S. Wharton Heterogeneity of Cellular Inflammatory Responses in Ageing White Matter and Relationship to Alzheimer's and Small Vessel Disease Pathologies	O11
12:15 – 12:45 - Lunch		
12:45 –14.00 POSTER DISCUSSION		
14.00-15.45 THIRD SCIENTIFIC SESSION – Neurodegeneration		
Chair: Seth Love, Stephen Gentleman		
14:00	<u>M Foiani</u> , T. Jackson-Morgan, C. Cicognola, N. Ermann, K. Ye, J. Kornhuber, N. Fox, P. Lewczuk, H Zetterberg, K. Blennow, K. Hoglund ³ , J. Rohrer ⁵ , T. Lashley Profiling of tauopathies in frontotemporal dementia	O12
14:15	<u>S. Kaalund</u> , K. Allinson, M. Spillantini, J. Rowe Quantification of neurons in the inferior frontal gyrus in frontotemporal lobar degeneration	O13
14:30	<u>B. Tilley</u> , M. Goldfinger, R. Pearce, S. Gentleman The role of tau and basal ganglia cholinergic pathology in the pathogenesis of Parkinson's disease motor subtypes	O14
14:45	B. Sonustun, C. Strand, S. Foti, T. Warner, H. Lashuel, J. Holton, <u>R. Bandopadhyay</u> Investigation of alpha-synuclein post-translational modifications in idiopathic Parkinson's disease and multiple system atrophy	O15
15:00	<u>C. Bettencourt</u> , S. Foti, T. Lashley, R. Balazs, E. Vire, J. Holton DNA methylation landscapes in patients with Multiple System Atrophy	O16
15:15	<u>C. Appleby-mallinder</u> , P. Heath, R. Highley DNA methylation in Amyotrophic Lateral Sclerosis (ALS)	O17
15:30	<u>Richard Cain</u> , Laura Palmer, Seth Love UK Brain Banks Network Database: a resource for researchers and brain banks	O18
15:45 – 16:15 Break		
16:15 – 17:00 BUSINESS MEETING		
17:00 – 18:00 PROFESSIONAL AFFAIRS MEETING		
19:30 SOCIETY DINNER AT THE CRYPT		

FRIDAY 8th MARCH 2019

08:00 registration

9:00 – 10:30 FOURTH SCIENTIFIC SESSION – Brain tumours

Chairs: Sebastian Brandner, Kathreena Kurian

- 9:00 S. Nagaraju, *U. Pohl*
Characterisation of gliomas with rare IDH mutation at QEHB during 2016-2018 O19
- 9:15 K. Loveson, P. Singh, K. Allinson, *H. Fillmore*
Paediatric brainstem glioma and its intersection with brain development: role of tumour microenvironment O20
- 9:30 I. Smolicz, A. Fairchild, J. Pickles, T. Stone, J. Chalker, J. Gonzalez Zapata, L. Wilkhu, S. Yasin, D. Hargrave, N. Sebire, T. Jacques
The biology of paediatric brain tumours at post-mortem: a national cohort O21
- 9:45 *J. Pickles*, T. Stone, L. Brownlee, A. Merve, S. Yasin, L. Wilku, M. Kristiansen, D. Hargrave, J Chalker, T. Jacques
DNA methylation profiling in paediatric CNS tumours O22
- 10:00 *L. Laraba*, S. Ferluga, E. Ercolano, C. Adams, A. Shivane, P. Edwards, M. Futschik, V. Lenis, J. Grimm de Guibert, S. Moller, O. Hanemann, D. Parkinson
The Cancer Stem Cell Marker ALDH1A1 is Upregulated and Drives Proliferation in Merlin Null Meningioma and Schwannoma O23
- 10:15 *O. Curran*, L. Gilroy, A.Torgersen, C. Smith, W. Al-Qsous
Intracerebral lymphoid malignancies: a retrospective study of morphological, immunohistochemical and molecular characteristics with survival correlation O24

10:30 – 11:00 Coffee break

11:00 – 12:15 FIFTH SCIENTIFIC SESSION- Neuroinflammation

Chairs: Colin Smith, Robin Highley

- 11:00 *J. Spencer*, J. Watson, G. Niblett, Y. Mahjoub, R. Yates, G. Hadley, C. Brendler-Spaeth, B. Kessler, R. Fischer, G. DeLuca
Extracellular matrix proteins interface with HLA genotype to modulate topographical variation of multiple sclerosis pathology O25
- 11: 15 *B. Ashford*, C. Appleby-Mallinder, P. Heath, J. Simpson, R. Highley
The Role of Immunity in Human Motor Neuron Disease O26
- 11:30 *D. Munoz*, M. Abdollahi
The Structural Basis of Susac's Syndrome O27
- 11:45 *R. Raha-chowdhury*, J. Henderson, A. Raha, E. Jones, R. Fincham, K. Allinson, A. Holland, S. Zaman
Choroid plexus acts a gatekeeper for TREM2, abnormal accumulation of ApoE and fibrillary tau in Alzheimer's disease and Down's syndrome O28

12.00 – 12:30 – Lunch

12:30 – 13:15 - POSTER DISCUSSION

13:15 – 14:00: Clinical update session:

Tumours of the sellar region - update on classification and new entities

Federico Roncaroli

14:00 – 14:30 DIAGNOSTIC SLIDE SESSION (INCORPORATING EQA)

14:30 – 15:00 break

15:00 – 16:15 SIXTH SCIENTIFIC SESSION – developmental neuropathology, epilepsy and skeletal muscle

Chairs: Janice Holton, Maria Thom

- 15:00 L. Chareyron, D. Gadian, F. Vargha-Khadem
Amygdala development in patients with hippocampal atrophy: associations with socioemotional status O29
- 15.15 K. Long, B. Newland, M. Florio, N. Kalebic, B. Langen, A. Kolterer, P. Wimberger, W. Huttner
How does the human neocortex fold? A novel role of the Extracellular Matrix O30
- 15.30 A. Sarkozy, S. Torelli, P. Ala, D. Ardicli, L. Feng, R. Mein, S. Aguti, H. Zhou, C. Sewry, R. Phadke, F. Muntoni
Quantitative flow cytometry in the diagnosis of Collagen VI-related disorders: 3-year experience from a tertiary referral centre O31
- 15.45 D. Chambers, A. Kumar, L. Feng, I. Hargreaves, A. Lam, S. Heales, A. Manzur, F. Muntoni, C. Sewry, J. Poulton, R. Phadke
A novel multiplex chromogenic immunoassay for evaluating mitochondrial respiratory chain complex I and complex IV defects in diagnostic muscle biopsies O32
- 16.00 J. Liu, M. Ellis, M. Thom, S. Sisodiya
Cellular expression of SCN1A mRNAs in patients with temporal lobe epilepsy O33
- 16.15 Announcement of the poster prize winners and closure of the meeting

POSTERS

- Microglia in cortical and subcortical autonomic brain regions in SUDEP**
M. Thom, A. Somani, S. Sisodiya P01
- Volumetric and neuropathological study of the medulla in SUDEP and correlates with 9.4T MRI**
M. Thom, S. Patodia, S. Sisodiya P02
- Medullary catecholaminergic neurones in Sudden Unexpected Death in Epilepsy**
M. Thom, S. Patodia, I. Tan, S. Sisodiya P03
- Adenosine kinase and Adenosine receptors A1 and A2A in temporal lobe epilepsy and association with risk factors for SUDEP**
M. Thom, B. Paradiso, S. Patodia, M. Garcia, B. Diehl, M. Ellis, O. Devinsk P04
- Identification of miRNA and mRNA regulatory networks in the ageing blood-brain barrier: Comparative gene expression studies in human and mouse**
V. Leach, E. Goodall, J. Cooper-Knock, C. Wang, J. Simpson, D. Baker, D. Drew, M. Saffrey, I. Romero, P. Heath, S. Wharton P05
- The effect of systemic atherosclerosis in the neurovascular unit**
M. Alejandra Rebollar, S. Wharton, S. Francis, J. Simpson P06
- Identification of miRNA and mRNA regulatory networks in the ageing blood-brain barrier: Comparative gene expression studies in human and mouse**
V. Leach, E. Goodall, J. Cooper-Knock, C. Wang, J. Simpson, D. Baker, D. Drew, M. Saffrey, I. Romero, P. Heath, S. Wharton P07
- Carotid Artery Disease, Strokes and Experimental Effects of Enriched Environment on Stroke Injury**
Y. Hase, T. Polvikoski, M. Hase, W. Stevenson, M. Ihara, L. Allan, K. Horsburgh, R. Kalaria P08
- Adrenergic receptors in the walls of cerebral vessels as possible targets for improving Intramural Peri-Arterial drainage in CAA**
A. Keable, D. Baseley, D. Zbarcea, M. Gatherer, D. Johnston, C. Smith, J. Attems, R. Weller, R. Carare P09
- Changes in the Intramural Peri-Arterial Drainage (IPAD) pathways after traumatic brain injury**
V. Unadkat, M. Gatherer, A. Pringle, A. Ahmed, R. Weller, R. Carare P10
- Iba-1-/CD68+ microglia are a prominent feature of age-associated deep subcortical white matter lesions**
S. Waller, L. Baxter, D. Fillingham, S. Coelho, J. Pozo, M. Mozumder, A. Frangi, P. Ince, J. Simpson, R. Highley P11

- Microglial Dynamics in the Developing and Early Postnatal Human Brain**
D. Menassa, L. Barry-Carroll, J. Nicoll, M. Chapman, T. Bloom, S. Lisgo, I. Adorjan, Z. Krsnik, I. Kostovic, T. Jacques, O. Ansorge, D. Gomez-Nicola P12
- A versatile, modular digital script for automated high-throughput multiparametric myofibre analysis in brightfield and epifluorescent paradigms**
M. Ellis, D. Scaglioni, F. Catapano, V. Sardone, D. Chambers, L. Feng, E. Curtis-Wetton, S. Saeed, A. Sigurta, N. Hill, M. Singer, C. Sewry, S. Torelli, J. Morgan, F. Muntoni, R. Phadke P13
- Recessive loss-of-function mutations in ITGA7 cause cardiac arrhythmia with or without structural cardiomyopathy and respiratory muscle weakness**
E. Bugiardini, R. Phadke, R. Maas, A. Pittman, B. Kusters, J. Morrow, M. Parton, A. Nunes, M. Akhtar, P. Syrris, L. Lopes, T. Fotelonga, H. Houlden, P. Elliott, M. Hanna, J. Raaphorst, D. Burkin, E. Matthews P14
- Congenital fatal cap-rod myopathy due to a de novo autosomal dominant pathogenic ACTA1 variant**
R. Phadke, B. Herron, D. Hurrell, S. Craig, B. Kelly, R. Mein, A. Sarkozy, C. Sewry, F. Muntoni, V. McConnell P15
- Extended phenotypic spectrum of VCP inclusion body myopathy: report of two cases with atypical early and late childhood-onset disease**
R. Phadke, A. Dean, M. Evans, A. Parker, D. Maxwell, C. Sewry, A. Sarkozy, F. Muntoni P16
- Diagnostic challenges in paediatric anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase necrotising myopathy (anti-HMGCR-NM)**
R. Phadke, M. Pal-Magdics, C. Pilkington, S. Maltby, A. Ismail, A. Chakrabarty, P. Munot, M. Wood, A. Manzur, F. Muntoni, A. Sarkozy P17
- A novel case of MSTO1-related congenital muscular dystrophy with cerebellar involvement**
D. Ardicli, I. Zaharieva, C. Deshpande, I. Bodi, A. Siddiqui, J. Marie U-King-Im, R. Phadke, A. Sarkozy, H. Jungbluth, F. Muntoni P18
- Prevalence of cytoplasmic bodies in a large series of diagnostic paediatric muscle biopsies**
M. Aizpurua Gomez, L. Feng, F. Leiva-Cepas, A. Sarkozy, A. Manzur, F. Muntoni, C. Sewry, R. Phadke P19
- SLONM: Sporadic Late-Onset Nemaline Myopathy. A case study and review of neuropathological findings.**
C. Turnquist, M. Hofer, D. Hilton-Jones P20
- Investigating senescence activation in response to oxidative DNA damage in neurones in vitro**
I. Vazquez-Villasenor, C. Garwood, P. Heath, J. Simpson, S. Wharton P21
- Advanced molecular characterization using Digital Spatial Profiling of immuno-oncology target expression in methylated versus unmethylated IDH-wildtype glioblastoma**
H. Barber, B. Lander, A. Daniels, A. Tofias, J. Gong, X. Ren, Y. Ren, P. White, K. Kurian P22
- Modelling high-risk paediatric brain tumour infiltration**
A. Fairchild, A. Rolland, Y. Li, T. Stone, J. Pickles, T. Jacques P23
- Characterisation of histone mutant gliomas in adults at QEHB 2015-2018**
U. Pohl, S. Nagaraju P24
- EGFR as a potential prognostic biomarker in adult IDH-wildtype Glioblastomas**
C. Cabral, R. Laxton, L. Doey, I. Bodi, A. King, L. Brazil, R. Bhangoo, K. Ashkan, S. Al-Sarraj P25

A rare case of fibroblastic reticular cell tumour in the spine <u>Z. Reisz</u> , J. Salisbury, D. Bell, I. Bodi, S. Al-Sarraj	P26
Audit of turnaround time (TAT) in glioma diagnosis Oliver Sargent	P27
Cerebral small vessel disease in multiple sclerosis <u>R. Geraldes</u> , M. Esiri, J. Palace, G. DeLuca	P28
Axonal protection by teriflunomide in an experimental lesion of Pattern III demyelination R. Desai, A. Davies, M. Tachrout, X. Golay, <u>K. Smith</u>	P29
Are visual hallucinations in Parkinson's disease a result of decreased perfusion of visual processing areas of the brain? <u>L. Sinclair</u> , J. Brenton, A. King Lun Liu, S. Gentleman, S. Love	P30
Do anti-cholinergic drugs increase Alzheimer-type pathology in Parkinson's patients? A retrospective post-mortem investigation <u>A. King Lun Liu</u> , Y. Mun Lim, R. Pearce, S. Gentleman	P31
Can early tau depositions in mixed Alzheimer's disease and Lewy body disease give insights into disease progression? <u>L. Walker</u> , E. Thomson, M. Thamed, K. McAleese, M. Johnson, J. Attems	P32
Serotonergic ¹²³I-FP-CIT binding is associated with depression and not neuropathology <u>A. Oliver-Evans</u> , S. Colloby, A. Thomas, D. Erskine, J. Attems	P33
White Matter Hyperintensities and Pathological Correlations in Alzheimer's Disease <u>P. Walsh</u> , D. Thomas, C. Sudre, J. Iglesias, S. Crampsie, Z. Jaunmuktane, J. Holton, N. Ryan, J. Barnes, T. Lashley	P34
DNA hydroxymethylation in Amyotrophic Lateral Sclerosis (ALS) <u>E. Schaber</u> , C. Appleby-Mallinder, R. Highley, P. Heath	P35
Increased calcyclin immunoexpression in ALS is seen within reactive astrocytes in corticospinal tracts and is not specific for ALS subtype <u>A. King</u> , C. Troakes, C. Shaw, V. Marchica, S. Al-Sarraj, B. Smith	P36
CLIPPERS: Chronic Lymphocytic Inflammation with Pontine Perivascular Enhancement Responsive to Steroids. A case study and review of neuropathological findings. <u>C. Turnquist</u> , M. Hofer, J. Halliday, R. Kerr	P37
A novel BMI1/Ephrin connection in human glioblastomas <u>T. Millner</u> , B. Ricci, X. Zhang, N. Pomella, G. Rosser, S. Marino	P38
Clinical and pathological features of multiple system atrophy and multiple system atrophy lookalikes <u>Y. Miki</u> , H. Ling, S. Foti, J. Holton	P39
Investigation of tau seeding activity in tauopathies <u>T. Bradshaw</u> , H. Ling, J. Holton, S. Wray, R. de Silva, T. Revesz	P40
The expression and presence of RNA binding proteins in FTLD-TDP <u>S. Foti</u> , L. Gittings, C. Toomey, Y. Asi, T. Lashley	P41
A Single cell omics approach to neurodegeneration <u>L. Greensmith</u>	p42