

# The Swedish National Center *for* Rett syndrome & related disorders

Support and information to patients, families, care assistants, and other professionals



**NATIONELLT CENTER**  
*för* RETT SYNDROM & NÄRLIGGANDE DIAGNOSER

**Rett syndrome, CDKL5 disorder, FOXP1 disorder, Pitt Hopkins syndrome, MECP2 duplication syndrome, Angelman syndrome, Mowat Wilson syndrome, and Phelan McDermid syndrome**

## ASSESSMENTS

- Three days' assessment at the Center by a child neurologist, a physiotherapist, an occupational therapist, a music therapist, a psychologist, a speech language pathologist and an orthopaedist.
- Report via telephone to family and colleagues
- Written report
- After four months, meeting with the support network in the hometown
- Focus: prevention of comorbidities and support to development

## CONSULTATIONS

- Consultations can be carried out at the Center or in the hometown. Social media and e-health options has opened up possibilities to reach individuals, families and professionals nationally and globally.
- Pilot project to develop online courses and online support is currently running, with participants from Sweden and other European countries.
  - More accessible ways to get answers to questions about symptoms and treatments are being developed
  - The aim is to provide support to families who do not have access to specialist care where they live.

## BASIC EDUCATION

Families and personal assistants provide the day by day care, training, and support. They also assist individuals to varied and stimulating activities. Basic education in Rett syndrome is given yearly in collaboration with the interest organisation Rett syndrome in Sweden (RSIS). The format and content varies according to their needs. The first basic education in Angelman syndrome is held in Stockholm in April 2018.

### Topics:

- Management of seizures and sleep disturbances
- To promote motor function and prevent loss of function
- Feeding
- Active and recovering activities,
- Enriched environment
- The impact of music
- Positive behaviour support
- Augmentative and alternative communication (AAC)

## CASE STUDIES

- 5-year project with five planned case studies examining interventions and their impact on active participation. In 2018 manuscripts for two cases will be submitted in collaboration with researchers from ??:
- Orthopedic surgery
  - Eye gaze technology

## NORDIC RETT CONFERENCE

- Held in Stockholm in April, 2018
- Topics: Health, Motor function, Communication and education, Quality of life
- Research & clinical experience
- Presenters from Denmark, Finland, Norway, Sweden
- 75 attendants from representing Sweden, Norway, Denmark

## TOPIC-SPECIFIC EDUCATIONS

- 2018:
- Workshop in AAC
  - Music every day
  - Positive behaviour support
  - Motivational Interview

**Combination of research, clinical work and disseminating knowledge,  
national and international collaboration  
for best possible health and development throughout life.**

Monika Dolik-Michno, Helena Wandin, National Center for Rett syndrome & related disorders,  
Region Jämtland Härjedalen, Sweden

[www.nationelltcenter.se](http://www.nationelltcenter.se)