Appointment discussion guide:
Rett syndrome

Rett syndrome (RTT) is a complex, progressive, multisystem condition with a wide range of symptoms. Understanding the impact of RTT from the caregiver's perspective may have important implications for clinical practice.[1,2] This tool can help weave caregiver insights into your comprehensive management plan by guiding collaborative conversations during appointments.

Topic	Suggested discussion starters	Background information
 Physical symptoms	Review the physical symptoms of RTT with patients and their caregivers. Are there any areas that could use more attention and focused care?	RTT may lead to loss of expressive language and purposeful hand use, impaired ambulation, stereotypical hand movements, microcephaly, seizures, and severe motor deficits.[1,3] RTT is a heterogeneous condition; the timing and severity of symptoms varies widely between patients.[4,5]
 Behavioral symptoms	Review behavioral symptoms of RTT with patients and their caregivers. Are there any areas that could use more attention and focused care?	Behavioral manifestations include anxiety, mood disturbances, screaming episodes or spells, and inconsolable crying.[6]
 Developmental milestones	Has she/he experienced any new life milestones? - Examples include attending school, making a friend, etc. Has she/he experienced any milestone regression? - Examples include loss of purposeful hand movement, regression in motor skills, etc.	Developmental regressions, such as reduced hand use and language, typically stops between 2 and 3 years of age.[1] However, significant life events may require changes to the existing care plan.[7]
 Communication	How does she/he communicate? Have there been any regressions or improvements in her/his ability to communicate? Are there any methods of communicating you're curious about or want to discuss?	Communication challenges vary between patients.[2] Communication methods might include eye pointing, vocalizations, or using an eye gaze device.[1]

Topic	Suggested discussion starters	Background information
Goal setting and tracking	Discuss realistic and attainable goals to set. - For example, a reduction in teeth grinding[1] Are you currently keeping track of her/his progress or regression? - Suggest keeping a daily log of symptoms to review at the next visit	To guide the discussion, you may suggest caregivers keep a log of certain symptoms or challenges they encounter each day to review with you at the next visit.
Care team	What other specialists does she/he see? Are there specific areas of care where she/he needs more support? Ask about any documents or test results that could be shared between specialists.	A multidisciplinary team of physicians, specialists, and specialized therapists is needed to help manage the wide-ranging symptoms.[1]
Social interactions	Discuss the types of accommodations that are being made for her/him in social settings, like school. What have been some recent successes/challenges?	Care plans that provide social interaction and physical activity at all ages may help reduce age-related deterioration.[1]
Impact on family	What aspects of caring for a loved one with RTT have been especially challenging? Discuss overall family involvement in providing care at home.	For example, if a patient with RTT has sleeping difficulties, it can impact life for the entire family and, in particular, lead to stress and anxiety in caregivers.[2]

This guide is provided by Acadia for educational purposes only. It is an example and may be used as part of a full assessment. Always use your clinical judgment when developing a care plan for patients with Rett syndrome.

References: 1. Fu C, Armstrong D, Marsh E, et al. Consensus guidelines on managing Rett syndrome across the lifespan. *BMJ Paediatr Open*. 2020;4(1):e000717. **2.** Palacios-Ceña D, Famoso-Pérez P, Salom-Moreno J, et al. "Living an obstacle course": a qualitative study examining the experiences of caregivers of children with Rett syndrome. *Int J Environ Res Public Health*. 2019;16(41):1-13. **3.** Neul JL, Kaufmann WE, Glaze DG, et al for the RettSearch Consortium. Rett syndrome: revised diagnostic criteria and nomenclature. *Ann Neurol.* 2010;68(6):944-950. **4.** Percy A. Rett syndrome: coming to terms with treatment. *Adv Neurosci.* 2014;2014:345270. **5.** Tarquinio DC, Motil KJ, Hou W, et al. Growth failure and outcome in Rett syndrome: specific growth references. *Neurology*. 2012;79(16):1653-1661. **6.** Gold WA, Krishnarajy R, Ellaway C, et al. Rett syndrome: a genetic update and clinical review focusing on comorbidities. *ACS Chem Neurosci*. 2018;9(2):167-176. **7.** Lotan M. Rett syndrome. Guidelines for individual intervention. *Sci World J*. 2006;6:1504-1516.

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