



# 5<sup>th</sup> T2IRS International Conference

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Promoting Research Excellence  
in Down Syndrome

**Rome, Italy**  
**June 5 – 8 2024**

**Preliminary  
Scientific Program**



# 5th T21RS International Conference

Promoting Research Excellence in Down syndrome

Rome, ITALY | June 5-8 2024

Under the Patronage of

Supporters & Partners

ROMA



REGIONE LAZIO



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 **Wednesday, 5 June 2024**

**Sapienza University of Rome**

	<b>Aula Magna - Rettorato</b>	<b>Aula Magna - Lettere</b>
08:00 AM – 06:00 PM	Registration	
09:00 AM – 10:40 AM	Satellite Symposium 1 Preclinical committee <i>(Open session)</i>	Satellite Symposium 2 Clinical committee <i>(Open session)</i>
10:40 AM – 11:05 AM	Coffee break	
11:05 AM – 12:35 PM	Satellite Symposium 1 Preclinical committee <i>(Open session)</i>	Satellite Symposium 2 Clinical committee <i>(Open session)</i>
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** *(Open session)*

*Chairs: Bruna Zampieri, Brazil, Randall Roper, USA*

09:00 AM – 09:05 AM

**Introduction by the chairs**

09:05 AM – 09:30 AM

***C. Elegans***

*Author: Jon Pierce, USA*

09:30 AM – 09:55 AM

***D. Melanogaster***

*Author: Bing Ye, USA*

09:55 AM – 10:20 AM

***Zebrafish***


*Author: Summer Thyme, USA*

10:20 AM – 10:40 AM

***iPSCs & organoids.***

*Author: Aoife Murray, UK*

10:40 AM – 11:05 AM

Coffee break 

11:05 AM – 11:55 AM

**Satellite 1 by Preclinical committee:**

**Model strengths and limitations** *(Open session)*

11:05 AM – 11:30 AM

**New Rodent models**

*Authors: Eugene Yu, USA*

11:30 AM – 11:55 AM


**Human post-mortem material**

*Authors: Sylvia Pérez, USA*

11:55 AM – 12:35 PM

**Panel discussion, all speakers**

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**

*Chair: María del Mar Dierssen Sotos, Spain*

03:00 PM – 03:30 PM

**The topological chromatin remodeling in trisomy 21**

*Author: Stylianos Antonarakis, Switzerland*

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- 03:30 PM – 04:00 PM **Gene dosage in four dimensions: insights from the spatiotemporal deconvolution of copy number variations in human neurodevelopment**  
*Author: Giuseppe Testa, Italy*
- 04:00 PM – 04:30 PM **HDAC inhibitor treatment restores transcriptome, chromatin accessibility, and memory deficits in a trisomic mouse model**  
*Author: César Sierra, Switzerland*
- 04:30 PM – 05:00 PM **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** *(Open session)*  
*Chairs: Brian Skotko, USA; Alberto Costa, USA*
- 09:00 AM – 09:05 AM **Introduction by the chairs**
- 09:05 AM – 09:35 AM **Managing healthy lifestyles in individuals with Down syndrome**  
*Author: Nicolas M. Oreskovic, USA*
- 09:35 AM – 10:05 AM **Down syndrome Clinic to You: a new platform bringing healthcare information to families around the globe**  
*Author: Brian Skotko, USA*
- 10:05 AM – 11:40 AM **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 – 12:35 PM****Satellite 2 by Clinical committee** (Open session)

11:05 AM– 11:35 AM

**Understanding cognitive decline in the transition to Alzheimer's Disease in Down syndrome***Author: Sigan L. Hartley, USA*


11:35 AM– 12:05 PM

**The development and onset of neurodegeneration in people with Down syndrome–the spectrum***Author: Shahid Zaman, UK*

12:05 PM– 12:35 PM

**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*

03:00 PM – 03:30 PM

**Exploring structural brain development in fetuses with Down syndrome using state-of-the-art in utero magnetic resonance imaging***Author: Abi Fukami – Gartner, UK*

03:30 PM – 04:00 PM

**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*


04:00 PM – 04:30 PM

**Advanced diffusion magnetic resonance imaging modelling: dissecting white matter changes in Down syndrome and Alzheimer's disease***Author: Fedal Saini, UK*

04:30 PM – 05:00 PM

**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 Recent advances in understanding Alzheimer's disease in adults with Down syndrome	Symposium 4 Heart development and disease in Down syndrome	Symposium 5 The relationship between sleep, epilepsy 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 Pathogenesis, omics, inflammation, biomarkers and SARS-Cov-2 in Down syndrome	NANO-S2 Molecular and cellular mechanisms of Down syndrome
03:15 PM – 04:00 PM	Keynote Lecture 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: Recent advances in understanding Alzheimer’s disease in adults with Down syndrome: Identifying clinical progression and biomarker accumulation**

*Chairs: Sharon J. Krinsky-Mchale, USA*

08:30 AM – 09:00 AM

**Longitudinal evaluation of promising outcome measures of mild cognitive impairment and the transition to clinical dementia in adults with Down syndrome: Results from the ABC–DS study**

*Authors: Sharon J. Krinsky-Mchale, USA*

09:00 AM – 09:30 AM

**Longitudinal trajectories of cognitive decline related to imaging biomarkers of Alzheimer’s disease in Down syndrome**

*Authors: Sigan L. Hartley, USA*

09:30 AM – 10:00 AM

**Characterizing Alzheimer’s disease cognitive decline in Down syndrome: A comprehensive analysis from the DABNI Cohort**


*Authors: Laura Videla Toro, Spain*

10:00 AM – 10:30 AM

**Longitudinal rates of accumulation for MRI–based cerebrovascular disease biomarkers**

*Authors: Patrick Lao, USA*

10:30 AM – 11:00 AM

Coffee break 


11:00 AM – 11:45 AM

**The EMBO Keynote Lecture  
Downs and Alzheimer’s  
Disease: Time for Treatment**

*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 Lecture 2  
Modeling brain development in Down syndrome**

*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*

04:30 PM – 05:00 PM

**In vivo gene therapy and genome editing with AAV vectors***Author: Alberto Auricchio, Italy*

05:00 PM – 05:30 PM

**Normalizing the expression of Has21 genes to elucidate their role in Down syndrome pathogenesis***Author: Antonella Izzo, Italy*

05:30 PM – 06:00 PM

**Development of gene therapy for intellectual disabilities in Down syndrome using new genome editing technologies***Author: Yasuji Kitabatake, Japan*

06:00 PM – 06:30 PM

**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*

08:30 AM – 09:00 AM

**Congenital heart defects in Down syndrome mice are caused in part by increased dosage of DYRK1A***Author: Victor Tybulewicz, UK*

09:00 AM – 09:30 AM

**Modelling cardiomyocyte development using Down syndrome human induced pluripotent stem cells***Author: Andreia Sofia Bernardo, UK*


09:30 AM – 10:00 AM

**Genetics, development and congenital heart disease in Down syndrome***Author: Roger H. Reeves, USA*


10:00 AM – 10:30 AM

**Birth defect causation in Down syndrome: Diminished response to Hedgehog signaling in Trisomy 21 cardiac progenitors***Author: Ivan Moskowitz, 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-S1: Pathogenesis, omics, inflammation, biomarkers and SARS-Cov-2 in Down syndrome**  
*Chair: Elliott Mufson, USA*
- 01:45 PM – 02:00 PM **Developmental and aging pathogenesis in Down syndrome**  
*Author: Sylvia Perez, USA*
- 02:00 PM – 02:15 PM **Neuropathological sequelae in glial and neuronal elements after a COVID-19 infection**  
*Author: Ann-Charlotte Granholm, USA*
- 02:15 PM – 02:30 PM **Deciphering ACE2 and TMPRSS2 changes in Down syndrome and Alzheimer's dementia: concerns regarding COVID-19**  
*Author: Javier Sáez-Valero, Spain*
- 02:30 PM – 02:45 PM **Low TGF- $\beta$ 1 plasma levels are associated with cognitive decline in Down syndrome**  
*Author: Filippo Caraci, Italy*
- 02:45 PM – 02:55 PM **Transient site-specific copy gains of the Chr21 IFNR locus are suppressed by Trisomy 21**  
*Speaker: Jason Wilson, USA*
- 02:55 PM – 03:05 PM **Variegated overexpression of chromosome 21 genes reveals molecular and immune subtypes of Down syndrome**  
*Author: Micah Donovan, USA*
- 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*
- 04:30 PM – 05:00 PM **Biochemical markers of metabolic health in persons with Down syndrome – data from Go-DS21**  
*Author: Leda Bianchi, UK*
- 05:00 PM – 05:30 PM **The role of physical activity and systemic inflammation in Alzheimer's disease in Down syndrome**  
*Author: Victoria Fleming, USA*
- 05:30 PM – 06:00 PM **Weight loss interventions for adolescents and adults with Down syndrome**  
*Author: Lauren Ptomey, USA*



06:00 PM – 06:30 PM

**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*

08:30 AM – 09:00 AM

**Sleep disturbances and behavioral profiles in children and adolescents with Down syndrome***Author: Stefano Vicari, Italy*

09:00 AM – 09:30 AM

**Impact of obstructive sleep apnea on sleep architecture, epilepsy, and Alzheimer’s Disease biomarkers***Author: Sandra Giménez, Spain*

09:30 AM – 10:00 AM

**Diagnosis and treatment of late-onset myoclonic epilepsy in Down syndrome (LOMEDS)***Authors: María Carmona-Iragui, Spain; Fedele Dono, Italy*

10:00 AM – 10:30 AM

**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 &amp; Poster presentations 🍴

01:45 PM – 03:15 PM

**NANO-S2: Molecular and cellular mechanisms of Down syndrome**


01:45 PM – 02:00 PM

**Single-cell multi-omics analysis reveals the unique features of mosaic Down’s syndrome***Author: Weihong Song, USA, China*

02:00 PM – 02:15 PM

**Amyloid- $\beta$  mediated inhibition of  $\gamma$ -secretase activity impairs endolysosomal and autophagy functions: implications in Alzheimer’s disease and Down syndrome pathogenesis***Author: Utpal Das, USA*

- 02:15 PM - 02:25 PM **Dysregulation of astrocyte–secreted pleiotrophin contributes to neuronal structural and functional deficits in Down syndrome**  
*Author: Ashley Brandebura, USA*
- 02:25 PM - 02:35 PM **Neural precursors cells derived from trisomy 21 iPSCs show a deficiency in mitophagy**  
*Author: Nunzia Mollo, Italy*
- 02:35 PM - 02:45 PM **Stabilizing effects of myelin repair drugs on dysbalanced oligodendroglial differentiation upon C21orf91 overexpression**  
*Author: Laura Reiche, Germany*
- 02:45 PM - 02:55 PM **Deciphering the interplay between the unfolded protein response and insulin resistance in Down syndrome neuropathology**  
*Author: Francesca Prestia, Italy*
- 02:55 PM **T21RS Thesis award**

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*

04:30 PM - 05:00 PM **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*

05:00 PM - 05:30 PM **Let's have a 'Tête-à-Tête': clonal hematopoiesis in people with Down syndrome**

*Author: Edward Evans Jr., USA*

05:30 PM - 06:00 PM **From Down syndrome to a potential treatment of leukemia and prostate cancer**

*Author: Shai Izraeli, Israel*

06:00 PM - 06:30 PM **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 approaches on human biosamples	Parallel Program for Families
10:30 AM – 11:00 AM	Coffee break			
11:00 AM – 11:45 AM	Keynote Lecture 3 Kelly Sullivan			Parallel Program for Families
11:45 AM – 01:45 PM	Lunch & Poster presentations			
01:45 PM – 03:15 PM	NANO-S3 Understanding DS-AD disease mechanisms using rodent models	NANO-S4 Genomic and epigenetic mechanisms, therapeutic approaches and diagnosis	Workshop 1 Developing data ecosystems for collaborative Down syndrome research	Parallel Program for Families
03:15 PM – 04:00 PM	Keynote Lecture 4 Montserrat Trueta Award			
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 disorder	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**

*Chairs: Randall Roper, USA; Antonella Tramutola, Italy*

08:30 AM – 08:50 AM

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

*Author: Randall Roper, USA*

08:50 AM – 09:10 AM

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

*Author: Tarik Haydar, USA*

09:10 AM – 09:30 AM

**Environmental issues with mouse models**

*Author: Mara Dierssen, Spain*

09:30 AM – 09:50 AM


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

*Author: Yann Herault, France*

09:50 AM – 10:30 AM

**Panel discussion, all speakers**

10:30 AM – 11:00 AM


Coffee break 

11:00 AM – 11:45 AM

**Keynote Lecture 3  
The interferonopathy of Down syndrome**

*Author: Kelly Sullivan, USA*

11:45 AM – 01:45 PM

Lunch break & Poster presentations 

01:45 PM – 03:15 PM

**NANO-S3: Understanding DS-AD disease mechanisms  
using rodent models**

*Chair: Antonella Tramutola, Italy*

01:45 PM – 02:00 PM

**The Cytogenetic and Down syndrome models resource at The  
Jackson Laboratory**

*Author: Jenny Morgenweck, USA*

Friday, 7 June 2024



- 02:00 PM – 02:15 PM **Unraveling deficits in neural encoding of space in Down syndrome mice**  
*Author: Andrea Contestabile, Italy*
- 02:15 PM – 02:30 PM **Triplication of HSA21–encoded genes modulates Alzheimer’s disease–related phenotypes in vivo via interferon signaling**  
*Author: Kyndal Schade, USA*
- 02:30 PM – 02:45 PM **Changed neuroimmune biology in mouse models of Down syndrome–Alzheimer’s disease**  
*Author: Paige Mumford, UK*
- 02:45 PM – 03:00 PM **Decoding plasma metabolic signature linking obesity and trisomy 21 in Dp(16)1Yey mice**  
*Author: Pinku Halder, France*
- 03:00 PM – 03:15 PM **The pharmacological increase of protein O–GlcNAcylation rescue Alzheimer signatures in Down Syndrome mice: A proteomics analysis of molecular target**  
*Author: Chiara Lanzillotta, Italy*
- 03:15 PM – 04:00 PM** **Keynote Lecture 4**  
**Montserrat Trueta Award**  
*Author: to be announced @ the conference*
- 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?**  
*Chairs: Daniella Balduino Victorino, France; Laurent Meijer, France*
- 04:30 PM – 04:50 PM **Elucidating the dose–dependent effects of DYRK1A on Alzheimer’s disease pathology in mice**  
*Author: Daniella Balduino Victorino, France*
- 04:50 PM – 05:10 PM **Functional MRI in DS models: impact of DYRK1A inhibitors**  
*Authors: Laura–Adela Harsan; Julien Caillete, France*
- 05:10 PM – 05:30 PM **DYRK1A/EGR1 pathway in vascular endothelial cells predispose to developing pulmonary arterial hypertension in Down syndrome**  
*Author: Hidehiro Suginobe, Japan*
- 05:30 PM – 05:50 PM **Role of DYRK1A in trisomy 21 megakaryopoiesis**  
*Author: Stella Chou, USA*
- 05:50 PM – 06:10 PM **DYRK1A inhibitors restore pancreatic functions in animal model of diabetes**  
*Author: Jamileh Movassat, France*



06:10 PM – 06:30 PM

**Phase 1 Clinical study of leucettinib-21, a dyrk1a kinase inhibitor drug aiming at the correction of cognitive disorders**

*Author: Laurent Meijer, 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*

08:30 AM – 09:00 AM

**Understanding normative change in executive functioning**

*Author: Anna Esbensen, USA*

09:00 AM – 09:30 AM

**Language measures to assess treatment efficacy**

*Author: Leonard Abbeduto, USA*

09:30 AM – 10:00 AM

**Memory measures in Down syndrome**


*Author: Hailey Van Vorce, USA*

10:00 AM – 10:30 AM


**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: Genomic and epigenetic mechanisms, therapeutic approaches and diagnosis**

*Chair: Floriana Costanzo, Italy*

01:45 PM – 02:00 PM

**The efficacy of Bumetanide for cognitive improvement in children and adolescents with Down syndrome: study protocol of a randomized, double-blind, placebo-controlled trial**

*Author: Floriana Costanzo, Italy*

02:00 PM – 02:15 PM

**Early detection and treatment of obstructive sleep apnea in infants with Down syndrome: a prospective, controlled clinical study**

*Author: Clotilde Mircher, France*

02:15 PM – 02:25 PM

**Rare potential loss of function variants in children with DS-associated congenital heart defects**

*Author: Elizabeth Feldman, USA*

02:25 PM – 02:35 PM

**Metabolic differences in Down syndrome basal forebrain cholinergic neurons**

*Author: Nicole West, USA*

Friday, 7 June 2024





02:35 PM - 02:45 PM **Variants in immune regulatory genes in Down syndrome regression disorder**  
*Author: Natalie Boyd, USA*

02:45 PM - 02:55 PM **Novel XIST minigenes locally repress Down syndrome critical region genes in an iPSC model of Trisomy 21**  
*Author: Eric Larsen, USA*

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*

04:30 PM - 05:00 PM **Catatonia and psychiatric phenotypes in DSRD**  
*Author: Cathy Franklin, Australia*

05:00 PM - 05:30 PM **Neurocognitive disorder differentiation in DSRD**  
*Author: Diego Real de Asua, Spain*

05:30 PM - 06:00 PM **Diagnostic work up in DSRD: when and how quickly to perform**  
*Author: Stephanie Santoro, USA*

06:00 PM - 06:30 PM **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*

08:30 AM - 09:00 AM **AD-Related Lipid and cellular energy dysmetabolism Occurs early in people with Down syndrome**  
*Author: Mark Mapstone, USA*

09:00 AM - 09:30 AM **Decoding genetic and sporadic forms of Alzheimer's disease: a multi-layered transcriptomic odyssey**  
*Author: Vivek Swarup, USA*

09:30 AM - 10:00 AM **Biofluid markers of Alzheimer's disease-associated CNS inflammation in adults with Down syndrome**  
*Author: Olivia Belbin, Spain*



10:00 AM – 10:30 AM **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*

01:45 PM – 02:00 PM **NIH perspective on international cohort development and data sharing**  
*Author: Dr. Huiqing Li, USA*

02:00 PM – 02:15 PM **Assembly and analysis of multidimensional datasets in the INCLUDE Data Hub**  
*Author: Dr. Joaquín Espinosa, USA*

02:15 PM – 02:30 PM **Multi-omics analysis of delayed development and early ageing in Down syndrome**  
*Author: Dr. Micah Donovan, USA*

02:30 PM – 02:45 PM **Standard criteria for database development and application of machine learning-based approaches: an Italian pilot study**  
*Author: Dr. Giuseppe Ramacieri, Italy*

02:45 PM – 03:00 PM **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*

04:30 PM – 05:00 PM **Altered bile acid metabolism in Down syndrome**  
*Author: Lauren Dunn, USA*

05:00 PM – 05:30 PM **Metabolic dysregulation in Down syndrome mouse models**  
*Author: G. William Wong, USA*



05:30 PM – 06:00 PM

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


06:00 PM – 06:30 PM

**Metabolic dysfunction associated to fatty liver disease in 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-S5 Physiological mechanisms of exercise intolerance in adults with Down syndrome	NANO-S6 Neurodegeneration and Ageing	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 Lecture 5 Miia Kivipelto			
02:15 PM – 03:30 PM	Science & Society meeting			
03:30 PM – 04:00 PM	Coffee break			
04:00 PM – 05:30 PM	Science & Society meeting			
07:00 PM	Closing Ceremony & Social Event: 10th Anniversary Dinner & Awards at Palazzo Brancaccio			



08:00 AM – 06:00 PM

Registration 

Saturday, 8 June 2024

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

**08:30 AM – 10:30 AM**

**Symposium 15: Immunomodulatory therapies in Down syndrome**

*Chair: Angela Rachubinski, USA*

08:30 AM – 09:00 AM

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

*Author: Angela Rachubinski, USA*

09:00 AM – 09:30 AM

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

*Author: Susan Pereira Ribeiro, USA*

09:30 AM – 10:00 AM

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


*Author: Jessica Bloom, USA*

10:00 AM – 10:30 AM

**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-S5: Physiological mechanisms of exercise intolerance in adults with Down syndrome**

*Chair: Thessa Hilgenkamp, USA*

11:00 AM – 11:15 AM

**Epidemiological approach to physical activity, fitness and healthy aging in older adults with intellectual disabilities**

*Author: Alyt Oppewal, The Netherlands*

11:15 AM – 11:30 AM

**Consequences of central mechanisms/autonomic dysfunction for work capacity in adults with Down syndrome**

*Author: Bo Fernhall, USA*

11:30 AM – 11:45 AM

**Oxygen uptake kinetics during submaximal exercise in adults with Down syndrome**


*Author: Victor Beck, USA*

11:45 AM – 12:00 PM

**The effects of exercise on peripheral blood flow regulation in adults with Down syndrome**


*Author: Thessa Hilgenkamp, USA*



- 12:00 PM - 12:15 PM **Exercise in a mouse model of Down syndrome, implications of metabolic energy source**  
*Author: Cristina Perez-Tenero, UK*
- 12:15 PM - 12:25 PM **Altered lower limb blood flow regulation during exercise in individuals with Down syndrome**  
*Author: Sara Sherman, USA*
- 12:30 PM - 01:30 PM Lunch break & Poster presentations 
- 01:30 PM - 02:15 PM** **Keynote Lecture 5**  
*Miia Kivipelto, Sweden*
- 02:15 PM - 03:30 PM** **Science & Society meeting**
- 02:15 PM - 02:20 AM **Introduction and opening**
- 02:20 PM - 03:00 PM **Presentations: Lifestyle and Health**  
*Chair: Lotta Granholm, USA*
- Finger-DS & lifestyle**  
*Author: Andre Strydom, UK*
- GO-DS study**  
*Author: Yann Herault, France*
- INCLUDE initiative**  
*Author: Sujata Bardhan, USA*
- Raising awareness of Down syndrome in the Global South**  
*Author: Eimear McGlinchey, Ireland*
- Discussion/questions**
- 03:00 PM - 03:30 PM **Talks: Independence and quality of life**  
*Chair: Anna Contardi, Italy*
- Motivational speech.**  
*Author: Sheri Brynard, South Africa*
- Debate with participants with Down syndrome.**  
**Michele Comai, appointed councilor;**  
**Riccardo Maino, Italian national Fisdor gymnast, world champion;**  
**Arianna Sacripante, synchronized swimming athlete, Italy**



03:30 PM – 04:00 PM

Coffee break **04:00 PM – 05:30 PM****Science & Society meeting**

04:00 PM – 04:50 PM

**Panel Clinical trials with anti-amyloid therapies and research participation***Chairs: Annie Cohen, USA ; Isabel Barroeta, Spain***Presentation Global DS***Author: Michelle Sie Whitten, USA***Anti-amyloid clinical trials in Down syndrome***Author: Isabel Barroeta, Spain***Research participation***Author: Annie Cohen, USA***Video participant research study (Lumind)****Debate**

04:40 PM – 05:10 PM


**Videos Co-researchers & health challenges****Co-researchers***Author: Eric Rubenstein, USA***Health Challenges***Author: Sujay Ghosh, India*

05:10 PM – 05:15 PM

**Closing remarks***Author: Anne-Sophie Rebillat, France ; María Carmona-Iragui, Spain  
Co-Chairs of the Science & Society Committee T21RS*

05:15 PM – 05:30 PM


**Cultural program: Rhythmic gymnastics***Author: Alice Pasini, Fisdir rhythmic gymnast, Italy***07:00 PM****Closing Ceremony & Social Event: 10th Anniversary Dinner & Awards at Palazzo Brancaccio** **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*

- 08:30 AM – 09:00 AM **Down syndrome in the Geroscience perspective: a focus on trajectories of epigenetic aging**  
*Author: Maria Giulia Bacalini, UK*
- 09:00 AM – 09:30 AM **Dissecting immune aging in people with DS**  
*Author: Bernard Khor, USA*
- 09:30 AM – 10:00 AM **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*
- 10:00 AM – 10:30 AM **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-S6: Neurodegeneration and ageing**  
*Chair: D. Allan Butterfield, USA*
- 11:00 AM – 11:15 AM **Fetomaternal transfer during Down syndrome pregnancies induces maternal cognitive decline in an APP-dependent manner**  
*Author: Eitan Okun, Israel*
- 11:15 AM – 11:30 AM **Hypothalamic structural involvement in the development of Alzheimer's disease in Down syndrome**  
*Author: Stephanie Brown, UK*
- 11:30 AM – 11:40 AM **Down syndrome and Alzheimer's disease: sleep deficits and dsRNA**  
*Author: Ryan Milstead, USA*
- 11:40 AM – 11:50 AM **Longitudinal retinal thickness in adults with Down's syndrome**  
*Author: Malwina Filipczuk, UK*
- 11:50 AM – 12:00 PM **Obstructive sleep apnea and Alzheimer's disease in Down syndrome**  
*Author: Dasoo Yoon, USA*
- 12:00 PM – 12:10 PM **Age-Associated changes of mitochondrial dysfunctions and UPRmt activation in the brain unravel potential mechanisms responsible for cognitive deficits in DS**  
*Author: Simona Lanzillotta, Italy*






12:30 PM – 01:30 PM

Lunch break & Poster presentations 

03:30 PM – 04:00 PM

Coffee break 

**07:00 PM**

**Closing Ceremony & Social Event: 10th Anniversary 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. Louis Valay, France*

08:30 AM – 09:00 AM

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

*Author: Adam M. Brickman, USA*

09:00 AM – 09:30 AM

**Blood–brain barrier pathophysiology in individuals with Down syndrome**

*Author: Lisi Flores-Aguilar, USA*

09:30 AM – 10:00 AM

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


*Author: Louis Valay, France*

10:00 AM – 10:30 AM

**Fluid biomarkers for vascular cognitive impairment**

*Author: Donna 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 


**07:00 PM**

**Closing Ceremony & Social Event: 10th Anniversary 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 



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