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

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
<table>
<thead>
<tr>
<th>Time</th>
<th>Session/Activity</th>
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| 12:00      | R. Waller, 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 |
| 12:15 – 12:45 | Lunch                                                      |
| 12:45 – 14:00 | 14.00 POSTER DISCUSSION                                    |
| 14:00 – 15:45 | THIRD SCIENTIFIC SESSION – Neurodegeneration                |
| Chair: Seth Love, Stephen Gentleman |
| 14:00      | M Foiani, T. Jackson-Morgan, C. Cicognola, N. Ermann, K. Ye, J. Kornhuber, N. Fox, P. Lewczuk, H Zetterberg, K. Blennow, K. Hoglund, J. Rohrer, T. Lashley  
             | Profiling of tauopathies in frontotemporal dementia          |
| 14:15      | S. Kaalund, K. Allinson, M. Spillantini, J. Rowe        |
| 14:30      | B. Tilley, M. Goldfinger, R. Pearce, S. Gentleman     |
| 14:45      | B. Sonustun, C. Strand, S. Foti, T. Warner, H. Lashuel, J. Holton, R. Bandopadhyay  
             | Investigation of alpha-synuclein post-translational modifications in idiopathic Parkinson's disease and multiple system atrophy |
| 15:00      | C. Bettencourt, S. Foti, T. Lashley, R. Balazs, E. Vire, J. Holton  
             | DNA methylation landscapes in patients with Multiple System Atrophy |
| 15:15      | C. Appleby-mallinder, P. Heath, R. Highley             |
| 15:30      | Richard Cain, Laura Palmer, Seth Love                  |
|            | UK Brain Banks Network Database: a resource for researchers and brain banks |
| 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                              |
| 20:00      | 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**  
The biology of paediatric brain tumours at post-mortem: a national cohort  
*O21*

**9:45**  
DNA methylation profiling in paediatric CNS tumours  
*O22*

**10:00**  
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-Qous  
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**  
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**  
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

   *How does the human neocortex fold? A novel role of the Extracellular Matrix*  O30

   *Quantitative flow cytometry in the diagnosis of Collagen VI-related disorders: 3-year experience from a tertiary referral centre*  O31

   *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

Volumetric and neuropathological study of the medulla in SUDEP and correlates with 9.4T MRI
*M. Thom*, S. *Patodia*, S. Sisodiya

Medullary catecholaminergic neurones in Sudden Unexpected Death in Epilepsy
*M. Thom*, S. *Patodia*, I. Tan, S. Sisodiya

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

Identification of miRNA and mRNA regulatory networks in the ageing blood-brain barrier: Comparative gene expression studies in human and mouse

The effect of systemic atherosclerosis in the neurovascular unit
*M. Alejandra Rebollar*, S. Wharton, S. Francis, J. Simpson

Identification of miRNA and mRNA regulatory networks in the ageing blood-brain barrier: Comparative gene expression studies in human and mouse

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

Adrenergic receptors in the walls of cerebral vessels as possible targets for improving Intramural Peri-Arterial drainage in CAA

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

Iba-1-/CD68+ microglia are a prominent feature of age-associated deep subcortical white matter lesions
Microglial Dynamics in the Developing and Early Postnatal Human Brain
P12

A versatile, modular digital script for automated high-throughput multiparametric myofibre analysis in brightfield and epifluorescent paradigms
P13

Recessive loss-of-function mutations in ITGA7 cause cardiac arrhythmia with or without structural cardiomyopathy and respiratory muscle weakness
P14

Congenital fatal cap-rød myopathy due to a de novo autosomal dominant pathogenic ACTA1 variant
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)
P17

A novel case of MSTO1-related congenital muscular dystrophy with cerebellar involvement
D. Ardicli, I. Zaharieva, C. Deshpande, I. Bodí, 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

SLOMN: 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. Vozquez-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 QEH 2015-2018
U. Pohl, S. Nagaraju
P24

EGFR as a potential prognostic biomarker in adult IDH-wildtype Glioblastomas
C. Cabral, R. Laxton, L. Doeby, I. Bodí, A. King, L. Brazil, R. Bhangoo, K. Ashkan, S. Al-Sarraj
P25
A rare case of fibroblastic reticular cell tumour in the spine
Z. Reisz, J. Salisbury, D. Bell, I. Bodi, S. Al-Sarraj

Audit of turnaround time (TAT) in glioma diagnosis
Oliver Sargent

Cerebral small vessel disease in multiple sclerosis
R. Geraldes, M. Esiri, J. Palace, G. DeLuca

Axonal protection by teriflunomide in an experimental lesion of Pattern III demyelination

Are visual hallucinations in Parkinson’s disease a result of decreased perfusion of visual processing areas of the brain?
L. Sinclair, J. Brenton, A. King Lun Liu, S. Gentleman, S. Love

Do anti-cholinergic drugs increase Alzheimer-type pathology in Parkinson’s patients?
A retrospective post-mortem investigation
A. King Lun Liu, Y. Mun Lim, R. Pearce, S. Gentleman

Can early tau depositions in mixed Alzheimer’s disease and Lewy body disease give insights into disease progression?

Sertanergic¹²³I-FP-CIT binding is associated with depression and not neuropathology
A. Oliver-Evans, S. Colloby, A. Thomas, D. Erskine, J. Attems

White Matter Hyperintensities and Pathological Correlations in Alzheimer’s Disease
P. Walsh, D. Thomas, C. Sudre, J. Iglesias, S. Crampsie, Z. Jaunmuktane, J. Holton, N. Ryan, J. Barnes, T. Lashley

DNA hydroxymethylation in Amyotrophic Lateral Sclerosis (ALS)
E. Schaber, C. Appleby-Mallinder, R. Highley, P. Heath

Increased calcyclin immunoexpression in ALS is seen within reactive astrocytes in corticospinal tracts and is not specific for ALS subtype
A. King, C. Troakes, C. Shaw, V. Marchica, S. Al-Sarraj, B. Smith

CLIPPERS: Chronic Lymphocytic Inflammation with Pontine Perivascular Enhancement Responsive to Steroids. A case study and review of neuropathological findings.
C. Turnquist, M. Hofer, J. Halliday, R. Kerr

A novel BMI1/Ephrin connection in human glioblastomas
T. Milner, B. Ricci, X. Zhang, N. Pomella, G. Rosser, S. Marino

Clinical and pathological features of multiple system atrophy and multiple system atrophy lookalikes
Y. Miki, H. Ling, S. Foti, J. Holton

Investigation of tau seeding activity in tauopathies
T. Bradshaw, H. Ling, J. Holton, S. Wray, R. de Silva, T. Revesz

The expression and presence of RNA binding proteins in FTLD-TDP
S. Foti, L. Gittings, C. Toomey, Y. Asi, T. Lashley

A Single cell omics approach to neurodegeneration
L. Greensmith