

A nighttime photograph of the London skyline. The London Eye is prominent on the left, illuminated in red. Several large fireworks are exploding in the dark sky. The River Thames flows through the center, with various bridges and buildings illuminated. The Big Ben clock tower is visible on the right, lit up. The overall scene is vibrant and festive.

9th Nuclear Envelope Disease and Chromatin Organisation Meeting  
and 3rd International Meeting on Laminopathies  
September 2-5, 2019 Kings College London

Programme  
and  
Posters list

9<sup>th</sup> UK Nuclear Envelope Disease and Chromatin Organization Meeting  
3<sup>rd</sup> International Meeting on Laminopathies

September 2 2019	afternoon	Time
<b>Registration</b>		15:00 - 15:30
<b>Progeroid Laminopathies</b> <i>Chairs: Cathy Shanahan and Delphine Larrieu</i>	<b>Cells</b> <i>sponsored session</i>	15:30 - 18:00
<b>Introduction and greetings</b>		15:30 - 15:40
<b>MADaM syndrome, a novel severe progeroid mandibuloacral dysplasia syndrome linked to primary mitochondrial and secondary nuclear defects</b> Annachiara DeSandre Giovannoli - France		15:40 - 16:00
<b>Progress, Lessons and the Future of Clinical Trials for Hutchinson-Gilford Progeria Syndrome</b> Leslie Gordon - USA		16:00 - 16:20
<b>Unexpected effects of interleukin 6 inhibition in progeroid cells</b> Giovanna Lattanzi - Italy		16:20 - 16:40
<b>Molecular and cellular mechanisms driving cardiovascular disease in Hutchinson-Gilford progeria syndrome</b> Vicente Andrès Garcia - Spain		16:40 - 17:00
<b>Prelamin A disrupts nuclear F-actin spatial organisation and prevents its association with sites of DNA repair</b> Andrew Martin Cobb - UK		17:00 - 17:15
<b>Structural defects at the nuclear periphery associated with envelopathies</b> Sophie Zinn-Justin - France		17:15 - 17:30
<b>Vulnerability of Progeroid Smooth Muscle Cells to biomechanical forces is mediated by an enzyme</b> Patricia R. Pitrez - Portugal		17:30 - 17:45
<b>Shaping of nuclear structure during mitosis impacts stem cell survival</b> Pamela K. Geyer - USA		17:45 - 18.00
<b>Welcome drink and POSTER SESSION I</b>		18:00-19:30

September 3 2019	morning	Time
<b>Muscular Laminopathies</b>		09:00-11:30
<i>Chairs: Gisèle Bonne and Gabriele Siciliano</i>		
<b>A Multistage Sequencing Strategy Pinpoints Novel Candidate Alleles for Emery-Dreifuss Muscular Dystrophy and Supports Gene Misregulation as its Pathomechanism</b> Eric Schirmer - UK		09:00-09:20
<b>Congenital laminopathies: what have clinicians learnt from 2008?</b> Susana Quijano-Roy - France		09:20-09:40
<b>Impairments in contractility and cytoskeletal organisation cause nuclear defects in skeletal muscle contractile disorders</b> Jacob Alexander Ross - UK		09:40-09:55
<b>Nuclear envelope abnormalities in myotonic dystrophy primary myoblasts</b> Peter Meinke - Germany		09:55-10:10
<b>Nesprin-1-alpha2 associates with kinesin at myotube outer nuclear membranes, but is restricted to neuromuscular junction nuclei in adult muscle</b> Ian Holt - UK		10:10-10:25
<b>Coffee break</b>		10:25-10:45
<b>Mechanically-induced nuclear damage and increased p53 signaling lead to myofiber dysfunction in skeletal muscle laminopathies</b> Tyler John Kirby - USA		10:45-11:00
<b>Modelling skeletal muscle laminopathies using human IPS cells and bio-engineered skeletal muscles</b> Francesco Saverio Tedesco - UK		11:00-11:15
<b>Defective physical coupling between the nucleus and cytoskeleton in Emery-Dreifuss muscular dystrophy</b> Lori L. Wallrath - USA		11:15-11:30
<b>Lunch</b>		13:00-14:15

September 3 2019	afternoon	Time
<b>Dynamic nucleo-cytoskeletal interactions and mechanotransduction</b> <i>Chairs: Qiuping Zhang and Joe Swift</i>		14:30 – 16:25
LINC-ing the nucleus and the cytoskeleton Patricia Davidson - France		14:30 - 14:50
Response to mechanical demand: from nucleus to matrix Joe Swift - UK		14:50 - 15:10
LINC complex disruption in epithelia increases cell malleability, and cohesive cell migration in space-restrictive 3D environments Iakowos Karakesisoglou - UK		15:10 - 15:25
Loss of cytoplasmic keratins in colonocytes downregulates nuclear lamins and compromises nuclear lamina integrity and cell cycle regulation Joel Nyström - Finland		15:25 - 15:40
Nesprin-1a2 mediates motor protein recruitment to the nuclear envelope to control myonuclear position Alice Catriona Hardie Haworth - UK		15:40 - 15:55
TorsinA is a multi-tool AAA+ protein within the nuclear envelope that acts through distinct functional assembly states. Gant Luxton - USA		15:55 - 16:10
SUN-KASH complexes undergo 6:6 back-to-back assembly to mediate force transduction across branched LINC complex networks Owen Richard Davies - UK		16:10 - 16:25
<b>Coffee break and POSTER SESSION II</b>		16:30 - 17:00 16:30 - 18:00

September 4 2019	morning	Time
<b>Cardio-Laminopathies</b>		09:00 - 11:30
<i>Chairs: Howard Worman and Elisa Di Pasquale</i>		
Lamina-associated polypeptide 1 in striated muscle and liver function Howard Worman - USA		09:00 - 09:20
Risk stratification for sudden death in laminopathies Karim Wahbi - France		09:20 - 09:40
Lessons from Lamin A mutants in cardiolaminopathies: from pathophysiology to molecular targets Monica Carmosino - Italy		09:40 - 09:55
Using CRISPR/Cas technology to study the role of LMNA exon 4 in the development and treatment of laminopathies Ignacio Pérez de Castro - Spain		09:55 - 10:10
<b>Coffee break</b>		10:15 - 10:45
The nuclear-cytoskeleton connection and nuclear positioning during muscle formation Bruno Cadot - France		10:45 - 11:00
Epigenetic inhibition of SCN5A by K219T-Lamin A/C induces myocardial conduction defects in an iPSC-based human model of cardiac laminopathy Elisa Di Pasquale - Italy		11:00 - 11:15
Establishing new role(s) for the nuclear envelope in the mammalian heart Matt Stroud - UK		11:15 - 11:30
<b>Lunch</b>		13:00-14:00

September 4 2019	afternoon	Time
<b>New nuclear envelope signalling pathways and mechanisms/DNA damage</b> <i>Chairs: Glenn Morris and Pascale Bertrand</i>		14:30 - 18:15
Altered cytoskeleton in cardiac disease caused by nuclear A-type lamins gene mutations Antoine Muchir - France		14:30 - 14:50
Mechanisms of Prelamin A induced Cardiovascular Ageing Catherine Shanahan - UK		14:50 - 15:10
Exploring synthetic rescue pathways in premature ageing syndromes Delphine Larrieu - UK		15:10 - 15:30
<i>Short presentation, Matt Wheeler</i> <i>Vectorbuilder</i>		15:30 - 15:40
<b>Coffee break and POSTER SESSION III</b>		15:40 - 17:00
Lamin B1, a new factor controlling the recruitment of 53BP1 to DNA damage Pascale Bertrand - France		17:00 - 17:15
P38MAPK mediates Senescence-induced Lamin B1 Degradation Rhys Anderson - UK		17:15 - 17:30
The maintenance of nuclear envelope integrity by a new regulator of nucleo-cytoplasmic transport Mike Schertzer - France		17:30 - 17:45
The front-rear polarity of cell nucleus Paolo Maiuri - Italy		17:45 - 18:00
Targeted Perturbation of Nuclear Envelope Integrity with Vapor Nanobubble-mediated Photoporation Gaëlle Houthaève - Belgium		18:00 - 18:15
<b>Drink reception</b>		<b>18:30 - 19.30</b>
<b>Standing Dinner</b>		<b>19:30 - 21:00</b>

September 5 2019	morning	Time
<b>Lipodystrophic laminopathies and new pathogenetic mechanisms</b> <i>Chairs: David Araujo-Vilar and Rafal Czapiewski</i>		09:00 - 11:25
The R482Q Lmna mutation in a mouse model of FPLD inhibits adipogenesis Colin Stewart - Singapore		09:00 - 09.20
LMNA-linked lipodystrophy: new insight on cardiovascular phenotypes Corinne Vigouroux - France		09:20 - 09.40
Nicotinamide riboside extends lifespan in progeroid mice Chen-Yu Liao - Kennedy's Lab - USA		09:40 - 09:55
The Inner Nuclear Membrane protein Nemp1 is specifically required for fertility Didier Michel Hodzic - USA		09:55 - 10:10
Membranes regulating function and the multiple personalities of AKTIP protein Isabella Saggio - Italy		10:10 - 10:25
<b>Coffee break</b>		10:25 - 10:45
FETCH1/Tmem120A Directs Spatial Genome Organization in Adipogenesis and Knockout Mice Have a Lipodystrophic Phenotype Rafal Czapiewski - UK		10:45 - 11:00
Monitoring of chromatin organization in live cells using a novel imaging tool, FRIC Einar Hallberg - Sweden		11:00 - 11:15
<i>Short presentation Kanika Coleman</i> <i>Transnetyx</i>		11:15 - 11:25
<b>Lunch</b>		13:00 - 14:00

September 5 2019	afternoon	Time
<b>Genome organisation and regulation from the nuclear envelope/Cancer</b> <i>Chairs: Eric Schirmer and Patricia Davidson</i>		14:00 - 17:30
SAMMY (Sequential Analysis of MacroMolecules accessibilitY)-seq to detect chromatin structure alterations Chiara Lanzuolo - Italy		14:00 - 14:20
Lack of Active Chromosome Relocation in Senescent and HGPS Fibroblasts after an external stimulus is recapitulated in a Freshwater Molluscan Model Organism Joanna Bridger - UK		14:20 - 14:40
Interplay between chromatin and the nuclear envelope modulates spatial genome topology in health and disease Philippe Collas - Norway		14:40 - 15:00
Genomic instability in laminopathies: new mechanisms to target therapeutically Susana Gonzalo-Hervas - USA		15:00 - 15:15
<b>Coffee break and POSTER SESSION IV</b>		15:15 - 16:45
Downregulation of Lamin A triggers cell migration and invasion in Ewing Sarcoma Francesca Chiarini - Italy		16:45 - 17:00
Increased lamin B1 levels promote cancer cell migration by altering perinuclear actin organization Gabi Gerlitz - Israel		17:00 - 17:15
<b>Conclusions</b>		17:15 - 17:30
<b>END OF MEETING</b>		



## POSTERS

### Progeroid laminopathies

- 1 Deborah Cardoso, France  
**Role of parylation on cardiovascular disease in progeroid syndromes**
- 2 Maria Rosaria D'Apice, Italy  
**MDPL Syndrome: the role of POLD1 gene in the DNA repair process**
- 3 Solenn Guilbert, France  
**Pathological modelling of vascular phenotype associated to HGPS using IPSC- derived VSMCs**
- 4 Elisabetta Mattioli, Italy  
**PCAF involvement in lamin A/C-HDAC2 interplay and its recruitment at the NE by lamin A/C**
- 5 Tobias Zech, UK  
**Interrogation of Acto-myosin Mediated Nuclear Force Coupling in HGPS**

### Muscular laminopathies

- 6 Giulia Ricci, Italy  
**A case of LMNA-LGMD1B and muscle biopsy's changes suggestive of calpain 3-related myopathy**

### Dynamic nucleo-cytoskeletal interactions and mechanotransduction

- 7 Paloma Alvarez-Suarez, Poland  
**Schwann cells pericentromeric heterochromatin is involved in neuromuscular junction morphological remodeling during development**
- 8 Ed Battey, UK  
**The missing LINC to human healthy ageing**
- 9 Emma Carley, USA  
**Elucidating the role of nuclear envelope proteins in the regulation of pro-fibrotic gene expression**
- 10 Pietro Salvatore Carollo, France  
**Linc complexes are mechanotransducers that fine-tune beta-catenin signaling along the EMT spectrum through  $\alpha$ -catenin nuclear translocation**
- 11 Glenn Morris, UK  
**Nesprin-1-alpha2 at the outer nuclear membrane**
- 12 Kleopatra Papa, UK  
**An investigation into variations in the LINC complex in breast cancer**

### Cardiolaminopathies

- 13 Shanella Amelia de Silva, UK  
**Investigating a novel role for the nuclear envelope LINC complex in cardiomyocyte mechanotransduction**

### New NE signalling pathways and mechanisms / DNA damage

- 14 Aurelie Bellanger, Norway  
**Identification and characterization of novel proteins involved in nuclear envelope repair**
- 15 Cristina Capanni, Italy  
**Emerin post-translational modifications are affected during early phase of oxidative stress response**
- 16 Charles R Dixon, UK  
**A nuclear envelope function for STING in mediating innate immunity to RNA viruses?**
- 17 Louise Ingeman Petersen, Norway  
**Unstrained ESCRT-II drives chromosome fragmentation and micronuclear catastrophe**
- 18 Ana Marques Leal, Belgium  
**A-type lamin status determines recovery from compression-induced nuclear envelope rupture**
- 19 Marine Jeanne Prissette, USA  
**Genome wide CRISPR/Cas9 screen identifies genetic modifiers of tau aggregation that contribute to nuclear envelope integrity**
- 20 Emilie Rass, France  
**Lamin B1, a new factor controlling the recruitment of 53BP1 to DNA damage**
- 21 Caroline Louise Stoten, UK  
**Regulation of ESCRT-III-dependent nuclear envelope regeneration during mitotic exit**

### Lipodystrophic laminopathies

- 22 Evdokila Pitolitsyna, Norway  
**Role of LncRBA HOTAIR in nuclear architecture remodeling during adipose differentiation**

Genome organisation and regulation from the NE /Cancer

- 23 Davide Andrenacci, Italy  
**Transposable element dysregulation as a consequence of NE impairment**
- 24 Daniel Horton, UK  
**Effects of ageing on nuclear organisation and induced genome reorganisation in a molluscan model organism**
- 25 Muhunden Jayakrishnan, UK  
**Lamin A/C regulates the heat shock induced expression of Hsp70 gene locus by assisting its movement away from nuclear periphery**
- 26 Yaiza Mariela Jung Garcia, UK  
**Nuclear envelope dynamics during cell migration in melanoma**
- 27 Celine Labouesse, UK  
**Lamin A overexpression in murine embryonic stem cells blocks neural lineage specification**
- 28 Rositsa Maystorova, Czech Republic  
**Characterisation of lamin A/C interaction with phosphoinositides**
- 29 Aakila Sammy, UK  
**Ovarian cancer cytogenomics and nuclear motors**

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