







COMMITTEES

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CME AND ANIMAL EXPERIMENTER ACCREDITATIONS



Accreditation Statement

The 3rd International Conference of the Trisomy 21 Research Society, Barcelona, Spain, 06/06/2019-09/06/2019 has been accredited by the European Accreditation Council for Continuing Medical Education (EACCME®) with 30 European CME credits (ECMEC®s). Each medical specialist should claim only those hours of credit that he/she actually spent in the educational activity.

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Live educational activities, occurring outside of Canada, recognised by the UEMS-EACCME® for ECMEC®s are deemed to be Accredited Group Learning Activities (Section 1) as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada.

Continuing Education in Animal Handling

The following sessions will offer the possibility to participants to be accredited as Continuing Education in Animal Handling:

Mouse master class (9:00-13:00, June 6, 2019) *4 hours

SESSION 2A: New disease-relevant behavioral perspective in preclinical intellectual disability studies (9:30-11:30, June 7, 2019) *2 hours

SESSION 4: Investigating Down syndrome phenotypes and mechanisms through comprehensive mouse modeling (18:00-20:00, June 7, 2019) *2 hours

For the Continuing Education in Animal Handling is obligatory to scan at the entrance and exit from the meeting room the QR code of your badge.



	Wednesday, June 5, 2019
15.30 - 19.00	Registration
	Room: TAU
16.00 - 18.00	Industry Session Top Challenges Facing the Pharmaceutical Industry in Down syndrome Drug Development.

		Thursday, June 6, 2019	
08:30-18:00		Registration	
	Room: ALPHA	Room: BETA	Room: GAMMA
		PRE-MEETING SATELLITE SYMPOSIA	
	CLINICAL SES	SIONS	PRECLINICAL SESSION
09:00-13:00	Health co-morbidities in older adults with Down syndrome - assessment and diagnosis Chair: Andre Strydom (University College London,UK) T21RS Committee for clinical research (UK)	Health co-morbidities in children with Down syndrome Chairs: Stephanie Sherman (Emory University, USA), Stefano Vicari (Bambino Gesu' Children's Hospital, IT)	Mouse master class Chair: Yann Hérault (Institute de Génétique et de Biolog Moléculaire et Cellulaire, FR) T21RS Committee for Preclinical research
		Room: AUDITORIUM	
14:00		START OF MEETING	
14:00-14:30		WELCOME NOTE Mara Dierssen (CRG, SP) President T21RS and local authorities	
14:30-16:30	SCIENTIFIC SESSION 1 (Parallel) Three of a perfect pair: stereotyped homeostatic compromises in trisomies Chair: Pablo Helquera (INIMEC-CONICET, AR)		
16:30-18:00	Poster Session 1 and selected Nanosymposia with coffee 16: 30 - 18:00 Poster Session 1: PO18 - PO61, PO145 (Foyer Hypatia) 17:00 - 17:30 Nano-symposium #8: PO13 - PO17 (Room Gamma) 17:30 - 18:00 Nano-symposium #3: PO01 - PO06 (Room Alpha) 17:30 - 18:00 Nano-symposium #6: PO07 - PO012 (Room Beta)		
		Room: AUDITORIUM	
18:00-19:00	Chair:	JEROME LEJEUNE PLENARY LECTURE Genetics of Human Chromosome 21 Stylianos Antonarakis (University of Geneva, CH) Thierry de la Villejegu (Jerome Lejeune Foundation,	FR)
19:00- 19:10	Montserrat Trueta Award Mara Dierssen (President T21RS), Katy Trias (Fundació Catalana Sindrome Down, SP)		
		Room: AGORA	
19:30-19:50	Pro	"I am able" sentation of an art project by Quim and Jesús Vilama	ó
		Room: AUDITORIUM	
20:00-21:00		"Cactus, sólo muere lo que se olvida" ng actors with Down syndrome, directed by Victor Igr npañía Nacional de Teatro Manantial de Ilusión (Chile	
		Foyer: HYPATIA (AMBAIXADES)	

	Friday, June	e 7, 2019
08:00-18:00	Registra	ition
	Room: AUD	TORIUM
	THE EMBO KEYN Human-specific genes, neural stem cell amplification, and ne	
08:30-09:30	Wieland Huttner (Max P. Chair: Anita Bhattacharyya (Un	
	Room: AGORA	Room: AUDITORIUM
	SCIENTIFIC SESSIO	DN 2 (Parallel)
	Session 2A	Session 2B
09:30-11:30	New disease-relevant behavioral perspective in preclinical intellectual disability studies	Biomarkers for Alzheimer's disease in Down syndrome Chairs:
05.30-11.30	Chairs: María Martínez de Lagrán (CRG, SP) Anna Vázquez (University Pompeu Fabra, SP)	Bradley Christian (University of Wisconsin, USA) María Carmona Iragui (Hospital Sant Pau, SP)
11:30-12:00	Coffee break - Foyer : H	ypatia (Ambaixades)
	Room: AGORA	Room: AUDITORIUM
	SCIENTIFIC SESSION	DN 3 (Parallel)
	Session 3A	Session 3B
	New directions in human trisomy 21 stem cell research	Alzheimer's disease in the context of the trisomy of
12:00-14:00	Chair: Anita Bhattacharyya (University of Wisconsin, USA)	Hsa21 - preclinical studies Chairs:
	Ainta Brattacharyya (omversity of Wisconsin, osa)	Frances Wiseman (University College London, UK) William Mobley (University of California-San Diego, USA)
14:00-15:00	Lunch b	reak
	Room: AUD	TORIUM
15:00-16:00	T21RS General	Assembly
	Poster Session 2 and selected	
16:30-18:00	16:30 - 18:00 Poster Session 2: PO75 - PO	
10.50-10.00	16:00 - 17:00 Special symposium "Microbiota, nutrition and 17:00 - 17:30 Nanosymposia #9 F	
	17:30 - 18:00 Nanosymposia #4	· · · · · · · · · · · · · · · · · · ·
	Room: AUDITORIUM	Room: ALPHA
	SCIENTIFIC SESSIO	DN 4 (Parallel)
	Session 4A	Session 4B
	Investigating Down syndrome phenotypes and mechanisms	Cognitive outcomes in children with Down syndrome
18:00-20:00	through comprehensive mouse modeling Chairs:	Chairs: Anna Esbensen (Cincinnati Children's Hospital Medical Cente.
18.00-20.00	Elizabeth Fisher (University College London, UK) Veronique Brault (Institut de Génétique et de Biologie Moléculaire et Cellulaire, FR)	USA) Lisa Jacola (St Jude Children's Research Hospital, USA)



	Saturday, J	lune 8, 2019	
08:00-18:00		tration	
	Room: Al	JDITORIUM	
08:30-09:30	PLENARY LECTURE 3 Alzheimer's disease mechanisms and therapeutics Li-Hue Tsai (Massachusetts Institute of Technology, USA) Chair: Victor Tybulewicz (Francis Crick Institute, London, UK)		
	Room: AUDITORIUM	Room: AGORA	
	SCIENTIFIC SESSION 5 (Parallel)		
09:30-11:30	Session 5A Modifications of autophagy-lysosomal-endosomal pathways in Down syndrome and other neurodegenerative diseases Chairs: Marie Claude Potier (ICM, FR) Eric Hamlett (Medical University of South Carolina, USA)	Session 5B Language Outcome Measures for Treatment Studies Chairs: Len Abbeduto (University of California-Davis, USA) Laura del Hoyo (University of California-Davis, USA)	
11:30-12:00	Coffee break - Foyer	: Hypatia (Ambaixades)	
	Room: Al	JDITORIUM	
12:00 - 15:35	SCIENCE & SOCIETY SYMPOSIUM - PROGRAM FOR FAMILIES T21rs Committee for Science & Society Peter Paul De Deyn (University Medical Center Groningen, NL)(Chairman), Alain Dekker (University Medical Center Groningen, NL), Juan Fortea (Fundació Catalana Síndrome de Down, SP), Sebastian Videla (Fundació Catalana Síndrome de Down, SP), Lotta Granholm (University of Denver, USA), Cindy Lemere (Brigham and Women's Hospital, Harvard Medical School, USA)		
13:30	Lunch break and Meet the Scientist		
15:45-17:45	The Role of Inflammation and NGF dysfunction in cognitive Cl Cl Jorge Busciglio (Universi	c Session 6 decline and Alzheimer's disease pathology in Down syndrome nair: ty of California-Irvine, USA) sité de Montreal, Canada)	
17:45-19:00	Poster Session 3 and selected Nanosymposia with coffee 17:45 - 19:00 Poster Session 3: PO127 - PO166 (Foyer: Hypatia) 18:00 - 18:30 Nanosymposia #2: PO121 - PO 123 (Room: Beta) 18:00 - 18:30 Nanosymposia #7: PO124 - PO126 (Room: Gamma) 18:30 - 19:00 Nanosymposia #1: PO118 - PO120 (Room: Alpha)		
	Room	AGORA	
19:00 - 20:00	MEET THE EXPERTS SESSIONS with aperitif Li-Huei Tsai (Massachusetts Institute of Technology, USA)		
20:00 - 21:00	How should we study Down syndrome: pro T21RS Committee fo <i>Renata Bartesaghi</i> (Univrsity of Bologna, IT) (Chair	s and cons of mouse models and human iPSCs r Education & Training:), Carmen Martínez-Cué (University of Cantabria, SP), ndrome, USA), Tarik Haydar (Boston University, USA)	

	Sunday, Jun	ie 9, 2019	
08:00-18:00	Registra	ation	
	Room: AUD	ITORIUM	
	PLENARY LE	CTURE 4	
	Alzheimer's disease in Down syndrome: update from the LonDowns consortium		
08:30-09:30	Andre Strydom (University College London, UK)		
	Chair: Juan Fortea (San	t Pau Hospital, SP).	
00-20 10-15	Annette Karmiloff-Smith and Michael	Harpold Thesis Awards Ceremony	
09:30-10:15	Renata Bartesaghi (Unive	ersity of Bologna, IT)	
10:15-11:00	Coffee break - Foyer : H	ypatia (Ambaixades)	
	Room: AGORA	Room: AUDITORIUM	
	SCIENTIFIC SESSIC	N 7 (Parallel)	
	Session 7A	Session 7B	
	Leukemia in Children with Down syndrome: why does it	Metabolic defects in Down syndrome: from peripher	
11:00-13:00	hapen and what can we do about it?	to the brain	
	Chairs:	Chairs:	
	John Crispino (Northwestern, USA)	Eugenio Barone (Sapienza University of Rome, IT)	
	Sebastien Malinge (Telethon Kids Institute, USA)	Marzia Perluigi (Sapienza University of Rome, IT)	
13:00-14:00	Lunch b	reak	
	Room: AUD	TORIUM	
	SCIENTIFIC SESSION 8		
	Correction of circuit specific GABAergic over-inhibiti		
14:00-16:00	Chair: Floriana Costanzo (Bambino Gesù Children's Hospital, IT)		
	Fioriana Costanzo (Bambino Gi Jean Maurice Delabar (Paris Sor		
	Jeun Maurice Delubur (Puns 30)	boline Oniversite, Civis, FN)	
16:00-16:30	Coffee break - Foyer : H	ypatia (Ambaixades)	
16:30-17:00	Melissa Parisi: updates on US NIH f	unding (ABC-Connect, INCLUDE)	
16:30-17:00	Chair: Roger Reeves (Johns Hopkins U	niversity School of Medicine, USA)	
	Room: AUD	ITORIUM	
	SCIENTIFIC S		
	Endpoints for cl	inical trials	
Endpoints for clinical trials 17:00-19:00 Chair:			
	Diana Bianchi (NIH	I-NICHD, USA)	
	Alain Dekker (University Medi	cal Center Groningen, NL)	
	CLOSING CEI	REMONY	
19:00	Posters awards and Honorar	y Members presentation	
	with aperitif (Foy	ver: Hypatia)	
	END OF M		



SCIENTIFIC PROGRAMME

Wednesday, June 5, 2019

15.30 - 19.00 Registration

Room: TAU

16:00 - 18:00 Industry session. (Page 24)

Thursday, June 6, 2019

08.30 - 18:00 Registration

09:00 – 13:00 PRE-MEETING SATELLITE SYMPOSIA (parallel)

CLINICAL SESSIONS

T21RS Committee for clinical research: Andre Strydom (King's College London, UK) (chair)

Room: ALPHA

CLINICAL SESSION 1: Health co-morbidities in older adults with Down syndrome – assessment and diagnosis

Adult clinical committee: Shahid Zaman (University of Cambridge, UK), Juan Fortea (Fundació Catalana Síndrome de Down and Hospital San Pau, SP), Wayne Silverman (Kennedy Krieger Institute and Johns Hopkins University School of Medicine, USA), Tonnie Coppus (Radboud University Medical Center, NL), Ben Handen (University of Pittsburgh, USA), Elizabeth Head (University of Kentucky, USA), Weihong Song (University of British Columbia, CA)

- 09:00 09:30 Registration / coffee
- 09:30 09:45 Welcome and Introduction. Andre Strydom (King's College London, UK)
- **09:45 10:15** Overview of the health needs of older adults. *Tonnie Coppus (Radboud University Medical Center, NL)*
- 10:15 10:45 Sleep Apnea. Dr. Anne-Sophie Rebillat (Institut Jerome Lejuene, FR)
- 10:45 11:00 Break
- 11:00 11:30 Cognitive function in aging adults. Sharon Krinsky-McHale (New York State Institute for Basic Research in Developmental Disabilities, USA)
- 11:30 12:00 Dementia management and maintaining functional abilities. (Professor Mary McCarron, Trinity college, Dublin, Ireland)
- **12:00 12:30** Summary and discussion

Room: BETA

CLINICAL SESSION 2: Health co-morbidities in children with Down syndrome

Child/developmental subcommittee: Stephanie Sherman (Emory University, USA) — co-chair; Stefano Vicari (Bambino Gesu' Children's Hospital, IT) — co-chair, Cecile Cieuta-Walti (Institut Jerome Lejuene, FR), Brain Skotko (Massachusetts General Hospital, USA), Julie Korenberg (University of Utah, USA), Silvia Sacco (Institut Jerome Lejuene, FR), Rafael de la Torre (IMIM-Hospital del Mar Medical Research Institute, SP), Jamie Edgin (University of Arizona, USA), Stefania Veltri (IT)), Andre Strydom (King's College, London, UK), Maria Stanley (University of Wisc, USA), Floriana Costanzo (Bambino Gesu' Children's Hospital, IT)

09:00 - 09:30 Registration / coffee

09:30 – 10:00 Welcome; Overview of Medical Research in Down Syndrome. Stephanie Sherman (Emory University, USA)

10:00 – 10:35 Medical comorbidities of childhood and their potential impact on cognition and development. George Capone (Kennedy Krieger Institute, USA)

10:35 - 10:50 Break

10:50 – 11:25 Psychiatric comorbidities of childhood/adolescents and their association with cognitive impairment. Stefano Vicari (Ospedale Pediatrico Bambino Gesu, IT)

11:25 – 12:00 Interve<mark>ntions from early childhood to adolescents—how we can help. Rafael de la Torre Fornell, (Universitat Pompeu Fabra, SP)</mark>

12:00 - 12:30 Panel: Q/A and discussion

PRECLINICAL SESSION

T21RS Committee for Preclinical research: Yann Hérault (Institut de Génétique et de Biologie Moléculaire et Cellulaire, FR)(Chair), William Mobley (University of California-San Diego, USA), Elizabeth Fisher (University College London, UK), Marie-Claude Potier (ICM, FR), Stylianos Antonarakis (University of Geneva, CH), Jean-Maurice Delabar (CNRS, FR), Eugene Yu (Roswell Park Cancer Institute, USA), Katheleen Gardiner (University of Colorado, USA), Mara Dierssen (Centre for Genomic Regulation (CRG), SP), Anita Bhattacharyya (University of Wisconsin, USA), Randall Roper ((Indiana University-Purdue University Indianapolis, USA)

Room: GAMMA

Mouse master class

09:00 – 09:15 Registration / coffee

09:15 – 09:45 Overview of currently available preclinical mouse models. *Yann Hérault (Institut de Génétique et de Biologie Moléculaire et Cellulaire, FR)*

09:45 – 10:15 Genetic quality control, maintenance and breeding procedures. Eugene Yu (Roswell Park Cancer Institute, USA)

10:15 – 10:45 Classical behavioral phenotyping. Mara Dierssen (Centre for Genomic Regulation (CRG), SP)

10:45 - 11:00 Break

11:00 – 11:30 Craniofacial and skeletal phenotyping. *Randall Roper (Indiana University-Purdue University Indianapolis, USA)*



11:30 – 12:00 Electrophysiology. William Mobley (University of California-San Diego, USA)

12:00 – 12:30 Preclinical testing. Jean-Maurice Delabar (CNRS, FR)

12:30 – 13:00 Discussion on key information about models and their availability (moderated by Lizzy Fisher) and perspective on new animal models for Down syndrome research. Roger Reeves (Johns Hopkins University School of Medicine, USA) and Victor Tybulewicz (Francis Crick Institute, UK)

Room: AUDITORIUM

14:00 START OF THE MEETING

14:00 - 14:30 Welcome note.

Mara Dierssen (President T21RS)

Angel Font (Corporate Director of Research and Strategy "la Caixa" Foundation)

Gemma Tarafa (Commissioner for Health and Functional Diversity, City Council of Barcelona)

Carmen Vela (Director of Projects Collaborative Projects, INGENASA- Eurofins Tecnologies and former Secretary of State of R+D, President of the Honorary Committee)

Presentation of the T21RS Honorary Member Prof. Marie-Odile Rethoré

14:30 - 16:30 SCIENTIFIC SESSION 1

SESSION 1: Three of a perfect pair: stereotyped homeostatic compromises in trisomies

Chair: Pablo Helguera (INIMEC-CONICET, Argentina)

- 1. Metabolic dependencies of aneuploid cells. *Eduardo Torres* (University of Massachusetts-Worcester, USA)
- 2. Comparative analysis of autosomal human trisomies. *Jorge Busciglio* (University of California-Irvine, USA)
- 3. Aneuploidy-associated stress; another therapeutic target for pathophysiology in Down syndrome. *Yasuji Kitabatake (Osaka University, JP)*

Short talks:

PO105. Multi-Omics Data-Driven Systems Biology approach for the identification of molecular targets to rescue Down syndrome-associated cognitive. *Mohit Rastogi (University Of Genoa, Genova, IT).*

PO113. A Comprehensive Proteomics-Based Interaction Screen Links Dyrk1a to RNF169 and to the DNA Damage Response. *Chiara Di Vona (Center For Genomic Regulation, Barcelona, SP)*

Foyer: HYPATIA

16:30 – 18:00 Poster SESSION 1 and selected Nano-symposia with coffee

Poster Session 1: PO18 – PO61, PO145

17:00 – 17:30 Nano-symposium #8: PO13 – PO17

17:30 – 18:00 Nano-symposium #3: PO01 – PO06

17:30 – 18:00 Nano-symposium #6: PO07-PO12

Room: GAIVIIVIA

Room: ALPHA

Room: BETA

Barcelona 6-9 June 2019

Room: AUDITORIUM

18:00 – 19:00 JEROME LEJEUNE PLENARY LECTURE: Genetics of Human Chromosome 21. Stylianos Antonarakis (University of Geneva, CH)

Chair: Thierry de la Villejegu (Jerome Lejeune Foundation, FR)



19:00 – 19:10 Montserrat Trueta Award Mara Dierssen (President T21RS), Katy Trias (Fundació Catalana Síndrome Down, SP)

19:30 – 22:00 Social Programme – Opening ceremony. (Page 28)

Friday, June 7, 2019

08:00 - 18:00 Registration

Room: AUDITORIUM

08:30 – 09:30 THE EMBO KEYNOTE LECTURE: Human-specific genes, neural stem cell amplification, and neocortex expansion in development and human evolution, *Wieland Huttner (Max Planck Institute, GER)*

Chair: Anita Bhattacharyya (University of Wisconsin, USA)



09:30 - 11:30 SCIENTIFIC SESSION 2 (parallel)

Room: AGORA

SESSION 2A: New disease-relevant behavioral perspective in preclinical intellectual disability studies

<u>Chairs: Maria Martínez de Lagran (Centre for Genomic Regulation, SP) and Anna Vazquez (University Pompeu Fabra, SP)</u>

- 1. Social Learning in the Ts65Dn Mouse Model of Down Syndrome: New Evidence for Hippocampal Dysfunction, Nicholas Santiago (University of California-Santa Cruz, USA)
- 2. Sleep and EEG abnormalities in DS mouse model, effects of single gene dosage correction. *Charles Hoeffer (University of Colorado, USA)*
- 3. Using mouse genetics to deconstruct Down syndrome phenotypes. *Victor Tybulewicz (Francis Crick Institute, London, UK)*



Short talks:

PO102. Genetic and Epigenetic Analysis of Down Syndrome: New insights from human studies and mouse models. Eugene Yu (Roswell Park Comprehensive Cancer Center and University at Buffalo, State University of New York. USA)

PO163. Treatment with Granulocyte-Macrophage Colony-Stimulating factor improves cognition in aging mice and in a mouse model of Down syndrome. *Md Mahiuddin Ahmed (Linda Crnic Institute For Down Syndrome. Aurora, US)*

Room: AUDITORIUM

SESSION 2B: Biomarkers for Alzheimer's disease in Down syndrome

<u>Chairs: Bradley Christian (University of Wisconsin, USA) and María Carmona Iragui (Hospital de la Santa Creu i Sant Pau, SP)</u>

- 1. Cognitive correlates of amyloid PET in Down syndrome. Sigan Hartley (University of Wisconsin, USA)
- 2. Alzheimer's-related MRI markers of cerebrovascular disease and dementia in Down syndrome. Adam Brickman (Columbia University, USA)
- 3. Cerebrovascular neuropathology, aging and Alzheimer disease in Down syndrome. *Elizabeth Head* (University of Kentucky, USA)
- 4. Plasma Neurofilament Light and Alzheimer's Disease Biomarkers in Down syndrome: Results the Down Syndrome Biomarker Initiative (DSBI). *Michael S. Rafii (University of Southern California. USA)*

11:30 – 12:00 Coffee Break Foyer: HYPATIA

12:00 - 14:00 SCIENTIFIC SESSION 3 (parallel)

Room: AGORA

SESSION 3A: New directions in human trisomy 21 stem cell research

Chair: Anita Bhattacharyya (University of Wisconsin, USA)

- 1. (Epi)genetic approaches towards resolving trisomy 21 gene-dosage contributions. *Stefan Pinter* (University of Connecticut, USA)
- 2. Consequences of trisomy 21 on the epigenome of different iPSC-derived cell types of the brain. *Hiruy Meharena (Massachusetts Institute of Technology, USA)*
- 3. CRISPR-enabled functional genomics approaches in Down syndrome iPSCs. *Ernst Wolvetang* (University of Queensland, AUS)

Short talks:

PO87. A System to Unravel Effects of APP Dosage in Trisomy 21 Cerebral Organoids. *Melvys Valledor (Umass Med Scool, Worcester USA)*

PO98. Human iPSC-Derived Down Syndrome Astrocytes Display Genome-Wide Perturbations in Gene Expression, Alterations in Chromatin Accessibility, and an Altered Adhesion Profile. *Blandine Ponroy (Mcgill University, Montreal, CA)*

Room: AUDITORIUM

SESSION 3B: Alzheimer's disease in the context of the trisomy of Hsa21 – preclinical studies

<u>Chair: Frances Wiseman (University College London, UK), William Mobley (University of California-San Diego, USA)</u>

- 1. The role of Dyrk1A in tau pathogenesis in Down syndrome and Alzheimer's disease. Fei Liu (New York State Institute for Basic Research in Developmental Disabilities, USA)
- 2. Mechanisms of neurodegeneration in mouse models of Down syndrome. *Carmen Martínez-Cué* (*University of Cantabria, SP*)
- 3. The role of genes other than APP in Alzheimer's disease development. *Frances Wiseman (UCL-UK)*.

Short talks:

PO85. Maternal DNA-Immunization strategy targeting Amyloid-β related neuropathology and dementia in Down-syndrome. *Tomer Illouz (Bar-Ilan University, Ramat Gan, IL)*

PO86. Alzheimer-like pathology in trisomy 21 cerebral organoids reveals BACE2 as a gene-dose-sensitive AD-suppressor in human brain. Aoife Murray (Lee Kong Chian School Of Medicine, Ntu, SG)

14:00 - 15:00 Lunch break

Room: AUDITORIUM

15:00 - 16:00 T21RS general assembly

- 1. Welcome and opening of the General Assembly, Mara Dierssen, President T21RS
- 2. Presentation and Approval of Annual Report 2018 incl. Financial Report 2018, *Marie-Claude Potier, secretary and Alain Dekker, treasurer T21RS.*
- 3. Society announcements and discussion
- 4. Closing

Fover: HYPATIA

16:00 – 18:00 Poster SESSION 2 and selected Nano-symposia with coffee

Poster Session 2: PO75-PO117, PO146, PO167

17:00 – 17:30 Nano-symposium #9: PO71 – PO74

17:30 – 18:00 Nano-symposium #4: PO62 – PO66

16:00 – 17.00 Special symposium "Microbiota, nutrition and dietary supplements in Down syndrome'

Room GAMMA

Room ALPHA

KOOM BEIA

- 1. Diet and Intellectual disabilities. *Dr. Rafael de la Torre (Integrative Pharmacology and Systems Neurosciences. IMIM, Barcelona, SP)*
- 2. Citizen-Science maps the oral microbiome in Down Syndrome. *Dr. Toni Gabaldon (Comparative Genomics. CRG, Barcelona, SP)*
- 3. Plant polyphenols as natural drugs for the management of Down syndrome. *Dr. Rosa Anna Vacca* (Institute of Biomembranes and Bioenergetics, Italian National Council of Research, Bari, IT.)



4. Dietary Supplement Use in Children with Down syndrome. Dr. Amy Feldman Lewanda (Division of Genetics & Metabolism, Children's National Health System, Washington, DC, USA)



Symposium sponsored by:

18:00 - 20:00 SCIENTIFIC SESSION 4 (parallel)

Room: AUDITORIUM

SESSION 4A: Investigating Down syndrome phenotypes and mechanisms through comprehensive mouse modelling

<u>Chair: Elizabeth Fisher (University College London, UK), Veronique Brault (Institut de Génétique et de Biologie Moléculaire et Cellulaire, FR)</u>

- 1. Route 66, an exhilarating ride. *Yann Herault (Institut de Génétique et de Biologie Moléculaire et Cellulaire, FR)*
- 2. Understanding gene-phenotype relationship of skeletal abnormalities in Down syndrome using several mouse models. *Randall Roper (Indiana University-Purdue University Indianapolis, USA)*
- 3. A new, humanized mouse model of Down syndrome. *Roger Reeves (Johns Hopkins University School of Medicine, USA)*

Short talks:

PO33. Investigating DYRK1A gene dosage effect in glutamatergic and Gabaergic Neurons in a mouse model of Down syndrome. *Veronique Brault (Igbmc Cerbm Gie, Illkirch, FR)*

PO71. Altered Hippocampal-Prefrontal Neural Dynamics in the Dp1Tyb model of Down syndrome. *Phillip Muza (University College London, UK)*

Room: ALPHA

SESSION 4B: Cognitive outcomes in children with Down syndrome

<u>Chair: Anna Esbensen (Cincinnati Children's Hospital Medical Center, USA) and Lisa Jacola (St Jude Children's Research Hospital, USA)</u>

- 1. Neurocognitive outcomes for survivors of childhood leukemia with Down syndrome. *Lisa Jacola (St Jude Children's Research Hospital, USA)*
- 2. Cognitive profiles in children with Down syndrome. Silvia Lanfranchi (University of Padova, IT)
- 3. Relationship between executive functioning and behavior in children with Down syndrome. *Anna Esbensen (Cincinnati Children's Hospital Medical Center, USA)*

Short talks:

PO90. New-rocognitive? Assessing the reliability and utility of the new DSM-5 neurocognitive disorder criteria in a Down Syndrome population. *Sarah Pape (King's College London, UK)*

PO138. Mortality reduction in institutionalized children with intelectualintellectual disability and social vulnerability: results of 2 years improving basic health care. *Beatriz Shand (Pontificia Universidad Católica, Santiago, Chile)*

21:00 GALA DINNER. (Page 28)

Saturday, June 8, 2019

08:30 - 18:00 Registration

Room: AUDITORIUM

08:30 – 09:30 PLENARY LECTURE 3: Alzheimer's disease mechanisms and therapeutics. *Li-Huei Tsai* (Massachussets Institute of Technology, USA)

Chair: Victor Tybulewicz (Francis Crick Institute, London, UK)

09:30 - 11:30 SCIENTIFIC SESSION 5 (parallel)

SESSION 5A: Modifications of autophagy-lysosomal-endosomal pathways in Down syndrome and other neurodegenerative diseases

Chair: Marie Claude Potier (ICM, FR) and Eric Hamlett (Medical University of South Carolina, USA)

- 1. Dysfunction of the Autophagy-Lysosomal Pathway as a common mechanism of neurodegeneration. Steve Finkbeiner (University of California- San Francisco, USA)
- 2. Neuronal lysosomal dysfunction releases exosomes harboring APP C-terminal fragments and unique lipid signatures. *André Miranda (University of Minho, PRT)*
- 3. Excess synaptojanin 1 contributes to place cell dysfunction and memory deficits in the aging hippocampus in Down syndrome. *Catherine Marquer (Columbia University, USA)*

Short talks:

PO109. Targeted Proteomics of the Endosomal Compartment: Spatio-temporal localization in human fibroblasts. *Laura Xicota (ICM, Paris, FR)*

PO119. Mitochondrial fusion and function are elevated in T21 iPSCs. Beverly Rothermel (University of Texas Southwestrn Medical Center, Dallas, USA)

Room: AGORA

SESSION 5B: Language Outcome Measures for Treatment Studies

<u>Chair: Len Abbeduto (University of California-Davis, USA), Laura Del Hoyo (University of California-Davis, USA)</u>

1. Measuring spoken language to evaluate treatment efficacy for Down syndrome. *Len Abbeduto* (University of California-Davis, USA)



- 2. Early language and communication indicators of response to treatment. *Angela John Thurman* (University of California-Davis, USA)
- 3. Semantic verbal fluency as an early diagnostic tool of Alzheimer's disease in Down syndrome. Rafael de la Torre (IMIM-Hospital del Mar Medical Research Institute, SP)

Short talks:

PO134. Lexical Prediction Abilities in People with Down Syndrome: An Eye-Tracking task. *Natalia Arias*, (Unam, México, MX)

PO135. Language Skills as a Predictor of Cognitive Decline in Adults with Down Syndrome. *Margaret Pulsifer, (Massachusetts General Hospital/Harvard Medical School, Boston, USA)*

11:30 -12:00 Coffee break

Foyer: HYPATIA

Room: AUDITORIUM

12:00- 15:35 SCIENCE AND SOCIETY SYMPOSIUM - PROGRAMME FOR FAMILIES

This years' symposium primarily intends to have an interactive discussion between the speakers and the audience. To facilitate the interaction, English-Spanish and Spanish-English translations will be provided. This symposium is organized by the Science and Society Committee of the T21RS.

T21RS Committee for Science & Society: Peter Paul De Deyn (University Medical Center Groningen, The Netherlands) (Chairman), Alain Dekker (University Medical Center Groningen, The Netherlands), Juan Fortea (Fundació Catalana Síndrome de Down, Spain), Sebastian Videla (Fundació Catalana Síndrome de Down, Spain), Lotta Granholm (University of Denver, USA), Cindy Lemere (Brigham and Women's Hospital, Harvard Medical School, USA)

12:00 Welcome: *Peter De Deyn* (Chairman of the T21RS Committee for Science & Society)

12:15 Parent-reported needs for development of research on Down syndrome (10 minutes presentation + 20 minutes debate) Hampus Hillerstrom (LumindRDS, USA).

Results from the parents' surveys on knowledge and personal experiences towards Down syndrome studies. Conducted by LumindRDS in the USA and Fondation Lejeune in the European Union.

12:45 Clinical studies (10 minutes presentation + 20 minutes debate) Juan Fortea (Sant Pau Hospital, Spain).

General overview of clinical studies and what we can expect for the future. Future clinical trials on Alzheimer's disease will be discussed. The voices of families and associations are encouraged.

13.15 How Global is Supporting Science & Possibilities for Collaboration. Michelle Whitten (Global Down Syndrome, USA)

13:30 Lunch break and Meet the Scientist

During lunchtime, individuals with Down syndrome, their families and caregivers will have the opportunity to discuss and ask questions to junior researchers about their work and other aspects of research. For young investigators, it will be and opportunity to get to know individuals with Down syndrome and their families.

14:10 Participation in research: pros and cons. How to involve families in research? (30 minutes debate)

Peter De Deyn (University Medical Center Groningen, The Netherlands)

Discussion with the audience about the needs and targets for research on Down syndrome. What is the point of view of family members and caregivers about participating in research?

14:40 Ethics: autonomy of people with Down syndrome (10 minutes presentation + 20 minutes debate)

Jesús Florez (University of Cantabria, Fundación Iberoamericana Down21, Spain)
How can we promote autonomy in people with Down syndrome? What should -and should not-expect from this autonomy?

15:10 Human Rights Assembly. Fundació Catalana Síndrome de Down, Spain

15:25 Summary and closure. Peter De Deyn (University Medical Center Groningen, The Netherlands)

Room: AUDITORIUM

15:45 - 17:45 SCIENTIFIC SESSION 6

SESSION 6: The Role of Inflammation and NGF dysfunction in cognitive decline and Alzheimer's disease pathology in Down syndrome

Chair: Jorge Busciglio (University of California-Irvine, USA), Florencia Iulita (Université de Montreal, CN)

- 1. Inflammation and NGF deregulation across the life span in Down syndrome. *Florencia Iulita* (Université de Montreal, CN)
- 2. Molecular and cellular mechanisms of degeneration in Down syndrome neurons: a link between ABeta pathological changes, inflammation and NGF dysmetabolism. *Maria Lioudyno (University of California-Irvine, USA)*
- 3. Analysis of biomarkers of cognitive decline and neurodegeneration in plasma and CSF in a DS cohort. Juan Fortea (Fundació Catalana Síndrome de Down and Hospital San Pau, SP)

Short talks:

PO149. Disturbed Expression of Inflammation-Related Genes Caused by A Triplication of Erg Gene in The Prenatal Developing Brain with Down Syndrome. *Ryohei Shimizu (Kyoto Pharmaceutical University, Kyoto, JP)*

PO150. SNX27 Deficiency Limits Antigen Dependent Activation and Potentiates CXCR4 Dependent Migration in T Lymphocytes. Cristina Rodríguez (National Centre for Biotechnology, Madrid, SP)

Foyer: HYPATIA

17:45 – 19:00 Poster SESSION 3 and selected Nano-symposia with coffee break

Poster Session 3: PO127-PO166

18:00 – 18:30 Nano-symposia #2: PO121-PO123

18:00 – 18:30 Nano-symposia #7: PO124-PO126

18:30 - 19:00 Nano-symposia #1: PO118-PO120

Room: BETA

Room: GAMMA

Room: ALPHA



Room: AGORA

19:00 – 20:00 Meet the Experts Sessions: Li-Huei Tsai (Massachusetts Institute of Technology, USA) with aperitif

Chair: Renata Bartesaghi (University of Bologna, IT)

Room: AGORA

20:00 – 21:00 Education Committee session: "How should we study Down syndrome: pros and cons of mouse models and human iPSCs"

T21RS Committee for Education & Training: Renata Bartesaghi (University of Bologna, IT) (Chair), Carmen Martinez-Cué (University of Cantabria, SP), Tom Blumenthal (Linda Crnic Institute for Down Syndrome, USA), Tarik Haydar (Boston University, USA)

Sunday, June 9, 2019

08:00 - 18:00 Registration

Room: AUDITORIUM

08:30 – 09:30 PLENARY LECTURE 4: Alzheimer's disease in Down syndrome: update from the LonDowns consortium. *Andre Strydom (King's College London, UK)*

Chair: Juan Fortea (Sant Pau Hospital, Spain).

09:30 – 10:15 Annette Karmiloff-Smith and Michael Harpold Thesis Awards Ceremony - *Renata Bartesaghi* (University of Bologna, IT)

10:15 – 11:00 Coffee break Foyer: HYPATIA

11:00 – 13:00 SCIENTIFIC SESSION 7 (parallel)

Room: AGORA

SESSION 7A: Leukemia in Children with Down syndrome: why does it happen and what can we do about it?

Chair: John Crispino (Northwestern, USA), Sebastien Malinge (Telethon Kids Institute, AU)

- 1.Targeting constitutive Ras/Mapk pathway activation in Down syndrome associated B Cell leukemia. Sebastien Malinge (Telethon Kids Institute, AU)
- 2.Trisomy 21 silencing in hematopoietic and neural cells: new insights and prospects. *Jeanne Lawrence* (University of Massachusetts-Worcester, USA)
- 3. HMGN1 in phenotypes of trisomy 21 hematopoietic stem/progenitor cells and leukemia. *Andrew Lane* (Dana Farber Cancer Institute, USA)

Short talks:

PO106. Using Single-Cell Sequencing to Assess DNA Damage in Trisomy 21 Blood. *Rebecca Silberman* (Massachusetts Institute of Technology, Cambridge, USA)

PO127. Uncovering The Origin(s) Of Down Syndrome-Associated Leukemia. *Sara Martina Maffioletti* (Ospedale San Raffaele/San Raffaele Telethon Institute For Gene Therapy, IT)

Room: AUDITORIUM

SESSION 7B: Metabolic defects in Down syndrome: from periphery to the brain

Chair: Eugenio Barone (Sapienza University of Rome, IT) and Marzia Perluigi (Sapienza University of Rome, IT)

- 1. Plasma and urinary metabolomic profiles of children with Down syndrome. *Maria Chiara Pelleri* (*University of Bologna, IT*)
- 2. Trisomy 21 activates the kynurenine pathway via increased dosage of interferon receptors. *Kelly Sullivan* (University of Colorado, USA)
- 3. Alzheimer's disease biomarkers in neuron-derived extracellular vesicles and cognitive decline in Down syndrome. *Aurelie Ledreux (University of Denver, USA)*

Short talks:

PO47. Reduced Insulin Receptor Levels in Neuronal-Derived Exosomes Highlights Early Alterations of Brain Insulin Signaling In Down Syndrome. *Eugenio Barone (Sapienza University of Rome, IT)*

13:00 -14:00 Lunch

Room: AUDITORIUM

14:00-16:00 **SCIENTIFIC SESSION 8**

SESSION 8: Correction of circuit specific GABAergic over-inhibition and dendritic alterations in Down syndrome

<u>Chair: Floriana Costanzo (Child Neuropsychiatric Unit, Department of Neuroscience, Bambino Gesù Children's Hospital, IT) and Jean Maurice Delabar (Paris Sorbonne Université, CNRS, FR)</u>

- 1. GABA neurotransmission in a mouse model of Down syndrome, *Trevor G. Smart* (University College London, UK)
- 2. Cortical inhibitory circuits in Down syndrome, *Javier Zorrilla de San Martin* (ICM, INSERM, CNRS, ICM,FR)
- 3. Modulation of dendritic α 5-GABAA receptors rescues impaired NMDA receptor activation in a mouse model of Down syndrome, *Jan Michael Schulz (University of Basel, Switzerland)*

Short talks:

PO51. In vivo modeling of human neuron dynamics and Down syndrome. *Vincenzo De Paola* (*Imperial College London, UK*)

PO54. Altered development of interneurons in Down syndrome. *Anita Bhattacharyya (University of Wisconsin-Madison, USA)*

16:00-16:30 Coffee break

Foyer: HYPATIA



Room: AUDITORIUM

16:30-17:00 Melissa Parisi: updates on US NIH funding (ABC-Connect, INCLUDE)

Chair: Roger Reeves (Johns Hopkins University School of Medicine, USA)

Room: AUDITORIUM

17:00-19:00 SCIENTIFIC SESSION 9

SESSION 9: Endpoints for clinical trials

Chair: Diana Bianchi (NIH-NICHD, USA) and Alain Dekker ((University Medical Center Groningen, NL)

- 1. Clinical trials in children with Down syndrome, the experience of the Jerome Lejeune Institute. Clothilde Mircher (Institut Jerome Lejeune, FR)
- 2. Endpoints for clinical trials from the Horizon 21 Consortium. *Bessy Benejam (Fundació Catalana Síndrome de Down, SP)*

Short talks:

Down Syndrome Clinical Trials Network (LuMind)

PO10. Diagnosing Cognitive Decline Due to Alzheimer's Disease in Down Syndrome Patients with Neuropsychological Testing. Laura Videla (Alzheimer-down Unit (FCSD&HSP), Barcelona, SP)

PO95. CSF NPTX2 as A Synaptic Biomarker of Alzheimer's Disease in The Down Syndrome Population. *Meifang Xiao (Johns Hopkins University, JHMI, USA)*

19:00 CLOSING CEREMONY. Poster awards presentation with aperitif



PROUS INSTITUTE for Biomedical Research

20:00 END OF MEETING

INDUSTRY SESSION

Wednesday, June 5, 2019

ROOM: TAU

16:00-18:00 Top Challenges Facing the Pharmaceutical Industry in Down syndrome Drug Development

The Market: Is it large enough to justify drug development? (Pier-Vincenzo Piazza, AELIS Pharma)

Translationality: Are we using the most adequate tools in the evaluation of drug effects in the transfer from preclinical studies to clinical trials

Mouse-based genetic modeling of Down syndrome. (William C Mobley, USC San Diego)

Bordeaux Maze Test (Rafael de la Torre, IMIM)

Clinical Trials Outcomes: Difficulties in finding primary outcomes acceptable for regulatory authorities (FDA, EMA)

The NIH Toolbox Cognitive Battery for intellectual disabilities (Len Abbeduto, MIND Institute)

Regulatory Aspects: The EMA perspective in drug development of orphan drugs targeting neurodevelopmental disorders (Fernando Mendez, EMA Committee for Orphan Medicinal Products)

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GENERAL INFORMATION



Conference venue

The 3rd International Conference of the Trisomy 21 Research Society takes place at the CosmoCaixa in Barcelona.

CosmoCaixa Barcelona C/Isaac Newton, 26 08022 Barcelona Tel. +34 932 12 60 50

The meeting rooms where the sessions of the conference will be held are: Auditorium, Agora, Alpha, Beta, Gamma and are located at the -2 floor. The coffee breaks will be served at Foyer Hypatia (Ambaixades)



How to get there

Bus lines: V13, V15, V17, V19, H2, H4, 196

Train: FGC - Line 7 - Av. Tibidabo



Free WI-FI internet is provided by the venue.



Registration

Registrations have been managed by Bco Congresos. The registration desk is located at the -1 floor of the venue. General registration fee includes: access to the scientific sessions and exhibition area, conference documentation, coffee breaks, lunch boxes, welcome cocktail and gala dinner (for those who have confirmed it). It excludes fee for Satellite symposia and the Industry Session.



Registration desk opening hours:

Wednesday, 5 June: 15.30 - 19.00Thursday, 6 June: 08.30 - 18.00Friday, 7 June: 08.00 - 18.00Saturday, 8 June: 08.00 - 19.00Sunday, 9 June: 08.00 - 18.00



N Badges

At the conference venue, a name badge will be provided on-site with your registration documents. Participants are requested to wear their badge at all times inside the venue and at the Gala Dinner organised within the conference.





The complete Abstract book will be available online on the Conference website.



Certificate of attendance

A certificate of attendance will be sent by e-mail to all participants after the conference.



Disclaimer

All best endeavours will be made to present the programme as published. However, the 3rd International Conference of the Trisomy 21 Research Society and its agents reserve the right to alter or cancel, without prior notice, any arrangements, timetables, plans or other items relating directly or indirectly to the conference, for any cause beyond its reasonable control, the 3rd International Conference of the Trisomy 21 Research Society and its agents are not liable for any loss or inconvenience caused as a result of such cancellation.

Delegates are advised to take out their own travel insurance and to extend their policy to cover personal belongings as the conference does not cover individuals against cancellation of bookings or theft or damage to belongings.



്യൂപ്പ് Workspace during the conference

The TAU meeting room will be accessible to participants who would like a space to work or have a meeting. Please consult the registration desk for availability as there are already programmed meetings.



Public transportation passes

Multiple public transportation passes can be bought in all metro stations. Passes can be used for both the metro and bus network. The most popular pass is the ten-ride card, called T-10. With a T-10 card you may use several subway lines or buses if less than 1h15m have passed since you went through the turnstile. The current price for a single ride ticket is 2,20 € and 10,20 € for a T-10 card.

Barcelona travel cards (Hola BCN!): These cards allow unlimited journeys on Barcelona public transport over 2, 3, 4 or 5 consecutive days on a single ticket. They also include subway journeys between the airport and the city centre. More info www.tmb.cat/en



Metro network opening hours:

Monday to Thursday 05.00-24.00 Friday 05.00-02.00 Saturday 24 hours Sunday 05.00-24.00

SOCIAL PROGRAMME

Thursday, June 6, 2019

Room: AGORA

19.30 – 19:50 *I am able: Presentation of an art project by Quim and Jesús Vilamajó.* Meet the artists Quim, a young man with Down syndrome, and his father Jesús, who have been working for several years in a singular, collaborative, ambitious and transmedia project. Through art, they want to change the way society views people with disabilities. The exhibition will be on display during the whole meeting. For more information: #IamAbleProject, https://vimeo.com/205878515

Room: AUDITORIUM

20:00 – 21:00 *Cactus, sólo muere lo que se olvida:* Shakespeare's play Romeo and Juliette adapted by Compañía Nacional de Teatro Manantial de Ilusión (Chile). Inclusive theatre play featuring actors with Down syndrome and directed by Victor Ignacio Romero Rojas. Subtitles in English.

Fover: HYPATIA

21:00 - 22:00 Opening ceremony. Welcome cocktail

Friday, June 7, 2019

21:00 Gala Dinner. Restaurant Mirabé (Address: Carrer Manuel Arnús, 08035 Barcelona). Within walking distance of the meeting venue, Cosmocaixa. Enjoy food and music over extraordinary views of the city of Barcelona. Concert: From Lost to the River + Els cracks d'Andi.

*Not included on the following registration fees: One Day, Corporate/ Industry, Satellite simposium, Science and Society.

A bus service will be available for the attendees to take them from the restaurant to central points of Barcelona.

Sunday, June 9, 2019

19:00 Closing Ceremony

Free entrance at museums:

During the days of the conference the Ayuntament de Barcelona has offered free entrance to all the conference attendees of the 3rd International Conference of the Trisomy 21 Research Society to the following museums by showing the conference badge. This free entrance is personal and non-transferable.

List of museums: Muhba, Museu Marès, Museu del Disseny, Museu Etnològic i Cultures del Món, Born, Monestir de Pedralbes



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Wolvetang, Ernst	Session 3A	15
X		
Xiao, Meifang	PO95, Short talk Session 9	16-23
Xicota, Laura	PO109, Short Talk Session 5A	16-18
Xie, Peng	PO123	20
Y		
Ye, Bing	PO121	20
Yu, Eugene Z	PO102, Short Talk Session 2A	16 - 15
Zammit, Matthew	PO124	20
Zampieri, Bruna L	PO154	20
Ziogas, Ilias	PO162	20
Zorrilla De San Martin, Javier	Session 8	22

NOTES



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