



124th Meeting of the British Neuropathological Society

The View, Royal College of Surgeons of England, Lincoln's Inn Fields, London

WEDNESDAY 1st March, 2023

Symposium: Skeletal muscle disorders – recent advances in pathology, genetics and therapy

Chairs: Professor Francesco Saverio Tedesco (UCL-GOSH-Francis Crick Institute, UK) and Dr Ashirwad Merve (GOSH-UCLH, UK)

12:30 Registration

13:00-13:05 Opening remarks

Francesco Saverio Tedesco (UCL-GOSH-Francis Crick Institute, UK) and Ashirwad Merve (GOSH-UCLH, UK)

SESSION I Chair: Caroline Sewry (RJA Orthopaedic Hospital, Salford Royal Hospital & UCL-GOSH Dubowitz Neuromuscular Centre, UK)

13:05-13:35 Doug Millay (Cincinnati Children's Hospital Medical Center, USA): **Muscle cell fusion during disease and opportunities for therapies**

13:35-14:05 Graziella Messina (University of Milan, Italy): **The transcription factor Nfix in skeletal muscle development and muscular dystrophies**

14:05-14:35 Marco Sandri (University of Padova, Italy): **Mechanisms of muscle mass regulation in health and disease**

14:35-15:00 Coffee break

SESSION II Chair: Fabrice Chretien (Institute of Myology & Hôpital Sainte Anne, Paris, France)

15:00-15:30 Francesco Saverio Tedesco (UCL-GOSH-Francis Crick Institute, UK): **Advanced skeletal muscle disease and therapy modelling**

15:30-16:00 Benedicte Chazaud (Institut NeuroMyoGène, Lyon, France): **Macrophage functions in skeletal muscle regeneration and in degenerative myopathies**

16:00-16:30 Jordi Diaz-Manera (Newcastle University, UK): **Fibro-fatty degeneration of skeletal muscles: causes, treatment and monitoring in muscular dystrophies**

16:30-17:00 Francesco Muntoni (UCL-GOSH, UK): **Genetic therapies for muscular dystrophies: successes and limitations** Benedicte

17:00-17:05 Concluding remarks

Francesco Saverio Tedesco (UCL-GOSH-Francis Crick Institute, UK) and Ashirwad Merve (GOSH-UCLH, UK)

17:05 Short Break

17:15 Alfred Meyer Memorial Lecture - The many faces of Myositis - results of a multidisciplinary approach
Werner Stenzel, Charité – Universitätsmedizin Berlin, Germany

THURSDAY 2nd March, 2023

- 08:00** **Registration**
- 08:50 - 09:00** **OPENING OF MEETING**
Stephen Wharton, BNS President
- 09:00 - 10:35** **FIRST SCIENTIFIC SESSION – Brain Tumours**
Chairs: Kathreena Kurian, Sebastian Brandner
- 09:00 **O1 Which meningiomas benefit most from methylation profiling?**
Fernanda Ruiz, Andrew Lowe, Monika Dutt, Peravin Mariathan, Ashirwad Merve, Zane Jaunmuktane, Sebastian Brandner
- 09:15 **O2 High-grade gliomas occurring in teenagers and young adult patients comprise novel molecular subgroups**
Matthew Clarke, Rita Pereira, Alan Mackay, Yura Grabovska, Amelia Clarke, James Nicholl, John Procter, Olumide Ogunbigi, Thomas Jacques, Lynley Marshall, Fernando Carceller, Zita Reisz, Safa Al-Sarraj, Maria Vinci, Kathreena Kurian, Ho-Keung Ng, Sebastian Brandner, Chris Jones
09. 30 **O3 Implementation of routine genomic profiling of cell-free DNA from cerebrospinal fluid: technical considerations and institutional experience across a broad spectrum of malignancies**
Richard Hickman, Alexandra Miller, Justin Jee, Si-Yang Liu, Elena Pentsova, Dara Ross, Subhiksha Nandakumar, Chad Vanderbilt, Satshil Rana, Katherine Hill, Kiana Chabot, Charli Hertz, Carl Campos, Neerav Shukla, Maria Diaz Ordonez, Khedoudja Nafa, Tejus Bale, Matthias Karajannis, Bob Li, Michael Berger, Marc Ladanyi, Ingo Mellinghoff, Adrienne Boire, Rose Brannon, Maria Arcila
- 09:45 **O4 Can p16 immunostaining predict the CDKN2A deletion status in IDH-mutant gliomas?**
Kathryn Urankar, **Gabrielle Cruickshank**
- 10.00 **O5 DNA methylation profiling of adult spinal glial tumours; results from a large single centre study**
Matthew Clarke, Rita Pereira, Alan Mackay, Yura Grabovska, Renee Cruickshank, Monika Dutt, Peravin Mariathan, Atul Kumar, Adrian Casey, Ashirwad Merve, Zane Jaunmuktane, Chris Jones, Sebastian Brandner
- 10:15 **O6 High-dimensional characterisation of the cellular and molecular changes in the human brain after radiotherapy**
Thomas Oliver Millner, Pratistha Panday, Nicola Pomella, Sara Bododi, Silvia Marino
- Flash talk*
- 10.30 **P13 Decision making for reporting health related research outcomes altering diagnosis in a paediatric brain tumour population**
Jessica Chiara Pickles, Kristian Aquilina, Jane Chalker, Christine Dahl, Abel Devadass, Kshitij Mankad, Ashirwad Merve, Munaza Ahmed, James AR Nicoll, Tabitha Bloom, David A Hilton, Neil J Sebire, Darren Hargrave, Thomas S Jacques
- 10:35 - 11:05** **Coffee break**
- 11:05 – 12.00** **SECOND SCIENTIFIC SESSION – Cerebrovascular and Neuroinflammation**
Chairs: Roxana Carare, Colin Smith
- 11.05 **O7 Pyramidal neurons of the medial prefrontal cortex in post-stroke, vascular and other ageing-related dementias**
Dan Jobson, Jeremy Cheong, Tuomo Polvikoski, Yoshiki Hase, Louise Allan, Raj N Kalaria

11.20 **O8 Hippocampal pericytes in post-stroke, vascular and neurodegenerative dementias**
Jeremy Cheong, Yoshiki Hase, Kelvin Gotama, Luciana Maffei, Ren Ding, Karen Horsburgh, Raj Kalaria

11.35 **O9 Exploring the Neuroinflammatory Response in Human Motor Neuron Disease**
Bridget Ashford, Charlotte Dawson, Charlie Appleby-Mallinder, Martyna Matuszyk, Paul Heath, Wenbin Wei, Mark Dunning, Delphine Boche, Julie Simpson, J Robin Highley

Flash Talks

11.50 **P6 Quantification of Oligodendrocyte (OLs) and Oligodendrocyte Precursor cells (OPCs) in the aging and pathological human brain using CD140A and SOX10**
W. Y. Stephanie Fang, Jamuna Jeevahan, Leslie. R. Bridges, Atticus. H. Hainsworth

11.55 **P26 The microglial TSPO phenotype – implications for clinical use**
Emma F. Garland, Laura Kulagowska, Thomas Scott, Henrike Antony, Michel Bottlaender, James A.R. Nicoll, Delphine Boche

12.00 - 13.00 Lunch

13:00 - 14.30 POSTER DISCUSSION

14.30 - 15.30 THIRD SCIENTIFIC SESSION – Epilepsy and Neuromuscular
Chairs: Maria Thom, James Nicoll

14.30 **O10 Delineating the neurodegenerative mechanisms underpinning epilepsy in Alpers' syndrome**
Laura Smith, Chun Chen, Alasdair Blain, Robert Taylor, Nichola Lax, Daniel Erskine, Robert McFarland

14.45 **O11 The effects of resistance exercise training on mitochondrial dysfunction in myotonic dystrophy type 1 patients**
Valeria Di Leo, Conor Lawless, Marie-Pier Roussel, Tiago B Gomes, Grainne S Gorman, Helen A Tuppen, Oliver Russell, Elise Duchesne, Amy E Vincent

15.00 **O12 Intra-individual variability in oxidative phosphorylation deficiency and mitochondrial DNA genetics in m.3243A>G-related Mitochondrial Myopathies**
Tiago Bernardino Gomes, Charlotte Warren, Valeria Di Leo, Jordan Childs, Grainne Gorman, Doug M Turnbull, Conor Lawless, Amy E Vincent

Flash Talks

15.15 **P8 Hyperphosphorylated tau protein, autophagy inhibition and somatic mutations in mTOR pathway in Focal Cortical Dysplasia type II (FCDII)**
Alicja Mrzyglod, Maria Thom, Joan Liu, James Mills

15.20 **P9 Evidence of chronic traumatic encephalopathy in the ERUK Corsellis Epilepsy Collection**
Maritchka Ryniejska, Hanaa El Hachami, Alicja Mrzyglod, Alyma Somani, Maria Thom

15.25 **P10 Amygdala Pathology In Temporal Lobe Epilepsy and Correlations with Diffusion MRI, Peri-Ictal Apnoea and SUDEP risk**
Hou Wang Lam, Smriti Patodia, Claudia Zeicu, Yau Lim, Hui Zhang, Antoine Legouhy, Beate Diehl, Maria Thom

15:30 - 16:00 Coffee Break

16:00 - 17:00 PROFESSIONAL AFFAIRS MEETING

17.00 - 17:45 BUSINESS MEETING

19:30 - late SOCIETY DINNER AT THE VIEW

FRIDAY 3rd MARCH 2023

- 08:00** **Registration**
- 09:00 - 10:35** **FOURTH SCIENTIFIC SESSION – Neurodegeneration**
Chairs: Lauren Walker, Stephen Wharton
- 09:00 **O13 Mitophagy impairments in Lewy body dementia – a protective role for Lewy bodies?**
Daniel Erskine, David Koss, Faye McLeod, Fiona LeBeau, Tiago Outeiro, Johannes Attems
- 09:15 **O14 Mitochondrial dysfunction in parvalbumin cells triggers juvenile-onset severe neurological disorder in vivo**
Elizaveta Olkhova, Yi Shiao Ng, Grainne S. Gorman, Fiona E. N. LeBeau, Carla Bradshaw, Nichola Lax
- 09:30 **O15 Brain DNA methylomic analysis of frontotemporal lobar degeneration identifies common dysregulated signatures across pathological subtypes**
Katherine Fodder, Megha Murthy, Patrizia Rizzu, Christina E. Toomey, Rahat Hasan, Jack Humphrey, Towfique Raj, Katie Lunnon, Jonathan Mill, Peter Heutink, Tammaryn Lashley, **Conceicao Bettencourt**
- 09:45 **O16 Prevalence, distribution, and severity of cerebral amyloid angiopathy differ between Lewy body diseases and Alzheimer’s disease**
Lauren Walker, Harry Simpson, Alan Thomas, Johannes Attems
- 10:00 **O17 Weighted gene co-methylation network analysis in post-mortem white matter tissue reveals shared and unique DNA methylation signatures associated with Parkinsonian diseases**
Megha Murthy, Katherine Fodder, Yasuo Miki, Eduardo De Pablo Fernandez, Emmanuelle Viré, Thomas Warner, Tammaryn Lashley, Conceição Bettencourt
- 10:15 **O18 Clonal hematopoiesis in Parkinson’s disease**
Karl Frontzek
- Flash Talk*
- 10.30 **P23 APOE genotyping from formalin fixed and paraffin-embedded tissue – Maximising the use of donated human brain tissue**
James Minshull, Yvonne Davidson, Andrew Robinson, Federico Roncaroli
- 10:35 - 11:00** **Coffee break**
- 11:00 - 12:30** **POSTER DISCUSSION**
- 12.30 - 13.30** **Lunch**
- 13:30 - 14:50** **CLINICAL UPDATE SESSION**
- 13.30 Glioneural Tumours
- 14.10 Meningiomas
- Arie Perry, University of California, San Francisco (UCSF)
- 14:50 - 15:05** **Coffee Break**
- 15:05 - 15.50** **DIAGNOSTIC SLIDE SESSION (INCORPORATING EQA)**
- 15.50** **PRIZE ANNOUNCEMENTS AND CLOSE OF THE MEETING**

Posters

P1 Anaplastic Neuroepithelial Tumour with Condensed Nuclei: a Case Report

Simon Deacon, Matthew Clarke, Zane Jaunmuktane, Stuart Smith, Richard Ashpole, Adam Thomas, Robert Goldspring, Ian Scott, Simon Paine

P2 Primary Leptomeningeal ALK-Negative Anaplastic Large Cell Lymphoma

George Johnson, Patricia Haylock-Vize, Paul Smith, Kathryn Urankar

P3 Primary central nervous system non-Langerhans cell histiocytosis with a NTRK1-LRRC71 gene fusion – a novel case report

Alexandra Rui Wan Kang, Chris Van Vliet, Jason Dyke

P4 Three rare cases of paediatric primary non-Langerhans cell histiocytosis of the CNS with associated molecular alterations

Alexandra Rui Wan Kang, Chris Van Vliet, Jason Mark Dyke

P5 Efficacy of PD-1/PD-L1 immunotherapy on brain metastatic non-small-cell lung cancer (BM-NSCLC) and treatment-related adverse events: a systematic review and meta-analysis

William J Phillips, Zak A Thornton, Lily J Andrews, Richard Daly, Gareth Ayre, Julian P T Higgins, Philippa Davies, Kathreena M Kurian

P6 Quantification of Oligodendrocyte (OLs) and Oligodendrocyte Precursor cells (OPCs) in the aging and pathological human brain using CD140A and SOX10

W. Y. Stephanie Fang, Jamuna Jeevahan, Leslie. R. Bridges, Atticus. H. Hainsworth

P7 Characterising the neuropathological features of cerebellar degeneration in Alpers' syndrome

Laura Smith, Elizaveta Olkhova, Nichola Lax, Daniel Erskine, Robert McFarland

P8 Hyperphosphorylated tau protein, autophagy inhibition and somatic mutations in mTOR pathway in Focal Cortical Dysplasia type II (FCDII)

Alicja Mrzyglod, Maria Thom, Joan Liu, James Mills

P9 Evidence of chronic traumatic encephalopathy in the ERUK Corsellis Epilepsy Collection

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P10 Amygdala Pathology In Temporal Lobe Epilepsy and Correlations with Diffusion MRI, Peri-Ictal Apnoea and SUDEP risk

Hou Wang Lam, Smriti Patodia, Claudia Zeicu, Yau Lim, Hui Zhang, Antoine Legouhy, Beate Diehl, Maria Thom

P11 Immune-mediated oxidative/nitrosative stress in metachromatic leukodystrophy

Daniel Erskine, Pawel Palmowski, Andrew Porter, Robert McFarland

P12 HnRNP K white matter pathology in neurodegenerative diseases

Xinwa Jiang, Xiangyi Du, Yazeed Buhidma, Ariana Gatt, Tammarn Lashley

P13 Decision making for reporting health related research outcomes altering diagnosis in a paediatric brain tumour population

Jessica Chiara Pickles, Kristian Aquilina, Jane Chalker, Christine Dahl, Abel Devadass, Kshitij Mankad, Ashirwad Merve, Munaza Ahmed, James AR Nicoll, Tabitha Bloom, David A Hilton, Neil J Sebire, Darren Hargrave, Thomas S Jacques

P14 The effects of resistance exercise training in mitochondrial myopathy patients

Valeria Di Leo, Conor Lawless, Jane Newman, Fiona Robertson, Sarah Pickett, Gavin Hudson, Yotam Levy, Julien Ochala, Helen A Tuppen, Grainne S Gorman, Oliver Russell, Amy E Vincent

P15 Fetal brain anomalies linked to MRPS22 mutations

Georges Tarris, Sophie Nambot, Faratanjona Harizay, Thierry Rousseau, Bernard Aral, Emmanuel Simon, Laurence Faivre, Christel Thauvin, **Homa Adle-biassette**

P16 Identification of Lewy Body Pathology Propagation Patterns In Lewy Body Disorders, Using Automated Digital Image Analysis

Toby Curless, Haruka Kawamura, Raquel Real, Alice Rockliffe, Sebastian Brandner, Yau Mun Lim, John Hardy, Huw Morris, Zane Jaunmuktane

P17 Differential Tau Burden in hTau Mice, Following Intracerebral Inoculation with Brain Extracts from Distinct Phenotypes of Corticobasal Degeneration

Toby Curless, Filipa Lourenco de Almeida, Ziang Wang, Justyna Zareba-Paslawska, Aran Mohan, Theisha Bradshaw, Per Svenningsson, Rohan de Silva, Zane Jaunmuktane

P18 The Role of Lipid Metabolism Across Cell Types in Parkinson's Disease

Kirsten Ebanks, Rina Bandopadhyay, Thomas Warner

P19 Exploring oligodendrocyte-related DNA methylation signatures in frontotemporal lobar degeneration

Katherine Fodder, Megha Murthy, Patrizia Rizzu, Peter Heutink, Tammaryn Lashley, Conceicao Bettencourt

P20 The role of Cystatin C in ALS

Sarah Granger, Kurt De Vos, J, Robin Highley

P21 Investigating the presence of tau pathology in Parkinson's disease

Joanne Lachica, Christina E Toomey, Sonia Gandhi

P22 DNA Methylation and One-Carbon Metabolism in Human Motor Neuron Disease

Farah Mahiddine, Robin Highley, Heather Walker, Guillaume Hautbergue

P23 APOE genotyping from formalin fixed and paraffin-embedded tissue – Maximising the use of donated human brain tissue

James Minshull, Yvonne Davidson, Andrew Robinson, Federico Roncaroli

P24 Developing scalable, in-vitro multicellular spinal cord models for assessment of neuropathological responses to nanotherapies

Shahzaib Shamsi

P25 Demystifying the gluten brain: understanding the development of gluten sensitivity-related neurological disorders

Mara-Luciana Floare, Julie E Simpson, Stephen B Wharton, Daniel Aeschlimann, Marios Hadjivassiliou

P26 The microglial TSPO phenotype – implications for clinical use

Emma F. Garland, Laura Kulagowska, Thomas Scott, Henrike Antony, Michel Bottlaender, James A.R. Nicoll, Delphine Boche