OASI MARIA SS.
ISTITUTO DI RICOVERO E CURA A CARATTERE SCIENTIFICO
OSPEDALE CLASSIFICATO

L'Arangio

17TH TROINA MEETING
on GENETICS OF NEURODEVELOPMENTAL DISORDERS
27TH - 29TH APRIL 2023

PROGRAM

MEETING2023.OASI.EN.IT

with the unconditional contribution of
International Society for Developmental Neuroscience (ISDN) – Montreal
Simons Foundation – Autism Research Initiative – (SFARI) – New York

SECRETARIAT
U.O.S.
FORMAZIONE PERMANENTE
E ECM
ecm@oasi.en.it - Tel. 0935 936461/2

17TH TROINA MEETING ON GENETICS OF NEURODEVELOPMENTAL DISORDERS

27th-29th April 2023

Thursday Morning 27th April

Opening Remarks
9.00 – 9.30
Phenotyping and AI (Chair Bert de Vries)
9.30 - 10.15 Peter Robinson (Framington, USA) "*The Human Phenotype Ontology: AI and Semantic Algorithms for Phenotype Driven Translational Research and Genomic Diagnostics*" peter.robinson@jax.org
10.15 - 10.45 Tzung-Chien Hsieh (Bonn, Germany) "*GestaltMatcher Database – a FAIR database for medical imaging data of rare diseases*" thsieh@uni-bonn.de

10.45 - 11.15 Break

11.15 - 11.45 Alexander Hustinx (Bonn, Germany) "*Face2HPO: CNN-based HPO Labeling and Disorder Classification of Syndromic Face*" alexanderhustinx@gmail.com
11.45 - 12.15 Lex Dingemans (Nijmegen The Netherlands): "*PhenoScore: AI-based phenomics to quantify rare disease and genetic variation"* a.dingemans@radboudumc.nl
12.15 - 12.45 Panel discussion: *"Role of AI in Genetics"*

Short communications session 1
12.45 - 13.00 Noraly Jonis (Nijmegen, The Netherlands): *"Human Disease Genes website series; developments and achievements"* Noraly.jonis@radboudumc.nl
13.00 - 13.15 María del Rocío Pérez Baca (Ghent, Belgium): *"A novel neurodevelopmental syndrome caused by loss-of-function of the Zinc Finger Homeobox 3 (ZFHX3) gene"* MariadelRocio.PerezBaca@UGent.be

LUNCH

Thursday Afternoon 27th April

Speech and ASD related disorders (Chair Heather Mefford)
14.30 - 15.00 Thomas Bourgeron (Paris, France) "*Phenotypic effects of genetic variants associated with autism beyond diagnosis*" thomasb@pasteur.fr
15.00 - 15.30 Bregje van Bon (Nijmegen, The Netherlands): "*The different faces of SETBP1"* Bregje.vanBon@Radboudumc.nl
15.30 - 16.00 Angela Morgan (Melbourne, Australia) "*Genetic basis of severe child speech disorder*" amor@unimelb.edu.au

16.00 - 16.30 Break

Short Communications Session 2
16.30 -16.45 Davide Aprile (Milan, Italy): *"Benchmarking cerebellar organoids development and their use to model autism spectrum disorder: the CHD8 paradigm"* Davide.aprile@external.fht.org

16.45 - 17.00 Lisa Hamerlinck (Ghent, Belgium): *"Structural variants disrupt a critical regulatory region downstream of FOXG1"* Lisa.Hamerlinck@UGent.be
17.00 - 17.15 Dale Annear (Antwerp, Belgium): *"CGG Short Tandem Repeat Expansions are Overrepresented in Neurodevelopmental Disorders"* Dale.Annear@uantwerpen.be

Friday Morning 28th April

Therapy session (Chair Frank Kooy)
9.30 - 10.15 Ype Elgersma (Rotterdam, The Netherlands): *"Angelman syndrome: Shifting from mechanisms towards ASO treatment"* y.elgersma@erasmusmc.nl
10.15 - 10.45 Giuseppe Testa (Milan, Italy): "*Translating brain organoids endophenotypes: from mechanisms to scales*" giuseppe.testa@fht.org

10.45- 11.15 Break

Short Communications Session 3
11.15 -11.30 Mathijs van der Lei (Antwerp, Belgium): *"A Combination of the Live Mouse tracker (LMT) and Multi-Electrode Array (MEA) As Versatile Drug Screening Platform for Fragile X syndrome"* Mathijs.vanderLei@uantwerpen.be
11.30 – 11.45 Oliviero Leonardi (Milan, Italy): *"Single-cell dissection of CHD2 dosage disruption effects on autophagy and cortical development".* oliviero.leonardi@fht.org
11.45 - 12.00 Imke Schuurmans (Nijmegen, The Netherlands): *"Modelling genetic disorders of lysine metabolism in a dish"* Imke.Schuurmans@radboudumc.nl

Friday Afternoon 28th April

Modelling session (Chair: Gemma Carvill)
14.30 - 15.00 Bassem Hassan (Paris, France): "*The brain on time: links between development and degeneration"* Bassem.hassan@icm-institute.org
15.00 - 15.30 Sofia Puvogel Lutjens (Nijmegen, The Netherlands): *"Towards functional and molecular understanding of human-derived networks in neurodevelopmental disorders"* sofia.puvogellutjens@radboudumc.nl
15.30 - 16.00 Christel Depienne (Essen Germany): *"Predicting gene-dosage associations at the genome scale using machine learning: the example of the X chromosome"*
christel.depienne@uk-essen.de
16.00 -16.30 Break

Short Communications Session 4
16.30 – 16.45 Federica Marinaro (Milan, Italy): *"The "High throughput brain organoid longitudinal profiling from patient cohorts of neurodevelopmental disorders" study: seeding an international and multidisciplinary effort for transforming our understanding of neurodevelopmental conditions and neurodiversity"* federica.marinaro@fht.org
16.45 - 17.00 Xiuming Yuan (Nijmegen, The Netherlands): *"Human stem cell-derived glutamatergic neurons display homeostatic plasticity at network and single-cell level"* Xiuming.Yuan@radboudumc.nl
17.00 – 17.15 Claudio D'Incal (Antwerp, Belgium): "*The chromatin remodeler ADNP regulates the cellular response to autophagy and mitochondrial activity: a suggested role of SIRT1 in the cerebellum of a deceased toddler with Helsmoortel-Van der Aa syndrome*" claudio.dincal@uantwerpen.be

Saturday Morning 29th April

Epilepsy session (Chair: Jozef Gecz)
9.30 - 10.00 Gemma Carvill (Chicago, USA) "*Molecular and neuronal phenotypes of CHD2 dosage sensitivity in neurodevelopmental disorders*" gemma.carvill@northwestern.edu
10.00 - 10.30 Sarah Weckhuyzen (Antwerpen, Belgium) *"Precision therapy development for KCNQ2-encephalopathy: a coordinate European effort"* sarah.weckhuysen@uantwerpen.vib.be
10.30 - 11.00 Heather Mefford (Memphis, TN, USA) "*Epigenetic approaches to epilepsy diagnosis*" Heather.Mefford@STJUDE.ORG

11.00 – 11.30 Break

Various (Chair: Corrado Romano)
11.30 – 12.00 Evan Eichler (Seattle, USA) *"Long-read sequence and assembly of patient genomes"* eee@gs.washington.edu
12.00 - 12.30 Jozef Gecz (Adelaide, Australia) *"Genetic, molecular and mouse model investigations of broad neurodevelopmental impact of deleterious variants of the TREX mRNA export complex subunits"* jozef.gecz@adelaide.edu.au

12.30 – 12.45 Questions and Comments
12.45 Closing remarks

Scientific Committee

Bert de Vries, Leader Research group Genomic profiling in intellectual disability Radboudumc Donders Institute for Brain, Cognition and Behaviour - Nijmegen, The Netherlands

Frank Kooy, Full professor Medical genetics departement – Research leader - University of Antwerp, Belgium

Heather Mefford, Center for Pediatric Neurological Disease Research - St. Jude Children's Research Hospital Leader Mefford lab - Memphis, USA

Corrado Romano, Head of Research Unit of Rare Diseases and Neurodevelopmental Disorders Oasi Research Institute-IRCCS – Associate Professor of Medical Genetics University of Catania

Responsabile scientifico ECM

Secretariat

Continuing Medical Education Unit – ecm@oasi.en.it

with the unconditional contribution of

International Society for Developmental Neuroscience (ISDN) – Montreal

Simons Foundation – Autism Research Initiative – (SFARI) – New York

Meeting2023.oasi.en.it