HSD101

The Key Role of Therapeutic Professionals in Rett Syndrome Care Pathway

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INTRODUCTION

- Rett syndrome (RTT) is a genetic neurological and developmental disorder that predominantly affects females. It is a rare condition, with an estimated incidence of 1 in 10,000 female births.[1]
- RTT is characterized by normal early development followed by regression beginning around 12–18 months of age.[2] Patients with RTT develop debilitating symptoms that render them severely disabled, with almost 40% of adult women with RTT being unable to walk.[1,3]
- There is currently no cure for RTT, and treatment focuses on symptom management.[4]

OBJECTIVES

- This study aimed to describe the care pathway for patients with RTT in the United States (US), particularly the role of therapeutic professionals (TPs).

METHODS

- A series of semi-structured interviews with TPs (physical therapists [PT], occupational therapists [OT], and speech and language therapists [ST]) and clinicians who treat patients with RTT (and may refer them to TPs) were conducted.
- To maintain impartiality of the collected evidence, all interviews were conducted in a double-blinded fashion.
- The target sample aimed to include 20–25 TPs and 8–10 referring clinicians, and provide a good representation of different care settings (RTT Centers of Excellence [COEs], non-COE outpatient and home health, and school-based care).
- Participants (both TPs and clinicians) were required to practice in the US and have >3 and <25 years experience in their role.

RESULTS

Participant characteristics

- Twenty-six healthcare professionals were interviewed, including 17 TPs (6 PT, 6 OT, 5 ST), and 9 clinicians (3 pediatricians and 6 pediatric neurologists).
- The TPs worked in community-based (n=10), school-based (n=5), and other (n=2) settings.

Evolution of RTT care pathway over the patient's lifetime

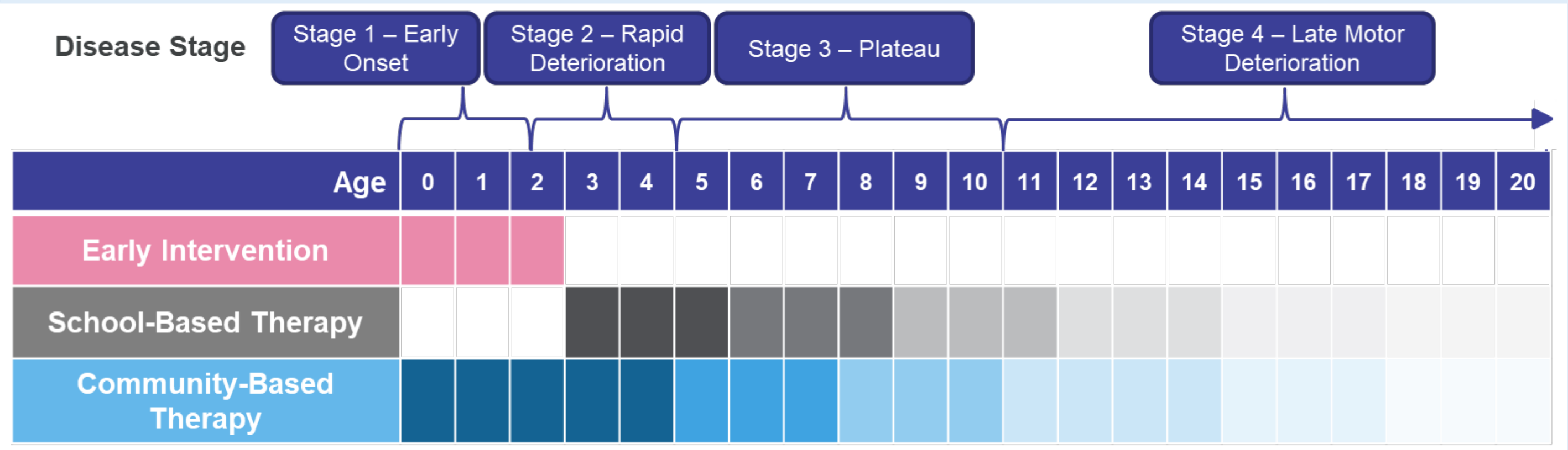
- The care pathway for patients with RTT was found to be largely driven by disease stage, age and symptoms experienced by the patient, with evolving needs for ongoing therapy services in later disease stages (**Figure 1**).
- Consistent with the changing needs of patients with RTT, the care pathway could be divided into three phases (**Figure 2**):

1. Early intervention services that aim to improve functional skills through parental training and terminate at the age of 3.
2. School-based therapy, focused on enabling access to education and integration in school, begins at the age of 3 and may continue over the child's education.
3. Community-based therapy to support functioning at home and in the community may continue over the patient's lifetime, but usually tends to decline by age 5–10.

Figure 1. The Changing Needs of Patients with RTT

RTT Stage	Common Symptoms and Therapy Focus
Stage 1 – Early Onset *Ages: ~6-18 months*	• Developmental delays in sitting and crawling • Loss of interest in toys • Less eye contact • Early Interventions services focus on parental training and support
Stage 2 – Rapid Deterioration *Ages: ~1-4 years*	• Loss of previous skills (such as sitting, standing, walking, and self-feeding) • Rapid onset of RTT symptoms (such as slowed head growth, abnormal hand movements, communication challenges) • Therapy services focus on parental training and minimizing functional deterioration
Stage 3 – Plateau *Ages: ~5-10 years*	• Mobility problems persist • Hand use and communication improve • Seizures often present • Therapy services focus on optimizing function and prevention of regression in latter stage
Stage 4 – Late Motor Deterioration *Ages: ~10+ years*	• Progressive decline in muscle strength and control resulting in decreased mobility • Communication and hand skills remain stable • Seizures may lessen • Need for therapy continues to address long-term optimal functional maintenance but may be subject to more barriers from payers

Figure 2. Therapy Continuum for Patients with RTT



Key stakeholders involved in the RTT care pathway

- Optimal treatment for patients with RTT was found to utilize a multidisciplinary care team approach in which TPs are integral:
  - **Community-based TPs** provide frontline support for patients, focused on functional mobility and contracture prevention (PT), participation in activities of daily living (OT), and addressing communication problems and dysphagia (ST).
  - **School-based TPs** act as educational enablers, addressing functional mobility (PT), fine motor skills (OT), and supporting communication and socialization at school (ST).
  - **Pediatricians and pediatric neurologists** act as care team leaders and decision makers.
  - **Other medical specialists** who support the patient and family. Gastroenterology, orthopedics, and endocrinology support is commonly needed to help manage symptoms such as scoliosis, dysphagia, and osteopenia. Other specialties may also be engaged depending on patient's symptoms and needs.
- Notable differences in the organization of care and stakeholder engagement were found between COE and non-COE settings:
  - COEs foster an integrated care model that facilitates access to care and coordination of services for patients with RTT, potentially also reducing caregiver burden.
  - Outside of COEs, care is more compartmentalized, which could require greater effort from caregivers to facilitate coordination.
- Identified areas for improvement of care organization outside of the COE setting included geographic accessibility, disease-specific experience, integration of care plan, communication across the care team, and support for caregivers.

Limitations of the RTT care pathway and efforts for improvement

- Inherent limitations in coordination of care, communication, and standardization were identified in the RTT care continuum, creating potential barriers to access for patients (**Table 1**). In particular, reimbursement and access issues were identified as important barriers to optimal provision of early intervention and community care, while school-based care was less affected, being funded by the school district.

Table 1. Characteristics and limitations of the RTT care continuum

	Early Intervention	School-Based Therapy	Community-Based Therapy
Goals of Treatment	• Addresses early functional skills, such as crawling, walking, talking, eating	• Access to education and integration into neurotypical classrooms	• Functional activities to support home and limited community function
Eligibility and Referral	• Physician diagnosis of developmental delay or disability, and referral to therapy	• Standard criteria of disability limiting access to education	• Physician referral for services based on symptoms and patient/family needs
Payment / Reimbursement	• Health insurance OR • Sliding scale available for those who private pay	• Services paid for by school district • No OOP costs to family	• Health insurance or private pay • Some OOP costs to family • Insurance may have visit limits • Request for additional therapy approval may be screened by AI
Characteristics	• One-to-one sessions with much of therapist time focused on parental training	• Integrated into school day • Limited burden on parents for delivery of services • Focused only on academic needs	• Intensive, one-to-one therapy • Driven by patient/family goals for home function
Limitations	• High demand for services – there may be a waiting list and/or limited availability of services	• Often takes place in a group setting • Frequency and intensity may be lacking	• Logistic burden on parents to get patient to clinic • High demand for services with a possible waiting list

Abbreviations: AI, artificial intelligence; OOP, out-of-pocket.

- Barriers to optimal provision of care for patients with RTT were identified at every level. Notable challenges included limited experience with the RTT population and fragmentation of care potentially delaying referral to TPs, an issue which COEs may be empowered to mitigate (**Figure 3**).

Figure 3. Barriers and Facilitators to Optimal Care Provision

	Barriers	Facilitators
Macro level ***(Diagnosis and standardization)***	• Rare disease with only symptomatic treatment • Lack of standard assessments and treatment	• Identification of disease etiology • Research and engagement
Meso-Level ***(Awareness and Access to Specialized Care)***	• Geographic location of COEs creates some areas with limited access • Limited disease awareness and experience among non-COE clinicians • Fragmentation and limited coordination/communication across the therapy continuum • Medical policy may not support habilitative services	• Growing presence of COE model, increasing centralization of treatment • Emergence of targeted drug therapies • Insurers typically supportive of funding rehabilitative treatment for patients with RTT
Micro-Level ***(Coordination and Communication)***	• Heavy burden of coordinating care and services • Burnout from caregiving • Potential social factors influencing access and continuation of care	• Highly engaged caregiver community • Support sources for caregivers available through patient organizations

Abbreviations: COE, center of excellence.

- Several factors that facilitate the efforts to improve the care pathway were also evident, including a very engaged caregiver community and growing centralization of care with the adoption of the COE model (**Figure 3**).

CONCLUSIONS

- Outside of the limited number of COEs, patients with RTT face barriers to optimal care provision by TPs, arising largely from lack of integrated, standardized care by RTT specialists familiar with this rare condition.
- Reimbursement and access issues may prohibit patients with RTT from accessing early intervention and community-based therapeutic services.
- Overall, TPs, who play a key role in day-to-day care for patients with RTT treated within COEs, are currently underutilized outside of the COE setting. This is likely driven by lack of standardized assessment of patient's therapeutic progress and response to treatment, insufficient integration of care, and limited access to care provided by TPs.
- Ongoing efforts to optimize the care pathway for patients with RTT and a growing adoption of the COE model with a strong engagement of TPs may lead to improved clinical outcomes and quality of life for patients with RTT and lessening the burden on their caregivers.

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DISCLOSURES

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