ECiDC Early Childhood Intervention Doctoral Consortium

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

Case Study: Joubert Syndrome

Part 1: Getting to Know Maya

Maya and Her Family

Maya (36 months) lives in a quiet residential neighborhood with her mother Priya (32), father Arjun (34), and older brother Kiran (7). The Patel family identifies as South Asian and speaks both Gujarati and English at home. English is used primarily for school and medical appointments, while Gujarati is spoken during family meals and cultural gatherings.

Priya left her job as a dental assistant to care for Maya full-time after her diagnosis. Arjun works as a truck driver and often takes extra shifts to help cover medical costs. The family's financial resources are limited, and they rely on public insurance and local clinics for Maya's care. Their pediatrician has been supportive but unfamiliar with Joubert Syndrome, which has made specialist referrals more challenging.

Maya enjoys quiet play and is fascinated by lights and textures. She often watches ceiling fans spin and enjoys playing with soft fabrics. Her brother Kiran is protective and tries to include her in games, but Maya prefers solitary activities and becomes overwhelmed by loud sounds or unpredictable changes. Priya has created a structured daily routine with sensory-friendly activities, including water play, music time, and visual schedules.

Despite her efforts, Maya's speech remains limited. She uses gestures and vocalizations to communicate, and Priya has started using picture cards to support her understanding. The family has expressed feelings of isolation, especially when trying to explain Maya's rare diagnosis to extended relatives who are unfamiliar with genetic disorders.

Medical and Developmental History

Priya's pregnancy with Maya was uneventful, but Maya was born with low muscle tone and irregular breathing. She was diagnosed with Joubert Syndrome at 18 months following an MRI that revealed the Molar Tooth Sign (MTS) which is a distinctive brain malformation associated with JS. Genetic testing confirmed variants in one of the known JS-related genes.

Maya has congenital hypotonia, which affected her ability to feed and meet motor milestones. She sat independently at 14 months and began walking at 30 months with support. Her gait remains unsteady due to ataxia. She receives physical therapy to improve coordination and strength.

Her expressive language is significantly delayed. She does not yet form words but responds to familiar routines and recognizes her name. Maya exhibits abnormal eye movements (nystagmus and strabismus), and her vision is being monitored. She also has kidney abnormalities and sees a nephrologist annually.

Behaviorally, Maya shows sensory-seeking behaviors, such as tapping and rocking, and becomes distressed during transitions. She has occasional tantrums and throws objects when overwhelmed.

Discussion Prompts:

- 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?
- How can professionals support families navigating unfamiliar medical terminology and fragmented care systems?
- Should Maya and her family have so many developmental professionals coming to their home? Describe other options to meet Maya's needs?

Part 2: Screening and Assessment

At 18 months, Maya's pediatrician referred her for neuroimaging due to concerns about delayed motor milestones, abnormal eye movements, and irregular breathing patterns. A brain MRI revealed the characteristic Molar Tooth Sign (MTS), a hallmark of Joubert Syndrome, indicating cerebellar vermis hypoplasia and brainstem malformation. This confirmed the diagnosis of Joubert Syndrome.

Following imaging, genetic testing was conducted using chromosomal microarray and targeted gene panels. Results identified a mutation in one of the known autosomal recessive JS-related genes, confirming the molecular diagnosis.

The family met with Dr. Mehta, a genetic counselor, who explained the implications of the diagnosis. She provided bilingual resources in English and Gujarati and connected the Patels with the Joubert Syndrome & Related Disorders Foundation (JSRDF). Priya described the meeting as helpful but emotionally overwhelming. Arjun asked practical questions about therapy and long-term care, while Priya focused on how to explain the diagnosis to extended family members.

Following Maya's diagnosis of Joubert Syndrome at 18 months, her care team initiated a comprehensive multisystem evaluation to identify and monitor potential complications associated with the condition. This approach was guided by clinical recommendations from the Joubert Syndrome & Related Disorders Foundation and aimed to address the syndrome's wide-ranging effects on neurological, visual, renal, hepatic, respiratory, and endocrine systems.

Maya's developmental pediatrician began with a detailed neurological assessment, focusing on her motor coordination, hypotonia, and signs of ataxia. Given the cerebellar malformations associated with Joubert Syndrome, ongoing monitoring of her cerebellar function and developmental progress was recommended to guide therapeutic interventions and track emerging needs.

An ophthalmologic evaluation followed, including an electroretinogram (ERG) to assess retinal dystrophy and Leber's congenital amaurosis both conditions that can accompany JS. Maya also underwent regular eye exams to monitor her nystagmus, strabismus, and potential optic colobomas. These assessments helped determine the extent of her visual impairment and informed the use of visual supports in her daily routines.

Because kidney abnormalities are common in children with JS, Maya was referred to a pediatric nephrologist. A renal ultrasound was performed to screen for nephronophthisis and cystic kidney disease. Her care plan now includes annual kidney function tests to monitor for any progression toward renal insufficiency.

To evaluate her liver health, Maya underwent liver function tests and an abdominal ultrasound. These screenings aimed to detect early signs of hepatic fibrosis or structural abnormalities, which can occur in some JS subtypes.

Given Maya's history of irregular breathing patterns, a sleep study was conducted to assess for apnea and other respiratory concerns. Her pediatrician also recommended ongoing respiratory monitoring, especially during sleep and illness, to ensure her safety and well-being.

Finally, Maya received an endocrine and craniofacial evaluation. This included screening for tongue hypertrophy, oral anomalies such as multiple frenulae, and polydactyly. Her growth patterns and hormonal development were also monitored for signs of hypothalamic dysfunction, which can affect children with JS.

These assessments were coordinated across multiple specialties, requiring careful scheduling and communication among providers. Priya and Arjun expressed gratitude for the thoroughness of the evaluations but also shared feelings of being overwhelmed by the number of appointments and the complexity of the information. The care team worked closely with the family to ensure that results were explained clearly and that follow-up plans were manageable and culturally responsive.

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: Home-based Services and Family Support

With Maya's diagnosis of Joubert Syndrome and her complex developmental and medical needs, home-based services became a cornerstone of her early intervention plan. These services were designed not only to support Maya's growth but also to empower Priya, who spends most of her time caring for Maya at home.

A team of professionals, which included a bilingual early intervention specialist, a speech-language pathologist, an occupational therapist, and a physical therapist who visited the Patel home regularly. Their approach emphasized parent-implemented interventions, recognizing Priya's deep involvement and commitment to Maya's daily care.

During sessions, therapists modeled strategies that Priya could integrate into everyday routines. For example, the speech therapist introduced visual supports and gesture-based communication tools to help Maya express her needs during mealtime and play. Priya learned to use picture exchange systems (PECS) and simple sign language, which helped reduce Maya's frustration and increase her engagement.

The occupational therapist worked with Priya to create sensory-friendly spaces in the home, using soft textures, calming lights, and predictable routines to support Maya's sensory regulation. Together, they developed a toolkit of activities such as water play, music time, and tactile exploration that Maya could enjoy safely and comfortably.

Physical therapy sessions focused on strengthening Maya's core muscles and improving her balance. Priya was taught exercises she could do with Maya during diaper changes, bath time, and outdoor walks. These embedded strategies helped Maya make progress while fitting naturally into the family's daily life.

Beyond direct services, the early intervention team provided parent coaching and emotional support. They recognized Priya's feelings of isolation and uncertainty and offered resources in Gujarati and English to help her explain Maya's diagnosis to extended family. The team also connected Priya with local support groups and online communities through the Joubert Syndrome & Related Disorders Foundation, giving her a sense of belonging and access to shared experiences.

Through consistent collaboration, the home-based services not only supported Maya's development but also strengthened Priya's confidence and capacity as a caregiver. The team emphasized that Priya's insights and cultural values were central to Maya's care, and they worked to ensure that all interventions were respectful, responsive, and sustainable within the family's routines.

Discussion Prompts:

- Given the wide range of potential complications in Joubert Syndrome, how should early childhood special education professionals prioritize developmental goals while remaining responsive to emerging medical needs?
- What strategies can be used to build trust and help 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?
- How can the team utilize a transdisciplinary team approach to minimize the number of direct service professionals serving Maya and her family?