Trisomy 21 Research Society

2018 Annual report

Executive board of T21RS

The executive board is formed by the president, past-president, secretary and treasurer of T21RS, as well as the chairs of the committees:
President:
Mara Dierssen (Center for Genomic Regulation, Barcelona, Spain)
Secretary:
Marie-Claude Potier (Brain and Spine Institute, Paris, France)
Treasurer:
Alain Dekker (University Medical Center Groningen, Groningen, The Netherlands)
Committee chairs:
Program Committee: Anita Bhattacharyya
Committee for Science & Society: Peter Paul De Deyn
Committee for Sponsoring: Jean Delabar
Committee for Fellowships, Education and Training: Renata Bartesaghi
Committee for Pre-clinical Research: Yann Herault
Committee for Clinical Research: André Strydom

A new working group for Communication was initiated by the Executive Board, chaired by Dr. Mariluz Montesinos from the University of Sevilla, Spain.

The Trisomy 21 Research Society (T21RS) is the first non-profit scientific organization of researchers studying Down syndrome, founded to promote basic and applied research on Down syndrome, stimulate translational research and apply new scientific knowledge to develop improved treatments and cures.

The society aims to:

- ✓ Facilitate the permanent interaction between researchers studying Down syndrome by means of our website, scientific meetings, publications in journals and the two-yearly T21RS International Conference.
- ✓ Establish common protocols both for basic research (mice studies, stem cells studies) and translational research (for clinical trials with biomarkers, cognitive paradigms etc.).
- ✓ Support education and training of young researchers in all stages of their careers, including undergraduates, graduates and postdoctoral fellows that are interested in Down syndrome, by providing training programs and grants to young scientists Stimulate research on Down syndrome.
- ✓ Explain (recent) findings in Down syndrome studies to the general public and to inform legislators and other policymakers about new scientific knowledge and recent developments and their implications for public policy and society.
- ✓ Promote the interaction between scientists and patient associations, foundations and pharmaceutical industries

The society was created in April 2014 and statutes were registered on 17th April 2014 in Groningen, The Netherlands, under the auspices of Mr. Albert Kraster, civil-law notary practicing in Groningen and member of The Royal Dutch Association of Civil-law Notaries. These statutes were revised the 11th July 2017. A Governance document has been produced by the Executive Board of T21RS to facilitate operation. The society is a non-profit organization for the stimulation of scientific research on Down syndrome, operating under Dutch law. The original deed in Dutch and the English translation can be downloaded from the society website, t21rs.org, as well as the Governance document.

The society has organised a 1st International Conference in Paris on June 4-8 2015, a 2nd in Chicago June 7-11 2017 and the 3rd in Barcelona 5-9 2019.

An electronic ballot was organized for the election of the next General Secretary and the Chairs of the Clinical, Preclinical and Education committees. There were several nominations that were all accepted by the Executive Board. The list of nominees was the following:

Secretary: two nominees **Dr. Maria Martinez de Lagrán** from CRG Barcelona Spain and **Dr. Bing Ye** from University of Michigan US.

Clinical Committee: two nominees **Dr. Alberto Costa** from Case Western University US and **Prof. Rafa de La Torre** from PRBB Barcelona Spain.

Preclinical Committee: only one nominee **Prof. Elizabeth Fisher** from UCL London UK together with **Prof. Eugene Yu** from University Buffalo US.

Education and Training Committee: only one nominee **Dr. Sandra Guidi** from University Bologna Italy.

A new General Secretary has been appointed from summer 2019– **Dr. Maria Martinez de Lagrán** from CRG in Barcelona, Spain.

A new chairman of the Clinical Committee has been appointed from summer 2019– **Dr. Alberto Costa**.

A new chairman has been appointed from summer 2019– **Prof. Elizabeth Fisher**, together with **Dr. Eugene Yu**.

A new chairwoman has been appointed from summer 2019– **Dr. Sandra Guidi** from University of Bologna.

Results will be announced at the next General Assembly during the meeting in Barcelona Friday the 7th of June 3PM to 4PM.

Report of the Presidency

In January 2018 Mara Dierssen became the T21RS third president, after Jean Delabar and Roger Reeves. She was serving before as Secretary General, Chair of the Program committee, and President Elect.

Actions and advances: New activities and initiatives

Since its launch, T21RS has worked to expand the international community of researchers committed to understand the effects of trisomy 21, to recruit new generations of investigators, and provide to researchers in the field opportunities to collaborate and network, learn from experts, explore the newest tools and technologies, and discover great career opportunities. We have launched several new activities to this purpose. The Executive board also thoroughly revised the Society Statutes and finalized the Governance document.

1. **Improving communication**. We have **created a Communication Workgroup** (see report below) that has build up regular newsletters, an improved website and new communication strategies with patient associations and stakeholders. This **monthly newsletter** has increased the participation of the members in the Society, and has served to foster communication, discussion and debate on research topics amongst the T21RS members and to meet new members. Members contributed with their perspective, no matter the research interests in the field of Down syndrome, and ideas to the activities of the Society, working with colleagues from around the world to advance the progress of Down syndrome research. We have also worked on the branding of the Society and created a new logo with the company Mr. Brightsight.

2. **Attracting new members and ensuring the sustainability of the Society** has also been a priority. T21RS takes on that endeavor, has worked very hard to strengthen our membership and expand it worldwide and very specially with young investigators, as we are faced with a progressive leakage of critical mass in the field. We created two working groups to identify and attract new members. The number of members is now 276 with a steady growth in the last year (April 2018: 149; July 2018: 163).

3. **Promoting activities organized by T21RS members to engage local communities.** Our interest to create a strong community and to boost Down syndrome research visibility has led to the creation of a **Call for t21rs grants for the organization of scientific events for T21RS members.** This program is designed to contribute to the funding of scientific activities such as talks, meetings and events organized by members of T21RS, which will contribute to increase the public visibility of Down syndrome research and of T21RS. The range of activities funded includes from local activities on specific topics to multidisciplinary activities with a strong international participation. The maximum amount funded is 500€, depending on the availability of funds and the outcome of the evaluation. Eligible expenses: speaker travel and housing, venue and or equipment rental, catering etc.

4. **Advocacy: roadmapping the unmet needs of Down syndrome research**. T21RS is committed to lead our field as it confronts the difficult situation of reduced interest in treatment developments for intellectual disability disorders. We know how little funding goes into Down syndrome research as compared with other areas of medicine. Rather than waiting for something to change we need to be proactive. We have taken specific actions to make the public, policy makers, and relevant stakeholders aware of these research inequities and change that situation. For promoting advocacy and public communication about Down syndrome T21RS has contributed to several advocacy actions at the European parliament, European Commission, NIH. Several meetings were held to this purpose.

5. **Supporting members' applications and initiatives**. T21RS has supported or participates as partner in different grant applications, has promoted member's visibility, and has improved the publication of events organized and positions offered by T21RS members.

6. Ensuring that **young researchers** see the excitement and potential of a career in a rapidly advancing area of research. Thus, T21RS already launched the Annette Karmiloff-Smith Dissertation Award, and launched the Michael Harpold Dissertation Award in 2019, to honor the memory of these distinguished scientists. Besides we have almost **triplicated the amount of money devoted to support young researchers to attend the 3rd T21RS International Conference**.

7. **T21RS committees** have been working to continue promoting international pre-clinical and clinical research on Down syndrome, to stimulating translational research to develop improved treatments and cures, and have designed new actions to improving education of researchers in the field. The Science and Society has working to disseminate research news to families and understand

their needs (see below). The plan for next term is **to assign a specific budget to concrete actions proposed by the committees.**

8. **Increasing and improving the relationships with Down syndrome associations**. M Dierssen and J Delabar have worked very hard to re-establish the links with EDSA and initiated conversations with other associations.

9. **Institutional relationships.** T21RS has expended its institutional relationships, joining IBRO. This gives access to T21RS members to all IBRO open calls for funding and to IBRO resources.

10. The 3rd International Conference of the Trisomy 21 Research Society "Building the future therapies for Down syndrome" will be held on 6-9 June 2019 in Barcelona (http://t21rs2019.com/).

The meeting in Barcelona is an important part of our activities. We have a very strong and professional technical secretariat (BCO; https://www.bcocongresos.com/) and a local organizing committee (http://t21rs2019.com/organizing-committee/) consisting of excellent scientists and professionals who make sure that everything runs smoothly all year long for our congress. We created a devoted website and have carefully planned the event planning marketing strategy that has boosted delegate numbers this year. We created a Conference Logo in which we capture the essence of Barcelona (the "Trencadis") with the T21S logo.

This year, we will have exciting activities for anyone interested in Down syndrome research. We have planned a variety of sessions in new formats, making our scientific program even more diverse and interesting. The meeting will host four plenary lectures, and parallel sessions. Regular symposia, pre-meeting satellites, a Science and Society symposium, and "Meet the Expert" and "Education Committee" sessions are some of the offerings from which to choose. Another novelty this year is a new nanosymposium format in which selected groups of posters will get the opportunity to present their poster during a 30-minute session.

Although we planned hosting around 200 delegates from all over the world, based on the attendance to our previous meeting, in April we already reached 292 delegates and 37 families/association have registered to the new Families Program.

Young Investigators Program

This year, T21RS will kick off a Young Investigators Program, with many different activities (http://t21rs2019.com/young-investigator-programme/). Young investigators are an integral part of the T21RS Conference and T21RS has installed a variety of measures to ensure that their participation is made as fruitful as possible. The young investigators will have the opportunity to apply for travel grants, join the volunteers program, attend focused interest sessions, participate in the "Meet the Scientist" session or even visit during one month a research lab investigating on Down syndrome in Spain to learn new techniques or simply get to know the lab and discuss about job opportunities. They will also have plenty of opportunities for their career development.

Families Program at T21RS: connecting with Down syndrome needs

We continue with the Science and Society symposium (http://t21rs2019.com/science-society/), that this year has reached the highest number of registrations, with 231 attendees already planning to attend (data in April). This Program has expanded this year, to a full day program to be held on Saturday, June 8, 2019. This is attracting many families who did not want to travel to only attend a 3-hour session. Also we reduced the fee to 20 Eur, in agreement with the S&S committee. The full

program is composed by three sessions: a session on Advocacy, Awareness and Fundraising; the Science and Society Symposium, and the Bridging Knowledge Session.

09:00-11:30 ADVOCACY, AWARENESS & FUNDRAISING SESSION

Through a series of short presentations and Q&A, this session will address important issues for self-advocates, family members, and Down syndrome organizations regarding three areas that are intricately related – advocacy, awareness & fundraising for Down syndrome research. To facilitate the interaction, English-Spanish and Spanish-English translations will be provided. This symposium is organized by Global Down syndrome.

09:00 Welcome: Mara Dierssen (President, T21RS) and Jean Delabar (Chair of the Fundraising and Membership Committee)

09:15 The Importance of DS Research & How DS Organizations and Families Can Advocate for Government Funding: Michelle Sie Whitten (T21RS Chair of Morning Session, President & CEO of Global Down Syndrome Foundation, Parent)

09:50 Self-Advocates with Down Syndrome Make it Happen!: TBD Megan Bomgaars (Emmy-award winning Actor, Entrepreneur, Motivational Speaker, Self-Advocate)

10:15 Scientists with Family Members with Down Syndrome – Things to Consider Before Participating in Research: Keith Smith (Crnic Institute Human Trisome Project Program Manager, Brother) & Katherine Waugh (Crnic Institute Postdoctoral Fellow, Sister-in-law, Down Syndrome World magazine science contributor)

10:50 Engaging the Press, Self-Advocates & Celebrities to Raise Awareness & Funds: Michelle Sie Whitten (T21RS Chair of Morning Session, President & CEO of Global Down Syndrome Foundation, Parent) and TBD Guest speaker from Spain or Europe (head of a DS fundraising organization)

Final Q&A/Interactive Discussion 11:30 END

11:45- 14:30 SCIENCE AND SOCIETY SYMPOSIUM (see below report of the S&S Committee)

This symposium is organized by the Science and Society Committee of the T21RS

15:00-18:00 BRIDGING KNOWLEDGE SESSION

This session is organized and supported by Jesús Flórez (University of Cantabria, Fundación Iberoamericana Down21, Spain), Carmen Martínez-Cué (University of Cantabria, Spain)

Doctors and scientists may be brilliant in their fields, but some may not be great at communicating their knowledge to the lay people. A good communication is important when research scientists are presenting their work aiming to reach non-scientific participants. With that goal, a team of experts directed by Profs. Jesús Flórez and Carmen Martínez-Cué will offer excerpts and summaries in Spanish of the relevant findings and discoveries reported during the conference. Profs. Flórez and Martínez-Cué will simplify the technical language to lay terms so that the main findings can reach families and other professionals of the Down syndrome associations.

The excerpts will be used as templates for the Bulletin

Local advocacy

Our biennial meeting is a unique opportunity to increase public awareness of the progress and benefits of Down syndrome research at the local level. In Spain the Trisomy 21RS International Conference is being a year-long event intended to bring together scientists, families, schools, and communities to promote Down syndrome research. The meeting has been recognized by the local

authorities as shown by the very important policymakers in our Honorary Committee: http://t21rs2019.com/honorary-committee/

I- Program committee

Program Committee members: Anita Bhattacharyya, Tom Blumenthal, John Crispino, Yasuji Kitabatake, Victor Tybulewicz, Bradley T. Christian, Weihong Song, Juan Fortea, Anna Esbensen, Pablo Helguera, and Roger Reeves

The Program Committee was formed in April 2018 to help craft the scientific content of the T21RS meeting in Barcelona in June 2019. The duties of this Committee include submission of symposia proposals, review and selection of symposia sessions, providing feedback and suggestions for schedule and plenary speakers, as well as selection of late breaking abstracts.

Following is the current schedule of the meeting:

T21RS2019 Program (Updated March 13, 2019)

Wednesday, June 5, 2019
17:00 – 20:00 Registration and bridging knowledge sessions

Thursday, June 6, 2019
9:00 – 18:00 Registration

09:00 – 13:00 PRE-MEETING SATELLITE SYMPOSIA (parallel)
CLINICAL SESSIONS
T21RS Committee for clinical research: Andre Strydom (University College London, UK) (chair)

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)

9:00 – 9:30 Registration / coffee
9:30 – 9:45 Welcome and Introduction – Andre Strydom (University College London, UK)
9:45 – 10:15 Overview of the health needs of older adults – Tonnie Coppus (Radboud University Medical Center, NL)
10:15 – 10:45 Sleep Apnea – 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 – TBC
12:00 – 12:30 Summary and discussion

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)

9:00 – 9:30 Registration / coffee
9:30 – 9:45 Welcome and Introduction – Stephanie Sherman (Emory University, USA)
9:45 – 10:15 Overview of the health needs of children with Down syndrome - Jesus Florez (Affiliation—to be confirmed)
10:15 – 10:45 Medical comorbidities of childhood and their potential impact on cognition and development - George Capone, Kennedy Krieger Institute, USA-to be confirmed)
10:45 – 11:00 Break
11:00 – 11:30 Psychiatric comorbidities of childhood/adolescents and their association with cognitive impairment - Stefano Vicari (Ospedale Pediatrico Bambino Gesu, IT)
11:30 – 12:00 Interventions from early childhood to adolescents--how we can help - Rafael de la Torre Fornell, (Universitat Pompeu Fabra, SP)
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 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)

Mouse master class
9:00 – 9:15 Registration / coffee
9:15 – 9:45 Overview of currently available preclinical mouse models – Yann Hérault (Institut de Génétique et de Biologie Moléculaire et Cellulaire, FR)
9:45 – 10:15 Genetic quality control, maintenance and breeding procedures - Eugene Yu (Roswell Park Cancer Institute, USA)
10:15 – 10:45 Classical behavioural 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 Delabar (CNRS, FR)
12:30 – 13:00 Discussion on key information about models and their availability (moderated by Eugene Yu) and perspective on new animal models for DS research Roger Reeves (Johns Hopkins University School of Medicine, USA) and Victor Tybulewicz (Francis Crick Institute, UK)

14:00 – START OF MEETING

14:00- 14:30 Welcome note (Mara Dierssen, President T21RS and local authorities)

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)

16:30 -18:00 Poster SESSION 1 + Coffee break

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

19:30 -20:30 Romeo and Juliette: Theater play by Down syndrome directed by Victor Ignacio Romero Rojas, from the Compañía Nacional de Teatro Manantial de Ilusión (Chile)

21:00 – 22:00 Opening ceremony (Musical Ensemble + welcome cocktail reception)

Friday, June 7, 2019
8:00 – 18:00 Registration

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

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

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

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

1. Touchscreen learning in Fmr1, Ube3a, Ts65Dn and Mecp2 mouse models of neurodevelopmental disorders with intellectual disabilities, *Prescott Leach* (Biogen, 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)

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

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

1. Cognitive correlates of amyloid PET in Down syndrome, *Sigan Hartley* (University of Wisconsin, USA)
2. MRI-derived 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)

11:30-12:00 Coffee Break

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

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)

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

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

1. 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).

14:00 – 15:00 Lunch break

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

16:30 – 18:00 Poster SESSION 2 with coffee

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

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

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

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)

SESSION 4B: Cognitive outcomes in children with Down syndrome

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

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, ItT)

3. Relationship between executive functioning and behavior in children with Down syndrome, *Anna Esbensen* (Cincinnati Children's Hospital Medical Center, USA)

21:00 GALA DINNER

Saturday, June 8, 2019

9:00 – 18:00 Registration

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

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, France) 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, *Gil DiPaolo* (Danali, USA)
3. Excess synaptojanin 1 contributes to place cell dysfunction and memory deficits in the aging hippocampus in Down syndrome, *Catherine Marquer* (Columbia University, USA)

SESSION 5B: Language Outcome Measures for Treatment Studies.

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

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 Thurma*n (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)

11:30-12:00 Coffee break

11:45- 14:30 SCIENCE AND SOCIETY SYMPOSIUM

T21RS Committee for Science & Society: Peter De Deyn (University Medical Center Groningen, NL) (Chair), 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)

11:45 Welcome and introduction: *Peter De Deyn (chairman T21RS Committee for Science & Society)*

12:00 Parent-reported needs for research (10 min. presentation + 20 min. debate)
Hampus Hillerstrom, LumindRDS, USA.
Results will be presented from the parents surveys on knowledge, attitude and behavior towards Down syndrome research, conducted by LumindRDS in the USA and Fondation Lejeune in the European Union.

12:00 Clinical studies (10 min. presentation + 20 min. debate)

Juan Fortea, Fundació Catalana Síndrome de Down and Hospital San Pau, SP.
Overview of clinical studies and what we can expect in the (near) future. Are new clinical trials coming up, e.g. in the Alzheimer's disease field? Plenary discussion with families and foundations.

13:00 Lunch break (junior investigators will be available to meet with families)

13:45 Participation in research: pros and cons. How to involve families in research? (30 min. debate)
Peter De Deyn, University Medical Center Groningen, NL
Discussion with audience about needs and wishes for research. What's the view of family members and caregivers on participation in research?

14:15 Ethics: autonomy of people with Down syndrome (10 min. presentation + 20 min. debate)
Jesus Flórez, University of Cantabria, Spain
How can we promote autonomy of people with Down syndrome? What should – and should not – be expected from this autonomy?

14:45 Human Rights Assembly *Fundació Catalana Síndrome de Down, SP*

15:00 Summary and closure *Peter De Deyn*

15:30 – 17:30 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 AB 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)

17:30-19:00 Poster SESSION 3 with hors d'oeuvres

19:00-20:00 Meet the Experts: To be determined (parallel)

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

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

09:30-10:15 Annette Karmiloff-Smith and Michael Harpold Thesis Awards Ceremony *(Renata Bartesaghi)*
10:15-10:30 Montserrat Trueta Award (*Mara Dierssen*)

10:30 – 11:00 Coffee break

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

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. Novel therapeutics for B-cell acute lymphoblastic leukemia in Down syndrome, S*ebastien Malinge* (Telethon Kids Institute, AU)
2. Hematopoietic skewing induced by trisomy 21, *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)

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 drives production of neurotoxic tryptophan catabolites via the interferon-inducible kynurenine pathway, *Kelly Sullivan* (University of Colorado, USA)
3. Exosome biomarkers reflecting brain changes in Down syndrome with Alzheimer's disease, *Aurelie Ledreux* (University of Denver, USA)

13:00 –14:00 lunch

14:00-16:00 SCIENTIFIC SESSION 8

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

Chair: Jean Delabar (Paris Sorbonne Université, CNRS, FR) and Floriana Costanzo (Bambino Gesù Children's Hospital, IT)

1. GABA in the brain, *Trevor Smart* (University College London, UK)
2. Modulating cortical networks in physiological and pathological conditions by tuning GABAα5 receptors, *Javier Zorrilla de San Martin (*ICM, FR)
3. GABA targeting drugs and clinical trials, *Marie-Claude Potier* (UPMC Université, INSERM, CNRS, FR)

16:00-16:30 Coffee break
16:30-17:00 Melissa Parisi: updates on US NIH funding (ABC-Connect, INCLUDE)
17:00-17:30 Stephane Hogan: long-term Horizon Europe budget in neuroscience and cognition

17:30-19:30 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)
3. Outcome measures for Alzheimer's disease trials in Down syndrome, *Mike Rafii* (University of Southern California), USA)
4. Down Syndrome Clinical Trials Network (LuMind) *short talk*

19:30 CLOSING CEREMONY. Poster awards with aperitif

20:30 END OF MEETING

II- Clinical Research committees

Adult/ Alzheimer's committee members

Andre Strydom UK (Chair)
Shahid Zaman (UK),
Ira Lott (USA),
Tonnie Coppus (NL),
Juan Fortea (SP),
Weihong Song (CA - CN)
Elizabeth Head (US)

The T21RS adult clinical committee has continued to encourage collaborations in cohort and biomarker studies of Alzheimer's disease (AD) in Down syndrome (DS) globally. This has included active support to several projects and input at key meetings:

Development of the Lumind-funded DS clinical trials network (DS-CTN) at a successful kick-off meeting in Boston on the 9th of March 2018. We have been involved in developing the cognitive test protocol that will be used in this network.

We have informed discussions by the NIH to fund an Alzheimer's disease in Down syndrome clinical trials network - https://www.nih.gov/sites/default/files/research-training/initiatives/include/INCLUDE-full-meeting-summary.pdf . We have also provided input at a subsequent workshop DS/AD workshop on the 12/13th of March 2019.

Most of the committee members have participated in funding applications to the NIH INCLUDE call(s) to develop clinical trials in DS that resulted from these discussions - https://www.nih.gov/include-project

A project funded by the LeJeune Foundation and Lumind-DSRF to identify, refine and validate clinical outcome measure(s) that will be used in clinical trials of treatment to prevent or delay dementia in individuals with Down syndrome is ongoing, involving collaborators from the Horizon21 consortium in Europe (Andre Strydom, Shahid Zaman, Juan Fortea, Johannes Levin, Tonnie Coppus and Anne-Sophie Rebillat) as well as several US research groups (Ben Handen, Ira Lott, Elizabeth Head).

A systematic review of the psychometric properties of IQ tests and adaptive behaviour scales in adults with Down syndrome to help with matching between studies is now in press.

The committee has plans to meet in person and to present some of the ongoing work from associated research groups at the AAIC meeting in Los Angeles and at the T21RS meeting in the summer of 2019.

The Committee has organized a satellite meeting on **Health co-morbidities in older adults with Down syndrome – assessment and diagnosis** in the T21RS Congress in Barcelona.

Contact Information:

Dr. Andre Strydom, Chairman T21RS Clinical Research Committee – Adult/Alzheimer's

Andre.strydom@kcl.ac.uk

A new chairman has been elected from summer 2019– Dr Alberto Costa. He will present his strategic plan at the General Assembly and the Clinical Committee Meeting In Barcelona

Developmental committee members:

Stephanie Sherman (US) & Stefano Vicari (IT) (co-chairs)
Rafael de la Torre (ES)
Julie Korenberg (US)
Cécile Cieuta-Walti (FR)
Alfieri Paolo (IT)
André Strydom (UK)
Stefania Veltri
Jamie Edgin (US)
Sophie Durand (FR)
Silvia Sacco (FR)
Brian Skotko (US)
Maria Stanley (US)

The developmental committee has as it goal to create a body of investigators interested in the cognitive and behavioral profile of individuals with DS and the developmental trajectory.

The committee is continuing to establish an International Down Syndrome Brain and Behaviour Consortium, staring with harmonization of existing datasets, and to establish a pilot project. Next steps include prioritizing questions that can be answered with these data sources (both pilot data and existing data).

The Committee has organized a satellite meeting on **Health co-morbidities in children with Down syndrome** in the T21RS Congress in Barcelona.

Contact Information:

Drs. Stephanie Sherman & Stefano Vicari, Chairwoman and Chairman T21RS Clinical Research Committee – Developmental

ssherma@emory.edu

stefano.vicari@opbg.net

III- Preclinical Research committee

Committee members:

Antonarakis, Stylianos Stylianos.Antonarakis@unige.ch (2015)
Bhattacharyya, Anita Bhattacharyy@waisman.wisc.edu (2018)
Delabar, Jean-Maurice jeanmaurice.delabar@icm-institute.org (2015)
Dierssen, Mara Mara.Dierssen@crg.eu (2015)
Fisher, Elizabeth M elizabeth.fisher@ucl.ac.uk (2015)
Gardiner, Katheleen (Katheleen.Gardiner@ucdenver.edu) (2015)
Herault, Yann herault@igbmc.fr (2015-CHAIR)
Mobley, William williammobley7@gmail.com (2015)
Potier, Marie-Claude marie-claude.potier@upmc.fr (2015)
Reeves, Roger rreeves@jhmi.edu (2015)
Roper, Randall J rjroper@iupui.edu (2017)
Yu, Y. Eugene Yuejin.Yu@RoswellPark.org (2015)

The committee is still pursuing his ambitious objectives:

1. to facilitate the access to cellular and animal models: build a simple nomenclature, create a resource sharing plan, harmonization of generation and storage protocols, set up an accessible virtual repository (database)
2. to establish common protocols for preclinical research: behavioural analysis, cellular characterisation, and breeding schemes for Down syndrome models
3. to capture and make available data from phenotyping including OMICS data; joining international initiative such as IMPC www.mousephenotype.org
4. to validate protocols for preclinical and translational medicine: establish and validate new disease-relevant phenotypes and protocols for preclinical pharmacology studies, publish position papers on new tools for manipulating new targets in cellular and mouse models

In 2018 we welcome Anita Bhattacharyya as a new member of the committee for the iPS preclinical model.

In the context of the work of the Preclinical Committee of the Trisomy 21 Research Society, several objectives have been recognized to improve the preclinical research in our field:

1. The list of DS mouse models is updated with current published models and a few that are in progress in rats (YH Personal communication, Birling et al., Sci rep. 2018). An updated list has been put on the website.
2. We generated a new table with trial assays (successful or not) on DS mouse models. The update table is on the website.
3. The document for craniofacial and skeleton defects have been delayed.
4. We have organised a mouse master class to present DS mouse models and their use at the next Barcelona T21RS meeting in 2019 with contribution from the main participants of the PC committee
5. We have elected a new chair with a co-chair that will serve after the Barcelona T21RS meeting in 2019
6. We should think about renewing the panel of members especially for those (at least 4 persons) who are never attending the monthly conference call.

The Committee has organized a satellite meeting on **Mouse Master Class** in the T21RS Congress in Barcelona.

Contact Information:

Dr. Yann Hérault, Chairman T21RS Preclinical Research Committee
yann herault@igbmc.fr

A new chairman has been appointed from summer 2019– Prof. Elizabeth Fisher, together with Dr. Eugene Yu. They will present their strategic plan at the General Assembly and the Clinical Committee Meeting In Barcelona.

IV- Education, Training and Fellowships committee

Committee members:

Renata Bartesaghi
Tom Blumenthal
Carmen Martinez Cué
Tarik Haydar

- In the year 2017, the Committee for Fellowships, Education and Training had launched the "Annette Karmiloff-Smith Thesis Award Program, for outstanding Ph.D. thesis. Two prizes of 1,000 euro each. Eligibility. Participation in the 2017 competition open only to candidates who obtained their Ph.D. title during the period January 1, 2016-December 31, 2017. Application Deadline: June 30, 2018. The winners will present results of their theses during the T21RS meeting in Barcelona, June 2019.

- In the first months of 2018, the Committee publicized the Award Program through e-mails sent to all members of T21RS.

- In a short article published in the Newsletters 2018 of T21RS, the Committee outlined the history and the purpose of the Award Programs for Outstanding Ph.D. Thesis, in order to make T21RS members and, in particular, young people aware of this initiative.

- For the 2017 Competition, the Committee received five applications. For the evaluation, the Committee has taken into account the originality and contribution to the literature, importance of the research, innovation, employed techniques; general presentation of the thesis and academic curriculum of the applicant. The PhD theses were first examined individually by each member of the Committee and then evaluated in a collegial skype meeting that took place on September 2, 2018. At the end of the skype meeting the Committee decided unanimously to assign the awards to the thesis by Nadine Aziz entitled "HISTOLOGICAL, CELLULAR, AND MOLECULAR ABNORMALITIES IN FOREBRAIN AND SPINAL CORD OF THREE DISTINCT MOUSE MODELS OF DOWN SYNDROME" and to the thesis by Eric Hamlett entitled "INVESTIGATIONS AT THE CROSSROADS OF DOWN SYNDROME AND ALZHEIMER'S DISEASE".

- The results of the 2017 Competition will be announced by the Committee during the T21RS Barcelona meeting, June 2019, in a dedicated session where the winners will present their work.

- In synergy with the Committee for Science and Society, the Committee for Fellowships, Education and Training has invited the two winners of the 2017 Competition to write a short outline of their thesis work to be published in the Bullettin of T21RS.

- The Committee has conducted an e-mail survey among T21RS young members in order to organize actions aimed to foster the scientific development of the young scientists of T21RS and to involve them in the T21RS Barcelona meeting, 2019.

- In the framework of the T21RS Barcelona meeting, 2019, the Committee has organized an educational session entitled “How should we study Down syndrome: pros and cons of mouse models and human iPSCs”.

Contact Information:

Dr. Renata Bartesaghi, Chairwoman T21RS Education, Training and Fellowships Committee
renata.bartesaghi@unibo.it

A new chairwoman has been appointed from summer 2019– Dr. Sandra Guidi from University of Bologna. She will present her strategic plan at the General Assembly and the Clinical Committee Meeting In Barcelona

V- Sponsoring committee

Chair: Jean Delabar (FR)
Members: Michelle Whitten (GDSF, USA), Michael Harpold (Lumind, USA), Marie-Claude Potier (FR). In March 2017 we lost **Michael Harpold** who was very influential at the interface of Pharma and Academic Research and had a major impact on health care through Lumind RDS grants program

Communication between members has been maintained via conference calls and emails

During 2018 research of sponsors had three objectives:

1. to maintain functioning of the society to cover basic expenses (web, notary);
2. to fund travel grants and research awards;
3. to fund the biennial meeting to be held in Barcelona in 2019.

Sponsors can be classified in five circles:

1. The first one is the circle of founding supporters: Lumind, Global Down Syndrome, Lejeune Foundation, Matthews Foundation, Trisomie 21 France, who committed themselves to give 5000 € per year for 5 years (2014-2019); during 2018 Trisomie 21 France was not able to contribute. Thus Trisomie 21 France has lost its Founding Member Status
2. The second circle is including organizations that have accepted to commit themselves to give 5000€ per year for the next five years (2020-2025): Down España, contacted by M Dierssen, has accepted this commitment.
3. The third circle concerns foundations that gave specific support for the Barcelona meeting including founding supporters Fondation Jerome Lejeune (application by M Dierssen, 34.500€), Lumind (contact M Dierssen, JM Delabar; 17.000 €) and other foundations contacted by M Dierssen like La Caixa (18.000 €), Down syndrome UK, The Fundación Iberoamericana Down21 (3.000,00 €), Down España (5.000,00 €), the company of Biologists (2.223,00 €), EMBO (1.000,00 €), PROUS Institute (2.400 €), Aelis Pharma (5000 €), Fundación IMIM (2.000,00 €), CRG, FECYT, BBVA, or MC Potier AMIPI (2000€) to a total in April of 92.123,00 €.
4. The fourth circle is the institutional granting agencies: an NIH grant was obtained by A Bhattacharrya, R Reeves, M Dierssen and J Busciglio to fund travels of PhD and Postdoc US members of T21RS for the 3rd International Meeting of T21RS in Barcelona in June 2019.
5. The fifth circle put together Pharmaceutical and Biotech companies with an interest in developing treatments for DS

Concerning **membership** the committee has developed benefits for members and has tried to attract new members. We created to subcommittees (Asian and Latin-American) who have helped to identify possible scientists in those regions interested in joining the T21RS. We contacted most of them.
The committee is also maintaining a contact list (more than 1000 names) of scientists and clinicians who are or have been involved in DS research. This directory is a valuable resource to publicize the work of T21RS committees and to advertise the biennial meeting of the Society.

Contact Information:
Dr. Jean-Maurice Delabar
Chairman T21RS Sponsoring and Membership Committee
sponsoring@T21RS.org

VI- Science and Society committee

1. General information

The T21RS Committee for Science & Society aims to be in contact with regional and (inter)national Down syndrome associations to disseminate and explain recent scientific findings in understandable language to family members and caregivers. Moreover, the Committee for Science & Society, chaired by prof. Peter Paul De Deyn, receives input from these associations on key issues that you feel should be investigated.

Address: T21RS Groningen Office
University Medical Center Groningen (UMCG)
attn. prof. PP De Deyn (AB51)
PO Box 30.001, 9700 RB Groningen, The Netherlands

Chairman: Peter Paul De Deyn (Belgium)
Committee members (2017): Alain Dekker (The Netherlands), Juan Fortea (Spain), Lotta Granholm (USA, Sweden), Cindy Lemere (USA) and Sebastián Videla (Spain). Diana Bianchi (USA), the new director of the American National Institute of Child Health and Human Development, contributed in 2017 with respect to the organization of the Science & Society Symposium in Chicago, June 2017.

2. Initiatives and achievements 2017

In the fourth year of T21RS, the Committee concentrated its efforts on 3 major initiatives:

1) T21RS Science & Society Bulletins
   → Publishing 4 bulletins
2) T21RS Science & Society Symposium 2019
   → Preparations for the Symposium during the T21RS International Conference 2019 in Barcelona
3) Contributing to The Dementia Table Initiative in The Netherlands

3.1. T21RS Science & Society Bulletins

The Committee for Science & Society is strongly committed to introducing scientific research and explaining recent findings in an understandable way through their T21RS Science & Society Bulletins. The Bulletins are short communications introducing the broad, non-scientific readership to a specific topic related to Down syndrome (research). The Bulletins do not aim to provide all ins and outs about a specific study with a lot of reservations. Instead, the Bulletin aims at introducing the readers to the current state-of-the-art knowledge about the topic of the article (mini-review) including the highlighting of important findings from recent research.
T21RS members are kindly invited to submit a T21RS Science & Society Bulletin to the Committee for publication on the T21RS website (author guidelines). Given the fact that only few members, so far, proposed to write a Bulletin, the Committee decided to focus on invited Bulletins as well. In 2018, a new policy started, asking the T21RS Thesis Award Winners and the elected Honorary Members to write a Bulletin.

In 2018, a total of 4 Bulletins was published (open access) on our website. Below you will find the 4 titles and abstracts. Direct link for full (and free) downloads for members and non-members:
https://www.t21rs.org/science-society/t21rs-science-society-bulletin

T21RS Science & Society Bulletin, 2018 (1)

A clue to Alzheimer's disease in Down syndrome: a summary of my PhD thesis

by M. Florencia Iulita (Montreal, Canada)

Abstract: Alzheimer's disease is a common medical problem in older adults with Down syndrome. Given that the brain changes that lead to Alzheimer's disease develop decades before signs of dementia manifest, studies focusing on the early mechanisms of Alzheimer's onset might offer valuable clues for diagnosis and treatment. The recent discovery of a group of molecules responsible for the health of memory brain cells, and their dysregulation in Alzheimer's disease and Down syndrome brains, could provide new insights for the development of new biomarkers or the identification of novel therapeutic targets.

T21RS Science & Society Bulletin, 2018 (2)

Is it possible to rescue maturation of nerve cells in Down syndrome with early pharmacological treatment?

by Fiorenza Stagni (Bologna, Italy)

Abstract: Intellectual disability in Down syndrome represents a major concern for families and society. Alterations of generation and maturation of nerve cells starting from early phases of brain development are key determinants of intellectual disability. Based on previous evidence that pharmacological treatment with the antidepressant fluoxetine in the neonatal period restores the reduced generation of new nerve cells in a mouse model of Down syndrome, the objective of my PhD thesis work was to establish whether fluoxetine also restores the abnormal maturation of nerve cells. I focused on the hippocampus, a region severely impaired in Down syndrome and fundamental for learning and long-term memory.
My study shows that early treatment with fluoxetine fully restores maturation of hippocampal nerve cells. Importantly, treatment also restores the communications between hippocampal nerve cells. Taken together, the results show that treatment with fluoxetine is sufficient to rescue the two major alterations of brain development in a Down syndrome mouse model.

T21RS Science & Society Bulletin, 2018 (3)

Prof. Marie-Odile Rethoré, Her Patients and Research – *a Life* –

by Aime Ravel, Clotilde Mircher, Daniel Satgé and Catherine Lemonnier (Paris, France)

Abstract: The Trisomy 21 Research Society (T21RS) awarded an honorary membership to professor Marie-Odile Rethoré. Marie-Odile Rethoré (Paris, France) is 89 years old and a well-known clinician who devoted all her life to patients with Down syndrome during her clinics. During her life, Marie Odile Rethoré took care of people with Down syndrome. Day after day (and sometimes night after day) she gave advice and support to family members and other caregivers. She initially worked with prof. Jérôme Lejeune in the Trousseau Hospital in Paris and continued at the Jérôme Lejeune Institute. She was full-time in charge of young children with intellectual disabilities, mostly Down syndrome, and followed her patients' life from birth until old age. She has more than 160 publications, of which almost 40 are on Down syndrome research. She provided trainings to caregivers, physicians and families. She is member of many well-known scientific academies, in particular the Academy of Medicine in Paris.

T21RS Science & Society Bulletin, 2018 (4)

'What are you proud of?' 'Everything'

by Claudia Cannavo (London, United Kingdom)

Abstract: LonDownS Consortium is an association of researchers working on different aspects of Down syndrome. We celebrated World Down Syndrome Day 2018 by organising a day of activities, scientific talks, and games to thank people with Down syndrome that took part in our studies.

The 21st of March (21/3) is World Down Syndrome Day – as people with Down syndrome have 3 copies of chromosome 21.

The LonDownS Consortium is a group of researchers working on different aspects of Down syndrome, that come together to build a comprehensive approach on Down syndrome research.

To celebrate World Down Syndrome Day 2018, LonDownS Consortium organised a day of talks, games, and activities for people with Down syndrome and their family and friends, aimed at bringing science closer to the non-scientific community

3.2. Preparation for T21RS Science & Society Symposium 2019 in Barcelona

Defined in the articles of association, T21RS aims, among others, to promote the understanding of, and involvement in Down syndrome research among the general public, as well as stimulate interactions between scientists and Down syndrome (family) associations. Next to publishing Science & Society Bulletins, the Committee, therefore, organizes a bi-annual T21RS Science & Society Symposium.

After a very successful kick-off symposium in 2015 in Paris (Delabar et al., 2016) and the lively second edition in 2017 in Chicago (Reeves et al., 2018), the Committee is now organizing a third edition during the T21RS International Conference 2019 in Barcelona.

Based on evaluations and feedback on the 2015 and 2017 editions, and suggestions from (founding) members, the Committee drafted a new program. The Science & Society Symposium is embedded within a larger families program. Importantly, the Symposium aims to stimulate interaction between families and scientists, and thus no parallel scientific sessions are scheduled, to facilitate scientists to join. After various rounds of refinement together with the Program Committee, the 2019 preliminary program is as listed above in the full program (page 12-13)

3.3. Dementia Table initiative

In The Netherlands, the committee contributed to organizing two Dementia Table evenings in 2018. During these evenings, a variety of topics in the field of dementia and intellectual disabilities/Down syndrome is discussed in an easy-accessible way and in a pleasant ambiance. In 2018, we focused, among others, on 1) different ways to support/aid people with intellectual disabilities and dementia, and 2) the multidisciplinary process from early changes to a diagnosis of dementia. The Dementia Table initiative is very successful in Groningen (120 participants each time) and a perfect example of interaction between science and society. More info: www.sdtg.nl (in Dutch)

Contact Information:

Prof. dr. Peter Paul De Deyn

Chairman T21RS Committee for Science & Society

Department of Neurology & Alzheimer Center, University Medical Center Groningen, The Netherlands p.p.de.deyn@umcg.nl

VI- Communication Workgroup

Members:

María Luz Montesinos, Chair

Eric Hamlett

We started our activity as Communication Working Group on June 2018.

1. We are in charge of producing a monthly Newsletter, which is distributed to the T21RS members who signed to receive it (currently 281) and also among a list of interested persons (currently 88) who subscribed through our web site.
2. We update the web site (English version) with news, events and job offers when solicited. A simplified version of the Spanish web site has also been set-up.
3. Finally, we act as Community Managers of the T21RS Twitter account. We have presently 229 followers (around 90 new followers since June 2018).

Contact Information:

Dr. Maria Luz Montesinos
Departamento de Fisiología Médica y Biofísica
Universidad de Sevilla, SPAIN
mlmontesinos@us.es

Financial Report 2018
01-01-2018 – 31-12-2018

1. Treasury

T21RS is the first non-profit scientific society (*Dutch:* vereniging) for Down syndrome research. T21RS is officially established in Groningen (NL), and operates under Dutch law.

Official statutory address:	T21RS Groningen Office University Medical Center Groningen *attn. P.P. De Deyn & A.D. Dekker (AB51)* PO Box 30.001, 9700 RB Groningen The Netherlands
RSIN identification number (NL):	853938283
KvK Chamber of Commerce number (NL):	60501162
Current treasurer (2016-present):	Dr. A.D. (Alain) Dekker University Medical Center Groningen, The Netherlands
Past-treasurer (2014-2015):	Dr. A.M.W. (Tonnie) Coppus Radboud University Medical Center Nijmegen, The Netherlands
Operational currency:	Euro (€)
Number format:	Continental European Example: 40.000,25 *(forty thousand and twenty-five cents)*

2. Summary of 2018

Given the fact that the T21RS International Conference is organized every two years, 2018 can be considered a quiet off-year in-between the Chicago conference (2017) and Barcelona conference (2019). Income in 2018 primarily concerned membership fees and annual sponsoring obtained from the Founding Sponsors. Moreover, preparations started for the T21RS International Conference 2019 in Barcelona. Two organizations (Fundación Iberoamericana Down21 and The Company of Biologists) have already provided their conference sponsorship to T21RS in 2018. On the other hand, expenses have been reduced to an absolute minimum in this off-year and concerned operational costs and renewal of the society's logo. In 2018, T21RS changed its payment module connected to the website: we moved from Multisafepay to the user-friendly payment operator Mollie.

3. Revenues

T21RS main revenues consist of membership fees and sponsoring. We distinguish two types of membership: (1) full membership for researchers and clinicians, and (2) associate membership for DS associations/foundations. A 50% reduced membership fee applies to individuals living and working in countries with low-, low-middle and upper-middle income economies (as defined by the Worldbank).

<u>Full membership for researchers and clinicians</u>

- Master/PhD student € 40,-
- Postdoctoral fellows € 80,-
- Academic staff members / clinicians € 100,-

<u>Association</u>

- Associate member € 50,-

After registering on www.T21RS.org, new members automatically proceed to a secure payment module. In 2018 we choose for incorporation of a new payment module, operated by the international payment operator Mollie.

Sponsors

T21RS is very grateful to five non-profit organizations that support the establishment of the society and its aims. These Founding Sponsors have committed themselves to 5 years of support at a level of € 5000,- per year:

- Lumind Research Down Syndrome Foundation (USA)
- Fondation Jérome Lejeune (France)
- The Matthews Foundation (USA)
- Global Down Syndrome (USA)
- Trisomie 21 France (France)

Conference sponsoring

The organization of the T21RS International Conference 2019 Barcelona has started in 2018. Although most sponsorship will be received in 2019, we already gratefully thank two organizations for their generous sponsoring:

- Fundación Iberoamericana Down21
- The Company of Biologists

4. Expenses

In this quiet off-year in-between the Chicago conference (2017) and Barcelona conference (2019), expenses were reduced to a minimum. Expenses related to general ongoing expenses (running costs) for update/maintenance of our website and banking fees. At the beginning of 2018, the society's logo was renewed.

5. Profit and loss statement

Profit and Loss Statement

For the period from 01/01/2018 to 31/12/2018

Accrual basis

	31/12/2018
Income	
Interest	**7,04**
Membership fees	**16 051,40**
Sponsoring (conference)	**5 285,80**
Sponsoring (general)	**25 000,00**
Total - Income	**46 344,24**
Less: Expenses	
Operational costs	**2 946,99**
Net profit (loss)	**43 397,25**

6. Balance

Balance Sheet

As at 31/12/2018
Accrual basis

	31/12/2018
Assets	
Cash & cash equivalent	**161.139,65**
Net assets	**161.139,65**
Equity	
Retained earnings	**43.397,25**
Starting balance equity 01/01/2018	**117.742,40**
Total - Equity	**161.139,65**
Total equity	**161.139,65**

7. Conclusion

The year 2018 has been closed with a net profit of € 43.397,25 resulting in a positive balance of € 161.139,65.