



RETT SYNDROME IN NORDIC LIGHT

STOCKHOLM 19-20 APRIL 2018

Nordic Rett Conference – Research and Practice

PRESENTERS

Katarina Öryd, Chairman of the Board for The Association for Rett Syndrome In Sweden, (RSIS).

Malin Rohdin, MD, PhD, is a paediatrician who works with children with breathing problems and lung disease as well as with respiratory support at Astrid Lindgren Children's Hospital, Stockholm, Sweden.

Ola Skjeldal, Professor in neurology at Oslo University, Norway.

Dr Jenny Downs is a paediatric physiotherapist and Co-Head of the Child Disability Group/Senior Research Fellow at the Telethon Kids Institute, Perth. She uses database methodologies for rare disorders, including Rett syndrome, to investigate natural history and treatment effectiveness. She also develops and validates robust outcome measures for gross motor function, physical activity and quality of life that can be used to monitor clinical progress and as an outcome measure in clinical trials.

Mari Wold Henriksen, MD, PhD student, Neurological Department, Drammen Hospital, Norway

Lotta Sjögren PhD is a language pathologist at Mun-H Center, a clinic that provides specialist dental care in Region Västra Götaland, Sweden. Her clinical work and research focuses on orofacial disorders in children, adolescents and adults with rare disorders.

Michelle Stahlhut, physiotherapist, PhD student, Center for Rett syndrome, Kennedy Center, Department of Paediatrics and Adolescent Medicine, Rigshospitalet, Denmark.

A focus on life-long health promotion is important for individuals with Rett syndrome. Michelle will present results and experiences from the Danish Center for Rett syndrome, with a special emphasis on the daily patterns of sedentary time/physical activity and participation in 'uptime' activities in girls and women with Rett syndrome.

Ann-Marie Bisgaard, MD, PhD, Center for Rett syndrome, Kennedy Center, Department of Paediatrics and Adolescent Medicine, Rigshospitalet, Denmark.

Presentation 1: Motor development is often impaired in children with Rett syndrome. We will present a study of 25 girls aged three to 12 years focused on sitting, standing and walking skills before, during and after the regression period. The aim of the study was to answer the question "Can girls with Rett syndrome (re)learn gross motor skills after regression?" in order to improve counselling including targeted habilitation.

Presentation 2: The symptoms of Rett syndrome are multifaceted and they change over time. We will present our experiences from the Danish Center for Rett syndrome based on our knowledge of about 100 individuals with Rett syndrome from diagnosis and up to nearly 65 years of age.

Birgitta & Jessica Green are leaders for "Gymnastics for all - Alla kan gympa", a section of the Swedish Gymnastics Federation. They are also mother and sister, respectively, to a girl with Rett syndrome.

Everyone should have the opportunity to move according to their own ability. The world of gymnastics offers so many possibilities, including improved heart rate, strength and balance. It also helps us learn about body perception, colours, shapes, to communicate and to show respect for each other. Most of all it gives us the chance to experience the joy of motion, the joy of music and friendships.

Lisbeth Nilsson, PhD is an occupational therapist that has studied activity, development and learning in people with significant disabilities, to understand how to learn and use tools and strategies that enable learning.

Helena Wandin is a speech language pathologist at National Center for Rett syndrome & related disorders in Sweden, and a PhD student at Uppsala University. Her clinical and research interest is communication and strategies to support communication.

Poul Jørgen Jennum, Professor, Chief Physician, DMSc, Danish Center for Sleep Medicine, Clinic of Clinical Neurophysiology, University of Copenhagen, Glostrup, Denmark.

Up to 80% of children with neurodevelopment disorders (NDDs) are reported to have disrupted sleep; subsequent deleterious effects on daytime behaviours of the child are commonly reported. These include autism spectrum disorder, cerebral palsy, Rett syndrome and others. The aetiology of sleep disorders in children with NDDs is largely heterogeneous and disease specific. A central core of the wake-sleep involvement is likely to include

complex mechanisms including the sleep-wake and circadian regulatory system. The diagnosis and management of sleep disorders are complex. Managing sleep disorders in children is critical both for the child and for the family, but it is often frustrating due to the refractory nature of the problem. Sleep hygiene must be implemented as first-line therapy; if sleep hygiene alone fails, it should be combined with pharmacologic management. This presentation will discuss mechanisms for sleep-wake disorders in Rett syndrome and other NDDs, as well as the potential evaluation, management and research agenda.

Stefanie K. Sachse, PhD has studied special education, specialised in AAC and has professional experience in AAC consultation and intervention. She currently works remotely for the University of Cologne, Germany. Her special interests are vocabulary selection and literacy in AAC.

Even though the children have complex communication needs, this does not mean that they cannot make progress on their literacy path. This lecture will address some of the relevant activities, adaptations, tools and programmes, and also look at the topic of prerequisites or readiness skills. In this regard, the emergent literacy viewpoint provides a different understanding and approach. The only prerequisite for literacy is the belief that everyone can make progress on his or her literacy path and learn to the best of his or her ability. In line with the “Literacy Bill of Rights” it is our responsibility and obligation to provide a wide range of high quality, i.e. evidence-based, individualised and meaningful literacy activities.

Eva Holmqvist, is a specialist in occupational therapy. She has a specific interest both clinically and in research in the field of gaze-controlled computers, especially for children with severe multiple disabilities.

Jane Lunding Larsen, MA (Ed) in Educational Psychology, Center for Rett syndrome, Kennedy Center, Department of Paediatrics and Adolescent Medicine, Rigshospitalet, Denmark.

Parents of girls with Rett syndrome experience more stress than parents of non-disabled children and they are at risk of having a reduced health-related quality of life. This session will focus on what parents of girls and women with Rett syndrome worry about and how preoccupied they are with these worries based on a study of parental worrying among Danish parents.

Åsa-Sara Sernheim, occupational therapist at National Center for Rett syndrome & related disorders, Sweden, PhD student at the University of Linköping, Sweden. Her clinical work and projects focus on the importance of activity.

Lotta Lintula, speech language pathologist, Tampere University Hospital, PhD student, Tampere University, Finland. Lotta’s research interest is eye gaze technology and Rett syndrome.

Maria Dellskog, is a member of the board for The Association for Rett Syndrome In Sweden (RSIS). She was a founding member of the Rett Section in 1986, which later became RSIS. She is mother to a 38-year-old woman with Rett syndrome who is the greatest mentor in Maria's life.

EXHIBITORS

- Abilia
- Kom i kapp
- Kompis Assistans
- Multivib AS
- Tobii Dynavox
- Made for Movement

