

Spencer's Story

Spencer is a 9-month-old baby with Cornelia de Lange Syndrome (CdLS). The diagnosis was made shortly after Spencer was born. He was born with very short arms and missing fingers on one of his hands. He also had a cleft palate, which was repaired two weeks after his birth. Spencer's parents, Nicola and Ricky, met with a geneticist a few days after Spencer was born. Although the genetic testing was not complete, the geneticist talked with them about CdLS.

Nicola and Ricky live in the Milwaukee area. At the time Spencer was born, Ricky worked for an overnight custodial service, and Nicola was unemployed. They were 18 and 19 years old when Spencer was born, unmarried, but living together in a one-bedroom apartment. Nicola had wanted to go to school to be a certified nursing assistant (CNA). When she found out she was pregnant, she did not enroll in the classes at their high school that would have helped her get into the program at the community college. Neither Nicola nor Ricky finished high school. Nicola's mother works at the Women, Infants, and Children (WIC) office greeting families and doing clerical work. Nicola's father left the family when she was a baby and has not been heard from since. Ricky's parents are not supportive of Nicola and Ricky. They tried to convince Nicola and Ricky that they should put the baby up for adoption after they found out that Nicola was pregnant. When the couple said they wanted to keep the baby, Ricky's parents shut them out.

At 9 months of age, Spencer is looking a lot like a 3- to 4-month-old in his development. He can hold his head up, and he gives them quick smiles when they talk to him but looks away and sometimes screeches. He is not yet rolling, and Nicola wonders if this is because his arms are so short or if he just isn't strong enough. He does use his "normal" hand to hold a toy and can get it to his mouth. He occasionally brings his other arm up to try to grab it. He sometimes likes to snuggle, but when he is out of sorts, he is very hard to console and does not seem to want to be touched or held. He arches and screams. This is really hard for Nicola, especially when she is home alone with him. Spencer likes things that light up, and sometimes the only way he will settle is when she holds a light-up toy right in front of his face.

Spencer has been on a g-tube since birth. Nicola and Ricky got the go-ahead to try to feed him some pureed baby foods when he was about 7 months old. They have not had much success; Spencer just doesn't seem to want or hold anything in his mouth. They either hold him or prop him with pillows in a high chair to feed him. Neither position makes much difference in his willingness to take food. The gastroenterologist (GI doctor) is concerned that Spencer is not making progress in his feeding. He is gaining weight and growing because of the g-tube. Nicola thinks if she could just get Spencer to eat, things would get better; he'd be more settled and their lives would be easier.

Spencer is making a few sounds but not really babbling. Nicola and Ricky were told they needed to get his hearing tested, but they have not made that appointment yet. Spencer's eyes are both goopy and crusty. Nicola and Ricky missed an appointment with the ophthalmologist because Spencer was sick and haven't had time to reschedule it.



Typical and Atypical Child Development
Module 1: Birth through 3 Years of Age
Case Study

Spencer is getting Birth to 3 services. An occupational therapist (OT) comes once a week for an hour. They work on feeding and sensory concerns. Nicola is not sure she is getting what she needs from Birth to 3, because Spencer is not feeding well and is so hard to settle.

Nicola and Ricky feel like they are very lucky in that Spencer does not show any signs of having a heart problem, but they know that this could still show up. For Nicola, it's one more thing that could go wrong and probably will.

When things get really tough at home during the day, Nicola puts Spencer in the stroller and walks over to the WIC office to be with her mom. This both helps and depresses Nicola. Her mom and the staff are always welcoming, but seeing other babies who look and act "normal" is so hard. She doesn't feel like "a mom." She can't breast feed. Her baby is so hard to settle, has funny looking arms that don't hold toys or reach for her, and screeches more than he babbles.

Summary

Nicola and Ricky are most concerned about Spencer's feeding. They want him off the g-tube, but they can't get him to eat. He just gets fussy and refuses. Spencer's moods and fussiness are a huge problem. Nicola is exhausted. She knows she is not getting to appointments but wishes Ricky would be more helpful. Ricky is unavailable during the day because he sleeps.

Spencer's diagnosis of CdLS puts him at risk for global developmental delays and sensory impairments. His feeding tube may become permanent. He has the potential for developing cardiac problems.

Follow-Up Questions

1. *How often is Spencer fed through his g-tube? When feeding orally, are they also trying to give him formula through a bottle, or are they only trying pureed solids? How long does an oral feeding typically last?*
2. *He is noted to be hard to console – how long does it typically take to calm him down, and how often is he so irritable that he is difficult to console?*