Case Study: Rett Syndrome

Part 1: Getting to Know Emma

Emma and Her Family

Emma is a 20-month-old girl who lives in a suburban neighborhood in central Texas with her mother, Rachel (29), father, Ben (31), and newborn brother, Noah (2 months). The family identifies as White and middle-class. Rachel works remotely as a graphic designer, and Ben is a high school science teacher. They live in a cozy home near Rachel's parents, who provide regular support and help with childcare, especially since Noah's birth.

During her first year, Emma was a calm and curious baby. She met early milestones such as babbling, sitting independently, and reaching for toys. She enjoyed music, responded to her name, and smiled frequently during interactions. However, around 14 months, Rachel and Ben began noticing subtle changes. Emma stopped using the few words she had learned, became less engaged with toys and people, and developed repetitive hand movements like hand-squeezing and finger-tapping. She also seemed less responsive to her name and began having difficulty with balance and coordination.

Medical and Developmental History

Emma was born full-term following an uncomplicated pregnancy and delivery. Her early infancy was typical, with no immediate concerns. She breastfed successfully and transitioned to solid foods at around six months, though she was always a slow eater and occasionally gagged on certain textures. Her pediatrician noted mild hypotonia at her 9-month check-up but attributed it to individual variation.

By 12 months, Emma's regression had become more pronounced, and her pediatrician referred her to a developmental-behavioral pediatrician for an evaluation. In addition to not meeting age-appropriate milestones, the pediatrician also noticed that Emma's head growth had slowed. By the time Emma was seen by the developmental-behavioral pediatrician and her team at 14 months, she had lost several skills, including saying words, using her hands to grasp objects, and had stopped walking. She was also becoming increasingly irritable.

After a multidisciplinary evaluation, the developmental behavioral pediatrician referred Emma and her family to a pediatric geneticist. The geneticist suspected Rett syndrome (RTT), a condition associated with mutations in the MECP2 gene, and recommended blood work to confirm the diagnosis. When the test was completed, it showed that Emma had Rett Syndrome. The diagnosis was a shock to the family, who had never heard of RTT before. They had also never heard of early intervention (EI), which both the developmental/ behavioral pediatrician and the pediatric geneticist recommended to them. They told Rachel and Ben that Emma's diagnosis would allow them to request developmental services and therapies for Emma without needing a developmental screening or evaluation for eligibility.

Discussion Prompts:

- What additional questions would help you understand Emma's family dynamics and support system?
- How can early intervention providers build trust and empower families in navigating a new diagnosis?
- What strategies can be used to support Emma's engagement in daily routines and play?

Part 2: Assessment

Emma began early intervention (EI) with a comprehensive developmental evaluation conducted by interventionists from early childhood special education and speech pathology. The EI team noted hallmark features of RTT, including regression in previously acquired skills, repetitive hand movements, limited verbal communication, and emerging motor difficulties. Emma's parents described her as increasingly irritable and anxious, with disrupted sleep and frequent episodes of hyperventilation. They also reported that mealtimes were particularly challenging. She no longer fed herself with her fingers and often resisted anyone attempting to get anything into her mouth during mealtimes.

The team observed Emma in her home and noted that she had limited purposeful hand use and difficulty initiating movement. She often looked away before attempting to engage. Her communication was primarily through eye gaze and vocalizations, though her response time was delayed. She also refused to eat anything. The team did notice Emma's strong connection with her family and her enjoyment of music and visual stimuli.

After documenting Emma's strengths and needs, the EI team recommended weekly home visits to provide intervention to Emma and support to Rachel in their natural environment. Together, with the family, the EI team developed an Individualized Family Service Plan (IFSP) focused on communication, motor development, and feeding.

Discussion Prompts:

- What challenges might arise when distinguishing RTT from other neurodevelopmental disorders?
- How can providers support families in understanding genetic testing results and implications?
- What are the benefits of observing children in both clinical and home settings?

Part 3: Working with a Feeding Specialist

Feeding quickly became a central concern for Emma's family. She had begun refusing certain textures, gagging during meals, and showing signs of fatigue while eating. Her weight gain had slowed, and Rachel reported that mealtimes were becoming stressful for the entire family.

The IFSP team referred Emma to a feeding specialist, a pediatric dietitian with expertise in neurodevelopmental disorders. The feeding evaluation revealed oral motor challenges, sensory sensitivities, and signs of gastroesophageal reflux, which, combined with Emma's repetitive hand movements and difficulty with motor planning, further complicated self-feeding.

The feeding specialist collaborated with the speech-language pathologist and occupational therapist to develop a comprehensive feeding plan.

Strategies included:

- **Oral Motor Support**: Exercises to strengthen Emma's jaw and tongue movements, using tools like chewy tubes and textured spoons.
- **Sensory Integration**: Gradual exposure to different textures and temperatures, paired with calming sensory input before meals.
- **Positioning and Equipment**: A supportive highchair with lateral supports and footrests to improve posture and reduce fatigue.
- **Family Coaching**: Training Rachel and Ben on pacing, physical and verbal prompting, and recognizing Emma's signs of discomfort or readiness.

The team also monitored Emma's nutritional intake and growth, considering supplemental options if needed. Mealtimes were reframed as opportunities for connection and communication, with Emma using eye gaze to choose between foods and express her preferences.

Over time, Emma developed a tolerance for a variety of foods and began participating more actively in family meals. Her parents reported reduced stress and greater confidence in their ability to support her feeding needs.

While the journey was not without challenges, such as adjusting to medication side effects and managing occasional nighttime seizures, the family found comfort in the school's commitment to inclusive care. Emma thrived in her classroom, forming bonds with peers and making progress in communication and motor skills, all while receiving the attentive nursing care she needed

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Discussion Prompts:

- How can early childhood teams collaborate with feeding specialists to support children with complex feeding needs?
- What role does sensory regulation play in feeding success for children with RTT?
- How can mealtime routines be adapted to promote communication, independence, and family engagement?