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**NEUROLOGY, EPISTOP &  
TAND (TSC-Associated Neuro-Psychiatric Disorders)**  
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Friday 19 October, 2018  
&  
Saturday 20 October, 2018

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### **Remembering Loren Leclezio PhD (1981-2018)**

Loren was a research developmental neuropsychologist who made significant contributions to the study of TAND, and in particular, validation of the TAND Checklist and generation of natural TAND clusters. She worked in the corporate health communications sector before completing a BA in Psychology (cum laude) and an honors in neuropsychology (cum laude) at the University of South Africa. She then worked with Prof Petrus de Vries on an MSc(Med) in Neuroscience at the University of Cape Town, and focused on development and validation of the TAND Checklist. She progressed to a PhD with Prof de Vries on identification of natural TAND clusters.

In her short academic career, Loren published 7 peer-reviewed papers, set up a South African TSC support group, and represented South Africa on TSCi (Tuberous Sclerosis International). She was a passionate champion for community-based and socially-responsive participatory research. Loren died in January 2018 after a short battle with cancer, but her legacy will live on in the TAND work that will follow from her fundamental contributions.

# Program

Friday, October 19

Venue: Convent of Chièvres, Herestraat 49, 3000 Leuven

Morning Sessions

9h30 – 10h20	<p><b>1. TSC and the brain</b></p> <ul style="list-style-type: none"><li>• <u>General introduction - update on clinical recommendations for TSC epilepsy</u> Prof. Liesbeth De Waele, University Hospital Leuven, Belgium <i>APOLOGIES, due to a technical defect we can only offer the slides of this presentation.</i></li><li>• <u>Latest data on SEGA from TOSCA</u> Prof. Anna Jansen, University Hospital Brussels, Belgium</li><li>• Epilepsy surgery <i>APOLOGIES, NOT AVAILABLE DUE OF TECHNICAL DEFECT</i> Dr. Vaz, UCL/Saint-Luc, Belgium</li><li>• Q&amp;A</li></ul>
10h20 – 11h10	<p><b>2. EPISTOP</b></p> <ul style="list-style-type: none"><li>• <u>EPISTOP: general overview</u> Prof. Sergiusz Józwiak, Medical University of Warsaw, Children's Memorial Health Institute, Warsaw, Poland</li><li>• <u>EPISTOP: EEG results</u> Prof. Lieven Lagae, University Hospital Leuven, Belgium</li><li>• Q&amp;A</li></ul>
11h10 – 11h30	BREAK
11h30 – 12h40	<p><b>3. TSC Associated Neuro-Psychiatric Disorders - TAND</b> organized around the natural TAND clusters, identified in Loren Leclizio's investigations</p> <ul style="list-style-type: none"><li>• <u>General introduction on TAND, TAND-checklist, TAND-clusters</u> Prof. Petrus de Vries, University of Cape Town, South Africa</li><li>• <b>Overactive/impulsive cluster &amp; neuropsychological cluster</b> Prof. Petrus de Vries, University of Cape Town, South Africa</li><li>• <u>Behavioral Dysregulation cluster</u> Lucy Wilde, research fellow, Cerebra Centre for Neurodevelopmental Disorders, University of Birmingham, UK</li><li>• Q&amp;A</li></ul>
12h40 – 13h40	LUNCH BREAK

# Program

Friday, October 19

Venue: Convent of Chièvres, Herestraat 49, 3000 Leuven<sup>m</sup>

## Afternoon Sessions

13h40 – 15h10	<ul style="list-style-type: none"><li>• <u>ASD-like cluster (focusing primarily on paediatric care)</u> Prof. Ilse Noens, University of Leuven, Belgium</li><li>• <u>Mood/anxiety cluster (primarily focussing on adult care)</u> Dr. Liesbeth Santermans/Dr. Nathalie Vanderbruggen, adult Psychiatrists TSC Team in Brussels, Belgium</li><li>• <u>Scholastic cluster</u> Prof. Annick Vogels, paediatric psychiatrist, University of Leuven, Belgium</li><li>• <u>Eat/sleep cluster</u> Prof. Anna Jansen, University Hospital Brussels, Belgium</li><li>• Q&amp;A</li></ul>
15h10 – 15h25	BREAK
15h25 – 16h40	<ul style="list-style-type: none"><li>• <u>From adolescent to adult care</u> Dr. Agnies van Eeghen, Expertisecentre ENCORE, Erasmus Medical Centre, Rotterdam, the Netherlands</li><li>• <u>RAPIT research results: the effect of Everolimus on children with learning and behavioural disabilities</u> Iris Overwater, research fellow, Department of Neuroscience, Erasmus MC, Rotterdam, the Netherlands</li><li>• <u>BATSCH-study (Bumetanide to Ameliorate TSC-Associated Hyperexcitable Behaviours)</u> Dorinde van Andel, research fellow, UMC Utrecht, the Netherlands</li><li>• Q&amp;A</li></ul>

# Program

**Saturday, October 20**

**Day 2** offered an extension to the DEA meeting with a closer focus on quality of life and case studies illustrating all possible manifestations of TAND. In addition to the professional public of day 1, day 2 was open to patients, parents, carers and professionals in family support, etc.

**Venue: the University Hospital of Leuven  
Administrative Building Bruckner, Herestraat 49, 3000 Leuven**

9h00 – 9h30	REGISTRATION AND COFFEE
9h30 – 11h00	<ul style="list-style-type: none"><li>• <b>Patient and parent perspective:</b> narrative resulting from in depth interviews during The Mind the Gap project by The King Baudouin Foundation Annemie T'Seyen, King Baudouin Foundation</li><li>• <b>TSC and quality of life :</b> results from the TOSCA register + results from 2016 survey by ASTB <u>Prof. Anna Jansen, University Hospital Brussels, Belgium + Eva Schoeters, be-TSC vzw</u></li><li>• <b><u>TAND (TSC Associated Neuro-Psychiatric Disorders): setting the scene: why and what?</u></b> Prof. Petrus de Vries, University of Cape Town, South Africa</li><li>• <b>Support for families living with TSC:</b> making use of the 'competence model for balance between skills and vulnerabilities Expertise team TSC &amp; Behaviour (Leonore Kuijpers, physician - Richard Slinger, system coach - Marije Offringa, psychologist)</li><li>• <b>Q&amp;A</b></li></ul>
11h00 – 11h15	BREAK

11h15 – 12h45

**Panel discussion of case studies** illustrating the various clusters in TAND  
Issues when investigating each case:

- Identifying the problem  
methodology (hospital setting vs home setting, questionnaires, observations, ...), expertise required, multidisciplinary approach...
- Best practices in addressing the problem:  
methodology, toolkits, family support, ...
- Expected outcomes and long term management

*The case studies illustrated the diverse clusters and age groups.*

*The case study discussion was spread over the morning and afternoon session.*

*These sessions were not recorded. Apologies.*

12h45 – 13h45

LUNCH

### **Biographies of all speakers (in order of appearance)**

**Liesbeth De Waele** is Assistant Professor at the Faculty of Medicine at the University of Leuven (KUL, campus Kulak in Kortrijk), and Deputy Head of Clinics in the Department of Paediatric Neurology of the University Hospitals Leuven, Belgium.

Liesbeth De Waele coordinates the TSC-clinic at UZ Leuven. She has participated as principal investigator in clinical trials with mTOR-inhibitors for the treatment of SEGA and TSC-related epilepsy. She is a principal investigator for the TOSCA registry and has contributed to the Dutch translation of the TAND-checklist.

With her TSC-colleagues in Leuven, she published a review on TSC. Together with the colleagues of the TSC-teams at UZ Leuven and UZ Brussel, she co-authored a retrospective cohort-study on the renal progression factors in young patients with TSC. Her current TSC research focusses on ambulant blood pressure measurement in patients with TSC, and metabolomics in TSC.

In addition, Liesbeth De Waele is medical advisor for be-TSC and she has contributed to the Multistakeholder Dialogue for Priority Setting in TSC-research in collaboration with the King Baudouin Foundation.

### **Anna C. Jansen, PD, PhD, pediatric neurologist, University Hospital Brussels, Brussels, Belgium.**

Professor Anna Jansen is a pediatric neurologist at UZ Brussels and is affiliated with the Neurogenetics Research Group at Vrije Universiteit Brussel. Together with dr. Peter Janssens, nephrologist, she runs a multidisciplinary TSC clinic for both children and adults. She has participated as principal investigator in clinical trials in the field of epilepsy and TSC. Dr. Jansen is member of the EPISTOP Steering Committee and is responsible for the dissemination of the project results ([www.epistop.eu](http://www.epistop.eu)). She is vice-chair of the TOSCA Scientific Advisory Board and coordinates the TOSCA SEGA research project. Together with the King Baudouin Foundation she initiated a multi-stakeholder dialogue for priority setting in TSC research.

**Geraldo Vaz** is Chef de Clinique in neurosurgery department of Cliniques Universitaires Saint-Luc (UCL). He is awarded a diploma in Biological Sciences and in Medicine. He made his training in several

countries (Brazil, Portugal, France and Switzerland). He works in the department of neurosurgery (Pr Raftopoulos) since the year 2000. Since 2004 he teaches the neurosurgery to the nurses of 4th of operating room, 4th anesthesia, to the pharmacists and He is the secretary of the jury and responsible for the organization of the examinations of access to the training in neurosurgery in the UCL. He has extensive experience in the field of the neurosciences and he wrote and participated in the writing of several papers published in scientific journals in neurosurgery. His activities include integrates several multidisciplinary groups (Center of Refractory Epilepsy, Neurovascular Center, Neuro-oncology Center, group of Spine and Chronicle Pain).

He is responsible for the project of study of monitoring of the intra-cranial pressure for the patients presenting signs of Normal pressure hydrocephalus (NPH), as well as of the program of Intraoperative magnetic resonance imaging of 3 Tesla (iMRI) for brain tumors.

**Sergiusz Jozwiak, MD, PhD, pediatric neurologist, Chair, Department of Neurology, Children's Memorial Health Institute Warsaw, Poland.**

Sergiusz Jozwiak is a professor and the head of the Department of Pediatric Neurology at the Warsaw Medical University. He received his medical degree from the Medical University Warsaw in 1983 and doctoral degree from the Children's Memorial Health Institute in 1990. Professor Jozwiak completed his habilitation at the same institute in 1995 and was appointed Head of Pediatric Neurology and Epileptology Department in 1997 in the same institution. He held his position till May 2015, when he moved to Warsaw Medical University. In the years 2009-2014 he served as a National Consultant in Pediatric Neurology. Professor Jozwiak's work focuses mainly on neurocutaneous disorders and epilepsy, especially infantile spasms. For more than 20 years, he has led a special program for TSC patients and worked out practical guidelines for TSC management. In 2009 professor Jozwiak received the prestigious Manuel Gomez Award established by TS Alliance for "creative or pioneering efforts that have appreciably improved either the understanding of the disease or the clinical care available for individuals with TSC". Currently he is coordinator of the large-scale European Commission Project EPISTOP evaluating clinical and molecular biomarkers of epileptogenesis in a genetic model of epilepsy-TSC ([www.epistop.eu](http://www.epistop.eu)). Professor Jozwiak is an active member of numerous international organizations and has published more than 300 papers in national and international peer reviewed journals. Professor Jozwiak is on the editorial boards of several professional journals as Pediatric Neurology (USA), European J. Paediatric Neurology (Amsterdam) and Journal of Child Neurology (USA).

**Lieven Lagae** is Full Professor at the University of Leuven, Belgium (KUL), Head of the Paediatric Neurology Department of the KUL University Hospitals, and Director of the Childhood Epilepsy Program at the KUL University Hospitals. Lieven Lagae is the immediate past President of the European Pediatric Neurology Society and serves as an elected Board Member of the International Child Neurology Association (ICNA). He chairs the Finance Committee of the ICNA board and the Taskforce on Medical Treatment of Childhood Epilepsy of the International League against Epilepsy (ILAE). From 2004 to 2015, he was the Editor-in-Chief of the European Journal of Paediatric Neurology. In 2017, he was the recipient of the 'Cures within Reach patient impact Clinical Award'. Current epilepsy research projects include: 1/ translational research in Zebrafish models of epilepsy; 2/ new anti-epileptic drugs in childhood epilepsy and especially in Dravet syndrome; 3/ brain stimulation in childhood epilepsy; 4/ preventive treatment of epilepsy in tuberous sclerosis complex. He published >200 papers in peer reviewed scientific journals and is the editor of the book: Cognition and Behaviour in Childhood epilepsy (Mac Keith Press 2017). He serves in many editorial boards of epilepsy and neurology journals.

**Petrus de Vries, MBChB, MRC-Psych, PhD, developmental neuropsychiatry, University of Capetown, Division of Child and Adolescent Psychiatry, Rondebosch, Cape Town, South Africa, South African representative of TSCi.**



Petrus de Vries is the Sue Strenghmann Professor of Child & Adolescent Psychiatry and Director of the Centre for Autism Research in Africa and the Adolescent Health Research Unit at the University of Cape Town. He trained in medicine at Stellenbosch University in South Africa before moving to the UK where he completed his clinical training in Psychiatry and Child & Adolescent Psychiatry and a PhD in Developmental Neuroscience at the University of Cambridge. Professor de Vries has a particular interest in TSC-Associated Neuropsychiatric Disorders (TAND) and led the team who developed the TAND checklist.

**Lucy Wilde** is a Research Fellow in the Cerebra Centre for Neurodevelopmental Disorders, School of Psychology, University Birmingham. Her work focusses on delineating social, emotional, cognitive and behavioural phenotypes of rare genetic disorders including tuberous sclerosis complex and Smith-Magenis syndrome.

**Ilse Noens** is an associate professor affiliated with the Parenting and Special Education Research Unit of the Faculty of Psychology and Educational Sciences at the University of Leuven (KU Leuven), specialised in autism spectrum disorder and family-based assessment and interventions. She is also head of PraxisP, the clinical expertise centre of the Faculty of Psychology and Educational Sciences, coordinator of Leuven Autism Research (LAuRes), an interdisciplinary consortium at the University of Leuven (KU Leuven), and member of the international Autism Ethics Network (AEN).

**Liesbeth Santermans, MD**, works as a psychiatrist in UZ Brussel, where she is responsible for the psychiatric outreachteam. She completed her master in medicine in 2002 at the KULeuven and ended her psychiatric training in 2007. Ever since she works at UZ Brussel, having experience in a broad range of psychiatric illnesses. She worked at the acute inpatient psychiatric service and has experience in liaison psychiatry and emergency psychiatry. She assists with the clinical teaching and supervision of psychiatrists in training. She participates in the expertise centre for adults with a developmental disorder (EVO) and is recently active as a member of the tuberosecleroseclinic at UZ Brussel.

**Nathalie Vanderbruggen, MD**, is a psychiatrist practising in UZ Brussel where she is responsible for the acute inpatient psychiatric service and assists with the clinical teaching and supervision of psychiatrists in training. She completed her master in medicine in 1994 at the Vrije universiteit Brussel and ended her psychiatric training in 1999. Dr. Vanderbruggen works ever since in UZ Brussel where she has years of experience across a variety of different functions. She operated for 8 years as a liaison and emergency psychiatrist, she is active as a member of the Klinefelter multidisciplinary group and recently as a member of the TSC multidisciplinary group at UZ Brussel. She started a private practice where she gains more than 15 years of experience as an eclectic psychotherapist. Enjoying the benefits of combining psychiatric and psychotherapeutic skills, she uses a blend of modalities which incorporates best practice of evidence based psychiatry, psychodynamic therapy, motivational interviewing, psycho-education and solution focused therapy. With this holistic approach she hopes that people can be challenged to experience change.

**Annick Vogels** obtained her Medical Degree at the Catholic University of Leuven. She was trained in clinical genetics in de Centre for Human Genetics at the University Hospital of Leuven. She was trained in child psychiatry at the University Hospital of Leuven and at the Royal Hospital for Sick Children in Bristol. In 2001, she completed her PhD on "Psychosis in Prader-Willi Syndrome" at the Catholic University of Leuven. She has been working in the Department of Clinical Genetics since August 1st, 1993.

Annick Vogels is a child psychiatrist with a subspecialisation in clinical genetics and has an outpatient clinic for children and adults with a genetic disorder and/or intellectual disability and psychiatric

problems.

Her main research fields are the genetic examinations in children and adults with a dual diagnosis of intellectual disability and psychiatric problems and behavioural and psychiatric problems in patients with intellectual disability and a genetic disorder.

Her main clinical work includes the genetic examination in persons with a dual diagnosis of intellectual disability and psychiatric problems and the multidisciplinary clinics for patients with amongst others Neurofibromatosis, Williams Syndrome, Prader-Willi Syndrome and Tuberous Sclerosis Complex.

**Agnies van Eeghen, MD, PhD**, works as an Intellectual Disability Physician at the Erasmus Medical Center in Rotterdam, as well as in the Amsterdam region. She is specialized in the neurobehavioral phenotypes of genetic syndromes such as Tuberous Sclerosis Complex (TSC) and Fragile X Syndrome (FXS).

After obtaining her medical degree, she followed the Intellectual Disability specialization at the Erasmus Medical Center in Rotterdam. Subsequently, she worked as a research fellow at Massachusetts General Hospital in Boston, where she obtained a PhD on the neuropsychiatric phenotype of Tuberous Sclerosis Complex. After moving back to the Netherlands, she rejoined the Erasmus Medical Center as coördinator of the adult expertise center of TSC.

Agnies focuses her clinic and research on manifestations and treatment of genetic neurodevelopmental disorders in teenagers and adults. She takes part in the international TAND expert panel, and is chairman of the guideline committee of the Dutch association for intellectual disability physicians.

**Iris Overwater** is a doctor of medicine, currently working in the department of neurology. She did extensive research on TSC as a PhD student. Her research focused on examining the mTOR inhibitors sirolimus and everolimus as treatment for epilepsy and intellectual disability in children with TSC. She also examined patient characteristics predicting intelligence in TSC patients and the correlation between lesions on brain MRI and genetic mutations. After her PhD, she remains part of the Encore TSC expertise center in the Erasmus MC-Sophia Children's Hospital in Rotterdam, among a team of neurologists, pediatricians, psychiatrists and many more, to provide the best care for patients with TSC, and to continue the search for more knowledge and better treatments for TSC patients.

#### **Dorinde van Anandel**

As a psychologist and PhD-student, Dorinde van Anandel (1991) is currently conducting research at the Sensory Processing Program (SPP) of the Brain Center Rudolf Magnus, UMC Utrecht. Within this program she focuses on sensory processing deficits over different neurodevelopmental disorders, including autism, ADHD, TSC and epilepsy. By integrating EEG measurements, cognitive tests, behavioral questionnaires and clinical observations the SPP provides an individually tailored Sensory Profile ("prikkelprofiel"), which is used to test novel treatments and to develop prognostic biomarkers. Besides coordinating two clinical trials, Dorinde is developing a patient-reported outcome measure to assess change in sensory reactivity over time. Ultimately, she aims to bring more objective measures into the clinic and combine these with subjective measures to improve quality of life for patients with a developmental disorder. Dorinde has a background in Psychology (Groningen University) and graduated with a Research Master degree in Cognitive and Clinical Neuroscience from Maastricht University (both cum laude). She spent a semester at SUNY Geneseo, New York studying cognitive psychology. In her free time, Dorinde enjoys going to indie-folk bands, travels the world, likes running, field hockey and cycling and tries to explore the nature as much as possible.

**Annemie T'Seyen**, joined the King Baudouin Foundation in 2010 after a career of 25 years as a

medical journalist. As a project manager in the Action Domain Health, she became responsible for the management of the Health Funds. At that time, they were 10 in total. Today, the King Baudouin Foundation is managing nearly 100 Funds in the domain of health, of which over 70 concentrate on research.

The research Funds support projects in very diverse fields such as cancer, ageing, cardiology, neurology, AIDS, rare diseases... The Funds are managed in such way that the donor's wishes are considered paramount and a tailor approach is developed. At the same time, the aim is to make the best possible use of the resources available and to create the greatest possible impact. With a constantly growing number of Funds, especially in the field of rare diseases, a global strategy for supporting health research and for innovative philanthropic tools are major challenges. In that context, Annemie T'Seyen was responsible for the pilot project: "Priority Setting in research by Multistakeholder dialogue- TSC".

**Eva Schoeters** is currently president of the Belgian patient association for Tuberous Sclerosis Complex and of RaDiOrg, Rare Diseases Belgium. However she was trained as an art historian and worked in the field of preservation of historical monuments for many years. She is currently on full-time leave from her normal daytime job.

After her son, Pepijn (now 10) received the diagnosis TSC in 2010, Eva and her husband Gert gradually became more and more involved in the TSC community. This resulted in the establishment of the Belgian TSC association in 2015.

Following from this grew Eva's commitment to advocacy for rare diseases in general. After working at the sidelines as a volunteer in the Belgian Rare Disease Organisation, RaDiOrg, she became president in March of this year. It has become a mission to contribute actively to creating greater awareness and better care for all living with a rare disease, whether it be TSC or another one of the 7000 defined rare diseases.

**The TSC and Behavioural Expertise Team (ETSCG)**, set up in 2003, the ETSCG is a team comprising a range of experts. It focuses specifically on providing and co-ordinating assistance to people with TSC and sharing knowledge on the subject with their nearest. To reflect the fact that medical issues, behavioural problems (including in some cases psychiatric ones) and practical problems all feature in the support provided, various disciplines are represented in the team.

The team members are:

- **Richard Slinger** has worked in care for people with mental disabilities since 1987, in a variety of job posts and for a range of target audiences. In 1996, as a 'Z-nurse' (a nurse trained to care for people with mental disabilities), he became involved in organising care for someone with TSC and mental health issues. Since 2000, he has worked for the Centre for Consultation and Expertise (CCE) as a consultant and case manager on the practical implementation of advice given.
- **Leonore Kuijpers - de Blicck**, a physician, has been working in care for people with mental disabilities since 1990. Since 2002, she has chiefly specialised in diagnostics and treatment of challenging behaviour and mental health issues. She is also a consultant for the Dutch Centre for Consultation and Expertise (CCE).
- **Marije Offringa - Lansbergen** is a psychologist. From 2008 to 2012, she was an academic lecturer and researcher at Erasmus Medical Centre, Rotterdam. From 2008 until present, she has been working in care for people with mental disabilities, largely specialising in challenging behaviour, particularly on serious cases. Since 2016, she has been a member of

the TSC and Behavioural Expertise Tam. She also works as a consultant at the Centre for Consultation and Expertise (CCE), providing advice in cases where challenging behaviour is a factor (elderly care, mental disability care, and children's and young people's psychiatry).

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This event was made possible by the generous contributions of all speakers, by the help of UZ Brussels and UZ Leuven and by the support of our sponsors. **However it would not have taken place without the loving care of the grandparents of Pepijn, aged 10, with Tuberous Sclerosis Complex.**