

Conference Programme ENCALS 2019

**Wednesday 15 May 2019**

**13.15-14.00 WELCOME & YOUNG INVESTIGATOR AWARD**

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

**SESSION 1 GENOMICS**

**1400-14.30 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 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 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)**

**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)**

**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**

**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**

**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 iPS-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 & POSTERS

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

### SESSION 5 BIOMARKERS

#### 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.  
**Andrea Lizio, 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)**

### **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 (C4G<sub>2</sub>)<sub>n</sub> 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**

### **20.00- GALA DINNER**

**Friday May 17 2019**

**SESSION 7 THERAPEUTICS**

**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**

**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**

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

**Stefan Dukic, Dublin Ireland & Utrecht Holland**

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

**Harold Tan, Utrecht Holland**

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**

**14.00          Satellite meeting: TRICALS Meeting**