1 in 10,000: Love between family, girl with genetic disorder inspires

By Victor R. Martinez / El Paso Times El Paso Times
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There are times -- many times – when Lily Lopez grows tired of the curious looks she gets when she takes her 10-year-old daughter Kayla out in public. "It's hard enough on us at is without all those people staring at us, saying 'Oh my God, did you see the baby? Did you see her?' I still haven't gotten used to it and it still gets on my nerves, but I've learned to ignore it better."

Lopez's daughter was born with Cornelia de Lange Syndrome (CdLS), a genetic disorder that creates physical, cognitive and medical challenges. The occurrence of CdLS is estimated to be 1 in 10,000 live births.

"The doctors thought something was wrong with her when I was about six months pregnant," Lopez said. "She was missing a limb and she was very small, but they had to wait until she was born to diagnose her. When she was born she didn't have a right arm and she only had two digits on her left hand."

In 2004, researchers at The Children's Hospital of Philadelphia and the University of Newcastle identified a gene named NIPBL on chromosome 5 that causes Cornelia de Lange Syndrome when it is mutated or changed. Since then, two additional genes -- SMC1A and SMC3 -- have been found that cause CdLS when changed.

"We loved her from the start," said Maria Ochoa, Kayla's grandmother said. "The doctor said she might not live a long time but she is 10 years old and she goes to school. She's just like a normal kid -- she sleeps in late and doesn't like going to school."

Ochoa said the most difficult part of the process was when the doctors told the family that Kayla had CdLS.

"That was really, really difficult for us because we did not understand why this happened to her," she said. "After she was born, we knew she was going to have problems, we knew she was going to be sick so we were prepared for it."

Common characteristics of CdLS include low birth weight (usually, but not always, under 5 pounds), delayed growth and small stature, and small head size.

Typical facial features include thin eyebrows which frequently meet at midline, long eyelashes, short upturned nose and thin, down-turned lips.

"She really doesn't have a full understanding of what's going on around her," Lopez said. "She is very mentally delayed; even now, she barely started looking at me in the eyes. She is actually a very happy girl. The only thing that bugs her is very loud noises or when people touch her unexpectedly."

Kayla enjoys painting with her 3-year-old brother Jose and loves listening to alternative rock.

"She smiles and taps her toes when I listen to Green Day," Lopez said. "She loves her brother and you can tell he loves her. To him, she is perfectly normal. He doesn't see her as different."

Kayla has a regular pediatrician in El Paso and also sees a specialist in Albuquerque.

"She was born in Las Cruces and three days after that, we had to move to Albuquerque for three months," Lopez said. "I was a teen mom, so I had to drop out of school and leave everything behind to be with Kayla."

Lopez eventually earned a high school degree and then started college, where she was studying to be a nurse.

"I was at the hospital for three months from 6 in the morning to 7 at night and I saw everything the nurses do for the kids," Lopez said. "They inspired me to become a nurse so can help children also."

But until then, Lopez has a full-time job taking care of Kayla.

"She is going to be dependent for the rest of her life," she said.

"She doesn't talk, she doesn't eat by mouth, she has a feeding tube, she doesn't walk on her own, so I'm going to have to watch over her for the rest of her life," she said. Lopez feels she is blessed to have a daughter like Kayla. "Nobody can take care of her or love her the way I do," she said. "A few years ago, the doctor found a lump on my breast and I thought I was going to die of cancer. Right away I thought about Kayla and who would take care of her? Who would love her? That was scary. Nobody else in this world could take care of her like I could."

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Characteristics of CdLS

Individuals with Cornelia de Lange Syndrome (CdLS) may have many of the following traits, or a select few. Geneticists establish the diagnosis after evaluating all the criteria:

Birth weight, growth and head size: The average birth weight for children with CdLS is 5 pounds, 1 ounce, but birth weights have been reported ranging from 1 pound, 2 ounces to 10 pounds. The average birth length is approximately 18 inches. Children with CdLS are often short and below average in weight when compared to others their age. Small head size (microcephaly) is a feature commonly associated with the syndrome.

Developmental delays: The vast majority of children diagnosed with CdLS are intellectually delayed, with the degree ranging from mild to severe. Learning disabilities and severe language delays are often present. Although intellectual delay is considered essential for diagnosis, there have been cases of people with CdLS who have near normal to normal intelligence.

Gastroesophageal Reflux Disease (GERD): It's estimated that 85 percent of people with CdLS experience some type of gastroesophageal reflux. The pain that can accompany GERD can make eating unpleasant and lead to a variety of behavioral problems.

Behavioral issues: People with CdLS may exhibit a number of behavioral problems such as self-injury (head-banging, hand-biting, etc.), compulsive repetition, and/or autistic-like behaviors. Anxiety, Obsessive Compulsive Disorder, Attention Deficit Disorder and Attention Deficit Hyperactivity Disorder have also been noted.

The following are secondary characteristics of CdLS:

Facial features: Prominent facial features include thin eyebrows that often meet at the midline (synophrys), long eyelashes, short upturned nose, thin downturned lips, low-set ears, and high-arched palate or cleft palate.

Limb differences: Small hands and feet, in-curved fifth fingers (clinodactyl), partial joining of the second and third toes, proximally placed thumbs, and upper limb abnormalities, including missing fingers, hands or forearms.

Other system abnormalities: Eye ailments such as blepharitis (inflammation of the eyelid), faulty or nonexistent tear ducts, ptosis (droopy lids), and extreme nearsightedness (myopia) can occur. Other medical concerns can include hearing loss, communication delays, feeding difficulties, seizures, heart defects, bowel abnormalities, undescended testes, and purplish discoloration of the skin (cutis marmorata).

Source: www.cdlsusa.org.