Case Study: Chromosomal Deletion

Part 1: Getting to Know Leo

Leo and His Family

Leo (30 months) lives in a modest suburban neighborhood with his mother Sofia (33), father Daniel (35), and older sister Mia (6). The Garcia family identifies as Hispanic and maintains strong cultural traditions, including weekly family gatherings, shared meals, and bilingual communication at home. Spanish is spoken primarily with extended family, while English is used more frequently in public settings and with Mia's schoolwork.

Sofia is a full-time caregiver who left her job as a preschool assistant to focus on Leo's needs after his developmental delays became more apparent. Daniel works long hours as a mechanic at a local auto shop, often taking weekend shifts to help cover medical expenses. The family's income is modest, and they face challenges accessing specialized healthcare services due to limited insurance coverage and transportation barriers. Most of Leo's care is coordinated through their trusted pediatrician, who has been supportive but has limited experience with rare genetic disorders.

The Garcias live in a two-bedroom apartment near a busy street. Mia is a bright and nurturing sibling who often tries to include Leo in her imaginative games, though he prefers solitary play and becomes overwhelmed by loud noises or unpredictable changes. Sofia has noticed that Leo is drawn to music and lights, often spending long periods watching ceiling fans or tapping rhythmically on surfaces.

Family routines are structured around meals, naps, and evening story time. Sofia incorporates sensory-friendly activities into Leo's day, such as water play and soft-textured toys, and she uses simple Spanish and English phrases to encourage communication. Despite her efforts, Leo's expressive language remains limited, and he often uses gestures or vocalizations to communicate needs. Sofia has expressed feelings of isolation and uncertainty, especially when trying to explain Leo's diagnosis to extended family members who are unfamiliar with chromosomal disorders.

Medical and Developmental History

Sofia's pregnancy with Leo was largely uneventful, with routine prenatal screenings showing no major concerns. However, she recalls that Leo was less active in utero compared to Mia, and she experienced mild polyhydramnios in the third trimester. Leo was born full-term via vaginal delivery, but his Apgar scores were slightly lower than expected due to poor muscle tone and weak initial cries.

From birth, Leo exhibited signs of hypotonia, or low muscle tone,, which made feeding difficult. He struggled with sucking and swallowing, leading to early concerns about weight gain and hydration. A feeding specialist was consulted within the first few weeks, and Sofia began using specialized techniques to support Leo's nutrition. Despite these efforts, Leo remained underweight for much of his first year.

Developmentally, Leo showed delays in motor milestones. He did not sit independently until 12 months and did not crawl until nearly 18 months. Walking was achieved at 24 months, but his gait remains unsteady. His muscle tone has gradually improved with physical therapy, though he continues to require support for balance and coordination.

Leo's expressive language is significantly delayed. He uses a few vocalizations and gestures but does not yet form words. His receptive language appears stronger; he responds to familiar routines and recognizes his name. Behaviorally, Leo exhibits sensory-seeking behaviors, such as rocking and tapping, and becomes easily overwhelmed by loud sounds or crowded environments. He has occasional self-injurious behaviors, including head banging when frustrated, and throws objects during transitions.

At 12 months, Leo's pediatrician referred him for genetic testing due to his distinctive facial features such as small head, deep-set eyes, flat nasal bridge, and a small, pointed chin as well as developmental delays.

Discussion Prompts:

- What systemic barriers might the Garcia family face in accessing specialized care for Leo's delays?
- How can professionals advocate for equitable access to genetic counseling, developmental services, and transportation resources?
- What questions might the early intervention team ask to better understand the Garcia family's priorities, routines, and cultural values?
- Considering Leo's sensory sensitivities and the family's living environment near a busy street, what environmental modifications might help reduce overstimulation and promote comfort? How can early intervention professionals collaborate with the family to create sensory-friendly spaces at home and in community settings?

Part 2: Screening and Assessment

At 12 months, Leo's pediatrician referred him for genetic testing due to his distinctive facial features such as small head, deep-set eyes, flat nasal bridge, and a small, pointed chin. Additionally, Sofia and Daniel had shared their concerns about his limited motor skills, lack of verbal communication, and unusual behaviors, such as prolonged fixation on lights and difficulty transitioning between activities to their pediatrician, Dr. Patel, during a routine well-child visit.

Dr. Patel administered the Ages and Stages Questionnaire (ASQ) 12 months, a developmental screening tool designed to identify delays across multiple domains. Leo's scores raised red flags in communication, motor development, and problem-solving. Based on these results and Leo's distinctive facial features Dr. Patel referred the family for genetic testing.

The chromosomal microarray analysis confirmed a diagnosis of 1p36 Deletion Syndrome, a rare microdeletion disorder affecting the tip of the short arm of chromosome 1. The family met with Genetic Counselor Elena Ruiz, who gently guided them through the implications of the diagnosis. Elena explained that children with 1p36 Deletion Syndrome often experience congenital hypotonia, feeding and growth challenges, developmental delays, and may be at risk for seizure disorders and intellectual disabilities. She provided printed resources in both English and Spanish and encouraged the Garcias to connect with support organizations such as 1p36dsa.org.

Sofia described the meeting with Elena as emotional but informative. She appreciated the counselor's warmth and clarity, though she admitted feeling overwhelmed by the medical terminology and uncertain about what the diagnosis would mean for Leo's future. Daniel asked practical questions about therapy options and long-term care, while Sofia focused on how to explain the diagnosis to extended family members.

Following the genetic confirmation, a team of developmental specialists visited the Garcia home to conduct a comprehensive evaluation. Rachel Kim, a bilingual early intervention specialist, and Jamal Thompson, a developmental psychologist, spent time observing Leo in his home. They noted his limited expressive language, reliance on gestures and vocalizations, and sensory-seeking behaviors such as rocking and tapping. Leo showed difficulty transitioning between activities and exhibited self-injurious behaviors, including head banging when frustrated.

Rachel engaged Sofia in conversation about Leo's daily routines, sensory preferences, and communication attempts. She praised Sofia's use of bilingual cues and sensory-friendly activities and emphasized the importance of embedding interventions into everyday moments. Jamal focused on Leo's emotional regulation and behavioral responses, recommending strategies to reduce frustration and support smoother transitions.

The assessment team concluded that Leo would benefit from a multidisciplinary early intervention plan, including speech therapy, occupational therapy, and parent coaching. They emphasized the importance of culturally responsive practices and ongoing collaboration with the family to ensure that interventions aligned with their values, routines, and goals.

Discussion Prompts:

- What strategies can genetic counselors use to ensure families feel supported, informed, and empowered when receiving a rare diagnosis?
- How can professionals ensure observations are both culturally sensitive and clinically informative?
- What strategies can providers use to ensure families understand the questions and feel comfortable sharing concerns?
- How can interdisciplinary teams support families who may be unfamiliar with developmental milestones or hesitant to disclose concerns?

Part 3: Transition Planning from IFSP to IEP

As Leo neared his third birthday, the early intervention team began preparing for his transition from an Individualized Family Service Plan (IFSP) to an Individualized Education Program (IEP). This process was initiated with a collaborative meeting that brought together a multidisciplinary team, including Elena Ruiz, the family's genetic counselor; Dr. Patel, Leo's developmental pediatrician; Rachel Kim, his bilingual early intervention specialist; a speech-language pathologist (SLP), an occupational therapist (OT), and the family's service coordinator.

During the meeting, Sofia and Daniel expressed both hope and concern. They were proud of Leo's progress, especially his increased engagement during play and his use of visual supports but remained deeply concerned about his limited expressive language and episodes of frustration. They emphasized the importance of culturally responsive services that respected their bilingual home and family traditions. The team reviewed Leo's current intervention strategies and discussed how they could be adapted and expanded within a preschool setting.

First, Parent-Implemented Interventions (PII) had been a cornerstone of Leo's progress. Sofia had learned to embed communication strategies into daily routines, such as mealtime and music time, using simple signs, visual cues, and positive reinforcement. The team discussed how these strategies could be modeled for preschool staff and incorporated into classroom routines. Additionally, Leo had responded well to Visual Supports and the Picture Exchange Communication System (PECS). These tools helped him express preferences and transition

between activities with less distress. The team recommended continuing PECS in his IEP, along with visual schedules and choice boards tailored to his sensory needs.

To address Leo's self-injurious behaviors, the team implemented ABC logs and Functional Behavioral Assessments (FBA). These tools helped identify triggers and develop proactive strategies, such as sensory breaks and calming routines. The IEP would include a behavior support plan informed by these assessments. Leo's physical therapy sessions focused on improving balance, coordination, and strength. While his muscle tone had improved, he still required support for gross motor tasks. The team recommended continued PT services within the school setting, with goals aligned to classroom participation. Finally, the team emphasized the importance of family education and support. Sofia and Daniel had benefited from resources provided by 1p36dsa.org and a local support group. The service coordinator offered to help them connect with school-based parent networks and advocacy organizations to ease the transition.

Discussion Prompt:

- How can early intervention teams ensure that the transition from IFSP to IEP remains family-centered and culturally responsive? What steps can be taken to support Sofia and Daniel in understanding their rights and roles in the IEP process?
- What strategies can be used to build trust and empower families to actively participate in decision-making?
- How should teams determine which EBPs are most appropriate for a child's developmental profile and family context?
- Leo has made progress with parent-implemented interventions, PECS, and sensory supports. How can these strategies be adapted and sustained in a preschool setting? What role should the early intervention team play in training school staff?