

Understanding Parent and Expert Concerns and Experiences in Rett Syndrome: Qualitative Research to Inform Outcome Measure Development

Wisnu A. Pradana¹, Daniel C. Tarquinio², Wing Yi Chan², & Usree Bhattacharya¹
¹University of Georgia; ²Center for Rare Neurological Diseases

Rett syndrome (RTT) is a neurodevelopmental disorder with no disease-modifying treatment and limited outcome measures. Using focus group interviews with caregivers and semi-structured interviews with RTT experts, we aimed to develop a novel scale of symptoms and skills for RTT based on their concerns and aspirations.

We found that many caregivers are dissatisfied with items on the existing standardized instruments used in RTT as they poorly capture the complexity and heterogeneous presentation of individuals across the lifespan. In addition to confirming ten current domains relevant to RTT diagnostic criteria and current literature, namely breathing, mobility, hand use, feeding/gastrointestinal, sleep, aggression/anxiety, non-verbal communication, verbal communication, scoliosis, and seizures, we identified two new categories of symptoms and skills that should be included in outcome assessment: pubertal concerns and independence/vulnerability concerns. Using these findings, we developed an item bank of more than 300 items. We administered these to two cohorts of caregivers. Results of the item bank surveys are being analyzed.