9th INTERNATIONAL MEETING

on CNVs and Genes in Intellectual Disability and Autism

April 4-5, 2014

La Cittadella dell'Oasi - Troina - EN (Italy)

Friday, April 4, 2014

9.15-9.30
OPENING ADDRESSES

9.30-11.30
INVITED SESSION 1
Chair: Bert de Vries (Nijmegen, The Netherlands)

9.30-10.15
Opening Keynote Lecture
Jozef Gecz (Adelaide, Australia)
Defining the role of mRNA export and mRNA decay in neurodevelopmental disorders, intellectual disability and autism

10.15-10.45
Vera Kalscheuer (Berlin, Germany)
X-linked intellectual disability genes and networks

10.45-11.15
Björn Menten (Ghent, Belgium)
Mate pair sequencing for the detection of chromosomal aberrations in patients with intellectual disability and congenital malformations

11.15-11.30
Discussion

11.30-12.00
Coffee Break

12.00-13.00
SHORT COMMUNICATIONS SESSION 1
Chair: Marco Fichera (Catania, Italy)

12.00-12.15
Sylwia Rzońca (Warsaw, Poland)
Array CGH analysis of a cohort of Polish patients with X-linked intellectual disability

12.15-12.30
Wigard P. Kloosterman (Utrecht, The Netherlands)
An inventory of de novo structural variants in the general population

12.30-12.45
Margot R.F. Reijnders (Nijmegen, The Netherlands)
Mutations in YY1 cause intellectual disability

12.45-13.00
Céline Helsmoortel (Antwerp, Belgium)
A recurrent SWI/SNF related autism syndrome caused by de novo mutations in ADNP

13.00-14.30
Lunch

14.30-16.15
INVITED SESSION 2
Chair: Frank Kooy (Antwerp, Belgium)

14.30-15.00
Alexandre Reymond (Lausanne, Switzerland)
Chromatin loops and CNVs: the complex spatial organization of the 16p11.2 locus

15.00-15.30
Sebastien Jacquemont (Lausanne, Switzerland)
Gene and expression dosage at the 16p11.2 locus modulates brain growth as well as key circuitry involved in social cognition and energy balance

15.30-16.00
Yann Herault (Strasbourg, France)
Modelling 16p11 and 17q21.31 copy number variants affecting intellectual disabilities in the mouse

16.00-16.15
Discussion

16.15-16.30
Coffe Break

16.30-17.15
SHORT COMMUNICATIONS SESSION 2
Chair: Paolo Bosco (Troina, Italy)

16.30-16.45
Agnieszka Charzewska (Warsaw, Poland)
Novel mutation in OPHN1 identified by chromosome X exome sequencing in a Polish family with intellectual disability

16.45-17.00
Daniëlle G.M. Bosch (Nijmegen, The Netherlands)
NR2F1 mutations cause optic atrophy with intellectual disability

17.00-17.15
Ana Rita Santos (Leuven, Belgium)
Dysfunctions of the CYFIP1 pathway leads to severe intellectual disability

Saturday, April 5, 2014

9.30-12.30
INVITED SESSION 3
Chair: Heather Mefford (Seattle, USA)

9.30-10.00
Frank Kooy (Antwerp, Belgium)
FRA2A is a CGG repeat expansion associated with silencing of AFF3

10.00-10.30
Reza Asadollahi (Zurich, Switzerland)
The use of small CNVs to identify novel disease genes in neurodevelopmental disorders

10.30-11.00
Lisenka Vissers (Nijmegen, The Netherlands)
Genome sequencing identifies major causes of severe intellectual disability

11.00-11.15
Discussion

11.15-11.45
Coffee Break

11.45-12.30
Closing Keynote Lecture
Evan Eichler (Seattle, USA)
Advances in Understanding the Genetic Basis of Autism and Intellectual Disability

12.45
Closing remarks

SCIENTIFIC COMMITTEE
B. de Vries - F. Kooy - H. Mefford - C. Romano

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