Characteristics and Etiology of Infants and Young Children with Disabilities

Chromosome Deletion

Case Study

Objectives:

- At the end of each section stop and assess: Is there an intervention that could occur? What areas could we assist the family with? Is there something else the medical professionals should consider?
- Look through the lens of the parents: Would you be overwhelmed? Worried? What questions would you have?

Birth

Lexi was born on February 18th, 2011. She weighed 4lbs 4oz and was 41 weeks gestation on her date of birth. Her mother's pregnancy was typical, all milestones were met, and it was an unremarkable pregnancy until a few weeks prior to birth. Her mother experienced contractions that felt like labor pains prior to her 38-week appointment. Her midwife was unavailable for an appointment, so she met with the on-call physician. Written off as Braxton-Hicks, mom went home. Lexi's mom continued to feel that something was different. On February 18th, mom went in for a stress test and was immediately admitted. When the time came for delivery, Lexi's heart rate would drop during each contraction. Doctors performed an emergency C-section and Lexi was delivered. Her APGAR tests were low, and she responded very slowly but was active. Prior to birth, her birth weight was estimated at seven pounds, which was significantly different than her actual birth weight.

Lexi's mother lost weight during the final weeks of pregnancy and Lexi was less active. It was determined that her labor pains were actual labor, and mom's body assumed the baby was delivered and no longer provided essential nutrients to Lexi. Lexi was moved to the Neonatal Intensive Care Unit (NICU) for monitoring. She was diagnosed with failure to thrive and hydrocephaly. Lexi was in the NICU for seven days before discharge.

Critical Thinking: Reflect on and discuss the discussion questions.

Early Development

Once Lexi was home, things felt different. She was breastfed but did not have a good suckle reflex. She did not move much but was responsive. Lexi began to gain weight but was small in stature. As appointments progressed, she continued to be monitored. Mom noticed that Lexi was not hitting her developmental milestones. She was not grasping, making eye contact, smiling, or attempting to hold her head up. Mom's initial gut feeling was that she was on the autism spectrum. This was mom's fourth child, and except for her brother who had health issues, all the previous children hit or exceeded the developmental milestones.

At the next doctor's appointment, Lexi's parents mentioned to the physician that something was different with Lexi. Her parents requested for her to be tested to attempt to figure out what was going on with their daughter. The doctor recommended that the parents wait, as most diagnoses of autism cannot be made until the age of two. Due to the perseverance of Lexi's mom, she was genetically tested which provided an answer to the family's concerns.

Critical Thinking: Reflect on and discuss the discussion questions.

Diagnosis

After genetic testing and a microarray test, it was determined that Lexi has 1p36 Microdeletion Syndrome. 1p36 is a genetic disorder where multiple base pairs have been deleted from the tip of the first chromosome. The deletions vary in size from person to person and cause global delays across all areas of the body. In Lexi's case, she was missing over 12 million base pairs on her first chromosome. Lexi's parents were relieved to know that this was the cause of her differences, but also sad and scared for the unknown challenges Lexi and her family would face. There are multiple iterations of this syndrome, and each individual is affected differently. Some children pass away as infants due to medical complications, some graduate college. There can be developmental and intellectual disabilities. There are often additional medical diagnoses that accompany the disorder like epilepsy, autism, cardiomyopathy, respiratory and vascular issues to name a few. This was a lot for Lexi's parents to take in and accept.

After the diagnosis, Early Intervention services would need to be initiated, therapies would begin, and figuring out how to communicate and understand their little one would begin.

Critical Thinking: Reflect on and discuss the discussion questions.

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