



CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.
Reaching Out, Providing Help, Giving Hope

Eye Care – The Families’ Perspective

By Lynn P. Audette, M.S.W., Family Service Coordinator

Like so many of us, children with CdLS rely on their vision to help them take in information about the world around them. This is especially true for people who do not communicate verbally but use communication systems like sign language, PECS (Picture Exchange Communication System), or computerized technology. The quality of their eyesight and health of their eyes is crucial to their ability to navigate in the world. The following family stories illustrate various eye conditions associated with CdLS and information on how families have dealt with them.

Kadin

Kadin (now two and a half) was diagnosed with ptosis (droopy eyelids) at the age of five months. His eyelids looked as if they were half closed or he was sleeping. Jeneene, Kadin’s mother, noticed that “whenever Kadin was looking at something, whether it was a toy or a person, his head was always tilted back with his chin up. We were referred to a pediatric ophthalmologist at our children’s hospital. Several different measurements of his eyelids were taken and the diagnosis was confirmed as ptosis. At the time of his diagnosis, we were only given two options: patching of his eyes or having surgery to lift his lids (fontalis slings). The doctor wanted to wait to do any type of surgery until he was a little older to see if maybe his muscles would tighten up a little. When they did not, Kadin had the fontalis slings surgically inserted.”

“The care after ptosis surgery required a lot of work and time and was emotionally difficult,” Jeneene shared. “The first few days we had to put ointment into Kadin’s eyes every three hours even when he was sleeping to prevent his eyes from drying out. A week later, we needed to put the ointment in every four to six hours. Two weeks later, Kadin was able to close his eyes fully so the ointment was required only every six hours during the day.” Jeneene felt that it was worth it despite the setback Kadin faced six weeks later when his left eyelid fell. “After one of our follow up visits we were told that it would have to be redone. The doctor did want to wait until six months after his surgery to make sure that his right eyelid did not also fall.” Kadin successfully had his left eyelid redone exactly six months later.

Jeneene feels the ptosis surgery has made it possible for Kadin to make great strides with gross motor skills. Before the surgery, Kadin always had to move his head to look up and as a result, was not able to sit, crawl, or walk. Jeneene now describes Kadin as “a running machine.” She reports that “He has achieved all of his goals in physical therapy. It’s amazing what a difference it has made for him. I would definitely recommend that any family who thinks their child has ptosis see a pediatric ophthalmologist who is familiar not only with



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children with CdLS but also with ptosis. Even knowing that the particular surgery that Kadin had is not a permanent surgery and will [likely] have to be redone in the future, it was worth it to us.”

Matthew

“It was easy for us to see Matthew’s eye problems,” shared his mother Janet. He had very crossed eyes (known as esotropia). He was referred to an eye surgeon at Children’s Hospital in Boston, MA. At eleven months of age, Matthew underwent his first eye surgery which loosened the interior eye muscles. “The same afternoon of this surgery he sat up without support for the first time and pulled himself up to standing position in his crib all afternoon and evening. (And we thought we were going to get some sleep!) I tell you this because his change from lying down to standing in one day was amazing!” recalls Janet. Matthew had a second surgery about a year later to tighten the outside eye muscles.

Today, at age seven, Matthew eyes still turn inward and outward sometimes, but are very straight when he wears his glasses. He has mild dry eye and has had two sties. Matthew also had nystagmus (shaky eyes), but it is minimal. Matthew contended with temporary excessive blinking occurring two years ago. No medical reason was found and Janet suspects that it may have been due to Matthew being nervous about a new school year or to a seasonal allergy.

Randy

This past year, Patty noticed that her son Randy (18 years old) was having some vision problems that rapidly worsened. She first suspected a problem when he began repeatedly taking out the wrong utensil from the drawer at snack time. He seemed to be good at covering the fact that he his vision was almost nonexistent, forcing him to rely a lot on his memory of where things were located.

Patty took Randy to their local hospital where he required a sedative to calm him down enough for the doctor to examine his eyes. He was then sent to an ophthalmologist followed by admission to Children’s Hospital. Sedation was required again for the specialists to perform a thorough evaluation of his eyes. He was diagnosed with a detachment of the retina in his right eye. Patty shared that Randy’s self-injurious behavior involving his head likely caused the detachment and these issues were also discussed during the various evaluations.

A retina specialist presented surgery on Randy’s eye as the only treatment available. Like many other parents faced with this decision, they were extremely anxious. Not only would the procedure required anesthesia but they would also need to severely restrict Randy’s activity and mobility while his stitches healed. Going ahead with the procedure, doctors



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repaired the detachment of his retina as well as removed his cataract (cloudy lens). Much to Patty's relief, Randy's surgery was successful and Randy did better with his recuperation than they had predicted. Patty attributes this to the fact that Randy could immediately see much better!

Randy is scheduled to have the cataract from his left eye removed in the near future. He will also require a procedure to have blood flushed from that eye. According to Patty, the vitreous fluid will need to be replaced if the blood is not flushed. In spite of his rapid improvement immediately following surgery, Randy and his family have some hurdles to overcome. Randy has currently experienced difficulty in being weaned off the pain medication.

Sam

At the age of three months, Sam's eyes occasionally turned inward (this is normal before three months), prompting Debbie and John (Sam's parents) to have his eyes checked. Debbie recounted that on their initial visit the pediatric ophthalmologist dilated Sam's pupils and refracted his eyes. He could do this eye exam and make a diagnosis even though Sam was too young to communicate verbally. The doctor diagnosed Sam with myopia (near-sightedness) and recommended glasses.

For many children with CdLS, glasses can be an unwelcome but important aid. Fortunately, Sam willingly wore his glasses from a young age because they helped him see better. Although he occasionally took them off and they were broken a few times, he was generally good about wearing them. Debbie encourages parents to try elastic bands designed to fit onto the back of the glasses. These help to keep the glasses on the child's head. Debbie recommends "that parents be persistent and put the glasses back on each time your child takes the glasses off so he or she realizes that you are going to insist they keep wearing them."

When Sam turned five, he didn't want to wear his glasses anymore. "We took him to his ophthalmologist and his myopia had greatly improved (which is unusual). The doctor felt he didn't need them anymore. It was our little miracle with Sam!" Debbie concluded.

The eye issues of cataracts, detached retinas, esotropia, myopia, and ptosis are but a few that are common in CdLS. For further information please see the article "CdLS and the Eye" starting on the cover page. We thank the Hogland, Montecalvo, Nemeth, and Walks families for the information they have provided. Through their sharing, we are better able to understand what children with CdLS experience and what their parents encounter when addressing some of these health issues. If you would like to be in contact with any of these families or those whose children have faced similar eye issues, please contact Lynn at families@cclsusa.org or through the Foundation office at **1-800-753-2357**.