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AFSA, COMMUNICATIONS

17 OCTOBER 2014 - PARIS

3RD ANGELMAN SYNDROME INTERNATIONAL MEETING



AUDITORIUM INSTITUT DE MYOLOGIE - BÂTIMENT BABINSKI
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REGISTRATION

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SPEAKERS

ARTHUR L. BEAUDET, M.D.

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STORMY J. CHAMBERLAIN

DEPARTMENT OF GENETICS AND DEVELOPMENTAL BIOLOGY
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YPE ELGERSMA PH.D.

DEPARTMENT OF NEUROSCIENCE, DIRECTOR OF ENCORE EXPERTISE CENTER
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ANGELA MABB, PH.D.

DEPARTMENT CELL BIOLOGY & PHYSIOLOGY,
UNIVERSITY OF NORTH CAROLINA - CHAPEL HILL, NC, USA

UGO MAYOR,

IKERBASQUE RESEARCH PROFESSOR, GROUP LEADER,
CIC BIOGUNE- BILBAO, SPAIN

SILVIA RUSSO, PH.D.

DEPARTMENT OF MOLECULAR GENETIC,
ISTITUTO AUXOLOGICO ITALIANO - CUSANO MILANINO (MI), ITALY

MARTIN SCHEFFNER, PH.D.

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DAVID SEGAL

GENOME CENTER, DEPARTMENT OF BIOCHEMISTRY & MOLECULAR MEDICINE
UNIVERSITY OF CALIFORNIA - DAVIS, CA, USA

HARALD H. SITTE, PH.D.

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VIENNA MEDICAL UNIVERSITY - VIENNA, AUSTRIA

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DR. SYLVIANE PEUDENIER, PRESIDENT, NEUROPEDIATRICIAN, CHRU BREST

DR. ARMAND BOTTANI, GENETICIST, CHU GENÈVE (SWITZERLAND)

DR. PERRINE CHARLES, NEUROLOGIST, HÔPITAL DE LA PITIÉ SALPÉTRIÈRE, PARIS

DR. NATHALIE DORISON, NEUROPEDIATRICIAN, HÔPITAL TROUSSEAU & HÔPITAL
ST JOSEPH/INSTITUT LEJEUNE, PARIS

DR. DELPHINE HERON, GENETICIST, HÔPITAL DE LA PITIÉ SALPÉTRIÈRE, PARIS

DR. MARIE-ODILE LIVET, NEUROPEDIATRICIAN, CH D'AIX EN PROVENCE

PROF. ANNE MONCLA, GENETICIST, HÔPITAL DE LA TIMONE, MARSEILLE

DR. NICOLE SARDA, NEUROBIOLOGIST, INSERM SCIENS'AS, LYON

PROF. LOUIS VALLEE, NEUROPEDIATRICIAN, CHRU LILLE

PROGRAMME

8.30 REGISTRATION

9.00 WELCOME ADDRESS

- **DR. SYLVIANE PEUDENIER**, NEUROPEDIATRICIAN,
PRESIDENT OF AFSA SCIENTIFIC ADVISORY COMMITTEE
(FRANCE)

- **MR. NICOLAS VIENS**, AFSA, BOARD MEMBER (FRANCE)

9.30 MONOAMINE TRANSPORTERS AS TARGETS OF CAMKII α : IMPLICATIONS FOR ANGELMAN SYNDROME
HARALD H. SITTE, PH.D. (AUSTRIA)

10.00 MODEL SYSTEMS TO STUDY UBE3A/E6AP FUNCTION
BEN DISTEL, PH.D. (NETHERLANDS)

10.30 A CRITICAL ROLE FOR UBE3A IN BRAIN DEVELOPMENT
YPE ELGERSMA (NETHERLANDS)

11.00 COFFEE BREAK

11.30 MECHANISMS OF UBE3A IMPRINTING
ANGELA MABB, PH.D. (USA)

12.00 CHARACTERIZATION OF UBE3A MUTATIONS IN ANGELMAN PATIENTS
SILVIA RUSSO, PH.D (ITALY)

12.30 LUNCH BREAK

14.00 NEWS AND VIEWS ON UBE3A/E6AP AND ITS ROLE IN UBIQUITIN
CONJUGATION
MARTIN SCHEFFNER PH.D. (GERMANY)

14.30 UBE3A ROLE IN MODEL SYSTEMS OF ANGELMAN SYNDROME: REGULATION OF PROTEIN HOMEOSTASIS
UGO MAYOR (SPAIN)

15.00 USING HUMAN INDUCED PLURIPOTENT STEM CELLS TO MODEL ANGELMAN SYNDROME
STORMY J. CHAMBERLAIN (USA)

15.30 USE OF OLIGONUCLEOTIDES TO ACTIVATE THE EXPRESSION OF THE PATERNAL GENE
ARTHUR L. BEAUDET, M.D. (USA)

16.00 COFFEE BREAK

16.30 TARGETED REACTION OF UBE3A IN A MOUSE MODEL OF ANGELMAN SYNDROME
DAVID SEGAL (USA)

17.00 CLINICAL TRIAL OF LEVODOPA APPLIED TO ANGELMAN SYNDROME: OBJECTIVES, METHODOLOGY, ASSESSMENT CRITERIA AND FOLLOW-UP ISSUES
PERRINE CHARLES, M.D., PH.D. (FRANCE)

17.30 BRAINSTORMING (CLOSED SESSION)

THIS IS A PRE-PROGRAMME, LAST MINUTE CHANGES MAY OCCUR

MODERATORS

PERRINE CHARLES / NATHALIE DORISON ; ARMAND BOTTANI / ANNE MONCLA