

MOLLY BLACK: Hi, my name is Molly Black, and I'm one of two family liaisons with the Pennsylvania Deaf-Blind Initiative. I am also the parent of a young man with Cornelia de Lange Syndrome, or CDLS, which is one of the etiologies that may contribute to deaf-blindness. It was discovered by Dr. Cornelia de Lange as a Dutch pediatrician who, in 1933, reported on two unrelated children with similar features.

According to the CDLS Foundation, as with many children with other syndromes, individuals with CDLS strongly resemble one another. Typical facial features include thin eyebrows that meet in the middle; long eyelashes; a short, upturned nose; and thin, downturned lips. Other characteristics include low birth weight, slow growth, small stature, and small head size. And other features may include excessive body hair and small hands and feet.

Common medical issues include gastroesophageal reflux disease, heart defects, seizures, feeding difficulties, vision problems, and hearing loss. Limb differences, including missing arms, forearms, or fingers are seen in about 25% of individuals with CDLS. Behavioral and communication issues and developmental delays often exist.

Today's focus, however, is on vision loss, eye disorders, and hearing loss. The most significant impacts to sight for individuals with CDLS are ptosis and severe nearsightedness. Ptosis is the mild to severe dropping of one or both eyelids, affecting approximately 44% of individuals with CDLS. The most common cause is a weakness of the upper eyelid muscle that is present at birth. Eyelids are meant to protect the eyeballs, and blinking lubricates and cleans the front of the surface of the eye.

However, if the eyelids is weak and drooping, vision can be blocked, which may lead to poor visual development, or what is called amblyopia, or lazy eye. If vision is compromised by a droopy eyelid, a child's head posture can change from the lifting of the chin in order for the child to see from under the droopy eyelids. Ptosis could lead then to interference with walking or inhibit interaction with the environment.

Nearsightedness, or myopia, is seen in almost two-thirds of patients with CDLS. Unfortunately, about half of children with CDLS who might benefit from glasses won't wear them. Persons with CDLS may have tactile or sensory issues that may lend themselves to an aversion to having something on or near the face, or they may not recognize the difference in their visual world enough to give them the incentive to wear their glasses.

High degrees of near-sightedness can result in an elongated shaped eyeball. This causes the inner lining of the eye, the retina, to be stretched, leading to retinal detachment. Retinal detachment can damage sight and even cause blindness, and requires immediate surgical repair. Head or eye trauma

from self-injurious behavior can also cause retinal detachment even if the individual is not near-sighted. The CDLS Foundation encourages discussion of any vision concerns or self-injurious behavior to the head and area with your child's pediatrician or doctor of ophthalmologist.

Another eye condition common in persons with CDLS is blepharitis. It is a condition whereby the eye has recurrent red eye, eye discharge, tearing, crusty eyelashes, styes, blinking, and/or eye rubbing. The eyelid glands that lubricate the eye flow slowly. While blepharitis is common in the general population, it's even more common in individuals with CDLS. The best treatment is washing the eyelash area with baby shampoo once or twice daily. If this treatment fails, then other causes such as infection or blockage of the tear ducts should be explored.

Although uncommon, glaucoma, high eye pressures, and congenital deformities in the shape of the eye have been reported in a small number of individuals with CDLS. Hearing loss has been widely reported in children with CDLS, and can be conductive or sensory-neural in nature. Most published reports suggest that sensory-neural hearing loss is most common in children with CDLS. However, degree of loss is unclear. Many families report that their young children have hearing loss along with hearing test results that confirm the finding.

Despite this, many families say that their child's hearing is better than the test results suggest, and that results in actually improved -- improvement as the child gets older. The mechanism behind this improvement is unclear, and it is an area that needs exploration. While it has not been studied, a diagnosis of auditory neuropathy may be something to consider in children with these findings. To confirm this diagnosis, children need to be tested with auto-acoustic emissions, in addition to an auditory brain stem response test.

A common cause for hearing difficulties are ear infections. A recent study looking at the incidence of ear infections in conductive hearing loss in children ages 1 to 18 with CDLS found that 94% had ear infections with fluid behind the eardrum, and associated conductive hearing loss. The study also found that the likelihood of having an ear infection was the same across the age range, suggesting that fluid behind the eardrums may persist in children with CDLS far longer than in typical children.

For children with moderate or severe nerve hearing loss, a trial with a hearing aid may assist in language development and/or understanding. In order to learn more about hearing difficulties in children with CDLS, a number of the children and adults have undergone computer tomography scans to look at the hearing bones and the middle ear structure. Researchers are hopeful that this information can provide insight into the nature and degree of hearing loss seen in individuals with CDLS.

The information reported here was obtained through publications and contact with the Cornelia de Lange Syndrome Foundation. If you suspect your child or your student may have CDLS, help and hope are best found by contacting the Cornelia de Lange Syndrome Foundation at info@cclsusa.org, or 860-676-8166, or 860-676-8255. Toll-free support lines at 1-800-753-2357 in the United States only, and 1-800-223-8355, also the United States only. Their mailing address is Cornelia de Lange Syndrome foundation, 302 West Main Street, #100, Avon, Connecticut, 06001.

For information regarding issues of deaf-blindness, please contact the Pennsylvania Deaf-Blind Initiative at 717-541-4960, extension 3115, or toll-free, 1-800-360-7282 in Pennsylvania. TTY, 800-654-5984.