Formazione a distanza sincrona – formazione residenziale

20TH TROINA MEETING ON GENETICS OF NEURODEVELOPMENTAL DISORDERS

La Cittadella dell'Oasi
Oasifad.it
15-18 aprile 2026

Rilevanza

Da 19 anni questo Meeting rappresenta un'occasione privilegiata per i partecipanti di scambiarsi le acquisizioni più recenti nel campo della genetica del neurosviluppo. Un Comitato Scientifico Internazionale seleziona ogni anno una lista di relatori invitati ai quali si associano comunicazioni brevi di giovani ricercatori, appositamente selezionate. Tutti i partecipanti possono interagire direttamente con i relatori e possono iniziare nuovi progetti di ricerca a partire dalle suddette interlocuzioni.

Finalità

Il Meeting intende aggiornare i partecipanti sugli ultimi studi e ricerche inerenti i disordini genomici e i nuovi geni coinvolti nell'eziologia dei Disturbi del neurosviluppo.

Obiettivi specifici

Grazie al Convegno i partecipanti saranno in grado di

- Ampliare le proprie conoscenze disordini genomici e i nuovi geni coinvolti nell'eziologia dei Disturbi del neurosviluppo
- Accrescere le proprie competenze nell'ambito della ricerca mutazioni e la genetica nell'ambito dei disturbi dello spettro dell'autismo, della disabilità intellettiva, delle epilessie.
- Confrontarsi con qualificati ricercatori provenienti dai maggiori centri di ricerca internazionali.

Obiettivo formativo ecm

18 -Contenuti tecnico-professionali (conoscenze e competenze) specifici di ciascuna professione, di ciascuna specializzazione e di ciascuna attività ultraspecialistica, ivi incluse le malattie rare e la medicina di genere

Metodologia didattica

Relazioni su tema

Modalità per la valutazione dell'apprendimento

Questionario a risposta multipla

Destinatari

L'evento è destinato alle seguenti figure professionali: biologo, farmacista, medico (tutte le discipline), tecnico di laboratorio biomedico, Studenti

Programma

15th Wednesday

09:00 Introduction
Corrado Romano, Bert De Vries, Frank Kooy, Christa Lese Martin, Heather Mefford

Session 1 Clinic:
Chair: Bert de Vries (Nijmegen, The Netherlands)

09:45 Opening speaker:
Jozef Gecz (Adelaide, Australia) jozef.gecz@adelaide.edu.au
"Cerebral Palsy, a new kid in the NDD Block"

10:15 Britt-Marie Anderlid (Stockholm, Sweden) britt.marie.anderlid@ki.se
"Whole Genome Sequencing in the Clinical Setting"

10:35 David Koolen (Nijmegen, The Netherlands) david.koolen@radboudumc.nl
"Two Decades of Advancing Knowledge in Koolen-de Vries Syndrome: From Mechanism to Clinical insight"

10:55 Break

11:15 Alexandre Reymond (Lausanne, Switzerland) alexandre.reymond@unil.ch
"Variable expressivity, reduced penetrance, pleiotropy and mode of inheritance"

11:35 discussion

12.00-13.00 Short communications 1

12.00 - 12.15 Tinatin Tkemaladze (Tbilisi, Georgia) T.tkemaladza@tsmu.edu
Rare and Ultra-Rare Diseases in Genetic Isolates from Georgia – Preliminary Study Results.

12.15 - 12.30 Jolijn Verseput (Nijmegen, The Netherlands) jolijn.verseput@radboudumc.nl
Recurrent missense variants in the endosomal trafficking protein RAB14 cause a neurodevelopmental disorder.

12.30 - 12.45 Lorenzo Perilli (London, UK) l.perilli@ucl.ac.uk
Expanding the Genetic Landscape of cHSP: Clinical Evidence for SYNRG as a Novel Disease Gene.

12.45 - 13.00 Alexander Dingemans (Nijmegen, The Netherlands) A.Dingemans@radboudumc.nl Deep phenotyping without compromise: open privacy-preserving AI extracts clinically relevant phenotypes from EHR data at scale.

13.00 – 15.00 Lunch

Session 2 Repeat disorders:

Chair: Frank Kooy (Antwerp, Belgium)

15.00 Opening speaker
Jean-Louis Mandel (Strasbourg, France) jlmandel@igbmc.fr
"Troina 1987-2013-2026: From FraX and XLID to some current general issues on XLID genes and NDDs"

15.30 David Nelson (Houston, USA) nelson@bcm.edu
"Fragile X, CGG and FMR1 at 35: remaining questions and prospects to therapy"

15.50 Egor Dolzhenko (San Diego, USA) edolzhenko@pacificbiosciences.com
"Genome-wide characterization of mosaicism in tandem repeat regions"

16.10 Christopher Pearson (Toronto, Canada) christopher.pearson@sickkids.ca
"What Molecular, Cellular and Clinical Phenotypes are actually connected to somatic repeat expansions?"

16.30 discussion

17.00 Break

17.30 -19.00 Short communications 2

17.30 - 17.45 Dale Annear (Antwerp, Belgium) dale.annear@uantwerpen.be
"Next-generation mouse phenotyping reveals multifaceted drug effects in a mouse model of Fragile X Syndrome"

17.45 - 18.00 Lisanne Wit (Nijmegen, The Netherlands) Lisanne.wit@radboudumc.nl
"Polygenic scores contribute to monogenic neurodevelopmental disorder variability: A literature review and pilot study"

18.00 - 18.15 Lara Colombo (Ghent, Belgium) lara.colombo@ugent.be
"Systematic comparative analysis of 3D genome structure between neuronal and clinically accessible tissues highlights tissue-specific chromatin features at disease loci"

18.15 - 18.30 Kyra Swildens (Rotterdam, The Netherlands) k.swildens@erasmusmc.nl
"The role of RNF12 in X-linked intellectual disability syndrome TOKAS"

18.30 - 18.45 Marina Hommersom (Nijmegen, The Netherlands)
Marina.Hommersom@radboudumc.nl
"Human neuronal networks on micro-electrode arrays as a tool to assess genotype-phenotype correlation in CACNA1A-related disorders"

18.45 - 19.00 Nikki Kolsters (Nijmegen, The Netherlands) Nikki.Kolsters@radboudumc.nl
"Assessing the excitatory-inhibitory balance in a human iPSC-derived neuronal model for neurodevelopmental disorders"

16th Thursday

Session 3 Psychiatry/autism and brain
Chair: Heather Mefford (Memphis, USA)

09:00 Opening speaker
Thomas Bourgeron (Paris, France) thomasb@pasteur.fr
"A multiscale stratification of autism: from medicine to neurodiversity"

09:30 Christa Lese Martin (Danville, USA) clmartin1@geisinger.edu
"Reporting Neuropsychiatric CNVs from a Population Genomic Screening in a Health Care System"

09:50 David Ledbetter (Tallahassee, USA) david.ledbetter@med.fsu.edu
"Clustering analysis of neurodevelopmental genes reveals distinct phenotypic profiles across cognitive, behavioral, psychologic and motor domains"

10:10 Hitoshi Okazawa (Tokyo, Japan) okznpat@tmd.ac.jp
"Innate immune functions of an ID gene: PQBP1"

10:30 discussion

11:00 Break

11:30 Sebastien Jacquemont (Montreal, Canada) sebastien.jacquemont.hsj@ssss.gouv.qc.ca
"Cell type and Brain network Architecture of autism and related Neurodevelopmental Disorders

11:50 Stephanie Baulac (Paris, France) stephanie.baulac@icm-institute.org
"Somatic Mutations and Mosaicism in Epilepsy and Cortical Malformations"

12:10 Joris Vermeesch (Leuven, Belgium) joris.vermeesch@uzleuven.be
"Unraveling the role of segmental Duplicons in predisposition and phenotypic variability of the 22q11.2 Deletion syndrome"

12:30 discussion

13.00 – 15.00 Lunch

Session 4 Epigenetics/RNA
Chair: Jozef Gecz (Adelaide, Australia)

15:00 Opening speaker
Rosanna Weksberg (Toronto, Canada) rweksb@sickkids.ca
"Episignatures for NDD: Impact on Delivering Precision Medicine"

15:30 Christel Depienne (Essen, Germany) christel.depienne@uk-essen.de
"Pathogenic variants in snRNA genes: a major cause of dominant and recessive neurodevelopmental disorders".

15:50 Angel Barco (Alicante, Spain) abarco@umh.es
"Epigenetic etiology of Intellectual Disability"

16:10 Peng Jin (Atlanta, USA) peng.jin@emory.edu
"RNA dysregulation in Neurodevelopmental Disorders"

16:30 Sarah Vergult (Ghent, Belgium) Sarah.Vergult@ugent.be
"Non-coding structural variants disrupt FOXG1 transcriptional regulation in early neurodevelopment"

16:50 discussion

17:10 Break

17:30-19:00 Short communications 3

17.30 - 17.45 Sebastian Leimbacher (Ghent, University) Sebastian.Leimbacher@ugent.be
"Regulatory dynamics of neurodevelopmental disease genes during early neural progenitor cell differentiation"

17.45 - 18.00 Mazen Khaddour (Milan, Italy) mazen.khaddour@fht.org
"Transcriptomics convergence and divergence of monogenic neurodevelopmental disorders"

18.00 - 18.15 Elsa Leitão (Essen, Germany) elsa.leitao@uk-essen.de
"Concepts and challenges in RNUopathies: the example of RNU2-2 disorders"

18.15 - 18.30 Claudio Peter D'Incal (Antwerp, Belgium) Claudio.DIncal@uantwerpen.be
"The Female Side of Autism: Sexual Dichotomies impacting the Helsmoortel-Van der Aa syndrome pathology"

18.30 - 18.45 Mara Graziani (Nijmegen, The Netherlands) Mara.Graziana@radboudumc.nl
"From genes to networks: *CHD2* haploinsuffiency in microglia shapes neuronal connectivity"

18.45 - 19.00 Katrin Linda (Nijmegen, The Netherlands) Katrin.Linda@radboudumc.nl
"Intercellular Autophagy: Astrocytes Degrade Neuronal Autophagosomes"

17th Friday: SOCIAL PROGRAM: TRIP TO ETNA

18th Saturday

Session 5 Modelling: cell and animal
Chair: Evan Eichler (Seattle, USA)

09:00 Opening speaker
Nael Nadif Kasri (Nijmegen, The Netherlands) Nael.nadifkasri@radboudumc.nl
"Multimodal mapping of neuronal network activity and transcriptomic convergence in neurodevelopmental disorders"

09:30 Giuseppe Testa (Milan, Italy) giuseppe.testa@fht.org
Tittle unknown

09:50 Silvia De Rubeis (New York, USA) silvia.derubeis@mssm.edu
"Sex-specific impact of *Ddx3x* alterations in developing and adult brain circuits"

10:10 Santhosh Girirajan (Penn State University Park, USA) sxg47@psu.edu
"Evolutionary history and structural diversity of disease-associated 16p12.2 locus"

10.30 - 11.45 Short communications 4

10.30 - 10.45 Nore Van Loon (Ghent, Belgium) nore.vanloon@ugent.be
Unravelling the functions of FOXG1 in neurodevelopment and disease.

10.45 - 11.00 Lukas Genbrugge (Ghent, Belgium) lukas.genbrugge@ugent.be
Modeling the choroid plexus – neural stem cell axis in neurodevelopmental BAFopathies using choroid plexus organoids.

11.00 - 11.15 Ann-Sofie De Meulemeester (Paris, France) as.demeulemeester@icm-institute.org
Modeling FCDII using inducible MTOR cortical organoids.

11.15 - 11.30 Cristina Gontan (Rotterdam, The Netherlands) m.gontanpardo@erasmusmc.nl
Resetting X Chromosome Inactivation in Female Human Pluripotent Stem Cells: A Dual Reporter Platform for Disease Modeling and Therapeutic Screening

11.30 - 11.45 Salma Amin (Milan, Italy) salma.amin@fht.org
Brain organoids from Cohen syndrome patients reveal a novel link between lysosomes and primary cilia in human brain

11:45 Break

Session 6:
Chair: Corrado Romano (Troina, Italy)

12:15 Evan Eichler (Seattle, USA) ee3@uw.edu
"From CNVs to genes: Understanding the genetic architecture of autism and developmental delay"

12:45 discussion

13:45 Closing remarks

Comitato Scientifico

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

Frank Kooy, Medical genetics departement – 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 – Responsabile UOSR di malattie rare e disturbi del neurosviluppo IRCCS Oasi Maria SS. – Troina – Professore Associato Università degli Studi di Catania – Responsabile scientifico ECM

Relatori

Stephanie Baulac, Research director and Group leader - Paris Brain Institute/Institut du Cerveau (ICM) - (Paris, France)

Thomas Bourgeron, Director of the unit "Human Genetics and Cognitive Functions" at the Neuroscience Department of the Pasteur Institute. Institut Pasteur - Assistant Professor at University Denis Diderot Paris 7, France)

Britt-Marie Anderlid, Associate professor and clinical geneticist, Department of Molecular Medicine and Surgery, Karolinska Institutet, (Stockholm, Sweden)

Angel L. Barco Guerrero, Group leader, Profesor de Investigación CSIC, Alicante, Spain

Christel Depienne, Associate Professor , Principal Investigator, Institute of Human Genetics, University, Essen, Germany

Silvia De Rubeis, Assistant Professor at the Seaver Autism Center for Research and Treatment, Department of Psychiatry, Mindich Child Health and Development Institute, and Friedman Brain Institute at the Icahn School of Medicine at Mount Sinai Hospital (New York – USA)

Egor Dohlezenko, Principal Bioinformatics Scientist, Pacific Bio Sciences (PacBio), (San Diego, Calfornia – USA)

Evan Eichler, Principal Investigator Eichler Lab - Professor of Genome Sciences, University of Washington (U.S.A.)

Jozef Gecz, Head Neurogenetics, Adelaide Medical School, Faculty of Health and Medical Sciences – University of Adelaide – Australia

Santhosh Girirajan, Professor of Genomics; Professor of Anthropology; Director Girirajan Lab, Department Head of Biochemistry and Molecular Biology, (Pennsylvania State University – USA)

Sebastien Jacquemont, Medical Geneticist, CHU Sainte Justine; Full Professor, Department of Pediatrics, Faculty of Medicine, University of Montréal (Canada)

Nael Nadif Kasri, Associate Prof. Dept Human Genetics Cognitive Neurosciences Radboudumc Donders Institute for Brain, Cognition and Behaviour - Nijmegen, The Netherlands

David Koolen, Clinical Geneticist - Radboud University Medical Centre - Nijmegen, The Netherlands

Peng Jin, Professor and Chair, Department of Human Genetics, Emory University School of Medicine – Atlanta (USA)

David H. Ledbetter, Professor, Departments of Pediatrics and Psychiatry, University of Florida College of Medicine Jacksonville (USA)

Jean-Louis Mandel, Professor of Genetics, Institute of Genetics and Molecular and Cellular Biology (IGBMC), University of Strasbourg, Strasbourg, France

Christa Lese Martin, Chief Scientific Officer / Director, Autism & Developmental Medicine Institute, Geisinger, Danville, USA

David L. Nelson, Professor, Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, USA

Hitoshi Okazawa, Professor and Chair, Department of Neuropathology, Medical Research Institute, Tokyo Medical and Dental University, Tokyo, Japan

Christopher Pearson, Genetics – University of Toronto – Canada

Alexandre Reymond, Professor, Past President of the European Society of Human Genetics and the current Chair of its Scientific Program Committee Lausanne, Switzerland

Giuseppe Testa, Direttore Laboratorio di Epigenetica delle Cellule Staminali – Istituto Europeo Oncologico – Milano – Professore Ordinario di Biologia Molecolare - Universita degli Studi di Milano - Direttore del Centro di Neurogenomica, Human Technopole, Milan – Italy

Joris Vermeesch, Full professor Faculty of Medicine - Department chair of department of Human Genetics - Head of the Laboratory for Cytogenetics and Genome Research - Katholieke Universiteit (KU) Leuven (Belgium)

Rosanna Weksberg, Professor of Paediatrics and Medical Genetics, Senior Associate Scientist, Genetics and Genome Biology - Hospital for Sick Children (SickKids) (Toronto, Canada)

Sara Vergult, Assistant professor - Faculty of Medicine and Health Sciences, Department of Biomolecular Medicine - Ghent University, Belgium

Segreteria organizzativa

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Informazioni generali

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c) Selezionare il corso e cliccare su "accedi". È possibile che il sistema richieda di effettuare l'iscrizione
d) Selezionare il menu "Accesso al meeting"

Si consiglia di scaricare l'ultima versione di zoom.

Fonti di finanziamento

Il corso è finanziato attraverso i fondi della ricerca.

Modifiche al programma e annullamento corso

In caso di mancato raggiungimento del numero minimo di iscritti o per altri giustificati motivi l'IRCCS si riserva la facoltà di annullare o rinviare il corso dandone comunicazione scritta, alla mail indicata.

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NOME COGNOME	PROFESSIONE	DISCIPLINA	ENTE DI APPARTENENZA/LIBERA PROFESSIONE	DESCRIZIONE ATTVITA'PROFESSIONALE/FORMATIVA
Stephanie Baulac	Medical Geneticist	Medical Genetics	Paris Brain Institute/Institut du Cerveau (ICM) - (Paris, France)	Research director and Group leader
Thomas Bourgeron	Director	Human Genetics and Cognitive Functions	Pasteur Institute / University Denis Diderot Paris 7	Director of unit and Assistant Professor
Britt-Marie Anderlid	Associate Professor	Medical Genetics	Karolinska Institutet, (Stockholm, Sweden)	Clinical geneticist, Dept. of Molecular Medicine and Surgery
Angel L. Barco Guerrero	Profesor de Investigación CSIC	Neuroscienze	Alicante, Spain	Group leader
Christel Depienne	Associate Professor	Human Genetics	Institute of Human Genetics, University, Essen, Germany	Principal Investigator
Silvia De Rubeis	Assistant Professor	Psychiatry / Neuroscience	Seaver Autism Center, Icahn School of Medicine at Mount Sinai (New York, USA)	Mindich Child Health and Development Institute
Egor Dohlezenko	Principal Scientist	Bioinformatics	Pacific Bio Sciences (PacBio), (San Diego, California – USA)	Principal Bioinformatics Scientist
Evan Eichler	Professor of Genome Sciences	Genome Sciences	University of Washington (U.S.A.)	Principal Investigator Eichler Lab
Jozef Gecz	Head Neurogenetics	Neurogenetics	Adelaide Medical School, University of Adelaide – Australia	Faculty of Health and Medical Sciences

Santhosh Girirajan	Professor / Director	Genomics and Anthropology	Pennsylvania State University – USA	Director Girirajan Lab, Dept. Head of Biochemistry and Molecular Biology
Sebastien Jacquemont	Medical Geneticist / Full Professor	Pediatrics / Genetics	CHU Sainte Justine; University of Montréal (Canada)	Department of Pediatrics, Faculty of Medicine
Nael Nadif Kasri	Associate Prof.	Human Genetics / Cognitive Neurosciences	Radboudumc Donders Institute (Nijmegen, The Netherlands)	Donders Institute for Brain, Cognition and Behaviour
David Koolen	Clinical Geneticist	Medical Genetics	Radboud University Medical Centre - Nijmegen, The Netherlands	Attività clinica specialistica
Peng Jin	Professor and Chair	Human Genetics	Emory University School of Medicine – Atlanta (USA)	Direzione Dipartimento di Genetica Umana
David H. Ledbetter	Professor	Pediatrics and Psychiatry	University of Florida College of Medicine Jacksonville (USA)	Attività accademica dipartimentale
Jean-Louis Mandel	Professor of Genetics	Genetics	IGBMC, University of Strasbourg (France)	Institute of Genetics and Molecular and Cellular Biology
Christa Lese Martin	Chief Scientific Officer / Director	Autism & Developmental Medicine	Geisinger, Danville, USA	Autism & Developmental Medicine Institute
David L. Nelson	Professor	Molecular and Human Genetics	Baylor College of Medicine, Houston, USA	Ricerca e insegnamento
Hitoshi Okazawa	Professor and Chair	Neuropathology	Tokyo Medical and Dental University (Japan)	Medical Research Institute

Christopher Pearson	Ricercatore	Genetics	University of Toronto – Canada	Attività nel settore genetica
Alexandre Reymond	Professor	Genetics	Lausanne, Switzerland	Chair of Scientific Program Committee (ESHG)
Giuseppe Testa	Professore Ordinario	Biologia Molecolare	IEO Milano / Human Technopole / Univ. di Milano	Direttore Laboratorio di Epigenetica e Centro di Neurogenomica
Joris Vermeesch	Full professor	Human Genetics / Cytogenetics	Katholieke Universiteit (KU) Leuven (Belgium)	Department chair and Head of Laboratory
Rosanna Weksberg	Professor of Paediatrics	Medical Genetics	Hospital for Sick Children (SickKids), Toronto, Canada	Senior Associate Scientist, Genetics and Genome Biology
Sara Vergult	Assistant professor	Biomolecular Medicine	Ghent University, Belgium	Faculty of Medicine and Health Sciences

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- di aver fornito all'interessato l'informativa sul trattamento dei dati personali (art. 13 del Regolamento europeo 2016/679; artt. 68, 70, 76, 96 Accordo Stato-Regioni 2017 "La formazione continua nel settore salute"- Rep. Atti 14/CSR del 2.2.2017 - Par. 4.6, lett. j) Manuale Nazionale di Accreditamento per l'Erogazione di Eventi ECM);
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