19th International Workshop on Fragile X and other Neurodevelopmental Disorders

Wednesday, September 18, 2019 – Day 1

14:00-16:00	**Arrival & Registration & Poster Set-Up** (poster mounting Wednesday PM through Thursday AM)
16:00-16:15	**Welcoming remarks** **Meeting Chair: Maria G. Miano** (Italy)
16:15-17:00	**Keynote Lecture** **Bekim Sadikovic** (Canada) *DNA methylation signatures in mendelian developmental disorders as a diagnostic bridge between genotype and phenotype*
17:00-19:00	**Fragile X Syndrome and *FMR1* related disorders** **Chairs: Barbara Bardoni** (France) and **Gary J. Bassell** (USA)
17:00-17:15	**Frank Kooy** (Belgium) *Single-Cell and Neuronal Network Alterations in an in vitro Model of Fragile X Syndrome*
17:15-17:30	**Francesco Longo** (USA) *Cell type-specific disruption of cortico-striatal circuitry drives repetitive and perseverative behaviors in Fragile X Syndrome model mice*
17:30-17:45	**Nisha Raj** (USA) *Cell-type-specific profiling of molecular defects in a human induced pluripotent stem cell model of Fragile X Syndrome*
17:45-18:00	**Paul Hagerman** (USA) *Relationship between Fragile X protein (FMRP) and IQ using a quantitative FRET-based method for determining FMRP levels*
18:00-18:15	**Pietro Chiurazzi** (Italy) *Methylated premutation of the FMR1 gene in three sisters: correlating CGG expansion and epigenetic inactivation*
18:15-18:30	**Andrew McKechanie** (UK) *Functional magnetic resonance imaging in idiopathic intellectual impairment and Fragile X Syndrome*
18:30-18:45	**Veronica Nobile** (Italy) *Altered mitochondrial function in cells carrying a premutation or unmethylated full mutation of the FMR1 gene*
18:45-19:00	**Veronica Martínez-Cerdeño** (USA) *New discoveries in the pathology of FXTAS*
19:00 20:00	**Welcome cocktail**
20:30	**Dinner**

Thursday, September 19, 2019 – Day 2

07:00-08:00	**Breakfast**
08:00-09:30	**Clinical studies in Fragile X Syndrome and X-linked Intellectual Disabilities** **Chairs: Flora Tassone** (USA) and **Vincent des Portes** (France)
08:00-08:15	**Angela Peron** (Italy, USA) *Cardinal Signs of Snyder-Robinson Syndrome*
08:15-08:30	**Aurore Curie** (France) *A French cohort of 187 patients with X-Linked Intellectual Disability (XLID): developmental trajectories, physical and cognitive assessment, impact on primary care-giver*
08:30-08:45	**David R. Hessl** (USA) *NIH Toolbox Cognitive Battery Validation for Individuals with Intellectual Disabilities*
08:45-09:00	**Alessandra Murgia** (Italy) *Gait analysis in Fragile X Syndrome*
09:00-09:15	**Lisa Cordeiro** (USA) *Evaluating trajectories of developmental and behavioral outcome measures in Fragile X Syndrome (FXS) across the lifespan: informing treatment trial design in FXS*
09:15-09:30	**Marta Arpone** (Australia) *Intellectual Functioning and Behavioural Features Associated with Mosaicism in Fragile X Syndrome*
09:30-10:15	**Coffee break**
10:15-11:00	**Keynote Lecture** **Chair: Charles E. Schwartz** (USA) **Hans van Bokhoven** (The Netherlands) *The Genetic landscape of Intellectual Disability: Extreme Genetic Heterogeneity Converging onto Shared Molecular and Cellular Disease Pathways*
11:00-12:50	**X-linked Intellectual Disability** **Chairs: Cheryl Shoubridge** (Australia) and **Hilde van Esch** (Belgium)
11:00-11:25	**Hilde van Esch** (Belgium) *Defective DNA polymerase alfa-primase leads to X-linked intellectual disability associated with severe growth retardation, microcephaly and hypogonadism*
11:25-11:50	**Roger E. Stevenson** (USA) *Phenotypic Consequences of Duplication of Genes Associated with X- Linked Intellectual Disability*

11:50-12:05 **Mike Fields** (Australia)
Xq13 chromosomal duplications including the neurocognitive gene RLIM are associated with intellectual disability, recognizable facial features and epilepsy in males

12:05-12:20 **Giovanni Neri** (Italy, USA)
XLID: Phenotype of Female Carriers

12:20-12:35 **Cheryl Shoubridge** (Australia)
Heterozygous loss of function of IQSEC2 /Iqsec2 leads to increased activated Arf6 and severe neurocognitive seizure phenotype in females

12:35-12:50 **Anna Fliedner** (Germany)
Investigating the pathomechanisms of Borjeson-Forssman-Lehmann Syndrome

13:00-14:30 Lunch and relax time

14:30 16:25 Autosomal syndromic and non-syndromic Intellectual Disability - I
Chairs: Anita Rauch (Switzerland) and **Alessandra Murgia** (Italy)

14:30-14:55 **Christiane Zweier** (Germany)
CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum

14:55-15:10 **Ype Elgersma** (The Netherlands)
Angelman Syndrome-associated mutations reveal the mechanism and importance of UBE3A nuclear targeting

15:10-15:25 **Paranchai Boonsawat** (Switzerland)
Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly and delineation of novel candidate genes

15:25-15:40 **Massimo Zollo** (Italy)
A genotype-phenotype correlation study in Microcephaly affected families carrying biallelic homozygous mutations (p.D106N) in the PRUNE-1 locus

15:40-15:55 **Anais Begemann** (Switzerland)
Clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and new insights into the underlying pathomechanism

15:55-16:10 **Gerarda Cappuccio** (Italy)
Genetic testing of a large cohort of patients with neurodevelopmental disorders

16:10-16:25 **Jean-Louis Mandel** (France)
Delineation of natural history and comorbidities in genetic forms of neurodevelopmental disorders through GenIDA, an international participatory database: identification of previously unreported respiratory problems in a large cohort study of Koolen-deVries Syndrome patients and initial analysis of response to antiepileptic drugs

16:30-18:00	**Coffee break & Poster Session I (odd numbers)**
18:00-19:55	**Autosomal syndromic and non-syndromic Intellectual Disability- II** **Chairs: Christiane Zweier** (Germany) and **Frank Kooy** (Belgium)
18:00-18:25	**Alexandre Reymond** (Switzerland) *Genome architecture and diseases: the 16p11.2 paradigm*
18:25-18:40	**Amélie Piton** (France) *De novo missense variants in genes encoding proteins involved in mRNA repression, AGO1 and DDX6, in Intellectual Disability*
18:40-18:55	**Danny Huylebroeck** (The Netherlands) *Multi-functional and multi-modal actions of the Mowat-Wilson Syndrome transcription factor ZEB2*
18:55-19:10	**Emanuela Leonardi** (Italy) *Mutations in PURA gene are related to Rett-like features, movement disorder, epilepsy and myelin function anomalies*
19:10:19:25	**Irma Järvelä** (Finland) *Phenotypic spectrum associated with a CRADD founder variant underlying frontotemporal predominant pachygyria in the Finnish population*
19:25-19:40	**Nadif Kasri Nael** (The Netherlands) *How to measure E/I balance in IPSC-derived neuronal models. A case for CDH13 deficiency*
19:40-19:55	**Madhura Bakshi** (Australia) *Application of Whole Genome Sequencing technology for molecular diagnosis of Intellectual Disability in a multiethnic cohort-initial experience and findings on reanalysis*
20:30	**Dinner**

Friday, September 20, 2019 – Day 3

07:00-08:00 **Breakfast**

08:00-09:25 **Autism Spectrum Disorders**
Chairs: Jean-Louis Mandel (France) and **Pietro Chiurazzi** (Italy)

08:00-08:25 **Yuri Bozzi** (Italy)
Aberrant somatosensory processing in mouse models of ASD

08:25-08:40 **Annette Schenck** (The Netherlands)
Kismet, the Drosophila orthologue of ID/ASD genes CHD7 and CHD8, is required for sleep integrity

08:40-08:55 **Gaelle Hayot** (France)
Autism Comorbidities: Role of CHD8 during the Development of the Enteric Nervous System

08:55-09:10 **Aditi Bhattacharya** (India)
Closing the loop on dysregulated translation in Autism: What does data from Neuroligin 3 rat model tell us?

09:10-09:25 **Emma Baker** (Australia)
Incomplete Silencing of Full Mutation mRNA from Males with Fragile X Syndrome is Associated with More Severe Autistic Features

09:30-11:00 **Coffee break & Poster session II (even numbers)**

11:00-11:45 **Keynote lecture**
Chair: Claudia Bagni (Italy, Switzerland)
Silvia Cappello (Germany)
Modeling neurogenesis and neuronal migration with human cerebral organoids

11:45-13:10 **Mechanisms of disease using animal models and human cells - I**
Chairs: David Nelson (USA) and **Hans van Bokhoven** (The Netherlands)

11:45-12:10 **Peng Jin** (USA)
The loss of Fragile X mental retardation protein alters the development of human forebrain organoids

12:10-12:25 **Edoardo Penna** (Italy)
Cystatin B involvement in synapse physiology of rodents' brain and human cerebral organoids

12:25-12:40 **David Picketts** (Canada)
Characterization of a mouse model of the NEDDFL Syndrome

12:40-12:55 **Bozena Kuzniewska** (Poland)
Mitochondria biogenesis in the synapse is supported by local translation

12:55-13:10 **Cecilia Laterza** (Italy)
Modelling Fragile X Syndrome with iPSCs

13:10-14:45	**Lunch and relax time**
14:45-16:15	**Mechanisms of disease using animal models and human cells - II** **Chairs: David Picketts** (Canada) and **Yuri Bozzi** (Italy)
14:45-15:00	**Arielle Valdez** (USA) *Novel role of Cdh1-APC as a regulator of FMRP and protein synthesis at the synapse*
15:00-15:15	**Giorgia Pedini** (Italy) *FMRP affects glioblastoma progression regulating invasion-associated genes*
15:15-15:30	**Małgorzata Drozd** (France) *The first spontaneous mouse model of epilepsy of infancy with migrating focal seizures*
15:30-15:45	**Anne Gregor** (Germany) *Genetic Interaction screen for severe neurodevelopmental disorders reveals a functional link between Ube3a and Mef2 in Drosophila melanogaster.*
15:45-16:00	**Ilaria Meloni** (Italy) *High efficiency of CRISPR/Cas9-mediated gene editing for the correction of pathogenic mutations in Rett spectrum disorders*
16:00-16:15	**Francois Bolduc** (Canada) *Deciphering neurodiversity: Why translational analysis of Neurodevelopmental disorders requires a multimodal quantitative approach*
16:15-20:30	**Free time**
20:30	**Gala Dinner**

Saturday, September 21, 2019 – Day 4	
07:00-08:30	**Breakfast**
08:30-10:10	**Therapeutic perspectives – I** **Chairs: Randi Hagerman** (USA) and **Roger E. Stevenson** (USA)
08:30-08:55	**Daman Kumari** (USA) *Therapeutic Potential of CRISPR/Cas9 Mediated Deletion of CGG repeats for FMR1 Gene Reactivation in Fragile X Syndrome*
08:55-09:10	**Christina Gross** (USA) *Dysregulated protein phosphorylation as biomarker and treatment target in Fragile X Syndrome*
09:10-09:25	**Alberto Martire** (Italy) *Adenosine A2A receptor inhibition reverses synaptic and behavioral abnormalities in Fmr1 KO mice*
09:25-09:40	**Heather Bowling** (USA) *New protein expression-based blood biomarkers for Fragile X Syndrome*
09:40-09:55	**Vitaly Klyachko** (USA) *Understanding and correcting neuronal excitability defects in Fragile X Syndrome*
09:55-10:10	**Maria G. Miano** (Italy) *Histone demethylase KDM5C is a HDACi-sensitive central hub at the crossroads of transcriptional axes involved in multiple neurodevelopmental disorders*
10:10-10:45	**Coffee break**
10:45-12:25	**THERAPEUTIC PERSPECTIVES – II** **Chairs: Christina Gross (**USA) and **Paul Hagerman** (USA)
10:45-11:00	**Randi Hagerman** (USA) *Controlled Trial of Lovastatin and PILI Intervention in children with Fragile X Syndrome*
11:00-11:15	**Flora Tassone** (USA) *Biomarkers predictive of metformin treatment in Fragile X Syndrome*
11:15-11:30	**Poonnada Jiraanont (Thailand)** *Molecular Biomarkers Predictive of Sertraline Treatment Response in Young Children with Autism Spectrum Disorder*
11:30-11:55	**Norifumi Shioda (Japan)** *G-quadruplexes as a therapeutic target for ATR-X Syndrome*
11:55-12:10	**Cheryl Shoubridge (Australia)** *Short term estradiol treatment reduces seizure severity but does not improve cognitive measures in mouse models of congenital epilepsy and intellectual disability*
12:10-12:25	**Lucia Verrillo (Italy)** *Phytocannabinoid treatment in a mouse model of West Syndrome with spontaneous seizures*

12:25-13:15	**Best paper/poster Award & Plans for the next Workshop** **Chair: Giovanni Neri** (Italy, USA)
13:15-14:30	**Lunch Box & Departure**

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