

**13 & 14 juin
2024**

STAV

**ICM Institut
du Cerveau**

Progrès en neuro-génétique ***Update in Neurogenetics***

*ICM- Institut du Cerveau - Paris Brain Institute, Paris
47 Bd de l'hôpital, 75013 Paris*



Coordinateurs :

Alexandra Durr, Christophe Verny, Mathilde Renaud, Cyril Goizet, Gaetan Lesca.

Progrès en neuro-génétique/ Update in Neurogenetics

DAY 1 - 13 June 2024

8h00-8h45 : Welcome

8h45 : Introduction to the day : Alexandra Durr, Christophe Verny

Session 1 : Genetics in frequent neurodegenerative diseases

Chair : Claire Ewenczyk (Paris) and Mathieu Anheim (Strasbourg)

- **9h00-9h20 : Elisabeth Tournier-Lasserre**, Genetics of cerebrovascular events (CERVCO, University Paris 7, Paris)
- **9h20-9h40 : Nick Wood**, Parkinson Disease is it inherited ? (UCL, London)
- **9h40-10h00 : Gael Nicolas**, Alzheimer disease: what's new and more than single genes ? (IRIB, Normandy University Rouen, Rouen)
- **10h00-10h30 : General discussion**

10h30-11h00 : Coffee break - stands

Session 2 : Neurodevelopment and neurogenetics

Chair : Alexis Brice (Paris) and Alexandra Durr (Paris)

- **11h00-11h20 : Thomas Bourgeron**, Genetic risk for autism (Institut Pasteur, Paris)
- **11h20-11h40 : Sandrine Humbert**, Neurodevelopment and Huntington Disease (ICM, Paris)
- **11h40-12h : Renzo Guerrini**, Florence Epileptic encephalopathies and progressive neurodegeneration (Children's Hospital A. Meyer-University of Florence, Florence)
- **12h-12h30 : General discussion**

12h30 -13h30 : Lunch Break - stands

13h30-14h30 : Symposium

Session 3 : Mechanisms and new paradigms

Chair : Odile Boesflug (Paris) and Gaetan Lesca (Lyon)

- **14h30-14h50 : Stéphanie Baulac**, Cortical dysplasia and treatment. (ICM, Paris)
- **14h50-15h10 : Luc Dupuis**, Amyotrophic lateral sclerosis and heritability (USIAS, University of Strasbourg, Strasbourg)
- **15h10-15h30 : Frances Platt**, Oxford Treatment of Niemann Pick disease type C. (Department of Pharmacology, University of Oxford, Oxford)
- **15h30-15h50 : Radhia Kacher**, Somatic instability in repeat expansion disorders (ICM, Paris)
- **15h50-16h10 : General discussion**

16h10-16h30 : Coffee break - stands

Session 4 : Genetic therapy

Chair : Cecilia Marelli (Montpellier) and Giovanni Stevanin (Bordeaux)

- **16h30-16h50 : Nicole Deglon**, Gene editing in SCA3 (LNCM, University of Lausanne, Lausanne)
- **16h50-17h10 : Isabelle Desguerre**, Antisense approach for neuromuscular diseases: where are we in 2023 ? (IMAGINE, University Paris Cité, Paris)
- **17h10-17h30 : Rebecca Schüle**, One variant one therapy example POLR3A (University of Heidelberg, Heidelberg)
- **17h30-17h50 : Guy Lenaers**, Pre-mRNA trans-splicing as a gene therapy solution for OPA1-related diseases (CHU d'Angers, Angers)
- **17h50-18h10 : General discussion**

End of Day 1

DAY 2 - 14 June 2024

8h00-8h30 : Welcome

Session 5 : Expansions in Neurogenetics

Chair : Karine Nguyen (Marseille) and Christophe Verny (Angers)

- **8h30-8h50 : Christel Depienne**, Expansions in Neurogenetics : How to get them ? (Institut für Humangenetik, Universitätsklinikum Essen, Essen)
- **8h50-9h10 : Franco Taroni**, Role of intermediate alleles in TBP (Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan)
- **9h10-9h30 : Mathilde Renaud**, SCA27B a frequent GAA among late onset ataxias (University of Lorraine, Nancy)
- **9h30-9h50 : Vincent Huin**, RFC1: motifs and phenotypes (CHU de Lille, Lille)
- **9h50-10h20 : General discussion**

10h20-11h : Coffee break - stands

11h -12h : Symposium

Session 6 : Round Table for pluridisciplinary genome interpretation (CV)

12h -13h : Jean Madeleine de Sainte Agathe (Paris), **Cyril Goizet** (Bordeaux), **Virginie Pichon** (Angers), **Christel Thauvin** (Dijon)

End of Day 2