ECiDC Early Childhood Intervention Doctoral Consortium

Creating Leaders in Intensive Interventions for Infants, Young Children and Their Families

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.

Emma was a calm and curious baby during her first year. 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 squeezing and 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 around six months, though she was always a slow eater and occasionally gagged on certain textures. Her pediatrician noted mild hypotonia at her 12-month check-up but attributed it to individual variation.

By 16 months, Emma's regression became more pronounced. She lost previously acquired words, began hyperventilating during periods of distress, and exhibited increased irritability. Rachel and Ben also noticed that Emma's head growth had slowed, and she was no longer meeting motor milestones like cruising or attempting to walk. Concerned, they returned to their pediatrician, who referred them to a developmental specialist.

A comprehensive evaluation, including genetic testing, confirmed a diagnosis of classic Rett Syndrome, linked to a mutation in the MECP2 gene. The diagnosis was a shock to the family, who had never heard of RTT before. They were connected with early intervention services and began working with a multidisciplinary team to support Emma's development.

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: Screening and Assessment

Emma's diagnostic journey began with a comprehensive developmental evaluation. The team noted hallmark features of RTT: 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.

During clinical observation, Emma showed limited purposeful hand use and difficulty initiating movement. She often looked away before attempting to engage, a behavior consistent with apraxia. Her communication was primarily through eye gaze and vocalizations, though her response time was delayed. The team emphasized the importance of waiting and interpreting her nonverbal cues.

A neurological exam confirmed slow head growth and hypotonia. Genetic testing revealed a common MECP2 mutation associated with classic RTT. The team conducted home visits to observe Emma in her natural environment, noting her strong connection with family members and her enjoyment of music and visual stimuli.

The evaluation team included a developmental pediatrician, neurologist, speech-language pathologist (SLP), occupational therapist (OT), physical therapist (PT), and early intervention specialist. Together, they 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 everyone.

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

The feeding specialist collaborated with the SLP and OT 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 high chair with lateral supports and footrests to improve posture and reduce fatigue.
- **Family Coaching**: Rachel and Ben received training on pacing, cueing, 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 preferences.

Over time, Emma showed increased tolerance for a variety of foods and began participating more actively in family meals. Her parents reported reduced stress and greater confidence in supporting 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. Devon thrived in his classroom, forming bonds with peers and making progress in communication and motor skills, all while receiving the attentive nursing care he needed.

Discussion Prompt:

- 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?