

# The Role of Therapeutic Professionals and Tools for Assessing Therapeutic Progress in Rett Syndrome

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**PURPOSE:** Rett syndrome (RTT) is a rare genetic neurological and developmental disorder that predominantly affects females. This study aimed to describe the care pathway for RTT in the United States, particularly the role of therapeutic professionals (TPs), and to identify the tools that TPs use for assessment of progress in patients with RTT.

**DESIGN AND METHOD:** A series of semi-structured interviews were conducted with TPs (physical therapists [PT], occupational therapists [OT], and speech and language therapists [ST]) and clinicians who treat RTT patients. The target sample aimed to include 20–25 TPs and 8–10 clinicians and provide a good representation of different care settings.

**RESULTS:** 26 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. The care pathway for RTT patients was driven largely by disease stage and age, and comprised three phases 1) early intervention for children ≤3 years of age aiming to improve functional skills through parental training, 2) school-based therapy for patients aged >3 years, focused on enabling access to education and integration in school, and 3) community-based therapy throughout the patient's lifetime to support functioning at home and in the community. TPs reported the use of 22 therapeutic progress assessment tools, including 6 RTT-specific tools (3 global function assessments and 3 tools focused on mobility and motor function). Tools unspecific to RTT were used to measure global function (n=4), activities of daily living (n=6), and cognition, communication, and psychosocial functioning (n=6).

**CONCLUSION:** TPs play a key role in day-to-day care for patients with RTT. Policy and practice efforts to enhance integration and standardization of care could facilitate care provision by TPs in this rare disease.

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