Case Study: Intellectual Disability

Part 1: Getting to Know Eli

Eli and His Family

Eli (age 4) lives in a small, rural town with his mother, Rachel (29), father, Marcus (31), and younger brother, Noah (18 months). The Thompson family identifies as White and values outdoor activities, community involvement, and a simple lifestyle. They frequently spend time hiking, gardening, and attending local library events. English is the primary language spoken at home.

Rachel is a part-time librarian who adjusted her work schedule to accommodate Eli's therapy appointments. Marcus works full-time as a carpenter and often takes seasonal jobs to supplement the family's income. The Thompsons have limited access to specialized services. They rely on yearly visits to update Eli's medical and developmental needs at a regional developmental pediatric clinic, which is two hours away.

Eli is a gentle and curious child who enjoys water play, stacking blocks, and watching birds. He struggles with transitions and becomes distressed by loud noises or crowded environments. Rachel has noticed that Eli prefers predictable routines and toys that have repetitive actions. He communicates through gestures and a few vocalizations and uses a Picture Exchange Communication System to express basic needs. Rachel uses visual schedules to support Eli's understanding of daily events.

Family routines are structured around meals, outdoor time, and quiet evenings. Despite her efforts, Eli's expressive language remains significantly delayed, and he often becomes frustrated when he is unable to communicate effectively. Rachel has expressed feelings of guilt and isolation, especially when comparing Eli's development to his peers or explaining his needs to extended family.

Medical and Developmental History

Rachel's pregnancy with Eli was complicated by gestational diabetes and mild preeclampsia. Eli was born at 38 weeks via emergency cesarean section due to fetal distress. His Apgar scores were low, and he required oxygen support for the first 24 hours.

Eli struggled with sucking and swallowing, leading to poor weight gain and frequent hospital visits during his first year. Low muscle tone delayed his motor milestones. He sat independently at 8 months, crawled at 12 months, and began walking at 18 months with an unsteady gait. His fine motor skills affected his ability to use utensils or manipulate small objects. Eli's expressive language was also delayed, and at 15 months, he could only vocalize a few sounds.

The nurse at the local health clinic was concerned about Eli's development at his 6-month checkup. She requested that a pediatrician at the clinic screen Eli during his next weekly visit.

The pediatrician evaluated Eli at 7 months and completed a developmental screening. Initially, he was not overly concerned and felt Eli needed more time to progress. However, three months later, he grew concerned when he saw that Eli had made little progress in meeting developmental milestones. He referred Eli and his family to an early intervention program and a regional developmental pediatric clinic, both located approximately two hours away from their home.

The early intervention program visited Eli and his parents in their home two months after the referral. They conducted an authentic assessment and documented that Eli had global developmental delays. Eli began receiving home-based early intervention services at 13 months.

Eli's family was unable to secure an appointment at the regional developmental pediatric clinic for four months. At the initial visit, the developmental pediatrician at the regional center suggested genetic testing due to Eli's persistent delays and distinctive facial features, including a broad forehead, low-set ears, and a flattened nasal bridge. Results confirmed a diagnosis of Smith-Magenis Syndrome when Eli was 16 months old, a rare chromosomal disorder associated with intellectual disability, sleep disturbances, and behavioral challenges.

Discussion Prompts:

- What systemic barriers might the Thompson family face in accessing specialized care for Eli's Smith-Magenis Syndrome?
- What questions might the early intervention team ask to gain a better understanding of the Thompson family's routines, values, and priorities?
- Considering Eli's sensory sensitivities and rural environment, what strategies could help him engage more comfortably in community settings?
- How can teams effectively coach parents to become primary interventionists, especially in rural or underserved communities?

Part 2: Assessment

Following the genetic diagnosis, Eli's developmental pediatrician referred the family for a comprehensive developmental evaluation at the regional developmental pediatric clinic. The team consisted of a speech-language pathologist (SLP), an occupational therapist (OT), and a developmental psychologist. All assessments were conducted at the regional center staff whom neither Eli nor his parents had met before. Assessments included the Battelle Developmental Inventory, the Preschool Language Scale, and the Vineland-3 Adaptive Behavior Scales, which confirmed significant delays in communication, motor skills, and social-emotional development. They diagnosed Eli with global developmental delays and recommended continued early childhood intervention services, including speech therapy, physical therapy, and behavioral and developmental support as provided by the local early intervention program.

The team observed Eli and noted his reliance on visual support, difficulty with transitions, and sensory-seeking behaviors. Rachel shared her strategies, which included using calming music and taking nature walks. The team praised her parenting and emphasized the importance of embedding interventions into daily routines.

The team connected the Thompsons with a regional parent-to-parent support network and provided resources on Smith-Magenis Syndrome, helping the family feel more comfortable with Eli's diagnosis.

Discussion Prompts:

- How can professionals ensure that assessments are culturally and contextually sensitive for each family?
- What strategies can providers use to help families feel comfortable sharing concerns and participating in the evaluation process?
- How could the team ensure that an authentic assessment process is followed during an interdisciplinary assessment?
- How can interdisciplinary teams support families who may be unfamiliar with developmental milestones or are hesitant to seek services?

Part 3: Transition Planning from IFSP to IEP

When Eli began early intervention, he received home-based visits once a month from both a pediatric physical therapist and speech therapist (each separately). An early childhood special educator visited every other week. The team coordinated their visits and the input they provided to the family. Each visit included a combination of assessing Eli's progress, listening to and clarifying Rachel's concerns, discussing and practicing interventions with her, reviewing data collection strategies, and determining how the interventions could be integrated into the family's daily routines. As Eli's developmental profile became more defined, the early intervention team shifted their focus toward transitioning him into the public school's preschool special education program. Although the Thompsons lived in a rural location, they were assured that the interdisciplinary team from the regional center was available for teleconsultations at the public school.

During the first transition meeting with the early intervention program staff and the local education agency staff, Rachel shared her hopes for Eli. She wanted him to be able to express his needs and preferences, especially during moments of frustration. She hoped he could participate more in family routines, like mealtimes and outdoor play, and learn to navigate transitions without distress. She also wanted him to have friends from the community.

The team listened closely and helped Rachel shape these hopes into three primary goals to be implemented in preschool:

1. Increase Eli's ability to communicate using visual supports, gestures, words, and an augmentative speech device.
2. Support Eli's participation in daily routines, both at home and in school.
3. Reduce frustration and promote emotional regulation during transitions and activities.

The preschool team described their model of using naturalistic, play-based interventions in an inclusive classroom comprising both preschoolers with and without disabilities. They also described the behavioral strategies that guided their classroom grounded in positive behavioral interventions. They demonstrated to the parents how they used data to track the events leading up to a meltdown and the outcomes afterward, helping to identify Eli's triggers and prevent future episodes. They also showed how they reinforced Eli's appropriate communication, such as using a preferred toy or giving a hug, to encourage positive behavior.

Throughout the process, the team emphasized culturally responsive practices, validating the strengths Rachel reported and adapting strategies to fit the family's lifestyle.

Discussion Prompts:

- What are the benefits and limitations of embedding interventions into daily routines versus using structured therapy sessions?
- What strategies can be used to build trust with families and encourage their active participation in decision-making?
- What tools and techniques can be used to monitor progress and adjust interventions in a telehealth model?