Linköping Studies in Health Sciences, Thesis No. 133

Time for Activities for Girls and Women with Rett Syndrome

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Linköping 2018

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Printed in Sweden by LiU-Tryck, Linköping, Sweden, 2018

ISBN 978-91-7685-232-3

ISSN 1100-6013

Have a good time!

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ABSTRACT

Irrespective of the great individual variation, people diagnosed with RTT largely rely on support from others to be able to do and participate in activities throughout their lives. This thesis focuses on which activities are done and liked/disliked by girls and women with RTT in Sweden. The overall aim was to describe the everyday lives of female individuals with Rett syndrome.

Two studies are included in this thesis. The first is a descriptive study, using secondary data from three earlier questionnaires, encompassing data from 175 participants (girls/women) described by 365 informants (parents/staff). Content analysis was used to analyse the open-ended questions. In the second study a Time-geographic diary method and the software VISUAL-TimePACts computer program, DAILY LIFE 2011 were used. Ten participants (teenagers/young female adults) with RTT and their 63 informants participated in the diary study.

The main findings in the first study (I) were that the girls and women with RTT enjoyed activities that included aspects of 'contact', 'sensory impression' and 'motion'. The activities most enjoyed over the years were bathing/swimming, listening to music or being outdoors/walking. The parents and staff also liked to do the same activities that the girls or women enjoyed doing, described as sharing their joy. Of the few activities that were reported as being unenjoyable, most were daily care activities.

The diary study (II) showed that the most frequently reported activities were hygiene/toilet, moving around indoors, eating and getting dressed. Most time was spent in sleeping, daily care, medical and health care activities and also for travel/transportation. Little time remained for other kinds of activities especially for the young adults. Most time was spent with staff, thereafter with families, and the least time was spent with friends. The participant response that was reported most often during activities was 'interested', while 'opposed' was the least reported. Responses of 'opposition' were primarily seen during caring activities such as toileting, using the breathing mask, stretching, brushing teeth, being woken up, dressing and putting on orthoses. Responses of 'engagement' were noted in contexts of socialising, playing and communicating activities with friends or staff. Engagement responses were also reported during activities of 'motion' such as changing body position, moving in the water or

gymnastics, eating food and snacks, and even when watching/listening to films, books or music.

Thus, increased knowledge concerning the importance of activities for girls and women with RTT is essential for their well-being, participation and continued development. Increased knowledge could facilitate the choice of activities and a more varied use of activities.

Regardless of age, severity of symptoms or developed skills, it is important that not only basic needs such as sleep, daily care and medical health care activities are fulfilled for individuals with Rett syndrome. It is also essential for them to spend time with friends, family and staff doing enjoyable activities both at home and in other places.

Keywords: Activities of daily living, activity patterns, neurodevelopmental disorder, occupation, occupational therapy, Rett syndrome, time-geographical diary method, time use.

LIST OF PAPERS

This thesis is based on the following papers that are referred to in the text by their Roman numerals:

- I. Sernheim, Å-S., Hemmingsson, H., Witt Engerström, I., & Liedberg, G. (2016). Activities that girls and women with Rett syndrome liked or did not like to do. *Scandinavian journal of occupational therapy*, 1-11. Doi: 10.1080/11038128.2016.1250812
- II. Sernheim, Å-S., Hemmingsson, H., Lidström, H., Witt Engerström, I., & Liedberg, G.M. Rett syndrome – Teenagers' and young adults' activities, usage of time and responses during an ordinary week - a diary study. 2018. Submitted manuscript.

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ABBREVIATIONS

AAC	Augmentative and Alternative Communication
CA	Content analysis
ICF	International Classification of Functioning, Disability and Health
LSS	The Legal Act concerning Support and Service for Persons with Certain Functional Impairments
MeCP2	Methyl-CpG-binding protein 2
MECP2	Mutations of the <i>MECP2</i> gene
OT	Occupational Therapy
RTT	Rett syndrome
WFOT	World Federation of Occupational Therapists

DEFINITIONS

Activity(ies): An observable unit of behaviour, and recognizable sequence of actions taken together in a particular context; beyond tasks yet without the complexity of occupations in the simple to complex hierarchy of tasks, activities and occupations (Christiansen & Townsend, 2010).

Activity patterns: The building blocks of the activity pattern are recognisable sequences of actions and activities (Bendixen et al 2006).

Informant: Person who provides the information concerning the individual with RTT who is unable to speak for themselves. In this thesis the informants were a parent or a staff member of the individuals with RTT.

Occupation: All that people need, want, or are obliged to do; what it means to them; and its ever-present potential as an agent of change, it encapsulates doing, being and becoming (Wilcock, 2006).

Occupational balance: The right amount and variation of occupations (Wagman, Håkansson, & Jonsson, 2015).

Occupational science: The rigorous study of humans as occupational beings (Yerxa, 1990).

Participant: In this thesis, a female individual diagnosed with RTT.

PREFACE

This thesis is inspired by my encounters with girls and women diagnosed with Rett syndrome (RTT), a rare, genetic and severe neurodevelopmental disorder (Amir et al., 1999). Common consequences of the syndrome include partial or complete loss of spoken language, a dyspraxic gait or inability to ambulate, stereotypic hand movements and breathing disturbance (Neul et al., 2010). Co-morbidities often develop including scoliosis, seizures and gastrointestinal disorders (Percy, 2016). Irrespective of the great individual variation, all girls and women rely largely upon support in daily activities, which has consequences for families, the health care system and society.

One of the first persons diagnosed with RTT that I met as an Occupational Therapist and Visual Art Therapist, had an accompanying person with her that said during the assessment of the woman's ability to perform activities, "She can't do anything". This could be interpreted in two ways; either that I as the therapist couldn't change anything or that the person with RTT wasn't capable of doing anything. The accompanying person wanted to protect their girl/woman from having to try to perform tasks that were, for her, impossible. This happened in Östersund in 2008 at The Swedish National Rett Center today named National Center for Rett syndrome & related disorders (<http://www.nationelltcenter.se>).

The statement "She can't do anything" prompted me to ask the question: What does she like doing? It was also the starting point for my increased interest in what individuals with RTT do in their daily lives at home, since activities occur all the time (Reed, Hocking, & Smythe, 2010). Everyday activities and special occasion activities also influence the daily life of individuals with RTT, and that is something to take advantage of in supporting a good life for the individual with RTT.

As occupational therapy indicates, activities are valuable resources for exploration and learning, for joy and sharing, and for lifelong development (Law, 2002) and the primary goal of occupational therapy (OT) is to enable people to participate in the activities of everyday life (<http://www.wfot.org>). A focus on what people do is central to health (Wilcock, 2007). Key findings in a literature review of occupation and health describes that personal, social and other factors interact in complex ways to mediate the influence of occupation on health. A deeper understanding of the relationship between the activities that people with RTT take part

in and their well-being is of essential importance to promote good quality of life for them (Baum, 2003; Epstein et al., 2015).

This thesis is occupation-centred (Fisher, 2013) and has been conducted with an occupational perspective. In other words, it focuses on the activities people do and the significance that they attach to what they are doing and the context in question (Njelesani, Tang, Jonsson, & Polatajko, 2014). This thesis is inspired by occupational science theory (Pierce, 2012; Wilcock, 2001; Yerxa, 1990), occupational therapy (Fisher, 2013; Hocking & Wright-St Clair, 2011; Kielhofner, 2007) and ten years of clinical experiences of RTT (<http://www.nationalcenter.se>).

INTRODUCTION

Rett syndrome

Background

Rett syndrome (RTT) is a rare neurodevelopmental disorder primarily affecting females. It occurs worldwide and appears in approximately 1 in 9,000 female births (Fehr et al., 2011). RTT may also be found in males, although even more rarely and prevalence is unknown (Reichow, George-Puskar, Lutz, Smith, & Volkmar, 2015). The cause of RTT is usually loss-of-function mutations in the *Methyl-CpG Binding Protein 2 (MECP2)* gene on the X-chromosome (Amir et al., 1999; Percy et al., 2010). The syndrome is considered to be the most common cause of severe intellectual disability in females after Down syndrome (Gold, Krishnaraj, Ellaway, & Christodoulou, 2017).

During the foetal period, development of the baby appears normal, and the female babies are generally born full term and start to develop normally. At the age of 6 - 18 months, subtle signs and symptoms such as placidness, decreased muscle tone, lack of crawling, inability to take support on the feet, inappropriate hand, arm and mouth movements and loss of interest in play and communication may appear. Loss of skills/regression may start insidiously and progress slowly or rapidly (Hagberg, 2002; Witt Engerström, 1987). Movements become poorly coordinated and the girls seem unresponsive and frightened. Voluntary movement diminishes, and involuntary movements become obvious, twisting and patting initially mixes in with normal hand use, gradually including mouth-hand movements, tongue movements and tooth grinding. Trunk control deteriorates, and dystonic postures appear (Kerr & Witt Engerström, 2001). Loss of day-night rhythm and sudden, intense screaming for hours are a big burden. Regression particularly involves loss of expressive language skills, and the purposeful hand use changeover to stereotypic hand behaviour described as repetitive hand mouthing, hand wringing/clasping, hand clapping or finger rubbing (Dy et al., 2017).

Development usually begins to stabilize at 30 - 36 months of life, called the stabilization phase (Neul et al., 2010). The girls often seem to “wake-up”, to come back into contact and their cognitive and motor development may continue somewhat, parts of words and communication return on an immature level. Respiratory dysfunction and muscle dystonia appears as do swallowing dysfunction, gastrointestinal and nutritional problems with severe

constipation, and seizures - epileptic and non-epileptic. These are all problems that have to be understood, treated and prevented during the entire lifetime (Kerr & Witt Engerström, 2001).

Diagnosis

The diagnosis of RTT is based on clinical criteria. The essential diagnostic criteria required for both classic RTT/typical RTT and variant RTT/atypical RTT are the regression phase and the stabilization phase (Table 1) (Neul et al., 2010). The clinical diagnosis may be confirmed by a genetic test.

Diagnostic exclusion criteria include: brain injury, perinatal or postnatal trauma, neurometabolic disease, severe infection involving neurological functions and grossly abnormal psychomotor development in the first six months of life (Neul et al., 2010).

Table 1. The 2010 clinical criteria for classic/typical RTT and variant/atypical RTT.

Requirement for classic RTT also called typical RTT	<ol style="list-style-type: none"> 1. A period of regression followed by stabilization and recovery 2. All main criteria (and all exclusion criteria) 3. Supportive criteria not required although may be present
Requirement for variant RTT also called atypical RTT	<ol style="list-style-type: none"> 1. A period of regression followed by a stabilization and recovery 2. At least 2 of the 4 main criteria 3. 5 of the 11 supportive criteria
Main criteria	<ol style="list-style-type: none"> 1. Partial or complete loss of acquired purposeful hand skills 2. Partial or complete loss of spoken language 3. Dyspraxic gait or inability to ambulate 4. Stereotypic hand movements: hand mouthing, hand wringing/clasping, hand clapping, or finger rubbing
Supportive criteria for variant RTT/atypical RTT	<ol style="list-style-type: none"> 1. Periodic breathing during wakefulness 2. Bruxism while awake 3. Altered sleep pattern 4. Abnormal muscle tone 5. Peripheral vasomotor disturbance 6. Scoliosis/Kyphosis 7. Growth failure 8. Small cool/cold hands and/or feet 9. Inappropriate laughing or screaming spells 10. Delayed or diminished response to pain 11. Intense eye communication or “eye pointing”

Rett syndrome in everyday life

The characteristic signs of RTT appear in a predictable sequence, affecting growth, movement, cognition and autonomic control. They may lead to postural difficulties, joint

deformities, nutritional and feeding problems, seizures, epileptic and non-epileptic, and strange behaviour. Although there are apparent intellectual difficulties, the girls look alert, and seem to see, hear, feel and enjoy experiences, they continue to relate to other people and interact with them at an emotional level (Kerr & Witt Engerström, 2001).

There is a wide variety in the composition and degree of the symptoms between the individuals with RTT and also in how they progress over time, but all individuals need lifelong support. The *partial or complete loss of acquired purposeful hand skills* and the *stereotypic hand movements* (Table 1) cause difficulties in controlling movement, for example playing with toys and using cutlery and other tools (Qvarfordt, Engerström, & Eliasson, 2009). Washing themselves, toileting and dressing are considerably problematic as well (Cass et al., 2003; Downs et al., 2010). Feeding problems may also include manipulation of food in the mouth, swallowing issues, gastroesophageal reflux, delayed gastric emptying, constipation, and gallbladder dysfunction (Percy, 2016). Effects of *bruxism/tooth grinding* may influence dental health and eating. International guidelines have been published to facilitate nutritional and digestive health (Leonard et al., 2013).

Because of *partial or complete loss of spoken language*, parents and supporting staff are usually the informants on behalf of the individuals. Research shows that the girls/women frequently use eye gaze for communication and parents report that the girls/women also express pleasure and discomfort and make requests and choices through vocalizations and body movements (Urbanowicz, Downs, Girdler, Ciccone, & Leonard, 2016; Urbanowicz, Leonard, Girdler, Ciccone, & Downs, 2016). It is nonetheless difficult to evaluate cognitive capacity.

Intense eye communication or “eye pointing” may improve an individual’s ability to express and communicate. Use of Augmentative and Alternative Communication (AAC), for example gaze-based assistive technology, may support communication (Wandin, Lindberg, & Sonnander, 2015) as proposed in the International guidelines of communication and RTT. (Townend, Wandin, Barlotta, Urbanowicz & Curfs, 2018).

Dyspraxia, including inability to ambulate hampers initiating of activities and mobility and causes dependence on support for moving indoors and outdoors. Many girls and women with RTT are able to sit independently and about half walk independently or with some degree of support (Larsson, G., Lindström, & Witt Engerström, 2005; Stahlhut, Downs, Leonard, Bisgaard, & Nordmark, 2017). Support in standing up and starting to walk is common as is

the use of assistive technology devices in daily care and physical activities, including for walking (e.g. foot orthoses), standing (e.g. standing shells or standing frame), changing body position (e.g. ceiling or mobile hoists) and wheelchairs. There is a high propensity to bone fracture, which requires vigilance in all activities (Jefferson et al., 2016).

An *Abnormal muscle tone* may impact range of motion, physical mobility ability and, in turn, everyday life activities. RTT comorbidities such as *scoliosis or kyphosis* are common and affect girls'/women's activities to a varying extent. One study showed that scoliosis frequency and severity increase with age and more than 85 % of people with RTT show signs of scoliosis by the age of 16 (Killian et al., 2017). Bracings may be used as support for moderate curves, surgery is recommended for severe curves. Scoliosis surgery is an example of a health care intervention that has positive effects on daily activities (Larsson E.-L., et al., 2009). International RTT guidelines exist for the management of scoliosis (Downs et al., 2009).

Altered sleep patterns such as waking up at night is the most prevalent sleep problem, affecting over 80% of individuals with RTT (Boban et al., 2016). A connection between sleep disturbance and severe seizures was also described by Boban et al.

Periodic breathing during wakefulness interferes with or interrupts activities, and it might be visible or barely noticeable. Breathing dysfunction such as apnoea, hyperventilation, breath holding, deep breathing, and valsava breathing may also interfere with on-going activities and affect the metabolism of the body (Julu & Engerström, 2005; Julu et al., 2008).

The importance of noticing an individual's pain, and whether it is a *delayed or diminished pain response* is emphasized. In a recent study, pain responses were tested using measurement instruments (O'Leary et al., 2017) and individuals with RTT appeared to experience pain (Barney, Feyma, Beisang, & Symons, 2015).

Nutritional, physical and occupational therapies and overall management strategies have been presented as reasons for over 50% of females with RTT surviving until the age of 50 (Percy, 2016). The ability to walk with or without assistance; maintenance of adequate weight; and effective seizure control all promote survival (Tarquinio et al., 2015). There is on-going research into RTT comorbidities, the diversity of the syndrome, relations between genotype-phenotype correlations, and to find cures and better alleviation of the symptoms of RTT (Gold et al., 2017; Halbach et al., 2013; Leonard, Cobb, & Downs, 2017).

Rehabilitation

An individual diagnosed with RTT needs interventions throughout their life. Research and new knowledge about relations between genotype-phenotype correlations have increased our understanding of the syndrome diversity and variation, but there might be a risk for too low expectations on development and learning in individuals with RTT (Halbach et al., 2012). We have to be mindful that no one person with a specific diagnosis like RTT is the same as another, and they all need rehabilitation (Ronen & Rosenbaum, 2016). Individual goal setting and use of an enriched environment with interventions is required in order to create the conditions to reduce the functional deficit due to RTT (Downs et al., 2018). Interventions may include: cognitive engagement; sensory stimulation; social stimulation; timing of activities and gross motor practice and exercise (Downs et al., 2018). Another study highlights the need to reduce sedentary time by developing tailored interventions and increasing physical activity, not least for adults and the elderly (Stahlhut, Downs, Aadahl, et al., 2017). There is a need for early and regular intervention, preventive management and an individualized approach at every age, even for adults and the elderly with RTT (Anderson, Wong, Jacoby, Downs, & Leonard, 2014; Halbach et al., 2008). Parents and all professionals supporting individuals with RTT also need support to be able to cope (Lamb et al., 2016; Retzlaff, 2007).

OT interventions may increase participation for the RTT individuals. Occupational therapists use occupation as the primary therapeutic agent as well as a goal of intervention (Fisher, 2013). The opportunity to take part in activities differs substantially between girls and women with RTT due to a range of reasons that are not solely related to the degree of severity (dyspraxia, latency, limited communication skills, sitting-standing-walking capacity, breathing disturbances, seizures, stereotypies). Other reasons may include; age, interests, motivation, earlier experiences, support and network, aids, financial situation, place of residence, health care, school or daily work resources (Andrews, Falkmer, & Girdler, 2015).

Support and service

The Swedish Act on Support and Services for Persons with Certain Functional Impairments (known as the LSS in Sweden) is an entitlement law and encompasses supplementary support for persons with significant and long-term functional disabilities (LSS, 1993:387, 2001). The law applies to people who, as a result of other serious and permanent functional disabilities

that are clearly not the result of normal ageing, have considerable difficulties in everyday life and a great need of support. Girls and women with RTT and their families can therefore apply for assistance. Clause 9 of the LSS includes ten forms of assistance such as personal assistance for dressing, managing hygiene, eating or communication with others (LSS,1993:387, 2001). The LSS provides the opportunity to apply for support that would enable participation and social inclusion beyond the home (Boren, Granlund, Wilder, & Axelsson, 2016).

Theoretical framework

In occupational therapy, the concepts of both occupation and activity are commonly used (Müllersdorf & Ivarsson, 2011). In the two studies included in this thesis, the concept of activity was used, but there is a need to use both concepts in reflection and discussion. The concept of activity can be derived from Activities of Daily Living (ADL) (Jonsson, 2008; Tornquist & Sonn, 2014). International OT research and occupational science almost always use the term occupation. Research is also on-going in which topics are connected to occupation and not the term activity, for example, justice and deprivation (Kosma, Bryant, & Wilson, 2013; Nilsson & Townsend, 2010).

Activity and occupation

In occupational therapy, the concept of activity is used to describe activities in general, and the concept of occupation is when people are involved, doing activities and describing experiences and what they mean for them (<http://www.arbetssterapeuterna.se>).

In the two studies included in this thesis, the concept of activity was selected to describe daily life for individuals with RTT. The International Classification of Functioning, Disability and Health (ICF) has a commonly-used description of activity: “Activity is the execution of a task or an action by an individual” (World Health Organisation, [WHO], 2001). This description emphasises performance and only includes the idea that activity is a process for one person. Doing an activity can also be a process when two or several people are involved, people may choose to do something with others as well as to do something for others (Hammell, K.,

2014). The concepts of activity and occupation are not synonyms, but they are used in similar contexts. A definition from Christiansen and Townsend describes these similarities well and this definition of activity is used for the thesis: “Activity(ies) is an observable unit of behaviour, and recognizable sequence of actions taken together in a particular context; beyond tasks yet without the complexity of occupations in the simple to complex hierarchy of tasks, activities and occupations” (Christiansen & Townsend, 2010, p. 417). One can say that the concepts of task, activity and occupations overlap each other and that the concept of occupation is the overarching concept of the three. Occupation has also been described as when a human being transforms a simple idea of doing into a complete occupation consisting of numerous actions intertwined with each other in complicated patterns (Bendixen et al., 2006, p. 4). Occupation occurs at family, community, national and global levels, and for cultural, social and political purposes, and as Wilcock described – we are, in fact, all occupational beings (Wilcock, 2001, p. 413).

Infants, children, teenagers, young adults, adults and the elderly, are all human occupational beings but the concept of occupation has rarely been described in connection with a severe disability. In a study of children with autism and other developmental disabilities as occupational beings, occupation was defined as “a set of directed actions connected by physical movement, materials, space or purpose within a time period, in a way that is meaningful to the individual executing them” (Spitzer, 2003, p. 72). This definition highlighted children with disability as occupational beings from an occupational science perspective. In the present thesis the choice of definition of the concept of occupation was based on that fact that the definition needed to be applicable regardless of age, illness or autonomy. The following definition was selected: “All that people need, want, or are obliged to do; what it means to them; and its ever-present potential as an agent of change, it encapsulates doing, being, and becoming” (Wilcock, 2006, p. 343). In this definition the “doer” is not one single person, it is a ‘we’. So, this definition suits the purposes of the thesis well because individuals with RTT are highly dependant on another person’s help and support in order to participate in almost any activity.

Occupying one’s own life, deciding and controlling what is possible to do in all different situations is something that even people with a life-long need of support desire (Hammel, J., et al., 2008; Hammell, K., 2004). It is impossible to predict what the potential of the process ‘to be, to do, to become and belong’ means for exploration and discovery, however this process is a prerequisite for all humans to develop (Hitch, Pépin, & Stagnitti, 2014).

Time use

When the concept of time is used it may include people, placements and the context of a past, present or the future (Pemberton & Cox, 2011). The activities of all people are performed in contexts in relation to time and have a relationship to health, as illustrated in occupational therapy theories (Hunt & McKay, 2015a). There is a connectivity between doing, being, rhythm and balance and time is a vital aspect of occupation (Pemberton & Cox, 2011). Peloquin (1991) regards the connection between time and occupation in OT as fundamental; people act in time. It is expected that individuals with RTT will grow up to be adults and elderly and also spend time (Tarquinio et al., 2015). RTT research exists presenting statistics about life span and mortality but little research contains content regarding how life is spent (Anderson et al., 2014; Cianfaglione et al., 2015).

A Time-geographic approach

Researchers in time geography also emphasise the importance of time as fundamental as well as a resource. All people's lives and every activity are performed in a time-space context (Ellegård, 1999). The approach Time Geography is applicable in many research topics and is described as an interdisciplinary field, though it originated in human geography (Persson & Ellegård, 2012). Its space-time dimension may, for example, facilitate the understanding of research concerning human life and activities and health (Hunt & A McKay, 2015a; 2015b). Investigating individuals' activity patterns may also clarify the occupational balance of activities for stimulation, development, rest and recovery (Christiansen & Townsend, 2010, pp. 57-74). The time-geography methodology is relevant for studies of individuals' daily life activities and displays different geographical (physical places) and social environments as well as the time use component (Hunt & McKay, 2015a). Usually the diary is self-reported but, in this thesis, (study II) informants were used as diary writers, in the form of parents/staff.

The time-geographical diary method is derived from time geography and is a qualitative method with the possibility to present quantitative data through charts and visual graphs (Orban, Edberg, & Erlandsson, 2012). The method was developed in the late 1980s and early 1990s (Ellegård, 2006). An example of a visual graph is presented in Figure 1.

Figure 1. Example of a graph visualising a person's time use over a 24-hour period (a Saturday). Time spent on activities is shown to the left. The vertical sections in Activities show the time use and the horizontal sections indicate that the participant stops one activity and starts another. Physical location e.g. where the activities took place is described under Locations and social companionship is described under With whom. Finally, the participants' responses during activities are shown in Responses.

The methodology has no predefined time-slots; it is flexible to the individual's unique everyday life. The method aims to register all 1440 minutes of a day through the diary notes. It shows occurrence and frequencies of the diary variables (the headlines) by presenting percentage and number of minutes, or through visual graphs. If data is missing the quantitative chart's summary of the minutes shows the mistakes and the coder must go back and correct the missing data.

The method's coding system has been discussed and compared in a study by Ellegård (Ellegård, 2006). In that study, the empirically generated category scheme of the seven activity spheres was compared with the analytically derived category scheme used by Statistics Sweden for coding diaries in the Swedish national use surveys. The study demonstrated that a time-geographical diary method might be useful in occupational science studies because it comprises an entire day, it presents more in-depth results, and it appears to be useful for individual rehabilitative purposes as well as for describing everyday life at an individual, household and population level, as well as for democratic dialogue between policy makers and citizens (Ellegård, 2006). Occupational therapy researchers have previously used

this method to study different activity areas and participants' everyday activities (Björklund, Erlandsson, Lilja, & Gard, 2015; Kellegrew & Kroksmark, 2000; Kroksmark & Nordell, 2001; Liedberg, Hesselstrand, & Henriksson, 2004; Nilsson, Blanchard, & Wicks, 2013; Orban et al., 2012).

Activity pattern

Activity or occupational patterns are concerned with how people organise their tasks, activities and occupations (Bendixen et al., 2006). A week's activities are often planned especially during school or working time. However, it is when looking back at what we did and how time was used that the final activity pattern will become visible (Orban et al., 2012). In a review of the linkage of the two concepts 'patterns of daily occupations' and 'occupational balance' the authors described the concepts as separate but interacting phenomena (Eklund et al., 2017). An individual's pattern of daily occupations is influenced by time, space, and an arrangement of temporal and spatial elements connected with experiences. It is also influenced by the social context in which the individual lives and acts. The individual capacity or skills to participate in and perform activities was not described as influencing patterns of everyday activities in this review, as is the reality for those diagnosed with RTT. For girls and women with RTT, it is very difficult for them to participate in activities such as communicative and social activities, getting fed and moving or walking outdoors, without support. Problems in initiating body movement are common due to latency, dyspraxia or stereotypic movements, and they therefore need support and time to increase their chances of participating in an activity (Downs & Leonard, 2016). One of the interesting points from the review was the linkage between the more objective concept 'patterns of daily occupation' and what was described as the more subjective concept, 'occupational balance' (Eklund et al., 2017). It is up to the individual to decide if their patterns of activities also feel balanced or not. In this thesis the following description of activity pattern was used: "The building blocks of the activity pattern are recognizable sequences of actions and activities" (Bendixen et al 2006, p. 8).

RATIONALE

Irrespective of health or illness humans are occupational beings (Hammell, K., 2015) and have similar needs and rights (<http://www.ohchr.org>). The Convention of the rights of people with disabilities provides that a citizen has the right to full and effective participation and inclusion in society, for example to get education and health care, Sweden International Agreement SÖ 2008:26 (<http://regeringen.se>) People with a severe neurodevelopmental disorder such as RTT possess civil rights but cannot assert their own rights.

Studies of RTT with an occupational approach and/or OT intervention are few. An occupational therapist contributed to one RTT multidisciplinary research study that confirmed that RTT is not a degenerative condition and indicated that interventions and support to maintain and increase motor skills, daily living skills, and cognitive and communicative functioning are appropriate goals for girls and women with RTT (Cass et al., 2003). Other early research focused on elbow splints, hand splints and feeding skills (Kubas, 1992; Sharpe, 1992; Sharpe & Ottenbacher, 1990). Research into how to promote eating through the use of guided eating or feeding has been carried out (Qvarfordt et al., 2009), as well as in the use of equipment and respite services in Australia (Urbanowicz et al., 2011). More recent research has focused on communication abilities and the use of eye gaze and gestures to communicate (Urbanowicz, Downs, et al., 2016; Urbanowicz, Leonard, et al., 2016). A mapping of skin injuries and joint contractures of the upper extremities was recently presented from Japan (Hirano & Taniguchi, 2018) and from USA a first colour tracker was developed to monitor hand and arm motion of a woman with RTT (McAmis, Foreman, Himmelrich, Diener, & Engsberg, 2017). Since RTT is a rare disorder, research that includes other diagnoses has also been conducted, such as a review of community participation interventions (Andrews et al., 2015). Other health care professionals' interventions may also improve the RTT individual's capacity to participate in activities as described by Stahlhut (2018) as well as scoliosis surgery that showed positive effects for participating in daily activities (Larsson, E.-L., et al., 2009).

Thus, there is a need to investigate and increase the knowledge of activities in everyday life for people with RTT. For the most part, the activities that individuals with RTT like to do, and what they may experience or receive support to take part in, remains unknown. Knowledge of activities and RTT may facilitate and hopefully improve the quality of life for people with RTT.

AIMS

The overall aim was to describe the everyday lives of female individuals with Rett syndrome.

The specific aims were:

To investigate over time the activities that girls/women with RTT liked or did not like to do (I).

To investigate and describe how teenagers and young adult females with RTT spent their time during a period of seven ordinary days as communicated by parents or support staff, how much time they spent doing the activities identified, in which locations the activities occurred and with whom. In addition, the study captured the responses that were observed during activities, in the form of visible and/or audible reactions expressed by the teenagers/young adults (II).

METHODS

This thesis is based on two descriptive studies, one questionnaire study (I) over a time period of 15 years, and one diary study (II) containing a week's activities for the participants (Table 2).

Table 2. Overview of the two studies.

Study	I	II
Research design	Descriptive study of secondary data from questionnaires	Descriptive study with a diary method
Sample selection	Available questionnaires	Random sampling
Inclusion criteria	Swedish citizenship, female gender of any age, clinical diagnosis of RTT. The selected questions to study from the questionnaires had to have been answered.	Swedish citizenship, female gender, diagnosis of RTT, born between 1983 and 1988 (young adults) or between 1993 and 1998 (teenagers). Parent/s as custodians.
Participants	175 girls/women with RTT. Age, years: mean, $\pm$ SD, (range) Q1: 19, $\pm$ 12, (2-54) Q2: 26, $\pm$ 14, (2-66) Q3: 27, $\pm$ 14, (2-66)	10 teenagers/young adults (females) with RTT. Age, years: mean, $\pm$ SD, (range) 4 teenagers, 18, $\pm$ 1.9, (15-20) 6 young adults, 27, $\pm$ 1.7, (25-30)
Informants	Parents/staff n, total: 365 Q1: 207 (111 mothers, 64 fathers, 32 staff). Q2: 96 (35 mothers, 16 fathers, 45 staff). Q3: 62 (22 mothers, 6 fathers, 34 staff).	Parents/staff n, total: 63 10 mothers, 9 fathers and 44 staff
Data collection	Three questionnaires were used. Q1: The Swedish Rett Center survey 1996. Q2: Cross-professional investigation, years 2008-2011 Q3: Occupational therapy questionnaire, years 2009-2011	Time-geographical diaries were used. Parents and staff involved selected their diary week containing seven days during a period of an ordinary week. Time: May 2013 until January 2015.
Data analysis	Descriptive statistics Content analysis	Descriptive statistics Software VISUAL-TimePacTs computer program, DAILY LIFE version 2011

Sample selection

For study I secondary data from three earlier questionnaires at the Swedish National Rett Center (since 2017 named National Center for Rett syndrome & related disorders) was used.

For study II a random sampling (Polit & Beck, 2012) was used. The Swedish National Rett Center medical files were used to sample the females diagnosed with RTT, born between 1983 and 1988 (young adults) or between 1993 and 1998 (teenagers). The names of all females were written on pieces of paper, teenagers and young adults separately, which were then placed upside down and mixed. Every fifth selected RTT female teenager and young adult fulfilling the inclusion criteria was contacted by letter. One request was sent to the families. Families were contacted by letter sequentially until data for ten individuals with RTT was collected. A total of 18 families were contacted.

Inclusion criteria

Study I: Swedish citizenship, female gender of any age, clinical diagnosis of RTT, and that the selected questions to study from the questionnaire had been answered.

Study II: Swedish citizenship, female gender, diagnosed with RTT, born between 1983 and 1988 (young adults) or between 1993 and 1998 (teenagers), and having their parent/s as custodians.

Participants and their informants

None of the participants in this thesis, girls/women, teenagers/young adults with RTT could answer a survey or write diary notes for themselves, therefore mothers/fathers and staff were used as informants. A brief overview of the characteristics and demographic data of the female RTT participants and their informants, parents and/or staff, is shown in Table 3.

Table 3. Characteristics and demographic data.

Study	I (n)	II (n)
Females with RTT, n	175	10
Age, years: mean, (range)	21 (2-66)	23 (15-30)
Mobility		
Can walk	79	1
Moves short distances with support and/or aids	21	1*
Uses a wheelchair	68	9
Unspecified or unanswered	7	-
Type of housing		
Parental home, n	113	8
Group accommodation, n	47	1
Own flat, n	15	1
Informants, total, n,	365	63
Mothers, n	168	10
Fathers, n	86	9
Staff, n	111	44

* Is pushed in a wheelchair for longer distances.

The study I participants were clinically diagnosed with RTT. Medical file data for study II participants showed a clinical diagnosis of classic RTT/typical RTT and for nine participants mutations were also confirmed (Table 4).

Table 4. Data concerning genetic investigations of nine of the ten participants in study II.

Data of mutations	Teenager or young adult, n
p.R168X	1
MECP2 deletion 1084-1197 (del 113)	1
p.R270X	2
p.R255X	1
Large MECP2 deletion from exon 4 (c.1396) into 3'UTR (c.670) and c.1461+5349).	1
p.R106W	1
p.Gln128X	1
p.R306C	1

Data collection

Questionnaires

Secondary questionnaire data was used for the first study (I). The first questionnaire (Q1) provided data on 123 girls/women with RTT, the second (Q2) on 52 and the third questionnaire (Q3), on 39. The 39 girls/women were also among the 52 girls/women in the Q2 questionnaires (total participants 123+52=175). Informants were parents and/or staff, in total 365 informants. The open-ended questions related to activity were selected from the questionnaires and data from a period of 15 years was studied.

Q1 was the Swedish Rett Center Survey developed with the aim of contacting all custodians in Sweden of females with RTT to follow-up already known girls/women and newly diagnosed RTT cases. The survey was also aimed at collecting data for further RTT research (Larsson, G., et al., 2005). The survey (Q1) was sent to all females that were known to have RTT at that time. This amounted to 178 persons in Sweden and 125 (70%) of the custodians responded. Of those 123 answered the question regarding liked activities and 80 answered the question concerning disliked activities (Table 5) used in study I.

The Q2 questionnaire was used to gather information prior to cross-professional assessments at the center, and the Q3 questionnaire was used during the week of cross-professional assessments and used by the occupational therapist. The response rate for Q2 was 95%, and for Q3 67% answered the question (Table 5).

Table 5. Questions included from the three questionnaires.

Questionnaire	Data from year/s	Open-ended question/s studied
Q1	1996	What does she prefer and like to do, most of all? What does she not like to do?
Q2	2008 - 2011	What does she like to do?
Q3	2009 - 2011	What activities do you like to do with her?

Time geographic diaries

Patterns of activities were assessed using time-geographical diaries (Ellegård 1999, 2006). The usefulness of this method has been tested in previous studies, aimed at facilitating reflection on patterns of daily activities/patterns (Liedberg et al., 2004; Nordell, 2002).

Activities at home and at school, and activities during daily work, leisure time and night time were described and collected in notebooks (diaries) during a period of seven days for 24 hours per day. The diaries had an open format with the following prepared headings; *time*, *activity*, *location* and *with whom* (Ellegård, 1999). In addition, visual and/or audible reactions were described (response words) inspired by Slåtta (Horgen, Slåtta & Gjermestad, 2010). Finally, there was space for the informants' own comments. Headlines can be added depending on the focus and aim of the study.

A pilot study was conducted with one adult with RTT, along with her family and support staff. It facilitated the choice of the diaries' pre-defined headlines of response words and enabled a pre-understanding that many people would be involved as informants. The result of the pilot study was not included in the diary study (II).

Before filling in the diaries, the informants were asked to describe in their own words how they interpreted the response words used in the diary; engaged, interested, uninterested, and opposed (Table 6). In that way all involved informants in one family had the opportunity to discuss and reach a sense of consensus about the response words before starting to fill in the diaries, which may act to strengthen the internal validity (Merriam & Tisdell, 2015) of the response words used.

Table 6. Informants' descriptions of the four response words.

Response words	Descriptions
Engaged	Body: Relaxed, engaged and participates, active and makes contact, smiling Sound: May laugh Eyes: Open eyes, intense glance, looking intently at what she likes
Interested	Body: Leans forward, wants to get closer, seems satisfied Sound: No sound but seems to be listening and following the conversation Eyes: Looking on with interest
Uninterested	Body: Passive, apathetic Sound: May whine somewhat Eyes: Sparse eye contact, looks away, closed eyes
Opposed	Body: Kicks with legs, writhes, beats with arms. Resists. Turns away Sound: Makes a lot of sounds, may cry or scream Eyes: May have intense eye contact, looks scared

The diary week was self-selected by the family. Parents and staff involved chose their diary week containing seven days in an ordinary week i.e. not holidays. All diary notes were written in the informants' own words during activities and when the informants assisted the teenager/young adult with RTT. When observing a response to an activity, the informants

made a check mark under one of the four available response word alternatives in the diary, or wrote their own comments. Once the diary week was complete, the diaries were returned by mail. Data collection took place from 20 May 2013 until 11 January 2015 (Table 7).

Table 7. Time period for the self-selected diary weeks.

Year	Jan	Feb	Mar	Apr	May	June	Jul	Aug	Sept	Oct	Nov	Dec
2013					T T	T					Y Y	
2014		T	Y						Y		Y	
2015	Y											

T = teenagers, Y = young adults

Data analysis

Descriptive statistics were used in both studies to present demographics and frequencies of activities and time use. For study I, the Statistical Package for the Social Sciences SPSS 19.0 (IBM Corp., Armonk, NY) was used. For the second study (II) the software VISUAL-TimePAC's computer program, DAILY LIFE version 2011 was used for calculating frequencies and time use.

For study I the open-ended questions about activities were analysed using a CA approach. Each question, two from Q1, one from Q2 and one from Q3 was analysed separately. All answers were copied, anonymised and read. The short sentences and words were considered as manifest data. Identified verbs and/or described activity contexts were marked in code units and subcategories were sorted by hand by using post-it notes (Polit & Beck, 2012).

Informants were free to report any number of activities. The development of subcategories and categories concerning activities was evolved through continuous discussions between three of the four authors during the analysis process. Similarities and differences between the subcategories were analysed and finally resulted in three categories shown in Table 8.

Table 8. The three categories emerging from the result and their stipulated definitions.

Categories	Definitions
Being in motion	Activities involving body motions and experiences of being moved
Receiving impressions	Activities that provide sensory stimulation while requiring little physical effort
Having contact	Affirmative activities in a communicative context that involve and include people

A Time-geographic diary method

In study II the diaries were first coded using the numerical codes of the seven activity spheres included in the diary computer method, using the software VISUAL-TimePacTs computer program, DAILY LIFE version 2011. The coding scheme has seven main categories: Care for oneself; Care for others; Household care; Recreation; Travel; Procure and prepare food; and Gainful employment (Liedberg et al., 2004). It is also a hierarchical structure with five levels of detail of activity codes that facilitate coding of detailed diary notes and overall descriptions (Ellegård, 1999; Ellegård & Nordell, 1997). The coded diaries' data was converted into charts and graphs, in line with the time geographic diary method. Similarities and differences between the time-use data for weekends and weekdays were noted at a group level.

Location codes were also used with the numerical codes included in the program as well as individual codes created by the first author, such as location for daily work. Individual codes were created in every diary to register with whom the activities were performed during the days. Examples of individual codes included father, mother, parents, siblings and staff.

Numerical codes for responses to activities were created for this study as follows: 1 Opposed, 2 Uninterested, 3 Interested, 4 Engaged, 5 Own comment, and 6 No comment at all. Then the data was converted into charts and graphs.

Thereafter, contact was made with each family followed by a visit in their homes by the first author of the study, in order to confirm the data collected from the diaries.

The 'own comments' notes were manually collected and briefly sorted into described general health conditions and notes that observed tiredness or happiness.

ETHICAL CONSIDERATIONS

The project including the two studies was approved by EPN, Umeå 29-05-2012, reference number 2012-204-31 Ö.

For study I in 1996, the returned and filled-in questionnaires were synonymous with the parents' informed consent. For the two other questionnaires used, the parents and staff decided themselves whether they wanted to complete the questionnaires both before and during the assessment days at the Swedish Rett Center. If parents or staff felt compelled to respond, then the responses can be seen to be less credible. However, the first author's comprehension during assessment was that the parents and staff wanted to describe the girl's/woman's health status and current difficulties.

In study II an introductory letter was sent out, and if the parents decided to allow their teenager or young adult with RTT to participate then the parents signed the consent form and sent it back. Further, the participants' parents were asked to inform their teenager/young adult about the diary week. Parents were encouraged to use the teenager's/young adult's communicative devices to inform the participant. To support parents in this, a picture-based AAC description (Appendix) was sent out with the introductory letter in the diary study, taking into account the participants' cognitive and significant communicative difficulties.

It is an ethical dilemma to have to ask someone else other than the person in question, and in turn, whether parents and staff see, understand and interpret the girl or woman with RTT truly and fairly. However, even if the informing parents and staff may fail to interpret and describe responses correctly, the people closest to the person with RTT were those chosen to ask questions about everyday life.

In all documentation in study II, the names and telephone numbers of the responsible research personnel were provided, which gave the parents the opportunity to ask questions, before, during and after the project. It was emphasised that withdrawal at any time, or a refusal to participate in the study, would have no effect on current or future contact with the center. The analysis and presentation of the data were done in a way that concealed the participants' as well as the informants' identity, thus, ethical considerations according to the Declaration of Helsinki (WMA, 2013) were followed. Further, all data was stored in a locked place to which only the first author had access.

RESULTS

The individuals with RTT (the participants) were referred to as girls/women (I) and teenagers/young adults (II) in this thesis, and their parents/staff were referred to as the informants.

Study I: Liked and disliked activities

The results of Q1 showed that the informants for girls/women with RTT reported the most frequently enjoyed activities as being bathing/swimming, listening to music, being outdoors/walking and horse riding. In 2008 - 2011 activities such as listening to music, bathing/swimming, being outdoors/walking and watching TV/DVDs/films were the most reported enjoyed activities. Figure 2 shows the twelve most frequently reported liked activities over time (Q1, Q2) presented as a percentage and according to these categories: (m) being in motion, (i) receiving impressions, (c) having contact. Differences in activities between 1996 and 2008 - 2011 were small.

Figure 2. The twelve most frequently reported activities over time that the girls/women with RTT liked to do, presented as a percentage.

Informants were free to report any number of activities. Table 8 shows the three categories and subcategories of activities that girls/women with RTT enjoyed (Q1, Q2) and activities the informants liked to do with their girl or woman (Q3), as reported by parents and/or staff.

Table 8. Occurrences and frequencies of liked activities from the three questionnaires.

Q1, 1996 Activity statement, n, (%)	Q2, 2008–2011 Activity statement, n, (%)	Q3, 2009–2011 Activity statement, n, (%)
BEING IN MOTION		
Bathing/swimming, 72(59) Being outdoors/walking, 39(32) Horse riding, 37(30) Going out in a car, 19(16) Dancing, 18(15) Walking around, 12(10) Going to town, 12(10) Swinging/rocking, 6(5) Gymnastics/training, 6(5) Travel/holiday, 2(2) Sport events, 1(1)	Bathing/swimming, 22(42) Being outdoors/walking, 19(37) Horse riding, 10(19) Going out in a car, 7(14) Walking around, 6(12) Going to town, 6(12) Dancing, 4(8) Gymnastics/training, 4(8) Swinging/rocking, 3(6) Travel/holiday, 2(4)	Being outdoors/walking, 42(67) Bathing/swimming, 21(33) Going to town, 16(25) Dancing, 12(19) Going out in a car, 10(16) Horse riding, 8(13) Travel/holiday, 7(11) Swinging/rocking, 5(8) Gymnastics/training, 4(6) Walking around, 2(3)
RECEIVING IMPRESSIONS		
Listening to music, 48(50) Watching TV/DVD/films, 23(19) Eating food or snacks, 21(17) Relaxing/being cuddled, 12(10) Sensory stimulation/massage, 11(9) Computer, 11(9) Baking/preparing meals, 5(4) Cultural event, 3(2) Pets/animals, 3(2) Art and crafts, 2(2) Showering, 1(1) Hair care, 1(1) Dressing, 1(1)	Listening to music, 25(48) Watching TV/DVD/films, 19(37) Sensory stimulation/massage, 12(23) Eating food or snacks, 6(12) Pets/animals, 5(10) Computer, 4(8) Baking/preparing meals, 4(8) Relaxing/being cuddled, 4(8) Cultural event, 4(8) Art and crafts, 2(4)	Listening to music, 18(29) Relaxing/being cuddled, 18(29) Eating food or snacks, 13(21) Watching TV/DVD/films, 10(16) Baking/preparing meals, 8(13) Cultural event, 7(11) Art and crafts, 6(10) Sensory stimulation/massage, 5(8) Pets/animals, 3(5) Showering, 1(3) Hair care, 1(3)
HAVING CONTACT		
Being with family and friends, 23(19) Singing and playing music, 23(19) Receiving attention/closeness, 22(18) Playing and comical situations, 13(11) Talking/interaction, 12(10) Reading, 9(8) Looking at photos/pictures, 4(3) Parties/celebrations, 1(1)	Being with family/friends, 11(21) Singing and playing music, 11(21) Playing and comical situations, 9(17) Reading, 7(14) Talking/interaction, 4(8) Looking at photos/pictures, 4(8) Receiving attention/closeness, 2(4) Parties/celebrations, 2(4)	Singing and playing music, 28(44) Talking/interaction, 10(16) Sharing her happiness, 9(14) Receiving attention/closeness, 8(13) Playing and comical situations, 8(13) Being with family/friends, 8(13) Reading, 3(5) Looking at photos/pictures, 1(2)

Few disliked activities were reported (Q2) and answers such as ‘I don’t know’ were reported. The findings from Q2 showed that the activities linked to daily care such as hair care, dental hygiene, showering, going to the toilet and getting dressed were described most frequently as disliked activities. Further, from the same category ‘Receiving impressions’, sensory stimulation such as massage was described as a disliked activity. From the Being in motion category, gymnastics/training was described as disliked.

Q3 showed that the informants themselves enjoyed doing similar activities as were described as liked by the participants (Q1, Q2). The most frequently enjoyed activities to do together

with the participants were being outdoors/walking, singing and playing music, bathing/swimming, listening to music and relaxing/being cuddled.

Study II: Time use for activities during an ordinary week

For the ten participants in the diary study (II), most time (Figure 3) was spent in sleeping (range 8 h 24 min – 10 h 17 min) and rest during day time (range 0 - 8 h 8 min), daily care (range 2 h 13 min – 7 h 50 min), medical and health care activities (range 14 min – 5 h 2 min) and outdoor travel/transportation between places such as home and school or daily work (range 48 min – 2 h 41 min).

Figure 3. Activity categories and mean values of the time for the ten participants, presented in 24-hour averages in h (hours) and min (minutes).

On a 24-hour average, at a group level of the ten participants, little time remained for other activities, 4 h and 17 min, (range 1 h 57 min - 6 h 57 min). The remaining time was spent in the following ways: receptive (range 6 min – 2 h 32 min), physical (range 3 min – 2 h 8 min), social/creative (range 0 – 3 h 6 min), communication (range 0 – 1 h 43 min), school/daily work

(range 0 – 1 h 23 min) and domestic chore (range (0 – 48 min) activities. Overall, the teenagers spent more time doing these activities than the young adults.

When comparing weekend and weekdays at a group level the activities and time use were similar, apart from school/daily work activities that only took place on weekdays. Further, all weekday activities showed little differences at a group level (Table 9).

Table 9. Activity spheres and categories: mean value of the ten participants' time use over the weekdays and weekends, presented in 24-hour averages with h (hours) and min (minutes).

Activity sphere	Activity category	Mon-Fri	Sat-Sun
Care for oneself	Sleep	9 h 21 min	9 h 51 min
	Daily care*	6 h 04 min	6 h 47 min
	Daytime rest	0 h 52 min	0 h 53 min
	Physical activities	1 h 01 min	0 h 40 min
	Medical health care	1 h 44 min	1 h 32 min
Care for others	-	-	-
Household care and Procure and prepare food	Domestic chores	0 h 18 min	0 h 22 min
Reflection/Recreation	Receptive activities	1 h 04 min	1 h 37 min
	Communication	0 h 40 min	0 h 39 min
	Social/Creative activities	0 h 34 min	0 h 54 min
Travel	Travelling/Transportation	1 h 40 min	0 h 45 min
Gainful employment	School/Daily work	0 h 42 min	0 h 00 min

* Eating, dressing, hygiene, toilet and moving about indoors activities.

Time use for activities spent in different locations and with whom

Most time was spent at home. During the weekdays the teenagers were at home for 15 h per 24 h and the young adult for 18 h per 24 h. At the weekends teenagers were at home for 20 h and the young adults for 22 h per 24 h. It was rare for the participants to be in public environments and places outside of school or daily work, especially for the young adults. During the weekdays, it was sometimes not clear from the diary if an activity was a specific school, therapy, or leisure activity or a combination. The participants were outdoors for a mean of half an hour per day. They spent most of their time with staff, thereafter with families, and the least time was spent with friends.

Responses observed during activities

In the diaries short time expressions of responses were common during activities – as short as one minute occurred frequently – as was a variety of responses during the same activity. Data from the diaries showed the following mean values per day for responses: 5 h 33 min for ‘interested’, 2 h 55 min for ‘uninterested’, 2 h 20 min for ‘engagement’ and 57 min for ‘opposed’.

Responses of ‘engagement’ were noted during contexts of socialising, playing and communicating activities with families, friends or staff; when changing body position, for example moving in the water or gymnastics; during meals or when watching/listening to films, books and music.

Responses of ‘opposition’ were noted during the following activities; toileting, use of breathing mask, stretching, brushing teeth, being woken up, dressing and putting on orthoses. When the informants noted a comment beyond observed responses, it mainly related to the participant’s daily fitness such as for example being tired, constipation problems, a seizure, or notes of a generally satisfied expression.

DISCUSSION

This thesis focused on what girls and women diagnosed with RTT liked or did not like to do. It also examined the activities that RTT females, teenagers or young adults do during an ordinary week, and their responses during the activities.

Discussion of results

Liked activities included motion, contact and sensory impressions

The girls and women were reported to like activities that contained aspects of motion, contact and sensory stimulation (I), for instance, bathing, horse riding or going out in a car. In these examples it can be assumed that assistance and family/friends are physically close to the girl/woman and thus opportunity for contact may occur. These are also activities that have good opportunities for everyone do the same thing and share the moment. There is also a clear activity context of what is about to happen, for example a riding helmet and the stable, swimsuits and the pool, a specific seat in a familiar vehicle. This may provide a feeling of safety and give a pre-understanding of what is expected for the girl/woman. The activities in themselves include a high degree of movement and sensory input. Bathing/going swimming and horse riding are valuable activities, not least for minimizing sedentary time and increasing physical activity (Lotan & Barmatz, 2009; Stahlhut, 2018). RTT research emphasises that advancing age and reduced walking skills have been associated with longer sedentary time and efforts are needed to change this (Stahlhut, Downs, Leonard, et al., 2017). It is important to highlight that it was obvious that girls/women with RTT enjoyed activities that included aspects of contact, sensory impressions or motion despite dyspraxia, mobility and speech limitations (Neul et al., 2010).

Disliked activities may be connected to touch and sensitivity

Some of the enjoyed activities could also be disliked, and vice versa, by the girls/women but some activities were more often described as disliked (I). Activities that were reported as being unenjoyable (I) were linked to daily care activities such as hair care, dental hygiene, showering, going to the toilet and getting dressed. A few girls/women were also described as disliking gymnastics/training. The activities that were reported to elicit most responses of

opposition (II) were daily care activities and medical health care activities. These were activities such as: toileting, use of breathing mask, stretching, brushing teeth, being woken up, dressing and putting on orthoses. When comparing the disliked activities from study I, and the activities that elicited responses of opposition from study II, it appears that they share the fact that they involve touching of body parts that are sensitive. Therefore, the need for careful assistance needs to be emphasised as does being observant for signs of pain (Barney et al., 2015; O'Leary et al., 2017).

The ordinary week's activities

One of the main results from the diary study (II) was that a large proportion of time was spent for sleep and rest, daily care, medical health care activities, and to travel between home, school or daily work. It seems problematic that those activities were time-extensive, and time became sparse for other kinds of activities. In the two studies in this thesis, information regarding the specific health status or condition of the participants was not collected. It is therefore not known to what degree comorbidities such as seizures, sleep disturbances, scoliosis, restricted mobility, nutrition and gastrointestinal problems affected the everyday life of the participants of this study. However, these comorbidities are common among individuals with RTT (Percy, 2016) and this thesis is performed in a context of care-taking and managing health issues, for example seizures, nutrition, stomach functioning and performance of standing/walking and gymnastics. The informants' own comments in the diaries (II) were mainly related to topics such as tiredness due to a seizure or constipation problems that may immobilise the person. In everyday life for individuals with RTT, it is necessary to find the best solutions possible to manage these problems caused by RTT comorbidities, in order to optimize the possibilities for the individual to take part in activities for joy per se, and to increase wellbeing. Individuals with RTT have complex needs and require collaborative professional health care teams to find good strategies and solutions together with the girls'/women's parents and staff (Ronen & Rosenbaum, 2016).

The activities reported in study II were largely the same regardless of weekday. A monotonous and perhaps an institutionalised everyday life of nursing activities (Maes, Vos, & Penne, 2010) appeared from the diaries, for example, the 30 minutes per day outside and few activities away from home after dinner and during the weekend. This is not expected in people between the ages of 15 – 30 years, such as the participants. There is a need for

routines, not least when activity support is necessary as with girls and women with RTT, and when several people are part of supporting and assisting. In an RTT interview study from Australia, parents described their daughters' quality of life in relation to daily activities (Epstein et al., 2015). Parents of 21 girls and teenagers (7-18 years) reported that stability in daily routines were important and referred to sequences of activities that were consistently followed. However, the extent to which activities and daily routines must be the same and similar between weekdays and weekends must be questioned for every individual with RTT. It is also important to examine what determines how time is spent in their everyday life in relation to the activities the girls/women need, want and must do (Wilcock, 2006). It was also a somewhat surprising result that the same activities were enjoyed during childhood and adulthood (I). The joy from recognising an activity, and positive memories from earlier experiences of the activity, is important. However, if the same activities are used year after year it might be difficult for the individual to have new experiences and develop new interests.

Time spent on a variety of activities, places and people

A child, teenager or adult with a diagnosis of RTT needs support with the basic needs, but they may also need support with other activities and need something to look forward to. Time was spent mostly at home and with staff, especially for the young adults. There might be a risk for deprivation if an inactive lifestyle is what is expected after school years (Hryniewiecka-Jaworska, Foden, Kerr, Felce, & Clarke, 2016). At an individual level for a teenager or a young adult with RTT, there might be a greater need to provide opportunities for visiting many different places, meeting other people and doing a variety of activities to a higher degree (II) as they can not take such initiatives by themselves. It is important to increase time for enjoyed activities (I), do more such activities, develop them and deepen them and introduce new activities, irrespective of age.

Living with RTT means that principally other people decide the pattern of activities (II). How the individual with RTT communicates and shows response can perhaps impact the choice of activity pattern. To observe responses, for example during activities of engagement (II), is highly important. In the diary study (II) the teenagers/young adults showed engagement when socializing, playing and communicating, and also when they were supported to move, eat food and snacks and when they were listening to and watching films, books and music. At an

individual level, engagement, interests and what may be enjoyed (I) must be explored to be possible to put into effect and used. The results of this thesis could inspire and guide to further development of activities in everyday life for girls and women with RTT.

Methodological consideration

These studies contribute new knowledge about activities for females diagnosed with RTT, nevertheless the results and methodological limitations need to be discussed and critically reflected upon. In this thesis the following concepts are used: reliability, described as “the extent to which there is consistency in the findings”; validity, both internal validity “the extent to which research findings are credible”, and external validity “the extent to which the findings of a qualitative study can be generalized or transferred to other situations” (Merriam & Tisdell, 2015, p. 265). These concepts are commonly used in quantitative studies but have also been used in qualitative studies (Polit & Beck, 2012).

Sample size and response rate

The representativeness of the studies is high with a selection from a specialized centre with established diagnoses. The national Swedish Rett Center survey from 1996 was sent to all known individuals (178, n) with RTT at that time. The response rate to the question of liked activities was 123 (69%), and 80 (45%) to the question of disliked activities. Q2 was used before a cross-professional investigation (2008 - 2011), informants of 52 RTT participants responded, yielding a response rate of 95% to the question regarding what the participants liked to do. This means that the external validity can be expected to be high. Q3 was used during cross-professional investigations (2009 - 2011). Parents and/or staff of 39 RTT participants were asked what activities they liked to do with the girls/women with RTT. Of 92 informants, 62 responded (67%).

In study II the pilot study showed that it is possible to use the Time-geographical method to collect data on activities during everyday life (II), which strengthens the method's internal validity. A random sampling was used, and families were contacted until diaries for ten participants were filled in during a self-selected ordinary week. The informants were well

acquainted with the participant's everyday life and can therefore be expected to be able to choose a representative week as instructed. However, the validity and reliability of time use diaries are directly affected by the subject's motivation, in this case the motivation of the parents and staff, and their conviction of the importance of collecting the data and sense of involvement in the study. Parents and staff involved showed a high commitment both in completing diaries as well as in the member checks (Merriam & Tisdell, 2015; Polit & Beck, 2012). Further, that all the diaries were sent in with seven completed days may strengthen the reliability of the results.

A limitation of the thesis is that it was not possible to ask the participants themselves. The informants answered the questionnaires based on their own interpretation and experiences of their girl's/woman's activities (I). Including a description of what the informants based their interpretations of the girl's/women's reactions as positive and/or negative upon, may have strengthened the validity of study I. The diary notes (II) were written continuously by a parent or staff member to describe the teenager's or young adult's activities and responses during the different activities. This is currently the best way to collect information about the participants' activities.

However, there is a risk that the informants in study I answered questions based upon how they thought they were expected to respond. For study II, the informants' traditional role as a caregiver might be a reason for the frequent notes related to personal care activities. Clearer instructions on the writing of the diaries, emphasizing school, daily work and leisure time activities may have shifted the focus of the notes to these activities, and also resulted in these activities being described more in detail.

A descriptive study using secondary questionnaire data

For the descriptive questionnaire study (I), content analysis was used to describe the answers to the open-ended questions (Nayar & Stanley, 2014). The CA used in this study was qualitative, considering that the categories were grounded in the material and not classified in predetermined codes. Counts in qualitative CA represent a summarization and localization of patterns emerging in the material (Morgan, 1993; Pope & Mays, 1999; Weber, 1990). Development of subcategories and categories was continuously discussed between the authors during the analysis process. The process of analysis continued until the findings were robust,

and there was an agreement between the authors to strengthen the reliability of the results (Merriam & Tisdell, 2015; Polit & Beck, 2012). The Statistical Package for the Social Sciences SPSS 19.0 (IBM Corp., Armonk, NY) was used to present occurrence and frequencies of the 33 developed subcategories, presented in numbers and percentage. Presenting data by quantification has been described as increasing the reliability of a result (Berg, 2004; Krippendorff, 2004). The studied data over time confirmed the results of liked activities for the girls and women and this may support the reliability of the results. Knowledge of disliked activities was only possible to study from the years 2008 – 2011 and with fewer answers than enjoyed activities. These responses should therefore be viewed with caution. Overall, the reliability of the results may be regarded as high due to the large sample size in study I.

A Time-geographic diary study

For the second study (II) the time-geographical diary method was used (Ellegård, 1999, 2006). Before starting, the informants had the possibility to discuss and describe the four response words of the diary headlines (engaged, interested, uninterested and opposed) with the aim of facilitating the diary writing of responses during activities. This procedure might have strengthened the internal validity of the reported responses during activities and in turn the study results. Using the response words in combination with the method's headlines (time, activity, location, with whom) was a first attempt at capturing the teenager's or young adult's opinion of the activity. The advantage of this is that the participant might have been given a higher degree of attention and that the use of the response words strengthens the validity of using the method for studies with individuals with RTT. The risk of interpreting the participant incorrectly should be considered in the interpretation of these results. It should also be noted that the results do not differentiate between whether an engagement response was seen because the teenager/young adult enjoyed the other people around them during the activity, and/or whether they really liked the place where the activity occurred, rather than liking the activity per se.

The transparency provided by several informants per participant writing in the same diary can be interpreted as increasing the reliability of the notes; informants were able to notice if an activity or something else was missed and could correct missing data. The member check meeting (Merriam & Tisdell, 2015; Polit & Beck, 2012) at the home location prevented

misunderstandings in interpretation of the data written in the diaries and provided an opportunity for informants to verbally develop and clarify events written in the diary, which may strengthen the reliability of the study results.

The reliability of the time-geographic method is strengthened by the hierarchical coding system and the manual about how to use it (Ellegård, 1999, 2006). This increases the chance of achieving a similar result if the data were to be recoded or if the diary notes were to be coded by another person.

A risk for bias

Pre-understanding on the part of the first author is an important aspect to address. The first author's personal knowledge of the topic, as well as many years of experience working with this group, may have influenced the research process (Patton, 2014). The use of secondary questionnaire data (I) and the discussions with all authors during the analysis process may have minimized this influence. For study II, the random sample selection of participants was chosen instead of contacting families that were well known to the first author. After the sample selection, all contact with informants was conducted by letter. During the member check meetings (Merriam & Tisdell, 2015; Polit & Beck, 2012), the first author met the participant with their informants and the data collection and coding had already been performed. Only small changes in notes were necessary after the meetings, which had been due to difficulties in reading handwriting.

CONCLUSION AND CLINICAL IMPLICATIONS

- Variation of activities may facilitate well-being and quality of life for female individuals with RTT.
- The amount, choice and use of activities need to be tailored individually for female individuals with RTT
- To enhance participation and well-being of girls and women with RTT, good support in activities is important. Communicating and interpreting the individual's responses during activities is an important aspect of support.
- To shift away from sameness of days, sedentary days and an inactive life style for young adults, changes are needed in the form of support for activities that include contact with other people, receiving impressions and motion.

The results of this thesis are described at a group level and the new knowledge concerning everyday lives for female individuals with RTT might be helpful for developing a richer everyday life for such individuals. Activities that are enjoyed may differ between individuals, but having contact, receiving impressions and being in motion were present in the liked activities. Moreover, the need for careful assistance cannot be under-estimated. The results indicate that professionals, parents and staff supporting a girl or a woman with RTT may need to explore and review what the days contain and how time is used. From an overall occupational perspective, asking questions about the activities occurring each day and why such activities are selected, may facilitate a more individual selection of activities that are planned and used to achieve several objectives. For example, if decreased participation is observed in already used activities, it may be useful to adapt the activities to include a higher degree of contact, impressions and motion. If there is a risk for an inactive life style, interventions including careful planning of content and amount of activities may be warranted. There could also be a need for different kinds of interventions, medical as well as rehabilitative.

Receiving input and new experiences is important all through life. Young adults with RTT need something to look forward to, and should have support in getting new social contacts, doing motivating activities for them, visiting new places and so on. This is not too much to ask of life, is it?

SVENSK SAMMANFATTNING

Diagnosen Rett syndrom (RTT) är sällsynt men förekommer över hela världen, prevalensen är 1:9 000 födda flickor och ett fåtal pojkar/män får diagnosen. Syndromet beskrivs som en utvecklingsneurologisk störning. I Sverige föds ca sex flickor per år som vid 6–18 månaders ålder ses utveckla symtom på syndromet. RTT är en klinisk diagnos, för många kan diagnosen idag även bekräftas genetiskt. Vanligtvis ses en förändring, en mutation i ett genetiskt arvsanlag på X-kromosomens långa arm. Syndromet kan utvecklas hastigt eller gradvis. Tecken på RTT är delvis eller fullständig förlust av; förvärvade funktionella handförmågor, talspråk eller joller samt avvikande gångmönster eller avsaknad av gångförmåga. Vanligt är också vridande handrörelser och svårigheter att göra viljemässiga rörelser. Syndromet varierar i svårhetsgrad mellan individer. Vanliga följder av syndromet är exempelvis andningsstörning, anfall, oliksidig muskelspänning och skolios. RTT är inte en fortskridande sjukdom. I Sverige finns kvinnor med syndromet som är över 60 år. Oavsett ålder har personer med RTT stort behov av stöd och insatser.

Arbeterapi och Aktivitetsvetenskap visar på betydelsen av aktivitet och utgår ifrån att människor är aktiva, gör saker och deltar i aktiviteter livet igenom. Människor gör saker på egen hand eller tillsammans med andra. Att leva sitt liv med diagnosen Rett syndrom och dess följder innebär att behovet av assistans och individuellt anpassade aktiviteter är mycket omfattande. För att delta i aktiviteter behövs assistans och hjälpmedel. I Sverige har LSS lagen (1993:387) om stöd och service till vissa funktionshindrade stor betydelse för personer med RTT. Verksamhet enligt LSS ska främja jämlikhet i levnadsvillkor och full delaktighet i samhällslivet för personer som ingår i lagens personkrets, vilket personer med RTT gör.

Avhandlingens övergripande mål var att beskriva vardagen för kvinnor med Rett syndrom. Forskningsprojektet godkändes av etiksprövningsnämnden (EPN) i Umeå, 2012 och två deskriptiva studier varav en med data från enkäter och en dagboksstudie genomfördes.

I den första studien var syftet att över tid undersöka vilka aktiviteter flickor och kvinnor med Rett syndrom tyckte om eller inte tyckte om att göra. Forskningsdata hämtades från tre enkäter (Q1, Q2, Q3) som samlats in av Rett Center på Frösön, nuvarande: Nationellt Center för Rett syndrom och närliggande diagnoser. Den första enkäten Svensk Rett-Kartläggning (Q1) skickades ut till alla personer med RTT som då var kända i Sverige 1996. Den andra

enkäten (Q2) hämtades från journalanteckningar med uppgifter från åren 2008–2011, insamlade data inför tvärprofessionella utredningsveckor vid centret. Den tredje enkäten (Q3) användes under tvärprofessionella utredningsdagar på centret åren 2009–2011. Samtliga enkäter besvarades av informanter; föräldrar och personal till personer med RTT. Frågor om gillade och ogillade aktiviteter studerades som manifest data. Aktiviteters likheter och skillnader studerades, diskuterades, sorterades och tre teman utvecklades från datamaterialet. Förekomst och omfattning av aktiviteter i form av deskriptiva data presenterades.

I den andra studien var syftet att kartlägga och beskriva aktiviteter och hur tonåringar och unga kvinnor med Rett tillbringade sin tid under en vanlig vecka beskrivet av föräldrar/personal. Tidsanvändning för de olika aktiviteterna undersöktes samt var och med vem aktiviteterna utfördes. Dessutom beskrevs responsen som deltagarna gav uttryck för i samband med aktiviteter. Ett randomiserat urval från patientjournaler vid centret utfördes och familjer kontaktades vartefter per brev. Samtyckesformulär användes där vårdnadshavare gav sitt samtycke att deras dotter skulle delta. 4 tonåringar (15-20 år) samt 6 unga vuxna (25-30 år) deltog.

En tidsgeografisk dagboksmetod modifierades för att beskriva aktiviteter, lokalisationer och med vem aktiviteten utfördes under sju dagar för tonåring/ung vuxen med RTT. Föräldrar valde vecka för dagboksnotat och informerade sin tonåring/unga vuxen med RTT och alla andra som var involverade. Anteckningsböcker användes som dagböcker och med följande rubriker inskrivna: *Datum/tid, Plats, Aktivitet, Vem/vilka, Respons* (Engagemang, Intresse, Ointresse, Motstånd) samt *Egen kommentar*. Kortfattade dagboksanteckningar skrevs löpande av informanter; föräldrar eller personal under veckan. Dagböcker kopierades och avidentifierades. Software VISUAL-TimePACts och dataprogrammet DAILY LIFE 2011 användes för kodning av anteckningar och dataanalys.

Resultatet av den första studien med data från enkäter visade att flickor och kvinnor i åldrarna 2–66 år med diagnos RTT gillade aktiviteter med innehåll av sensoriska intryck, kontakt och rörelse. Aktiviteter som gillades mest var simma/bada, lyssna på musik, vara utomhus/gå, rida, vara tillsammans med familj eller vänner. Aktiviteter eller sammanhang som flickor/kvinnor med RTT inte gillade var hårvård, tandborstning, duscha, toalettbesök och assistans med klädsel. För några få beskrevs även gymnastik/stretchning som ogillade aktiviteter. Föräldrarna och personal gillade i hög grad att göra samma aktiviteter tillsammans med flickan/kvinnan med RTT som hon själv gillade att göra.

Resultatet av den andra studien med data från dagböcker presenterade resultat främst på gruppnivå och medelvärde för de tio deltagarnas aktivitetssfärer över ett dygn. Mest tid använde tonåringar och unga vuxna med RTT till att sova, till personlig vård, medicinsk hälsovård, vila på dagtid samt resor till skola eller daglig verksamhet. Det fanns begränsat med tid kvar till andra aktiviteter. 4 timmar och 17 minuter per dag till övriga aktivitetssfärer: Receptiva/återhämtande aktiviteter, Fysisk träning, Sociala/kreativa aktiviteter, Kommunikation och Skola/Daglig verksamhet. Mest tid tillbringade tonåringar/unga vuxna hemma och tillsammans med personal. Flest notat om respons under aktivitet var respons av intresse och minst antal notat var om motstånd. Vid aktiviteter som umgänge, lek och samtal med familj, vänner och personal noterades respons av engagemang. Även när de stod upp, gick eller assisterades till rörelse vid bad eller gymnastik och under måltider, vid film- och TV-tittande, bokläsning och musik noterades engagemang hos deltagarna. Respons av motstånd noterades vid toalettbesök, användande av andningsmask, stretchning, tandborstning, väckning, klädsel och vid påtagning av ortoser.

Sammantaget understryker avhandlingen vikten av att ge möjligheter och tid för uppskattade aktiviteter i en högre grad. Variation av aktivitet kan öka välbefinnandet och därigenom livskvaliteten för flickor och kvinnor med RTT. Val och omfattningen av aktiviteter behöver anpassas individuellt. Att ge stöd till ökad delaktighet i aktivitet och att därmed främja välbefinnandet är viktigt. Likaså att kommunicera och tolka individens respons under aktiviteter är ett betydelsefullt sätt att ge stöd. Åtgärder kan behöva vidtas om personer med RTT har enformiga dagar, mycket stillasittande tid och en inaktiv livsstil. Exempelvis att erbjuda aktiviteter och anpassa aktiviteter, skapa kontakt och att använda sig mer av rörelse och av aktiviteter som medför sinnesupplevelser.

ACKNOWLEDGEMENTS

First, I would like to thank all the families and staff who responded to the questionnaires concerning activities (I). Thereafter, I also want to thank the adult with RTT who participated in the pilot study, and the family and staff, and the second study ten participants (teenager/young adults with RTT) and their families and staff who wrote all the diary notes (II). Thank you, all of you for sharing your precious experiences.

Warm and special thanks, to my three amazing supervisors. Gunilla Liedberg, my main supervisor, for your heart full of patience, for your constructive criticism, engagement and guidance. Without your support this thesis would not have been completed. Another very warm thank you goes to Helena Hemmingsson for your perspicacity, for supporting us when we got stuck and needed a second opinion or needed to discuss overall issues, and to Ingegerd Witt Engerström with your wide-reaching expert knowledge of the diagnosis RTT and your interest for the topics of this thesis. Thank you also to my co-author in the second study, Helene Lidström for interesting discussions and new perspectives on the studied data, for joy and friendship.

Thanks to all the OT and Nursing PhD students and researchers at the ISV, campus Norrköping, Linköping University. When I first started there were only two OT PhD students, Vedrana Bolic Baric and Maria Borgestig, and with those two people I had a great time, in particular at the world congress for Occupational Therapists in Yokohama, meeting clinicians and Occupational Therapy researchers from all over the world. As my work place is in Östersund I have visited Campus Norrköping now and then but always felt welcome at ISV by my supervisors, teachers and the growing OT PhD student group. Thank you everyone for solid support and a lot of learning.

A special thanks to my colleagues at the National Center for Rett syndrome & related disorders, Frösön, Sweden for joy and hearty laughter, for sharing research competence and encouragement. A big hug to Helena Wandin for the exchange of the research topic discussions as well as linguistic support. Thanks to all of you, new and earlier colleagues; Thomas Aronsson, Märit Bergström Isacson, Monica Dolik-Michno, Annelie Ericsson, Elisabet Gabrielsson, Eva Hamner, Martina Holmbom, Gunilla Larsson, Gun-Marie Nyström, Inga Qvarfordt, Magnus Starbrink, Elisabeth Sundeman and Lena Svedberg.

Thank you for such great support dear siblings, friends and our big family, Sernheim ++, Rebecka, Mikael, Ida, Majken, Jacob, Maria, Folke, Gustav, Adam, Emelie, Olle and Assar. Last, and most of all, I thank my beloved husband, Johan for your patience and support.

Åsa-Sara Sernheim

This thesis was supported by grants from the Linnéa and Josef Carlsson's Foundation, Helsingborg, Sweden and the Folke Bernadotte Foundation, Stockholm, Sweden. Thank you very much for supporting descriptions of everyday life for girls and women with RTT.

APPENDIX

tid

skriva

göra

berätta

morgon

kväll

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