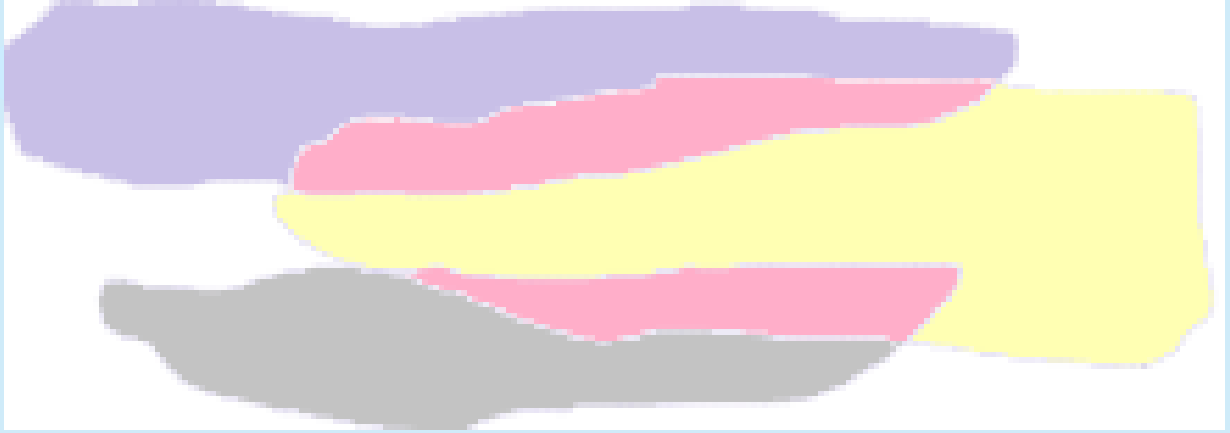

Can girls with Rett syndrome (re)learn gross motor skills after regression?

RETT SYNDROME IN NORDIC LIGHT
Stockholm 19-20 April 2018

Nordic Rett Conference – Research and Practice

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Can girls with Rett syndrome (re)learn gross motor skills after regression?

BACKGROUND INFO

- What is our background for talking about this?
  - Center for Rett syndrome in Denmark
- What do we know in the RTT world about the abilities to sit, stand and walk throughout life?
- Useful knowledge about the gross motor skills in adults
- Do gross motor skills change in the adults?

- Part 2: Can girls with Rett syndrome (re)learn gross motor skills after regression?

REGION

Center for Rett syndrome in Denmark (since 2007, with an interest since the 80's)

- We offer clinical follow-up of individuals with RTT in Denmark on a livelong basis as a highly specialized service
- We offer multidisciplinary counseling and support to all Danish patients and families and
- advice on management of individuals with RTT for local health care and social authorities
- Research is included in the daily clinical practice

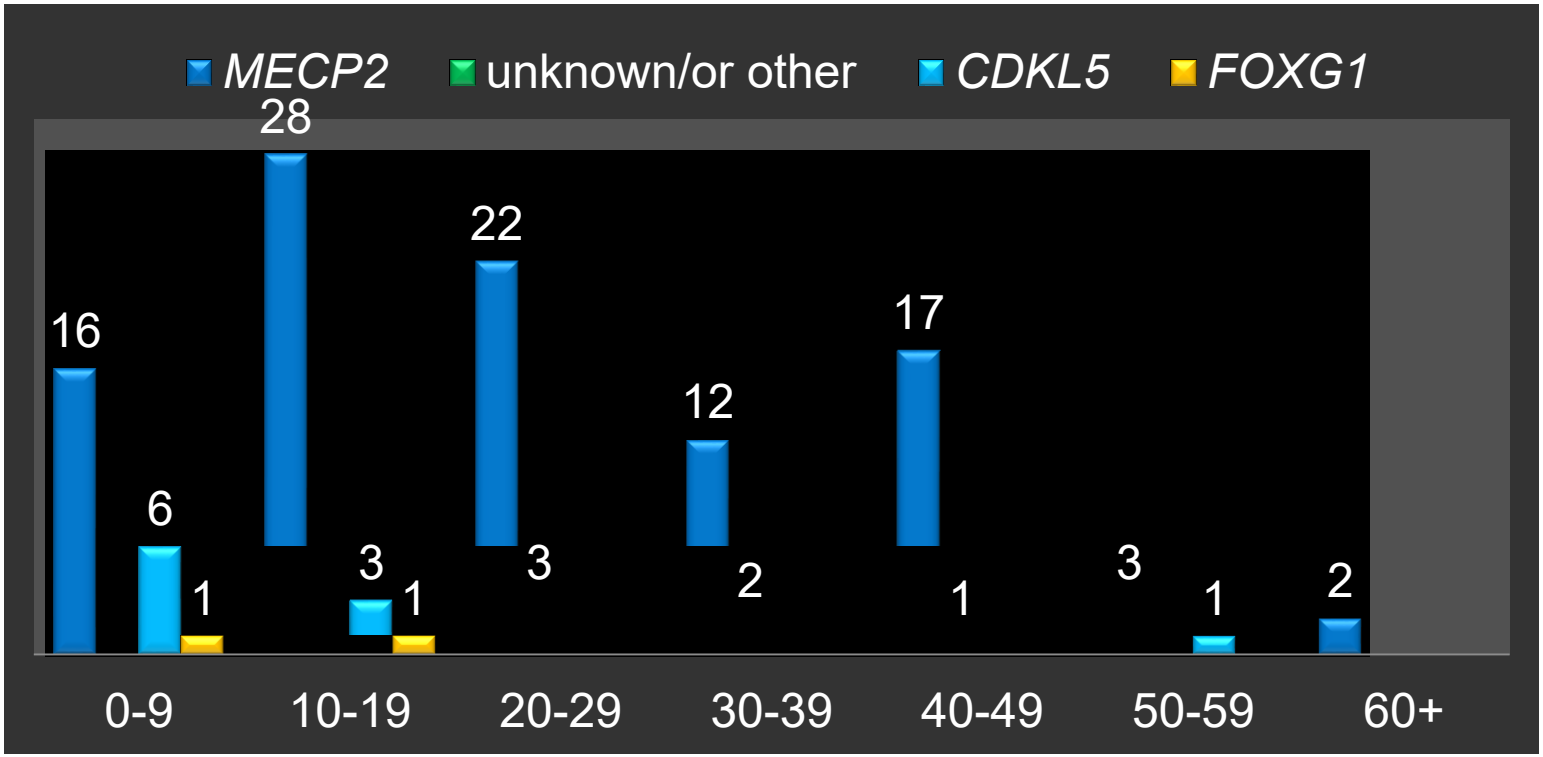
REGION

Danish Center for Rett syndrome, who are we?

- A multidisciplinary team, which in spring 2018 includes
  - Neuropaediatrician
  - Educational psychological adviser
  - Clinical physiotherapist
  - Research physiotherapist (PhD student)
  - Research assistant, geneticist (PhD student)
  - Social worker (part-time, 33%)
  - Access to a nurse, a dietician, a secretary, and to the laboratory

REGION

We have knowledge of 118 individuals - the majority has typical RTT with a *MECP2* variant



What do we know in the “RTT world” about the abilities to sit, stand and walk throughout life?

Cross-sectional study of Gross Motor Skills in a cohort of 77 children and adults i 2015

Rett syndrome Gross Motor Scale (Downs J, 2008)

Rett Syndrome Gross Motor Scale - RSGMS

Validating the Rett Syndrome Gross Motor Scale.

Downs J, et al. PLoS One. 2016 Jan 22;11(1):e0147555. doi: 10.1371

Item	Level of assistance				Comments
	Maximal (0 points)	Moderate (1 point)	Minimal (2 points)	None (3 points)	
1. Sitting on floor for 10 seconds					
2. Sitting on a chair with a back 10 seconds					
3. Sitting on a stool 10 seconds (no back support)					
4. Sit to stand					
5. Standing 3 seconds					
6. Standing 10 seconds					
7. Standing 20 seconds					
8. Walks 10 steps					
9. Side steps					
10. Turns					
11. Walks on a slope					
12. Steps over obstacle (height of a door frame)					
13. Stands up from the floor					
14. Bending to touch the floor and returns to standing					
15. Runs					

Cross-sectional study of Gross Motor Skills in a cohort of 77 children and adults i 2015

Gross Motor Scale (Downs J, 2008)

•To sit independently	81%
•To stand independently at least 20 sec	51%
•To walk independently at least 10 steps	40%

The ambulation level in the Danish population of 94 girls and women, aged 3-60y, with RTT and a *MECP2* mutation using HAS

- Community ambulators and household ambulators
  - 58% walked independently or with assistance
- Therapeutic ambulators
  - 14% walked a few steps with substantial assistance
- Non-ambulant/Standers
  - 28% were non-ambulant

60-70% are able to walk without or with support

Other studies show

- Downs J et al, 2008 (99 individuals, aged 1.5-28y)

Videos, RSGMS

  - The majority of individuals with RTT are able to sit independently
  - 43% walked independently and 27% with support

- Pidcock FS et al, 2015 (96 individuals, children, aged?)

PAMS

  - Ambulation in 57% (differences between mutationtypes)

- Vignoli A et al, 2011 (84 individuals, aged 14-42y)

questionnaire

  - 20% walked independently, 42% with assistance; 23% non-ambulatory, 15% had lost the ability

REGION

Useful knowledge about the gross motor skills in adults

DISABILITY AND REHABILITATION, 2016
http://dx.doi.org/10.3109/09638288.2016.1170896

Taylor & Francis
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ORIGINAL ARTICLE

Functional abilities in aging women with Rett syndrome – the Danish cohort

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ABSTRACT

Rett syndrome (RTT) is a neurodevelopmental disorder, which mainly affects females and results in multiple disabilities. Many clinical descriptions of the symptoms and functional abilities have been made medically, though mainly in children with RTT. Previous reports have established that even though the syndrome causes severe psychomotor disability, women with RTT can live long into adulthood.

Purpose: We aim to describe what to expect from aging women with RTT regarding some of the basic functional abilities that are used in daily activities and that could have an impact on quality of life in these women.

Methods: A team of two medical doctors, a physiotherapist and an educational psychological adviser, performed clinical evaluations of 27 women with RTT in Denmark above 30 years of age and confirmed *MECP2* mutation.

Results: We found that 63% of the women were able to walk outside their homes and only 11% were not able to walk at all. However, 67% could not transfer from sitting to standing position without support. There was profound difficulties communicating, but 85.1% of the women could either consistently point with their hand or eyes to things of their interest.

Conclusions: Women with RTT are very dependent on caregivers who maintain and rehabilitate their functional abilities. They can often walk short distances unassisted, but do have trouble transferring and thus getting up from a chair on their own. They have severe problems communicating and they often perform subtle signs that can be difficult to recognize.

ARTICLE HISTORY
Received 29 September 2015
Revised 22 March 2016
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KEYWORDS
Aging; communication; functional ability; hand function; intellectual disability; *MECP2;* methyl CpG binding protein 2; Rett syndrome

Do gross motor skills change in the adults?

visit 1 follow-up

6 years - 8.4 years

- **24** females with clinical RTT and a *MECP2* mutation
- Evaluated twice
  - follow-up period from 6 years to 8.4 years (mean 7.2 years)
- Clinically assessed, video filmed and scored for gross motor skills
  - in 10 of the 15 items from the validated RTT Gross Motor Scale
- Age at follow-up ranged from **30.5 years to 62 years** (mean age 43 years)

Change in gross motor functions

Items	Maintained ability, N	Improved ability, N	Decreased ability, N
Sitting on the floor	15	0	9 (4 lost)
Sitting on chair with back	21	0	3
Sitting on chair without back	23	0	1
Standing for three seconds	19	0	5 (1 lost)
Standing for ten seconds	20	0	4 (3 lost)
Standing for 20 seconds	18	0	6 (2 lost)
Getting up from sitting on chair	15	0	9 (2 lost)
Walking ten steps	17	0	7 (2 lost)
Side step	7	0	17 (4lost)
Turn 180 degrees	14	0	10 (2 lost)
Total	2	0	22

Change in gross motor functions

Items	Maintained ability, N	Improved ability, N	Decreased ability, N
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Total	2	0	22

In relation to the children, we concluded from the study that

- It is important to learn the children to move as much as possible in the daily life and
- to motivate them to do as much

  as they can and

  so to give them the best conditions

  to maintain their abilities

  into adulthood

Can girls with Rett syndrome (re)learn gross motor skills after regression?

- In the regression period the children
  - Loss some learned skills – mainly regarding hand function and spoken language
  - Might get periods of distress and might scream for no obvious reason
  - Might withdraw socially and avoid eye contact
  - Get other symptoms - not focused on in this talk

Questions and Objectives

- Parents often ask – and especially at the time of diagnosis
  - What is the probability that my child will learn to walk?
  - Will my child loss the ability to walk?
  - Is it useful to exercise?

- To be able to ask these questions and to improve our counselling including targeted habilitation
  - We did at study and looked in to changes in gross motor skills in children with Rett syndrome in relation to regression

Methods

- Review of records and clinical evaluations regarding sitting, standing and walking
  - in a cohort of 25 girls
  - aged three to 12 years with
  - RTT and a *MECP2* mutation
  - data were related to parent-reported time of regression
  - quality of movement was not reviewed

Gross motor skills before and after regression for 25 girls as a group

	Before regression	After regression*
To sit	22 (88%)	23 (92%)
To stand with assistance	17 (68%)	18 (72%)
To stand independently	8 (32%)	12 (48%)
To walk with assistance	14 (56%)	14 (56%)
To walk independently	8 (32%)	11 (44%)

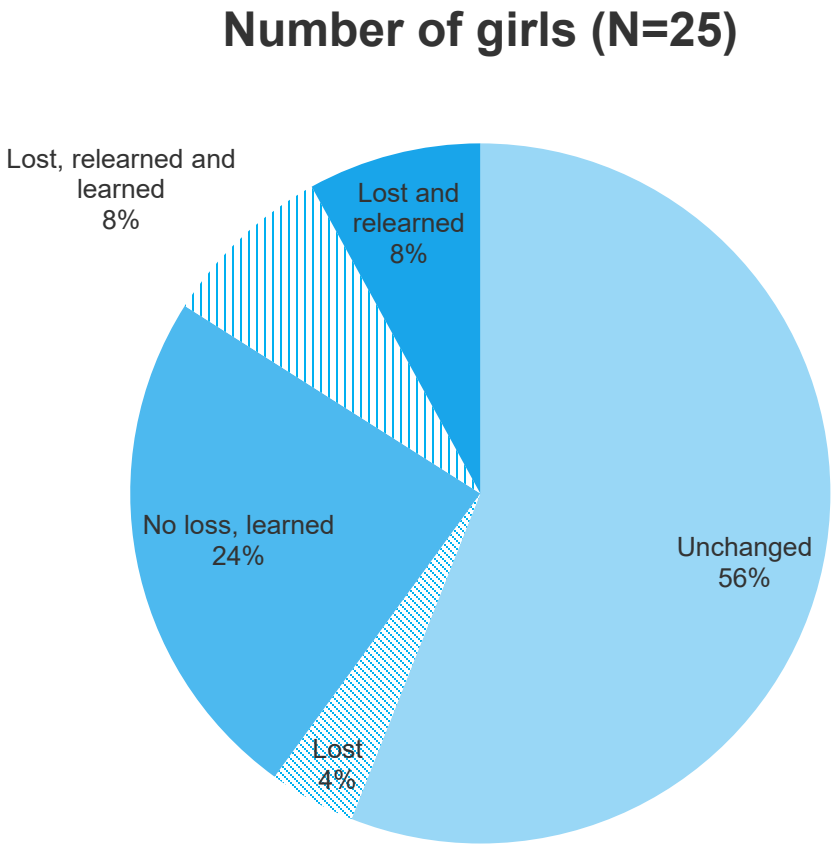
* Observation period 0.2-9.5 years from regression (mean 4 years, median 3 years)

Changes in gross motor skills sit, stand and walk

	Number of girls (N=25)
Unchanged*	14 (56%)
No loss, learned	6 (24%)
Lost	1 (4%)
Lost, relearned	2 (8%)
Lost and relearned and learned	2 (8%)

* Some lost the ability to crawl or to turn around

Changes in gross motor skills sit, stand and walk



Changes in gross motor skills sit, stand and walk *during regression* at the individual level

- Five girls (20%) ***lost*** gross motor skills during regression; two of the girls lost more than one skill
  - sit (1)
  - stand with support (2) or independently (2)
  - walk with support (1) or independently (2)

Changes in gross motor skills sit, stand and walk *after regression* at the individual level

- Four girls (16%) ***relearned*** skills:
  - to sit (1)
  - stand with support (1) and independently (1)
  - walk with support (1) and independently (1)

Changes in gross motor skills sit, stand and walk *during* *regression* at the individual level

- Four girls (16%) ***learned*** gross motor skills ***during*** regression; two of the girls learned two skills:
  - to sit (1)
  - stand independently (3)
  - walk independently (2)

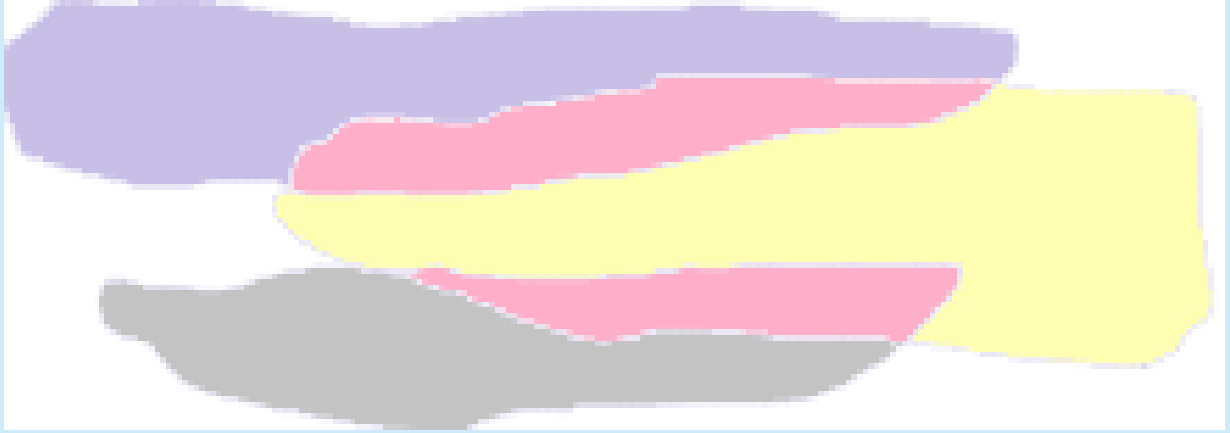
Changes in gross motor skills sit, stand and walk *after regression* at the individual level

- Five girls (16%) learned new skills:
  - standing with support (3) and independently (1)
  - walk with support (1) and independently (1)

Can girls with Rett syndrome (re)learn gross motor skills after regression?

- The answer is

Yes



REGION

Can girls with Rett syndrome (re)learn gross motor skills after regression?

Video examples

Conclusion

- Girls with RTT can develop gross motor skills during and after the regression period
- It emphasises the importance of continued and targeted physiotherapeutic counselling and training
    - for maintenance but
    - also for promotion of new gross motor skills