

Wednesday, September 29th, 2010

- 09:00 - 12:00 Meeting of the Leopoldina Senate
(*by invitation only*)
- 12:15 - 13:30 Reception to welcome new
Leopoldina members (*by invitation only*)
- 14:00 - 14:30 **Welcome and Introduction**
Leopoldina Early Career Award
(*funded by the Commerzbank-Stiftung*)

Session I: Dominant and X-linked MR Chair: Wieacker/Wieczorek

- 14:30 André **Reis**
Dominant forms of mental retardation
- 15:00 Hans-Hilger **Ropers**
X-linked forms of mental retardation
- 15:30 Simone **Berkel**
Mutations in the SHANK2 synaptic scaffolding
gene in autism spectrum disorders and mental
retardation
- 15:45 Georg **Rosenberger**
Mutations in GRIN2A and GRIN2B encoding
regulatory subunits of NMDA receptors cause
variable neurodevelopmental phenotypes
- 16:00 Luciana **Musante**
The XLMR protein PQBP1 plays a role in
alternative splicing

Session II: Autosomal Recessive MR Chair: Ropers/Engels

- 16:45 Andreas **Kuss**
Unravelling the molecular basis of autosomal
recessive intellectual disability:
The Berlin experience
- 17:15 Laurence **Colleaux**
Unravelling the molecular and pathophysiological
bases of autosomal recessive mental
retardation: The Paris experience
- 17:45 Anita **Rauch**
Disturbed Wnt signalling due to mutations in
CCDC88C causes autosomal recessive non-
syndromic hydrocephalus
- 18:00 Andreas **Tzschach**
Next generation sequencing in a family with
autosomal recessive Kahrizi syndrome reveals
a homozygous frameshift mutation in SRD5A3

Keynote Lecture I Chair: Strom

- 18:30 - 19:15 Matthew **Hurles**
Next generation sequencing - promises and
pitfalls
- 19:30 – Get together and Poster exhibition**

Thursday, September 30th, 2010

Session III: Copy number variation and MR Chair: Rauch/Schröck

- 09:00 Nigel **Carter**
Deciphering Developmental Disorders
- 09:30 Stylianos **Antonarakis**
Genomics of Down Syndrome
- 10:00 Denise **Horn**
Identification of FOXP1 deletions in three
unrelated patients with mental retardation and
significant speech and language deficits
- 10:15 Janneke H.M. **Schuurs-Hoeijmakers**
Identifying MR genes associated with CNVs

Keynote Lectures II & III Chair: Rappold/Reis

- 11:00 - 11:45 Daniel **Geschwind**
Autism genetics: at the crossroads of
genomics and cognitive neuroscience
- 11:45 - 12:30 Danielle **Posthuma**
Genetic foundations of human intelligence

Session IV: Neurodevelopment and MR Chair: Antonarakis/Geschwind

- 14:00 Markus **Missler**
Synaptic cell-adhesion molecules and
the aetiology of ASDs/MR
- 14:30 Jamel **Chelly**
Neuronal migration defects
- 15:00 Jacques L. **Michaud**
De novo mutations in synaptic genes
associated with non-syndromic mental
retardation

Friday, October 1st, 2010

Session V: Molecular Networks in MR I Chair: Schenck/Hempel

- 09:00 Eric **Legius**
RAS-pathway
- 09:30 Linda **van Aelst**
Role of Rho regulators in neurodevelopment
and disease
- 10:00 Patrizia **D'Adamo**
Mutations in the small GTPase gene RAB39B
are responsible for X-linked Mental Retarda-
tion associated with autism, epilepsy and
macrocephaly
- 10:15 Christian **Thiel**
Disruption of the histoneacetyltransferase
MYST4 leads to a Noonan syndrome-like
phenotype and hyperactivated MAPK signaling

Session VI: Molecular Networks in MR II Chair: Rieß/van Aelst

- 11:00 Annette **Schenck**
Dissection of molecular networks and
common mechanisms in *Drosophila*
- 11:30 Hans **van Bokhoven**
Genetic and epigenetic networks in
cognitive dysfunction (GENCODYS)
- 12:00 Merel **Oortveld**
A first systematic evaluation of MR gene
function in neuronal development

Keynote Lecture IV Chair: Reis

- 12:15 - 13:00 Stephen T. **Warren**
Fragile X syndrome: Molecular mechanisms
and therapeutic implications
- 13:00 André **Reis**
Closing remarks

MRNET

MRNET is a research network aiming at the systematic identification of novel genes involved in mental retardation and the functional investigation of the respective proteins as well as the delineation of genotype-phenotype correlations. MRNET is funded by the National Genome Research Network (NGFNplus) of the Federal Ministry of Education and Research.

Project coordinator: André Reis, Erlangen

Organizers

André Reis, Erlangen (chair)
Maja Hempel, Munich
Gudrun Rappold, Heidelberg
Anita Rauch, Zurich
Hilger Ropers, Berlin
Annette Schenck, Nijmegen
Evelin Schröck, Dresden

Conference venue

Hörsaalgebäude Medizin, Ulmenweg,
91054 Erlangen, Germany

Registration and abstract submission

Registration is free, but capacity is limited.

Please visit our website: www.german-mrnet.de

Conference Secretariat

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Invited Speakers

Linda **van Aelst**, Cold Spring Harbor
Stylios **Antonarakis**, Geneva
Hans **van Bokhoven**, Nijmegen
Nigel **Carter**, Cambridge
Jamel **Chelly**, Paris
Laurence **Colleaux**, Paris
Daniel **Geschwind**, Los Angeles
Matthew **Hurles**, Cambridge
Andreas **Kuss**, Berlin
Eric **Legius**, Leuven
Jacques L. **Michaud**, Montréal
Markus **Missler**, Münster
Danielle **Posthuma**, Amsterdam
André **Reis**, Erlangen
Hans-Hilger **Ropers**, Berlin
Annette **Schenck**, Nijmegen
Stephen T. **Warren**, Atlanta

Sponsors



German Mental Retardation Network (MRNET)

International Conference on Genetics and Neurobiology of Mental Retardation

Biennial Conference of the German Academy of Sciences Leopoldina National Academy of Sciences



29th September - 1st October, 2010
Erlangen, Germany

