



5th T2IRS International Conference

Promoting Research Excellence
in Down Syndrome

Rome, Italy
June 5 – 8 2024

**Preliminary
Scientific Program**



 **Wednesday, 5 June 2024**

Sapienza University of Rome

	Aula Magna - Rettorato	Aula Magna - Lettere
08:00 AM – 06:00 PM	Registration	
09:00 AM – 10:40 PM	Satellite Symposium 1 Preclinical committee	Satellite Symposium 2 Clinical committee
10:40 AM – 11:05 AM	Coffee break	
11:05 AM – 12:35 PM	Satellite Symposium 1 Preclinical committee	Satellite Symposium 2 Clinical committee
12:35 PM – 02:00 PM	Lunch break	
02:00 PM – 03:00 PM	T21RS General Assembly	
03:00 PM – 05:00 PM	Symposium 1 Beyond the gene-dosage hypothesis in trisomy 21	Symposium 2 From fetus to old age
05:00 PM – 05:30 PM	Coffee break	
05:30 PM – 07:00 PM	Opening ceremony	
07:00 PM	Welcome reception	



08:00 AM – 06:00 PM

Registration 

Wednesday, 5 June 2024

 **Aula Magna – Rettorato** – Sapienza University of Rome

09:00 AM – 10:40 AM

**Satellite 1 by Preclinical committee:
Model Strengths and Limitations**

Chairs: Frances Wiseman, UK; Randall Roper, USA

Introduction by the chairs

C. Elegans

Author: Jon Pierce, USA

D. Melanogaster

Author: Bing Ye, USA


Zebrafish

Author: Summer Thyme, USA

iPSCs & organoids.

Author: Aoife Murry, UK

10:40 AM – 11:05 AM

Coffee break 

11:05 AM – 11:55 AM

Model Strengths and Limitations

New Rodent models.


Authors: Eugene Yu, USA

Human post-mortem material.

Authors: Sylvia Pérez, USA

Panel discussion, all speakers

12:35 PM – 02:00 PM

Lunch break 

02:00 PM – 03:00 PM

T2IRS General Assembly

03:00 PM – 05:00 PM

Symposium 1: Beyond the gene-dosage hypothesis in trisomy 21

Chair: María del Mar Dierssen Sotos, Spain

The topological chromatin remodeling in trisomy 21

Author: Stylianos Antonarakis, Switzerland



Gene dosage in four dimensions: insights from the spatiotemporal deconvolution of copy number variations in human neurodevelopment

Author: Giuseppe Testa, Italy


Epigenetic mechanisms contributing to cognitive deficits in Down syndrome

Author: César Sierra, Switzerland

Contribution of retrotransposition to Down syndrome

Author: María Martínez de Lagrán, Spain

05:00 PM – 05:30 PM

Coffee break 

05:30 PM – 07:00 PM

Opening Ceremony

10 Years of promoting excellence in Down syndrome research. Welcome messages from T21RS President, Institutions and stakeholders

07:00 PM

Welcome Reception at Rettorato Terrace

 **Aula Magna – Lettere** – Sapienza University of Rome

09:00 AM – 10:40 AM

Satellite 2 by Clinical committee

Chairs: Brian Skotko, USA; Alberto Costa, USA

Introduction by the chairs

Managing healthy lifestyles in individuals with Down syndrome.

Author: Nicolas M. Oreskovic, USA


Down Syndrome Clinic to You: a new platform bringing healthcare information to families around the globe.

Author: Brian Skotko, USA

Clinical evaluation, treatment, and support for patients with Down syndrome and Alzheimer's disease.

Author: Stephanie Santoro, USA

10:40 AM – 11:05 AM

Coffee break 



11:05 AM – 12:05 PM

Satellite 2 by Clinical committee

Emerging pharmacological and immunological strategies to treat Alzheimer's disease in persons with DS.

Author: Michael S. Rafi, USA


The development and onset of neurodegeneration in people with Down syndrome—the spectrum.

Author: Shahid Zaman, UK

Potential challenges and barriers to implement recently approved pharmacological strategies to treat Alzheimer's disease in the general population to persons with DS

Author: Alberto Costa, USA

12:35 PM – 02:00 PM

Lunch break 

03:00 PM – 05:00 PM

Symposium 2: From fetus to old age: advanced neuroimaging biomarkers and their predictive power for cognitive outcomes across the lifespan in Down Syndrome

Chair: Abi Fukami – Gartner, UK

Exploring structural brain development in fetuses with Down syndrome using state-of-the-art in utero magnetic resonance imaging

Author: Abi Fukami – Gartner, UK

Frontal lobe structure and function in youth with Down syndrome: Lessons from magnetic resonance imaging, diffusion tensor imaging, and functional near-infrared spectroscopy

Author: Nancy Raitano Lee, USA


Advanced diffusion magnetic resonance imaging modelling: dissecting white matter changes in Down syndrome and Alzheimer's disease

Author: Fedal Saini, UK

White matter alterations in early Huntington's disease

Author: Herminia Diana Rosas, USA

05:00 PM – 05:30 PM

Coffee break 

07:00 PM

Welcome Reception at Rettorato Terrace

 **Thursday, 6 June 2024**

Roma Convention Center 'La Nuvola'

	Plenary Room	Room 1	Room 2
08:00 AM – 06:00 PM	Registration		
08:30 AM – 10:30 AM	Symposium 3 Alzheimer's disease in Down syndrome	Symposium 4 Heart Development and Disease in Down Syndrome	Symposium 5 The relationship between sleep, epi- lepsy and cognition in Down Syndrome
10:30 AM – 11:00 AM	Coffee break		
11:00 AM – 11:45 AM	The EMBO Keynote Lecture John Hardy		
11:45 AM – 01:45 PM	Lunch & Poster presentations		
01:45 PM – 03:15 PM	Industry Session	NANO-S1 Young Investigators	NANO-S2 Young Investigators
03:15 PM – 04:00 PM	Keynote 2 Anita Bhattacharyya		
04:00 PM – 04:30 PM	Coffee break		
04:30 PM – 06:30 PM	Symposium 6 Is gene therapy a perspective in Down syndrome?	Symposium 7 Metabolic health in persons with Down syndrome	Symposium 8 Hematopoietic Dysregulation in People with Down syndrome



08:00 AM – 06:00 PM

Registration 

Thursday, 6 June 2024

 Plenary room – Roma Convention Center ‘La Nuvola’

08:30 AM – 10:30 AM

**Symposium 3: Alzheimer’s disease in Down syndrome:
Updates on Natural History, Biomarkers and Clinical Trials**

Chairs: Sharon J. Krinsky-Mchale, USA; Michael Rafii, USA


Recent Advances in Understanding AD Biomarkers in DS
Authors: Juan Fortea, Spain; Beau Ances, USA

Clinical Trials for AD in DS
Authors: Sarah Walter; Michael Rafii, USA

Characterizing cognitive decline in Alzheimer’s disease in
adults with Down syndrome Results from the ABC-DS and DABNI
cohort study
Authors: Sharon J. Krinsky-Mchale, USA; Laura Videla Toro, Spain

Imaging Biomarkers of Alzheimer’s Disease and their
Relation to Cognitive
Authors: Sigan L. Hartley, USA; Patrick Lao, USA

10:30 AM – 11:00 AM

Coffee break 

11:00 AM – 11:45 AM

The EMBO Keynote Lecture

John Hardy, UK



11:45 AM – 01:45 PM

Lunch break & Poster presentations 

01:45 PM – 03:15 PM


Industry Session

03:15 PM – 04:00 PM

Keynote 2

Anita Bhattacharyya, USA

04:00 PM – 04:30 PM

Coffee break 

04:30 PM – 06:30 PM

Symposium 6: Is gene therapy a perspective in Down syndrome?

Chair: Prof. Antonella Izzo, Italy



In vivo gene therapy and genome editing with AAV vectors

Author: Alberto Auricchio, Italy

Normalizing the expression of Has21 genes to elucidate their role in Down syndrome pathogenesis

Author: Antonella Izzo, Italy

Development of gene therapy for intellectual disabilities in Down syndrome using new genome editing technologies

Author: Yasuji Kitabatake, Japan

A dynamic in vitro model of Down Syndrome neurogenesis with Trisomy 21 gene dosage correction

Author: Stefan F. Pinter, USA

 **Room 1 – Roma Convention Center ‘La Nuvola’**

08:30 AM – 10:30 AM

Symposium 4: Heart Development and Disease in Down Syndrome

Chair: Ivan Moskowitz, USA

Modelling cardiomyocyte development using Down syndrome human induced pluripotent stem cells

Author: Andreia Sofia Bernardo, UK

Mechanisms of altered cardiac differentiation control in Trisomy 21 human induced pluripotent stem cells

Author: Ivan Moskowitz, USA


Cardiac Progenitor Development in Organoid Models

Author: Chulan Kwon, USA


Congenital heart defects in Down syndrome mice are caused in part by increased dosage of DYRK1A

Author: Victor Tybulewicz, UK

10:30 AM – 11:00 AM

Coffee break 

11:45 AM – 01:45 PM


Lunch break & Poster presentations 

01:45 PM – 03:15 PM

NANO-S1: Young Investigators



04:00 PM – 04:30 PM

Coffee break 

04:30 PM – 06:30 PM

Symposium 7: Metabolic health in persons with Down syndrome: causes, consequences, and treatment

Chair: Lauren Ptomey, USA

Biochemical markers of metabolic health in persons with Down syndrome – data from Go-DS21

Author: Leda Bianchi, UK

The role of physical activity and systemic inflammation in Alzheimer's disease in Down syndrome

Author: Victoria Fleming, USA

Weight loss interventions for adolescents and adults with Down syndrome

Author: Lauren Ptomey, USA

Remote exercise for the promotion of physical activity and cardiovascular health in adults with Down syndrome

Author: Amy Bodde, USA

 **Room 2 – Roma Convention Center 'La Nuvola'**

08:30 AM – 10:30 AM

Symposium 5: The relationship between sleep, epilepsy and cognition in Down Syndrome

Chairs: María Carmona-Iragui, Spain; Sandra Giménez, Spain; Renata Bartesaghi, Italy

Sleep disturbances and behavioral profiles in children and adolescents with Down syndrome

Author: Stefano Vicari, Italy

Impact of obstructive sleep apnea on sleep architecture, epilepsy, and Alzheimer's Disease biomarkers

Author: Sandra Giménez, Spain

Diagnosis and treatment of late-onset myoclonic epilepsy in Down syndrome (LOMEDS)


Authors: María Carmona-Iragui, Spain; Fedele Dono, Italy

Mechanisms and outcome of sleep disruption in Down Syndrome: insight from mouse models


Authors: Charles Hoeffler; Chiara Berteotti, USA




10:30 AM - 11:00 AM

Coffee break 

11:45 AM - 01:45 PM

Lunch break & Poster presentations **01:45 PM - 03:15 PM****NANO-S2: Young Investigators**

04:00 PM - 04:30 PM

Coffee break **04:30 PM - 06:30 PM****Symposium 8: Hematopoietic Dysregulation in People with Down syndrome***Chair: Edward Evans Jr., USA*

Too much and too little: how does an extra chromosome 21 initiate leukemia and hematopoietic dysfunction before birth in children with Down syndrome?

Author: Irene Roberts, UK

Let's have a 'Tête-à-Tête': Clonal Hematopoiesis in People with Down Syndrome

Author: Edward Evans Jr., USA

From Down syndrome to a potential treatment of leukemia and prostate cancer

Author: Shai Izraeli, Israel

Germline susceptibility factors associated with risk of leukemia in Down syndrome

Author: Karen Rabin, USA

 **Friday, 7 June 2024**

Roma Convention Center 'La Nuvola'

	Plenary Room	Room 1	Room 2	Families Room
08:00 AM – 06:00 PM	Registration			
08:30 AM – 10:30 AM	Symposium 9: Rodent Models of Down Syndrome	Symposium 10 Cognitive Outcome Measures in Down syndrome Across the Lifespan	Symposium 11 Understanding Alzheimer's disease in Down syndrome using omics appro- ches on human biosamples	Parallel Program for Families
10:30 AM – 11:00 AM	Coffee break			
11:00 AM – 11:45 AM	Keynote 3 Kelly Sullivan			Parallel Program for Families
11:45 AM – 01:45 PM	Lunch & Poster presentations			
01:45 PM – 03:15 PM	NANO-S3 Young Investigators	NANO-S4 Young Investigators	Workshop 1 Developing data ecosystems for collaborative Down syndrome research	Parallel Program for Families
03:15 PM – 04:00 PM	Keynote 4 Award Keynote Lecture Montserrat Trueta			
04:00 PM – 04:30 PM	Coffee break			
04:30 PM – 06:30 PM	Symposium 12 The role of DYRK1A in Down syndrome and related comorbidities: what is new	Symposium 13 Clinical and Neurodiagnostic Biomarkers in Down Syndrome Regression Disord	Symposium 14 Liver Dysfunction in Down Syndrome	Parallel Program for Families



08:00 AM – 06:00 PM

Registration 

 Plenary room – Roma Convention Center ‘La Nuvola’

08:30 AM – 10:30 AM

**Symposium 9: Rodent Models of Down Syndrome:
Present and Future Considerations**

Chair: Randall Roper, USA

Sex differences in mouse models recapitulate those seen in humans with DS

Author: Randall Roper, USA

New insights into rodent models of DS – what exactly is the prenatal phenotype?

Author: Tarik Haydar, USA

Modeling Down syndrome using human mouse chimeras

Author: Hiruy Meharena, USA

Environmental issues with mouse models

Author: Mara Dierssen, Spain


The future of Down syndrome mouse models—where are we going?

Author: Yann Herval, France

Panel Discussion—all speakers and chairs will participate in the panel

Author: Frances Wiseman, UK

10:30 AM – 11:00 AM


Coffee break 

11:00 AM – 11:45 AM

Keynote 3

Kelly Sullivan, USA

11:45 AM – 01:45 PM

Lunch break & Poster presentations 

01:45 PM – 03:15 PM

NANO-S3: Young Investigators

03:15 PM – 04:00 PM

Keynote 4

Award Keynote Lecture

Montserrat Trueta, Spain

Friday, 7 June 2024



04:30 PM – 06:30 PM**Symposium 12: The role of DYRK1A in Down syndrome and related comorbidities: what is new?***Chairs: Daniella Balduino Victorino, France; Laurent Meijer, UK*

Elucidating the dose-dependent effects of DYRK1A on Alzheimer's disease pathology in mice

Author: Daniella Balduino Victorino, France

Functional MRI in DS models: impact of DYRK1A inhibitors

Authors: Laura-Adela Harsan; Julien Caillete, France

DYRK1A/EGR1 pathway in vascular endothelial cells predispose to developing pulmonary arterial hypertension in Down syndrome

Author: Hidehiro Suginobe, Japan

Role of DYRK1A in trisomy 21 megakaryopoiesis

Author: Stella Chou, USA

DYRK1A & congenital heart defects in DS

Author: Laurent Meijer, UK

DYRK1A inhibitors restore pancreatic functions in animal model of diabetes

Author: Jamileh Movassat, France **Room 1 – Roma Convention Center 'La Nuvola'****08:30 AM – 10:30 AM****Symposium 10: Cognitive Outcome Measures in Down syndrome Across the Lifespan***Chair: Laura del Hoyo Soriano, Spain*

Understanding normative change in executive functioning

Author: Anna Esbensen, USA

Language Measures to Assess Treatment Efficacy

Author: Leonard Abbeduto, USA


Memory Measures in Down syndrome

Author: Hailey Van Vorce, USA


Cognitive measures in prodromal Alzheimer disease and Alzheimer disease Dementia in Down syndrome

Author: Laura del Hoyo Soriano, Spain


10:30 AM - 11:00 AM

Coffee break 

11:45 AM - 01:45 PM

Lunch break & Poster presentations **01:45 PM - 03:15 PM****NANO-S4: Young Investigators**

04:00 PM - 04:30 PM

Coffee break **04:30 PM - 06:30 PM****Symposium 13: Clinical and Neurodiagnostic Biomarkers in Down Syndrome Regression Disorder: Diagnostic and Therapeutic Implications***Chair: Jonathan Santoro, USA*

Catatonia and Psychiatric Phenotypes in DSRD

Author: Cathy Franklin, Australia

Neurocognitive Disorder Differentiation in DSRD

Author: Diego Real de Asua, Spain

Diagnostic Work Up in DSRD: When and How Quickly to Perform

Author: Stephanie Santoro, USA

Therapeutics in DSRD: The Choice Between Immunotherapeutics and Psychotropics

Author: Jonathan Santoro, USA **Room 2 - Roma Convention Center 'La Nuvola'****08:30 AM - 10:30 AM****Symposium 11: Understanding Alzheimer's disease in Down syndrome using omics approaches on human biosamples***Chair: Cliona Farrell, UK; Lisi Flores-Aguilar, USA*

AD-Related Lipid and Cellular Energy Dysmetabolism Occurs Early in People with Down Syndrome

Author: Mark Mapstone, USA

Decoding genetic and sporadic forms of Alzheimer's Disease: A Multi-Layered Transcriptomic Odyssey

Author: Vivek Swarup, USA


Biofluid markers of Alzheimer's disease-associated CNS inflammation in adults with Down syndrome

Author: Olivia Belbin, Spain


Proteomic analysis reveals increased Apolipoprotein E abundance in the Alzheimer's disease-Down syndrome brain compared to early-onset AD

Author: Cliona Farrell, UK

10:30 AM - 11:00 AM

Coffee break 

11:45 AM - 01:45 PM

Lunch break & Poster presentations 

01:45 PM - 03:15 PM

Workshop 1: Developing data ecosystems for collaborative Down syndrome research in the age of artificial intelligence

Moderator: Dr. Angelo Carfi, Italy

NIH perspective on international cohort development and data sharing

Author: Dr. Huiqing Li, USA

Assembly and analysis of multidimensional datasets in the INCLUDE Data Hub

Author: Dr. Joaquín Espinosa, USA

Multi-omics analysis of delayed development and early ageing in Down syndrome

Author: Dr. Micah Donovan, USA


Standard criteria for database development and application of machine learning-based approaches: an Italian pilot study

Author: Dr. Giuseppe Ramacieri, Italy

AI and medicine: the case study of the co-occurring conditions of Down syndrome

Author: Dr. Pietro Lió, UK

04:00 PM - 04:30 PM

Coffee break 

04:30 PM - 06:30 PM

Symposium 14: Liver Dysfunction in Down Syndrome

Chair: Lauren Dunn, USA

Altered bile acid metabolism in Down syndrome

Author: Lauren Dunn, USA




Metabolic Dysregulation in Down Syndrome Mouse Models
Author: G. William Wong, USA

Novel insights from fetal and placental phenotyping in
3 mouse models of Down syndrome
Author: April Adams, USA


Nonalcoholic Fatty Liver Disease in Children with Down Syndrome
Author: Diletta Valentini, Italy

 **Families Room – Roma Convention Center ‘La Nuvola’**


08:30 AM – 10:30 AM **Parallel Program for Families**

10:30 AM – 11:00 AM Coffee break 

11:00 AM – 11:45 AM **Parallel Program for Families**

11:45 AM – 01:45 PM Lunch break & Poster presentations 

01:45 PM – 03:15 PM **Parallel Program for Families**

04:00 PM – 04:30 PM Coffee break 

04:30 PM – 06:30 PM **Parallel Program for Families**



 **Saturday, 8 June 2024**

Roma Convention Center 'La Nuvola'

	Plenary Room	Room 1	Room 2	Families Room
08:00 AM – 06:00 PM	Registration			
08:30 AM – 10:30 AM	Symposium 15 Immunomodulatory therapies in Down syndrome	Symposium 16 Understanding progeroid biology of Down syndrome throughout lifetime	Symposium 17 Interplay between Alzheimer's disease and cerebrovascular pathology in individuals with Down syndrome	Parallel Program for Families
10:30 AM – 11:00 AM	Coffee break			
11:00 AM – 12:30 PM	NANO-S6 Young Investigators	NANO-S7 Young Investigators	Workshop 2 NIH and EU-funded projects	Parallel Program for Families
12:30 PM – 01:30 PM	Lunch & Poster presentations			
01:30 PM – 02:15 PM	Keynote 5 Miia Kivipelto			
02:15 PM – 03:30 PM	Science & Society meeting			
03:30 PM – 04:00 PM	Coffee break			
04:00 PM – 05:15 PM	Science & Society meeting			
05:15 PM – 06:15 PM	Closing ceremony			
07:30 PM	Gala dinner & awards at Palazzo Brancaccio			



08:00 AM – 06:00 PM

Registration 

 **Plenary room** – Roma Convention Center ‘La Nuvola’

08:30 AM – 10:30 AM

Symposium 15: Immunomodulatory therapies in Down syndrome

Chair: Angela Rachubinski, USA

The safety and efficacy of a JAK inhibitor for diverse clinical endpoints in Down syndrome

Author: Angela Rachubinski, USA

Multi-Omics approaches to dissect mechanisms of Down syndrome immune dysfunctions and vaccine responses

Author: Susan Pereira Ribeiro, USA


Immunomodulatory therapies in children with Down syndrome: a rheumatologist’s perspective

Author: Jessica Bloom, USA

In utero immune dysregulation in Down syndrome: implications for prenatal treatments

Author: Joaquín Espinosa, USA


10:30 AM – 11:00 AM

Coffee break 

11:00 AM – 12:30 PM

NANO-S6: Young Investigators

12:30 PM – 01:30 PM

Lunch break & Poster presentations 

01:30 PM – 02:15 PM


Keynote 5

Miia Kivipelto

02:15 PM – 03:30 PM

Science & Society meeting

03:30 PM – 04:00 PM

Coffee break 

04:00 PM – 05:15 PM

Science & Society meeting

05:15 PM – 06:15 PM

Closing ceremony

07:30 PM

Gala dinner & awards at Palazzo Brancaccio

Saturday, 8 June 2024



Room 1 – Roma Convention Center ‘La Nuvola’

08:30 AM – 10:30 AM

Symposium 16: Understanding progeroid biology of Down syndrome throughout lifetime

Chairs: Maria Giulia Bacalini, Italy; Dean Nizetic, UK

Down syndrome in the Geroscience perspective: a focus on trajectories of epigenetic aging

Author: Maria Giulia Bacalini, UK

Dissecting immune aging in people with DS

Author: Bernard Khor, USA


Multi-system biological age of individuals with DS is increased from earliest childhood and dose imbalance of DYRK1A provides one of possible causative mechanisms

Author: Dean Nizetic, UK

The DREAM complex regulates the DNA repair capacity of somatic tissues

Author: Arturo Bujarrabal-Dueso, Germany


10:30 AM – 11:00 AM

Coffee break 


11:00 AM – 12:30 PM

NANO-S7: Young Investigators

12:30 PM – 01:30 PM

Lunch break & Poster presentations 

03:30 PM – 04:00 PM

Coffee break 

07:30 PM

Gala dinner & awards at Palazzo Brancaccio

Room 2 – Roma Convention Center ‘La Nuvola’

08:30 AM – 10:30 AM

Symposium 17: Interplay between Alzheimer’s disease and cerebrovascular pathology in individuals with Down syndrome

Chair: Lisi Flores Aguilar, USA

Blood-brain barrier pathophysiology in individuals with Down syndrome

Author: Lisi Flores Aguilar, USA



The role of endothelial cells in maintaining the blood-brain barrier in Down syndrome and APPdup

Author: Louis Valay, France


Cerebrovascular dysfunction and Alzheimer's disease pathogenesis in adults with Down syndrome

Author: Adam M. Brickman, USA

Fluid biomarkers for vascular cognitive impairment

Author: Donna M. Wilcock, USA


10:30 AM - 11:00 AM

Coffee break 


11:00 AM - 12:30 PM

Workshop 2: NIH and EU-funded projects

12:30 PM - 01:30 PM

Lunch break & Poster presentations 

03:30 PM - 04:00 PM

Coffee break 

07:30 PM


Gala dinner & awards at Palazzo Brancaccio

 **Families Room** - Roma Convention Center 'La Nuvola'

08:30 AM - 10:30 AM

Parallel Program for Families

10:30 AM - 11:00 AM

Coffee break 

11:00 AM - 12:30 AM

Parallel Program for Families

12:30 PM - 01:30 PM

Lunch break & Poster presentations 

03:30 PM - 04:00 PM

Coffee break 



Organized by



Technical Secretariat

