2017

# T21RS Science & Society Symposium

# Bulletin

## T21RS Science & Society Symposium – Saturday June 10th 2017

**Sponsored by the National Down Syndrome Society (NDSS)**

*Location:* Feinberg Conference Center, Feinberg Pavilion, Northwestern Memorial Hospital at 251 E. Huron Street, Chicago, Illinois 60611, USA

Session coordination: T21RS Committee for Science & Society
Session chair: Peter De Deyn

**T21RS Committee for Science & Society**
Peter Paul De Deyn (Belgium, chairman), Alain Dekker (NL),
Juan Fortea (SP), Sebastian Videla (SP), Lotta Granholm (US, SW),
Cindy Lemere (USA), Diana Bianchi (USA)

*Overview of Down syndrome associations presenting at the T21RS Science & Society Symposium during the T21RS International Conference in Chicago*

t21
RESEARCH
SOCIETY

# Program

**17:00 Welcome and Introduction**

*Peter De Deyn (chairman T21RS Committee for Science & Society)*
*Kandi Pickard (Chief of Staff, NDSS)*

**T21RS Committee for Science & Society: aims & achievements**

*Peter De Deyn*

**17:15 The pros and cons of having my relative with Down syndrome participate in clinical research**

*Sub-session chair: Diana Bianchi*

Two families are invited for a debate: one family in favor of, and one family against participating in scientific research + discussion with panel and plenary audience

**17:45 Medical policies for people with Down syndrome**

*Sub-session chairs: Juan Fortea and Cindy Lemere*

Four initiatives to integrate care and research with social aspects for patients/clients and family members + discussion with panel and plenary audience

- Sebastian Videla (Catalan DS Foundation)
- Melissa Parisi (NIH/NICHD)
- Michelle Whitten (Global Down Syndrome):
  *Medical care for adults with Down syndrome - lifting barriers*
- Kandi Pickard (NDSS)
  *ABLE Act – Progress Report*

**18:40 Break**

**18:55 Association introduction round: DS associations as research partners**

Each DS association will briefly introduce themselves (3 minutes) by focusing on how they contribute to research, with the aim to get acquainted, share ideas and facilitate discussion.

1. National Down Syndrome Society (USA), *Kandi Pickard*
2. Global Down Syndrome (USA), *Michelle Whitten*
3. Fondation Jérôme Lejeune (France), *Catherine Lemmonier*
4. LumindRDS (USA), *Hampus Hillerstrom*
5. The Matthew Foundation (USA), *John Blascovich*
6. Trisomie 21 France (France), *Renaud Touraine*
7. Down's Syndrome Association (UK), *Gillian Bird*
8. Band of Angels (USA)
9. Alana (Brazil), *Claudia Moreira*
10. Catalan Down Syndrome Foundation (Spain), *Bessy Benejam*
11. Down Syndrome International (UK/international), *Helen Powell*
12. Association Française pour la Recherche sur la Trisomie 21 (France), *Jean Marc Richard*
13. Down Syndrome Hungary (Hungary), *Agnes Toth*
14. AMIPI-Bernard Vendre (France), *Marie-Laure Blandin*
15. Down Syndrome OPTIONs (USA), *Alexandria Durkin*

**20:05 Summary** *Peter De Deyn*
**20:10 End**

# National Down Syndrome Society (USA)

*Speaker: Kandi Pickard, Chief of Staff*

The National Down Syndrome Society (NDSS) was founded in 1979 as an official nonprofit organization for all individuals with Down syndrome. Throughout the years, the NDSS has supported Down syndrome research and worked on increasing general public awareness and acceptance of people with Down Syndrome. In the 1980s, the NDSS awarded annual research grants (via their Science Scholar Award Program) to excelling post-doctoral scientists interested in gaining a better understanding for Down syndrome. Additionally, two successful projects were launched, namely Project Mainstream and Project Child. The NDSS introduced "The Buddy Walk" in 1995, and to date there have been over 250 walks. A major research project with the National Institute for Child Health & Human Development (NICHD) and National Institute for Neurological Disorders and Stroke (NINDS) was launched in the 1990s, with six grants awarded to fund research on development and treatment for individuals with Down syndrome. In the 2000s, the NDSS set up its National Policy Center in order to work with members of Congress and federal agencies to develop and improve laws for people with DS. In 2014, the Achieving a Better Life Experience (ABLE) Act was passed, aiding individuals with DS to live better lives all round. The NDSS envisions a world where people with Down syndrome have the opportunity to enhance their quality of live, realize their life aspirations, and become valued into welcoming communities.

***Key research links***: www.ndss.org/Resources/Research/Research-Down-Syndrome/

national down syndrome society
ndss

*More information?*
*Go to www.ndss.org*

# Global Down Syndrome Foundation (USA)

*Speaker: Michelle Sie Whitten, President & CEO*

Established in 2009, Global Down Syndrome Foundation is a public nonprofit organization dedicated to significantly improving the lives of people with Down syndrome through research, medical care, education and advocacy. Global supports two affiliates which together constitute the only academic home in the United States committed solely to research and medical care for people with Down syndrome – the Crnic Institute for Down Syndrome and the Sie Center for Down Syndrome.

The Linda Crnic Institute for Down Syndrome is the first medical and research institute with the mission to provide the best clinical care to people with Down syndrome, and to eradicate the medical and cognitive ill effects associated with the condition. Established in 2008, the Crnic Institute consists of over 100 researchers working in 33 labs and is a partnership between the University of Colorado School of Medicine, the University of Colorado Boulder, and Children's Hospital Colorado. Headquartered on the Anschutz Medical Campus in Denver, the Crnic Institute includes the Anna and John J. Sie Center for Down Syndrome at Children's Hospital Colorado. It partners both locally and globally to provide life-changing research and medical to improve the lives of people with Down syndrome. The Crnic Human Trisome ProjectTM is a discovery accelerator for treatments or even cures for diseases like Alzheimer's and Leukemia that are more prevalent in people with Down syndrome. The Crnic Institute is made possible by the generous support of the Anna and John J. Sie Foundation, and relies on the Global Down Syndrome Foundation for fundraising, education, awareness, and government advocacy. It is a research and medical-based organization without political or religious affiliation or intention.

***Key research links:*** www.globaldownsyndrome.org/research-medical-care/

GLOBAL
DOWN SYNDROME
FOUNDATION®

*More information?*
*Go to* www.globaldownsyndrome.org

# Fondation Jérôme Lejeune (France)

*Speaker: Catherine Lemonnier, MD & Research Director*

Professor Jérôme Lejeune (1926-1994), discovered in 1959, the cause of Down syndrome (Lejeune/Gautier/Turpin). Subsequently, the Jérôme Lejeune Foundation, given public interest status in 1996, has been working for individuals affected by genetic intelligence disorders (Down syndrome, Williams-Beuren, Fragile X, “Cri du chat”, and other unexplained intellectual disability). At the service of affected individuals and their families, the Jérôme Lejeune Foundation is driven by three objectives: Research, Care, and Advocacy. It implements and supports research programs to develop treatments for the above described diseases of genetic origin, under the supervision of the Scientific Board (25 international experts in Fundamental, Translational and Clinical Research). The Jérôme Lejeune Foundation develops and funds programs in France and abroad thanks to a Research Funds allocation of 3,5 to 4 M€ per year (all funds are private). It initiates (as major examples) Projects on Protein to Protein Interactions, on animal models -new Rat models- (with the Bettencourt-Schueller Foundation), on Dyrk1a and CBS genes located on Chromosome 21, Clinical trials (Tesdad), and allocates Prizes (Young Searcher Awards and Sisley d’Ornano-J.Lejeune Postdoctoral Award). It recently focuses its Research Strategy on cross pathologies (DS -Autism, -Alzheimer and -Oncology). As the Jérôme Lejeune Foundation defends Life, it is highly vigilant about Bioethical issues: in a context in which ever-accelerating scientific progress and ideological pressure pose fundamental questions for society, the Foundation provides scientific expertise together with its Ethical Values.

***Key research links***: www.institutlejeune.org
Created in 1997 and funded mainly by the Jérôme Lejeune Foundation, the Jérôme Lejeune Institute is a clinical center for specialized medical and paramedical consultations, operating around three goals: Care, Research, Training. The Institute is dedicated to Down syndrome patients and to patients with other intellectual disabilities from genetic origins, and to their families. The Jérôme Lejeune Institute proposes a consultation with a referring physician (geneticist, pediatrician, neuro-pediatrician or geriatrician), supplemented if necessary by a paramedical consultation (neuropsychologist, speech therapist, social worker, psychologist...), blood sampling or consultation.

FOUNDATION
Jérôme Lejeune
search, healing, defend

*More information?*
*Go to* www.fondationlejeune.org

# LuMind RDS (USA)

*Speaker: Hampus Hillerstrom, Vice Chair*

The vision of the LuMind™ Research Down Syndrome Foundation (LuMind RDS) is to be the global leader in funding a comprehensive portfolio of research to meaningfully improve memory, cognition, and independence in individuals with Down syndrome. LuMind RDS' sole focus is to discover groundbreaking Down syndrome research in four scientific categories: 1) Prevent Alzheimer onset; 2) Improve cognition; 3) Develop gene therapies; 4) Advance understanding. Since 2004, LuMind RDS has been the largest source of private funding for Ds cognitive research—granting more than $15M to leading academic institutions and biopharmaceutical companies, nationally and internationally and leading to millions more in concurrent funding from the National Institute of Health, industry and other sources. LuMind RDS cares because they believe in Cognition And Research Empowers Success (CARES).

***Key research links:*** www.lumindrds.org/research/active-research-projects/

*(learn more about LuMind RDS' work in down syndrome research)*

: www.lumindrds.org/get-involved/

*(learn how you can support Down syndrome research)*

*More information?*
*Go to* www.lumindrds.org

# The Matthew Foundation (USA)

*Speaker: John Blascovich, President & Chair*

The Matthew Foundation’s mission is to improve the quality of life for people with Down syndrome throughout their lifetime. The areas of focus are supporting research and care, promoting inclusive practices, and developing employment programs because the foundation appreciates that different needs are present during different periods in a person’s life. The Matthew Foundation makes deep impact through specific programs. These are carefully selected to ensure that they can either provide very meaningful impact across a broad population base or the impact is multi-layered. And the foundation only takes on programs where the team has unique expertise and can be successful given its relationships and capabilities. Examples include: the establishment of the first and currently only endowed fund for Down syndrome research at Stanford University and Super Fest, an inclusive football and cheerleading clinic the day before the Super Bowl where people with and without developmental disabilities can participate together. Super Fest has gained national US media coverage from ESPN, support from NFL executives and alumni including a head coach, general manager, team president, entire cheerleading teams, and political leaders such as President George H. W. Bush.

***Key research links:*** http://dsresearch.stanford.edu/

The Matthew Foundation

*More information?*
*Go to* www.thematthewfoundation.org

# Trisomie 21 France (France)

*Speaker: Renaud Touraine, Vice President*

**Main goals**

- To support people with Down Syndrome and their families and to make the social participation of people with Down-Syndrome effective, through mainstream schooling, independent living, work inclusion.
- To favour the exercise of full citizenship, self-advocacy, and peer-advocacy for people with Down Syndrome.
- To contribute to scientific research in order to improve the well-being and the quality of life of people with Down-Syndrome (T21 France has a scientific committee)

Trisomie 21 France offers counselling, locally for the families and nationally for the associations, as well as rehabilitation (speech therapy, physiotherapy, psychomotricity, medical, psychological and educational care). T21 France has a partnership with some French universities (Saint-Etienne, Bordeaux…) and organizes annual seminars. They also organize seminars for people with Down Syndrome and develops accessible material for people with intellectual disability.

T21 France publishes a regular journal “Trisomie 21”, brochures about schooling and medical care; the local associations publish a variety of information flyers. They also offer several informative videos (about work inclusion, independent living, health…) and short films and videos made by people with Down Syndrome. One Sunday in November is dedicated to Down-Syndrome awareness, throughout France. Every 18 months a symposium or a “summer academy” is organized by T21 France.

***Key research links:*** www.trisomie21-france.org/presse-publications

trisomie 21 FRANCE

*More information?*
*Go to www.trisomie21-france.org*

# Down’s Syndrome Association UK (UK)

*Speaker: Gillian Bird, Information & Training Services Manager*

The DSA is the only charity in England, Wales and Northern Ireland, focusing solely on all aspects of living successfully with Down’s syndrome. Our aim is to help people with Down’s syndrome live full and rewarding lives. Established in 1970, the DSA has evolved from a parent member organisation into a leading national charity. We provide services to everyone regardless of where they live; we have regional offices in England, Wales and Northern Ireland as well as a network of affiliated support groups across the country. Our services include an information helpline, information resources, training for parents and professionals covering preparation for parenthood, maternity services, education and development, puberty and adolescence, ageing and dementia. We also provide developmental assessments, assessments for social care provision and school consultations to foster better practice. In addition we host and facilitate advocacy groups, and offer a sports programme and an employment programme. We participate in a number of alliances to bring about change in education, health and social care practices, work with civil servants and politicians and engage with research projects relevant to Down’s syndrome.

***Key research links***: www.downs-syndrome.org.uk/about/research-campaigns/current-research-projects/

Down’s Syndrome
Association
A Registered Charity No. 1061474

*More information?*
*Go to* www.downs-syndrome.org.uk

# Alana (Brazil)

*Speaker: Claudia Moreira, Scientific Advisor*

The Alana Institute is a nonprofit civil society organization that brings together programs that pursue conditions for children to fully experience their childhoods. Founded in 1994, Alana has been maintained by an endowment fund since 2013. Its mission is to “honor the children”. The main location of the Alana Institute is São Paulo, Brazil.

The Alana Foundation, headquartered in the United States, is a fully philanthropic foundation created in order to invest in the cutting-edge and innovative research capable of changing the world. The Foundation is dedicated to promote research and enhance the community’s knowledge of child development, in general, and Down syndrome, in particular. Currently, Alana has partnership with Case Western Reserve University (CWRU), Massachusetts Institute of Technology (MIT) and Albert Einstein Education and Research Israeli Institute (Brazil). There are six years of research planned to increase the chances of developing new therapies for people with Down syndrome. The partnership brings together Dr Alberto Costa's (CWRU) 25 years of experience in Down syndrome research and Dr Li-Huei Tsai's (MIT) expertise in Alzheimer's research in order to achieve Alana’s objectives.

***Key research links***: *(sites of main researchers)*

http://casemed.case.edu/pediatrics/people/faculty-profile.cfm?people_id=179 (Dr Alberto Costa, CWRU)
www.tsailaboratory.mit.edu/research/ (Dr Li-Huey Tsai, MIT)

*More information?*
*Go to* www.alana.org.br/en

# Catalan Down Syndrome Foundation (Spain)

*Speakers: Bessy Benejam, Neuropsychologist*
*& Sebastian Videla, Head of Clinical Research*

The FCSD was born out of a common interest, shared by parents and professionals alike, to gain a better understanding of Down Syndrome (DS) by carrying out research and doing practical work in the fields of education, psychology, medicine and welfare. From its very beginning, outstanding international scientists specialized in subjects relating to DS have given the FCSD their valuable support and on some occasions even their personal participation, if necessary. The FCSD technical team is made up of a group of professionals specialized in psychology, education, and medicine and a group of scientific advisors. Due to its distinct pioneering spirit, the FCSD is regarded as a reference point and therefore, consulted by a large number of public and private organizations, both inside and outside Spain. The spirit which the FCSD wishes to convey is that people with DS have inherent capacities and are entitled to receive adequate resources to meet their individual needs so as to optimize their development.

***Key research links:***

- Cerebrospinal fluid biomarkers for Alzheimer's disease in Down syndrome. Dekker AD, Fortea J, et al. Alzheimers Dement 2017
- Cerebral Amyloid angiopathy in Down syndrome and sporadic and autosomal-dominant Alzheimer's disease. Carmona-Iragui M, Balasa M, et al. Alzheimers Dement 2017.
- Feasibility of lumbar puncture in the study of cerebrospinal fluid biomarkers for Alzheimer's disease in subjects with Down syndrome. Carmona-Iragui M, Santos T, Videla S, et al. J Alzheimers Dis 2017.
- Periodontal disease's contribution to Alzheimer's progression in Down syndrome. Kamer AR, Fortea JO, et al. Alzheimers Dement 2016.
- Safety and efficacy of cognitive training plus epigallocatechin-3-gallate in young adults with Down's syndrome (TESDAD): a double-blind, randomised, placebo-controlled, phase 2 trial. de la Torre R, de Sola S, et al.; TESDAD study group. Lancet Neurol. 2016.
- Patterns of Performance on the Modified Cued Recall Test in Spanish Adults With Down Syndrome With and Without Dementia. Benejam B, Fortea J, Molina-López R, Videla S. Am J Intellect Dev Disabil. 2015.
- A new cognitive evaluation battery for Down syndrome and its relevance for clinical trials. de Sola S, de la Torre R, et al.; TESDAD Study Group. Front Psychol. 2015.
- Epigallocatechin-3-gallate, a DYRK1A inhibitor, rescues cognitive deficits in Down syndrome mouse models and in humans. De la Torre R, De Sola S, et al. Mol Nutr Food Res. 2014.

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SÍNDROME DE DOWN

*More information?*
*Go to* www.fcsd.org/en

# Down Syndrome International (UK)

*Speaker: Helen Powell, Policy Development Manager*

Down Syndrome International (DSi) is a global network of member organisations and individuals from 136 countries across the world. DSi is involved in the dissemination of high quality research to its members, and to anyone in the international community seeking related information, in a clear, factual, understandable, sensitive and unbiased way. The DSi publishes selected research abstracts on their website quarterly. Additionally, the DSi is involved in the development of international consensus guidelines in healthcare, and in other areas that affect the lives of people with Down syndrome, with input from expert advisors and stakeholders. The DSi has worked on promoting T21RS by having a permanent link on their homepage of their website, and by featuring the T21RS Science & Society bulletins in their newsletter.

***Key research links:*** www.ds-int.org/research

*More information?*
*Go to* www.ds-int.org

# Association Française pour la Recherche sur la Trisomie 21 (France)

*Speaker: Jean-Marc Richard, Vice President*

AFRT, the French Association for Research on Trisomy 21, was created in 1990. In the early 90's, the idea was that Down syndrome was almost impossible to understand, as due to a whole chromosome. AFRT was among the ones trying to reverse that idea, to give some hope to the families. Even if T21 is complex and affects each person differently, it is only through research that one can make progress in studying this syndrome and find basic and pharmacological pathways to interfere with the deficits. This view is now more widely accepted, and recent results are encouraging. Since 1995, AFRT informs its members about recent and ongoing research through a publication: "News from Chromosome 21". In 2005, AFRT initiated the choice of March 21st as an emblematic day for T21, and decided to hold a workshop every March 21st. The meeting of March 2005 took place in Paris, with the participation of FAIT21 and Fondation Jérôme Lejeune. In June 2005, at an international meeting in Palma de Majorca (attended by some board members of DSi and EDSA), the president of AFRT proposed March 21st as the World Down Syndrome Day, which was later approved by WHO in 2007 and UN in 2011. Since 2005, the AFRT holds an annual meeting in France around March 21st. The 2016 meeting was in Marseilles and the 2017 one in Grenoble, each gathering about 200 participants. On the AFRT website, information on medical, scientific and societal topics is available for persons with T21, their families, and people involved in creating a better life for them. Besides collecting and spreading information about T21, AFRT promotes biomedical research through fellowships for students and grants for research teams. In recent years, support was mainly focused on the detection of sleep apnea, aging aspects of ocular problems, and nervous and mental breakdown.

***Key research links:*** www.afrt.fr/recherche.html

A.F.R.T.

*More information?*
*Go to* www.afrt.fr

# Down Syndrome Hungary (Hungary)

*Speakers: Agnes Toth, Medical Advisor*

The Hungarian Down Syndrome Association was founded in 1998 in order to support people with Down syndrome and other types of intellectual disabilities so that they can live meaningful, active and happy lives, fully integrated in society. They were the first to organize an event on the World Down Syndrome Day in Hungary in 2009. Their sensitivity program *0321 Challenge, a game without labels*, models the difficulties people with Down Syndrome may face, demonstrates how these can be overcome by proper support, and makes team members realize that achievements require cooperation. Their inclusive educational program *School4All,* allows students of special schools to take part in lessons of mainstream schools. The project is highly successful in demonstrating that children with intellectual disabilities can be integrated in a mainstream setting, and their presence may be beneficial for all the children and teachers involved. The Hungarian Down Syndrome Association will shortly open their second Castle of Miracles (www.csodavarfc.hu), an early intervention centre and integrated playhouse, where children with special needs not only receive therapy, but play together with typically developing children. Experiences like these lay the foundation of a more accepting and more tolerant society. Additionally, they operate an online informational center that contains up-to-date and balanced information for expectant parents, and the only one that provides support in case of a positive pre- or postnatal diagnosis. The Hungarian Down Syndrome Association regularly takes part at the national Down symposium and other professional forums to raise awareness about Down syndrome research among medical professionals. They are willing to promote scientific achievements and recruit volunteers for clinical research through its online platforms and live performances.

***Key research links:*** www.downegyesulet.hu/olvasosarok/tudastar
***:*** www.DownBaba.hu

*More information?*
*Go to* www.downegyesulet.hu

# AMIPI-Bernard Vendre (France)

*Speaker: Marie-Laure Blandin, Director in charge of learning*

"Learning Factories" operator, the AMIPI Foundation brings the conviction that cognitive deficiency can be reduced thanks to specific learnings through industrial activity. Researches have confirmed that learning through "the repetition of cognitive tasks" favors the development of the neuronal system, reinforcing and creating new synapses.

Since 1985, the activity of cable harness assembly was identified as the right mean for learning for persons with cognitive disorders. It allows them to make progress on their cognitive abilities such as manual movements, complex coordination and visual memory.

Today, the AMIPI Foundation employs 600 disabled workers in 7 factories, based in the regions "Pays de Loire" and "Centre – Val de Loire". In these factories, the operators develop their cognitive skills and professional behaviors. Then they prepare a personal and professional project towards individual integration at regular employers.

In the field of cognitive disorders, AMIPI Foundation demonstrates the absolute needs of behavior therapy through: 1/ usual traditional manual job, 2/ factory with learning methodology, and 3/ objectives of new job out of the Foundation

**Key figures:**

- Founded in 1965
- Annual turn over: 28 M€
- Production: 6,5 M electric cables and sticks, to high quality standards such as ISO9001 ans ISO16949
- 75 insertions since 2009
- More than 1500 individual integrations at regular employers since 50 years.

***Key research links:*** www.fondation-amipi-bernard-vendre.org/la-fondation/comite-scientifique/

FONDATION AMIPI
Bernard Vendre

*More information?*
*Go to* www.fondation-amipi-bernard-vendre.org

# Down Syndrome OPTIONs (USA)

*Speaker: Alexandria Durkin, President & Executive Director*

Down Syndrome OPTIONs' aims to increase awareness and educate parents and doctors on options for individuals with Down syndrome to help improve speech and learning as well as physical, emotional, social and intellectual growth and development. They aim to facilitate collaboration and networking for distribution of accurate information about integrative options for individuals with Down syndrome to families, government and professional agencies and nonprofit organizations worldwide. To fulfill their mission, they hold annual conferences for researchers, physicians and therapists to present cutting edge translational medical research and interventions to improve the health, cognition, behavior and development of individuals with Down syndrome.

***Key research links:***

- At one of their DS conferences, they recruited over 200% more participants for Dr. Starbuck's craniofacial development study than their original study
- Partnered with Faycal Guedj, Mother Infant Research Institute, to help him recruit prenatal moms
- Continue to help researcher, Barbara Strupp, reach the ever changing prenatal population
- Helping retired endocrinologist, Dr. Richard Levy, gather patient records and to disseminate, gather and record questionnaires on parent testimony about the effects of growth hormone treatment on their children. They will be eagerly broadcasting his results on their upcoming webinar.
- Their conferences provide a platform for researchers to share their information to parents and practitioners
- Regularly share requirement information to our members such as Dr. Feldman and Dr. Lee at UMass Memorial Children's Medical Center and their thyroid study.
- Provide pertinent research links for parents to share with their physicians.

Down Syndrome
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*More information?*
*Go to [www.downsyndromeoptions.org](http://www.downsyndromeoptions.org)*

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The Committee of Science & Society thanks all Down syndrome associations for their participation and contribution to the symposium.

For more information, visit our website at
http://www.t21rs.org/science-society