

Conference Programme ENCALS 2019

Wednesday 15 May 2019

LUNCH sponsored by: Mitsubishi Tanabe Pharma

13.15-14.00 WELCOME & YOUNG INVESTIGATOR AWARD

Patrice Diot, Dean of Tours University
Leonard van den Berg, Chair of ENCALS

SESSION 1 GENOMICS

CHAIRS: Jan Veldink, Russell McLaughlin

14.00-14.30 KEYNOTE SPEAKER: GUY ROULEAU

14.30-16.00 PLATFORM PRESENTATIONS

2084 MicroRNA genetic variants in human ALS;
Chen Eitan, Weizman Institute Israel

1950 Serum microRNA profiles reveal an involvement of FXR1 and FXR2 in amyotrophic lateral sclerosis
Axel Freischmidt, Ulm Germany

2053 C9orf72 intermediate expansions of 24–30 repeats are associated with ALS:
Alfredo Iacoangeli, KCL London UK

1929 GWAS in ALS identifies novel loci and insight into the genetic architecture:
Rick van der Spek on behalf of Project MinE, UMC Utrecht the Netherlands

2049 Opportunities and pitfalls in unravelling the role of DNA methylation in ALS: a large-scale collaborative effort within Project MinE
Paul Hop on behalf of Project MinE Consortium, UMC Utrecht the Netherlands

1949 Hexanucleotide repeat RNA-binding proteins as modifiers of RNA toxicity in C9orf72 ALS
Elke Braems, Leuven Belgium

16.00-16.30 COFFEE & POSTERS

SESSION 2 DISEASE MECHANISMS (1)

CHAIRS: Janine Kirby, Patrick Vourc'h

16.30-18.00 PLATFORM PRESENTATIONS

1955 Transcriptional alterations induced by polyDPRs overexpression in ALS-neuronal cell model.

Riccardo Cristofani, Milano Italy

1946 Heterozygous Tbk1 loss has opposing effects in early and late stages of ALS in mice

David Brenner, Ulm Germany

1974 Loss of C9orf72 alters the immunophenotype but does not change the motor neuron loss of SOD1G93A mice.

Caterina Bendotti, Milano Italy

2097 Gain and loss of function synergize in the C9ORF72 HRE pathogenicity

Hortense de Calbiac, Paris France

1982 TDP-43 peptide seeding in the context of aggregation and phase separation

Donya Pakravan, Leuven Belgium

1909 Cognitive deficits in ALS are a marker of localized TDP-43 cerebral pathology

Jenna M. Gregory, Edinburgh Scotland

18.00-19.30 POSTER SESSION (WITH CHEESE AND WINE)

FACILITATORS: Michael van Es, Orla Hardiman

18.30-20.00 Satellite Meeting: Mitsubishi Corporate

18.00-19.00 Project MinE meeting

Thursday 16 May 2019

7.00-9.00: Satellite meeting: FILSLAN Meeting

SESSION 3 CLINICAL AND EPIDEMIOLOGY

CHAIRS: Philippe Couratier, Dorthée Lule

8.45-9.15 KEYNOTE SPEAKER: GIAN CARLO LOGROSCINO

9.15-10.30 PLATFORM PRESENTATIONS

2021 Occupations, socio-economic position, and the risk of amyotrophic lateral sclerosis. The Euro-MOTOR study

Fabrizio D'Ovidio on behalf of EUROMOTOR, Torino Italy

2052 The D50 Progression Model for Amyotrophic Lateral Sclerosis: Extension to and Validation in Global Cohorts.

Julian Grosskreutz, Jena Germany

1997 Pulmonary Function and ALS Staging Systems

Philippe Couratier Limoges France

1964 Endophenotypes in ALS: Cognitive and psychiatric assessment of unaffected relatives of ALS patients.

Emmett Costello, Dublin Ireland

2012 Executive impairment among C9orf72 repeat expansion carriers: An under recognized phenomenon

Marie Ryan, Dublin Ireland

10.30-11.00 COFFEE

SESSION 4 DISEASE MODELS

CHAIRS: Rudolf Hergesheimer, Severine Boillee

11.00-11.30 KEYNOTE SPEAKER: DR. LAURA FERRAIUOLO

11.30-12.30 PLATFORM PRESENTATIONS

1993 Metabo-lipidomics of fibroblasts from ALS patients show alterations in purine, pyrimidine, energetic, and phospholipid metabolisms

Charlotte Veyrat-Durebex, Angers France

1983 C9orf72 and TDP-43 iPSC-derived motor neurons have alterations in calcium buffering

Ruxandra Dafinca, Oxford UK

1925 Using patient-derived astrocytes to unravel the nuclear role of SOD1 and its link with nucleus/cytoplasm shuttling

Noemi Gatto, Sheffield UK

2040 Epigenetic features of C9orf72 gene: DNA methylation and hydroxymethylation as RNA foci modifiers in iPSCs

Clara Volpe, Milano Italy

12.30-14.00 LUNCH sponsored by: Biogen Idec & POSTERS

12:30-13:15 Satellite Symposium EFFIK Italfarmaco: Riluzole and ALS: from the theory to the real world.

SESSION 5 BIOMARKERS

CHAIRS: Eran Hornstein, Peter Bede

14.00-15.30 PLATFORM PRESENTATIONS

2082 Prognostic and disease progression microRNA biomarkers for amyotrophic lateral sclerosis

Iddo Magen, Weizman Israel

1991 Glucose metabolic patterns along the phenotypic MND-FTD spectrum.

Joke De Vocht, Leuven Belgium

1965 Prognostic value of neurofilament light chain and C-reactive protein in serum of patients with ALS

Maxim de Schaepdryver, Leuven Belgium

1920 Cervical spinal cord MRI captures presymptomatic pathology in c9orf72 mutation carriers: a longitudinal neuroimaging study.

Giorgia Querin, Paris France

2000 The hypometabolic state is a good predictor of a better prognosis in ALS patients.

Christian Lunetta, Milano Italy

2028 Quantifying Executive Subdomain Dysfunction in ALS using EEG during the Sustained Attention to Response Task

Roisin McMackin, Dublin Ireland

15.30-16.00 COFFEE AND POSTERS

SESSION 6 DISEASE MECHANISMS (2)

CHAIRS: Catherina Bendotti, Luc Dupuis

16.00-17.30 PLATFORM PRESENTATIONS

1932 Molecular and cellular functions of the C9ORF72 protein and its potential implication in ALS

Manon Boivin, Ilkirch-Graffenstaden France

2098 Inclusions of Misfolded wt-SOD1 are common in ALS patients with mutations in c9orf72 and other ALS/FTD-associated genes

Karin Forsberg, Umea Sweden

1945 Antisense (C4G₂)_n RNA repeats from C9orf72 mutation bind RNA binding proteins

Mirjana Malnar, Slovenia

1984 The interactome of human mutant TDP-43 identifies key pathways dysregulated in amyotrophic lateral sclerosis

Emily Feneberg, Oxford UK

2009 Role of potential cell-to-cell transmission of dipeptide repeat proteins in C9ORF72 ALS/FTD pathology

Paolo Marchi, Sheffield UK

1943 Behavioral deficits, tdp-43 pathology and progressive neuropathology in cyclophilin a knock-out mice: a mouse model of frontotemporal dementia.

Laura Pasetto, Milano Italy

17.30-19.00 POSTER SESSION

FACILITATORS: Magdalena Kuzma, Blaz Koritnik, Ammar Al-Chalabi

20.00- GALA DINNER

Friday May 17 2019

SESSION 7 THERAPEUTICS

CHAIRS: Philippe Corcia, Ruben van Eijk

**9.00-9.30 KEYNOTE SPEAKER: VINCENT MEININGER
RILUZOLE 20 YEARS LATER, CAN WE DO BETTER?**

9.30-10.45 PLATFORM PRESENTATIONS

2081 Safety, PK, PD, and exploratory efficacy in single and multiple dose study of a SOD1 antisense oligonucleotide (BIIB067) in participants with ALS

Tim Miller, St. Louis USA, Biogen Idec

2029 Results of FORTITUDE-ALS: A Phase 2, Double-blind, Randomized, Placebo-Controlled, Study to Evaluate Efficacy, Safety and Tolerability of Reldesemtiv

Jeremy Shefner Phoenix Arizona USA, Cytokinetics

1939 : The REFALS-ES open-label extension study of oral levosimendan (ODM-109)

Ammar Al-Chalabi, London UK

2039 Long-term Outcome of Filgrastim (G-CSF) in ALS Patients in Reference to PRO-ACT

S. Johannesen, Heidelberg Germany

2002 Gene editing as a potential therapeutic approach for ALS/FTD-associated with expanded C9ORF72

Joao Cruzeiro, Sheffield UK

10.45-11.15 COFFEE

SESSION 8 PRECISION MEDICINE

CHAIRS: Caroline Ingre, Claude Desnuelle

11.15-12.15 PLATFORM PRESENTATIONS

1902 Using Artificial Intelligence to Uncover Novel Drug Therapies and Targets for Amyotrophic Lateral Sclerosis (ALS)

Monika Myszczyńska, Sheffield UK

2100 Evidence of New ALS Phenotypes based on Clustering of Spectral EEG Measures

Stefan Dukic, Dublin Ireland & Utrecht the Netherlands

2007 The distinct phenotypical traits of the UNC13A polymorphism in amyotrophic lateral sclerosis

Harold Tan, Utrecht the Netherlands

Platform Trials in ALS

Sabrina Paganoni, Jeremy Shefner & HEALEY Institute Boston

12:15-12:30 POSTER AWARDS

12:30-12:40 WELCOME IN EDINBURGH

12.40-12.45 CLOSE OF MEETING