Cathryn Ghena

PHYT 885 Module 5 Toolbox

**Rett Syndrome Assessment Toolbox**

Rett Syndrome (RTT) is a progressive neurological disease that affects females, with an onset of symptoms from 6-18mos. progressing throughout life. There are four stages through which indivdiuals typically progress—early onset, rapid regression, plateau, and late motor deterioration. Currently no outcome measures have been validated specifically for use in this population. However, outcome measures should still be used in a plan of care as they can provide valuable information for tracking disease progression and setting appropriate goals.

The following is a non-exhaustive list of measures that can be used to aid a therapist in his/her assessment of a child with RTT. Measures were chosen based on: 1) Utilization in research studies on patients with RTT and/or 2) Measures typically used for signs/symptoms commonly present in this population.

**RTT Specific Measures for Assessing Level of Impairment**

*Clinical Severity Scale (CSS):* This quantitative measure was developed specifically for use in the RTT population, although has yet to have psychometric properties validated. It is based on the diagnostic and developmental features of RTT, specifically onset and rate of regression, communication skills, hand behavior, and head size. It has been paired with quality of life measures such as the CHQ-PF50 (see quality of life section of toolbox) to track trends related level of motor impairment, family stress, and child quality of life.1

*Motor Behavior Analysis (MBA):* The MBA is considered a “more dynamic measure” as it focuses on the motor, behavioral, and respiratory dysfunction as compared to the more regression-focused CSS. The respiratory component is especially important in this population as girls with RTT tend to have abnormal breathing patterns most notably hyperventilation and breath holding. Scoring for the MBA is out of 148, with a higher score indicating more severe involvement.

*Rett Syndrome Severity Scale (RSSS):*RSSS asses seizures, respiratory patterns, scoliosis, ability to walk, hand use, speech, and sleep. It uses a Likert scale with higher scores indicating more severe involvement. Increases in scores are generally seen with increased age.

*Rett Syndrome Video Checklist*: Video analysis has been used to evaluate symptoms as well as track progression of RTT. Specific presentation to be recorded include eating, drinking, communication, hand function/movement, personal care, and mobility. This method of tracking disease progression allows for objective assessment of both function and behavior in turn helping to develop an appropriate plan of care.

**Measures to Assess Common RTT Impairments**

*Scoliosis*: It is suggested that >85% of people with RTT will acquire some degree of scoliosis; furthermore, presence of scoliosis is positively correlated with total MBA score as well as delay or loss of ambulation. Therapists can play a valuable role in tracking scoliosis over the course of life traditional scoliosis evaluation, having the patient forward flex at the hips while therapist is palpating for vertebral alignment and rib hump. Formal assessment of scoliosis can be done through use of radiographs and analyzing Cobb Angle.

*Modified Ashworth Scale:* The Modified Ashworth can be used to classify and track progression of tone in patients with RTT. The triceps surae are most commonly affected by tone, resulting in abnormal gait deviations such as toe walking and a decreased base of support. Formal gait analysis should be performed in conjunction with assessment of tone.

*Peabody Developmental Motor Scale:* While all components of the PDMS can provide valuable information, the Fine Motor Scale section can be especially informative as it assesses grasping, hand use, hand-eye coordination, and manual dexterity.Hand function should be monitored over the course of life as these skills tend to be impaired, with regression of skills typical occurring. Intervention can help slow this regression allowing girls to be as functional as possible.

*Earhardt Developmental Prehension Assessment*: This test can also be used to more thoroughly assess hand function. It looks at primary involuntary hand/arm patterns (i.e. clenching, wringing), primary voluntary movements (i.e. posture, reach, grasp, manipulation), and pre-writing skills. Score can then be compared to provide a functional age based on upper extremity manipulation ability. While it is not as common as the PDMS, it can provide more specific detail on fine motor abilities.

*Timed up and go (TUG)*: The TUG requires the patient to stand up from a chair and ambulate, providing the therapist with information on balance ability. While it has not been validated for use in children with RTT, it has been validated for use in children with cerebral palsy as well as healthy children 3-9y.o. This information could serve as a good baseline for balance expectations and goal setting.

**Other Impairment Based Measures**

1. Active and Passive Range of Motion
2. Manual Muscle Testing
3. Gait Analysis

**Activity/Participation Based Measures**

*Screen for Social Interaction (SSI)*: This scale is traditionally used in patients with autism or other developmental disordres to assess motor and communication impairment; however, the similarities between RTT and Autism suggests that this measure can be successfully used in this population. Furthermore, SIS score does not appear to be effected by age. SIS assesses caregiver connection (i.e. smiling, eye contact, facial expression, play), interaction/imagination (i.e. play, peer interaction), social approach/interest (avoidance, shyness), and agreeable nature (aggressiveness, sharing). Refer to Kaufmann et al. article for a copy of measure.

*Rett Syndrome Behavior Questionnaire (RSBQ):* Developed in 2002 specifically for RTT, this questionnaire assesses 8 factors found in most girls with RTT: body rocking/facial expression, hand behavior, repetitive face movement, night behavior, fear/anxiety, walking/standing, general mood, and breathing. The anxiety aspect of this scale is especially important as patients with RTT commonly present with increased anxiety, which in turn interferes with their ability and desire to participate in activities outside of the home. Such anxiety can also interfere with progress in therapy, emphasizing the importance of developing a trustworthy relationship with these girls and choosing calming, non-provocative interventions. RSBQ score does not appear to be related to age. Refer to Kaufmann et al. article for a copy of measure.

*Vineland Adaptive Behavior Scale:* This is a parent interview measure that looks at adaptive behavior tendencies in children specifically assessing communication, daily living, socialization, and motor skills. While this has been used in research on girls with RTT, the daily living and motor skills sections were not used secondary to the level of involvement of participants.

*Pediatric Functional Independence Measure (WeeFIM):* This functional independence measures tracts a child’s functional ability and its impact on activity and participation. Specific components include self-care, mobility, cognition, and communication. It has one of the few measures that has been successful and repeatedly used in research studies on children with RTT, suggesting that therapists should be utilizing the WeeFIM during initial and follow up evaluations.

*Pediatric Evaluation of Disability Inventory (PEDI):* While not as commonly used in research on children with RTT, the PEDI could is another option for assessing activity and participation. It would provide therapists with more information on equipment, modifications, and caregiver assistance.

*Community Balance and Mobility Scale*: This scale may be appropriate for higher functioning females with RTT, providing quantifiable information on the individual’s ability to ambulate and function with a community environment. Community participation is important in this population as it is correlated with higher patient outcomes.

**Quality of Life Measures**

*Non-Communicating Children’s Pain Checklist (NCCPC-R)*: Pain in this population is often overlooked despite its correlation with age and number of health problems. This measure has been validated for use in children 3-18 years of age who are unable to verbally communicate. Test administrator, commonly the caregiver, is to monitor the child’s activity over a 2 hour period specifically taking note of vocalizations, social interaction, facial features, level of activity, body language, physiological behavior (i.e. breathing, sweating, shivering, etc.), and eating/sleeping habits. A score >7 indicates that the child is in pain. [The Non-Communicating Children’s Pain Checklist Form](http://www.google.com/url?sa=t&rct=j&q=&esrc=s&source=web&cd=1&ved=0CC0QFjAA&url=http%3A%2F%2Fwww.aboutkidshealth.ca%2FEn%2FDocuments%2FAKH_Breau_everyday.pdf&ei=0wk7U_OaOsmI7AaptICwCA&usg=AFQjCNGriqh_HvnC3w52uVzE3iNOX_uc8w&bvm=bv.63934634,d.ZGU&cad=rja)

*Child Health Questionnaire 50 (CHQ-PF50):* This is a quality of life measure validated for use in children with a chronic illness, ages 5 to 18 years. With 14 different domains, it provides a holistic assessment of the child’s quality life considering vital physical and psychosocial components of life. There are two versions of it, allowing for either child report or parent report. The parent report version (PF50) is considered more appropriate for girls with RTT secondary to communicative and cognitive impairment. It is important to note that while parent report can be extremely informative, it is important to still provide holistic care based on the needs of the child. Furthermore, this measure should be completed by the same caregiver over the course of care for increased accuracy. Interestingly, higher scores on the psychosocial component of the CHQ have been associated with more impaired physical involvement on the CSS. Researchers suggest that this relationship could be secondary to increased behavioral issues (i.e. anxiety, self-abuse, aggression) in girls with higher motor function.1

* + <https://www.healthactchq.com/chq.php>

**Disease Progression**

The following graph is meant to serve as a quick guidelines as to what impairments are commonly see during the 4 different stages of RTT as well as common age of disease onset and progression.

