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## Health-enhancing participation in girls and women with Rett syndrome

  
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Center for Rett syndrome


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### Danish center for Rett syndrome

- 118 diagnosed with Rett syndrome, age 3-63 years
- ~ 80% annual follow-up
- 100 have a *MECP2* mutation, ♀ = 98, ♂ = 2



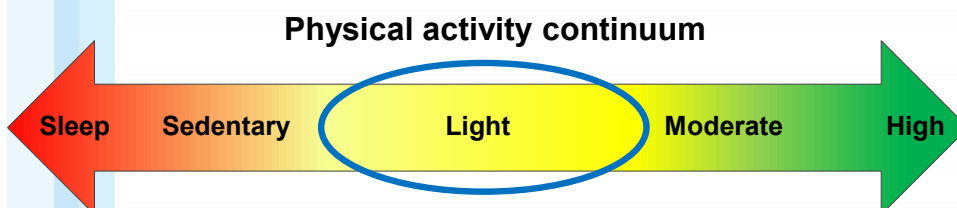
## Background

Girls and women with RTT experience:

- High dependency on caregivers in all areas of daily life
- Limited gross motor skills and risk of high levels of sedentary time and low levels of physical activity
- Resctricted participation in everyday and community activities but they enjoy physical and social activities


## Background

- Interventions to promote health throughout the lifespan in girls and women with RTT are lacking



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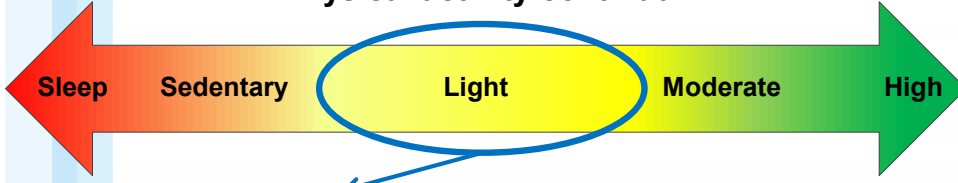
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## Background

- Interventions to promote health throughout the lifespan in girls and women with RTT are lacking

**Physical activity continuum**




Sleep Sedentary Light Moderate High

Photo of young woman in standing frame

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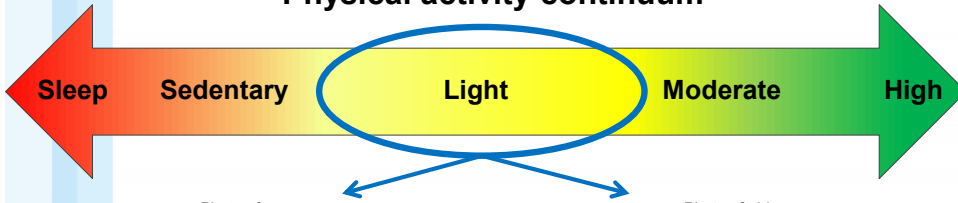
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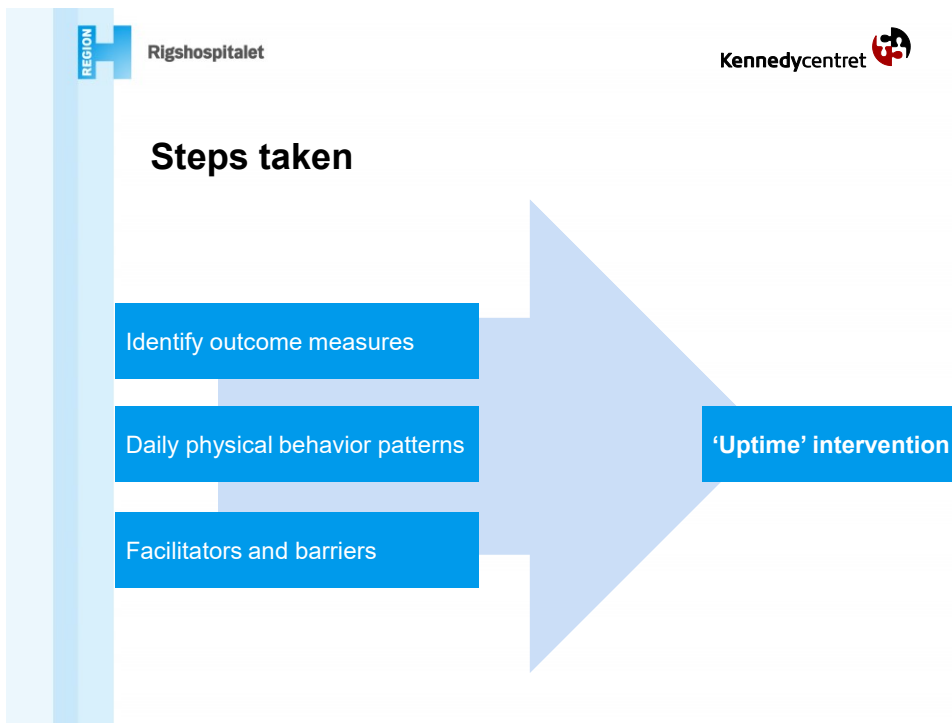
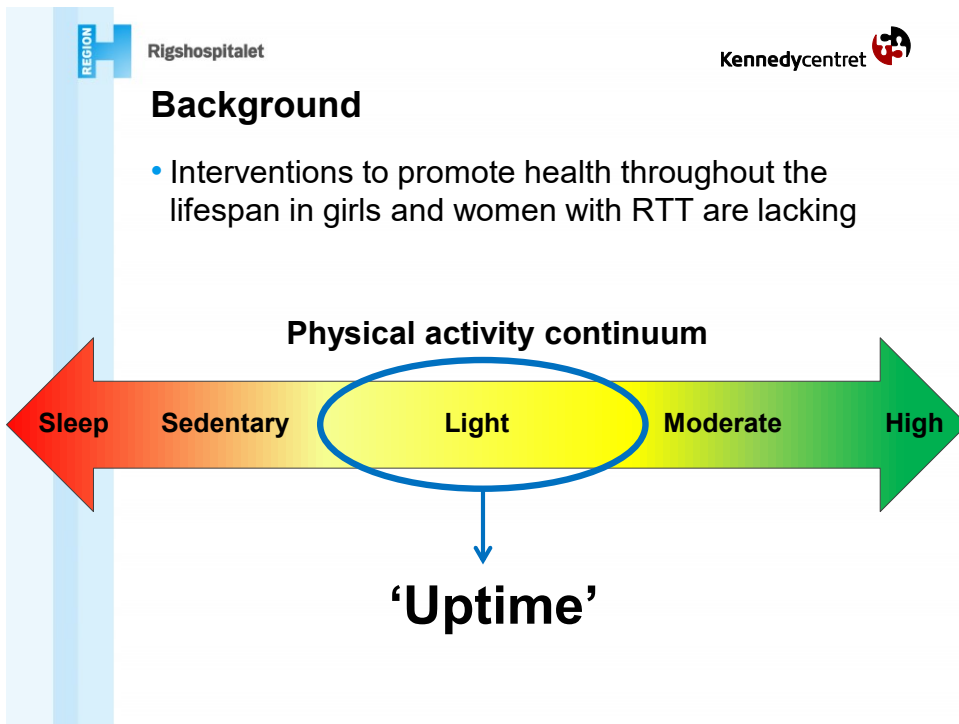
**Physical activity continuum**

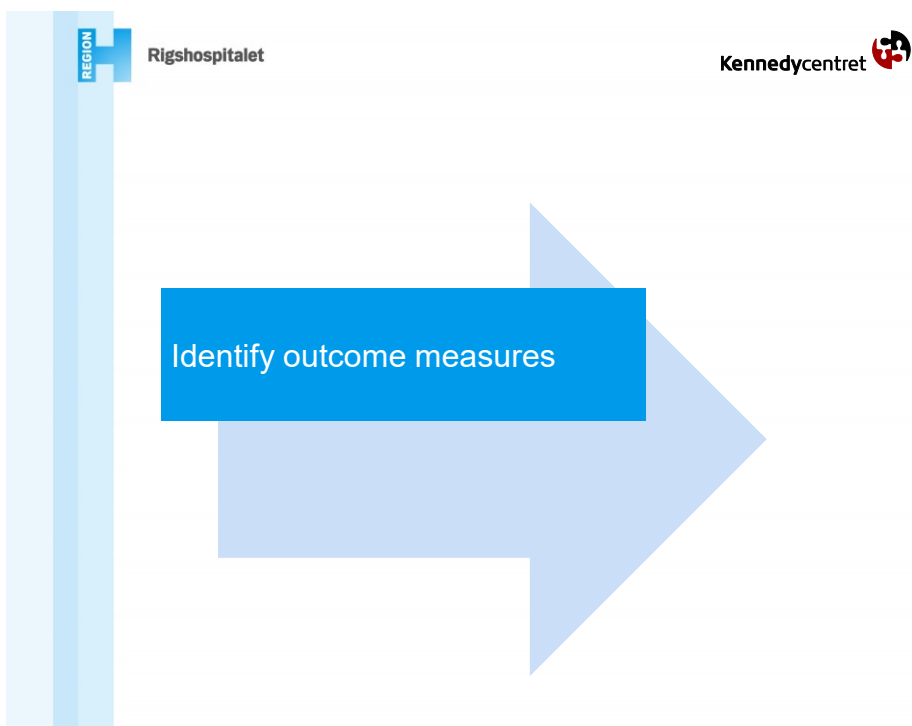


Sleep Sedentary Light Moderate High

Photo of young woman in standing frame

Photo of girl walking, holding someone's hand






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## Outcome measures

**Aim:**  
To modify two existing measures of walking and to examine their concurrent validity and test-retest reliability



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## Outcome measures

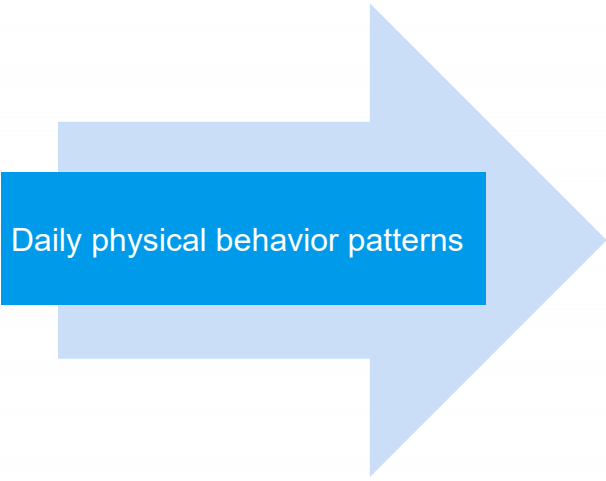
- Modification and test of two measures

### The Functional Mobility Scale – Rett syndrome (FMS-RS)

- N=42
- Concurrent validity – moderate to high correlations to comparison measures
- Test-retest reliability – ICC=0.94-0.99

### 2 Minute Walk Test (2MWT)

- N=27
- Concurrent validity – low to moderate correlations to comparison measures
- Test-retest reliability – ICC=0.86-0.98
- Minimal detectable difference – 38meters



Daily physical behavior patterns

## Daily physical behavior patterns

### **Aim:**

To describe patterns of sedentary time and steps in girls and women with Rett syndrome across ambulation levels



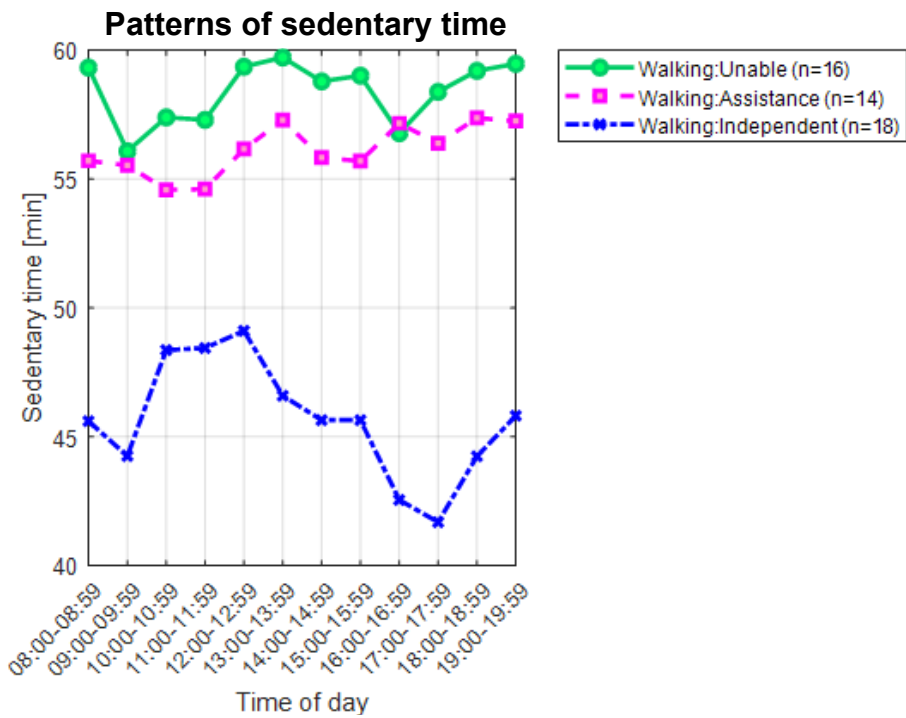
## Method

- Inclusion criteria
  - RTT and *MECP2* mutation
  - $\geq 5$  years
  - $\rightarrow n = 88$  eligible participants
- Minimum 4 days
  - ActivPAL
  - StepWatch Activity Monitor

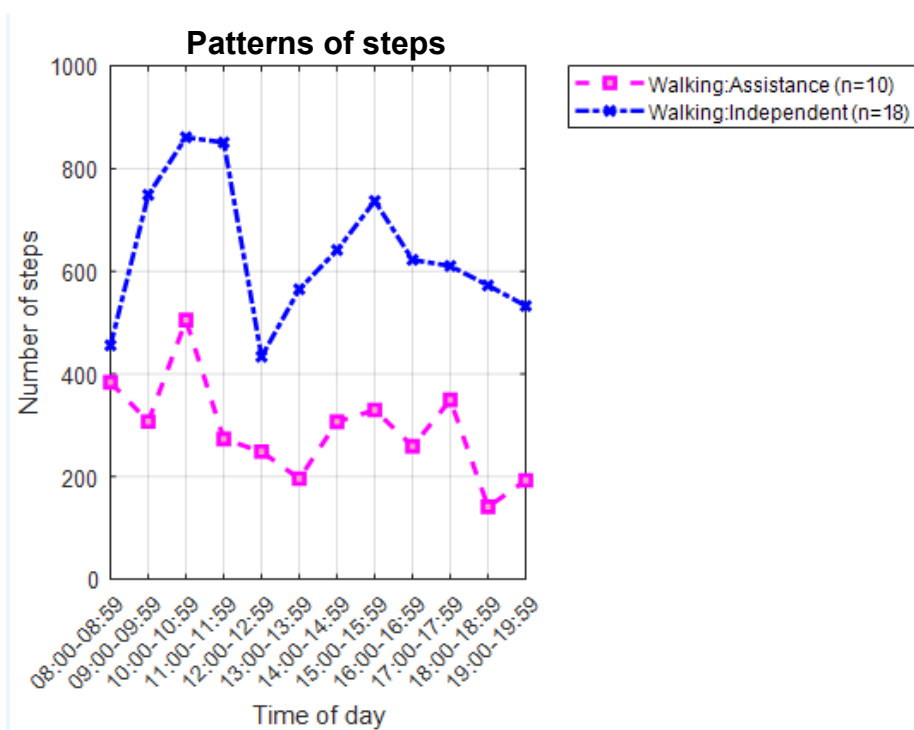


## Results

- n = 55, 5-60 years (median = 22 years)
- **Sedentary time** (n = 48):  
83 % (SD 14%) of waking hours
- **Physical activity** (n = 28):  
5,128 steps/day (IQR 2829-7704)





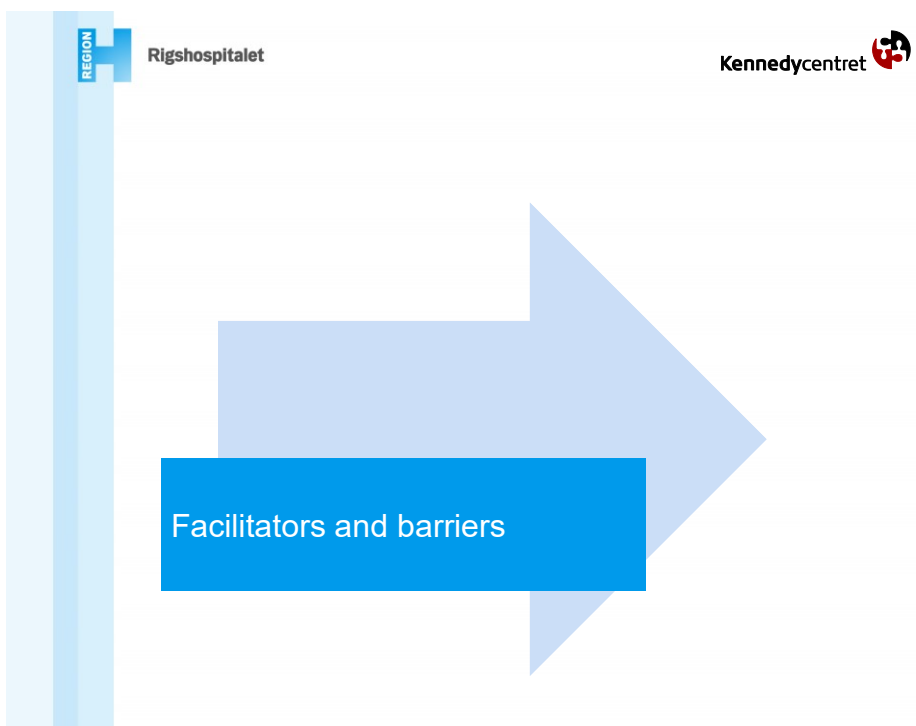


## Factors associated with sedentary time

- Univariate linear regression models
  - Increasing clinical severity
  - Decreasing gross motor skills
  - Inability to walk
  - Low maternal physical activity level
  - Absence of a physical activity policy

} sedentary time ↑
- Multivariate linear regression models
  - Assisted walking and inability to walk
  - Participants older than 33½ years

} sedentary time ↑




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## Facilitators and barriers to 'uptime' activities

**Aim:**  
To explore facilitators and barriers of participation in 'uptime' activities in girls and women with Rett syndrome from the perspectives of parents and professionals

  
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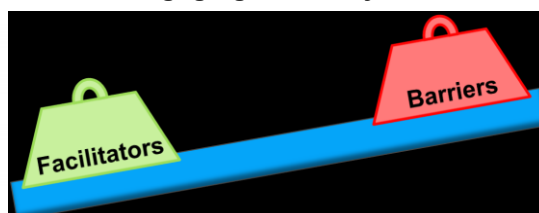
## Method

- 4 focus groups
  - Parents (n=9) } Home setting
  - Care assistants (n=4) }
  - Professionals at schools (n=7) } Day time setting
  - Professionals at day centers (n=5) }
  
- Purposive sampling



## A constant balance to do the best thing

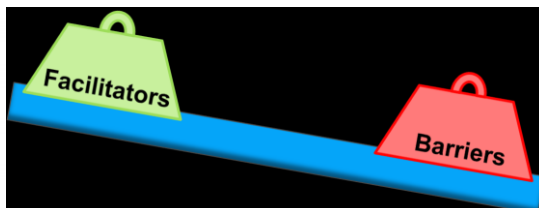
- The girl/woman engaging in activity → Individual factors



\*Walking independently  
\*Social interaction  
\*Positive reactions

## A constant balance to do the best thing

- The girl/woman engaging in activity → Individual factors

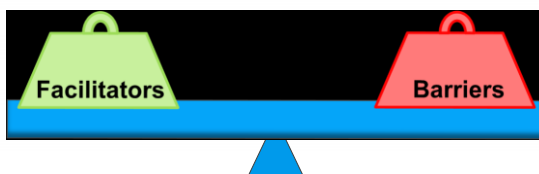


\*Walking independently  
\*Social interaction  
\*Positive reactions

\*Need of assistance  
\*Limited resources  
\*Lack of enjoyment

## A constant balance to do the best thing

- The girl/woman engaging in activity → Individual factors

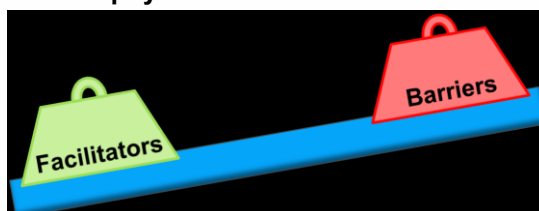


*"We have a young woman who lives here. She uses a wheelchair but can stand and walk somewhat unsteady...some of us [staff] walk with her but its very varying how tired she is"*

*Care assistant*

## A constant balance to do the best thing

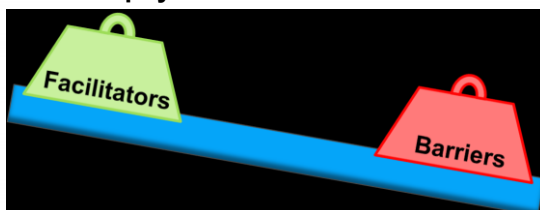
- Using aids and the indoor and outdoor environment →  
Factors in the physical environment



\*Standing/mobility aids  
\*Flexible environment  
\*Access to equipment

## A constant balance to do the best thing

- Using aids and the indoor and outdoor environment →  
Factors in the physical environment

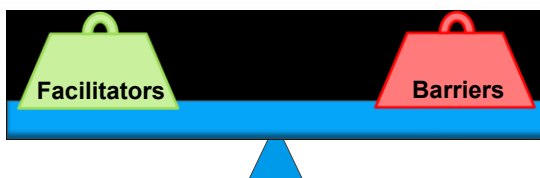


\*Standing/mobility aids  
\*Flexible environment  
\*Access to equipment

\*Difficulties getting  
aids granted  
\*Weather

## A constant balance to do the best thing

- Using aids and the indoor and outdoor environment →  
Factors in the physical environment

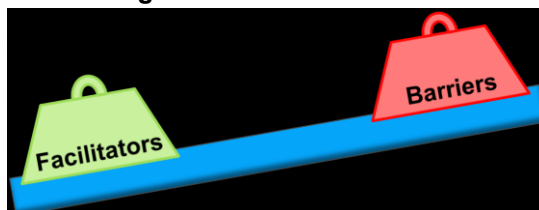


*"To have the mobility aids and equipment right here with the group, then it's ['uptime] doable that is what makes it a long-lasting solution and it makes sense in everyday life"*

*Pedagogue*

## A constant balance to do the best thing

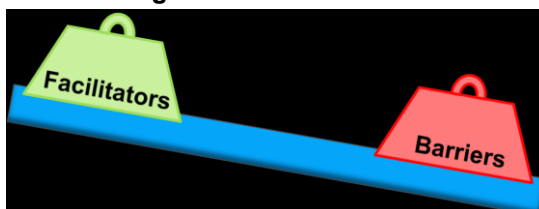
- Creating possibilities within the organizational structure →  
Factors in the organizational environment



\*Daily routines  
\*Individual strategies  
\*Having enough 'hands'

## A constant balance to do the best thing

- Creating possibilities within the organizational structure →  
Factors in the organizational environment

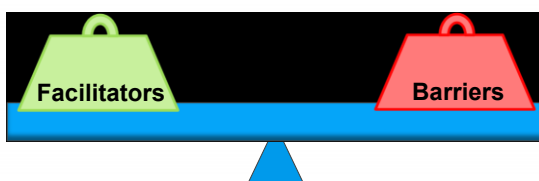


\*Daily routines  
\*Individual strategies  
\*Having enough 'hands'

\*Prioritizing activities  
\*Time constraints  
\*Limited staffing

## A constant balance to do the best thing

- Creating possibilities within the organizational structure →  
Factors in the organizational environment

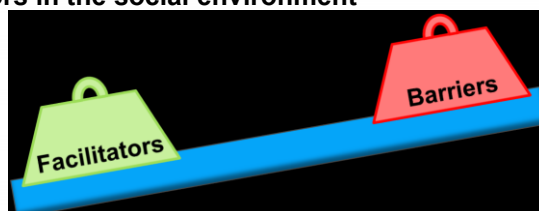


*"She can participate standing and at the same time the tube is used to feed her. She much rather want to be social than to sit down and eat with an adult. It's a balance as I say...it's a question of resources all the time"*

Parent

## A constant balance to do the best thing

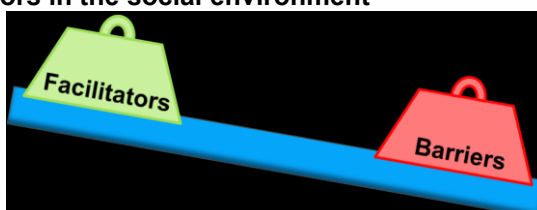
- Together we provide the best possible daily life →  
Factors in the social environment



\*Presence of a PT  
\*Collaboration

## A constant balance to do the best thing

- Together we provide the best possible daily life →  
Factors in the social environment



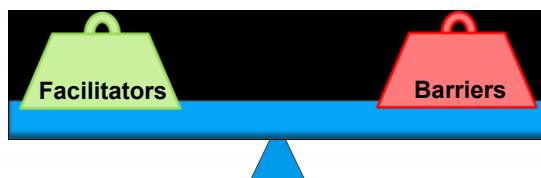
\*Presence of a PT  
\*Collaboration

\*Limited access to a PT  
\*Different priorities  
between stakeholders



## A constant balance to do the best thing

- Together we provide the best possible daily life →  
Factors in the social environment

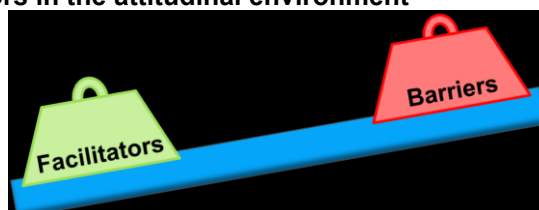


*“Often its other issues that are important to pedagogues and teachers, and that’s why it’s extremely important to be there [as a physiotherapist] to follow up on things because we prioritize differently”*

*Physiotherapist*

## A constant balance to do the best thing

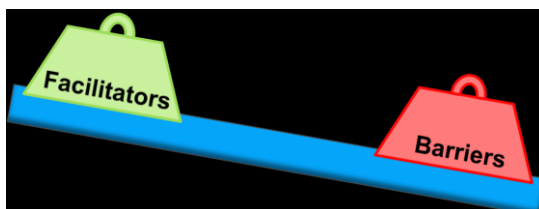
- Our knowledge, attitude and beliefs have an impact →  
Factors in the attitudinal environment



\*Motivation in stakeholders  
\*Positive attitude

## A constant balance to do the best thing

- Our knowledge, attitude and beliefs have an impact → Factors in the attitudinal environment

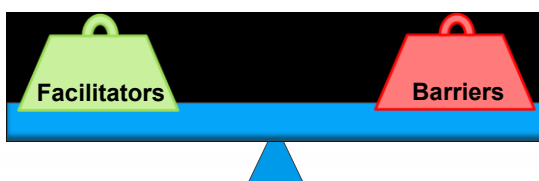


\*Motivation in stakeholders  
\*Positive attitude

\*Lack of knowledge  
\*Concerns about the right of self-determination

## A constant balance to do the best thing

- Our knowledge, attitude and beliefs have an impact → Factors in the attitudinal environment



*"I think it's important, I mean we know that physical activity is important...but I also think it's really important to respect it when they say 'no'. They have the right to say 'no' and to be tired and have a bad day"*

*Teacher*

## Clinical implications of first steps

- Provide health professionals with knowledge to develop tailored interventions that reduce sedentary time and promote 'uptime'



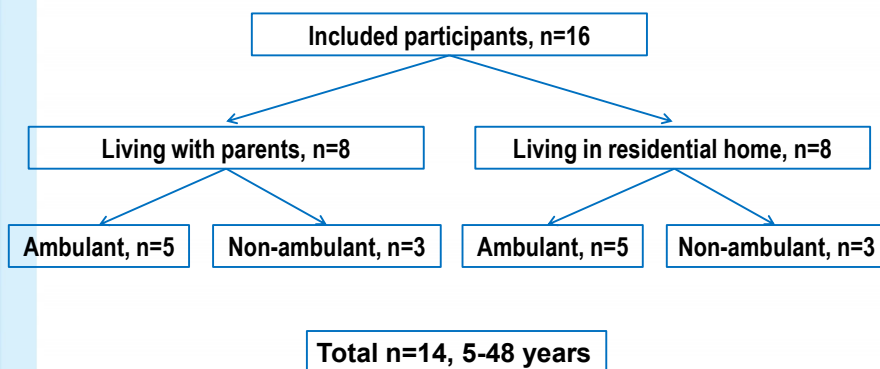
**'Uptime' intervention**

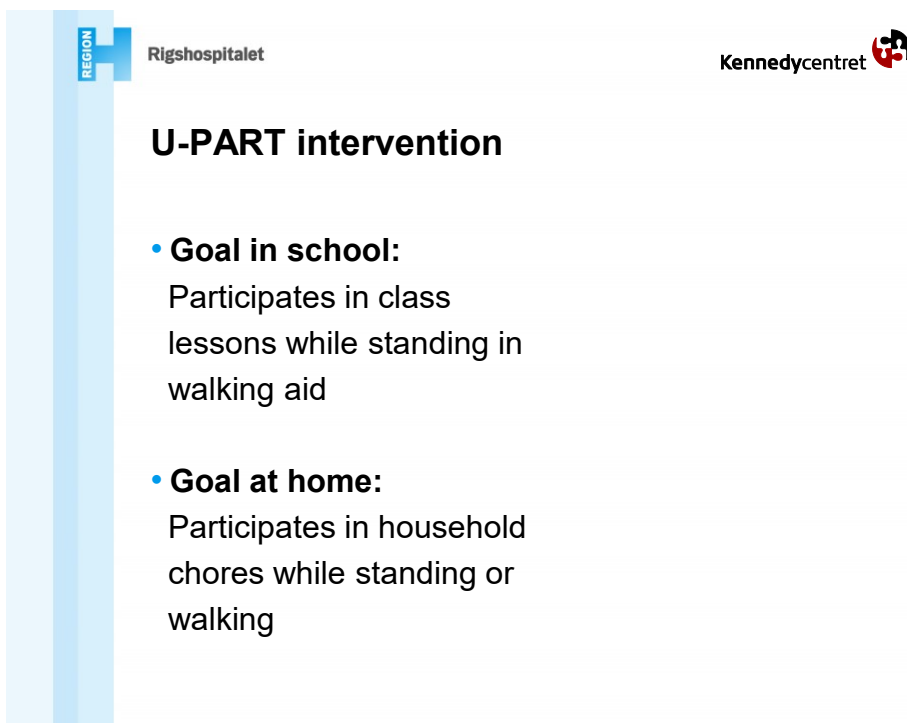
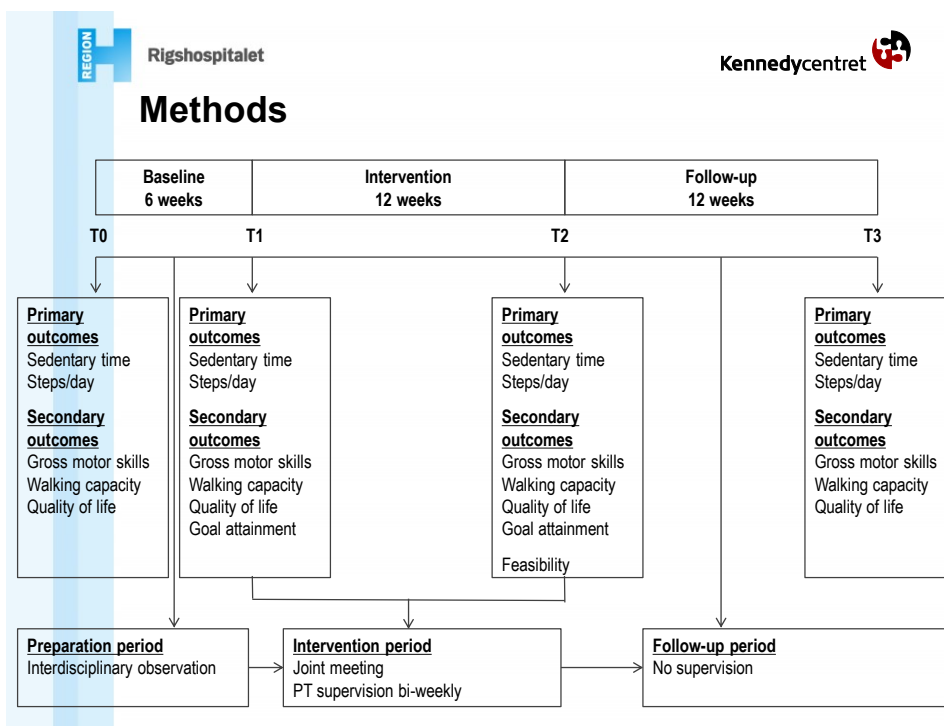
## 'Uptime' participation intervention (U-PART)

### Aim:

To evaluate the feasibility and health-related effects of an 'Uptime' Participation (U-PART) intervention in girls/women with Rett syndrome

## Participants





## Results

	Baseline	After intervention	After follow-up
Sedentary time	73.3%	67.9%	68.9%
Daily steps	4057	3989	4658
Walking capacity	54.8m	92.2m	87.6m
Quality of life	80.8	84.7	84.7

## U-PART intervention

- **Goal at home:**  
Participates in household chores while standing

Participates in games/  
arts and crafts  
while standing

## Results

	Baseline	After intervention	After follow-up
Sedentary time	94.4%	89.6%	85.8%
Quality of life	71.2	68.4	72.1

## U-PART intervention

- **Goal in day center:**  
Participates in daily walk around day center and greets other groups
- **Goal at home:**  
Participates in sport activity once a week

## Results

	Baseline	After intervention	After follow-up
Sedentary time	88.3%	84.3%	85.2%
Daily steps	3151	3961	3956
Walking capacity	113.5m	120.0m	118.7m
Quality of life	72.7	78.2	80.0

## U-PART intervention

- **Goal in day center**  
Walks to social activities with using a walking aid (assisted by two persons)
- **Goal at home:**  
Walks 5m to sofa to drink evening coffee



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## Results

	Baseline	After intervention	After follow-up
Sedentary time	96.6%	93.3%	95.6%
Quality of life	67.1	71.5	65.0

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## Results – Health-related effects

Primary outcomes	T0	T1	T2	T3
<b>Daily sedentary time</b>				
Sitting time (h)	9.79 (1.6)	9.67 (1.6)	9.13 (1.56)	9.21 (1.45)
Relative to waking h (%)	83.5 (10.7)	84.8 (10.5)	80.1 (10.2)*	80.9 (9.6)*
<b>Daily physical activity</b>				
Step count	4291 (2650)	4522 (2642)	5096 (2546)*	4700 (2665)
Secondary outcomes	T0	T1	T2	T3
Gross Motor Skills (/45)	21.6 (8.6)	21.3 (8.4)	21.5 (8.3)	21.1 (8.3)
Walking capacity (m)	81.9 (35.4)	93.6 (35.3)	106.5 (33.7)*	99.9 (34)*
Quality of life (/100)	78.3 (7.9)	78.6 (7.8)	81.4 (8.8)*	79.8 (10.6)

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After intervention: -4.09% ([95%CI -5.87,-2.32],  $p<0.001$ )  
 After follow-up: -3.36% ([95%CI -5.15,-1.58],  $p<0.001$ )

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After intervention: +708 steps/day ([95%CI 126,1290],  $p<0.019$ )

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After intervention: +18.94m ([95%CI 7.45,30.42],  $p=0.002$ )  
 After follow-up: +12.40m ([95%CI 0.87,23.29],  $p=0.036$ )

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After intervention: +2.81 points ([95%CI 0.5,5.11],  $p=0.018$ )

## Results – Goal Attainment Scaling

- Total of n=56 GAS goals (median = 4)

Score	Goal
-2 (baseline)	The girl <b>does not</b> perform any class chores (e.g. distributing lunch boxes to classmates)
-1 (less than expected)	The girl participates in class chores <b>once</b> a week while walking
0 (expected level)	The girl participates in class chores <b>twice</b> a week while walking
+1 (better than expected)	The girl participates in class chores <b>three</b> times a week while walking
+2 (much better than expected)	The girl participates in class chores <b>four</b> times a week while walking

## Results – Goal Attainment Scaling

- Total of n=56 GAS goals (median = 4)
  - n=33 goals (58.9%) showed expected improvements
  - n=21 goals (37.5%) showed minor improvements
  - n=2 goals (3.6%) did not change

## Feasibility

- Overall good feasibility
  - High degree of fulfilled expectations
  - High degree of appropriateness
  - High degree of intention to continue with 'uptime'
- Assessments
  - Acceptable and feasible
- Preparation (observation, joint meeting)
  - Acceptable, feasible and relevant
- Intervention
  - Supervision and 'uptime' activities were very feasible
  - Time consuming to some extent (no extra resources)
  - Difficult to provide the girl/woman with choice

## Conclusion

- The concurrent and test-retest reliability were established in the modified 2MWT and FMS-RS measures
- The level of sedentary time was high during awake hours and the daily step count was low. Most 'uptime' activities take place during the morning
- Optimal participation in 'uptime' activities was achieved when balancing facilitators and barriers within the individual and her physical, organizational, social and attitudinal environment
- The U-PART intervention was feasible and positive health-related effects were seen

## Clinical implications

- Building the repertoire of walking measures is valuable for researchers and practitioners for clinical monitoring and evaluation studies
- Knowledge of daily physical behavior patterns can aid health care professionals in promoting active lifestyles
- Interventions promoting 'uptime' activities should take the different facilitators and barriers into account to optimize participation across settings
- The U-PART intervention provided a systematic framework for the promotion of 'uptime' within the participant's natural environment

## Take home messages

- Many steps are necessary in the development of an optimal health-promoting intervention for girls/women with RTT
- Need of larger and longer clinical trials



## Thank you

## Key references

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