Program of 14th Troina Meeting on Genetics of Neurodevelopmental Disorders

Thursday Morning 4th April 2019

Opening Addresses

9.15-9.30

Silvestro Rotondo, President of the Oasi Research Institute

Raffaele Ferri, Scientific Director ofthe Oasi Research Institute

Angela Ragusa, Boardmember of Italian Society of Human Genetics (SIGU)

Corrado Romano, on Behalf of the Scientific Committee

Invited Session 1 on Autism Spectrum Disorders

Chair: Thomas Bourgeron (France)

9.30-10.15

Thomas Bourgeron (France)

"The genes and mechanisms underlying autism"

10.15-10.45

James Ellis (Canada)

"*SHANK2* mutations associated with Autism Disorder cause hyperconnectivity of human neurons"

10.45-11.00

Questions and Comments

11.00-11.30

Break

11.30-12.00

Gaia Novarino (Austria)

"Epigenetic Regulators in Autism Spectrum Disorders"

12.00-13.15

Short Communications Session 1

12.00-12.15

Alessandro Vitriolo (Italy)

"Multi-omic deconvolution of the regulatory networks underlying neurodevelopmental and autism spectrum disorders"

12.15-12.30

Thomas Rolland (France)

"Differences in the genetic background contribute to risk and resilience to autism"

12.30-12.45

Hande Kaymakçalan Çelebiler (Turkey)

"First prevalence study of PTEN mutations in Turkish children with autism spectrum disorders and macrocephaly"

12.45-13.00

Emanuela Leonardi (Italy)

"Next Generation Sequencing tools for the characterization of Rett-like spectrum disorders"

13.00-14.30

Lunch

Thursday afternoon 4th April 2019

Invited Session 2 on FMR1-related disorders

Chair: Frank Kooy (Belgium)

14.30-15.00

Maria Vincenza Catania(Italy)

"Neurotransmitter receptor dysfunctions and possible therapeutical interventions in Fragile X syndrome"

15.00-15.30

Paul J. Hagerman (USA)

“Pathogenesis of FXTAS” 30-minute talk

15.30-16.00

Randi J. Hagerman (USA)

“New Targeted Treatments in Fragile X syndrome” 30-minute talk

16.00-16.15

Questions and Comments

16.15-16.45

Break

16.45-18.15

Short Communications Session 2

16.45-17.00

Barbara Bardoni (France)

“New insights into the pathophysiology of Fragile X Syndrome: altered molecular and cellular Phenotypes”

17.00-17.15

Alessandra Murgia (Italy)

“Gaitanalysis in childrenwith Fragile X Syndrome: could this become a measurable outcome?”

17.15-17.30

Natália Oliva Teles (Portugal)

“Loss of amplification variation observed in six families detected in 22q11.2 region by multiplex ligation-dependent probe amplification”

17.30-17.45

Martina Servetti (Italy)

"Potential additive effects of multiple heterozygous copy number variants (CNVs) in patients affected by neurodevelopmental disorders (NDD)"

17.45-18.00

Giulia Rosti (Italy)

"Novel position effects through involvement of Topologically Associating Domains (TADs) in patients with neurodevelopmental disorders"

18.00-18.15

Nuno Maia (Portugal)

"A de novo *HDAC8* missense variant that promotes genome instability"

Friday Morning 5th April 2019

Invited Session 3 on Intellectual Disability

Chair: Bert de Vries (The Netherlands)

9.30-10.15

Fowzan S Alkuraya (Saudi Arabya)

"Autosomal Recessive Intellectual Disability: A Story of Stealth, Affliction, Heritage and Opportunities"

10.15-10.45

Bert BA de Vries (The Netherlands)

"From next generation sequencing to next generation phenotyping in intellectual disability"

10.45-11.15

Joseph D Buxbaum (USA)

"Drug development in Phelan-McDermid Syndrome"

11.15-11.30

Questions and Comments

11.30-12.00

Break

12.00-13.15

Short Communications Session 3

12.00-12.15

Michele Gabriele (Italy)

“Patient-specific disease modelling uncovers molecular convergence of enhanceropathies: Kabuki and Gabriele de-Vries syndromes”

12.15-12.30

Mike Field (Australia)

“Facematch.org.au: A computer face-matching technology to help aid diagnosis and phenotypic recognition”

12.30-12.45

Yan Shi (The Netherlands)

“A human model for Brunner syndrome reveals increased neuronal activity of dopaminergic neurons”

12.45-13.00

Katrin Linda (The Netherlands)

“ROS Dependent increased autofagosome formation mediates synaptic dysfunction in a patient derived model for Koolen-de Vries Syndrome”

13.00-13.15

Anouk Verboven (The Netherlands)

“MEA-seq for combined gene expression and neuronal network measurements in iPSC-derived neurons from Koolen-de Vries patients”

13.15-14.30

Lunch

Friday Afternoon 5th April 2019

Invited Session 4 on Epilepsy and Birth Defects

Chair: Heather Mefford (USA)

14.30-15.00

Ghayda Mirzaa (USA)

"Mosaic mutations in brain malformations"

15.00-15,30

Gemma Carvill (USA)

"Aberrant splicing of poison exons in patients with epilepsy"

15.30-16.00

Johannes Lemke (Germany)

"De novo variants in neurodevelopment disorders with epilepsy"

16.00-16.15

Questions and Comments

16.15-16.45

Break

16.45-18.00

Short Communications Session 4

16.45-17.00

Ginevra Zanni (Italy)

"Biallelic variants in the nuclear pore complex protein NUP93 are associated with non-progressive congenital ataxia"

17.00-17.15

Maria Cristina Aspromonte (Italy)

“Targeted gene panel for identification of genes in a diverse clinical phenotype of children with Movement Disorders and Cerebral Palsy”

17.15-17.30

Filomena Pirozzi(USA)

“Modeling brain growth disorders using cerebral organoids: the case of LIG4 syndrome”

17.30-17.45

Cristina Cheroni (Italy)

“Cortical brain organoids to dissect human corticogenesis in physiological and pathological conditions”

17.45-18.00

Ilse van der Werf (The Netherlands)

“NMDAR activity as a target for treatment of Kleefstra Syndrome”

Saturday Morning 6th April 2019

Session 5 on Genomics and Modelling

Chair: Corrado Romano (Italy)

9.30-10.00

Jozef Gecz (Australia)

“PCDH19 Girls clustering epilepsy, a disorder of cellular mosaics”

10.00-10.30

Hans van Bokhoven (The Netherlands)

“IMPACT: Identification of converging Molecular Pathways Across Chromatinopathies as Targets”

10.30-10.45

Questions and Comments

10.45-11.15

Break

11.15-11.45

NaelNadifKasri (The Netherlands)

“Modelling mitochondrial disorders and epilepsy using induced pluripotent stem cells”

11.45-12.15

Alejandro Lopez Tobon (Italy)

“One by one: single cell resolution illuminates disease pathogenesis in organoid models of neurodevelopmental disorders”

12.15-12.30

Questions and Comments

12.30

Closing Remarks